



Trinity College Dublin
Coláiste na Tríonóide, Baile Átha Cliath
The University of Dublin

Fathers' experiences of caring for a child with a chromosome abnormality and complex care needs

A thesis presented to Trinity College Dublin for the degree of Doctor in
Philosophy

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Declaration

I declare that this thesis has not been submitted as an exercise for a degree at this or any other university and it is entirely my own work. I confirm that the work has been completed in full compliance with relevant university policies, including the Academic Integrity policy, the Trinity Policy on Good Research Practice, and the guidance on AI and GenAI in Teaching, Learning, Assessment and Research.

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Summary

Background

Chromosome abnormalities commonly result in children having associated complex care needs. While a growing body of literature examines the experiences of families caring for a child with a chromosome abnormality and complex care needs, the focus has predominantly centred on mothers. This is despite international recognition of the importance of paternal involvement and the expectation that fathers share caregiving responsibilities equally. In response to this gap, the present study explores the lived experiences of fathers who care for a child with a chromosome abnormality and complex care needs, offering insight into their unique perspectives.

Aims and objectives

The aim of this study was to explore the experiences of fathers caring for a child with a chromosome abnormality and their associated complex care needs. It focused on four key objectives which were:

1. To examine fathers' experiences of how their child was diagnosed;
2. To understand the impact that this diagnosis has on fathers;
3. To explore the roles and responsibilities fathers take on in caring for their child;
4. To identify the types of support available to them, as well as how these supports influence their daily lives.

Methodology

This study was underpinned by a hermeneutic phenomenological approach guided by van Manen (1990). Following ethical approval, purposeful sampling was conducted through two routes (homecare providers and a range of support groups), that support families of children with a chromosome abnormality and/or complex care needs. Nine fathers participated and data were collected through both in person and online interviews via Zoom, with two participants also completing an optional journal. Data were analysed using van Manen's hermeneutic phenomenological method (van Manen, 2014a), and findings were presented through his five existential themes: lived body,

lived relation, lived space, lived things, and lived time. These themes were then examined as a whole experience.

Findings

Five themes emerged, each corresponding to one of van Manen's existential lifeworld's, within which 17 subthemes were identified. The first theme, "Receiving a diagnosis of a chromosome abnormality and complex care needs, and the subsequent fight for services (lived body)", captures fathers' multifaceted emotional responses to their child's diagnosis, as well as their experiences with professional support and the systemic barriers they encountered. The second theme, "The complexity of relationships (lived relation)", explores the intricacies of navigating relationships with their children, the child's mother, and extended family members such as grandparents. The third theme, "The physical and psychological space occupied by fathers (lived space)", reflects how fathers negotiate hospital admissions, the transition home, and the emotional and practical demands of complex care decision-making. The fourth theme, "The material aspects of care (lived things)", highlights the tension fathers experience in balancing employment, financial responsibilities, and the hands-on demands of caregiving. The fifth theme, "Adapting to their child's complex care needs over time (lived time)", illustrates how fathers' perspectives and coping strategies shift in response to their child's evolving needs. When each theme is examined as a whole, this study shows that fathers are important members of the caregiving team, whose experiences are dynamic and are influenced by the changing nature of their child's complex care needs, as well as by their reflections on the past, present and anticipatory future experiences.

Conclusion

When a child is diagnosed with a chromosome abnormality and complex care needs, it marks the beginning of an emotional journey for fathers. Delays in both diagnostic and support services have a direct impact on them. Fathers' support needs evolve in parallel with the changing nature of their child's care requirements. Fathers highlight the shift that often occurs from intensive medical care to managing their children's behavioural and psychosocial needs. While the term complex care needs is often associated with

medical complexity, this study reveals that these complexities also span health, social, and educational domains. There is a need for ongoing, responsive support for fathers in their caregiving roles. It is essential that stakeholders acknowledge the complexity of chromosome abnormalities and the impact this has on fathers.

Acknowledgements

There are a multitude of people that I am extremely grateful for, and without them this study would not have been possible, and I would like to express my sincere gratitude.

Firstly, I would like to acknowledge the nine fathers who took time out of their extremely busy lives to be part of this study. The love and admiration you all have for your children shines through, and I hope this study has brought that to light.

I am extremely grateful to my supervisors who have been an endless source of support and encouragement. Firstly, to Dr. Carmel Doyle, who has been an inspiration to me since my undergraduate degree, your knowledge, outlook and advice has been invaluable not just as part of this study, but also in encouraging me and indeed many other students to partake in nursing research. Secondly, I would like to thank Professor Mary Hughes, who took a chance on me two years into this study and whose advice has been invaluable. I would like to thank you both for your continuous encouragement, supervision and guidance over the years. Thank you for being as passionate about this topic as I am. Thank you to Professor Maria Brenner who was part of the supervision team for the first two years.

Thank you to my friends who have listened to me talk of this study endlessly. A special shout out to “the girls” - I promise I will be a more present friend going forward.

Thank you to my family, especially my mum, who always believed in me and encouraged me throughout this study, your advice was greatly appreciated. Thank you to my husband Ian, for the endless encouragement, fun and laughter along the way. Thank you, to the Brennan’s, Tadhg, Finn, Denise and Katie.

I would like to dedicate this study to my own father - just for being a great dad.

List of abbreviations

ACP	Advance care planning
AON	Assessment of need
CA	Chromosome abnormality
CACCN	Chromosome abnormality and Complex Care Needs
CAMHS	Child and Adolescent Mental Health Services
CAMHS-ID	Child and Adolescent Mental Health Services - Intellectual disability
CAs	Chromosome abnormalities
CCN	Complex care needs
CCNE	Centre of Children’s Nurse Education
CDNT	Children’s Disability Network Team
CHI	Children’s Health Ireland
DOH	Department of Health
DOHC	Department of Health and Children
DSFA	Department of Social and Family Affairs
EU	European Union
EUROCAT	European Surveillance of Congenital Anomalies
GOI	Government of Ireland
GP	General practitioner
HCP	Homecare Packages
HIQA	Health Information and Quality Authority
HSE	Health Service Executive
NCHD	Non consultant hospital doctors
NG (feeding)	Nasogastric
NHS	National Health Service
NIDD	National Intellectual Disability Database
NMBI	Nursing and Midwifery Board of Ireland
NMC	Nursing and Midwifery Council
PEG (feeding)	Percutaneous endoscopic gastrostomy
PIL	Participant information leaflet
RCN	Royal College of Nursing
ROI	Republic of Ireland
SIB	Self-injurious behaviours
SNMREC	School of Nursing and Midwifery Research Ethics Committee
TCD	Trinity College Dublin
UK	United Kingdom
USA	United States of America
UN	United Nations
WHO	World Health Organisation
Zoom	Zoom Video Communications Inc.

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1 Introduction

1.1 Introduction

The purpose of this research was to explore and gain a deeper understanding of the experiences of fathers caring for a child with a chromosome abnormality and complex care needs. This introductory chapter presents a short background to the study, establishing the significance of investigating paternal experiences of caring for a child with a chromosome abnormality and complex care needs. Furthermore, this chapter outlines the research aims and objectives, provides a brief overview of the methodological approach adopted, and concludes with a summary of the overall summary of the structure of the thesis.

1.2 Background

The aim of this study was to explore the experiences of fathers caring for a child with a chromosome abnormality (CA) and associated complex care needs (CCN). For the purposes of this research, children who have a CA and associated CCN will be referred to as children with a "CACCN". CAs can cause multi-domain disabilities that can include a combination of physical disabilities, health problems, intellectual disabilities, psychiatric/mental health disorders, and behavioural difficulties (Ajitkumar et al., 2020; Anzick et al., 2020; Boczek et al., 2018; Fermin Gutierrez et al., 2020; Kawai et al., 2024; Lizarazo et al., 2019; National Human Genome Research Institute, 2016; Schwartz et al., 2021; Uygur et al., 2026; Wilson et al. 2025). The combination of these disabilities can result in children having CCN. Life expectancy of children with often rare conditions have increased considerably in recent decades, albeit these children commonly survive with increased complexities (Abebe et al., 2020; Aitchison et al., 2020; Aleksander et al., 2025; Curran et al., 2020; Gulla et al., 2020; Kato et al., 2024; Marshall et al., 2017).

In the Republic of Ireland (ROI), there are 131,764 children with a disability, accounting for 10.8% of the child population (Department of Children, Disability and Equality, 2025). The majority of children with a CACCN reside within the family home, where parents

play a central role in providing direct care and ongoing support. According to the most recent Census, over one fifth (21.3%) of individuals with a disability in the ROI are children living in private households. While the Health Information and Quality Authority (HIQA) estimates that there are only about 192 children with disabilities living in residential care (HIQA, 2025). These figures highlight the importance of gaining a comprehensive understanding of parents lived experiences. Such insights are essential for healthcare professionals to appreciate the realities of parenting a child with a CACCN and to inform the development of holistic, family-centred care and targeted interventions (Barratt et al., 2025). However, the discourse on CACCN focuses predominantly on mothers, or mothers' voices are significantly outweighing those of fathers (Caples et al., 2018; Celik & Kara, 2023; Currie & Szabo, 2020; Desimpelaere et al., 2024; King et al., 2025; LeGrow et al., 2022; Nygård et al., 2025; Page et al., 2020; Patty et al., 2024; Richardson et al., 2021; Rossi et al., 2021; Thunberg et al., 2016; Whiting, 2013).

There is a disproportionate amount of empirical evidence between mothers and fathers in which mothers' voices outweigh fathers. This is despite the significant global social, economic, and demographic changes over the last 40 years suggesting that traditional parental roles are evolving (Barratt et al., 2025). There is increased acknowledgment that fathers, as well as mothers, should have a caring and nurturing relationship with their children, and that fathers tend to play a more active role in the parenting of their children than ever before (Cabrera & Tamis, 2013; Lamb, 2010; Planalp & Braungart-Rieker, 2016). Therefore, this study explores the experiences of fathers caring for a child with a CACCN.

1.3 Aims and objectives

The aim of this study was to explore fathers' experiences of caring for a child with a CACCN.

To meet this aim, the following objectives were established:

- To explore fathers' experiences of how their child was diagnosed;

- To develop an understanding of how this diagnosis impacts fathers;
- To explore the roles and responsibilities fathers have in caring for their child with a chromosome abnormality and complex care needs;
- To identify what supports fathers have and how these supports impact their everyday life.

1.4 Research design

There are a variety of different types of research designs that could be utilised in exploring the phenomenon of fathers' experiences of caring for a child with a CACCN. The hermeneutic phenomenology design allows researchers to develop a deep understanding of a phenomenon (van Manen, 1990). Hermeneutic phenomenology guided by van Manen is the approach chosen to support the exploration of fathers' experiences of caring for a child with a CACCN in this study. Van Manen believes that hermeneutic phenomenology is about uncovering the meaning of a lived experience by incorporating both interpretive and descriptive features and has been proven to be a beneficial research design in exploring the lived experience of parents of children with CACCN. Furthermore, van Manen (1990, 1997), understands and recognises the researcher in the research process, and acknowledge their own understandings and experiences; this is a significant aspect of this study. In addition, the element of van Manen (1990, 1997) hermeneutic phenomenology that is particularly useful, is his six guidelines to approaching hermeneutic phenomenology, which is supportive to a novice researcher in implementing the research process.

The lifeworld existential themes guided by van Manen, offers a lens through which to explore lived experience such as fathers' experiences of caring for their child with a CACCN. These existential themes allow this study to present the lived experience of fathers caring for a child with a CACCN and fosters a deep understanding of the nature and meaning of their everyday experience. Van Manen himself says that these are the "universal themes of life" (van Manen, 2014, p. 302). Chapter four provides a detailed explanation as to why hermeneutic phenomenology has been selected.

Data on fathers' experiences of caring for a child with a CACCN were collected by up to two individual semi structured interviews each and journal entries. Fathers were recruited through two routes; firstly, fathers of children who were receiving a homecare package from two homecare providers, and secondly from support organisations on specific CA.

Nine fathers of children with a CACCN took part in this study. This included fathers of two children with Down syndrome, two children with Williams syndrome, two children with Prader-Willi syndrome, one child with Turner syndrome, one child with Trisomy 13 (Patau syndrome), and one child with an unnamed abnormality (1.1mb missing on 14q.33 and slight loss on 22qxb22.33.12 and 12q22.1). Of the nine fathers that participated in the study; seven fathers completed two interviews while two fathers participated in one interview, resulting in a total of sixteen interviews. Two fathers opted to complete the journal entries. Eight fathers chose to partake in the interviews via the online platform Zoom and one father chose to be interviewed in his home.

1.5 Findings

Findings from this study are presented through van Manen's existential themes of lived body, lived relation, lived space, lived things and lived time. These themes are then presented through the totality they comprise. The experiences of fathers within the existential themes often fluctuate and intertwine with one another throughout different stages and elements of their lives. When all five of these existential themes are brought together a deep understanding of the father's experience emerges. This is what van Manen calls *balancing of the parts and the whole*. Through this process, meaning is not simply interpreted but embodied and evoked, allowing the essence of father's experiences to be felt as well as understood.

Findings from the existential theme of "lived body" reveal that when their child is diagnosed with a CA, it is a profoundly complex experience for fathers, encompassing both emotional turmoil and practical challenges. This theme traces their journey from initial concerns to the confirmation of a diagnosis, highlighting the unique experiences particularly in the context of a rare CA. Beyond the emotional weight, a central issue

that emerges is the delay in accessing timely and appropriate diagnostic and support services, barriers that significantly impact fathers' capacity to navigate their roles and advocate for their children within the Irish healthcare and disability support systems. Three core elements of the "lived body" show the critical role of how professionals communicate diagnostic information; the rarity of many CA and gaps in how these conditions are monitored and recorded; and the ongoing, often international struggle to access adequate disability services.

The theme of "lived relation" highlights the complex relational landscape fathers navigate when parenting a child with a CACCN. Fathers strive to balance their emotions and caregiving responsibilities between their child with a CACCN and their other children, seeking to be present and equitable in their roles. They also face the challenge of sustaining their relationship with their partner while managing the demands of caregiving, often needing to shift between being a supportive parent and a committed partner. Additionally, while grandparents can offer valuable support, fathers also recognise the importance of maintaining boundaries to protect the emotional and practical stability of the family unit.

The theme of "lived space" highlights both the emotional and physical space needed for fathers to care for their children with a CACCN. Fathers reflect on the transition from hospital to home, highlighting how national procedures particularly within hospital settings, discharge processes, and access to home care packages influence their sense of preparedness and support. Their concerns also extend into the future, with many expressing anxiety around long-term living arrangements for their child, including access to respite services and residential care planning. Additionally, fathers face complex decisions regarding their child's care and education, often having to navigate emotionally and logistically challenging choices about medical procedures and school placement.

The theme of "lived things" highlights the structural and financial pressures that shape fathers' experiences in caring for a child with a CACCN. Fathers reported significant out-of-pocket expenses related to medical equipment, therapies, and homecare supports

that placed substantial strain on family finances. In addition, their ability to engage fully in caregiving was often constrained by inflexible or limited parental and paternity leave policies, both in Ireland and internationally. These employment related challenges not only restricted fathers' participation in daily care but also tended to at times have the potential to reinforce traditional gender roles within the family. The findings underscore the need for more equitable leave policies and financial support that enable fathers to share caregiving responsibilities more fully and sustainably.

The theme of "lived time" reveals the enduring and cumulative impact of caregiving on fathers of children with CACCN. Fluctuations in their children's symptoms, particularly those affecting life expectancy carry a profound emotional toll. Fathers also face challenges with their children's increasing behavioural challenges, that fathers report at times are harder to manage than the medical complexities. These difficulties are compounded by a lack of accessible mental health support for their children. Moreover, caregiving is not confined to daytime hours as it is a continuous, 24-hour responsibility. Sleep deprivation is a common and deeply affecting feature of their experience, as night-time care routines further intensify the strain, particularly as many fathers reported that nighttime caregiving was a role they specifically undertake.

Fathers' responses to their roles in caring for a child with a CACCN involves continuous adaptations and adjustments. Yet, the love and joy their children bring to their families and the wider communities outweigh these challenges. Therefore, this study concludes that: Fathers' experiences of having a child with CCN, are constantly evolving as they are influenced by their child's specific CCN that changes over time, and are impacted by their past, present and anticipatory future experience.

1.6 Summary of chapters

A short summary of each chapter is presented to highlight the structure of the thesis.

Chapter one introduces this study by highlighting the importance of exploring father's experiences in caring for a CACCN. It outlines the research aims and objectives, briefly

describes the methodological approach, and provides an overview of the thesis structure.

Chapter two presents a detailed background and context to this study by presenting an overview of the phenomenon. This includes exploring the definition of a CA, discussing the prevalence, and explaining the typical diagnostic pathways. Chapter two then addresses the concept of CCN and highlights the significance within the context of a CA including the health and developmental complexities associated with a CACCN. Additionally, this chapter highlights the paucity of empirical evidence on father's experiences compared to mothers thus, giving context to this study.

Chapter three considers this phenomenon with relation to international literature. A scoping review of the literature to date on father's experiences of caring for a child with CACCN is presented. The aim of this chapter is to contextualise what is known and unknown about this experience and maps out the terms used throughout the literature describing children with a CACCN, as well as the different CA represented within the context of father's experiences. Three overarching themes and associated sub themes are explored. This chapter also highlights the lack of specific research on father's experiences of caring for a child with a CACCN.

Chapter four introduces the methodological framework that guides this study and the rationale for using a hermeneutic phenomenology guided by van Manen. An overview of the philosophical issues is presented in this chapter, with a description of the philosophical underpinnings of this study.

Chapter five provides the methods for carrying out this study. This includes identifying the aims and objectives of the study, the sample, the inclusion and exclusion criteria, as well as the methods used to collect and analyse the data. Ethical considerations are also discussed.

Chapter six presents the findings through the lens of the five existential themes body (corporeality), lived space (spatiality), lived time (temporality), lived human relation

(relationality), and lived things (Van Manen, 1990, 2016). Through this, five major themes and seventeen subthemes emerged which are presented in this chapter.

Chapter seven provides a critical discussion of the findings with reference to international literature. This chapter is structured around the main themes in the context of the five existential themes guided by van Manen (1990, 2016) and explains how the five existential themes make up the whole experience ensuring that the study aims and objectives were met.

Chapter eight is the final chapter in this thesis and presents a final summary of the results. The implications of this study that emerged from fathers' interviews and journals are discussed as well as presenting targeted recommendations.

1.7 Conclusion

This chapter provided a brief overview of this thesis by outlining the research aims and objectives, providing a brief overview of the methodological approach adopted, and concluded with a summary of each chapter highlighting the structure of the thesis. The next chapter presents a detailed background giving context to this study by presenting an overview of children with a CA and CCN as well as giving context to why fathers specifically have been selected.

2 Background

2.1 Introduction

This background chapter provides a contextual foundation for the research by offering a broad overview of the phenomenon under investigation. Furthermore, this chapter articulates the rationale for its exploration. This study explores the experiences of fathers' caring for a child with a CACCN. The chapter commences by presenting a definition of a CA, discussing the prevalence, and explaining the typical diagnostic pathways. The concept of CCN is addressed, acknowledging the ambiguous nature of the term and also examining its significance within the context of CAs. The chapter also outlines the range of complex health and developmental challenges experienced by children with a CACCN.

Finally, this chapter presents an overview of how disability services have changed in recent decades, highlighting the important roles families have in the care of children with a CACCN. More specifically, this chapter highlights the paucity of empirical evidence on fathers' experiences compared to mothers highlighting the need for fathers' experiences to be represented thus, giving context to this study.

2.2 Definition and incidence of a chromosome abnormality

In defining a CA, it is important to first understand what a chromosome is. A chromosome is a strand of DNA that is encoded with genes. Humans have 22 pairs of these chromosomes plus the two sex chromosomes (XX in females and XY in males) for a total of 46 (Queremel Milani & Tadi, 2023). Each chromosome contains thousands of genes, which make proteins that direct the body's development, growth, and chemical reactions (Genetic alliance, 2009; Padiath, 2025). The first 22 pairs of chromosomes are called autosomes. The 23rd pair consists of the sex chromosomes, X and Y. Half of these chromosomes are inherited from the mother and the other half from the father (Genetic alliance, 2009). CAs are the result of changes in either the number, or structure of a chromosome (Miles & Averil, 2015; Moorthie et al., 2018; Theisen & Shaffer, 2010).

There are an infinite number of CAs and over 100 reported chromosomal syndromes associated with a CA (Moorthie et al., 2018). The most common syndrome is Trisomy 21 (Down syndrome). According to EUROCAT, the prevalence of syndromic CAs, which includes all instances (live births, termination of pregnancy due to a congenital defect, and foetal deaths/stillbirths from 20 weeks gestation), was 45.28 per 10,000 births in 2019 (EUROCAT, 2022). While Census data (2022) indicates childhood disability in 45000 children, the breakdown of these figures is limited, and more detailed statistics are not available. Only one Irish study examined the rates of children with a CA focusing only on Trisomy chromosomal disorders (McDonnell et al., 2017). There were 394 cases from 80 894 total births, of which 289 were Trisomy 21, 75 were Trisomy 18 (Edwards syndrome), and 30 were Trisomy 13 (Patau's syndrome) (McDonnell et al., 2017). The total prevalence rate was 48.9/10 000 births. The specific birth prevalence was 35.7 for Trisomy 21, 9.3 for Trisomy 18 and 3.7 for Trisomy 13 per 10000 live births (McDonnell et al., 2017). The National Health Service (NHS) in England documented that the live birth prevalence for Down syndrome was one in every 854, Edward syndrome was one in every 13,591 and Patau syndrome was one in 23,001. This was relatively consistent with previous years for Down syndrome and Edwards syndrome but higher for Patau's syndrome relative to 2020 (NHS, 2024).

Although many CAs are considered rare, together they make up a significant contribution to human morbidity and mortality. Rare diseases have particularly low prevalence, with 70% starting in childhood (European Commission, 2025; Rodwell & Aymé, 2015). Currently there is no universal definition for a rare disease and the classification varies from nation to nation, which means point prevalence is the most appropriate indicator for rare diseases worldwide (Richter et al., 2015; Nguengang Wakap et al., 2020; Wainstock & Katz, 2023). The European Union, Australia and Canada define a disease, disorder or abnormality 'rare' when it affects less than 50 per 100,000 (one in 2,000 people) (Canadian Organization for Rare Disorders, 2025; European Commission, 2020; Rare Ireland, 2025). In the USA, the American Orphan Drug Act (1983) defined disorders as rare if they affect 86 per 100,000 of the population and this remains the current USA definition.

2.3 Aetiology of a chromosome abnormality

The exact cause of a CA is often unknown; however, a CA occurs when a cell divides in two. Sometimes this happens during the development of an egg or sperm cell and other times they happen after conception due to misdivision of cells as chromosomes are missed, inserted, or disrupted (Iqbal et al., 2025). Some changes in chromosome structures can be inherited, while others occur as random accidents during the formation of reproductive cells or in early foetal development. In other words, the chromosomes do not separate correctly, resulting in an incorrect number of chromosomes in the gametes. These variations can affect both the autosomes or the sex chromosomes (Moorthie et. al., 2018).

Although the cause of a CA is often unknown, risk factors have been identified. The birth prevalence of a variety of CAs increases with advancing maternal age (Table 1) (Cuckle & Benn, 2021; Frederiksen et al., 2024; Matorras et al., 2025; Yuning Zhu et al., 2016; Zhang X et al., 2020). Advancing maternal age is defined as childbirth at the age of 35 years or older (Afroz & Rajib, 2024); Zipori et al., 2020). Recently, an upward trend in the prevalence of CAs has been widely reported worldwide due to the increasing proportion of births to older mothers (Frederiksen et al., 2024; Irving et al., 2011; Loane et al., 2013). In Ireland in 2022, the average age of mothers giving birth was 32.2 and in 2022 5,137 births were to mothers over the age of 40 (Central statistics office, 2024). While in Canada, in 2023, the average age of mothers at the time of delivery was 31.7 years, an increase from 31.4 years in 2021 (Statistics Canada, 2025). In the US, in 1990 mothers younger than 30 accounted for 7 in 10 births (69.8%), while in 2023, they accounted for less than 1 in 2 (48.6%) (Driscoll et al., 2025).

Table 1: Increased maternal age and risk of a chromosome abnormality

Mothers Age	Risk of trisomy 21	Risk of other Chromosome Abnormalities/syndromes
20	1.1480	1.525
25	1.1340	1.475
30	1.193	1.384
35	1.353	1.178
40	1.185	1.162
45	1.35	1.18

Adapted from: American College of Obstetrics and Gynaecology (2016)

Paternal age is also linked to an increased risk of a CA. It is estimated that spermatogenic chromosomes are replicated 35 times by age 15, 150 times by age 20, 380 times by age 30, 610 times by age 40, and 840 times by age 50, increasing the risk of an abnormality occurring due to cell mutations such as deletions and insertions (Janeczko et al. 2020; Kong et al., 2012; Speicher et al., 2010). Similarly, to mothers, paternal age is also increasing worldwide adding to the increased rate of CAs. The average age of fathers in England and Wales increased in the last decade, with the most recent statistics in 2023 showing that they were on average almost a year older than their 2013 counterparts (32.8 to 33.8) (Office for National Statistics, 2024). The Irish Central Statistics Office figures indicate fathers of new babies in their 40s increased significantly in the past decade, with 12% of new fathers in 2007 to 17% in 2017 (Central Statistics Office, 2025).

In addition to maternal and paternal age, predisposed medical conditions, environmental exposures to both mothers and fathers such as smoking, drugs, alcohol and low economic income can increase the risk of having a child with a CA (Björkstедt et al., 2025; Kurita et al., 2020; Beal et al., 2017; Pereira et al., 2014). Ethnicity and family history can also influence the risk of a child having a CA. It is possible to inherit some types of CA, however, most chromosomal disorders are not passed from one generation to the next (National Human Genome Research Institute, 2020; Simpson, 2024).

2.4 Classification of a chromosome abnormality

There are approximately 19,900 genes used to produce proteins that are contained on the 46 chromosomes in each cell of the human body (National Library of Medicine, 2024). Small mutations can occur to specific genes within the 46 chromosomes which can cause few or mild problems to human development, while other mutations cause serious medical disorders such as Sickle Cell Anaemia, Cystic Fibrosis, and Muscular Dystrophy. However, a CA occurs from either a loss or a gain of entire chromosomes (aneuploidy), or parts of chromosomes (structural abnormalities) not the genes within them (Moorthie et al., 2018; Theisen & Shaffer, 2010). CAs can be classified as either numerical or structural (Iqbal et al., 2025).

2.4.1 Numerical abnormalities

Numerical abnormalities occur when there is a change in the number of chromosomes. They are either missing or have extra to the normal pair (more or fewer than 46) (National Human Genome Research Institute, 2020). Examples of numerical disorders include trisomy, monosomy and triploidy.

1. A trisomy CA happens when a critical region of a chromosome appears three times (trisomy) rather than twice. Sometimes, the CA may be present in only a percentage of cells (mosaicism) (McDonnell et al., 2017; Rohrlach et al., 2024). Examples of a Trisomy CA is Trisomy 21 (Down syndrome), Trisomy 18 (Edwards syndrome) and Trisomy 13 (Patau syndrome) (Rohrlach et al., 2024). Sex chromosome trisomy's are an additional X or Y chromosome, and examples of these are 47,XXY/Klinefelter syndrome, 47,XYY/Double Y syndrome or 47,XXX/Trisomy X syndrome (Hall, 2025).
2. Monosomy is a form of aneuploidy with the presence of only one chromosome from a pair. Partial monosomy occurs when a portion of one chromosome in a pair is missing (National Human Genome Research Institute, 2020). An example of this is Turner syndrome which is associated with partial or complete absence of the X chromosome (Sharma & Shankar Kikkeri, 2025).
3. Triploidy is an extremely rare CA and usually neonates with triploidy are miscarried early in the pregnancy. This is the presence of an additional set of chromosomes in the cell for a total of 69 chromosomes rather than the normal 46 chromosomes per cell (NORD, 2024; Toufaily et al., 2016; Walsh et al., 2022).

2.4.2 Structural abnormalities

Structural abnormalities occur when part of an individual chromosome is either missing, extra, switched to another chromosome, or turned upside down i.e. the genomic rearrangement of one or more chromosome (Genetic Alliance, 2009; Queremel & Tadi, 2023). Structural rearrangements are defined as balanced if the complete chromosomal set is still present, though rearranged, and unbalanced if information is additional or

missing. An example of a structural CA is Williams Syndrome, which is a deletion of a region on chromosome 7 (Wilson et al., 2025).

2.5 Diagnosing a chromosome abnormality

The diagnosis of a CA has had significant advances in recent years, with diagnostics technologies like next-generation sequencing and whole exome sequencing, chromosomal microarray analysis, and non-invasive prenatal testing (Armengol et al., 2012; Berman et al., 2012; Samura, 2020; Van den Veyver, 2016). These diagnostic tools have allowed for increased high-resolution detection of CAs with greater accuracy and speed than traditional methods. This diagnosis will either occur before birth or during pregnancy which is known as a 'pre-natal diagnosis' or after birth which is known as a 'post-natal diagnosis'.

2.5.1 Prenatal diagnosis of a chromosome abnormality

Diagnosing a foetus with a CA is one of the most important aspects of modern perinatology (Neagos et al., 2011; Van den Veyver, 2016). Chromosomal analysis of the foetus is a definite way to diagnose a foetus with a CA (Liehr et al., 2017; Santoli, 2025; Xia et al., 2020). Prenatal diagnosis of CAs has increased over the past few decades, with increased reporting and prenatal screening practices being the gold standard in most countries (Kagan et al., 2020; Xie et al. 2021). Approaches of prenatal diagnosis of CAs can be divided into two categories, non-invasive and invasive techniques. Screening tests are usually non-invasive which includes blood tests and ultrasounds (HSE, 2023). Invasive screening on the other hand involves, 'Chorionic Villus Sampling' (placing a needle into the uterus and placenta) or amniocentesis (needle placed into the uterus and amniotic fluid, which increased the risk of miscarriage), but is usually more precise in identifying CAs but can sometimes increase the risk of miscarriage (HSE, 2025).

2.5.2 Non-invasive techniques

The nuchal translucency scan is a method that estimates the risk of a CA and is carried out from 12.5 to 13.5 weeks. The likelihood of a CA is identified through factors such as, the amount of fluid behind the neck of the foetus, presence or absence of the foetal nasal bone and the presence or absence of any physical abnormalities. Although this

method is not a diagnosis it can help identify foetuses that are at risk of having a CA (HSE, 2023).

Non-Invasive Prenatal Screening (NIPS) screens for CA and is based on the analysis of cell-free foetal DNA in maternal blood (Goldwaser & Klugman, 2018; Sotiriadis et al., 2017). This isolates cell-free foetal DNA in maternal blood to detect a CA (Carlson, 2017). NIPS is the most recent development in prenatal screening (Hixson et al., 2015). NIPS was identified in the maternal plasma in 1997 (Lo et al., 1997), however, has only been clinically available for prenatal aneuploidy screening since 2011 (Sotiriadis et al., 2017). Data now shows that it is commonly used by obstetric providers in the diagnosis of CAs (Allyse et al., 2015). This is used to avoid the small risk of procedure-related miscarriage (1%) (Wulff et al., 2016). NIPs can be utilised to not only to identify or exclude a risk for genetic diseases in general, but also specifically, can address CAs in the following unbalanced chromosomal alterations: Trisomy 13, 18, 21, and X as well as other chromosome aberrations (Liehr, 2015). Furthermore, NIPS has a high detection rate with extremely low false positive rates (0.5%) (Abel et al., 2020). However, what cannot be addressed are, for example, triploidy, balanced chromosomal aberrations, and unbalanced rearrangements involving other genetic regions than the ones of the tested chromosomes (Liehr, 2017).

In Europe, 14 countries have implemented NIPS into a national screening policy/programme. Two countries (Belgium and the Netherlands) offer NIPS for all pregnant women, whereas most other European countries have implemented NIPS as an offer for higher risk women after first trimester screening (Gadsbøll et al., 2020). In the Republic of Ireland, NIPT tests are suggested if the foetus is deemed 'at risk', however, this is within the private sector as a voluntary opt-in service (Gadsbøll et al., 2020; HSE, 2025). The American College of Obstetrics and Gynecology (ACOG) released a statement indicating that all women, including those at low risk, should be offered NIPT as a screening option. Non-invasive a screening test, not a diagnostic one and an invasive test is needed for an accurate diagnosis.

2.5.3 Invasive prenatal techniques

Invasive procedures for prenatal diagnoses of CAs such as amniocentesis and CVS were created as diagnostic options for high-risk mothers; for example, mothers with advanced maternal age or a family history of genetic disorders (Beta et al., 2019; Carlson et al., 2017). During the 1980s chorionic villi began being sampled to perform foetal karyotyping to diagnose CAs prenatally. This shifted the prenatal diagnosis from the second trimester (using ultrasound) to the first trimester (Holzgreve et al., 1984; Carbone et al., 2021). Pregnant women who were deemed to be at high risk for foetal aneuploidy and those with major foetal defects diagnosed on ultrasound or NIPS are typically offered the option of invasive prenatal diagnosis (Beta et al., 2019; HSE, 2023).

CVS is a prenatal test in which a sample of chorionic villi is removed from the placenta for testing. This sample can be taken through the cervix (transcervical) or the abdominal wall (transabdominal). Chorionic villi sprout from the chorion to provide maximal contact area with maternal blood. CVS is offered as the procedure of choice up to 15 weeks' gestation (Beta et al., 2019) and is estimated to give a definitive result in 99 percent of all samples (NHS, 2023).

Amniocentesis is a procedure in which amniotic fluid is removed from the uterus for testing of CAs. Every cell in the amniotic fluid contains a complete set of the foetus DNA (HSE, 2023). Amniocentesis is offered after this gestational age of 15 weeks (Beta et al., 2019) with accuracy estimated at 98-99% of all tests. The risk of miscarriage is slightly lower than that of CVS and is estimated at 1% (UK National Health Service Fetal Anomaly Screening Programme, 2023). Since the commencement of NIPS, fewer invasive tests are performed worldwide (Gill & Prosen, 2017).

The aim of prenatal CA testing is to deliver information about the wellbeing of the foetus to prospective parents and their healthcare providers. This can potentially enable parents to make an informed choice regarding pregnancy terminations or allow parents warning of the potential complex needs associated with the CA (Carlson & Vora, 2017; Richardson & Ormond, 2018; Werner et al., 2016). Furthermore, a prenatal diagnosis can allow healthcare workers plan the delivery of the baby, especially where specialist

interventions and supports are required such as palliative care needs (Congenital Anomaly Statistics, 2021). Pre-natal testing affords parents the opportunity to make informed decisions about the management and treatment of their children. This means, they can facilitate both early diagnosis and early abortion of babies identified as having a CA (Grossman et al., 2020).

2.5.4 Post natal diagnosis of a chromosome abnormality

Genetic testing is required or necessary to be performed in a multitude of circumstances, often during the early postnatal period, but can be prolonged into early childhood (Berisha et al., 2020). Some CA remain undiagnosed for prolonged periods of time, examples of this is Klinefelter syndrome, despite being the most common chromosomal aberration in men, 75% will remain undiagnosed throughout their lifetime and only 10% will be diagnosed before puberty (Akcan et al., 2018). Medical professionals will use genetic testing when the baby or child are portraying signs of non-typical features and development which include but are not limited to, early growth and development such as developmental delays, Autism, failure to thrive, intellectual disabilities, dysmorphic features, multiple malformations, heart defects, short stature, and ambiguous genitalia (Moore et al., 2022; Berisha et al., 2020). When it is suspected that a neonate has an abnormality or medical condition, genetic diagnostic tests such as cytogenetic and molecular genetic techniques are applied in clinical genetics laboratories to detect CAs. Recent studies show it is now possible to obtain whole genome test results to confirm a CA in a median time of about 20 hours by using computer systems able to perform automated phenotyping and data interpretation, tasks that normally require human intelligence (Bell, 2018; Clark et al., 2019).

2.6 Children's complex care needs associated with a chromosome abnormality

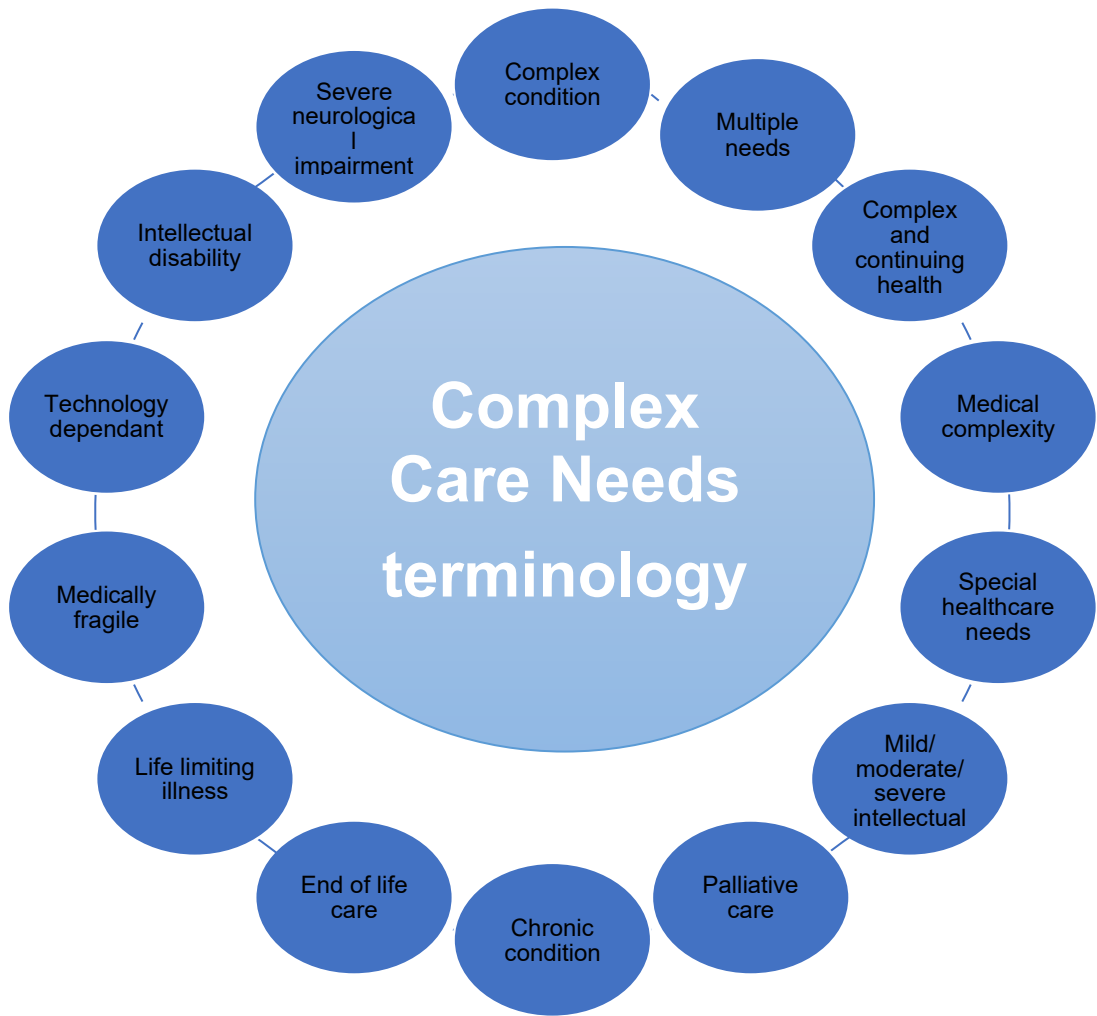
CAs can cause multi-domain disabilities that can include a combination of physical disabilities, health problems, intellectual disabilities, psychiatric/mental health disorders and behavioural difficulties (Ajitkumar et al., 2020; Anzick et al., 2020; Boczek et al., 2018; Fermin Gutierrez et al., 2020; Kawai et al., 2024; Lizarazo et al., 2019;

National Human Genome Research Institute, 2016; Schwartz et al., 2021; Wilson et al., 2025). The combination of these disabilities can result in children having CCN.

Children with CCN have increased chronic physical, developmental, behavioural or emotional illnesses and disabilities, who also require increased health and social care related services across multiple settings (Brenner et al., 2019; Cassidy et al., 2023). These increased services involve both treatment and monitoring of medical conditions, as well as caring for their psychosocial and psychological development through routine screening and testing as well as the development and implementation of continuously changing care plans (Brenner et al., 2015; Brenner et al., 2018; Luzi et al., 2019). One shared feature among children with CCN is the need for additional health care services compared with the general child population, however there is considerable variation in the depth and breadth of their needs and each child will have individual and varying person-centred specific needs with a need for multidimensional care (Brenner et al., 2016; Brenner et al., 2018; Cassidy et al., 2023; Dewan & Cohen, 2013).

It is clear within the literature, there is a significant range of terminology that is used to refer to children with CCN as displayed in figure 1. Brenner et al. (2018, p 1641–1652) completed a concept analysis and found that children's CCN 'refer to multidimensional health and social care needs in the presence of a recognized medical condition or where there is no unifying diagnosis. They are individual and contextualized, are continuing and dynamic, and are present across a range of settings, impacted by healthcare structure'. For clarity and continuity, the operational term this study uses to describe these children is 'Complex Care Needs' (CCN). This has been adopted as this definition is the first to systematically analyse and define CCN.

Figure 1: Terms associated with complex care needs



Children with CCN are an ever-growing population; although these children represent a small proportion of the child population, they have a significant impact on the delivery of integrated health and social care services (Cassidy et al., 2023; Cohen et al., 2023; Curran et al., 2020; Dunbar et al., 2019; Gay et al., 2016; Kieran et al., 2019; Mackay et al., 2020 2025; Russell et al., 2014). There is an increasing number of children living with CCN with improved quality of life due to the increased survival rates of children with

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often rare and complicated medical conditions (Cohen & Patel, 2014; Fraser et al., 2021). The increase in children living with CCN is due to the advances in technology, medicine and health care that provide lifesaving equipment, prolonged hospital admissions, medicines and services that increase life expectancy (Aitchison et al., 2021; Feinstein et al., 2019; Mack et al., 2024).

In Ireland, the exact number of children living with CCN is unknown, however it is estimated that there are approximately 4000 children with life-limiting conditions of which whom many will have CCN, yet statistics for this are based on data from 2005 which the national children's hospice highlight is significantly outdated (Laura Lynn Children's Hospice, 2019; Ling et al., 2015). In England, one study showed there are 121,626 young people aged 16 years or younger living with CCN or life limiting condition (Javi et al., 2022). Furthermore, from the year 2012/13 to 2018/19, the number of young people aged 14-19 years with a life-limiting condition diagnosed at age 16 years or younger increased from 20,363 to 34,307 (Javi et al., 2022). In Canada, 97,600 children and youth have been identified as living with medical complexity (Canadian Institute for Health Information, 2020). Unfortunately, the exact number of children worldwide living with CCN is unknown due to the lack of available data and specific complex needs databases coupled with the large variation in terminology used (Pinney, 2017). However, in the Republic of Ireland (ROI), there are 131,764 children with a disability, accounting for 10.8% of the child population (Department of Children, Disability and Equality, 2025).

The range of CCN associated with a CA can include reduced intellectual and impaired neurological functioning, impaired GI functioning, limitations to breathing and circulatory systems, altered physical ability, reduced life expectancy and increased mortality rate.

2.6.1 Intellectual functioning

There are similar symptoms associated with most CAs, that occur to varying degrees in many affected children. Intellectual disability often occurs in children with a CA, due to the genes located across all of these chromosomes that provide instructions for normal

development and function of the brain (Foster et al. 2014; Ilyas, 2020; Michelson et al., 2011, Wiltrout & Mefford, 2026). The incidence of a person having an intellectual disability due to a CA varies from 17% to 50% in this population (Batshaw et al., 2013; Deciphering Developmental Disorders, 2017; Moeschler & Shevell, 2006; Xu et al., 2024).

An intellectual disability is defined as having “significant limitations in both intellectual functioning and in adaptive behaviour, which covers many everyday social and practical skills”. This disability originates and is diagnosed before the age of 18 (American Association of Intellectual Disabilities, 2021 p. 33). Similarly, The WHO defines an intellectual disability as “a person who has significant reduced ability to understand new or complex information; this results in a reduced ability to cope independently due to impaired social and intellectual functioning, which begins before adulthood and has a lasting effect on development” (WHO, 2019). Despite varying definitions, the WHO’s definition is currently the internationally recognised definition. Limitations associated with an intellectual disability are presented in Table 2.

Table 2: Challenges associated with an intellectual disability

Limitations associated with an intellectual disability
1. Communication
2. Personal care
3. Home life
4. Social skills
5. Utilization of the community
6. Self-governance
7. Health and safety
8. Functional academic skills
9. Leisure time

10. Work

From American Association on Intellectual and Developmental Disabilities (AAIDD, 2013)

Intellectual disabilities include a broad spectrum of intellectual functioning which requires vast person-centred support throughout a person's lifetime. The most recent census data shows that 2% of the population in the ROI have an intellectual disability, making it the most common disability in the country (Census, 2023). As of 2024, approximately 349,000 children in the UK are reported to have an intellectual disability, which equates to about 2.5% of the child population (Mencap, 2024).

Intellectual disabilities classified as mild, moderate, severe and profound (Katz & Lazcano-Ponce, 2008; American Psychiatric Association, 2013). The severity of intellectual disability varies, depending on the different CA. For example, children with Klinefelter syndrome can have very mild intellectual disabilities (Blackburn et al., 2025). Yet, children with Trisomy 18 or 13 will typically have severe to profound intellectual disabilities. It is estimated that CAs account for approximately 20-25% of all children with a severe intellectual disability (Foster et al., 2015). Even within specific CAs there are variations, for example, children with Trisomy 21 can present with the entire range from mild to moderate intellectual disability with IQ ranging from 40- 70 (NHS, SCS Disability Pathway 2025).

Children living with a CACCN may face a range of significant health challenges that can impact their physical, emotional, and social well-being. These challenges are explored in further detail in the following section.

2.6.2 Neurodevelopmental and mental health

CAs are associated with an increased risk of seizure activity and epilepsy (Akasaka et al., 2020; Altuna et al., 2021; Izumi et al., 2014; Jaspersen et al., 2023; Kobow et al., 2020; Montenegro et al., 2025; Pascual-Morena et al., 2024). Children with some CAs are at an increased risk of Autism (Akshoomoff et al., 2015; Bettencourt et al., 2024; Carvalho & Mazza, 2020; Spinazzi et al., 2023; Wiśniowiecka-Kowalnik et al., 2019). Additionally, some CAs increase the risk of psychiatric co-morbidities such as schizophrenia, and

anxiety (Aman et al., 2025; Royston et al., 2021; Stephenson et al., 2014; Srinivasan et al., 2019).

2.6.3 Feeding and toileting

Children with CAs have an increased risk of developing feeding problems and swallowing dysfunction because of the complex interactions between anatomical, medical, physiological, and behavioural factors (Anil et al., 2019; Cooper-Brown et al., 2008; Daley et al., 2025; Fish et al., 2025; Rochedy et al., 2023; Roy et al., 2025). Challenges with oral-motor function may develop as a result of both functional problems, e.g., cleft lip/palate, hypotonia, dysphasia, (Anil et al., 2019; Cooper-Brown et al., 2008; Daley et al., 2024; Jackson et al., 2019; Klingel et al., 2017; Shenoy et al., 2018). A lack of opportunity to develop oral motor skills due to environmental factors such as not being given the opportunity to breast feed, surgeries, prolonged hospital admissions, and reduce the ability of children with CAs to develop oral motor skills (Cooper-Brown et al., 2008; Swillen & McDonald-McGinn, 2015).

These feeding difficulties result in children with CAs often relying on external feeding devices such as Percutaneous Endoscopic Gastrostomy (PEG), Percutaneous Endoscopic Gastric Jejunostomy (PEGJ), Nasogastric Feeding (NG) and Total Parental Nutrition TPN (Aviles et al., 2024; Boechler et al., 2024; Mannava et al., 2025; Poskanzer et al., 2020; Roy et al., 2024; Smith & Landman, 2019; Stanley et al., 2019). Additionally, CAs increase the risk of having disturbances in gastric functioning and are at an increased risk of developing gastroesophageal reflux disease (GERD) (Bermudez et al., 2019; Boechler et al., 2024; Bedeschi et al., 2025).

Associated systemic and localised comorbidities, intellectual functioning, interception and psychological challenges can result in children with a CA having bladder and bowel disorders, often requiring support with their elimination needs (Choi et al., 2019; Dreher et al., 2022; Kotcher et al., 2022; Stogren et al., 2025; Santucci et al., 2025; von Gontard et al., 2016; Phelan et al., 2022). Constipation has been reported to be a particular problem for those with many CAs (Boechler et al., 2024; Hussong et al., 2020; Ramsey &

Nichols, 2025; Giardino et al., 2014). Children with Trisomy 21 are at an increased risk of coeliac disease (Lattuada et al., 2025). Additionally, there is a higher incidence of lower urinary tract infections in children with CAs (Barnes & Carey, 2018; Chao et al., 2021; de Carvalho Mrad et al., 2014; Scheuerle et al., 2025).

2.6.4 Breathing, circulation and sleep

Lung development is a complex process, CAs can cause severe lung malformations that can result in conditions such as; hyperventilation syndrome, increased risk of sleep disordered breathing, apnoea, tracheal rings, tracheal stenosis and shortening (Amata, 2019; Cereda & Carey, 2012; Debs et al., 2025; Kettler et al., 2020; Kylat, 2019; Presti et al., 2025; Thamkittikun et al., 2025). Aside from distorted breathing, challenges with sleep have also been seen within the literature of children with a CA, in relation to insomnia (Bruni et al., 2025; Duis et al., 2022; Fish et al., 2025; Revana et al., 2024).

As a result of breathing challenges many children with CAs require oxygen therapy, non-invasive and invasive ventilation and suctioning. Furthermore, there have been an increasing number of children with a CA surviving with high medical needs, for whom tracheostomy and/or home ventilation is part of their chronic disease management (Hudson et al., 2022; Taira et al., 2020; Shibuya et al., 2018; Suco et al., 2025; Chindris et al., 2020; Ignatz-Hoover et al., 2025; Fockens et al., 2021; Smith et al., 2021). Additionally, children with CAs are at an increased susceptibility to repeated infections of the lungs due to physical abnormalities, weaker immune systems and repeated aspiration (Imataka et al., 2016; Ghezzi et al., 2024; Com et al., 2021; Wong et al., 2020).

CAs are commonly associated with heart defects and congenital heart disease (Abduljawad et al., 2020; Alba et al., 2024; Calcagni et al., 2017; Duarte & Singh, 2024; Green et al., 2024; Pfitzer et al., 2018). Congenital heart disease is a common cause of neonatal morbidity and mortality (Hopkins, 2019). Of all the global congenital heart disease deaths in 2017 (261,247 deaths) 180,624 occurred in infants younger than 1 year (Congenital Heart Disease Collaborators, 2020). In the ROI, 1 in every 100 children are born with a congenital heart condition (Heart Children Ireland, 2019). The estimated

occurrence of congenital heart defects is 2.4 million people in the United States of America (USA) with 1 million being children (Gilboa et al., 2016). The rate of CAs accounting for congenital heart defects is significant and ranges from 8-12% of all live births worldwide (Cowan, 2015; Goldmuntz et al., 2011; Odilova & Tilavova, 2025). These statistics can vary as in some countries, fetuses with a CA or heart defect are aborted for example, in the Czech Republic, of 2066 fetuses diagnosed in the second trimester with a congenital heart defect, 880 (42.6%) were terminated (Tomek et al., 2023).

One-quarter of all congenital heart defects are severe and will require treatment in order to survive (Hartman et al., 2011; Leirgul, 2014; Oster, 2013). Developments in cardiac surgery have significantly improved in terms of immediate and long-term outcomes for babies and children with a congenital heart disease associated with a CA (Alam et al., 2021; Çelik et al., 2023; Fuchs et al., 2020; Green et al., 2024; Kavarana, 2021; Peterson et al., 2021). A palliative care approach was the traditional guideline for infants with conditions such as Trisomy 18, however more clinicians are willing to discuss cardiac surgery options despite resulting in a child with such Trisomies living with subsequent CCN (Jacobs et al., 2016; Kaulfus et al., 2019; Kawasaki et al., 2021; Neubauer et al., 2020; Palloto et al., 2017; Staso et al., 2018).

2.6.5 Physical ability

A variety of physical disabilities can be associated with a CA. For example, hypotonia is a symptom of diminished tone of skeletal muscle associated with decreased resistance of muscles to passive stretching, which can be caused by abnormalities of the central nervous system, any element of the lower motoneuron, or both. This is associated with over 500 different genetic disorders many of which are CAs (Buddhdev et al., 2020; Chaudhry et al., 2023; Hidalgo Robles et al., 2024; Lisi & Cohn, 2011; Metzler & Brown, 2025; Tuysuz et al., 2014; Yang, 2024). Musculoskeletal anomalies are also common (Deepshikha et al., 2025; Balasundaram & Avulakunta, 2025; Verscaj et al., 2025) that can increase a child's support needs.

Cerebral palsy is a disorder of movement and posture due to nonprogressive disturbances that occur in the developing foetal and infant brain that is an umbrella term, primarily affecting posture and mobility, and causes variations in muscle tone, stiff muscles, spasticity, rigidity, ataxia, tremors or involuntary movements (Alpay Savasan et al., 2021; Blair & Cans, 2025). Some CA are associated with cerebral palsy (Michael-Asalu; Chen et al., 2012; Han et al., 2025). Globally, approximately 17 million people have cerebral palsy, (1 in every 500 neonates) of these up to 10% of cerebral palsy cases are the result of CA (Bartels et al., 2020; Cerebral Palsy Alliance, 2020; Menkes & Flores-Sarnat, 2006).

2.6.6 Life expectancy and mortality rates

Modern medicine and increased medical technologies have increased the life expectancies of many children with a CA, however, at times this requires the children to be technology dependent for survival (Armour et al., 2025; Hamill et al., 2021; Hudson et al., 2021; Ikeda et al., 2018; Schlosser et al., 2024). Children who are technology dependent need technology support for basic biological functioning (Brenner et al., 2020). However, despite these medical advances, some CAs still result in children having increased mortality rates (Aleksander et al., 2025; Bastos et al., 2025; Patterson et al., 2017; Schlosser et al. 2024; Tantaleán et al., 2025; Wojcik et al., 2019).

The increased risk of childhood death varies depending on the child's diagnosis. The National Paediatric Mortality Registers Annual Report (2025) in Ireland (between January 2019- December 2023), highlighted that the second most common cause of death in the neonatal period were CAs and congenital anomalies, which accounted for 38.3% of deaths (n=259), and 37% of post neonatal deaths (n=81) (National Office of Clinical Audit, 2025). In Canada, CAs and congenital malformations were the third leading cause of death in children between 0-4 years of age for the last four years (Statistics Canada, 2023). In Boston's Children's Hospital, a review examined 573 infant deaths (who died within the first year of life) 54% of which died with a CA (Wojcik et al., 2019).

In the United States of America (USA), CAs were the second leading cause of death in infants aged 1 to 4 years old. However, as the children's age category increased, the mortality due to a CA decreased. In school aged children, (5-9 years old) CAs became the third leading cause of death (Cunningham et al., 2018). Furthermore, for adolescents (10-19 years) CAs did not appear in the six-leading causes of death (Cunningham et al., 2018). This shows that if a child with a CA is going to die in childhood, this is typically within infancy or early childhood. However, the type of CA significantly influences the mortality rate and age of death in these children. For example, approximately 50% of infants with Trisomy 13 and 18 die during the first weeks of life, with the reported average survival for children with Trisomy 13 and Trisomy 18 being 5 days and 8 days respectively. The majority (74.5% and 62.8% respectively) die within the first month of life (Meyer et al., 2016; Milligan et al., 2020; Nelson et al., 2016). However, in comparison, children with Down syndrome can expect to live more than 60 years (Centres for disease control and prevention, 2024), with most countries have a decline in Trisomy 21 deaths among children and adolescents over the last 42 years (Ye et al., 2025).

2.7 Home environments for children with a chromosome abnormality and complex care needs

Care systems have made significant advances in recent decades, with the majority of children with a CACCN now living at home. This is a result of the deinstitutionalisation of care for people with disabilities followed the requirements of the United Nations Convention on the Rights of Persons with Disabilities (UN CPRD, 2006) and The United Nations Convention on the Rights of the Child (UN, 1989). European countries have implemented measures reinforcing the transition from institutional and formal public spaces to informal community-based services such as the home. This is at various stages worldwide and the ROI has not yet fully achieved this, however, now almost all children with disabilities live within the family home (Census, 2023). According to the most recent Census, over one fifth (21.3%) of individuals with a disability in the ROI are children living in private households.

Many children with a CCN require multifaceted home care management plans to address their medical, social and emotional care needs, in which parents take a large amount of responsibility when they are discharged from hospital (Page et al., 2020; LeGrow et al., 2022; Haspels et al., 2025). Parents undertake a caregiver role, completing tasks previously provided by nurses in hospitals or residential services, additional roles than parents of typically developing children (Woodgate et al., 2015; Currie & Szabo, 2019; Imperial-Perez & Heilemann, 2019; Page et al., 2020; Selzer et al., 2024).

These responsibilities include performing medical procedures, administering medications, managing emergencies (sometimes life-threatening), coordinating care and advocating for their child (Imperial-Perez & Heilemann, 2019; Page et al., 2020). Parents report feeling that they become nurses, therapists and caregivers to their children with CACCN (Currie & Szabo, 2019). Many of these parents report being the 'experts' in their child's condition albeit often without any medical background, some parents go as far as to say they 'educate' or 'train' nursing staff (Brenner et al., 2015; Callans et al., 2016; Doyle, 2022).

Many of these parent's report being afraid, anxious and concerned that a life-threatening situation may occur in their home. Often finding they have 'direct care overload' and 'psychological stress' (Conden, 2020; Gong et al., 2019; McDonald et al., 2017; Ronan et al., 2020). Specifically, many of these parents report high levels of distress and burden, low quality-of-life and report the need for more emotional and practical support (Collins et al., 2020). The transition of children with CCN from hospital to home is a challenging dynamic that is critically in need of further improvement and greater negotiation between the parent and health service providers (Brenner et al., 2015; Ronan et al., 2020). Many parents report that the hospital discharge process is complicated and even frustrating at times. They are often exhausted and stressed with the lack of support that they feel is available to them from both the hospital and community services (Callans et al., 2016; de Souza Esteves et al., 2015; Ronan et al., 2020).

Paediatric home health care is an essential bridge that supports the transition from hospital to home to allow for children with CCN to live outside the hospital safely (Boss et al., 2020; Fratantoni et al., 2019). The success of the home care process is built on knowledge, anticipation, early identification of needs, communication, care-coordination infrastructure, skilled home health providers, and the parent-provider relationship (Fratantoni et al., 2019). It has been well documented that families of children with CCN appreciate and significantly benefit from home health care that is provided in an integrated and coordinated manner, when the child is being treated in their home (American Academy of Pediatrics: Council on Children with Disabilities and Medical Home Implementation Project Advisory Committee, 2014). A comprehensive process is required to identify individual children's needs, develop options of care, create detailed care plans and to navigate these children's ever evolving needs (Fratantoni et al., 2019).

Many families of children with CCN rely on care coordinators to navigate the system, and report the need for improved coordination to facilitate transitions in care from hospital to home (Curren et al., 2020; Ware et al., 2020). The current approach to the management and integration of care of children with CCN in 30 countries across Europe, show that few systems are in place to identify all the health-care providers who deliver care to a child living with CCN, and that less than half of all countries (included in the study) have policies in place to support care coordination for these children (Brenner et al., 2018).

Community support to families is provided by a wide range of health, education and social care professionals, including community children's nurses, often through a home care package process (Whiting et al., 2019). Some children with CCN receive home nursing as part of home care services, either intermittently (nursing visits) or continuously (private and state funded) by a registered nurse at the child's home (Carter, 2012; Nageswaran & Golden 2017)). Families have reported the clear benefits of home care packages (HCP), but some challenges have been identified. For example, families have reported feeling that the nurse has poor training and is not always of high quality

and family routines being dictated by rostering schedules. Many parents discuss the lack of family privacy, particularly with shift nursing, feeling never truly alone in their home (Boss et al., 2020; Nageswaran & Golden, 2017; Nageswaran et al., 2017, 18).

In the Republic of Ireland (ROI), the HSE is dedicated to the on-going provision of paediatric HCP to support the discharge of children with CCN from acute hospital services into the care of their families in the community. In 2018, there were a total of 457 nursing HCP for children with CCN in the ROI, with a budget of 28.7 million euro, an increase of 6 million euro from 2018 (Office of the Head of Operations Primary Care, Community Operations, 2019; National budget, 2019). Home care packages can only be provided by approved home care providers in conjunction with the HSE, to ensure robust clinical governance is in place (HSE, 2013, HSE, 2021). However, despite the need for home care nurses for these children, there is a critical shortage of home nurses with the skills that are required to care for these children (Benscoter et al., 2019; Maynard, 2019; Sobotka et al., 2019).

2.8 The emerging importance of recognising fathers in paediatric complex care arena

Improvements in survival rates and quality of life among children with CACCN have also been accompanied by a notable shift in caregiving dynamics. Fathers are assuming increasingly more active roles in their children's care, due to significant reforms in social and political movements, which reject the traditional gendered roles within the family (Taylor & Scott, 2018). Men's interactions with their children were minimal until the 1960s, with their primary responsibility being the family provider (Adema & Clarke, 2017; Lomazzi, 2017). In the second half of the 20th century, there were a variety of significant political changes that brought about a change in the social and economic dynamics of the family unit (Carrillo Ripoll & Schvaneveldt, 2012). A recent survey named 'State of the World's Fathers' highlighted that today the majority of men do not agree that care giving is solely a women's responsibility (Equimundo Center for Masculinities and Social Justice, 2023). In the ROI, both men and women see caring for their children as being one of the most important and valued features of their lives (Men's Development Network, 2023).

One of the major substantial reforms relates to the labour workforce, in which there is an increase in female participation, and traditional gender roles have been re-examined (Fanelli Profeta, 2019). Historically, male roles within the family were centred around being the breadwinners, yet more women than ever before are taking on these roles. Although no economy has yet achieved full gender parity, there has been significant progress in areas of economic participation and opportunity as well as political empowerment (World Economic Forum, 2025). For example, in the ROI, a marriage bar in which a requirement that women working in certain jobs must leave that job when they marry was in place until the 1970's (Mosca & White, 2020). Now, women account for 41.9% of people in full-time employment, which has more than doubled since 1998 (Central Statistics Office, 2025). The U.S. Census Bureau Current Population Survey Annual Social and Economic Supplement data reported that in 2023, 45% of mothers were the main family earner.

Therefore, there is now increased societal agreement that fathers, just as much as mothers, should have a caring and nurturing relationship with their children (Cabrera & Tamis, 2013; Lamb, 2010; Planalp & Braungart-Rieker, 2016). There are substantial benefits for both the father and their children when fathers are actively involved in their care. Men's physical, mental and social health are directly and significantly impacted by fatherhood over the course of their lives (Kotelchuck, 2022). Research consistently demonstrates that fathers' involvement has a significant positive impact on children's overall well-being, social development, and behaviour (Choi et al., 2021; Zheng et al., 2025). Importantly, these positive influences are not limited to childhood but can extend into adulthood improving long term adjustment (Choi et al., 2021). Despite the international consensus that fathers are exceptionally important in caring for their children, research shows there are some persistent organisational and social barriers to them engaging in equal parental participation (Brooks & Hodgkinson 2020, Watkins et al., 2024). Sear (2021) highlights how the failure to recognise this change in the role of fathers in society can ultimately have adverse health effects on children. The creation of a father inclusive perspective should consider gender relations, the variety of family contexts and structures, best practices and relevant evidence, and the requirement for

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fathers and other carers to be explicitly included in policy (Leitão, 2022). Yet fathers are significantly underrepresented in parenting research in comparison to mothers (Bogossian et al., 2019; Cabrera et al., 2018; Parent et al., 2017). Understanding parental experiences in caring for children with CCN is vital in developing holistic, evidence-based healthcare related services.

As children with CACCN are living longer, in turn increasing the need for expanded parental responsibilities of children in their care, this means that fathers are playing a more vital role than ever before in providing this care. It is essential to study fathers' experiences, needs, and barriers in caring for children with a CACCN as research in this field can identify challenges, and legitimise fathers' roles as equal partners in the caregiving responsibilities. Like other areas of child research, within the CCN field, fathers are significantly underrepresented. Much of the research focuses on mothers of the children, even when “parents” are represented in the title (Bogossian et al., 2019; Lysak et al., 2024; Nygård et al., 2025; Patty et al., 2024). Somanadhan et al., (2025) identified the need to specifically engage with fathers. Researchers need to employ father targeted studies rather than parent specific to help increase empirical evidence on fathers' experiences (Deneault et al., 2025; Yaremych & Persky, 2023).

2.9 Conclusion

This chapter presented an overall background giving context for this study. Key terminology was identified in relation to children with CACCN. Specifically, this chapter presented the definition, aetiology and classification of a CA. Furthermore, how a CA is diagnosed was explored. This chapter then identified the working definition of children's CCN and highlighted that CCN that can be associated with a CA. This included impacting intellectual functioning, neurodevelopmental and mental health status, feeding, toileting, breathing, circulation, sleep, physical abilities and can impact life expectancy and mortality rates. Finally, this chapter identified why specifically fathers have been selected to be included in this study. The next chapter will present a scoping review on father's experiences of caring for a child with CACCN to scope what evidence is available

pertaining to fathers' experiences of caring for these children and to explore what CA are represented within the literature.

3 Scoping Review

3.1 Introduction

Chapter two presented an overall background to who children with a CACCN are and why fathers' specific experiences was selected to be explored. Chapter three follows on from the background and presents a scoping review of the literature to date titled "fathers' experiences of caring for a child with a CACCN". The aim of this chapter is to contextualise what is known and unknown about this experience. The detailed search strategy and framework for this scoping review is presented. Given the ambiguity of the terminology discussed in chapter two, this chapter maps out the terms used throughout the literature describing children with a CACCN, as well as the different CA represented within the context of fathers' experiences. Three overarching themes and associated sub themes are explored. Finally, this chapter highlights the lack of specific research on father's experiences of caring for a child with a CACCN.

3.2 Context for a scoping review

Most children with a CACCN are now living within the family home, with ever increasing complexities that are typically supported by their parents. Furthermore, as advances in diagnosing CAs have increased, more parents have children with a specific diagnosis which cause their children's disabilities. Chapter two highlighted that in general fathers of children with disabilities are less represented within the literature compared to mothers, despite their roles in caring for their children being equally as important. Fathers' experiences of caring for a child with a CACCN is unclear, and as described in chapter two, the range of terminology associated with this cohort of children is challenging to identify. Despite the growing number of fathers caring for children with a CACCN, no specific synthesis of the literature has been explored.

A scoping review is a method of systematically mapping diverse types of research, that has not been previously reviewed, which allows for the identification of ideas and concepts, in the available evidence, thus identifying gaps within the empirical literature

(Khalil et al., 2025; Munn et al., 2018; Sucharew & Macaluso, 2019). Given the often-multidimensional nature of caring for children with CCN, a scoping review is an appropriate method to systematically explore the breadth of available evidence pertaining to this experience. A scoping review presents the opportunity to complete a thorough synthesis of the available literature focused on the review question without being constrained by study quality or design of a complex phenomenon (Arksey & O'Malley, 2005; Davis et al., 2009). Scoping reviews are different to a systematic review, as typically a quality assessment is not completed (Rumrill et al., 2010), however the major difference between a scoping review and systematic review is that in terms of a review question as a scoping review has a broader “scope” (Munn et al., 2018). Therefore, a scoping review was completed as part of this study. The primary aim of this scoping review was to map out the existing literature and understand what was known about fathers’ experiences of caring for a child with CACCN, and to summarise the key themes identified across studies. The secondary aim was to identify areas for further research, thus giving context and rationale to this study. Scoping reviews are described as a useful step before undertaking a larger body of research (Mak & Thomas, 2022). Scoping reviews can help clarify and improve clarity for those planning, conducting or using research (Cambell et al., 2023).

3.3 Methods

An overview of fathers’ experiences of caring for a child with a CACCN is required to compile the international literature and to map out all available evidence pertaining to these fathers. The methods utilised in this review were guided by Arksey & O'Malley (2005) which was the first published methodological framework for scoping reviews, which was further developed by Levac et al. (2010), and Tricco et al. (2016). This incorporates five stages for completing a scoping review as displayed in Table 3 (Arksey & O'Malley 2005; Levac et al., 2010).

Table 3: The five stages for completing a scoping review

Five stages of completing a scoping review
1. Identifying the research question
2. Identifying relevant studies
3. Study selection
4. Charting the data
5. Collating, summarizing, and reporting results

3.3.1 Identifying the research question

The research question for this scoping review was established through an initial search of the literature which identified a lack of comprehensive synthesis of fathers' experiences of caring for a child with a CACCN. Population, concept and context was utilised to form the research questions. This was discussed with the supervisory team prior to commencing the search. It was noted that when exploring the experiences of parents, fathers were not represented equally within the data on CACCN.

Therefore, the overall research question is: What is known about fathers' experiences of caring for a child with a CACCN?

Moreover, the initial search showed that the range of terminology used to identify CNN, is vast and often challenging to navigate. This challenge therefore identified two further research questions:

1. What CAs are represented in the research?
2. What CCN are identified within the literature?

3.3.2 Identifying relevant studies

A systematic search strategy was employed using the assistance of an expert subject librarian in the School of Nursing and Midwifery to aid the identification of all available literature on the topic. A search using multiple sources is important, accessing the most appropriate electronic databases relative to the phenomenon being explored (Arksey &

O'Malley, 2005; Levac et al., 2010). The systematic search in this study was across multiple data bases including CINAHL Ultimate, Medline, Psycinfo and Embase. These data bases were selected and discussed with the supervisory team, and searches of the grey literature were also included. Controlled vocabulary terms were utilised depending on which database was being searched (Table 4). This review applied the population, concept, context framework to determine inclusion criteria, following the guidance provided by the Joanna Briggs Institute (Peters et al., 2020); population (fathers), concept (experience) context (chromosome abnormalities and complex care needs). The search was completed in November 2021-January 2022, and an updated search was completed in May 2025 to ensure all available literature was included.

Table 4: Data base and controlled vocabulary

Database	Controlled vocabulary terms
CINAHL Ultimate	CINAHL subject headings
Medline	MeSH terms
Psycinfo	APA descriptors
Embase	Emtree

An initial search was conducted to explore terminology used in the population of children with a CACCN to create key terms for the children within this scoping review. Keywords were also identified to broaden the search and index terms were identified to focus the search appropriately which were identified through the literature (The Cochrane Handbook, 2011). These index terms and keywords were then searched using Boolean operators creating search strings. Key search terms and derivatives of these search terms included fathers, experience, CCN and CAs (Appendix 1: Search term derivatives and search string).

3.3.3 Study selection

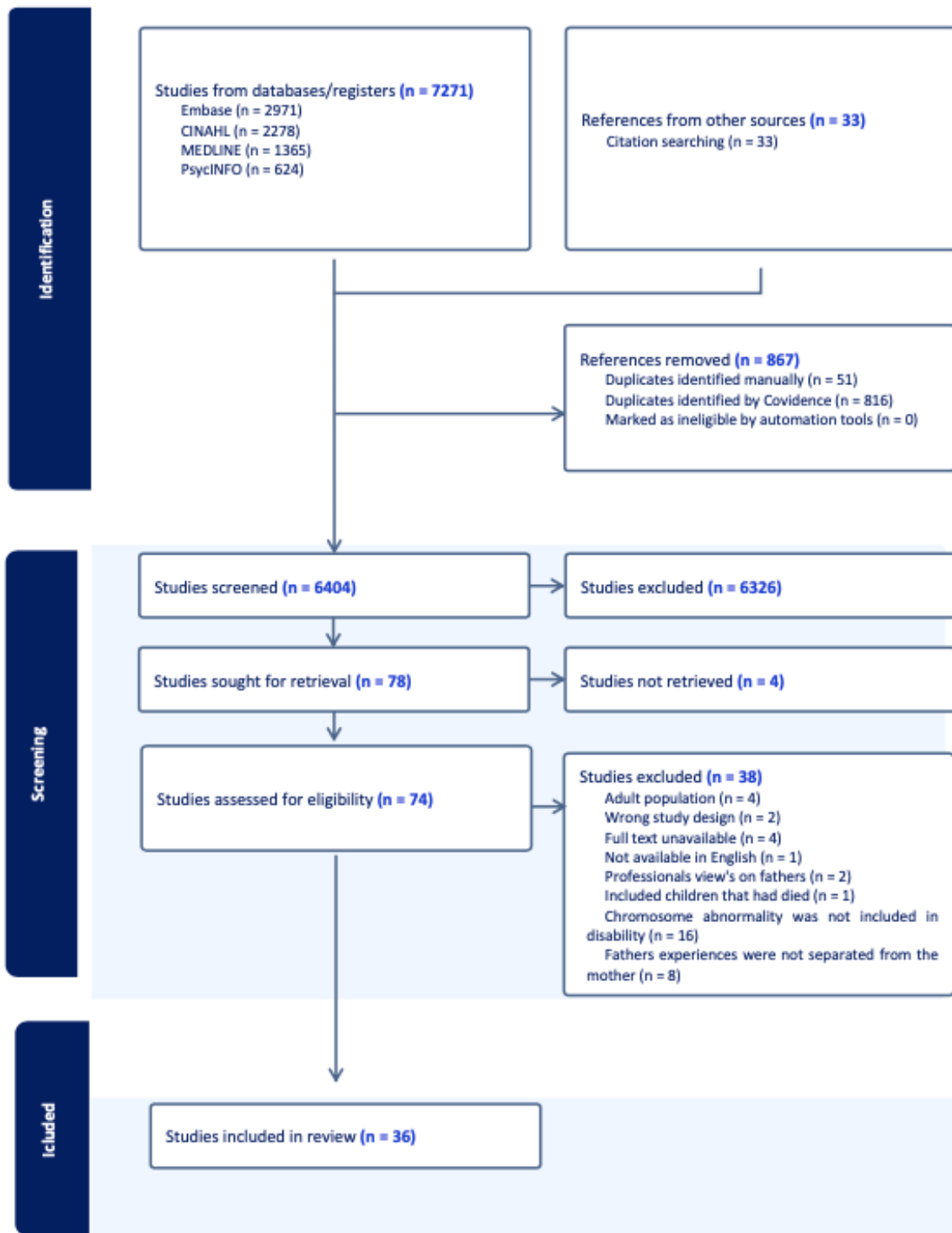
The search was completed across these data bases. A detailed inclusion and exclusion criteria were identified which included both the types of studies that would be included, and participation inclusion/exclusion criteria were devised. The first step of identifying the studies to be included in this scoping review was to review the titles and abstracts

of all yielded studies and they were screened for relevance. This was completed independently by the main researcher (Isobel Mahon). If there were any concerns regarding inclusion or exclusion, this was discussed with the supervisory team. The screening process was conducted using Covidence, a web-based review software. In all perceived eligible studies, the full text was reviewed from the relevant online database and assessed for eligibility. Additional citation and bibliographic searches were conducted on all included studies to identify additional relevant studies for inclusion.

3.3.3.1 Study inclusion and exclusion criteria

Inclusion and exclusion criteria are presented in Table 5. Due to the wide range of publication types that could be included in this scoping review, a meaningful assessment of methodological quality was not completed; this limitation is typical of scoping reviews and distinguishes them from systematic reviews (Munn et al., 2018). Studies were included if they were based on primary research only, to provide a description, summary and critical evaluation of father's experiences (Parahoo, 2014). Scoping reviews provide an overview of the existing evidence, regardless of quality; however, each included article was peer-reviewed or part of national government report/guidelines to ensure some quality (Peters et al., 2015). A time frame limitation was not put on the search. After duplicates were removed 6404 studies were screened for relevance from title and abstract of which 74 were selected for full text reading. The study selection process was documented using the Preferred Reporting Items for Systematic Reviews and MetaAnalyses (PRISMA) flowchart (Figure 2) in line with the PRISMA 2020 guidelines.

Figure 2: PRISMA flow chart



3.3.3.2 Participant inclusion and exclusion criteria

The aim of the scoping review was to explore what is known about the experiences of fathers of children with a CACCN. This review only included studies if the focus was fathers of children under 18, (however, studies were included if some of the participants had children over the age of 18, but the majority needed to be children). Studies were only included if the fathers were the main participants of the research. Therefore, studies that only included mothers or did not report data separately were excluded. If mothers and fathers were included in a large-scale study, but the paper was reporting on fathers' experiences alone, this was included. Only research that explicitly mentioned that it included fathers of children with a CA were included.

Table 5: Inclusion and exclusion criteria

Inclusion criteria	Exclusion criteria
● Fathers of children with a chromosome abnormality. ● Fathers of children with CCN if they included children with a chromosome abnormality. ● Primary research. ● In the English language. ● Peer reviewed. ● Study focus was on fathers of children.	● Mothers' experiences of having a child with complex needs/ or a chromosome abnormality. ● Included mothers and fathers but not specific to fathers. ● Studies that included complex needs but were not explicit in the inclusion of chromosome abnormalities. ● Studies that included chronic conditions but were not explicit in the inclusion of chromosome abnormalities. ● Study focused on adults. ● Not peer reviewed. ● Secondary research

3.3.4 Collecting, summarising and reporting the results

Collecting, summarising, and reporting the results of this scoping review includes a structured approach to mapping and synthesising evidence of fathers' experiences of caring for a child with a CACCN. This includes extracting data, charting the findings, and presenting the results in a clear and organised manner. The next part of this process was extracting data from the included research. This was completed by the main researcher, who created a data-charting form to decide which variables to extract that would best aid in addressing the research question of father's experiences of caring for a child with a CACCN. As recommended by Levac et al. (2010), the literature was reviewed using qualitative descriptive methods, and thematic analysis was employed to analyse the study results. The results were categorised thematically. This was undertaken using Microsoft Excel. The PRISMA Extension for Scoping Reviews (PRISMA-ScR) Checklist and Explanation was used as part of this review to ensure rigour of the results (Tricco et al., 2018), a guide to which scoping review results should be reported (Appendix 2).

3.3.4.1 Charting the data

Data including authors, year and location were compiled. The study population which included how many fathers participated, and children's diagnoses was also extracted. A table highlighting the exact CAs, and terminology used in relation to CCN was developed. Key findings in relation to the research question were also extracted. Detailed methodology, aims and objectives, and participant demographics, key findings and study limitations are presented in Appendix 3.

3.3.4.2 Results

The results yielded thirty-six studies that were included in this scoping review. Studies that were excluded were studies that focused on disabilities such as Autism, ADHD, liver disease, cardiac abnormalities, chronic diseases such as Cystic Fibrosis/Asthma, Fragile X syndrome and Spinal Muscular Atrophy. Included studies were undertaken in Ireland, United Kingdom, Canada, The Netherlands, Spain, Poland, Sweden, The USA, Canada, Australia, Japan, China, Bedouin and Brazil.

When exploring what CAs are represented in the literature, the CA that is predominantly reported on is Trisomy 21. Only five other named CAs were represented throughout the literature (Chromosome 14 deletion syndrome, Mosaic trisomy 14, Chromosome 15/Prader-Willi syndrome, 1P36 deletion syndrome and Williams syndrome). Twelve studies included only children with a diagnosis of Trisomy 21 (Barros Torquo et al., 2013; Bentley et al., 2015; Docherty & Dimond, 2018; How et al., 2019; Lowit & Averill, 2014; Marshak et al., 2019; Ridding & Williams, 2019; Sheldon et al., 2021; Takataya et al., 2016). The remainder of studies did not focus on a CA per se, but these children were included in studies that focused on children with other disabilities but included a CA in the inclusion criteria. Thirteen studies included children with an intellectual disability (Boström & Broberg, 2014; Cameron & Cooper, 2020; Cohen et al., 2016; Giallo et al., 2015; Huang et al., 2011; Huang et al., 2012; Ly & Goldberg, 2014; Mac Donald & Hastings, 2010; Marsh et al., 2018; Marsh et al., 2020; Ogourtsova et al., 2021; Vitiritti et al., 2015). One study explored fathers of children with genetic disorders (Rivard & Mastel-Smith, 2014), congenital abnormalities (Baumann & Braddick, 1999) and unspecific disabilities (Schippers et al., 2020; Uribe-Morales et al., 2022). The final study compared Trisomy 21 to different intellectual disabilities (Ricci & Hodapp, 2003). Trisomy 21 and Down syndrome are both used within the literature. The Term Trisomy 21 will be used throughout as this describes the genetic anomaly (Antonarakis et al., 2020).

In relation to the CCN of children with a CA, intellectual disabilities (including Trisomy 21) were the dominant care needs identified. The remainder included their care needs with other disabilities such as Autism, physical disabilities, genetic disorders, cerebral palsy, hydrocephaly, strokes and acquired brain injuries. Studies that focused on only Trisomy 21, were the only studies that the children's needs were specific to a CA. Table 7 presents an outline of the CA represented, the CCN that were identified and if they were specific to a CA. Some studies identified specific challenges the children faced such as behaviour issues, weight gain, poor sleep, challenges with eating and drinking, toilet training issues, severe multiple disabilities, medical issues, oxygen therapy, ICU admissions, complex heart surgeries, nasal tube feeding (Cohen et al., 2016; Giallo et al.,

2015; Lowit & Averill, 2014; Ogourtsova et al., 2021; Rivard & Mastel-Smith, 2014; Schippers et al., 2020; Sheldon et al., 2021; Takataya et al., 2016). The terminology that is used to describe the children that are represented in this review was also compiled. Intellectual disability/disabilities, Trisomy 21/Down syndrome, CCN, genetic disorders, learning disabilities, mental disorders, developmental challenges, mild/ severe and multiple intellectual disabilities, disabilities, physical disabilities, “children who are not the same” were all used throughout this review to describe children with CCN (Table 6).

Table 6: Chromosome abnormalities represented, terminology used to describe complex care needs and the specific complex care needs identified within the included studies

Study reference	Chromosome abnormalities represented	Terminology used	Any specific CCN identified?
Marsh et al., (2018)	Trisomy 21	Intellectual Disability; developmental delay; Associated severe physical disability; Autism	No
Marsh et al., (2020)	Trisomy 21	Intellectual disability; Global developmental delay; Associated severe physical disability; Autism	No
Barros Torquo et al., (2013)	Trisomy 21	Intellectual disability	Yes
Marshak et al., (2019)	Trisomy 21	Intellectual disability	Yes
Takataya et al., (2016)	Trisomy 21	Intellectual disability	Yes
Rivard & Mastel-Smith, (2014)	Trisomy 21 Prader-Willi syndrome	Intellectual disability; Genetic disorders; Complex medical needs	No
Boström & Broberg, (2014)	Trisomy 21 Deletion chromosome 14 Mosaic trisomy 14	Intellectual/developmental disability; Autism	No
Ridding & Williams, (2019)	Trisomy 21	Intellectual disability	Yes
Lowit & Averill, (2014)	Trisomy 21	Intellectual disability	Yes
Cohen et al., (2016)	Trisomy 21	Intellectual disability; Cerebral palsy; Undifferentiated developmental disability; Behaviour issues	No

Cameron & Cooper (2020)	Trisomy 21	Mild learning disability; Learning disability; Autism	No
Bentley et al., (2015)	Trisomy 21	Intellectual disability	Yes
Vitiritti et al., (2015)	Trisomy 21	Mental disability	No
Docherty & Dimond (2018)	Trisomy 21	Intellectual disability	Yes
Huang et al., (2012)	Trisomy 21 Chromosome abnormalities (unspecific)	Intellectual disabilities; Cerebral Palsy; Autism; Tetralogy of Fallot; Hydrocephaly; Stroke; Neurological sequelae of infection with enterovirus 71	No
How et al., (2019)	Trisomy 21	Intellectual disabilities	Yes
Huang et al., (2012)	Trisomy 21 Chromosome abnormalities (unspecific)	Intellectual disabilities; Cerebral Palsy; Autism; Tetralogy of Fallot; Hydrocephaly; Stroke; Neurological sequelae of infection with enterovirus; Time sensitive disability management	No
Huang et al., (2011)	Trisomy 21 Chromosome abnormalities (unspecific)	Intellectual disabilities; Cerebral Palsy; Autism; Tetralogy of Fallot; Hydrocephaly; Stroke; Neurological sequelae of infection with enterovirus	No
Baumann & Braddick, (1999)	Trisomy 21	Intellectual disabilities/ Significant intellectual disabilities; Congenital anomalies; Heart defects Cleft lip and pallet; Unpredictable conditions	No
Giallo et al. (2015)	Trisomy 21	Autism; Vision impairment; Hearing impairment; Cerebral palsy; Epilepsy or other seizure disorders; Psychiatric disorders; Attention deficit and hyperactivity disorder; Acquired brain injury; Behaviour problems; Health problems (i.e., asthma, food allergies)	No
Ricci & Hodapp, (2003)	Trisomy 21 Williams syndrome Prader–Willi syndrome	Intellectual disabilities; Cerebral palsy; Autism; Maladaptive behaviours	No

Schippers et al., (2020)	Trisomy 21	Intellectual disability; Physical disability; Physical and intellectual disability	No
Ly & Goldberg, (2014)	Trisomy 21	Autism; Intellectual disabilities; Attention deficit hyperactivity disorder	No
Mac Donold & Hastings (2010)	Trisomy 21	Autism; Cerebral palsy; Intellectual disabilities	No
Ogourtsova et al., (2021)	Trisomy 21	Epilepsy; Anxiety; Cognitive and language delay; Cerebral palsy; Autism; Attention deficit and hyperactivity disorder; Developmental coordination disorder; Behavioural challenges; Globoid cell leukodystrophy	No
Sheldon et al., (2021)	Trisomy 21	Intellectual disability; Challenging behaviour and health concerns such as challenges with toileting, feeding difficulties medical issues, and physical limitations).	Yes
Uribe-Morales et al., (2022)	1p36 deletion syndrome Trisomy 21	Hearing impairment; ADHD; Oppositional defiant disorder; Autism;	No
Pisula & Banasiak (2020)	Trisomy 21	Mild intellectual disability; Moderate intellectual disability; Severe intellectual disability; Profound intellectual disability	Yes
Dabrowska & Pisula (2010)	Trisomy 21	Autism; Intellectual disability; Behaviour	Yes
Rose, (2021)	Trisomy 21	Down syndrome; Medical complexity during diagnoses stage; Disability	Yes
Armijos-Yambay, (2020)	Trisomy 21	Intellectual disability; Developmental disability	Yes
MacDonald et al., (2010)	Trisomy 21	Autism; Cerebral Palsy; Behaviour problems	No
Manor-Binyamini, (2025)	Trisomy 21 Unnamed chromosomal condition	Developmental disabilities; Chromosomal conditions; Intellectual disability; Autism	No
Randolph (2023)	Trisomy 21	Intellectual disability; Intellectual impairments; Medical issues; Uncertain futures; Behaviours; Communication challenges	Yes

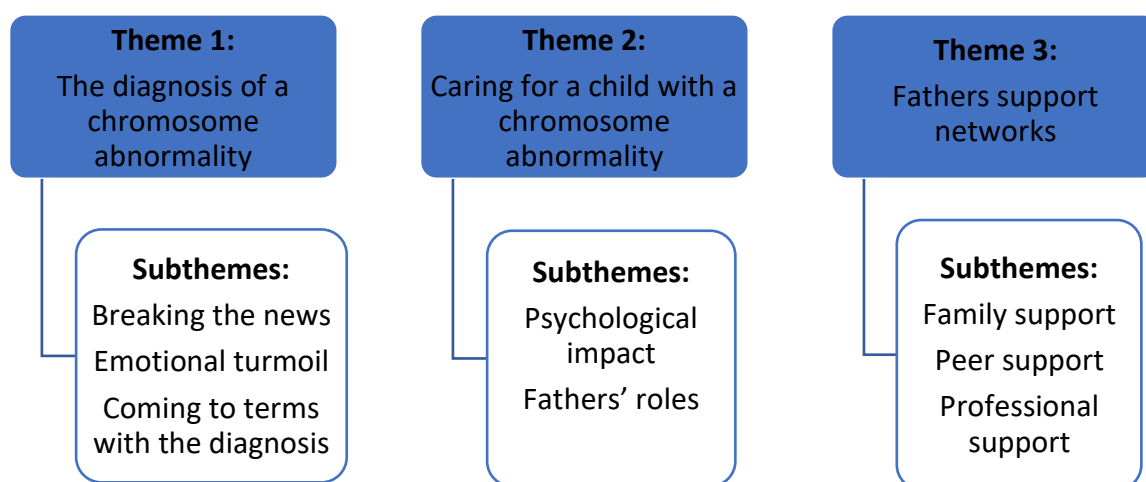
Woodgate et al., (2024)	Trisomy 21	Complex Care Needs (ongoing medical and/or pharmacological treatment needed as well as the condition's severity, rarity, and comorbidities)	No
Walker, (2012)	Trisomy 21	Intellectual disabilities; Physical disabilities	No

3.4 Thematic findings

A thematic analysis was undertaken to synthesise and interpret the key findings across the included literature on fathers' experiences of caring for a child with a CACCN. Following the full text review, the main experiences from each study were carefully examined and compared. Common patterns and recurring topics were identified through reading and re-reading the studies, allowing for the development of overarching themes that captured the core content of the literature. This approach enabled a structured and meaningful synthesis of the literature, highlighting key areas of focus and gaps in current research.

Following this thematic analysis, three themes, and eight subthemes were identified (Figure 3). The first theme, fathers reflect on how they were told about the diagnosis, their feelings and experiences following the diagnosis and their methods of coping with this diagnosis. The second theme refers to fathers' experiences caring for a child a CACCN; both the psychological impact this had, and their roles and responsibilities in caring for them. The final theme emphasises fathers' support networks.

Figure 3: Scoping review themes and subthemes



3.4.1 Theme one: The diagnosis of chromosome abnormality

The first theme in this review relates to the available evidence pertaining to fathers' experiences of how their child was diagnosed with a CA and/or CCN. There are three

subthemes related to this experience. The first subtheme fathers recall how the news was broken to them. The second subtheme delves into the emotional turmoil associated with the new diagnoses. The final subtheme refers to how fathers began to cope and come to terms with the new diagnoses. It is evident, though, that current research on fathers of children with a CA and CCN disproportionately investigates the perspectives of fathers raising children with Trisomy 21, while insights from those parenting children with other conditions remain underrepresented.

3.4.1.1 Breaking the news of the diagnosis

Fathers recall how they initially learned about their child's diagnosis, with many remembering explicitly how they received this information from professionals (Baumann & Braddick, 1999; Docherty & Dimond, 2018; Huang et al., 2012; How et al., 2019; Marsh et al., 2020). Generic challenges, such as poor bedside manners, were common throughout the literature (Rose, 2021; Docherty & Dimond, How et al., 2019; Lowit & Avrill, 2014). However, most of the evidence on this experience is shaped by the type of diagnosis that the children received. Much of the existing research is framed around the perspectives of fathers whose children were diagnosed with Trisomy 21 (Rose, 2021; Docherty & Dimond, How et al., 2019; Lowit & Avrill, 2014) or they are included within the research of fathers of children with an intellectual disability (Huang et al., 2011, 2012a, 2012b; Lowit & Avrill, 2014; Marsh et al., 2020).

A substantial body of literature has examined the language used by healthcare professionals when delivering a diagnosis of Trisomy 21. Fathers often perceive this language as overly medicalised, failing to reflect the individuality of their children. Many report that the emphasis on potential medical complications associated with Trisomy 21 felt irrelevant or disconnected from their personal experiences. For instance, Docherty & Dimond, (2018) presented an interpretative phenomenological analysis (IPA) exploring the experiences of seven fathers of children with Trisomy 21, focusing on their interactions with professionals. Findings highlight that the communication around diagnosis was paramount in influencing fathers' own personal views. Professionals used negative language which in turn resulted in fathers experiencing pessimistic feelings

towards their own children and their child's future. Fathers felt that the negative connotations around Trisomy 21 did not accurately reflect their child or their child's development (Docherty & Dimond, 2018). Similarly, a qualitative study that interviewed five fathers of children with Trisomy 21, presented fathers' views on the availability of non-invasive prenatal testing, and reported that fathers recall the pressure they felt for the mother to terminate the pregnancy following a prenatal diagnosis. Fathers highlighted the perceived over-medicalisation and clinical, often negative framing of Trisomy 21 by professionals, failed to adequately describe their child. This left some questioning whether they might have chosen to terminate the pregnancy if they had relied solely on the information provided by medical staff (How et al., 2019). Upon reflection, some fathers felt that professionals frequently underestimated their child's potential, focusing more on perceived limitations than capabilities (Barros Torquato et al., 2013).

However, these accounts may not reflect the experiences of fathers whose children with Trisomy 21 also presented with ongoing medical complexities. Rose (2021), using a narrative methodology for a PhD thesis, explored the experiences of 22 fathers of children with Trisomy 21. Many described similarly low expectations to those conveyed by healthcare professionals. Notably, fathers of children with medical complications during the neonatal period rarely commented on messages about Trisomy 21 itself. As medical severity and CCN increased, fathers tended to concentrate on the immediate health concerns of their newborns during the neonatal period rather than any other narrative detail, often to the exclusion of broader narratives including their own emotions and feelings when receiving the diagnosis. Rose's study identified a need for further research that distinguishes between fathers' initial reactions at the point of diagnosis and their later experiences in parenting, especially when medical complexities persist beyond the neonatal period.

The timing of a diagnosis emerged as a significant theme across the literature, particularly in studies focused on Trisomy 21, where diagnosis typically occurred prenatally or during the neonatal period. Huang et al., (2011, 2012a, 2012b) conducted

a hermeneutic phenomenological study, publishing findings from the same group of 16 fathers of children with developmental disabilities, drawing on the philosophies of Heidegger and Gadamer. Among the participants, four children had a CA, including Trisomy 21 and an unspecified CA. Fathers had differing experiences depending on the time of diagnosis. Some identified their child's CA at birth based on distinct phenotypic features, however some presented with delayed diagnosis based on their children missing developmental milestones which produced unique experiences and support needs depending on the time of diagnosis (Huang et al., 2011). Notably, the children diagnosed with developmental disabilities in this study because of missed milestones did not have a CA. Similarly, Marsh et al. (2019, 2020), completed a qualitative descriptive study with ten Irish fathers whose child had received a diagnosis of an intellectual or developmental disability. Six out of the ten children had a diagnosis of Trisomy 21 and all but one received a diagnosis straight after birth. The other four children had not received a diagnostic cause for their intellectual disability. None of the included studies identified the experiences of fathers when their child missed developmental milestones because of their child having a CA.

3.4.1.2 Emotional turmoil due to the diagnosis

Fathers reflect on the emotional aspect of receiving a diagnosis, which is mostly from the perspective of hearing that their child has a diagnosis of Trisomy 21. Fathers often described feelings of shock, disappointment and devastation (Marsh et al., 2018; Marsh et al., 2020; Rivard & Mastel-Smith, 2014; Uribe-Morales et al., 2022). The expectation of having a typically developing child was exciting for fathers, and when they discovered something was atypical this was met with feelings of fear, grief and confusion (Barros Torquo et al., 2013; Marsh et al., 2018; Marsh et al., 2020; Marshak et al., 2019; Rivard & Mastel-Smith, 2014; Ridding & Williams, 2019). The oldest of the included studies, Baumann & Braddick (1999) explored the experiences of eleven fathers of children who “were not the same” and reported the emotions of the “sad news”. This included five children with Trisomy 21 and four children with other anomalies not described. This study also included two children with “less severe anomalies” such as club foot and ADHD. The broad inclusion criteria limited the ability to isolate experiences specific to

distinct diagnoses. Moreover, this study consolidated the findings and did not describe experiences for each of the different conditions.

Some fathers reported grieving the loss of their imagined "typical" child, but the medical complexities during this period were often a greater challenge (Rose, 2021; Lowit & Averill, 2014; Rivard & Mastel-Smith, 2014; Takataya et al., 2016; Walker, 2015). Rivard & Mastel-Smith (2014) conducted a qualitative study reported on the lived experience of seven fathers living in the USA, whose children were diagnosed with genetic disorders - four unspecified, one with Trisomy 21 and one with Prader-Willi syndrome. Their study highlighted that the complexity of medical issues at diagnosis heightened initial fear and anxiety during the perinatal and neonatal period (Rivard & Mastel-Smith, 2014). Often this worry was during the neonatal period and soon subsided as the complexities stabilised. Adding to this, Lowit & Avrill (2014) published results of an exploratory study investigating the experiences of 13 fathers upon the postnatal diagnosis of Trisomy 21. This study found that fathers whose children did not require heart surgery experienced fewer negative emotions and recalled the diagnosis more positively. However, even for those requiring surgery, eventual positive outcomes helped reduce long-term anxiety (Lowit & Avril, 2014). Similarly, in a Japanese study exploring the perceptions and feelings of nine fathers of children with Trisomy 21, fathers expressed emotional relief once their child's immediate medical issues had resolved (Takataya et al., 2016).

The increased incidence of Trisomy 21 in comparison to other rarer conditions, meant that some fathers were aware of what Trisomy 21 might look like and what to potentially expect. In one large scale qualitative study that included 311 fathers who answered open-ended survey questions on the personal impact of raising children with Trisomy 21, found that less than a quarter of the participants reported experiencing sadness, loss and depression while trying to adjust to a life they did not expect (Marshak et al., 2019). Rose (2021) found that of the fathers, who were personally familiar with someone with Trisomy 21, the diagnosis was easier to manage emotionally. In another study by Lowit & Avrill, (2014) one father recalled fond memories of how the doctor diagnosing his child, also had a child with Trisomy 21, bringing comfort and hope to the situation.

3.4.1.3 Coming to terms with the diagnosis

Evidence highlights the significance of fathers reaching acceptance of their child's disability. A qualitative study by MacDonald et al. (2010) found that when fathers accepted their child's diagnosis, this was positively associated with their perception of the personal gains and positive experiences related to raising a child with an intellectual disability. Methods such as gaining knowledge of the diagnosis, changing their expectations of their children and remaining hopeful that their children would gain new skills allowed some fathers to cope with their new reality (Bentley et al., 2015; Cameron & Cooper, 2020; Rose, 2021; Huang et al., 2012; LaTanya & Randolph, 2023; Marsh et al., 2020; Marshak et al., 2019; Vitiritti et al., 2015). Often this included fathers being hopeful that their children with Trisomy 21 would achieve milestones that one might expect from typically developing peers. Armijos-Yambay, (2020) explored fathers' lived experiences of children with Trisomy 21 in a hermeneutic phenomenological study guided by van Manen's approach. Armijos-Yambay argues that having a hopeful perspective is vital to supporting a child with Trisomy 21, and encouraging a hopeful narrative can benefit both families and professionals who work with them. For some, their children with Trisomy 21 became like any "normal child" that just needed a little more dedication (Barros Torquo et al., 2013). Much of what is known though, is largely based on experiences of fathers of children with Trisomy 21 and this may not fully represent the experiences of those dealing with other, more medically complex or less common disabilities.

Boström & Broberg (2014) conducted a longitudinal qualitative study on fathers' experiences of parenting a child with an intellectual disability on seven Swedish fathers. This was an IPA that included the largest variety of CAs represented. Four of the seven children had a diagnosis of a CA (two children with Trisomy 21, deletion chromosome 14, mosaic trisomy 14). Fathers described developing emotional stability in relation to their child's diagnosis. When they accepted that their children's developmental pathways needed to go at their own pace, some of the fathers ceased attempting to avoid the impact of the disability. Feeling gratitude that their child's disability was not more severe was evident throughout the literature, with fathers feeling gratitude for

what their children could achieve because their children's disability was Trisomy 21. Some fathers reported feelings of relief that the diagnosis their child had was not a condition with perceived worse outcomes (Rose, 2021). Walker (2012) explored the experiences of eight fathers who have parented children with and without intellectual disabilities, using a qualitative descriptive design and one father reported being grateful that their children did not have complexities such as being in wheelchairs and could speak to them.

3.4.2 Theme two: Caring for a child with a chromosome abnormality and complex care needs

The second theme in this review relates to the available evidence pertaining to fathers' experiences of caring for their child. There are two subthemes related to this experience. The first subtheme highlights the psychological impact of caring for their children. Fathers discuss the joy that their children bring them, alongside at times challenging aspects of care. The second subtheme highlights the roles and responsibilities fathers assume in their caring roles. However, in a similar manner to theme one, studies on the experiences of caring for a child with a CACCN predominantly draw from experiences related to Trisomy 21, with little findings related to other less common CAs.

3.4.2.1 Psychological impact of having a child with a chromosome abnormality and complex care needs

The psychological impact that caring for their child has on fathers is described throughout the literature (Cameron & Cooper, 2020; Cohen et al., 2016; Giallo et al., 2015; Huang et al., 2011; Randolph, 2023; Marsh et al., 2020; Marshak et al., 2019; Ricci & Hodapp, 2003; Ridding & Williams, 2019). Much of the evidence of the physiological impact is based on diagnosis of Trisomy 21 and often highlights the joy that these children bring. For instance, Sheldon et al. (2021) conducted a survey with open-ended questions for fathers of children with Trisomy 21 about their parenting experiences. Their findings highlighted that the positive, fun personalities associated with Trisomy 21 brought fathers much joy.

Ricci & Hodapp (2003) explored whether different diagnoses influence paternal stress. This study presented fathers' perceptions of stress relating to children with Trisomy 21 versus those with other types of intellectual disabilities. Fathers of children with Trisomy 21 reported less overall child-related stress (Ricci & Hodapp, 2003). Another study compared parenting stress and coping styles in both mothers and fathers of pre-school children with Trisomy 21, Autism and typically developing children. The findings suggest that fathers of children with Autism have higher levels of stress than fathers of children with Down syndrome (Dabrowska & Pisula, 2010). Furthermore, there was no difference in the stress levels between those with Trisomy 21 and typically developing children (Dabrowska & Pisula, 2010). This was the only included study that was based on mothers and fathers, as this reported the results separately.

When Trisomy 21 is included within intellectual disability, fathers appear to be more at risk of poor mental health. Giallo et al., (2015) published a study exploring the risk factors associated with the mental health of fathers of children with an intellectual disability in Australia. This was a large-scale quantitative study that included fathers of children with a range of diagnoses leading to an intellectual disability, one of which was Trisomy 21. This study found that there is an increased risk of depression and stress symptoms for this group of fathers (Giallo et al., 2015). Cohen et al. (2016) presented a quantitative study exploring long-term psychological symptoms in fathers of children with and without intellectual disabilities (27.8% of whom had Trisomy 21) and reported that behavioural co-morbidities in the child were associated with elevated levels of paternal stress, anxiety, and depression.

Similar challenges in the literature relate to the co-morbidities associated with Trisomy 21, for example, a UK study using a Grounded Theory approach by Ridding & Williams (2019) explores the experiences of fathers parenting a child with Trisomy 21, and reported that some fathers were more stressed during the initial diagnosis period, and this reduced over time (Ridding & Williams, 2019), yet increased stress levels were more likely to persist if their children had co-morbidities. Randolph (2023) highlighted that behaviour and worries about previous health co-morbidities were barriers for fathers maintaining resilience in a generic qualitative study conducted in the USA.

Schippers et al. (2020) conducted a qualitative study involving Dutch fathers of children with various intellectual disabilities, including an unspecified number with Trisomy 21. Over half of the fathers reported difficulties coping, often linked to anxieties about their child's life expectancy. The underlying reasons for these fears, however, were unclear. This was largely due to the vague or non-specific nature of the diagnosis. The potential for a child's death or ongoing medical complexity beyond the initial diagnosis was not widely addressed in the included literature, despite recurring paternal concerns about long-term care. Woodgate et al. (2024) was the only study that specifically examined the experiences of fathers caring for a child with CCN. Although not limited to CAs, it included children with congenital and genetic conditions such as Trisomy 21. A key source of ongoing stress identified in this study was related to future caregiving responsibilities and the challenges of transitioning into adult healthcare systems.

The stigma in relation to their children's disability and the psychological toll this takes on fathers is also documented. Many of the fathers in the Randolph (2023) study, reported feeling guilty that they felt the need to hide their child due to their children's diagnosis of Trisomy 21. Similarly, Manor-Binyamini (2025) presented findings of Bedouin fathers of children with developmental disabilities. This study included children with Trisomy 21, and an unnamed CA and reported that some fathers experience negative affiliate stigma as a result of their child's diagnosis (Manor-Binyamini, 2025).

While there is considerable research on paternal experiences with Trisomy 21 and general intellectual disability, there remains a notable gap in focused, qualitative studies exploring the psychological impact and unique experiences of fathers caring for children with a CACCN. In particular, the intersection of medical complexity, future caregiving anxieties, and psychosocial stressors remains underexplored, especially in studies that disaggregate data specifically for fathers.

3.4.2.2 Fathers' roles in caring for their children

The empirical evidence on this cohort of fathers' highlights that most fathers report wanting to be involved in the care of their children. Many fathers perceive their caring roles as extremely important within the family unit (Docherty & Dimond, 2018; Huang

et al., 2011, 2012; Ricci & Hodapp, 2003; Schippers et al., 2020; Uribe-Morales et al., 2022), although some still reported that the mother was the primary carer (Barros Torquo et al., 2013).

One phenomenological study conducted in Spain explored fathers of children with generic disabilities, this included one child with 1P36 deletion syndrome and Trisomy 21. Findings affirm that mothers are no longer the only parent that wants to be involved, and fathers are dedicated to care routines (showering, feeding, and dressing) (Uribe-Morales, et al., 2022). Woodgate et al. (2024) described the overarching theme of “wanting to be there for their children” (p. 6). Fathers report that they adapt and do what needs to be done in the care of their children (Walker, 2012). Many fathers report being involved in all aspects of their children’s care such as nappy changes, bathing, feeding and medication preparations (Huang et al., 2012; Ogourtsova et al., 2021; Uribe-Morales et al., 2022; Woodgate et al., 2024). Some document how they share care responsibilities, in a tag team approach (Woodgate et al., 2024).

Even so, despite wanting to be involved, some fathers reflect on the concept of “masculinity” in their caring roles. Cameron & Cooper (2020) conducted an IPA in the UK of four fathers’ experiences of caring for children with both autism and a learning disability (a dual diagnosis), which included one child with Trisomy 21. This study did not explore CAs per se, but did add an additional diagnosis to the learning disability. Fathers pondered over the legitimacy of their caring roles compared to the mother of the child. For some the fact that their child with a disability was not “typical”, meant that fathers regretted the absence of the roles such as being an educator and/or supervisor which were often perceived as traditional fathering roles (Schippers et al., 2020). Furthermore, one Irish study explored the psychological variables that may explain some variation in parenting by fathers of children with intellectual disabilities (Mac Donold & Hastings, 2010). They conducted a quantitative questionnaire survey and found that fathers were more involved with socialisation related parenting tasks if their child was male (Mac Donold & Hastings, 2010).

Although existing studies acknowledge the growing role of fathers in caregiving, much of the literature remains focused on general intellectual or developmental disabilities, with limited attention to chromosome abnormalities or medically complex conditions. While some studies include children with Trisomy 21, few explore the unique challenges faced by fathers caring for children with rarer CA or CCN. Challenges related to masculinity, identity, and long-term caregiving responsibilities remain poorly understood

3.4.3 Theme three: Support

The third theme identified in this review explores the available evidence on fathers' experiences of support networks. Three subthemes emerged in relation to this theme. The first concerns the role of family support, which can exert both positive and negative influences on fathers' wellbeing. The second highlights the value of peer support, which many fathers described as an essential coping mechanism in their caregiving journey. The final subtheme focuses on interactions with professionals, emphasising that these relationships whether supportive or challenging can significantly impact fathers' experiences and sense of support. However, fathers of children with Trisomy 21 are well represented in the literature on fathers' support networks, there is a significant lack of research addressing the experiences of fathers managing less common conditions or more complex diagnoses.

3.4.3.1 Family support networks

Support from a spouse or partner is consistently identified as a key factor in reducing paternal stress, anxiety, and depression (Cohen et al., 2016; Randolph, 2023; Rivard & Mastel-Smith, 2014; Takataya et al., 2016). Some fathers reported that their child's diagnosis brought them closer to their partner (Sheldon et al., 2021; Uribe-Morales et al., 2022), while others experienced increased tension within their relationships (Ridding & Williams, 2019; Takataya et al., 2016; Uribe-Morales et al., 2022). In a Brazilian hermeneutic phenomenological study of fathers of children with a "mental disability" (which included Trisomy 21 within this cohort), Vitiritti et al. (2015) highlighted that at times differing views on management of their children for example, education around their sexuality is an area that can cause tension.

Extended family members were also noted as an important source of support for some fathers of children with Trisomy 21 (How et al., 2019; Randolph, 2023; Takataya et al., 2016). However, as with partners, family involvement can also be a source of distress when conflicting views or expectations arise, as has been identified in the context of Trisomy 21 and intellectual disability (Rose, 2021; Huang et al., 2011).

While the literature acknowledges the role of family support networks on paternal wellbeing, few studies specifically examine how fathers navigate these dynamics in the context of CA or CCN. The role of family conflict, shared decision-making, and emotional strain in shaping the father's caregiving experience remains underexplored.

3.4.3.2 Peer support

Peer support in the form of chromosome specific groups and specific organisations allowed fathers to meet parents in similar circumstances, and they deemed this fundamental in their coping. This is described well by Bentley et al. (2015) who found many fathers enjoyed getting involved in social support groups and Down syndrome organisations in a mixed method study that described fathers' attitudes and coping behaviours, responses to their child having Trisomy 21. Most of the available evidence on peer support for fathers, notably, centres on the Trisomy 21 community (Rose, 2021; Lowit & Averill, 2014; Marshak et al., 2019; Ridding & Williams, 2019).

Fathers of children with Trisomy 21 identified participation in support groups as beneficial, particularly for gaining insights from other parents, expanding their social networks, and fostering a sense of community engagement (Rose, 2021; Ridding & Williams, 2019; Sheldon et al., 2021). Social support is vital to helping fathers cope which in turn helps fathers engage with their child. This is highlighted in one quantitative Polish study that reported on fathers of children with Autism and Trisomy 21 in the context of the role of social support and coping with stress. This study revealed that the more supported fathers of children with Trisomy 21 felt socially, the more empowered they were in community involvement and engagement (Pisula & Banasiak, 2020). Fathers of children with CCN also appear to have a desire to be involved in philanthropic initiatives, community organisations, and parent associations in the only study included that

specifically included fathers of CCN (it must be noted that Trisomy 21 was the only CA in this cohort) (Woodgate et al., 2024).

Despite strong evidence of the benefits of peer support for fathers of children with Trisomy 21, there is a notable lack of research exploring how fathers of children with other CAs or broader CCN access and experience such support. Little is known about whether similar opportunities for connection and empowerment exist beyond the well-established Down syndrome networks.

3.4.3.3 Support from professionals

Communication between fathers and professionals plays a key role in shaping how supported fathers feel (Huang et al., 2011, 2012; Ly & Goldberg, 2014; Lowit & Averill, 2014; Rivard & Mastel-Smith, 2014; Takataya et al., 2016; Docherty & Dimond, 2018; How et al., 2019). Much of this research focuses on fathers of children with Trisomy 21, or Trisomy 21 is included within broader studies on fathers' experiences caring for children with intellectual disability.

Many fathers feel that they are treated unequally to the mother by professionals (Docherty & Dimond, 2018; Huang et al., 2012; Rivard & Mastel-Smith, 2014). Findings from a study by Ly & Goldberg (2014) presented the perspectives of fathers caring for a child with an intellectual disability, including Trisomy 21, using a mixed method approach of both semi-structured interviews and a questionnaire. This study reported that those fathers felt professionals considered them secondary parents to mothers. In further research, fathers described their roles in the family as not being acknowledged by professionals (Docherty & Dimond, 2018), and with their input often ignored or redirected to the mother (Huang et al., 2012; Ly & Goldberg, 2014; Rivard & Mastel-Smith, 2014). Similarly, fathers of children with CCN reported that professionals frequently deferred to mothers and wished to speak with them, disregarding the father's desire to share responsibilities equally (Woodgate et al., 2024).

When fathers are treated as equal partners in care, positive interactions with health professionals can be a vital source of support during challenging times (Huang et al., 2011). Ogourtsova et al. (2021) conducted a study in Canada, which explored the

barriers and facilitators to positive and empowering healthcare experiences, of fathers of children with a developmental disability (including Trisomy 21) and also experiences from professionals. This study used a mixed-method approach, using a quantitative survey and qualitative semi-structured interviews. Fathers emphasised the value of respectful, inclusive communication with professionals. Accessing health professionals through publicly funded services for children with CCN, though, has been identified as a particular challenge. For instance, Canadian fathers have reported inadequate provision and availability of such services (Woodgate et al., 2025), posing a barrier to accessing essential support.

In addition to feeling excluded from interactions with professionals, fathers also describe being overlooked in research. Marsh et al. (2020) specifically asked fathers what their motivation was in being part of research on raising a child with an intellectual disability (including Trisomy 21). Findings showed that fathers are motivated to participate in research about them, but that due to their experiences of being left out of conversations, they often need to be specifically asked to participate. The study concluded that fathers must be actively given opportunities to share their perspectives and be included in research on caregiving (Marsh et al., 2020). However, Uribe-Morales, et al. (2022) reported one of the limitations of their study was a lack of engagement and reported enormous difficulties in finding a sufficiently large sample of fathers who wanted to participate, even when they were invited (Uribe-Morales, et al. 2022).

The importance of father-professional communication is increasingly acknowledged, though existing research remains heavily focused on fathers of children with Trisomy 21 or general intellectual disability. There is a lack of targeted exploration into how fathers of children with other CA or CCN experience healthcare interactions and professional support. There is a need for more research that amplifies fathers' voices in healthcare and research contexts, particularly concerning CCN and CA aside from Trisomy 21.

3.5 Methodological considerations

This scoping review prioritises the exploration of meaning and lived experience over strict methodological evaluation. Each study offers a valuable perspective that deepens

the overall understanding of fathers' experiences. Therefore, limitations to each study were noted. A variety of methodological approaches were used within the included studies in this scoping review. These are presented in Appendix 3.

A large proportion of the included studies used a qualitative design, with a wide range of qualitative methodologies. Several adopted IPA to explore fathers' lived experiences (e.g., Rivard & Mastel-Smith, 2014; Boström & Broberg, 2014; Cameron & Cooper, 2020; Docherty & Dimond, 2018). These studies presented rich and detailed descriptions of how fathers make sense of fathering their children. Other qualitative studies used hermeneutic phenomenology, with some grounded specifically in Heideggerian philosophy (Vitiritti et al., 2015; Huang et al., 2011, 2012a,b), and Armijos-Yambay, (2020) used van Manen's hermeneutic phenomenology. Using hermeneutic phenomenological approach allowed these studies to present deep insights into fathers' lived experience by uncovering the existential meaning behind their narratives.

A number of studies used narrative inquiry to capture the temporal and storied nature of this experience (Marsh et al., 2018, 2020; Rose, 2021), or descriptive and exploratory designs aimed at producing insights without a strong theoretical lens (Barros Torquo et al., 2013; Takataya et al., 2016; Walker, 2012; Baumann & Braddick, 1999). Some studies were guided by a grounded theory approach (Ridding & Williams, 2019) or symbolic interactionism (How et al., 2019), while Woodgate et al. (2024) adopted an ethnographic approach to engage more creatively and culturally with fathers' experiences. Some studies used open ended survey responses as a form of qualitative data collection (Marshak et al., 2019; Sheldon et al., 2021), though these tended to offer less methodological transparency. Notably, a few studies (e.g., Schippers et al., 2020; Randolph, 2023) were described as generic or qualitative without a clear methodological or philosophical foundation, limiting interpretive clarity and theoretical alignment.

Several studies in this review employed quantitative methodologies to examine fathers' psychological wellbeing, parenting experiences, and support needs in the context of raising a child with a developmental disability. These studies typically used self-report questionnaires to measure factors such as stress, coping, child behaviour, and family

functioning (e.g., Cohen et al., 2016; Giallo et al., 2015; Pisula & Banasiak, 2020). Some focused on evaluating the outcomes of parenting interventions or support programmes, while others explored associations between paternal mental health and external factors such as spousal support or service use (MacDonald et al., 2010; Manor-Binyamini, 2025). Although this type of research provided insight into fathers' experiences, it tended to quantify them rather than offer a deep, reflective exploration of their lived experiences.

Three studies included a mixed method approach, in which self-reporting questionnaires were coupled with qualitative questionnaires or interviews (Bentley et al., 2015; Ly & Goldberg, 2014; Ogourtsova et al., 2021). One study only asked three open ended questions (Bentley et al., 2015), while the other two used a less structured interview style (Ly & Goldberg, 2014; Ogourtsova et al., 2021).

Across the studies reviewed, several common limitations emerged, including small and homogenous samples and at times a lack of methodological transparency. Many studies highlighted limitations in relation to the recruitment process, as fathers were recruited through support groups or specific services, resulting in self-selecting samples that may not represent more marginalised or under-resourced populations even within the Trisomy 21 co-hort. These issues suggest a need for more inclusive research on fathering in the context of CACCN.

3.6 Discussion

This scoping review aimed to explore what is known about the experience of fathers caring for a child with a CACCN. The findings highlight that fathers have unique experiences in caring for their children. Many fathers feel their roles are equally as important in caring for their children as the mother and want to be part of the care giving team. This is despite findings from a systematic review and meta-analysis on fathers caring for children with an intellectual disability, which indicated that fathers continue to play a limited role in care and questioned whether fathers want to be involved in caring for their children (Dunn et al., 2019).

Findings also show that Trisomy 21 is the predominant diagnosis identified within the literature; these findings are therefore largely specific to fathers of children with this

diagnosis. Trisomy 21 is the most frequent CA diagnosed at birth (Queremel Milani & Tadi, 2023). This scoping review found little information related to fathers' experiences of caring for a child with other CA that are often much rarer, but complexities can be greater, for instance Trisomy 13 and Trisomy 18. Most children with these conditions die within the first month of life (74.5% and 62.8% respectively) (Meyer et al., 2016; Milligan et al., 2020; Nelson et al., 2016). This means however, that 25.5% and 37.2% of these children are living beyond that age, often with extremely CCN. Research shows the importance of exploring the experiences that children with different disabilities and conditions have on fathers (Bailey-Pearce et al., 2017; Carpenter & Towers, 2008; Goble, 2004; Lashewicz et al., 2019; Ogourtsova et al., 2021; Postavaru et al., 2021; Waite-Jones & Madill, 2008). Understanding these experiences is seen as necessary to ensure provision of proper intervention and care, therefore further research with more representation of different abnormalities is required (Rivard & Mastel-Smith, 2014).

Only one study included in this review explicitly specified fathers of children with CCN as part of its inclusion criteria. While children with CCN were represented in several other studies, their presence was discernible only through narrative accounts of fathers' experiences within those individual studies, making the experience of fathers specifically with CACCN difficult to decipher. The vast nature of CA means that not all children with a diagnosis will have CCN, even within the most common CA Trisomy 21 children can have a range and varying degree of complexities. Trisomy 21 can result in increased risks of cardiovascular complications, mild to moderate intellectual disabilities, epilepsy, endocrine abnormalities, obesity, physical disabilities, digestive problems, celiac disease, and mental health difficulties (Visootsak et al., 2007; Mégarbané et al., 2013; Benhaourech et al., 2015; Foerste et al., 2016; Pavlovic et al., 2017; Whooten et al., 2018; Altuna et al., 2023), yet people with Trisomy 21 may also lead active, healthy and independent lives (Down syndrome Ireland, 2023).

The psychological impact of caring for a child with a CACCN, as well as their roles and responsibilities in their children's care significantly impacts fathers in this review. Yet, it is not known if this experience differs if a wider range of CA are included. When Ricci & Hodapp (2003) compared Trisomy 21 with other intellectual disabilities it was reported

that fathers of children with Trisomy 21 expressed less child-related stress, particularly in the areas of acceptability, adaptability and demandingness. This study hypothesised that this could be due to the positive perceptions parents have of Trisomy 21. Similarly, Sheldon et al. (2021) reported that the “enjoyable personality characteristics” which children with Trisomy 21 bring, have positive perceptions of fathers parenting. There is a term named the “Down syndrome advantage” which is a concept that children with Trisomy 21 are easier to care for, compared to children with other developmental disabilities (Corrice & Glidden, 2009; Mitchell et al., 2015). However, other research shows that this “advantage” can also be determined by children with Trisomy 21’s level of adaptive behaviour (Masters Glidden et al., 2014). Therefore, children with different levels of complexity may change their fathers experiences, and this is particularly important given that the life expectancy of children with Trisomy 21 is increasing. Globally deaths of children and adolescents with Trisomy 21 have decreased by 22.8% over the past 42 years (Ye et al., 2025), but this means at times children are living with increasingly more complex needs.

This scoping review highlights the relationships between fathers and professionals that have both a positive and negative impact on fathers. Some fathers reported professionals to be inappropriate, unsupportive and lacked knowledge in their children’s conditions. Again, this is predominantly with regards to Trisomy 21, this being the most common CA. Research is needed to determine what the relationship with professions is like, when more rare conditions are represented. Research shows the importance of having services with individual support, with professionals who have specialist knowledge (Brenner et al., 2021). Service provision of children with a CACCN is vital and additional research will allow an evidence base to determine if greater support is required and if professionals require further education and training.

Fathers in this scoping review highlight that they still feel significant inequalities between how professionals treat them, when compared to the way mothers of their children are treated. This discrepancy is also apparent in the level of research available pertaining to fathers. Research shows that there are unique differences between fathers and mothers, and fathers' contributions to child development are often independent

from the contributions of the mothers (Cabrera et al., 2018). However, within much of the literature on parents of children with CACCN, research is predominately focused on mothers (Whiting, 2013; Thunberg et al., 2016; Thomson et al., 2016; Caples et al., 2018; Currie & Szabo, 2020; Page et al., 2020).

3.7 Literature gap

It is clear from the results that there is a significant lack of empirical evidence on the experiences on this cohort of fathers as most studies only focus on Trisomy 21, or Trisomy 21 is included within intellectual disability research. Although it is evident that there has been an effort to include fathers in child healthcare research, their experiences are often conflated with the mothers, despite these results showing they often have unique experiences in fathering a child with a disability. Secondly, when looking at the inclusion criteria of the thirty-seven included studies, the results predominantly focus on fathers of children with an intellectual disability, or Trisomy 21. Within these studies, the complexities of the children are not well identified. Only one study specifically looked at fathers' experiences of caring for a child with CCN, however, was not specifically aimed at children with CACCN. No research specifically exploring fathers' experiences of caring for a child with CACCN was found at the time this search was conducted.

Despite the limited number of studies on this topic some common themes were identified which give an insight into father's experiences. Yet across all three themes and subthemes the gap in these father's experiences is evident. The first theme highlights the overall experience of receiving the diagnoses, which includes receiving the news, the emotional turmoil that this fathers face, and how they came to terms with their children's diagnoses. Yet, this understanding is limited to the fact that this experience included a range of disabilities such as intellectual disabilities (which always included CA but was never the major aim) or specifically were specifically related to Trisomy 21. There is a need to explore if fathers of children with less common CA have the same experiences during the diagnosis stage. Furthermore, within this review, the results are prominently related to the fact that their child would have an intellectual disability, not the CCN associated with this disability. The discussion section shows the

importance in understanding this experience in supporting families of children with disabilities during a diagnosis, and this review shows a significant gap in knowledge for these fathers.

The second subtheme discussed the impact of caring for their children has on fathers, both the psychological impact and their roles in their child's care. This highlights a gap related to a cohort of fathers. Although they highlight some of the challenges faced in their caring roles, for example, being at an increased risk of depression and anxiety, the psychological impact of these experiences, if their child has a diagnosis of a CA and definite CCN is not well explored. Developing a deeper understanding of fathers' experiences if their children have more complex needs than originally anticipated is needed.

The final theme and subtheme relate to the support fathers receive from their family members, peer support from other families of children with similar conditions and the support they receive from professionals. Yet, the support that fathers need may vary depending on their child's diagnosis due to the unique characteristics that their children's conditions present. For instance, a child with Trisomy 21, may or may not have CCN. Furthermore, specific CAs such as Trisomy 21 may have more established support networks and services, while other rare CAs are less understood, making it harder to find appropriate support due to the rare nature of such conditions. This experience needs to be further explored.

3.8 Limitations

This scoping review employed precise, transparent methods based on study and reporting guidelines (Arksey & O'Malley 2005; Levac et al., 2010). However, some limitations have been identified. Firstly, due to the infinite number of CAs, the various syndromes and conditions associated with them, there was a risk that some abnormalities may not have been included in the search terms, resulting in some studies not being included. Additionally, the language that is used in relation to CCN is challenging and there is a potential for ambiguity within the research terms (Brenner et al., 2018), thus resulting in potential studies not being included.

3.9 Conclusion

This chapter presented a scoping review of the available evidence of the experiences of fathers' caring for a child with a CACCN. This review highlights that caring for these children presents a unique experience and fathers feel a range of emotions such as additional fear, grief and pride in caring for their children. How and when a diagnosis is presented can have a significant impact in how fathers cope with their children's diagnosis.

Fathers of children with a CACCN have additional caring roles within the family unit and view their roles and responsibilities to be equally as important to those of the children's mothers. Fathers need support from family, friends and professions on a regular basis. Some fathers documented the lack of knowledge from professionals as a challenge in caring for their children. Understanding the experiences of these fathers is necessary to ensure provision of suitable intervention and care. Findings from this scoping review highlight the need for more research on fathers' experiences of caring for a child with a CACCN. In relation to the aim of exploring what CA are represented within the literature, a wider variety of CAs need to be included to ensure adequate support is provided to fathers of children with other CAs not just specifically Trisomy 21.

Finally, the concept of a child having a diagnosis of a CA as well as CCN is not well reported. There are currently no studies where the inclusion criteria specifically explore fathers' experiences of caring for a child with a CACCN. Although in many studies, the complex nature of the children's conditions are discussed. Further research on this unique experience is needed to develop our understanding. This could potentially create the opportunity to tailor supports and services for fathers, whose children have other often rarer conditions. Based on these conclusions, undertaking a study exploring the experience of fathers caring for a child with a CACCN is justified.

The next chapter will discuss the methodological underpinnings of the study. Hermeneutic phenomenology guided by van Manen is the approach chosen to support the exploration of fathers' experiences of caring for a child with a CACCN. Therefore, the

next chapter will explore the different types of research in order to provide context on how this methodology was selected.

4 Methodological Underpinning of the Study

4.1 Introduction

Chapter four introduces the methodological framework which guided this study and the rationale for why this methodological framework was selected. An overview of the philosophical issues are presented in this chapter, with a description of the philosophical underpinnings of this study. Hermeneutic phenomenology guided by van Manen is the approach chosen to support the exploration of fathers' experiences of caring for a child with a CACCN. Therefore, this chapter will explore the different types of research in order to provide context on how this methodology was selected. A discussion as to what phenomenology is, and why specifically hermeneutic phenomenology was the chosen methodology is presented. In order to rationalise why this was chosen, a historical overview of hermeneutic phenomenology is offered. More specifically, the principles of van Manen's (2014) hermeneutic phenomenology are explored to explain how this can support the exploration of the lived experience of fathers caring for a child with a CACCN.

4.2 Rationale for adopting qualitative research

The aim of this PhD study is to seek a greater understanding of the experiences of fathers who have a child with a CACCN by interpreting their views of their everyday life. The methodological underpinnings of this study must therefore enable the researcher to develop a complete approach to answer the research question, specifically exploring the experiences of fathers of a child with a chromosomal abnormality (Polit & Beck, 2018). The technique, principles, and procedures by which a researcher understands a phenomenon and finds answers to particular questions is referred to as methodology (Bogdan & Taylor, 1975). The term methodology refers to the general approach to researching a topic, whereas the method refers to the specific technique(s) used, and this will be addressed here in chapter four. The first decision that was made when contemplating the methodology to explore these fathers' experiences, was determining what type of research would best help answer the research question that had emerged from the literature, as previously discussed in chapter two. Research is characterised as

quantitative or qualitative in nature (Rutberg & Bouikidis, 2018) and the decision on whether qualitative or quantitative data would be most appropriate was made. The two research paradigms aim to seek a greater understanding of a phenomenon, but one of the distinguishing factors between qualitative and quantitative approaches is the nature of the intended outcome or findings. The aim of qualitative research is to explore the meaning of the peoples' experiences, cultures, or how people view particular experiences. Qualitative research is a multi-layered approach that examines culture, society, perceptions, experiences and behaviour through analysing people's words and actions to understand the social reality of individuals by developing non-numeric data (Parahoo 2024). Qualitative research, in general, is a strategy that allows researchers to investigate people's experiences in depth utilising a specific set of research methods such as interviews, focus groups, and observations (Hennink et al., 2020). This data enables a researcher to identify difficulties from the viewpoint of study participants, as well as to comprehend the meaning that they place on behaviour, events, or objects (Hennink et al., 2020). In using a qualitative approach, the researcher can strive toward thoughtful reflection that is "free from theoretical, prejudicial and suppositional intoxications" (van Manen 2007, pp. 12).

The aim of quantitative research on the other hand is to examine the relationship between variables, such as the dependent, independent variables, and extraneous and is based on numerical data (Creswell, 2009). Quantitative research uses a rigorous and controlled design to explore a phenomenon using a precise set of measurements (Polit & Beck, 2024). Although quantitative data also seeks to find patterns in data, it is used to quantify experiences. The aim of this study is not to quantify these fathers' experiences; therefore, a qualitative methodological approach is utilised for this study to answer questions regarding experience, meaning, and perspectives from the participant's perspective as it enables information rich, contextual data for this complex group of people.

Within qualitative research, there are multiple different approaches to produce nonnumeric data. Creswell (2016) describes these qualitative inquiry traditions as, a case study, a narrative research, phenomenology, grounded theory, ethnography.

Individually, each approach may help provide insight into the experiences of fathers of children with a CACCN, however, the most appropriate method had to be selected which resulted in careful consideration of each approach. A narrative research study would report a story about the personal experiences of an individual. Although, a narrative inquiry would be appropriate, as it could be used to tell the story of the lives of the fathers of children with a CACCN, finding a deeper understanding of the context of these fathers' lives is the aim of this study and therefore a narrative enquiry was not selected. Ethnography is one of the early qualitative approaches and is largely aimed at learning about people, in contrast to studying people, through immersion in native populations (Huot, 2014; Spradley, 1980). Typically, ethnography is described as in-depth observations of groups of individuals, in relation to influences of historical and cultural contexts on social interactions (Streubert & Carpenter, 2011), and as this was not the aim of this research, ethnography was not deemed the most appropriate method. Grounded theory involves collecting and analysing data in order to develop new theories on a particular phenomenon. The aim of this research is not to create a theory of this phenomenon, but allow fathers voices to be heard, and therefore grounded theory was also not selected. The study of phenomenology has its roots in philosophy and has been studied in various forms through the years (Carpenter, 2011). Aside from being a philosophy, phenomenology is a qualitative research approach and methodology (Burns & Grove, 2020; Dowling, 2007; Heinonen, 2015). Phenomenology aims to describe the essence of a phenomenon by looking at it through the eyes of individuals who have experienced it, within the world that they live in (Neubauer et al., 2019; Teherani et al., 2015). Like other concepts in social theory, phenomenology is a broad term and covers varied traditions and therefore, there are numerous philosophical traditions within phenomenology that are continuously being reinvented and added to (Carpenter, 2011).

Within nursing and healthcare research, phenomenology is a popular approach to undertaking research. The philosophy of phenomenology resides within the naturalistic paradigm; which attempts to explore, what a particular experience is like, what does a particular experience mean and how does the lived world present itself to the participant or to me as the researcher (Rodriguez & Smith, 2018). Phenomenology

seeks to understand the outside world as it is interpreted by the human consciousness. Phenomenology is an appropriate, powerful method of healthcare research, enabling healthcare professionals to gain deeper understanding of the experiences of other people (Neubauer et al., 2019).

A central part of the nursing profession is the holistic caring, advocating and support for the bio-psychosocial and spiritual well-being of individuals, families, and communities (Moxham & Patterson, 2017). The profession requires people to want to understand how the people in their care experience their health in order to give them the best possible standard of care (Berkowitz, 2016). Phenomenological research within the nursing profession reflects the nursing values, because, like nursing, it considers the whole person and values their experience (Morell-Scott, 2022). This study, focusing on the experiences of fathers, adopts a phenomenological approach, offering a way to begin to understand the ranges of factors that can affect parents caring for children with CCN from their perspective and experience, revealing meanings that can be 'hidden', rather than making inferences (Rodriguez & Smith, 2018). This knowledge has the ability to enhance the quality of holistic patient care (Patton, 2020). The phenomenology research design is distinguished by its concentration on a common phenomenon among a group of people, as the name implies (van Manen, 2016). Therefore, fathers of children with a CACCN share a common phenomenon: the human experience of being a father of children with complex conditions. More specifically, this study adopts a hermeneutic phenomenology approach. However, to truly understand the development of hermeneutic phenomenology, a full exploration of its origin is needed. Different philosophers have given their views on what phenomenology is, and how it is carried out. Researchers need to understand the philosophical underpinning of phenomenology and then choose appropriate methodological approaches to assist them in answering the research question (Dowling & Cooney, 2012) and to really comprehend hermeneutic phenomenology, one must first gain an understanding of the philosophies that underpin it (Neubauer et al., 2019).

4.3 A historical viewpoint of phenomenology

According to Barbera & Inciarte (2012), phenomenology is derived from the Greek word *phenomenon*, which means "anything that presents itself and can become visible by itself" (p. 201). Phenomenology is literally the study of "phenomena": the ways in which things seem to us, how we perceive them, and the meanings they have for us. Modern understanding of phenomenology was first explored in the early 20th century through the work of Edward Husserl (1859–1938), who sought to make philosophy "a rigorous science" and expanded upon later by scholars of his work including Heidegger and Merleau-Ponty (Gadamer, 1975, 1998; Heidegger, 1962; Merleau-Ponty, 1962). This originally started in the German language, and attempted to place philosophy to a scientific background, thus making it an empirical philosophy of science.

There are two main approaches to phenomenology: descriptive and interpretive (Sloan & Brain, 2014). Phenomenology has developed from descriptive phenomenology, which emphasises the 'pure' description of people's experiences, to the 'interpretation' of such experiences, as in hermeneutic phenomenology (Matua & Van Der Wal, 2015). Edmund Husserl developed descriptive phenomenology, while Martin Heidegger developed interpretive phenomenology (Husserl 1859-1938; Heidegger 1962). Each of these approaches produces information that reflects insights into the meaning of the phenomena under investigation.

In both interpretative and descriptive approaches, the researcher expects a willingness to listen to the participants' explanations of their lived experiences (Penner, 2008). According to Matua & Van Der Wal (2015), the descriptive approach is employed to shed light on parts of experiences that are not well known. The interpretive approach in contrast is used to analyse and contextualise aspects of an experience in relation to other influences such as culture, gender, employment or wellness of individuals or groups experiencing this occurrence.

4.4 Descriptive phenomenology

Phenomenology as we know it today originated as descriptive phenomenology, which emphasises the 'pure' description of people's experience. The historical viewpoint can

be used to highlight that there are various different ways to do phenomenology research that are embedded with different ways of conceiving the what and how of the human experience by adopting different philosophies.

4.4.1 Edmund Husserl

Edmund Husserl (1859-1938), was a German mathematician who developed what is known as Husserl's descriptive phenomenology, also known as transcendental phenomenology (Spinelli, 2005). Husserl developed phenomenology as an alternative to the empirically based positivist paradigm that was prominent during this time (Nelms, 2015). Husserl presented phenomenology as a philosophy and a way to comprehend human experience with the publication of his book *Logical Investigations* at the turn of the twentieth century (Husserl, 1913). This was because he believed that most people went about their daily lives without critically reflecting on their experiences, and that a scientific method was required to bring out the fundamental components of a group of people's lived experiences (Husserl, 1913). Husserl believed that phenomenology suspended all suppositions, was related to consciousness, and was based on the meaning of the individual's experiences (Creswell, 1994). The researcher does not make any interpretations using the descriptive method. Rather, they analyse participant descriptions and split them into meaning-laden statements, collecting the meanings that are critical to the construct of the phenomenon under investigation. As a result, the researcher can describe the structure of the phenomenon of interest in writing. This meant that there was an objectification of the meanings of human experiences (Smith et al., 2009). Husserl's aim for phenomenology was to capture experience in its primordial origin or essence, without interpreting, explaining, or theorizing.

Husserl valued rigor and attempted to mimic these characteristics through bracketing, also referred to as *epoché* (McConnell-Henry et al., 2009). Husserl (1952/1980) criticised psychology as a science that had gone wrong by attempting to apply methods of the natural sciences to human issues. Husserl first proposed bracketing in 1913, to help better understand other people's experiences. Bracketing requires the researcher to put aside their assumptions, including their scientific, philosophical, and cultural views

(McConnell-Henry et al., 2009). Husserl thought that this would provide objectivity to the research reducing bias (Koch, 1995; McConnell-Henry et al., 2009).

4.5 Interpretative/ hermeneutic phenomenology

Interpretative phenomenology developed when more philosophers added to Husserl's work by not just describing the results but also the 'interpretation' of such experiences. Hermeneutic phenomenology, also known as interpretive phenomenology, originates from the work of Martin Heidegger (1962) whose philosophical inquiry began in alignment with Husserl's original work, but later challenged several key elements of Husserl's transcendental phenomenology (Neubauer et al., 2019). The word hermeneutics comes from the Greek verb *hermeneuein*, which means "to interpret or decipher." (Cohen et al., 2000). The term derives from Hermes, who was a wing-footed messenger-God associated with the Delphic oracle, who was accountable for changing the unknowable to a form that humans could understand via language and writing. Comprehension can take many forms, including language, text, interpersonal interactions, personal experiences with oneself and society, and linguistic, religious, and artistic thoughts (Gadamer, 1976). Hermeneutics is a theory of interpretation that was originally used to translate and interpret Biblical texts (Crotty, 1998). Hermeneutics has been around since the 17th century. It ventured into a variety of fields of study and applied to texts other than the Bible, such as unpublished sources (McManus Holroyd, 2007). This is now used widely by researchers to understand lived experiences. This methodology elicits stories from participants as a source of understanding (Alan et al., 1986; Gyollai et al., 2020; Heidegger, 1962; Richardson et al., 1999). In general terms, hermeneutic phenomenology is the art and science of interpretation or meaning.

Hermeneutic phenomenology is a useful research method because it allows researchers to describe and articulate how people's ordinary, everyday practices and routines are shaped by their family relationships, childhood experiences and day-to-day challenges (Alan et al., 1986; Gyollai et al., 2020; Heidegger, 1962; Oerther, 2020; Richardson et al., 1999). This type of research aims to explain a phenomenon as it presents itself to the consciousness of the participant, and to return to the "things themselves" as they are initially presented to us (Husserl, 1970).

Hermeneutic phenomenology has been formed by the contributions of numerous philosophers and continues to evolve as a tool for qualitative research (McConnell-Henry et al., 2009). Different styles of hermeneutic phenomenology share a common interest in empirically understanding the nature of human experience but draw on foundational philosophies in different ways (Suddick et al., 2020). Heidegger, Sartre, and Merleau-Ponty are probably among the best-known originators, from whom van Manen has developed his ideals, although their work is not always easy to read and comprehend. Still, their original foundational writings offer fundamental insights which appear inexhaustible in their philosophical significance for those seriously interested in phenomenology, thus extensive exploration of their work is required, in order to fully appreciate how hermeneutic phenomenology guided by van Manen has developed.

4.5.1 Martin Heidegger

Martin Heidegger was also a German philosopher and was a student of Husserl's. Heidegger is a philosopher who built upon the foundation laid by Husserl by transforming the idea of intentional consciousness of the world into the concept of 'being in the world' (Heidegger, 2007). Like Husserl's phenomenology, this is concerned with human experience as it is lived. While Heidegger's philosophical investigation started in alignment with Husserl's views, he later challenged several key areas of Husserl's phenomenology. Heidegger developed hermeneutic phenomenology by extending hermeneutics through studying the concept of being in the world, rather than knowing or understanding the world. Which emphasises the importance of interpreting and understanding human experience from an ontological stance rather than merely describing (Polit & Beck, 2024).

In 1927 Heidegger released a text "Being and Time" which started a radical movement in the field. Being and Time is long and complex, which means readers often find it challenging, particularly when translated into English. However, he firmly opposed intentionality and favoured existentialism, which he dubbed "*dasein*", which translates to "being in the world." (Heidegger, 1958, 1962). He is one of the first philosophers to combine existential matters with phenomenological methodology and was influenced

by the Danish philosopher Soren Kierkegaard (1813-1855) who is generally regarded as the founder of existential philosophy (Valle et al., 1989).

Heidegger rejected the theory of knowledge known as epistemology, and adopted ontology, the science of being. With this focus on human experience and how it is lived, hermeneutic phenomenology moves away from Husserl's focus on 'acts of attending, perceiving, recalling and thinking about the world' (Lavery, 2003). Thus, expanding hermeneutics into the philosophy of interpretation, to create interpretive phenomenology (Creswell, 1994). This means that instead of focusing on how we know what we know, Heidegger believed in exploring what it means to live in a world that is experienced by everyone in their own unique manner which is both interpretable and relational. Therefore, using '*dasein*' is neither subjective nor objective.

Heidegger disagreed with Husserl on many counts, arguing that there is no absolute truth and, accordingly, that context must be considered in order to shape understanding (Holroyd, 2007; McConnell-Henry et al., 2009). Heidegger believed that human beings are actors in the world and so focused on the relationship between an individual and his/her lifeworld (Lopez & Willis, 2004). Heidegger's term lifeworld referred to the idea that 'individuals' realities are invariably influenced by the world in which they live. Heidegger recognised the researcher as a critical part of a research study and thus rejected Husserl's use of bracketing (McConnell-Henry et al., 2009). Heidegger believed that one can never bracket one's thoughts and that one's pre-understanding cannot be avoided in the development of understanding (Keller, 1999).

Distinct from the natural sciences, where a whole entity is quantifiable by the sum of each component, the holistic philosophical approach accepts that a sum of the parts does not necessarily equal the whole. Each individual part must be considered in its own context. A word on its own may have many meanings until given the context of a sentence. A sentence, similarly, can have multiple meanings depending on the context of the preceding sentence or paragraph or the following sentences for that matter. It is equally true the other way around, that a word can change the meaning of a sentence which in turn may change one's understanding of a paragraph. This idea of envisioning

a whole in terms of how the parts interact with each other, and how those parts interact with the whole is referred to as the hermeneutic cycle/circle, first proposed by Heidegger in 1927 in his book "Being and Time". From a phenomenological standpoint, Heidegger proposes hermeneutical phenomenology as a method of understanding being, through assessment of a phenomena of one's experiences. One can begin to form an understanding of being in the world, with the understanding that those experiences influence perception, reactions, and the behaviours all resulting from an experience and similarly behaviours, reactions and perceptions give context to each experienced phenomenon.

3.5.2 Merleau-Ponty (1958)

Maurice Jean Jacques Merleau-Ponty (1908–1961), was a French philosopher who also has an interest in phenomenology and was greatly influenced by Husserl and Heidegger. Merleau-Ponty aimed to redefine hermeneutic phenomenology and claimed that phenomenology as it was at the time had several inconsistencies, such as its endeavour to construct a rigorous science-based philosophy while also providing an account of space, time, and the world as humans perceive them (Merleau-Ponty, 1945). Merleau-Ponty developed phenomenology in his own direction and felt it required modernisation. He wanted to take phenomenology towards rediscovering first experiences. Merleau-Ponty focused on the problems of perception and embodiment as a starting point for clarifying the relation between the mind and the body, the objective world and the experienced world. Different from Husserl and Heidegger, Merleau-Ponty analysed some experimental psychology, he explored the concept of how amputees could feel sensation in their amputated limb and focused on rational construction of the world in the mind. In this way, Merleau-Ponty developed a rich version/form of phenomenology emphasising the role of the body in human experience.

Merleau-Ponty uses spatiality and orientation to explicate the notion of the human subject as being-in-the-world. Our world is not only scientific, but it is emotional and aesthetic (among other things). Merleau-Ponty (1997) developed four life-world existentials that are implicit to understanding lived experience: temporality (lived time),

spatiality (lived space), corporeality (lived body) and sociality (lived relationships). These life-worlds were of particular interest to van Manen as he took inspiration from these concepts and added an additional one 'lived things' (materiality). Merleau-Ponty was of huge influence on van Manen, and the ideas of the four life-worlds developed by Merleau-Ponty existentials are vital to understand how van Manen has developed his ideas on hermeneutic phenomenology. Below are the four lifeworld existentials first described by Merleau-Ponty and van Manen's agreement of them, and how they guide this study.

4.5.2 Temporality (lived time)

The existential of lived time can be understood as time as we experience it (Merleau-Ponty, 1962, 2012). This is composed of a subjective understanding of time as opposed to the more objective or "factual" time, and it refers to the ways in which we experience our world on a temporal level (Merleau-Ponty, 1962, 2012). The dimensions of past, present, and future are interconnected and have an accumulated effect on our lives becoming "a person's temporal landscape" (van Manen 1990, p.104). Temporality is subjective time as opposed to clock time. It is the way time appears to change depending on the situation we find ourselves in. The temporal things of the past determine the present and the future perceptions of the world. For example, past experiences such as trauma, culture and spiritual beliefs can affect the way one interprets an experience. The temporal things of the past determine the present and the future perceptions of the world. This is of note when exploring fathers' experiences of caring for a child with a CA, to determine if past experiences affect how they experience currently caring for their child.

4.5.3 Spatiality (lived space)

Spatiality in phenomenology is defined as felt space (Merleau-Ponty, 1962, 2012). Felt space affects our experience of the world. Van Manen believes that our lived space shapes how we experience our lives, he refers to lived space as felt space. It enables us to comprehend the world in which we live and how it affects our lives. Space is distinct in that it surrounds us, can be objective or subjective, and is influenced by mood, emotions, and geography. We each have our own special place(s) where we feel at ease

(van Manen, 1990 p.104). Exploring how fathers determine their lived space enabled the researcher to delve into how fathers experience time. This was important when exploring the space in which fathers of children with a CACCN find themselves, for example while in hospital/appointments, but equally can include the mental space needed to care for their children.

4.5.4 Corporeality (lived body)

Human beings are always “bodily in the world”. The lived body is our pivot of existence, thus Merleau-Ponty (1962, 2012) explained this as how people experience the world through their bodies. From the phenomenological perspective, corporeality means that we are always bodily in the world (Fredriksen, 2007; Tembo, 2016; van Manen, 1990). When an individual meets another in his or her lifeworld, they are introduced to each other first in a physical sense. As they interact, this first impression can grow and change. This was interesting to the researcher, to reflect upon how the body is present in conversations relating to fathers caring for their children.

4.5.5 Sociality (lived relationships)

According to both Merleau-Ponty (1962, 2012) and van Manen (1990), lived relationships are “the lived relations we maintain with others in the interpersonal space that we share with them” (van Manen, 1990, p. 10). How fathers experience their relationships around them, both with their children, family members, peers and professionals, enabled the exploration of this phenomenon of how these fathers care for their children in the context of their relationships.

4.5.6 Hans-Georg Gadamer

Hans-Georg Gadamer (1900-2002) was another German philosopher who had a special interest in hermeneutic phenomenology, whose ideas were connected with Greek philosophers. Gadamer aimed to develop historical hermeneutics and therefore grounded his own hermeneutics in the history of hermeneutic practice. Gadamer’s ideas were originally influenced by Martin Heidegger who exerted the greatest and enduring effect on Gadamer’s philosophical development. Gadamer, first met Heidegger before Heidegger was a professor and followed him to become one of his students and later a

colleague. The influence of Heidegger, is seen in his early work, called 'On the Idea of System in Philosophy' (1924), and 'Plato' (1931). However, Gadamer's insights into hermeneutics for the twentieth century were also influenced by some other phenomenological thinkers such as Husserl but also noted a shift to include the observer or researcher in the research. Gadamer produced his major work "Wahrheit und Methode: Grundzüge einer philosophischen Hermeneutik" (1960) which was then translated into English in 1975 (without the important subtitle) as Truth and Method. This is arguably one of the major 20th-century philosophical statements on hermeneutical theory, despite the evolution of his thought later in his life, this remains the most systematic account of his philosophical project. Today, Gadamer is recognised as one of the preeminent voices for philosophical hermeneutics.

Gadamer agreed with Heidegger that interpretation is impossible without our preconceptions; we cannot "extinct" ourselves when listening to someone or reading a text. However, according to Gadamer, it is critical to remain conscious of our own prejudices, to emphasise them, and to continuously examine the validity of our prior knowledge in light of new information (Gadamer, 2004). Understanding the historical contexts of text and reader, as well as the ways in which these interact to form a temporary shared meaning, is central to Gadamer's hermeneutical approach (Austgard, 2012). Gadamer argues that to understand a phenomenon, this is always dialogical. In other words, we begin to understand a phenomenon through conversations with others. Gadamer looked at how we rely on our innate foreknowledge and worldview while also opening ourselves up to alternate interpretations by others, which he called "*fusion of horizons*". When applying hermeneutics to the human process of interpretation Gadamer talks of a '*horizon*' as a way to conceptualise understanding (Clarke, 2008). He explains the concept of horizon suggests itself because it expresses the superior breadth of vision that the person who is trying to understand must have. To acquire a horizon means that one learns to look beyond what is close at hand - not in order to look away from it but to see it better (Clarke, 2008).

There are several concepts to explore on the way to developing our horizon as detailed by Gadamer. These are: pre-understanding, prejudices, fore conceptions, 'Bildung' or

openness to meaning, language, and imagination. Firstly, we come into an encounter with preconceived notions or an idea of a situation (Gadamer, 1966). Gadamer refers to this as 'prejudices' not in a negative sense, but: a judgement that is rendered before all elements that determine a situation have been finally examined (Gadamer, 2004). He explains that we have fore-conceptions (openness to meanings) that unless an interaction is entirely inexplicable, we always assume it has meaning. We are always looking ahead to gain a deeper knowledge. This occurs before we make our final decision on a phenomenon. We must remain open to allow us to move onto the next stage of understanding (Gadamer, 2004). To develop an understanding of a phenomenon that the past and present cannot exist without each other, and understanding is always the fusion of these horizons supposedly existing by themselves (Gadamer 2004). Gadamer identifies language as the medium for understanding and a means of sharing the complexities of human experience (Gadamer, 2004a). Suggesting that understanding was interpretation and vice versa (Gadamer, 2004a). For Gadamer, interpretation is fundamentally dialogic: the metaphor of an ongoing conversation is therefore extremely important. Gadamer states that 'understanding itself has a fundamental connection with language (Gadamer, 2004a). Gadamer does not offer the researcher a method or a set of rules to follow (Austgard, 2012) but more a philosophical stance.

4.6 Max Van Manen

Max van Manen is a Dutch-born Canadian scholar whose expertise lie in phenomenological research, and his ideas around hermeneutic phenomenology will be used to guide this study. The philosophical underpinnings of phenomenology according to van Manen involve both descriptive and interpretive hermeneutic phenomenology (van Manen, 2014). Van Manen (1990, 1997) describes methods for healthcare professionals to explore meaning in certain aspects of life; which allows the researcher to understand links between meaning, language and the world in which this meaning exists. Van Manen explains that to conduct phenomenological research it is consequently to investigate what an experience is like (1990, 2017) and suggests the basic phenomenological question is the question of "What is that experience like?" (van Manen, 2014 p. 35). The scoping review highlighted the need to explore what the

experiences of having a child with a CACCN is like, to add to the empirical evidence sounding their experiences.

Van Manen (2014) renamed this phenomenological method "*phenomenology of practice*," referring to inquiries that target and assist professional practitioners' practices as well as daily practices. Although his contribution to phenomenology appears to fall under the heading of new phenomenology, it also appears that it does not, as his writings combine Husserl's descriptive phenomenology with an emphasis on the study of the world before reflection and it also argue that it is scientific, while also asserting that it involves interpretation. As said by van Manen (2014) "More specifically, this phenomenology of practice is meant to refer to the practice of phenomenological research and writing that reflects on and in practice and prepares for practice" (van Manen 2014, p. 15).

As discussed above, Husserl (1936, 1970) introduced the concept of the lifeworld in which he explained the lifeworld as pre-reflective, in which our focus is on what we are perceiving rather than how we are perceiving it. Gadamer (1975) further expanded this concept explaining the lifeworld as something embedded in discourse, explaining that the world we live in is verbal in nature. Merleau-Ponty (1997) developed four life-world existentials that are implicit to understanding lived experience: temporality (lived time), spatiality (lived space), corporeality (lived body) and sociality (lived relationships). Van Manen argues that this lived experience needs to be explored across the whole range of modalities (van Manen, 1990). He was particularly influenced by Merleau-Ponty's concept of the fundamental life-worlds perspectives. Yet went further to expand these concepts and added a fifth, materiality (Lived things) (van Manen, 2014). In this study, the lifeworld existential offers a lens through which to explore fathers' lived experiences of caring for their child with a CACCN. Van Manen explains they are the "universal themes of life" (van Manen, 2014, p. 302). These existentials allow researchers access the lived experience of human beings and develop a deeper understanding of the nature and meaning of everyday experience such as caring for a child with a CACCN.

The idea of the lifeworld existentials offers a lens through which to explore lived experience such as fathers' experiences of caring for their child with a CACCN. Van Manen himself says that these are the "universal themes of life" (van Manen, 2014, p. 302). These existentials allow researchers access the lived experience of human beings and develop a deeper understanding of the nature and meaning of everyday experience such as caring for a child with a CACCN.

4.6.1 Lived things (materiality)

In addition to the earlier identified existentials, more recently, van Manen (2014) introduced the fifth existential 'lived things' or materiality. This relates to how people feel in relation to the topic being studied, emphasising the relevance of material objects and how they might aid us in gaining particular insights. This is interesting within the concept of this study, as by exploring this aspect of a father's life, it was possible to see what "things" influence fathers' perspectives of caring for their children. The lived experience is the focal point of phenomenological research and "transforming lived experience into a textual expression of its essence, - in such a way that the effect of the text is at once a reflective re-living and a reflective appropriation of something meaningful: a notion by which a reader is powerfully animated in their own lived experience" (van Manen, 1997/1990, p. 36).

4.7 Hermeneutic Cycle

The concept of the hermeneutic cycle was first established by Heidegger in 1927 in his book "Being and Time". The hermeneutical circle is one of the most fundamental aspects of hermeneutical theory (Grondin, 2015). This idea of envisioning a whole in terms of how the parts interact with each other, and how those parts interact with the whole, is the concept of the hermeneutic cycle. This means that the concept of the hermeneutic circle states that our comprehension of a text as a whole is dependent upon our comprehension of each component as well as our comprehension of how each component part pertains to the entire text or body of research. In other words, that understanding is in a circular movement. In adopting van Manen's (1990) approach to hermeneutic phenomenology, the hermeneutic cycle is a fundamental aspect to developing an entire understanding of the experiences of fathers caring for a child with

a CACCN. Following the six guidelines developed by van Manen (discussed further below), it allows the researcher to delve deeply into this phenomenon, by visiting and revisiting the data collected in a circular action, by reflecting on individual aspects of the text collected and reflecting on it as a whole. Furthermore, the existentials also illustrate a *fusion* of the objectivist hermeneutic circle (part-whole) and the alethic hermeneutic circle (pre-understanding), as they acknowledge the experience of a phenomenon in a whole experience, and also the researcher's role in the research process (van Manen, 1990).

4.8 Placing van Manen in the context of this study

Van Manen argues that to do phenomenological research, you are attempting to investigate what an experience is like (van Manen, 1990, p. 42; 2017a, p. 776). As the aim of this study is to explore fathers' experiences of caring for their child with a CACCN, hermeneutic phenomenology fits well to support this enquiry. According to van Manen, hermeneutic phenomenology is a human science that enables us to study a person's world as they live it, seeking to obtain a more complete understanding of our experiences and, consequently, what it means, in more depth, to live in the world (van Manen, 2015). This phenomenological study is hermeneutical in the sense that it does not aim exclusively to describe how the experience is given to consciousness, but seeks to find what the interviewees can reveal towards the studied phenomenon. Hermeneutic phenomenology is a good way to capture these lived experiences as it does not claim to solve problems (van Manen, 1990) but is helpful to gain a rich description that may resonate with others.

Although the concept of the lifeworld existentials is offering a method to explore these fathers' experiences, van Manen highlights challenges of phenomenological research, noting that the term phenomenology occurs in a confusing abundance, arguing the importance of not presenting the methodology of phenomenology in an overly technical manner (van Manen, 2017). Van Manen aims to make phenomenological research "doable by researchers who are not themselves professional philosophers" (van Manen, 2014, p. 18). However, there are some critiques to van Manen who argue that although he aspires to make phenomenology accessible, his ideas are certainly not a non-

technical, easily accessible introduction to phenomenological philosophy (Dahivi, 2019). However, van Manen (1997) offers a unique standpoint, as he describes a methodological approach to hermeneutic phenomenological research unlike many earlier writers. Another element significant to van Manen's stance on hermeneutic phenomenology is the concept of not bracketing. Van Manen (1990), does not strictly adhere to bracketing (*epoché*) one's own preunderstandings as used in other phenomenology approaches (e.g. Husserl), instead understands that the researcher is part of the research process. As the author is a nurse working within the CCN field this is an extremely valuable element to the study's methodological approach.

Van Manen (1990, 1997) also describes methods for healthcare professions to explore meaning in certain aspects of life; which allows the researcher to understand links between meaning, language, and the world in which this meaning exists (Van Manen, 2014). Van Manen discusses that there is no step-by-step model that will guarantee phenomenological insights and understandings (van Manen, 2017). However, he does provide six guidelines to carrying out hermeneutic phenomenology that will be utilised in this thesis. The application of the six research activities is portrayed in Table 7.

Table 7: The process of applying Van Manen's guidelines in the context of this study

Activity	Guideline	How this is incorporated in this study
Activity 1.	Turning to a phenomenon that interests the researcher	The development of the research question. When commencing and deciding the appropriate methods. The formulation of the research question: what is the experiences of caring for a child with a CACCN?
Activity 2.	Investigating experiences as its lived rather than how we conceptualise it	This activity is incorporated throughout the data collection process. Using both semi structured and structured interviews to explore and develop a rich understanding of the phenomenon of interest (fathers experiences of caring for a child with a CACCN). Optional journals for fathers to document their experiences.
Activity 3.	Reflecting on the essential themes	This step develops a deep understanding of the phenomenon. Emerging themes are determined by reflecting on the data by replaying the audio recordings, reading and re-reading the journal entries and interview transcripts.
Activity 4.	Describe the phenomenon through steps of writing and rewriting	This activity transforms the lived experiences of fathers caring for a child with a CACCN into a textual expression of its essence, thus giving voices to the fathers of these children.
Activity 5.	Remaining a strong relation to the phenomenon	This stage highlights the importance of remembering and sticking to the research question. This step is represented throughout the data collection, analysing of the data and during the writing of the results.
Activity 6.	Developing a research concept by concept or idea by considering parts and whole.	In this step, the researcher constantly assesses the overall design of the research study.

4.9 Van Manen's guidelines in the context of this study

Van Manen (1990) argues that these steps allow the researcher to reveal the phenomena within the lived experience. Van Manen argues that these guidelines are not a set of strict rules, instead they can be revisited as the enquiry continues (van Manen, 1990). Adopting van Manen's six guidelines, provides structure to this PhD study, while also incorporating the other elements of hermeneutic phenomenology described by van Manen.

4.9.1 Turning to a phenomenon that interests the researcher

The researcher began this study while working with families of children with CCN. Van Manen believes that every project of the phenomenological inquiry is driven by a commitment which turns to an abiding concern (van Manen, 1997). At the start of this study, the researcher began to question and think about what it must be like for a parent to care for a child with a CA. This is where the commencement of the background search began, and a clear research question emerged focussing on fathers' perspectives. A pre-understanding of the phenomena of the experience, influences interpretation and disclose the nature of the lived experience (van Manen, 1997). According to van Manen (1990), enquiries should be direct, specific, and written to stimulate detailed descriptions of the experience. Van Manen's guidelines remind the researcher to keep their '*gaze*'. This means the researcher will keep returning to the research question, to make sure the methodologies being used are appropriate in addressing the research question.

4.9.2 Investigating experience as it is lived rather than how we conceptualize it

This stage is to explore the originality of the lived experience of a phenomenon. This is undertaken by capturing the data through methods of investigation. Phenomenological research focuses on the meaning of lived experience, or "the world as we immediately experience it pre-reflectively rather than as we conceptualize, categorize, or reflect on it" (van Manen 2016, p. 9). In hermeneutic phenomenology van Manen (2016) believes that interviewing has two main purposes: (1) as a means to explore and develop a rich understanding of the phenomenon of interest; (2) to develop a conversation around the meaning of specific experiences. Van Manen (2016) suggests using conversational interviews that are semi-structured in nature, which will rely on the participants' memories and reflections in order to help participants revisit their experiences. The researcher therefore chose to do a series of interviews, the first being unstructured in nature and more conversational in nature, and then subsequent interviews having a more structured approach to collect this data. Additionally, fathers had a choice to complete journal entries if they wished to do so.

4.9.3 Reflecting on the essential themes

By reflecting on the essential themes that are characterised by the phenomenon, this will help the researcher find essential meaning and the essence of the phenomena of the lived experience (van Manen, 1990). Phenomenological research is the way to a thorough understanding of the phenomena of lived experience; reflection is therefore founded in it (Umair et al., 2020). Since structured meanings or themes are regarded as a reflection of an entire lived experience, van Manen emphasises the necessity of reflecting on them (van Manen, 1997). By reflecting on the transcripts from the interviews and the journal entries, it allowed the researcher to extract themes known as essential themes from the father's experience to try to reveal the true meaning of their experiences. The researcher's journal also helped to reflect thoughts and experiences while interviewing the fathers.

4.9.4 Describe the phenomenon through steps of writing and rewriting

The aim of this thesis is to make the experiences of the fathers caring for children with a CACCN visible and to truly document what it is like to care for these children. This requires the author to write a true representation from the verbal text represented in the writing of the thesis. Van Manen expressed that the aim of hermeneutic phenomenology is to transform lived experiences such as caring for a child with a CACCN into a textual expression of its essence, thus giving voices to the fathers of these children. This study also uses these five existentials in the writing and reporting of the findings. How these existentials are implemented will be further discussed in chapter five.

4.9.5 Maintaining a strong and oriented relation

This stage highlights the importance of remembering and staying with the research question. Van Manen (1990) highlights how carrying out phenomenological research can be lengthy, and distractions can happen. In this event, potential bias or the lack of a true representation can be possible. The nature of hermeneutic phenomenological research and the richness of data can easily pull the researcher from the focus of the question and notion of the lived experience. As the researcher attempts to interpret and describe the data, she ensured to document her own feelings and experiences to ensure

that the research can stay focused on the fathers' true experiences. Van Manen also highlights that if a professional has too many "pre-understandings" or beliefs about a phenomenon this can be an issue particularly if the researcher themselves does not acknowledge this (Van Manen, 1990). Therefore, the importance of the journal throughout the interviewing process must be acknowledged. The aim of this was to allow the researcher to undertake a deep reflection of emotions and interpretation of the interviews allowing the research to align with van Manen, supporting his idea that the researcher must identify the role they have to play in the exploration. The researcher frequently went back to this journal throughout the data analysis phase to ensure she was identifying her own personal experiences with each interview and to be self-aware of any potential bias or preconceived ideas.

4.9.6 Balancing research context by considering part as a whole

The data collected through hermeneutic phenomenology can present a large amount of information rich data, which in turn can result in a researcher looking at this data from a superficial standpoint and not delving into the true representation. As discussed above, the hermeneutic circle is a significant aspect of hermeneutic phenomenology guided by van Manen, and the steps of reflection and the art of writing and rewriting is highly significant throughout the hermeneutic process (van Manen, 1990). Hermeneutic phenomenology can be a lengthy process, with the risk of researchers becoming consumed by certain aspects of the data that is collected, van Manen encourages researchers to look at the entire body of the research to ensure certain aspects are considered in the context of the whole story or experience, thus the continuation of the hermeneutic circle throughout to provide a basis for a robust, thorough study.

4.9.7 Placing the researcher in this study

Van Manen differentiates between phenomenological research conducted by philosophers and by professionals of a practice (2014). Philosophers usually deal with philosophical phenomena that are mainly theoretical in nature, whereby professionals of practice (such as nurses), also use certain empirical and analytical methods required in social sciences (van Manen, 2014, 2017). Van Manen recognises that the researcher is an important aspect of a research study. He highlights that if a professional has too

many “pre- understandings” or beliefs about a phenomenon this can be an issue particularly if the researcher themselves does not acknowledge this (Van Manen, 1990). From exploring the different philosophers in phenomenology, the researcher felt it would be extremely challenging to bracket out their interpretation of this topic and van Manen’s six guidelines helped keep this potential bias in check throughout the whole research process. As a nurse, working with children with CCN and their families, the researcher is particularly interested in the phenomenon. Van Manen believes that it may be difficult to maintain the on-going and intensive involvement with the phenomenon, which is required to accomplish the project, if one is not deeply and personally interested in it (van Manen, 2014).

4.10 Conclusion

Chapter four presented an overview of the methodological framework that guides this study and the rationale for why hermeneutic phenomenology guided by van Manen is the chosen methodological approach to exploring fathers’ experiences of caring for a child with a CACCN. The variety of different types of research that could be utilised in exploring the phenomenon of interest were identified in this chapter. A discussion on both qualitative and quantitative research was also presented. The multiple research designs for conducting qualitative research were discussed, arguing why the decision to adopt a hermeneutic phenomenological approach was made. In doing so however, the complex and somewhat challenging nature of hermeneutic phenomenology was explored, and a historical viewpoint was developed which explains the two different research traditions of descriptive and interpretive phenomenology, in order to justify and truly understand the decision to adopt a hermeneutic phenomenological study guided by van Manen. Chapter five presents the methods that form the research design, participant recruitment, data collection, data analysis and ethical considerations.

5 Methods

5.1 Introduction

As discussed in chapter four, this study adopted a hermeneutic phenomenological approach guided by van Manen (1990, 1997, 2014) to explore fathers' experiences of caring for a child with a CACCN. Van Manen's method of hermeneutic phenomenology (1990, 1997, 2014) was chosen for this study as it aligned with the researcher's values and philosophical stance and allowed for a strong framework with which to explore fathers' experiences. Van Manen's (2014) hermeneutic phenomenological approach is a qualitative research methodology motivated by an epistemological belief in the significance of first-hand descriptions of lived experience. This chapter will describe the methods utilised to collect and analyse the data, while evaluating and justifying the methodological choices that were made throughout this study in order to demonstrate the research process. This fifth chapter will present the aims and objectives of this thesis, detail the sample, identify the inclusion and exclusion criteria, discuss the methods used to collect the data and discuss how this data was analysed. This chapter will also discuss the ethical considerations used throughout this study and will present how rigour was ensured.

5.2 Aims and objectives

As explained in chapter three, the scoping review highlighted that caring for a child with a CACCN presents unique experiences for fathers. Findings highlighted the need for more research on fathers' experiences of caring for a child with a CACCN. A wider variety of CAs need to be included to ensure adequate support is provided to fathers of children with other CA, not only Trisomy 21. Findings highlighted that further research should delve deeper into fathers' support needs, and coping strategies. The concept of fathers' experiences when their child has a diagnosis of a CA as well as CCN is not well reported. Findings highlighted that further research on this unique experience was needed to develop our understanding. Therefore, the aim of this study was to explore fathers' experiences of caring for a child with a CACCN. To address this aim, the following objectives were established:

- To explore fathers' experiences of how their child was diagnosed;
- To develop an understanding of how this diagnosis impacts fathers;
- To explore the roles and responsibilities fathers have in caring for their child with a chromosome abnormality and complex care needs;
- To identify what supports fathers have and how these supports impact their everyday life.

5.3 Sample

A population consists of all the people within a certain group from whom a researcher seeks to gain knowledge or information (Chadwich, 2017). Therefore, fathers of children with a CACCN are the population that were identified for this study. A subset of fathers of children with a CACCN were selected, this is called a sample (Chadwich, 2017). Qualitative research often relies on non-random sampling techniques due to the fact that these techniques provide deep information about the population of interest. Researchers often use purposeful sampling to select participants based on their particular knowledge of, and/or experience with, a particular inquiry (Robinson, 2024). Van Manen (2014) identifies the need for the participants of a study to have the relevant experiences of the specific phenomenon to enable them to create a narrative and reflect on a situation. There are many benefits to purposeful sampling and this method is seen as reliable, affordable and time effective (Andrade et al., 2020; Palinkas et al., 2015). Purposive sampling can ensure that a quality sample is located without biases (Nyimbili & Nyimbili, 2024) and therefore, was used as a method of recruitment of participants for this study.

As described in chapter two some CAs can be extremely rare, two routes of recruitment were established with the goal of allowing fathers of children with different abnormalities the opportunity to participate. Firstly, services in the community that provide direct support such as homecare service providers to fathers of children with a CACCN were contacted. Some fathers may not be accessing specific services or may be on waiting lists to access them due to a new diagnosis. Therefore, a second route via voluntary groups/support groups was also established. A gatekeeper was appointed for each avenue. The recruitment process is discussed in detail in section 5.5.

5.4 Inclusion and exclusion criteria

To ensure that there was a high-quality research procedure, establishing inclusion and exclusion criteria for study participants was an essential aspect of this study (Patino & Ferreira, 2018). Inclusion criteria are the key features of the target population that the researcher used to answer this research question and are needed in all types of research (Capili, 2021; Hulley et al., 2007). Fathers of children with a CACCN were the chosen population being studied. If a father had a child with CCN without a diagnosis of a CA they were not included in this study. Fathers caring for their child over the age of 18 with a CACCN may yield different results, and as the aim of this study was fathers of children, fathers needed to be caring for a child under the age of 18. Additionally, as interpreters were not available, fathers had to speak English to be included. It is important to note, that although there was an option to complete a journal, fathers were not required to have good written English and had the choice to not complete the journal if they did not feel comfortable writing in English.

5.4.1 Inclusion criteria:

- Fathers of children with a CACCN between 0-18 years;
- Fathers must provide regular care for their children with a CACCN;
- Fathers are required to speak English as interpreters are not available for this research study;
- Fathers are not required to have good written English.

5.4.2 Exclusion criteria:

- Fathers of children who do not have a CA;
- Fathers of children who have CCN but not a CA;
- Fathers who do not provide direct care for their children.

5.5 Recruitment and access

A robust recruitment plan increases trustworthiness and can also increase overall research success (Negrin et al., 2022). This study used two different methods of recruitment to ensure as many fathers as possible with children with different CA

diagnosis were afforded the opportunity to participate. The first route was from two service providers that provide HCP (both nursing and healthcare assistance) to children with CCN. The second route was through five support organisations/associations of different CAs.

5.5.1 Route 1: Homecare providers

The HSE Primary Care currently funds homecare nursing and healthcare assistance by using private healthcare providers, who deliver nursing care in the home environment to children with CCN (Department of Education, 2022). HCP supports the discharge of children with CCN from acute hospital services into the care of their families. A paediatric HCP is required when a child has medical and/or nursing needs that cannot be met by existing primary care services (such as therapies) and children's disability services (which may provide respite). Parents are still the main caregiver, but in-home respite is provided. In 2021, in Ireland, there were 533 packages of care for children with CCN and approximately 18,000 hours of homecare per year (HIQA, 2021). Therefore, homecare providers are significant contributors/influencers of fathers' experiences of caring for a child with a CACCN. There are five lead agencies that provide HCP for children with CCN. These agencies are on the national tender, in which successful providers enter a service arrangement with the HSE (HIQA, 2021). Service providers who carry out this care, were invited to be recruitment gatekeepers for this study.

5.5.2 Route 2: Support services for children with chromosome abnormalities

In the ROI not all children with CCN receive a HCP. Receiving a HCP is based on the individual child's clinical needs coupled with available resources (HIQA, 2021). This means that some families, although in need of in-home support may not yet be receiving these packages due to lack of resources, such as staff shortages, geographical location or their needs are not best met with a HCP. Therefore, to ensure that all fathers who are providing care for their children were included, a second route via voluntary groups/support groups was developed.

The HSE provides an extensive list of recognised support groups/websites for families of children with disabilities (HSE, 2025). This list was searched and support groups that

focus on supporting families of children with a CA in Ireland were selected. Additionally, a search looking for support groups of the most common CAs was completed and support groups contacted. Additionally, as many of the CAs are extremely rare, support groups for families of extremely rare conditions were also asked to participate. Twelve support/information websites were contacted and five agreed to take part. For the support groups that responded and wanted to take part, the administrator for the support group agreed to be the gatekeeper. Some support groups were services providing information websites or social media support pages and therefore they may not have direct contact with potential participants.

5.5.3 Gatekeeper

Gaining access to fathers of children with a CACCN through homecare companies and organisations/associations needed careful consideration as accessing an organisation to conduct research with its individual users can be difficult; and many healthcare organisations need healthcare researchers to go through a formal process to enter the organisation and gain access to potential participants (Singh & Wassenaar, 2016). A gatekeeper is a designated person who acts as a liaison between potential respondents and the data collector (Andoh-Arthur, 2019). Gatekeepers are in a position to manage who has access to a respondent and when, because of their personal or professional connection to the respondent (Andoh-Arthur, 2019; Gallo et al., 2012). Gatekeepers are used to help ensure the best interest of a population by maintaining their credibility and ensuring that the researcher does not jeopardise their relationships with the specific group of interest. The ethical concerns of participants can often be appropriately represented by gatekeepers, who are respected members of their communities (Eide & Allen, 2005; McFadyen & Rankin, 2016; Sanghera & Thapar-Bjorkert, 2008). A gatekeeper was therefore required to help gain access to fathers and to additionally ensure credibility of this study. Prior to gaining ethical approval, a letter was sent to all five agencies and 12 different support groups, and they were asked if they would be interested in being a gatekeeper to the fathers who receive support from their organisation (Appendix 4). Agencies and support groups email responses are displayed in Appendix 5.

At this point, any ethical concerns were discussed, explored and included in the research ethics application that was submitted to the School of Nursing and Midwifery Ethics Committee (SNMREC) in Trinity College Dublin. The service providers included in this study did not have their own ethics committee. Therefore, an agreement between the agencies who agreed to participate, and the researcher was put in place, so that the service providers would review the research proposal and approve on a basis of ethical approval from the SNMREC.

Once ethical approval was granted from the SNMREC, the researcher contacted the community service providers who responded to the letter requesting their organisation be a gatekeeper for this study. The individual organisations then appointed an independent gatekeeper within their organisation, who was then the point of contact for this study. The researcher either met with or spoke with potential gatekeepers to allow them time to ask any possible questions and explain in detail the aims and objectives and the method of the study. Once the gatekeeper was satisfied and all questions had been attended to, they then either addressed and sent out information packs, provided by the researcher, to potential participants, or uploaded the online advertisement poster to their websites. When instructed to, gatekeepers followed up with a reminder letter if required.

The researcher then distributed an information poster to the administrators who uploaded the poster to their sites, social media pages or displayed the poster within their organisation (Appendix 6). This poster had the researcher's contact details and the potential participants could contact the main researcher directly. Once the potential participants had contacted the main researcher she then sent (via email or post depending on the father's preference) the participant information pack to the fathers directly.

All fathers of children within the organisations who met the inclusion criteria were invited to participate. A letter of invitation and participant information pack was sent to each prospective participant outlining the title and purpose of the study, the amount of time it would take for them to engage with the study and informed them that

participation is voluntary (See Appendix 7 and Appendix 8). The names of the organisations were kept anonymous to ensure confidentiality of the fathers and the services were maintained.

One challenge of using a gatekeeper, is the recurring issue that may be related to the sensitive nature of the study and the deeply held attitudes and values of the individual gatekeepers (McFadyen & Rankin, 2016). Some research has shown that gatekeepers can hinder research on vulnerable populations as a result of their own attitudes or mistrust of researchers (Brodeur et al., 2025). This possibly led to some service providers choosing not to participate in the study. Five home care companies were asked to participate and two agreed to take part. One provider reported that these families are saturated with research projects.

5.5.4 Contact with the fathers and gaining consent

Contact was either made indirectly via the gatekeeper or directly with the researcher. All potential participants who had contact with the gatekeeper received the information pack through the gatekeeper. These fathers were invited to contact the researcher if they wanted some more information or if they wanted to participate in the study. Fathers who viewed the poster advertisement, either through social media pages or had been shown the poster through the various support groups, contacted the researcher directly to express interest in the study. The researcher then sent out the information pack to these fathers by email and they were informed to contact the researcher if they wanted more information or would like to participate. Fathers who wanted to participate were contacted by phone/Zoom explaining the consent process and any questions regarding participation in the study were discussed. This enabled them to make an informed decision when consenting to participate.

A written, signed, and dated consent form serves as the official record of informed consent (Manti & Licari, 2018). Informed consent is necessary when people such as parents, children or vulnerable populations are involved in the research; or when human genetic material, bodily samples, or personal data are used or collected in the research (Manti & Licari, 2018). A fully informed, competent individual gives their informed

consent when deciding whether to volunteer as a research subject. Therefore, informed consent from each participant was required in order to be part of the study. Informed consent means that a person has voluntarily opted into a research study, and it can only take place after they have been fully informed of all that the study will entail. Potential participants were therefore informed about the research, what was required of them, what would happen to their data and how it would be handled, as well as their rights as a participant, during the course of this research study. A timeframe of at least 7 days from when the participant was given the participant information pack to when the participant consented to participate was adhered to. This allowed the potential participant's time to reflect on the study, discuss the study with friends and family and develop any questions they may have regarding the study.

As this study offered the opportunity to complete online interviews via Zoom or in person interviews, two methods of informed consent were needed. When an in-person interview was completed a hardcopy of the consent form was signed by the participant. This was stored securely in a locked filing cabinet in a locked room within the School of Nursing and Midwifery. For the fathers who choose to complete an online Zoom interview, consent was gained through an online informed consent form via Qualtrics. The link for Qualtrics was sent via email. Additionally, a copy of the PDF informed consent was also sent via email/post to allow fathers to print off the informed consent to ensure they could read and understand all aspects of the consent form. The electronic data was stored on a password protected computer on an encrypted file backed up on a Trinity server. In addition to this, consent forms were downloadable from Qualtrics, and a hard copy was printed and stored with the manually collected consent forms within a locked filing cabinet in a locked room within the School of Nursing and Midwifery. Participants were invited to download a hard copy of the consent from Qualtrics for their own records, if they so wished. It must be noted that the consent form was read out by the researcher prior to the interview to ensure that if there were any literacy concerns, that every father had verbally heard and understood the consent form prior to commencing the study. Both the in person and online consent form is presented in Appendix 9 and Appendix 10.

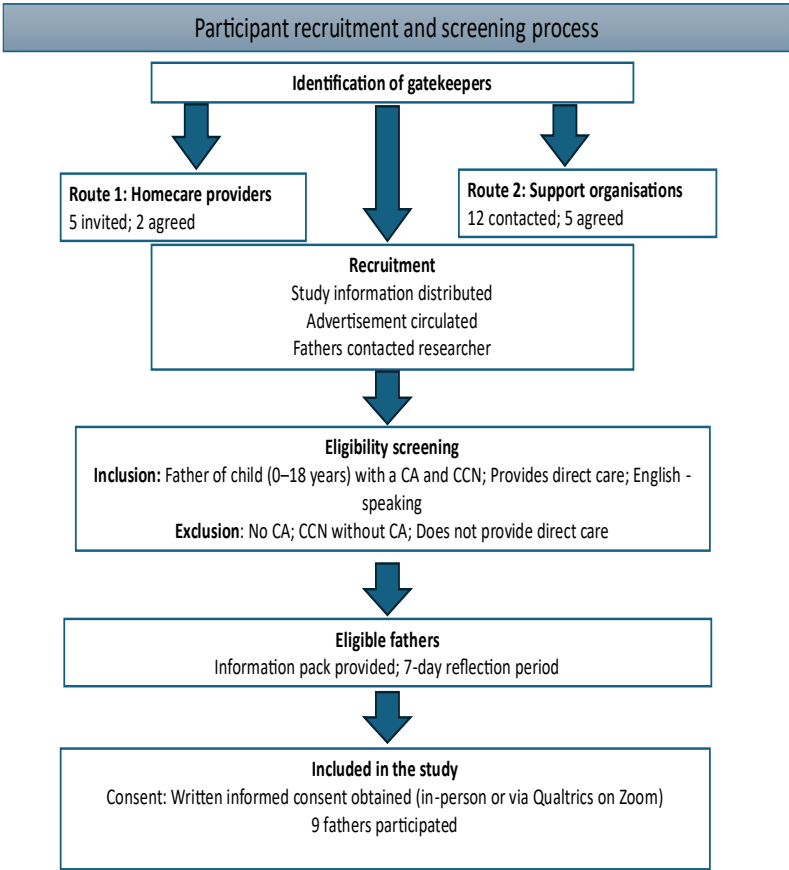
All participants were given the participation information leaflet which informed them of what data was going to be collected. Participants were informed of their rights during the research process.

1. Right of access: Participants had the right to access their transcripts and/or journals at any point up until the point of data analysis.
2. Right to rectification: Participants have the right to amend and rectify their transcripts and/or journals up until the point of data analysis. Only one father requested that they read their transcript.
3. Right to erasure: Participants had the right to erase their transcripts and/or journals up until the point of data analysis.
4. Right to object to processing based on legitimate or public interest: This was important for fathers who were invited to participate in the study through nominated gatekeepers and/or public advertising. The fathers were informed that they were not obliged to participate in this study as part of their child's ongoing care. Their decision to participate in the study or not will have no bearing on their child's ongoing care. Should a father initially choose to participate but change their mind, they have the right to withdraw from the study and have their personal information erased up until the point of data analysis. No father wished to withdraw from the study.
5. Right to data portability: Participants were informed that they had the right to request and receive their data in whichever document format is most amenable to them.
6. Right to object to profiling or making decisions about individuals by automated means: Fathers were informed that no clinical or otherwise significant decisions would be made about participants of this study on the basis of their data.

Figure 4 illustrates the participant recruitment and screening process used in this study. The figure details how study information was distributed, how fathers expressed interest, and how eligibility was assessed using specific inclusion and exclusion criteria. It also shows the informed consent process and final participant inclusion, providing a

transparent overview of how the sample of nine fathers was systematically identified and enrolled.

Figure 4: The participant recruitment and screening process used in this study.



5.6 Sample size

Van Manen (1990, 1997) does not indicate a specific sample size for his method, rather he explores the term ‘example’ over ‘sample’ expressing that example of experiential descriptions should be evident. Recruitment was conducted over a 12-month period and concluded after a consecutive three-month interval during which no new participants expressed interest. Phenomenologists tend to rely on a very small sample of participants, typically 10 or fewer (Polit & Beck, 2020). Therefore, after the 12 months it was deemed enough evidence had been collected to explore fathers’ experiences of caring for a child with a CACCN. This was known to the researcher when no new information was coming through the interviews.

5.7 Data collection

Two methods of data collection were completed as part of this PhD study, which included interviews and optional journal writing. As discussed in detail in chapter four, hermeneutic phenomenology allows researchers to describe and articulate how people's ordinary, everyday practices and routines are shaped by their family relationships, childhood experiences and day-to-day challenges (Alan et al., 1986; Gyollai et al., 2020; Heidegger, 1962; Oerther, 2020; van Manen, 2014). Van Manen argues that to do phenomenological research, you are "attempting to investigate what an experience is like" (van Manen, 1990: 42, 2017a: 776). Phenomenology is a way to capture lived experiences but does not claim to solve problems (van Manen, 1990) but is helpful to gain a rich description that may resonate with others. Data collection techniques are crucial because they affect the researcher's methodology and analytical approach, which in turn affect how the information is used and what explanations it might produce. Having a data collection procedure was vital in enabling the researcher to gather rich information and was crucial in allowing this study to meet its aims and objectives (Gunasena et al., 2024). This protocol included both the interview process and the journaling.

5.7.1 Interviews

Van Manen suggests that there are many means of data gathering for the analysis of lived experience, but he appears to favour interviewing individuals when gathering their reflective recollections (van Manen, 1990). One method of helping participants express and divulge their experiences, is by a series of semi-structured interviews, involving open ended questions with different interviews having a slightly different focus depending on the data collected in the previous interviews (Beven, 2014). Semi-structured interviews are the most common method of qualitative data collection in health services research (DeJonckheere & Vaughn, 2019). Specifically, with a hermeneutic phenomenology method in mind, van Manen (2016) believes that interviewing has two main purposes: (1) as a means to explore and develop a rich understanding of the phenomenon of interest; (2) to develop a conversation around the meaning of specific experiences. Van Manen (2016) suggests using conversational

interviews that are semi-structured in nature which will rely on the participants' memories and reflections to help participants revisit their experiences. Therefore, data were collected in interviews with the fathers to help answer the research question, what are father's experiences in caring for their child with a CACCN.

The first interview was unstructured and more conversational in nature, and then subsequent interviews had a more structured approach. The researcher was familiar with the areas of both CAs and CCN, which is recommended when carrying out semi-structured interviews (Jordan et al., 2021). A semi-structured interview guide was utilised for the interviews, including interview prompts (Flanagan & Beck, 2024). Following the first interview a time frame of at least three months was given before scheduling a second interview. This allowed the researcher time to explore the transcripts and delve deeper into the experiences and also allowed the participants time to reflect and add to their comments if they so wished (McGraw et al., 2019).

The interviews were audio recorded on a portable audio recording device as this allowed for an accurate summary of the interviews. Which is especially important when receiving in-depth information from semi-structured interviews. Online interviews, conducted via the Zoom videoconferencing system, were similarly audio-recorded and stored in an electronic data file. Audio recordings were downloaded in an electronic data format into an encrypted file on the researcher's login, on a password protected computer, backed up to a TCD server. This audio file was then stored in an encrypted file on a password protected computer backed up on a TCD server. This information was only accessible to the researcher.

Once ethical approval was gained and the sample had agreed to participate, the location of the interviews was determined depending on convenience and preference of the father (McGrath et al., 2018). The physicality of our situation or "space" can affect behaviour and responses at both conscious and subconscious levels, so choosing the location was important (van Manen, 1990). As discussed above, online interviews were also offered. This research study commenced in 2022, when COVID-19 guidelines and restrictions were still a possibility. COVID-19 transitioned in-person qualitative research

interviews to virtual platforms (Oliffe et al., 2021). Given that these families had children with CCN and increased vulnerability to Covid-19 offering online interviews was vital to ensure the safety of the children and their families. Eight fathers selected to do online interviews with one choosing face to face.

Only one father requested that the interview take place in person, and this father selected having it in their own home, although locations such as a quiet hotel foyer or a coffee shop were also suggested. This was very important to ensure that the fathers felt as comfortable as possible in participating. The researcher wanted to be sure that they felt they could openly discuss the true experiences of caring for their child with a CACCN. As these fathers were caring for their children in their own homes, and may have a busy household, it was important to ensure a space where they were not influenced by their surroundings and therefore left the choice of the interview location to the fathers. Additionally, a place that was quiet to allow the interview to be audio recorded was needed. These considerations impacted on the ability of the participant to relax and develop trust with the interviewer and the interview process.

Times were selected that suited the fathers. This allowed for some flexibility, some fathers preferred to do the interviews during their work breaks as they preferred to do the interview at a time that their children were in school or away from the house, while others selected evening interviews when their children had gone to bed. Additionally, some fathers gave an estimated time that suited them and were able to call the researcher when they had their children settled which resulted in them coming online at a time that was convenient to them. One father opted for Zoom to prevent having to get childcare during the interview. Two fathers had to cancel the interviews last minute due to having a sick child and we were able to reschedule the interview easily. One father was able to pause the interview and go to another room in the house to assist the child, recommencing soon after.

A time was agreed with the fathers and a Zoom link was sent via email. A reminder email was sent to the fathers the day of the agreed date. One interview was postponed due to unstable internet and was rescheduled the following day. Another interview was

postponed due to a child having an emergency hospital admission and another postponed due to receiving a cancellation bed for their child's surgery. All of these interviews took place at a later date.

5.7.1.1 The first interviews

Conversational interviews typically begin with a "grand tour question" (Spradley, 1979). Therefore, the first interview was unstructured in nature and began with a simple question of "Tell me about where your journey with [name of child] began". The aim of this interview was to allow it to flow like a natural conversation. This interview was non-directive and allowed the conversation to develop naturally into how fathers experienced caring for their child. The researcher works with children with CCN and has undertaken other research around CAs and was therefore well equipped to engage in this interview style. Some conversational questions were used by the interviewer to guide the conversation so that the interviewee was able to ponder and reveal their own moments of discovery. In preparing for the interview, the researcher wanted to ensure that she had prepared enough to allow the conversation to flow, without sounding like a rehearsed script. This was important, not just to enable a conversation but also to reduce the researchers own bias to the interview. In a phenomenological study, conversational interviewing calls for the researcher to reflect on their own perspectives, biases, and understandings; to be supportive of participants; to be receptive to learning from their experiences; and to thoughtfully share experiences and exchange them with participants through a dialogical interview process (van Manen, 2014). The researcher ensured that she knew which complex care needs are typically associated with the children's conditions.

Prior to the first interview the researcher was apprehensive as to how the conversation would develop. As the researcher is a nurse working with these families, she wanted to ensure her questioning style was that of a researcher and not a clinical nurse. When transcribing the first interview, the researcher noticed that she was a little apprehensive in her follow up questions but as interviews progressed her confidence grew, and the interviews became smoother. Fathers typically began to tell their stories in a somewhat

chronological order of the child's life, which allowed a natural flow to the interviews. The type of questions that were asked are displayed in Appendix 11.

5.7.1.2 The second interviews

Van Manen (1997) reports that transcripts require interpretive analysis by the researcher in order to produce a human science (phenomenological) description of the experience of the interviewee. The aim of the second and subsequent interviews are for the researcher to reflect and examine the data collected in the first interview, and create more clear, structured questions. Two fathers wished to only carry out one interview, which was acceptable, and their first interview was used in the analysis. This is extremely important to allow fathers the chance to opt out, which is especially relevant to fathers of children who have such complex needs that their situation may change quickly. All fathers were offered an opportunity to review their transcripts prior to the second interview with only one father taking this opportunity. The second interview took place at least 3 months after the first interview.

Transcripts were read and re-read and any areas that required further discussions were explored. Furthermore, the interview transcripts were compared with each other, determining if any fathers had mentioned points that were unusual or that other fathers had not mentioned for example; "I know you mentioned in our first interview that there is a huge cost to the medications that your child is on, and that the HSE pay for these medications and you are very grateful for these, have you noticed any additional costs that having a child with a CACCN has on you or your family?". This question was asked as other fathers had discussed the cost implication of having a child with CACCN, yet this father only discussed the cost to the state in his first interview. Van Manen (1990) suggests using probes such as "Can you think of a time?" or "tell me about this experience" to elicit personal memories and anecdotes from the participants. This was used throughout the second and subsequent interviews. This process continued until the researcher reached data saturation whereby there were no new ideas and insights or stories emerging from the data set.

5.7.2 Journaling writing

Van Manen (2014) discusses a variety of methods of collecting data from a sample that is vital in producing enough evidence to create meaning to the lived experience. Documenting experiences can assist a researcher in delving deeper into particular topics that may not be apparent through interviews alone. Journaling was first identified as a key form of data gathering by Verbrugge (1980), and it is now recognised as a successful approach of studying specific experiences in natural settings, particularly within hermeneutic studies (Hayman et al., 2012). Journals have been described as a data collection method that enables nurse researchers to gain a rich insight into a phenomenon of interest (Morrell-Scott, 2018). Fathers were given a journal at the time of the first interview to document their experiences and memories if they wished. As one of the aims of this study was exploring the experiences of these father's everyday life, the aim of the journal was to give fathers an opportunity to self-document aspects of their lives that they might find important to the study. This aimed to enable a father to document their own behaviours, experiences, or feelings which in turn could allow this research to delve deeper into the habits and behaviours of these fathers.

A recent scoping review that explored the use of personal journaling/ diary keeping and found that as a qualitative research method journaling is a practical means of generating first-hand and participant-directed data (Akinreni et al., 2024). This study included both the terms "diaries" and "journals" and these terms were at times interchangeable (Akinreni et al., 2024). In diary studies, participants are asked to repeatedly fill out questions about experiences, feelings, cognitions and behaviours (Kimhy et al., 2012).

The word diary comes from "diarium", which means daily allowance, entries are often arranged by date reporting on what has happened over the course of a day or other periods. A journal, however, refers to the process of participants sharing some thoughts, ideas, feelings and experiences through writing. Journals can be considered shorter and less restrictive in nature and are often not dated. The scoping review found that fathers of children with a CA have repeatedly reported that they are bound by time constraints which can pose a challenge to families (Cameron & Cooper, 2021; Cohen et al., 2016; Giallo et al., 2015; Huang et al., 2011; Marsh, 2020; Marshak et al., 2019; Ricci & Hodapp,

2003; Ridding & Williams, 2019) so therefore, shorter journal entries were considered to be more appropriate for this study as one complaint of research participants is that diaries can often be a challenge for those maintaining them but offer a personal insight which can be useful as a data collection method and also cathartic for the participant (Morrell-Scott, 2018), thus the researcher thought it would encourage fathers to complete the journals by asking for shorter entries.

In addition to the length and less restrictive nature of journalling compared to diary entries, the participants' gender was also taken into consideration. Research has shown that adults have exhibited some small but substantial gender variations in emotion expressions, with women displaying stronger emotional expressivity, while men can find it challenging to express emotions openly with others (Chaplin et al., 2015; Nolen-Hoeksema & Hilt, 2006). Journals are traditionally regarded as somewhat of a feminine activity (Starr, 2023). The term "journal" seems to be a more generic label than "diary," possibly making it more comfortable for men to engage with.

Like other methods, journals have their limitations. Participants must be willing to put in the necessary time, keep the journal updated on a regular basis, and adhere to any rules if applicable. In phenomenological research, diaries/journals can be either solicited or unsolicited (Akinreni et al., 2024; Corti, 2003), this means the solicited diary is a research technique in which informants actively participate in both recording and reflecting on their own thoughts, feelings, actions, and behaviour. Solicited diaries/journals are journals that are 'intentionally' created for the purpose of research, where participants are required to write diary entries regularly over a given period of time, reporting their own experiences and interpretations of events related to a particular research topic (Braun & Clarke, 2013; Cucu-Oancea, 2013). Due to the time constraints of these fathers, unsolicited journals were chosen.

The journal was offered as an optional form of data collection for these fathers. The journal was devised, by keeping it as simple as possible. In Ireland in 2023, 21% of adults have low literacy proficiency (Organisation for Economic Co-operation and Development, 2024). With this in mind, a short-written text was presented on the front

page of the journal explaining in simple English how to fill out the journal. This was also explained during the first interview. Additionally, if a second interview was completed without the journal entry, it was explained again. The decision to have this on an optional form of data collection was important should some potential participants not be able to read or write. Furthermore, the researcher was concerned that fathers who did not feel comfortable documenting their experiences in written format would not want to participate for this reason.

The design of the journal in this study aimed to be simple and easy to understand to aid completion. The journal was an A4 size booklet with the front page deliberately printed on white paper with the title “Journal” in blue, this was to make it distinguishable and remind fathers what this booklet was, but also to prevent it from ‘standing out’. Furthermore, no bright colours were used, as discussed above, some participants may have found this to be “feminine”. The journal is displayed in Appendix 12.

Fathers could write none, one or multiple entries depending on their own preference. Additionally, fathers may want to document more than one entry at the same time. The journal was left open to father’s wishes but had enough space that they could write long entries, or even one-word entries if they wished. Table 8 portrays the journal responses.

Table 8: Journal responses

Journal Responses	
Number of fathers who completed the journal	7
Number of fathers who did not complete the journal	2
Rationale for not completing the journal	• Time constraints • English not being the first language or not confident to write in the journal • Didn’t feel they could add anything in the journal • Forgot

5.7.3 Reflexivity and personal journal writing

Although not specifically part of the data collection, it is important to note that the researcher also engaged in journal writing. As the researcher is a nurse, working with children with CCN and their families, she was particularly interested in the phenomenon of fathers caring for their children with a CACCN. Van Manen believes that it may be difficult to maintain the ongoing and intensive involvement with the phenomenon, which is required to accomplish a study, if one is not deeply and personally interested in it (van Manen 2014). Reflexivity is an important aspect of hermeneutic phenomenology research, and the researcher needs to acknowledge their role. Van Manen (2014) highlights that if a professional has too many “pre- understandings” or beliefs about a phenomenon this can be an issue particularly if the researcher themselves does not acknowledge this (van Manen, 1990). Reflexive journals can help a researcher engage with the notion of creating transparency throughout their research and discover the impact of critical self-reflection (Ortlipp, 2008) which also ensured all decisions at all stages of the research were reflected upon (Olmos-Vega, 2022).

A reflexive journal can assist the researcher by allowing them to record and document their own thoughts and feelings and record the highs and the lows of data collection (Engin, 2011). This was particularly important when exploring father’s experiences, which in some cases was of a sensitive nature. Therefore, the researcher kept a journal that was taken with her throughout the interviewing process. This was completed before each interview to document any pre-understandings or concerns and then half an hour after each interview was taken as “quiet time” to reflect on each interview. For the eight interviews that took place over Zoom, this was sitting at my kitchen table with a cup of tea. For the in-person interview, this was at a coffee shop. The aim of this was to allow the researcher to do a deep reflection of her emotions and interpretation of the interviews allowing the study to align with van Manen, in his idea that the researcher must identify the role they have to play in the exploration. The researcher frequently went back to this journal throughout the data analysis phase to ensure she was identifying her own personal experiences with each interview.

The researcher used her own journal writing to focus a question in a second interview: “I remember looking around the families only sitting room, which now has the child’s hospital bed, and feeling somewhat awkward wondering where we would sit to record this interview that wouldn’t interrupt the family if they were to come home”. The researcher used this as a follow-on question regarding space in the home and the fact that the child’s bed was in their living room. It made her think of not just the concept of a hospital bed, but indeed the physical space that children’s equipment may take up within the family home.

The journal was also helpful, when reflecting on the interview transcripts. In one interview, a moment of silence was evident, while the father was deep in thought, remembering a sad and worrisome time in their lives, almost to the point of crying. The researcher wrote “As I’m listening to this transcript, I am trying to fill the silence, without just allowing the father to be silent in his own thoughts. I ask another follow-on question too quickly”. As van Manen was at the forefront the researchers mind, she thought about the concept of an epistemological silence is “the kind of silence we are confronted with when we face the unspeakable” (van Manen, 1990, p. 113), and reflected, that for the next interview, she would allow these silences to be part of the interview. This journal writing formed an integral part of the reflexivity throughout the research journey.

5.8 Data analysis

As this study adopted a hermeneutic phenomenological approach guided by van Manen (1990), this study therefore draws on van Manen’s approach to data analysis. The next section will detail how this was applied. This study used a rigorous, systematic method to analyse the data to ensure the studies’ trustworthiness (Ravitch & Carl, 2019).

5.8.1 Data transcription

The interview audio files were transcribed by the researcher by hand. Each interview was transcribed into a Microsoft Word document. Once transcribed the transcripts were then revised by the researcher to redact any identifiable information and replace names with appropriate pseudonyms and redact any sensitive personal information provided.

Once this was complete, the original transcripts were then deleted. Following each first interview, the researcher wrote a list of follow up questions that emerged from each transcript. Each interview was then compared to other interviews and any outlying points were noted and utilised as follow up questions or highlighted as areas for further exploration for the second interviews. The second interviews were then transcribed in the same format. For the father that requested to see his first transcript, all the identifying information was removed and then the transcript was posted to the father. This father used the transcription to prompt him in writing the journal entries. When the two journals were received, they were similarly transcribed onto a Microsoft word document and attached to the associated father's interview. These journals were then used as talking points for the second interview and follow up questions were also devised using these journal entries.

The decision to self-transcribe the interviews and not utilise an independent transcribing company was made for several reasons. Firstly, the researcher, being dyslexic, found that undertaking the transcription process personally allowed for deeper engagement with the data, particularly in discerning the nuances in the fathers' tones and expressions. Secondly, self-transcription aligned with van Manen's hermeneutic phenomenological approach, which emphasises the reflective process of writing and rewriting (1990, 2014).

5.8.2 Analysing the data in line with van Manen

In keeping with the methodological approach of hermeneutic phenomenology this study adopted van Manen's (1990, 1997, 2016) six research activities as a guiding framework for exploring the lived experiences of fathers caring for a child with a CACCN. Van Manen's method of thematic analysis, which incorporated three analytic steps was used to analyse the data in this study (van Manen 2007, 2014a). The three steps are "holistic reading, selective approach and detailed approach" and were applied within the six methodological guidelines, discussed earlier in section 4.10. This enabled the analysis to remain grounded in phenomenological inquiry while also allowing for rigorous and thoughtful interpretation of fathers' lived experiences of caring for a child with a CACCN.

This section will highlight how the data was analysed within van Manen's six guidelines. Table 9 highlights how the analytic steps were applied.

Table 9: Van Manen three analytic steps to data analysis

Analytic step	How it was applied
Holistic approach	The first step in the analytic process starts with a holistic or sententious reading of the text (van Manen, 2014). This method includes discovering, interpreting and reporting patterns and clusters of meaning within data (van Manen 1990, 1997). This step began following the first interview and was completed regularly throughout the data analysis phase. This was also undertaken with the journal entries.
Selective approach	The second step in this approach is to select or highlight key phrases or sections of the data that illustrate the meaning of this experience. Key segments were noted for their relevance in relation to fathers' experiences of caring for a child with a CACCN.
Detailed approach	The third step according to van Manen (2007, 2014a) is a detailed approach which incorporates a line-by-line analysis, by which every sentence is examined with the aim of capturing the essence of the entire lived experience.

Data analysis was conducted in a phased manner, consistent with the principles of hermeneutic phenomenology guided by van Manen. The process began following the first interview and continued iteratively throughout the data collection period. The phases involved in analysing the data are summarised in Table 10.

Table 10: Data analysis

Van Manen's (1990) guidelines	Application to the data analysis phase
Turning to the nature of lived experience	Transcribing the interviews Transcribing the journals Reading and re-reading both the interviews and the journals (Holistic approach)
Investigating experience as we live it	Created initial themes by noting key segments for their relevance (Selective approach)
Describing the phenomenon through the art of writing and re-writing	This incorporated a line-by-line analysis, by which every sentence was examined with the aim of capturing the essence of the entire lived experience (Detailed approach).
Maintaining a strong and orientated relation to the phenomenon	It was important to be reminded of this research study's aims and objectives while writing and re-writing.
Adopting an existential approach	Essential themes were then explored through the lens of the five existential themes; lived body, lived relations, lived space, lived things, lived time.
Balancing research context by considering part as a whole	Writing and rewriting the findings while balancing research context considering the parts and the whole

5.8.2.1 Turning to the nature of lived experience: Transcribing, reading and re-reading

The first step of analysing the data from the fathers' interviews and journals included listening and transcribing each interview, writing down initial thoughts and key points.

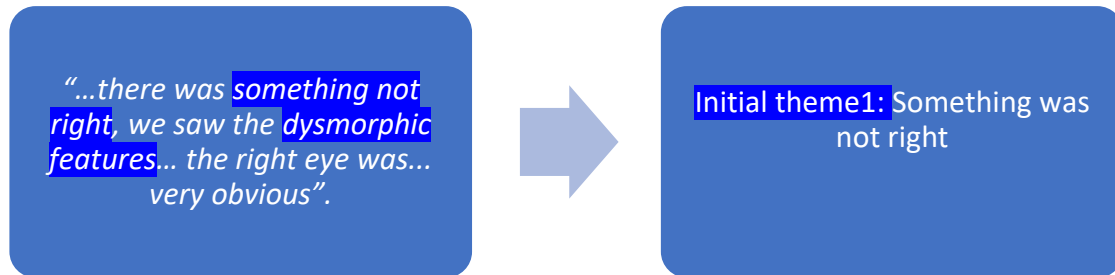
The interview recordings needed to be transferred to text. Each interview was transcribed shortly after they took place which was always ahead of the second interview. The journals were sent back from the fathers prior to the second interview commencing and were also transcribed. Each interview or journal was read multiple times prior to the second interview (reading and re-reading). A vital aspect of re-reading was listening to the audio recording alongside the transcript to note any shifts in tone, emotional inflections, and nuanced pauses that might not be fully captured in written text. *Epoché* is designed to stop researchers from jumping to conclusions too quickly, instead allowing researchers to examine the phenomenon more closely, giving time for what is not yet visible to manifest. Noting initial thoughts proved valuable in aiding the construction of follow up questions, or if certain topics needed clarification prior to the second interview. This was particularly useful in clarifying if different fathers had similar or differencing experiences. An example of this was: “Some fathers are noting...what are your thoughts on that?”

5.8.2.2 Investigating experience as we live it

The next step in the data analysis process was the creation of initial themes by noting key segments for their relevance (selective approach) (van Manen, 1990). The researcher read each transcript and highlighted segments that were particularly useful in understanding the phenomenon of the experiences of caring for a child with a CACCN. Each segment was grouped, and initial themes were developed by constructing keywords which eventually would become the essential themes.

The researcher listened to the recorded interviews and read the text several times searching for statements or phrases that seemed particularly essential or revealing about the experiences described by the participants. While van Manen (1997) suggests there is no need to use a software package, the researcher did explore the possibility of using a software package yet found doing it by hand allowed her to delve deeper into the data. An example of how the initial themes were compiled are outlined below in Figure 4.

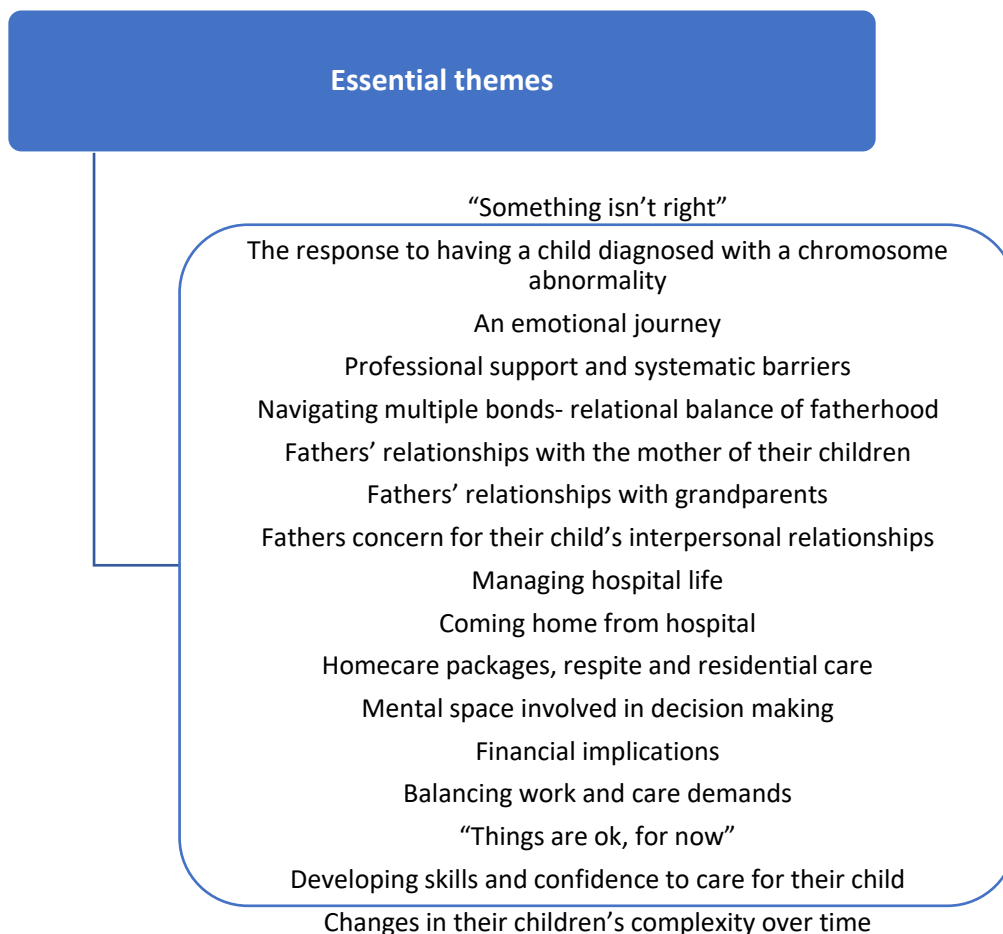
Figure 5: Method of key word identification



5.8.2.3 Describing the phenomenon through the art of writing and re-writing

The next step of the data analysis was writing and re-writing, which aided in the development of an understanding of the experiences of fathers caring for a child with a CACCN. This step also incorporated a line-by-line analysis, by which every sentence was examined with the aim of capturing the essence of the entire lived experience (detailed approach). Writing and rewriting allowed for the development of text giving context to the whole experience which were presented as the overall findings. The initial themes were then incorporated into larger essential themes. There were seventeen initial essential themes which are depicted in Figure 6. Direct quotations from fathers were incorporated within the essential themes to authentically capture the essence of their lived experiences. Every time a quote was selected, the entire transcript of their interview or journal entry was reread to ensure that the quotation was contextually appropriate and meaningfully aligned with the identified essential theme.

Figure 6: Initial essential themes



5.8.2.4 Maintaining a strong and orientated relation to the phenomenon

Ensuring that this study maintained a strong orientation to father's experiences of caring for a child with a CACCN was vital and is a fundamental element of van Manen's hermeneutic phenomenological approach. It was important that fathers' experiences were at the forefront. A major aspect of ensuring this was self-reflection, as although van Manen recognises the researcher as co-creator it was important that the findings were not from a healthcare professionals view and remained vigilant to maintain a strong orientation to father's experiences.

5.8.2.5 Adopting an existential approach

This study adopted van Manen's existential approach in which the five existential themes were incorporated into the analysis. Essential themes were explored through

the lens of the five existential themes; lived body, lived relations, lived space, lived things, lived time. These existential themes allowed for a deep reflection on the overall experience of caring for a child with a CACCN. Furthermore, the existential themes were used to present the findings. Each essential theme was mapped onto van Manen’s existential lifeworld dimensions, with an overall title of their experiences. These are displayed in Figure 7.

Figure 7: Lived existential themes (van Manen 2014, p. 302-310)

Lived body	Lived relations	Lived space	Lived things	Lived time
Receiving a diagnosis of a CACCN and the subsequent fight for services	The complexity of relationships	The physical and psychological space occupied by fathers	The material aspects of care	Adapting to their children’s CACCN changing over time

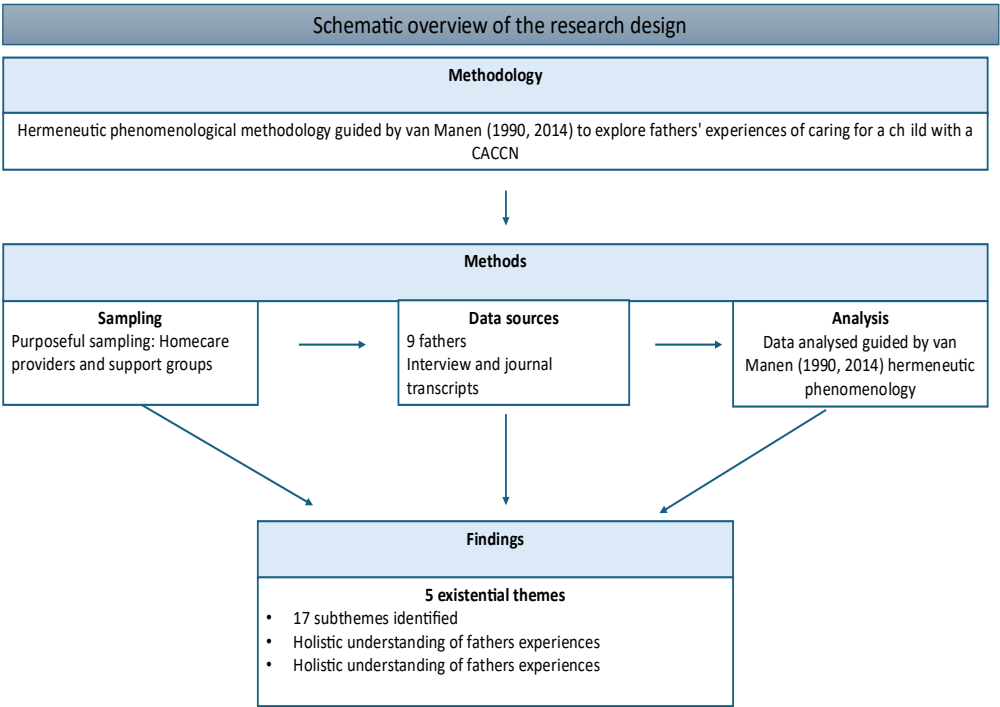
5.8.2.6 Balancing research context by considering part as a whole

Balancing the research context by attending to both the parts and the whole was a vital aspect of the data analysis. Although as discussed above that the data analysis commences with reading the transcripts, the art of writing and rewriting, reading and listening back to transcripts and then writing again allowed the researcher to go back to individual stories, quotes and passages (the parts) and then look at how they all fit together (the whole). This is important when examining the total meaning behind individual experiences. The hermeneutic cycle was evident throughout this process; this was not a linear process instead an iterative approach.

Figure 8 presents a schematic overview of the research design, illustrating how the methodology, methods, and data sources are interlinked within this study. The research is underpinned by a hermeneutic phenomenological methodology guided by van Manen (1990), which informed the selection of qualitative methods appropriate for exploring

fathers lived experiences of caring for a child with a chromosome abnormality and complex care needs. Purposeful sampling through homecare providers and support groups guided the recruitment of fathers, while data were generated through in-person and online semi-structured interviews, alongside optional reflective journals. These data sources produced rich experiential accounts, which were analysed using van Manen’s hermeneutic phenomenological method (2014a). Analysis was iterative and reflexive, with ongoing movement between data collection and interpretation, resulting in the identification of five existential themes and associated subthemes grounded in van Manen’s lifeworld existentials. This integrated approach ensured coherence across all stages of the research process and supported a holistic interpretation of fathers’ experiences.

Figure 8: Schematic overview of the research design



5.9 Ethical considerations

Ethical considerations in this research were a vital aspect of conducting this study, which ruled out any potential misconduct either with the method or the way results are presented (Correia, 2023). Within research, ethics is an essential measure to protect

society, and the earliest research ethics code emerged in the 19th-century (Vollmann & Winau, 1996). Ethics is a broad term that covers the study of the nature of morals and the specific moral choices to be made (Varkey, 2021). Ethical approval was received through the SNMREC, Trinity College Dublin in August 2022 (Appendix 13).

The Nursing and Midwifery Board of Ireland (NMBI) in partnership with The Department of Health developed “The Research Strategy for Nursing and Midwifery in Ireland” (NMBI & Department of Health and Children, 2015), which is a document regarding ethical conduct of nursing and midwifery research. The aim is to provide nurses and midwives with general guidance and overview of ethical matters that relate to nursing research and to ensure the protection of the rights of all those involved in the research process. Autonomy, beneficence, nonmaleficence, justice, fidelity, veracity, privacy and confidentiality (Ahmed, 2024; Beauchamp & Childress, 2001; International Council of Nurses, 1996; Polit & Beck, 2004; Rasheed et al., 2023; Storch et al., 2004; Varkey, 2021). Although this research explored fathers, who were all adults, the research focus included children and therefore, “The Ethics Journey in Children’s Research: Checklist” was also consulted (Department of Children, Equality, Disability, Integration and Youth, 2022). All of these aspects were given significant consideration throughout this research study and will be discussed in detail below. Table 11 highlights the ethical considerations that were considered prior to commencing the research study and were evaluated regularly throughout the research process.

Table 11: Ethical considerations

Key ethical considerations	Meaning	How it was applied to this study
Autonomy	Respecting individuals' rights to make informed decisions	Ensured fathers right to self-determination, which means that a fathers had the right to choose whether or not to participate in this research study, the right to full disclosure, ensuring that they received information outlining the nature of the study, including the likely risks and benefits, allowing them to make an informed choice and the participant has the right to withdraw at any time with no consequences
Privacy and confidentiality	Acknowledging individuals' will and preference	- To ensure confidentiality, data was collected on a coded basis with each participant receiving a unique identifier (study ID number) along with a pseudonym (both father and child). - The electronic data, including identifiable participant information, were stored on a password protected computer on an encrypted file backed up on a TCD server. - Fathers were informed that the interviews would be recorded and transcribed. Transcribed recordings and/or journals were then converted to an electronic data format, stored in an encrypted file on a password protected computer. - Location of the interviews were discussed with the fathers to ensure they took place in a location that fostered privacy and confidentiality.
Non-maleficence	Avoiding harm and prioritising safety	- Ensuring no harm to both the child and the father was vital. Prior to the study, relevant support groups contact details of specific support groups were devised for the fathers. A detailed plan was put in place in the event concerns for the father's wellbeing were noted throughout the data collection process. - Fathers were informed about the children's first policy and that the researcher was a mandated person. Child protection (children's first training) was completed by the researcher.
Beneficence	Minimising risks, and maximising benefits	- Fathers were informed in the PIL that they would not directly benefit from this project but their participation would add to the body of knowledge. - The aims and objectives of the research was outlined.

		● Dissemination of the results is an important element of beneficence.
Justice	Ensuring fairness and advocating for vulnerable individuals.	● Purposeful sampling was used, and only participants that met the inclusion criteria were included. ● A gatekeeper was also used.
Fidelity	Maintaining trust and honouring commitments	● Participant information leaflet was given to all participants ● Information about the background of the researcher was given to the fathers. ● A discussion with the researcher was conducted prior to the first interview to explain the study and develop a rapport.
Veracity	Being truthful in all communications.	● The researcher made every possible effort to ensure that all participants understood all aspects of the study. ● The PIL gave fathers the written explanation of all aspects of the study. ● They were given at least a period of seven days for reflection on the PIL. ● Fathers signed written consent, stipulating that they understood all aspects of the study. ● The researchers' contact information was given to all fathers in the event that questions arose along the way.
Trustworthiness	Credibility Transferability Dependability Confirmability	● Adequate time with each father was taken to build a rapport with them prior to the interview taking place. Fathers were offered two interviews, and an optional journal giving ample opportunities to develop an understanding of their experiences. A researcher's reflective journal was also used. ● Contextual information about the fathers in this study was presented in the demographics section of the results. ● Documenting and recording each step of this research process was vital, this was completed before the research took place and re-evaluated at each stage. ● The supervision team was instrumental in ensuring this element was adhered to. Both members of the team had a background in children's nursing and/or intellectual disability nursing and provided essential feedback throughout the writing process. Additionally, offering fathers the options to review their transcripts and having second interviews enabled the researcher.

5.9.1 Autonomy

Issues of anonymity are key considerations in ethical research practice and was a key element to this research study as it allowed the research participants' autonomy while also ensuring full disclosure of factors regarding the study (Barrow et al., 2022). The concept of autonomy calls for the ability of people to make their own decisions to be acknowledged and respected. As well as vulnerable populations having their autonomy protected by avoiding having undesirable decisions imposed upon them (Owonikoko, 2013). A key recommendation by NMBI (2015) is to treat participants as autonomous. Fathers were provided with complete information about the study which allowed them to decide on their own whether to enrol and to ensure this, all potential fathers were given a PIL which informed them of what data would be collected. Anonymity was preserved as fathers were informed of their rights within the PIL.

5.9.2 Privacy and confidentiality

Taking steps to ensure that research participants privacy and confidentiality is maintained is a crucial obligation of the researcher, this means ensuring that participants identity or data is protected and not shared to others (Bos, 2020). Thus, preventing any damage and fosters trust (Resnik, 2018). Extreme care was taken during the interpretation and presentation of the findings, which allows a study to ensure confidentiality, privacy and anonymity for the fathers and their children (Bera, 2018:2). To ensure confidentiality, data were collected on a coded basis with each participant receiving a unique identifier (study ID number) along with a pseudonym (both father and child considering the sensitivity of data collected). Manually collected data were secured in a locked filing cabinet in a locked room within the School of Nursing and Midwifery until data entry to electronic format. The electronic data, including identifiable patient information, were stored on a password protected computer on an encrypted file backed up on a TCD server. This was only accessible to the researcher and the research supervisors. Codified information for analysis (father's demographics) without patient identifiers, was stored on an encrypted file on a password protected computer accessible only to the researcher and research supervisors. A physical log was stored in a locked file cabinet in the same room as the computer, where data processors

could log how and when personal data has been consulted, altered, or erased. Disclosure of personal data was not used as part of this research study.

Fathers were informed that the interviews would be recorded and transcribed. Transcribed recordings and/or journals were then converted to an electronic data format, stored in an encrypted file on a password protected computer. Online interviews conducted via the Zoom videoconferencing system, also had the audio of the interview recorded and stored in an electronic data file. This audio file was also stored in an encrypted file on a password protected computer. This information is only accessible to the researcher.

5.9.3 Non maleficence

Non-maleficence' means seeking to do no harm. In all research, no excessive physical, emotional or psychological demands should be placed on participants (Polit & Beck, 2016). Researchers have a responsibility to balance potential benefits against potential risks to reduce possible risk and safeguard the protection of participants (Parahoo, 2014). There were no foreseen ethical issues for this study. However, it was possible that a father may become upset as they recall their experiences of caring for their child. The individual who was responsible for data collection is a Registered Children's Nurse/Registered Intellectual Disability Nurse and has 6 years of experience working closely with children with CCN and their families and would use this experience to support a father who became upset. Additionally, relevant contact details of specific support groups were provided. The researcher would advise the fathers to contact a relevant person to seek advice/support (i.e. key worker, liaison nurse, GP, pharmacist or relevant other) or with the father's permission the researcher would make contact with one of the above in the event that this occurred. Fathers were informed of this in the PIL. Although this was not necessary throughout the data collection process, having this in place before interviewing was a vital aspect in the data collection planning.

As this research explored fathers' experiences, children were the topic of conversation. Non-maleficence includes the need to avoid harm to children both through acts of commission or omission (Child Ethics, 2024). Safeguarding of the children was

considered and at the forefront of this study. The primary researcher completed relevant child protection training and was aware of the Children's First National Guidelines for the Protection and Welfare of Children (The Department of Children and Youth Affairs, 2017). A plan was put in place for the event that a father disclosed any information or evidence that their child is at risk of harm, the researcher would discuss this with the father and let them know that they had to report this information to the relevant personnel. The appropriate steps would be followed to safeguard the child.

5.9.4 Beneficence

Beneficence in health research is the twin concept of the non-maleficence demand that subjects should not be harmed through the conduct of the study, which means the researcher needed to ensure that there was minimum risk to fathers and to maximise the benefits of taking part in the study (Mondragón et al., 2022; Owonikoko, 2013). It is worth noting that, in difference to nonmaleficence, the language here is one of positive requirements. The principle calls for not just avoiding harm, but also to benefit patients and to promote their welfare (Varkey, 2021). In qualitative research, it is acknowledged that the participants may not directly benefit but the development of research or knowledge may benefit this population as a whole (Rossetto, 2014). Fathers were informed in the PIL that they may not directly benefit from this project; however, the aims and objectives of the research were outlined, and it was acknowledged the findings may be of benefit to future developments.

5.9.5 Justice

The principle of justice is defined as the ethical obligation to distribute the benefits and burdens of research fairly (Parahoo, 2014). This included ensuring fairness and advocating for vulnerable individuals (NMBI, 2015). This was applied by ensuring appropriate selection of research subjects, in which careful consideration was given. Purposeful sampling was used and only fathers who met the inclusion criteria were permitted to participate. Additionally, a gatekeeper was used who liaised with a potential respondent and the data collector, this was used as a protection to this population as they help ensure the best interest of a population.

5.9.6 Fidelity

Fidelity in research is the idea of trust (ICN, 1996; NMBI, 2015). In this study, fathers placed trust in researcher, and this necessitated a commitment to protect them. Trust was built with the fathers by firstly providing enough information through the PIL and answering any relevant questions they may have had prior to the study commencing. Secondly, the researcher is an experienced nurse who works in the area of CCN, which helped to build trust and credibility of the study.

5.9.7 Veracity

Veracity involves the responsibility of the researcher to tell the truth about the study and the absence of deception. Participants have the right to be told the truth and not to be deceived about any aspect of the study (Parahoo, 2014). The researcher made every possible effort to ensure that all participants understood all aspects of the study. The participation information leaflet (PIL) gave fathers the written explanation of all aspects of the study. They were given at least a period of seven days for reflection on the PIL. This gave fathers time to consider and discuss participation if they so wished with friends and family members. Fathers then signed written consent, stipulating that they understood all aspects of the study. The researchers' contact information was given to all fathers if questions arose along the way.

5.9.8 Trustworthiness

Bloomberg and Volpe (2012) described trustworthiness in qualitative research as being credible, dependable, confirmable, and transferable. Ensuring trustworthiness was crucial in establishing the credibility and reliability of the qualitative findings in this study (Ahmed, 2024). This study followed the pillars of trustworthiness in qualitative research by ensuring adequate credibility, transferability, dependability and confirmability to ensure this study was robust (Ahmed, 2024).

Qualitative research is a valued method of inquiry which is now a widely accepted by the global community of medical and nursing researchers (Ahmed, 2024; Norwell et al., 2017; van Manen, 2006). Qualitative researchers must document, systematise, and disclose the techniques of analysis in sufficient detail for the reader to assess the

process's credibility to show that the data analysis was undertaken in a precise, consistent, and exhaustive manner. In hermeneutic phenomenology, researchers assume their prior knowledge, experiences, and biases cannot be fully bracketed, as the researcher is the primary analytic tool; however, researchers engage in a process of explicating assumptions and preunderstandings as part of the process (van Manen, 2016).

5.10 Integrity of the study - quality and rigour

Ensuring that quality and rigour were maintained was a vital element of conducting this study to ensure that true representation of fathers' experiences of caring for a child with a CACCN was portrayed. Van Manen (1990) believes that rigour in a research study comes from being able to illustrate the connections between philosophical framework and findings as an interpretive process. Phenomenology uniquely emphasises depth, transparency and the preservation of the described lived experience (Tavakol & Sandars, 2025). Qualitative research must be original, relevant, have a clear aim and be rigorous (Stenfors et al., 2020). This study was original, given the fact that it is the first known study on fathers' experiences caring for a child with a CACCN. The relevance of the study was clearly highlighted following a detailed scoping review on the topic as seen in chapter three. Furthermore, the scoping review allowed for the development of clear aims and objectives as highlighted in section 5.2.

A recent review evaluating the methodological rigour of phenomenological studies examined the frequency and accuracy of phenomenological approaches and provided recommendations to enhance qualitative rigour in future research. It highlights the need for appropriate use of data collection and analysis methods aligned with phenomenological principles, and the need to clearly document the philosophical underpinnings of their study (Ago et al., 2024). The philosophical underpinnings of this study are clearly outlined and are guided by van Manen (1990) and are highlighted in detail in chapter four. Furthermore, the method of data collection as advocated by van Manen (1990) was ensured, by the implementation of prolonged engagement with the fathers and undertaking multiple interviews. Data were analysed using van Manen's hermeneutic phenomenological approach to data analysis (van Manen, 2014a). Findings

were also presented through van Manen's existential themes of lived body, lived relation, lived space, lived things and lived time.

Rigour, in the context of qualitative research, refers to the trustworthiness, credibility, and overall quality of the research process and findings (Saul, 2024). There is some critique about the concept of rigour in hermeneutic phenomenology research and it has been argued that this is not possible, rather "hermeneutic phenomenology lies in creating study reports that compel thinking and invite reinterpretations of life experiences" (Crowther et al., 2017: 834). However, van Manen (1990) believes that phenomenological rigour is not determined by methods such as member checking or triangulation but rather on the strength of insight, originality and the scholarly rigour of the interpretive process. Lincoln and Guba's (1986) principles of credibility, transferability, dependability, and confirmability were incorporated to enhance the rigour of this study. This is a widely accepted and recognised criteria that allows a study to demonstrate rigour (Nowell et al., 2017).

5.10.1 Credibility

The truth value of qualitative research, or whether the study's conclusions are true and accurate, is measured by its credibility. Prolonged engagement with the research field ensures that credibility is maintained as this allows qualitative research to capture data that may not be evident in short interactions (Ahmed, 2024). Adequate time was taken with each father to build a rapport with them prior to the interview taking place. Fathers were offered two interviews, and an optional journal giving ample opportunities to develop an understanding of their experiences.

Reflexivity and identifying the researchers own potential bias was also a significant element of ensuring credibility to this study. Keeping a reflective journal allowed the researcher time to achieve a deep reflection of her emotions and interpretation of the interviews allowing her to align with van Manen, in his idea that the researcher must identify the role they have to play in the exploration. Furthermore, using the transcript as well as the reflective journal as the data analysis took place. The researcher was conscious of the use of any leading questions, and searched for evidence of her own

presumptions in the conversation that had taken place within the interview. Van Manen's method of hermeneutic phenomenology (1990, 1997) was chosen for this project as the researcher felt it suited her values and was the best method to equip her to explore fathers' experiences of having a child with a CACCN. Van Manen's six basic activities in approaching research enabled her to ethically and vigorously create meaning from the lived experiences of these fathers to ensure credibility throughout.

5.10.2 Transferability

Transferability in research is the concept of applying information gathered by specific people and using this information to apply this information to others who have not been directly studied (Drisko, 2024; Riazi et al., 2023). Contextual information about the fathers in this study was presented in the demographics section of the results, pseudonyms were used to maintain confidentiality to enable readers to assess the transferability of findings. Additionally, the sampling strategies that were employed to recruit fathers are presented, which enables readers to identify whether the findings from these fathers' depictions are potentially transferable to other cohorts of people. However, while findings from this study can be transferable to similar populations, van Manen (2017) does not suggest the use of generalising results. He argues that "phenomenology is not to be confused with case studies, ethnographies, narrative inquiries, or empirical studies that aim to generalize their findings to a certain group or population, and so forth" (van Manen, 2017, p. 779). Therefore, while the results of this study exploring fathers' experiences of caring for a child with a CACCN may have relevance to comparable contexts, they are not considered generalisable.

5.10.3 Dependability

Dependability was ensured in this study by making sure it was repeatable and consistent. This allows the study to show trustworthiness as it pertains to the unwavering nature of the research findings across time (Ahmed, 2024). Documenting and recording each step of this research process was vital, this was completed before the research took place and reevaluated at each stage. Supervision records served as a method of traceability, in which detailed discussions around each step of the process were documented.

5.10.4 Confirmability

The concept of confirmability pertains to the impartiality and objectivity of the findings (Rasheed et al., 2023). The supervision team was instrumental in ensuring this element was adhered to. Both members of the team had a background in children's nursing and/or intellectual disability nursing, they provided essential feedback throughout the writing process. Additionally, offering fathers the options to review their transcripts and having second interviews enabled the researcher to clarify any points made by fathers to ensure their comments were accurately represented.

5.11 Conclusion

This chapter presented a detailed description of the methods adopted in this study. This included justifying the methodological choices that were made throughout, aligned with the chosen van Manen framework. The aims and objectives, the recruitment process and the inclusion and exclusion criteria were discussed. The use of interviews and the rationale for also using a journal was presented, as well as how the data was then analysed. The ethical considerations were also discussed. The next chapter, chapter six will present the findings.

6 Findings

6.1 Introduction

This chapter represents an interpretation and analysis of the data gathered in this study. Key findings are presented using the method of analysis described in chapter 5. Guided by van Manen's (1990, p. 39) concept of hermeneutic phenomenology, which aims to reveal "the lived experience... in such a way that we are now able to grasp the nature and significance of this experience in a hitherto unseen way". The analysis seeks to illuminate the fathers' unique perspectives. The transcripts were examined through the lens of the five existential themes as outlined in chapter 4: lived body (corporeality), lived space (spatiality), lived time (temporality), lived human relation (relationality), and lived things (van Manen, 2016). Through this, five major themes and seventeen subthemes emerged which will be presented in this chapter. The aim of this study is to explore the experiences of fathers' caring for a child with a chromosome abnormality and complex care needs.

6.2 Demographic data

Demographic data were collected as part of this study. Nine fathers participated in the study; seven fathers completed two interviews while two fathers participated in one interview, resulting in a total of sixteen interviews. Two fathers opted to complete the journal entries. Eight fathers chose to participate in the interviews via the online platform Zoom and one father chose to be interviewed in his home. Throughout the interviews fathers were asked for certain demographic information that helped develop a background into their lives and to give context to some of the themes. For example, all fathers noted that they worked or had worked outside the family home and that they all had other children. All fathers spoke of their relationship with the mother of the child. Eight of the fathers were in a partnership and/or were married to the mother of the child, one father separated with the mother in the first year of the child's life.

Fathers and their children with a CACCN were given a pseudonym to protect their identity and anonymity. Mothers and siblings are not given a pseudonym but will be referred to as "X's mother" or "X's sibling"; X being the child's pseudonym. Furthermore,

due to the small number of study participants, the children's specific CAs identified in this study are not associated directly with the pseudonyms to avoid potentially identifying specific participants. Additionally, identifying features such as hospitals and locations have been removed. Fathers and their children's pseudonyms are displayed in Table 12.

Table 12: Father and child pseudonym

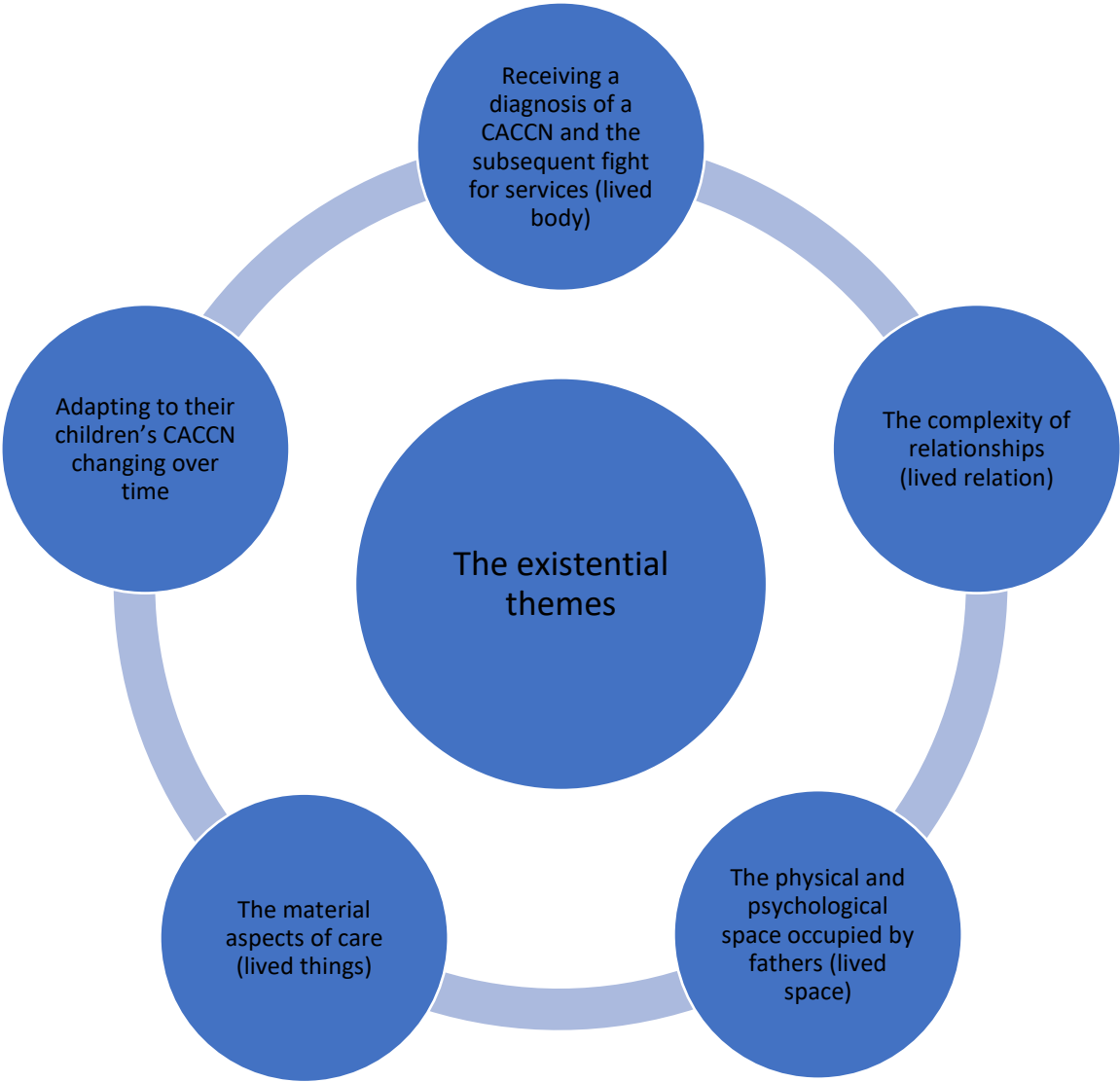
Participant number	Father's Pseudonym	Child's pseudonym
1	Peter	Rosie
2	Mike	Toby
3	Jan	Jakob
4	David	Joey
5	Alex	Amy
6	Rory	Jayne
7	Ali	Zara
8	Jamie	Lucy
9	Paul	Caitlin

Understanding the children's demographic data is also important to give context to the complexities the children face, thus giving context to the fathers' experiences. There were six female and three male children, diagnosed with a variety of CAs. All the children were under the age of 16. There were two children with Down syndrome, two children with Williams syndrome, two children with Prader-Willi syndrome, one child with Turner syndrome, one child with Trisomy 13, and one child with an unnamed abnormality (1.1mb missing on 14q.33 and slight loss on 22qxb22.33.12 and 12q22.1). Appendix 14 gives more detail and context about their CCN.

6.3 Presentation of the themes and subthemes

This study represents nine fathers' experiences and are presented through the lens of the five existential themes as outlined in chapter four: lived body (corporeality), lived space (spatiality), lived time (temporality), lived human relation (relationality), and lived things (van Manen, 2016). Within each of the five existential themes, an overarching title is presented, as seen in Figure 9.

Figure 9: The existential themes and their overarching title



Within each existential theme, a total number of seventeen subthemes or essential themes were identified through the process of hermeneutic data analysis outlined in chapter five. The seventeen subthemes help display the overall experience of fathers caring for a child with a CACCN. These are displayed in table 13.

Table 13: The existential themes and subthemes

(lived body)	(lived relation)	(lived space)	(lived things)	(lived time)
• “Something isn’t right...” • The response to having a child diagnosed with a chromosome abnormality • An emotional journey • Professional support and systemic barriers	• Navigating multiple bonds-relational balance of fatherhood • Fathers’ relationships with the mothers of their children • Fathers’ relationships with grandparents • Fathers’ concerns for their child’s interpersonal relationships	• Managing hospital life • Coming home from hospital • Homecare packages, respite and residential care • Mental space involved in decision making	• Financial implications • Balancing work with care demands	• “Things are ok, for now...” • Developing skills and confidence to care for their child • Changes in their children’s complexity over time

6.3.1 Theme one: Receiving a diagnosis of a chromosome abnormality and complex care needs and the subsequent fight for services (lived body)

The first existential theme that will be discussed is “lived body”. The emerging themes represent the physical and emotional impact of their children’s CACCN has on the father, particularly in relation to how fathers learned about the diagnosis and how they subsequently coped. Fathers’ experiences of their child being diagnosed is multifaceted

and despite all children having a CA, and in some cases a shared one, each individual experience was different.

This theme is further delineated into four subthemes. The first subtheme highlights how fathers realised that something was abnormal with their children, and this time frame varied between fathers resulting in unique experiences. The second subtheme portrays the fathers' emotional responses to receiving this news. The third subtheme explores the impact that healthcare professionals have on fathers during this time. The final subtheme describes how fathers came to terms with their children's diagnoses.

6.3.1.1 "Something isn't right..."

Fathers recall how they realised that their child had a CA. All nine fathers recounted where their journey of having a child with a CA began. Each interview started in chronological order of realising that something was not "right". All nine children were diagnosed with a CA postnatally, with only one father being told his child would have CCN prenatally. Five children received a diagnosis in the neonatal period due to the presentation of dysmorphic features, while three children missed developmental milestones and subsequently received a diagnosis in their toddler years (prior to starting school). Fathers recount this early experience, many of whom noted the physical features of their children at birth and were appreciative of healthcare professionals making this diagnosis early, while others who received a later diagnosis recall their need to search for answers, and at times feeling frustrated with professionals.

Caitlin was diagnosed with a heart condition prior to her birth. Paul was the only father who was told his child would have CCN prenatally:

"... Caitlin was diagnosed with left ventricular hypoplasia, so the left side of her heart, the pump side wasn't working, not developed properly, so as soon as she was born she would need an operation... They informed us after about a year that normally, people with the left heart condition usually have other issues, and that's when she was tested for [Caitlin's CA]." (Paul, interview 1)

Both Ali and Mike recalled having concerns that there might be something abnormal with the developing baby in the prenatal period. For instance, Ali felt that something

was wrong during his wife's pregnancy, as the mother of the child was experiencing continuous pain. He describes repeatedly going back to the maternity hospital as they felt something was wrong with their baby. Ali felt that his wife was dismissed during her pregnancy, and he believed that Zara's diagnosis should have been detected in prenatal scans:

"...and the hospital, maternity hospital was not able to identify that something was wrong until Zara was born... [mother's name] kept saying something is wrong, the baby is so big, but still, they did not recognise the symptoms... And they didn't listen; they thought [mother's name] was making it up... They should have seen it on the scan." (Ali, interview 1)

Five children received a hypothesised diagnosis (later confirmed on genetic tests) shortly after birth and all in the neonatal unit. For some, fathers noted the physical features, such as hypotonia, a protruding tongue or double digits:

"...there was something not right, we saw the dysmorphic features... the right eye was... very obvious". (Paul, interview 1)

Two fathers noted the fear and concern that their child wasn't alive and noted the lack of movement from their baby. Rory recalled Jayne being lifeless:

"... just lifeless in the bed... she was like stone". (Rory, interview 1)

Four fathers were then told by professionals that although they would have to wait for formal diagnosis with the blood tests, professionals named what CA they thought their child had. These fathers appreciated being told the potential diagnoses, even if they had to wait for formal results. Alex spoke of his gratitude that the dysmorphic features had been identified so early on, now knowing what he knows about the rare nature of his child's condition:

"We found out subsequently, that the reason why he knew the presentation of [Amy's CA], was because he worked over in the children's hospital in London where there were a lot more presentations. And fortunately, enough, he was on duty on that day..." (Alex interview 1)

Other fathers' experiences differed, and they identified that "something was not right", not because of physical features at birth, but rather their children were not meeting developmental milestones; thus, a diagnosis was made in the early childhood years. David and Peter spoke of the frustration of not having answers from healthcare professionals. They recall searching for answers to their children's symptoms. Peter remembers feeling like something was wrong and taking Rosie to multiple healthcare professionals. He shared his feelings of betrayal, noting that he had trusted professionals' expertise:

"We took her to a couple of different private paediatricians, none of them diagnosed her, and everyone missed the diagnosis... you're trusting that they know what they are on about." (Peter, interview 1)

David spoke of the frustration of having to seek non-medical avenues to find the "cause" for Joey's developmental delays and unexplained medical needs:

"like we were clutching at every straw, you know... [we tried] kind of heebie jeebies medicines, you know... homeopathy and all that..." (David, interview 2)

Fathers recalled vividly how their children were diagnosed with a CA, despite how much time had passed. They remembered the circumstances and the buildup to their children's diagnoses. Furthermore, many of these fathers identified the abnormal features before healthcare professionals.

6.3.1.2 The response to having a child diagnosed with a chromosome abnormality

Fathers reflected on their experience with the diagnostic process and explored how they exhibited distinct emotional responses upon receiving their child's diagnosis. This was shaped largely by the timing and context in which they learned about their child's CA. While all fathers expressed an emotional response to their child's CA, the immediacy or gradual nature of the diagnosis influenced their reactions. Some experienced a sudden shock immediately after birth, while others underwent a slower, more complex emotional journey that included worry, confusion, and eventual relief as they navigated the gradual discovery of their child's CA. Some fathers discussed the complexity and uncertainty surrounding their child's developmental trajectory or life expectancy.

For fathers who sensed something was wrong immediately after birth, reactions included sudden fear, panic, and heartbreak. When Jamie spoke of the moments after Lucy's birth, he described being physically frightened at the sudden fear that something was wrong with Lucy. He recalled knowing that something wasn't right, and this terrified him:

"It was like a nightmare, a horror show." (Jamie, interview 1)

Similarly, Rory likened the emotional response to being in physical pain. He recalled suddenly being faced with a CA after the elation of a seemingly uncomplicated birth:

"... it was like a punch... a sucker punch." (Rory, interview 1)

Confusion was a common response among fathers who were unfamiliar with their child's specific condition. Many fathers felt bewildered upon hearing the name of the CA. They suddenly felt confronted by unfamiliar medical terms which were extremely challenging to comprehend, particularly when the pregnancies had been normal. However, Jan equally felt a great sense of confusion, but not due to a new medical term. His child was diagnosed with a more common CA that he had heard of before. However, Jan struggled to understand this diagnosis as Jakob did not exhibit the usual physical features of this condition:

"I don't know, I didn't understand it... usually with people with [Jakob's CA] you can see in their face, but you couldn't see anything in his face. He doesn't look like that... I was so confused." (Jan, interview 2)

Fathers expressed particular concern about the sudden uncertainty about their child's future life. For Ali and Paul, the fear related to their newborn baby's survival. Ali recalled the distressing statistic that:

"97% of children with these syndromes would not pass three weeks... [this was a] ... shock, shock, shock." (Ali, interview 1)

However, some fathers were not fearful for their child's survival but instead were concerned about the unpredictability of their children's developmental outcomes due to the rarity of their conditions. Jamie compared Lucy's condition to Trisomy 21, noting that with more common conditions, there is often greater certainty regarding developmental milestones. In contrast, this unknown trajectory of Lucy's rare condition was particularly anxiety-inducing. He also felt that if Lucy had a different diagnosis, the diagnostic process would not have taken so long.

"It's not a very common one, so I'm not sure people with Down syndrome will be having the same issues... because it's so well known." (Jamie, interview 2)

Some fathers discussed the complex process of understanding that something was wrong with their child. David and Peter spoke of how the mother of the child was the first person to identify a developmental delay, which healthcare professionals dismissed. They themselves expressed feelings of guilt and regret for preferring to trust healthcare professionals, sometimes attributing maternal concerns to *"post-natal depression"* (David) or *"crazy post-natal wife."* (Peter). Both fathers then spoke of the journey of beginning to believe the mother of the child, having to push professionals to also believe the mother, and their shared relief when they were finally listened to.

Paul and Jamie spoke of complex decisions that had to be made during the diagnosis stage. Paul was the only father in this study who was offered the option to terminate the pregnancy due to a detected heart condition. He recounted his/their decision to proceed with the pregnancy, stating:

"We were given an option to terminate... but we decided to let the child fight her own battle... we are glad we did that. She is here now, 8 years old." (Paul, interview 1)

Jamie similarly recounted the stress of deciding to undertake genetic testing on Lucy to determine whether her condition was inherited or not. He acknowledged the emotional

toll of the process, as it involved the risk of pointing blame if the condition was found to be passed on by one of the parents. Reflecting on receiving the results, he shared:

“It was quite upsetting getting those results... it was pretty tough.” (Jamie, interview 2).

Overall, these reflections underscore the complex emotional journeys fathers navigate following a diagnosis and the range of emotions experienced, with responses shaped by factors such as timing, the perceived adequacy of healthcare communication, and the rarity of their child’s condition.

6.3.1.3 An emotional journey

The journey of coming to terms with the diagnoses was a significant aspect for many fathers. The specific nature of the children's CA significantly influenced how fathers coped with their children's diagnoses. Fathers spoke of how their mind-set had to change to help them come to terms with this information. For some, they felt they were able to complete this change quickly, with support from the people around them. For others, this took time. They realised that the strategies for managing the CCN, regardless of the time and effort put into them, would not change the fact that their child would still have to navigate the world with a disability. Others used their faith and religious beliefs to help them come to terms with the unknown nature of their children’s futures, especially lower life expectancies. Charity organisations were noted by some to be a helpful source of support, enabling them to come to terms with their children’s conditions. However, not all fathers agreed, and some felt that these organisations were harmful, and made it harder to come to terms with their children’s CA.

When Toby was diagnosed, Mike describes feeling distraught, he phoned his family and was standing on the street crying. A stranger stopped to support him. Mike told the stranger that Toby had just received this diagnosis. The response from this stranger, Mike reports gave him the strength to come to terms with this new journey:

“...she said you have just been given a miracle and you are going to have a child that will love you every single day of your life and all you have to do is learn what

[name of Toby's syndrome] is and the benefits you will get from it is amazing."
(Mike, interview 1).

Having to come to terms with their children's conditions not being 'curable' was something that was difficult for many. They spoke of making plans in the beginning to help their children overcome any difficulties that may arise. Rory recounted his initial plans to improve Jayne's quality of life, only to later realise that this approach was unrealistic. He then had to change his expectations, for both him and his daughter:

"...You know I had these great plans as to how we would fix it, and this wouldn't be a problem because we would just fix her muscle tone by building a gym in the house. And have her in the gym all the time and it is all just illogical and unrealistic actually in reality... [shaking his head]". (Rory, interview 1).

The uncertainty of their children's life expectancies was challenging for some in coming to term with the diagnoses. For both Ali and Paul, who were told that their child may not survive the postnatal period, they both spoke about how their religious beliefs brought them solace, in turn allowing them to accept what might happen is out of their control. For Ali, he feels the path for Zara's life is already written and finds comfort in praying that she will not suffer, not that she will not die:

"... we just pray and that is what keeps us strong. The belief that if she is with us, how long she is with us is already written [by God], so just pray that she doesn't suffer." (Ali, interview 1)

Paul similarly found comfort in pledging to give Caitlin the best life possible, even if that is for a short amount of time:

"Yes, it was scary, but we had a belief in the child - that if she wanted to be here, we would help, but if she didn't, we would be there to give her a good life for a short amount of time." (Paul, interview 1)

Many fathers utilised voluntary/charitable organisations dedicated to their children's specific conditions. Five fathers became actively involved in these associations or support groups, with three taking on leadership roles as chairs or deputy chairs. Others

used support groups more sporadically, attending as needed, for information and support. As one father put it:

"I was there on an information gather for myself to find out what's ahead of us...." (Rory, interview 2).

Conversely, David felt that organisations provided outdated information and were not particularly helpful for accepting Joey's diagnosis. He described the organisation he sought out as promoting a sentimentalised perspective referring to the children with a CA as "Angels", which he did not find practical.

"... it was called Angels at my table... you know, it was that kind of vibe." (David, interview 2).

Some fathers expressed ambivalence about syndrome-specific support groups, finding them to be a "double-edged sword". Peter noted that while these groups could and did provide him with valuable support in coming to terms with Rosie's diagnosis, seeing older children with the syndrome also highlighted its long-term impacts which he found difficult:

"...it's a double-edged sword; you're seeing other kids with [Rosie's condition] that have grown up, and, god forbid the expression, they start to look a little more like the syndrome..." (Peter, interview 2).

Rory reflected on how his relationship with his organisation had evolved. Initially, he sought out the organisation to gain knowledge to help him come to terms with Jayne's diagnosis. Over time, he found that while he no longer needed much support for Jayne from the organisation, he valued the social aspect of it:

"When we first went, it was inquisitive. Now, 13 years later, I'm thinking about Irish All Blacks rugby and how I can watch it in the bar." (Rory, interview 2).

Lucy's condition was rare, to the point that there is no national organisation for support with the condition. Jamie found this difficult and sought alternative resources to help him come to terms with the diagnosis. He eventually discovered a Facebook group that he said provided invaluable insights and a sense of connection:

"... so I found one on Facebook. It's a private group, and from that group, I have learned more than from anything else." (Jamie, interview 2).

The type of CA, and the specific CCN associated with this, impacted on how fathers came to terms with their children's diagnoses. For Mike, when Toby's health status differed from the expected health outcomes for a child with his syndrome, he questioned why his son had such CCN when this was not the typical trajectory of this syndrome:

"You know, he has the Tracky [tracheostomy], the leukemia, hypotonia, that's not all [Toby's CA]." (Mike, interview 2).

While for others, knowing that their children's complexities will only get more challenging over time, were concerned that they had not fully come to terms with their child's diagnosis. Alex questioned this:

"I actually don't know if I am still denying it to myself. Did I really just accept it? Will I ever accept it? Or is it just that we have to live with it?" (Alex, interview 2)

Within this subtheme fathers navigated the emotional journey of coming to terms with their children's diagnoses in complex and deeply personal ways, shaped by the specific nature of the CA, their personal beliefs, and the support systems available to them. For some, acceptance was an evolving process, requiring significant adjustments to their mindset and expectations as they grappled with the reality of their children's conditions and uncertain futures.

6.3.1.4 Professional support and systemic barriers

Fathers described how the reactions of healthcare professionals influenced their experiences and perceptions following their children's diagnoses. For some, this impact

began immediately after birth, while for others, it stemmed from a lack of follow-up response or lack of adequate information after the diagnosis was made.

The manner in which professionals communicated the diagnoses was highlighted. Both Peter and Mike for example, described the over-reliance on medical terminology when explaining their children's conditions which made understanding the diagnoses confusing:

"I don't talk medical jargon, break it down... everything can be broken down if you try." (Mike, interview 1)

Similarly, language barriers compounded difficulties for Jan, for whom English is a second language. He recounted struggling with medical terminology and not fully understanding the information provided. He felt especially misled when signing consent for a procedure, which he later felt this surgery had caused Jakob's CCN (Jakob had a stroke which required surgery to manage). He felt that he should have been offered an interpreter, or some form of reasonable accommodation, to properly explain the risks associated with the surgery.

David felt that during his search for answers, the doctor seemed more focused on the achievement of identifying a diagnosis than on the family's needs, describing his child as akin to a machine:

"I find that they just see the human body as a machine sometimes and don't connect the emotions with it... I think that doctor was just probably chuffed with herself... after finding a fault in the machine." (David, interview 2)

The lack of preparation and time allocated to giving them a diagnosis was noted by some fathers; this was in relation to both inpatient and outpatient appointments. Some noted that the mother of the child was alone while receiving the diagnosis, while others spoke of not being told that they were about to receive a diagnosis. Jamie recalled how his wife was told while she was alone, and how Jamie felt compelled to request another

meeting with the consultant, and, as advised by nurses, to conduct his own research in advance:

“...we had nothing scheduled or anything like that, and someone just walked in and said it's 14 deletion bla bla bla, and mum was like, what am I supposed to do with that information... we had to demand another meeting with the consultant, and, a comment was passed by one of the nurses to read up on it because they won't even entertain you unless you have done your homework on it.” (Jamie, interview 1)

Several fathers mentioned the need to conduct independent research on their children's conditions due to insufficient information provided by professionals. They described hearing the names of the CA and subsequently relying on the internet for further understanding. Many fathers humorously referred to themselves as “Dr. Google”. Ali researched his child's condition, explaining:

“We didn't know what that meant. We had to go and search for it on the internet.” (Ali, interview 1)

Alex also googled his child's condition, where he found an association for supporting Amy's specific CA. He spoke with them and was advised not to continue to look up the condition online. He later spoke about how he found the advice helpful as it allowed him to focus on getting to know Amy. Rory suggested improvements for healthcare professionals in supporting families after a diagnosis, advocating the need for a designated liaison to guide parents through the next steps:

“... when a child is born with [Jayne's CA], there should be a designated person who comes and liaises with the family and tells them where you are and what you need to do next.” (Rory, interview 2)

Systematic barriers quickly became apparent for most fathers in this study. Mostly in relation to disability services in Ireland. Many fathers spoke of having to fight the system particularly in relation to their children accessing support services, namely the CDNT's.

“It’s just another day of fighting the system today. I’ve spoken to disability services (or lack thereof) about finding out why we have NO speech and language therapy, NO physio, NO play therapy and NO music therapy for at least 18 months in [name of CDNT]. I understand that asking for music therapy is a bit of a push, but SLT and physio is a necessity! I am trying my best to do physio but seem to have just got to a certain level and can’t go anymore! I used to get so frustrated working in [name of company] but this is on another level” (Mike, interview 1).

Other fathers spoke of having to fight for cognitive assessments as the waiting lists to see psychology were too long, or spoke of needing to pay privately for such assessments. For example, Peter recalled having to use the syndrome specific support organisation as they had voluntary psychologists who completed cognitive assessments without which allowed his child to go to school.

“...the organisation had to do the cognitive [assessment] a psychologist’s sibling also had [Name of syndrome], so did the whole thing for free.” (Peter, interview 1)

Overall, these accounts depict the varied impacts of healthcare communication, with many fathers feeling underserved by professionals and compelled to seek out information independently, often facing additional barriers in the process.

6.3.2 Theme two: The complexity of relationships (lived relation)

This existential theme of “lived relation” highlights how having a child with a CA, can increase the complexity of their relationships thus shedding light on the relational dimensions of their experiences in raising a child with a CA. Within this existential theme, five subthemes emerged that included the relations fathers have with others in caring for a child with a CA.

The first subtheme shows the positive and unique bond between fathers and their children as the distinctive characteristics associated with their children’s CA often bring profound joy and pride. Within the second subtheme, fathers report that while the

mothers of the children frequently serve as crucial sources of emotional and practical support to them, mothers can also be a cause for stress and relationship strain. Within the third subtheme, fathers express deep concerns about the emotional impact their children's disabilities have on siblings, both in daily life and with regard to the long-term responsibilities these siblings might assume in the future. Within the fourth subtheme, fathers explore their relationship with the children's grandparents, expressing both appreciation and frustration in their varying levels of support. Within the final subtheme, fathers convey anxieties about their children's social interactions, friendships, and the potential challenges these relationships may pose.

6.3.2.1 Navigating multiple bonds- relational balance of fatherhood

Fathers articulated their experiences of fathering a child with CACCN in relation to their bond with that child, while also situating this relationship within the broader context of their roles as fathers to the child's siblings. All of the fathers in this study had other children without a CACCN.

Despite the fear and worry that comes with having a child with a CA, a profound sense of love, pride and fun emerges in every father's account of their lived experience with their child. The strong bond these fathers share with their children is evident. Some fathers attribute an enhanced connection with their child to unique characteristics associated with their CA.

Their children's personality traits, often associated with their specific CA, bring not only joy to these fathers but also to their wider communities. Peter reflected on the social ease associated with Rosie's CA saying:

"Yeah, well it's the feature; she's hypersocial, so she's really charming. She's a joy, really open, and she's lovely to bring places because people just light up when they meet her..." (Peter, interview 1)

Similarly, the strong will that is often linked to Amy's CA, is one of the characteristics that Alex admires and loves about her. He described her as extremely driven and caring:

“Extremely driven... She is very funny, very loving, caring for everyone... and I wouldn’t change her for a minute.” (Alex, interview 2)

The pride fathers feel in witnessing their children overcome challenges, whether medical or developmental, is palpable. Mike spoke with pride about Toby’s resilience and medical achievements, explaining:

“... because he is a miracle... the things he has done, I mean he has trained student doctors to be doctors... he got through his cancer therapy quicker than any of the doctors could imagine. Let’s give him a little bit of credit, let’s show him off to the world. You know, see this boy, he is my boy.” (Mike, interview 2)

Other fathers shared similar pride in their children when they met their developmental milestones, regardless of timing. Alex spoke about Amy and his happiness each time she reached a developmental milestone:

“... it’s all the little things, like sitting up, eating solid foods, all the milestones. Even though it is plus one year or plus six months, you’re still happy about it.” (Alex, interview 2)

However, alongside the fun and joy their children with CACCN bring, fathers equally expressed guilt, sadness, and concern over how their child’s condition may have affected their other children’s lives. Many fathers regretted missing aspects of their other children’s childhoods due to the demands of medical care and hospital admissions. Others worried about the premature responsibility their other children had to take on, or may have to take on, as caregiver for their child with a CA.

Fathers expressed feelings of both guilt and sadness at having to choose between their children, feeling firstly that they would not want to leave their child while they are in hospital, but equally they were acutely aware of what they were missing out on with their other children as a result. Mike expressed a sense of loss over missing out on pivotal moments with his other children, expressing:

"I regret... that I really missed out on being the dad I should have been to them..."

(Mike, interview 1)

Paul also felt this same regret and heartbreak of feeling torn as he was the primary carer for a long hospital admission. He elaborated on how this initially caused him to feel spending so much time in hospital with one child would affect his bond with the others, as they would look for their mother over him. However, he explained that this changed once he came home, and that now they no longer favour their mother since he has been able to spend time with them:

"They were more prone to their mother back then. But now, they seem to be more prone to me. They try to spend as much time with me now, especially the older one." (Paul, interview 1)

Fathers also reflected on how the complexities associated with their children's conditions impacted their other children. The psychological toll of medical emergencies on siblings was a significant aspect to some. The worry for siblings' emotional wellbeing during these critical moments was a shared concern among several fathers. Jamie recalled a traumatic incident where an ambulance was called due to seizure activity, and how he noted the effect this had on Lucy's sibling:

"... the impact this had on her... And people don't realise it enough, to be honest. Things like that were so scary for her [sibling's name]..." (Jamie, interview 2)

Similarly, Mike noted that one of his children managed by wanting to learn as much as possible about Toby's medical needs, to the extent of becoming proficient in using medical equipment. Although he was proud of his daughter, this also led to anxiety for Mike, knowing that this was too much responsibility for a child. This caused her to be constantly fearing for her brother's life, which affected her mental health. Mike remembered a time where she said:

"...no one understands what it's like having a sick brother that may die at any moment..." (Mike, interview 2)

Abi shared similar concerns about how the uncertainty surrounding his daughter Zara's condition impacted her siblings, in which he acknowledged the emotional toll of living with the constant possibility of loss. In the beginning, when his daughter Zara was in hospital and they were told that she would likely not survive, he was able to protect his other children somewhat. But now, the love Zara's siblings have for her means he feels when or if she dies, this will be harder for the children. He noted:

"If she were to die now, it would be much harder on the whole family than if she had died as a baby." (Ali, interview 1)

Both Rory and Alex spoke of the concern around food for their other children. They had to make decisions around locking presses, what treats their other children could or couldn't have, and how making these decisions fair was difficult.

Some fathers observed that their other children had to mature quickly, adapting to their sibling's needs. This early maturity, while necessary within the family unit, also meant that these siblings took on responsibilities typically reserved for adults. Paul described how his children had developed a heightened awareness of their sister's condition, taking on parental roles:

"We never shielded it from them. Mentally, they suffered as well. They are now not your normal 15- or 16-year-old... they took a teenager's role before they were 12." (Paul, interview 1)

For many fathers, the prospect of their other children taking on caregiving roles in adulthood was a source of enduring guilt. When discussing Jayne's future, Rory explained that his biggest worry was not for her, but rather that of her siblings. His fears were that the burden of care would fall to them:

"My biggest concerns weren't for Jayne it was for the siblings... I don't want the weight or burden of Jayne to fall back on her sibling's shoulders." (Rory, interview 2)

Similarly, Peter explained that he even considered the possibility of having another child solely to alleviate some of the sibling's future caregiving responsibilities:

"... It's a big ask, and I have terrible guilt that it might be on him... And that is a terrible reason to want to have a child because I feel we are overburdening him..."

(Peter, interview 2)

Through these reflections, despite their challenges, fears and concerns, these fathers experience immense joy and pride, finding strength in their children's unique qualities and accomplishments. This sense of love shows the unique bond and the positive relationships these fathers have with their children. Yet, fathers are acutely aware of the potential long-term impact a child's CACCN has on their siblings.

6.3.2.2 Fathers' relationships with the mothers of their children

Fathers' relationships with their children's mothers are complex and are impacted by the demands of caring for a child with a CA. Some fathers felt a need to protect the mother by downplaying the severity of their children's conditions, while others found their partners to be the stronger figures in managing the diagnosis. Some fathers reported that the stress sometimes brought them closer, although for some it ultimately contributed to marital strain. Overall, fathers discussed the unique personal choices and preferences in managing the diagnoses, and how this difference affected their relationship with their partner.

Some fathers attempted to protect the mother of their child from the reality of their child's diagnosis, feeling it was their role to be protective. Alex recalled:

"And my wife, you know, I was trying to mind my wife as well and tell her nothing was wrong..." (Alex, interview 1)

Rory, however, saw his partner as the stronger one, admitting that he was the more emotional of the two, and that he relied on her for this:

“... she broke down that night [when receiving the diagnosis] but that was kind of the only time. She is very strong... of the two of us, I would definitely be the more emotional.” (Rory, interview 1)

For some, the challenges associated with the diagnosis led to marital strain. Mike attributed part of his marriage’s breakdown to difficulties that his wife had bonding with Toby after receiving the diagnosis. This subsequently led to him taking on much of the caring role, and contributed to the marriage ended:

“That’s when it all changed... mum found it very, very hard to bond. She really, really did. And whenever we talk about it now [mother’s name] say’s it was the worst day of her life to get that news.” (Mike, interview 1)

Differences in management strategies and opinions on the best way to move forward following the diagnoses caused some strain. Jamie described a disagreement with his wife regarding his use of counselling to process the diagnosis. His wife did not believe he should go to counselling, so reluctantly, this resulted in him no longer attending:

“I had to go to counselling in the hospital, [mother’s name] didn’t like me going... I found it useful but she didn’t like it, so I stopped... she doesn’t believe in it.” (Jamie, interview 2)

Differences also were noted by some fathers in the use of the associations and support groups. Some found that they had different interests with regards to both joining and taking on leadership roles within the groups. Rory recalled that he felt more value in engaging with a support association than his partner did, sharing that:

“She didn’t like to deal with the association or anything.” (Rory, interview 2)

Peter remarked that he did not feel his child’s CA was the cause of any marital strain as any child, not only those with disabilities, can change a relationship. He emphasised that parenthood is relentless, but that ultimately, his relationship with the mother has become closer and they work better together as a team.

This subtheme highlights the complexities of fathers' relationships with their children's mothers, shaped by the challenges of raising a child with a chromosomal abnormality. Fathers described varied dynamics, from protecting their partners to relationship strain.

6.3.2.3 Fathers' relationships with grandparents

Grandparents played a complex role in supporting fathers caring for children with a CACCN. For some, grandparents provided crucial support, especially during extended hospital stays, but others experienced disappointment or hurt due to grandparents' lack of understanding or acceptance of their child's condition.

For those that received support, the children's grandparents (both their parents and the mother's parents) were essential to managing daily life, particularly in relation to looking after their other children during hospital stays and appointments. Jamie relied on his parents to look after Lucy's sibling during hospital admissions, saying:

"They helped a lot, she [sibling] would have to stay with my parents a lot." (Jamie, interview 1)

However, others were cautious about over-relying on grandparents, fearing they would exhaust their goodwill. Fathers also worried that the physical aspect of care may be too great for grandparents, given their advanced age. Peter expressed this concern, saying:

" But we don't, unless we really have to because (A) We don't want to abuse it and (B) we feel like you only want to wait until a proper emergency. It's like it's a depletable resource and you don't want to abuse it... and then physically, when she drops to the floor [due to a behavioural outburst], I can pick her up, my mum can't." (Peter, interview 1)

Furthermore, some fathers did not feel support from grandparents at all, and they perceived this lack of support as a lack of interest or understanding from the grandparents. Rory described his frustration, saying:

“Granny really doesn’t get it; she doesn’t ask questions, she isn’t interested...”
(Rory, interview 1)

For Mike, the emotional distance shown by Toby’s grandparents was particularly painful. He recounted an incident where he was extremely hurt by the language used by Toby’s grandfather, who made a comment that reflected a deeply insensitive perspective, which compounded Mike’s feelings of isolation:

“... And I always remember his line, because he was a small farmer, that if Toby was a calf, I would have put it down by now.” (Mike, interview 2)

Abi decided not to inform his own parents about his child’s diagnosis at all, as they did not live in Ireland. He explained that due to the distance and the cultural differences, by telling his parents what they were going through, he felt that he would bring more hurt to himself and his family aboard. He explained:

“... my parents would not understand [Zara’s condition]... We didn’t want other people in the family to suffer. If they are that far and every time the phone rings they would think [she died]... yeah, we decided not to.” (Ali, interview 1)

Overall, the relationship with grandparents presents emotional complexities in how fathers navigate family support. While some found grandparents to be invaluable, others struggled with the pain of unmet expectations or insensitivity.

6.3.2.4 Fathers’ concerns for their child’s interpersonal relationships

Fathers relayed how their children’s interpersonal relationships outside the family home were a source of anxiety. Some emphasised their concern in relation to making and maintaining friendships. Others worried about romantic relationships, particularly in relation to their intellectual disability and understanding of consent.

Fathers’ own anxieties for their children’s future loneliness were apparent. Peter’s biggest fear for Rosie was exactly that:

“My biggest fear is she won’t have friends.” (Peter, interview 1).

Rory wrote in a journal entry relating to his worry for the loneliness that Jayne may feel in a community where she is supposed to belong:

"It is clear that [Jayne's CA] is a lifelong condition that brings new challenges as the individual gets older... Loneliness in her own community, lack of opportunities in life, lack of integration, are all concerns for the future for Jayne." (Rory, Journal entry)

Although fathers all expressed their hope that their children will have typical friendships and relationships, they equally worried for their safety, particularly in relation to consent and understanding. David recounted an incident at one of the association conferences that led him to worry, for both the girl involved and Joey's safety:

"... I found him kissing in the bathroom... So I was like... What's going on here? He said... She's my girlfriend... So we did a kiss... It's kind of cute and all that... but it's also kind of... it's kind of difficult, you know, because, you're trying to explain to him like, look, she's not really your girlfriend... his comprehension of the whole situation is not there. I know it's not there, so if he was to figure out, you know, how to make babies. He'd probably just go for it. The fact is his girlfriend has an intellectual disability as well, so. You know... it's really complex." (David, interview 2)

Similarly, Rory wrote:

"Jayne is starting to talk about a 1st year in a mature way, boyfriend/girlfriend. It's part of life but not easy to manage... She has become very independent, but this means it is much more difficult to influence her and look after her welfare." (Rory, Journal Entry)

6.3.3 Theme three: The physical and psychological space occupied by fathers (lived space)

This theme displays the existential theme of "lived space", that explores the physical and mental space that caring for a child with a CACCN occupies in the lives of fathers. It captures the space they inhabit and the psychological landscapes they navigate as a

result of caring for a child with a CACCN. This dual occupation of space, both physical and psychological, highlights the profound impact of caregiving on fathers' lived experience.

Four subthemes have been identified. In the first subtheme, fathers described the realities of long hospital stays and the emotional challenges associated with this. In the second subtheme, fathers recount the subsequent adjustments needed upon returning home. These adjustments in lived space extend beyond physical adaptations; they also encompass the mental and emotional space these fathers dedicate to their caregiving responsibilities. The third subtheme shows that homecare packages, respite and residential care are vital in supporting fathers, in both current and future care. In the final subtheme, fathers explore the complex decisions and the emotional toll these decisions take on them.

6.3.3.1 Managing hospital life

The complexity of having their child in hospital was a significant challenge for many. The existential theme of "lived space" in the context of fathers' experiences with a hospitalised child, highlights the physical, mental, and emotional landscapes that are shaped by the demands of hospital care. Fathers described the complex logistics of balancing work and family, resulting in them feeling stretched across multiple responsibilities.

Paul recalls the fear of leaving the hospital that something would happen, and that Caitlin would die. He felt he needed to be constantly vigilant, which he found emotionally exhausting:

"... we were told she could go any time... if she was going to go it would be very quick, and someone needed to be there, so I spent most of my time in the canteen hospital and got food there." (Paul, interview 1)

Some fathers felt guilt or frustration over their inability to stay with their child all day, due to work and home duties. For others, such as Mike and Paul, staying at the hospital became a priority, yet this arrangement caused them to feel distanced from their partners. Mike explained that they were:

".. like ships in the night."(Mike, interview 2)

Ali shared the daily rhythm of managing multiple roles and responsibilities, reflecting how caregiving logistics while Jakob was in hospital, resulted in so much stress that he relied on alcohol to get through it. While his wife spent most of the time in the hospital, he managed their other son and work:

"Basically, his mum would be there [the hospital] from morning to night, and then I would switch out. I bring the kids to school and bring them back... she was in the hospital most of the time... I drank a lot." (Jan, interview 1)

Paul highlighted how power dynamics within hospital settings amplified his stress and emotional strain when he was already feeling under pressure. He recalled an incident when a nurse reprimanded him for not being present enough while he was away with work, leaving him feeling punished:

"... I was at a work event and the nurse came in and said that I wasn't there enough... that they were taking the chair. And I just flipped, I drove into the hospital and was like I don't know who you think you are, she's doing her best, I'm doing my best, we are in an absolute mess here, and you're treating us like children taking the chairs out of the room. I'll never forget it." (Jamie, interview 1)

The need to rely on other people for basic needs while in hospital was noted. Having to ask to use the toilet or go and get a coffee was something that Jamie recalled. For many, the inadequate accommodation further intensified the physical and emotional toll. Jamie expressed disbelief at the poor conditions:

"The accommodation was terrible; it was like being in a third world country." (Jamie interview 2)

In contrast, Mike appreciated the comfort provided from a charity that housed parents of children with CCN while in hospital, which contributed positively to his experience:

"... it was comfortable, you were looked after." (Mike, interview 1)

Financial strain was another significant concern, with costs for food, lodging and transport adding up quickly. Paul estimated an additional €400-€500 per week due to travelling to and from the hospital. This financial impact often compounded the worry and uncertainty that fathers faced as they navigated both the immediate needs of their child and the larger family's well-being.

The overwhelming nature of the hospital environment also took up considerable mental space, with fathers recounting feelings of helplessness with the constant influx of medical interventions that their child needed. Jan described the confusing experience of watching various medical procedures without fully understanding what was happening. This lack of clarity created a sense of psychological confinement, with fathers feeling trapped:

"They just kept bringing in different syringes, different medicines..." (Jan, interview 2)

Overall, the fathers' accounts highlight the significant emotional, logistical, and financial challenges of navigating their child's hospitalisation while maintaining their family and work responsibilities. The existential theme of "lived space" underscores how hospitalisation reshapes fathers' physical, mental, and emotional realities. Many fathers struggled with logistics, feeling stretched between multiple responsibilities.

6.3.3.2 Coming home from hospital

For many fathers, adjusting the home environment was essential to accommodate their child's needs upon returning home from the hospital. This process involved practical, emotional, and logistical challenges, and fathers often reported feeling that they had inadequate external support.

For most, returning home from hospital was a joyous occasion, but for Jan, this was bittersweet. Returning home with Jakob after his stroke was emotionally challenging as Jakob no longer recognised his home. This response was particularly painful for Jan, as it disrupted the sense of normalcy that they hoped to restore in bringing their son home:

“When we bring him back home, he wasn’t happy; he was crying... he didn’t recognise the place.” (Jan, interview 1)

Jan, like many fathers, discussed the need to complete renovations before being discharged from hospital. Jan recalled that when returning home from a long hospital admission with Jakob, who was now a full-time wheelchair user following a stroke, the physical environment proved challenging. Jan faced structural limitations when trying to install a shower in their downstairs bathroom as Jakob was no longer able to access the upstairs of the house. Despite this urgent need to provide a space where Jakob could be bathed, the size of the physical space prevented them from qualifying for financial assistance as the bathroom would not meet building regulations, which resulted in the family having to complete construction work themselves:

“The hardest part at the beginning was the bath... the area was too small to get a grant, so we just changed it ourselves.” (Jan, interview 2)

Fathers recalled the need to change rooms around in their homes, often medicalising the home environment. Some fathers spoke of having to put hospital beds in kitchens and sitting rooms, as these were the only spaces in the home that could hold the equipment necessary for their children to sleep, which led to a lack of space for the rest of the family. This affected families living both in social and private housing. Paul also spoke of the logistical challenge of storing medical equipment, occupying significant space in their home:

“We have a shed full of equipment...her bed was in the kitchen.” (Jamie, interview 2)

The geographic distance of their homes to the hospital added a layer of constant, underlying fear. Ali expressed how he was feeling anxious about leaving the hospital, as he worried that if something was to go wrong or Zara had a medical emergency, an ambulance might take too long to arrive. He feared something would happen that the family could not manage without medical support:

“An ambulance would take 20 minutes... she was very weak, and we didn’t know what would go wrong.” (Ali, interview 2)

Mike also shared his challenge with living far from Dublin and his perceived feeling that Dublin hospitals have a better ability to handle Toby’s condition. He had a lack of confidence in their local hospital’s ability as previously they had not been able to manage his son’s tracheostomy. This resulted in them feeling they needed to bypass their local hospital and travel across counties to a national children’s hospital. This led Mike to feel guilty about not wanting to travel to Dublin, and he spoke about sometimes slightly delaying a hospital visit due to this, hoping to manage at home with nursing support:

“You can get complacent sometimes, I felt terrible... I just do not want to have to go there.” (Mike, interview 2)

Two fathers expressed their worry for their future home environment, their children have hyperphagia (chronic hunger) which can result in obesity. Both fathers discussed their worries about this condition escalating and their need to modify their home environment by locking kitchen presses preventing their children having access to parts of their home. This brought them both anticipatory worry and guilt.

The transition from hospital to home presents fathers with significant emotional, logistical and practical challenges as they adapt their environments to meet their child’s medical needs. Structural changes, such as renovations for mobility aids, often place financial strain on families and disrupt routines. Distance from specialised care compounds these challenges, with fathers fearing emergencies and experiencing guilt when relying on local hospitals with limited expertise. Anticipatory concerns, such as managing conditions like hyperphagia, further illustrate the tension between ensuring safety in the home and fostering independence.

6.3.3.3 Homecare packages, respite and residential care

Five of the nine children in this study received a HCP. These HCP support the discharge of children with CCN from hospital to home, providing families with nursing and healthcare staff in the home environment. For these fathers, this served as a vital but sometimes challenging element of support. These HCP are essential for providing respite and hands-on care and brought both relief in alleviating some of the caring roles but were equally sometimes a cause for concern for these fathers.

Some expressed that there was a lack of communication from hospital staff about what services their children were entitled to. Ali recalls being frustrated that Zara was discharged home from hospital without availing of homecare support, which meant that her family had to look after all her medical needs, which her father said needed to be monitored 24 hours a day. This unfortunately led to exhaustion and Ali recalled that they were no longer able to manage, and Zara was readmitted to hospital while they waited for a HCP which should have covered 7 nights of nursing:

“The Palliative Care team got involved, but they never told us that Zara needed support 24/7... we had to do it alone... It took about eight months before she was fully home. And even then, it was like only ¾ nights covered.” (Ali, interview 1)

The decision to accept a HCP itself was often emotionally difficult. While some fathers initially resisted this package, wanting to retain autonomy over their caregiving, they later acknowledged the indispensable role of these services. Paul admitted that initial emotional resistance gave way to the realisation of the practical need:

“We got a little bit stroppy... we don’t need nurses, but... coming home, we weren’t prepared. We realised it’s going to take a city.” (Jamie, interview 1)

However, despite agreeing that they needed these HCP, the continuity and quality of the care provided by them presented ongoing concerns. Jan described the difficulty in constant nursing roster changes which meant a different nurse would arrive to take care of Jakob each night. He reflected on the persistent anxieties about entrusting their child’s care to unfamiliar staff members.

“People keep leaving the job...” (Jan, interview 2)

HCP also resulted in fathers feeling they lacked freedom. The legal requirement for a second trained person to be present when a homecare provider was in the house limited families’ ability to leave the home, which curtailed a sense of normalcy and independence. Jamie articulated this frustration as he felt that while respite allowed both himself and his wife to sleep, he equally felt that the respite was inadequate without the ability to leave the family home:

“There is respite, but there is more to life than sleep... people need a life.” (Jamie, interview 2)

Beyond immediate care, future planning for the child’s long-term needs is a significant area of continuous worry for several fathers. Some actively planned to adapt their living situations as the child grew. Peter strategically chose a home with adaptability in mind, anticipating future modifications for independent or semi-independent living:

“We specifically bought somewhere that we could change or convert for some level of independent living.” (Peter, interview 1)

Many other fathers discussed the need to plan for residential care facilities. For instance, Alex spoke of the need for specifically trained residential houses. He referenced stories from people within the specific association for his child’s CA, who were left vulnerable after the deaths of elderly parents:

“We hear of adults with [Amy’s condition] that when they [the parents] die, they [adults with Amy’s condition] are lost...” (Alex, interview 2)

In this subtheme, fathers noted their initial resistance to accepting HCPs, driven by a desire to maintain autonomy in caregiving. However, as the demands of caring for a child with CCN became apparent, many came to recognise the necessity of this support. Despite this, the perceived inadequate communication about entitlements and delays in securing HCPs, exacerbated family exhaustion and stress. Furthermore, frequent

changes in nursing staff undermined trust and stability in care delivery, while the legal requirement for a second trained person limited family mobility and independence. Future planning emerged as a persistent concern, with fathers anticipating the need for home adaptations or residential care to ensure their child's safety and independence as they grew older.

6.3.3.4 Mental space involved in decision making

The complex decisions involved in parenting a child with a CA is a fundamental aspect of care for these fathers. Fathers described making difficult decisions such as agreeing to “Do Not Resuscitate” (DNR) orders or consenting to surgeries with high risks of adverse effects. These choices required careful consideration of their child’s comfort and the impact on the family. Daily decisions, particularly around feeding, were also significant. Furthermore, decisions around school placements were often challenging.

Paul reflected on the numerous challenging decisions he faced, beginning even before Caitlin's birth. When offered the option to terminate the pregnancy, he and his wife unequivocally decided against this course, a decision that came naturally to them. However, the complexity of decision-making became immediately apparent after Caitlin’s birth, when she needed to be transferred to a different hospital due to her critical condition. At the same time, Paul’s wife required surgery, leaving him in a difficult position to decide whether to accompany his newborn or remain with his wife. Ultimately, he chose to stay by his wife’s side, meeting Caitlin the following morning. Decisions about surgery were also challenging, which led him to constantly weigh her comfort against the slim chance of survival. Only after Caitlin was home, did he fully comprehend the gravity of his decisions:

“In all her surgeries, we were given 20 percent of her actually coming out of it... We didn’t sit down and process it until we were home... and we were shocked and surprised that we had forgotten all that had happened.” (Paul, interview 1)

Mike reflected on the profound emotional and ethical challenges he faced when consenting to a DNR order for Toby. This decision was further complicated by the urgency of the situation, as it had to be made over the phone due to Mike’s inability to

reach the hospital in time for Toby's emergency surgery. The complexity of this decision was shaped by competing concerns - his desire to ensure Toby did not suffer, while also grappling with worries about Toby's mother and their other children. Ultimately, Mike prioritised Toby's comfort, stating:

"I didn't want him to be in pain or suffering... I was worried about mum... and the others... but we signed it." (Mike, interview 1)

Healthcare decision-making can be further complicated by a lack of trust in healthcare professionals, as evident in Jan's experience following Jakob's stroke. Feeling let down by the healthcare system in Ireland, Jan sought a second opinion and pursued alternative therapies in Poland, making this decision against the medical advice provided in Ireland. He believed this choice significantly improved Jakob's quality of life, particularly through the introduction of oral feeding, which had been deemed unsafe in Ireland. Reflecting on this, Jan noted:

"Here they wouldn't allow it... he has a PEG (Percutaneous Endoscopic Gastrostomy), but in Poland we had a physio with a speech therapist who showed us some massages for him and slowly, slowly so he is getting three meals. Two through the PEG and one through the mouth." (Jan, interview 1)

Like Jan, feeding decisions emerged as a significant theme for many fathers, reflecting both the medical and ethical complexities involved. For others, like Rory, these decisions were shaped by the unique challenges associated with specific symptoms, such as hyperphagia. Rory expressed growing concerns about external influences on food choices as his daughter Jayne matures, particularly considering the Assisted Decision-Making (Capacity) Act 2024. He highlighted the legal and practical difficulties he anticipates when Jayne turns 18, noting his inability to control her food intake under the new legislation:

"You're losing control... which is critical for a person with [Jayne's CA]... she has full control of everything she wants [talking about food], and we have to fight her"

legally to keep her healthy... it's a minefield, an absolute minefield." (Rory, interview 2)

Rory also critiqued the framing of decision-making legislation, particularly its emphasis on autonomy, which he argued overlooks the practical realities and cognitive challenges associated with certain conditions:

"The ads [advertisements] are quite inflammatory actually [the decision-making bill advertisements]. They mean well, they are by the person's [with Jayne's condition] perspective. I think they are, as much as you would love to have in an ideal world. Unfortunately, with some of these conditions, they live with challenges, emotionally, intellectually." (Rory, interview 2)

Educational decisions were also a source of concern. Many fathers sought inclusive education, but the process was often fraught with institutional barriers. Jamie shared the difficulty of securing a mainstream school placement for his daughter Caitlin, recounting how he had to advocate against professionals who resisted mainstream integration. Paul worried about the long travel times if his daughter were sent to a "special school" as she would have to get a bus to another county stating:

"So we were being put into huge pressure regarding school... the school is a 40-50 mins drive so she would have to get a bus on her own..." (Jamie, interview 1)

David spoke about the choice for mainstream schooling came from a want for his child to go to the same school as his sister. David spoke of being afraid that Joey would be sent away for education, away from his community. He recalled:

"I remember not wanting a big white bus... taking him away to a special school. Away from his community..." (David, interview 1).

Mike spoke of how Toby's medical complexities impacted his educational decision. Toby was academically able for mainstream, and Mike did not want him to miss out academically, but if he went to a special school, he would have a nurse to support his

medical needs. Mike explained how Toby ended up going to a mainstream school as part of a HSE initiative which allowed their homecare nurse to go to school with him. Mike described his struggles with interagency policies regarding nurse funding in mainstream schools. He eventually threatened to publicise his story to secure Toby's right to education accommodations, stating:

"... I have written to the Irish times [national newspaper] ... And all we want is a nurse, so I sent that letter and two hours later we were given a nurse." (Mike, interview 2)

These fathers discussed complex decisions spanning medical, ethical, educational and legal challenges. From agreeing to DNR orders and high-risk surgeries to navigating feeding complexities, they explored their need to balance their child's quality of life with family needs. Similarly, education choices involved wanting inclusive schooling while addressing practical concerns like travel, medical support and funding.

6.3.4 Theme four: The material aspects of care (lived things)

Fathers spoke about the impact of material aspects of care, covering the material items that they deemed necessary to care for their child. The first subtheme related to the financial implications of everyday care needs, and the financial worry for their children's futures. The second subtheme explores how balancing work with their care demands posed significant challenges for fathers.

6.3.4.1 Financial implications

Fathers caring for children spoke about the financial implications of caring for their child. Many expressed gratitude for the Irish healthcare system and the supports provided, such as medical equipment, allowances, and tax credits, though some felt the financial aid for daily care needs was insufficient. Fathers also spoke about the future financial implications for their children.

Many fathers appreciated the state's support in relation to equipment. Jan was particularly grateful as he felt that if Jakob's stroke had happened in Poland, they would have been responsible for supplying medical equipment:

“You know in Poland you would get nothing...here we got everything, bed, wheelchair, even that machine... 7k euro or something.” (Jan, interview 2)

Peter appreciated the Domiciliary Care Allowance (DCA) and how it facilitated upgrades to his home through the warmer home scheme. Mike believed that the family got everything they needed from a medical equipment perspective to provide care for Toby. He said:

“The support we have had from the HSE is second to none... Anywhere else in the world, you wouldn’t get the support we have had.” (Mike, interview 2)

However, Jamie highlighted hidden costs of essential supplies that were not covered by the HSE. Although he felt that they received some medical equipment, healthcare items that supported Lucy’s basic needs such as hygiene wipes were not covered. These were items that, had Lucy not had a CACCN, the family would not need to buy:

“They give you nappies but no wipes... when they all come together its expensive...” (Jamie, interview 2)

Planning for future finances was a significant aspect of father’s worries for their children. Fathers discussed their own mortality and the anxiety that this brings from a financial perspective. Many fathers explored the need to have a will and to plan for their children financially when they are no longer able to care for their child. This was challenging for some, as they were conflicted in knowing that their child with CCN would need financial protection which may cause challenges for siblings. Jamie explained:

“And it’s hard, because I have, I have other children...but I have to ensure Lucy’s future is boxed off if she is not self-sufficient, which will be hard for them.” (Jamie, interview 2)

Similarly, David expressed the guilt he had for having to have a trust fund for Joey and not for the other siblings, as he felt that his other children would be able to look after themselves financially. However, he also felt that fundamentally, this was unfair on his other children. He equally expressed his concerns that if he did leave money for Joey, that due to his intellectual disability, this could leave him vulnerable to being scammed:

“There's guilt associated with that as well [having a trust fund for Joey] but again, it comes back to those two [Joey's siblings] can definitely forage for themselves. People with an intellectual disability in particular are susceptible to kind of scam artists, and if they've got a lot of money. He could be scammed.”
(David, interview 2)

In summary, caring for children with CA frequently highlighted the financial implications of caregiving, acknowledging both the support provided by the Irish healthcare system and the ongoing challenges they faced. Many expressed gratitude for medical equipment, allowances, and tax credits, with some noting that such support would not have been available in other countries. However, hidden costs, such as essential supplies and private healthcare expenses for therapies not covered by the state, added significant financial strain. Planning for their children's future financial security was a major concern, often compounded by worries about their own mortality and the potential impact on siblings. Fathers grappled with the need to ensure financial stability for their children, balancing this against feelings of guilt and fairness toward their other children.

6.3.4.2 Balancing work with care demands

Balancing work with caregiving responsibilities was another common theme. Initially, all fathers were working full-time. Some took time off when their child was hospitalised while others had to continue to work. Some fathers spoke of the stress they felt daily being the financial provider, while others reflected on how they felt guilty at having to miss some of the care-taking roles as a result of being in work. Some fathers felt pride in how they balanced their caring roles with work.

Jamie described the pressure he felt the whole family were under while he maintained dual roles as both an employee, and carer for a child with a CACCN:

“The whole house is like a pressure cooker constantly... add the complexity of being a carer and working.” (Jamie, interview 1)

Aside from the pressure at home, fathers documented feeling torn between these two roles. They felt that by returning to work, they were not able to attend key medical

appointments, which they often regretted. Alex spoke of feeling guilty for missing appointments due to being away with work. This also led him to struggle to understand his daughter's speech, which others at home could comprehend:

"I probably went to one or two... because I just wouldn't have been around... I felt bad... The others in the house knew what she was saying and I was like, I haven't got a clue... it made me feel like crap." (Alex, interview 2)

Some fathers, however, were able to change roles in work, enabling them to be present for medical appointments. Rory was able to rearrange his work schedule to attend all appointments, feeling proud of his involvement:

"In terms of being the dad and being the provider, I just jumped into that role, not that I saw a lack in my wife, it is just a natural instinct to jump into that... I did the clinical appointments or any of her home therapies, any time she had an appointment I arranged that it was a day that I was off..." (Rory, interview 1)

Balancing work and caregiving responsibilities was a significant challenge for fathers, with many feeling torn between their roles as providers and carers. While some took pride in managing both roles effectively, others experienced stress and guilt, particularly when work commitments caused them to miss medical appointments or caregiving moments.

6.3.5 Theme five: Adapting to their children's CACCN changing over time (lived time)

Fathers have subjective experiences of temporality in the context of caring for a child with a CACCN. This theme captures the multifaceted nature of their daily lives, encompassing the management of medical responsibilities, addressing their children's mental health and behavioural challenges, and navigating the demands of employment alongside caregiving. The first subtheme, "things are okay for now," reflects how fathers adapted their mindsets in response to their children's evolving CCN particularly as changes in life expectancy reshaped their perspectives. The second subtheme examines the development of fathers' practical caregiving skills, illustrating how they gained proficiency and confidence over time as their children's needs evolved. The final subtheme underscores the dynamic nature of chromosomal abnormalities, highlighting

the necessity for fathers to continually adapt their caregiving roles in response to the changing demands of their children's conditions.

6.3.5.1 "Things are ok, for now..."

Many fathers recounted the evolution of their mindset over time, as they adapted to the demands of parenting a child with a CACCN. For some, the immediate medical challenges diminished as their child grew, but an underlying sense of impermanence and anxiety persisted.

Jamie described his approach with the phrase "for now," emphasising its importance in his daily outlook. This refrain, that was repeated throughout his interviews, reveals a continual anticipation that Lucy's stability may be temporary. He explained:

"We use the term 'for now' a lot... everything is ok - for now." (Jamie, interview 2)

Similarly, Paul articulated that as a result of being told that Caitlin may not survive, he now has an aversion to future planning and focuses instead on the present. This anxiety remained a constant factor for Paul, despite the fact that Caitlin's risk of death has reduced:

"We had a thing that we wouldn't look to the future. Everything was for now, every minute we had counted... Our future was non-existent. It was this hour, then look to the next hour... Now I suffer with anxiety and depression. But say I was going to plan anything; I can only plan 48 hours before because of my anxiety." (Paul, interview 1)

Over time, some fathers reported a shift from short-term to slightly longer-term thinking. Ali noted this transition, describing a gradual change in his outlook. He describes this change as:

"You realise, ok we probably have more than an hour now, we can plan. You suddenly have a change in mind-set." (Ali, interview 1)

Many fathers described how their mindset evolved over time as they adapted to the challenges of raising a child with a CACCN. While immediate medical crises often lessened as their children grew, an enduring sense of impermanence and anxiety remained.

6.3.5.2 Developing skills and confidence to care for their child

Fathers explored aspects of their children's care that initially caused fear and anxiety but over time, they began the process of gaining proficiency and adapted to complex caregiving responsibilities. Jamie reflected on his early confusion over Lucy's medical equipment which gradually gave way to practical skill acquisition, but not without moments of self-doubt and fear of errors:

*"And then the suction machine, we were like, what the hell is this? Peg feeding, what the f*** is this?"* (Jamie, interview 1)

Paul similarly recounted being initially intimidated by the medical equipment and clinical skills needed to care for Caitlin. Over time, he felt so confident in doing these that he began making sure healthcare professionals were not making mistakes:

"It was scary. She was so delicate, and all those tubes... she had feeding tubes, broviacs [central line], and other lines... We became doctors ourselves and did exactly what the nurses were doing... even making sure they weren't making mistakes." (Paul, interview 1)

Alex noted his concerns with over and underfeeding Amy through a feeding tube, but showcased how these fears alleviated as time went by:

"I was a little bit nervous about the tube... once you do it once or twice, it's a little bit more blasé - not blasé but a little more confident with it... When you're feeding her from the tube, I think we overdid it because she got quite chubby... But we were thinking we were overfeeding her, then underfeeding her and then overfeeding her again..." (Alex, interview 2)

In addition to gaining confidence in clinical skills, many fathers established roles within their households that facilitated the caregiving routine. Jan explained how these roles became crucial for the family to adapt to meet the demands of Jakob's needs:

"Yeah, mostly my wife is doing the feeds. She cooks, and I mostly do the nappies and things like this. Some things we do together, and it's easier. Like dressing, and you know, my wife is fifty-something kg, and he is sixty (kg)." (Jan, interview 2)

Although skills were gained, some fathers also described the repetitive and mundane aspects of caregiving. Mike, who journaled his experiences, expressed frustration and boredom in some of his caregiving roles. His son was off school sick again, which meant he could not go to work due to the "loco parentis", had to stay home with Toby, who enjoyed watching the same show repeatedly:

"If I hear that show one more time." (Mike, journal entry)

These reflections underscore the full spectrum of caregiving, from the acquisition of complex skills to the relentless, sometimes monotonous, routines that fathers faced.

6.3.5.3 Changes in their children's complexity over time

For several fathers, their children's CCN evolved over time, due to the specific nature of their conditions. Some fathers observed increasing complexity in their children's needs related to behaviour, intellectual functioning, and anxiety. Some children initially required medical interventions like nasogastric feeding or growth hormone injections, but as they grew, they faced escalating complexities, especially as behavioural and mental health issues emerged. Additionally, sleep deprivation was a profound and ongoing burden. Fathers used a variety of strategies, yet they collectively highlighted the severe impact of prolonged sleeplessness on their and their children's lives.

Fathers provided detailed accounts of how their children's anxiety impacted daily life. Peter noted a significant shift in his daughter Rosie since she started school, describing how school-related anxiety disrupted her sleep:

"... since she started school, the anxiety levels have risen. She's just really anxious, wanting us to stay in the room until she falls asleep, purposefully waking herself up, and coming in and out all night..." (Peter, interview 2)

David highlighted the severe anxiety that affects his son in his teenager years, noting that it manifests in behaviours like skin-picking. Witnessing their children's distress is challenging, particularly as their intellectual disabilities often hinder effective communication about the causes of their anxiety. David described the difficulty of seeing his son's self-harm due to unresolved anxiety:

"So, anxiety rules his life and ours at the minute... his fingers are picked to bits... his head is just so full... It's very hard to watch your own kid after picking off his own skin off his knuckles just because he's thinking about something. And he can't tell you what it is." (David, interview 2)

Other fathers discussed their children's food anxiety - some of which they were already managing and some of which they anticipated as a future concern. Alex referred to the early years as a *"honeymoon period"* before food-related anxiety began. He reflected on how, in Amy's early childhood, he could control her environment and needs more directly:

"Ok, so the honeymoon period is kind of zero to six, I would say. When everything is ok, delayed, but normal... when she was a baby and a child, you were in control of her and what happened to her. And with [Amy's CA], they always need a carer, and as an early-day parent, you are that person. But now that she is a teenager, you can't as much, and that is hard." (Alex, interview 2)

The challenges of their children's behaviours were a recurrent theme. Many fathers discussed the complexities of managing their children's behaviours, noting how people sometimes judged their parenting due to her "invisible" condition. Peter explained:

"... people who interact with people [name of a different syndrome] know they have it... but she doesn't look syndromic, that you wouldn't realise when meeting her that there is something wrong with her. And I find that challenging because

of her behaviour... these are the behaviours we are seeing more and more of, and it would just be easier if it was more visible.” (Peter, interview 2)

The additional complexities of puberty and hormonal changes presented further challenges for some fathers. David noted Joey’s experience of precocious puberty, and managing wet dreams which required ongoing personal care assistance. This issue was made more complex by intellectual disabilities as personal hygiene needed to be managed.

“So, I get up in the middle of the night and kind of help him through that [wet dreams].” (David, interview 1)

Sleep deprivation as a result of some CA symptoms, was a prevalent challenge for many fathers, closely linked to their children’s CCN. They used intense, war-like language to convey the severity, describing it as “torture” and likening it to being “in the trenches” or in “hell.” Peter noted:

“It was really, really hard... it’s one of the forms of torture, isn’t it?” (Peter, interview 1)

David spoke of sleep deprivation as an early sign of his child’s undiagnosed condition. He spoke about taking on night duties to let his wife rest. He mentioned that the effects were cumulative and exhausting:

“She needs a lot of sleep... so I was just trying to mind my wife... I didn’t need a lot of sleep... that’s fine for a couple of nights... but it doesn’t relent. I remember it impacted work. I was just not performing well... and that brings its own stress.” (David, interview 1)

Some sleep deprivation changed over time for the better, as some fathers found relief through home care packages, medications like Melatonin, and private sleep therapists. Peter noted:

“We found out about Melatonin... and our lives changed...privately of course.” (Peter, interview 2)

These fathers recounted the evolving nature of their children's conditions, highlighting increased challenges over time, particularly in behaviour, intellectual functioning, and anxiety. Initial medical interventions sometimes gave way to more complex issues, including severe anxiety, behavioural difficulties, and puberty-related changes. Sleep deprivation emerged as a persistent challenge, with fathers describing its profound impact on their lives. Fathers also detailed the emotional toll of witnessing their children's distress, exacerbated by intellectual disabilities that hinder communication. Despite these challenges, fathers found some relief through strategies like medication, professional support, and HCP.

6.4 Conclusion

This chapter presented the findings from nine fathers of children with a range of CACCN with the aim of gaining an understanding of father's experiences of caring for their child. Findings were reported in line with the five existential themes; lived body (corporeality), lived space (spatiality), lived time (temporality), lived human relation (relationality), and lived things (Van Manen, 2016). Overall, these findings highlight the complex nature of daily life.

The first existential theme of "lived Body" explores the physical and emotional impact on fathers learning of their child's CA and coming to terms with this diagnosis. Fathers' experiences were unique, influenced by the timing and context of the diagnosis. Many recalled when they noticed something was abnormal, which varied from birth to early childhood. Fathers described emotional responses, ranging from shock and fear to confusion and eventually gradual acceptance. Healthcare professionals significantly impacted fathers, with some noting inadequate communication or support and others highlighting the need for independent research. Fathers described coming to terms with the diagnosis, finding solace in faith, support groups, or adjusting expectations, while others struggled with uncertainty about their children's futures. This complex journey was shaped by individual circumstances, healthcare interactions, and personal coping strategies.

The second theme, "lived relation," explores the relational aspects of fathers' experiences in raising a child with a CA, which underscores the complexity of relationships shaped by their children's abnormalities. Fathers articulated their experiences of fathering a child with CACCN in relation to their bond with that child, while also situating this relationship within the broader context of their roles as fathers to the child's siblings. While mothers frequently provide critical emotional and practical support, they can also be sources of stress at times. Fathers' relationships with grandparents are marked by both gratitude and frustration regarding the support they provide and at times their insensitivity and lack of understanding was profound. Fathers equally had anxieties about their children's social interactions, friendships, and the challenges these relationships may entail as a result of their intellectual disability.

The third existential theme of "lived space" highlights the physical and psychological spaces that fathers navigate while caring for a child with a CA. Fathers describe the challenges of long hospital stays and the subsequent adjustments required upon returning home, which involve not only physical changes but also significant mental and emotional adaptations. The importance of homecare packages, respite, and residential care in supporting both current and future caregiving needs is evident. Additionally, fathers reflect on the mental space occupied by the complex decisions they must make in providing care for their children, such as signing DNR's or what schools they should send their children to.

The fourth existential theme of "lived things" explores fathers' experiences of caregiving in relation to material aspects, specifically the resources and items deemed necessary for their child's care. Fathers highlighted the financial implications of meeting daily care needs and the associated worries about their children's future financial security, some fathers were grateful for financial support they received, while others felt let down by basic items such as hygiene wipes not being supplied. Fathers discussed the additional challenges in balancing work responsibilities with the demands of caregiving.

The fifth theme of "lived time" encapsulates the evolving temporal experiences of fathers as they navigate the complexities of caring for a child with a CA. This journey

involves adapting to the dynamic and often unpredictable nature of their children's needs, which shape their daily routines, mindsets, and caregiving roles. Fathers demonstrated resilience and growth, as seen in their ability to adjust their perspectives in response to changing life expectancies and the demands of caregiving. They highlight their increasing proficiency and confidence in caregiving, their dedication and capacity for adaptation. Ultimately, this theme reflects the fathers' evolving relationship with time as they strive to balance the demands of caregiving with the broader dimensions of their lives.

The next chapter will discuss these findings in relation to the study's aims and objectives and will present a critical discussion in relation to the existing literature.

7 Discussion

7.1 Introduction

The aim of this study was to explore fathers' experiences of caring for a child with a CACCN. This was achieved through data collected via the interviews and journals of the nine fathers that participated. This chapter discusses the main findings in relation to the study's aims and objectives and presents a critical discussion in relation to the existing literature. Van Manen (1990, 2014) highlights the importance of balancing the research, by considering the relationships between the *parts and the whole*. Therefore, to fully understand the experience of fathers caring for a child with a CACCN, it requires a dynamic interaction between examining parts of this experience in each existential theme and determining the overall meaning.

The findings from this study provides a comprehensive understanding of fathers' experiences caring for children with a CACCN. Using van Manen's existential themes as a framework, the findings highlight that fathers' caregiving roles are multifaceted, continuously evolving, and deeply intertwined across emotional, relational, material, and temporal dimensions. Collectively, these insights highlight both the challenges, and the meaning fathers derive from their caregiving responsibilities.

The theme of lived body shows that fathers experience profound emotional responses to their child receiving a diagnosis. Furthermore, they report experiencing practical difficulties, particularly related to delayed diagnoses and fragmented access to healthcare and support services specific to their children's CACCN. In the theme of lived relation, fathers negotiate complex family dynamics, balancing the needs of their child with CACCN, their other children, the mother of their children, while maintaining relationships with extended family.

Lived space highlights the influence of physical and emotional environments, including hospital-to-home transitions, homecare, respite and residential care availability, and planning for future care and education needs. In lived things, fathers face structural and financial pressures, such as out-of-pocket medical costs and restrictive leave policies, which can limit their ability to fully participate in caregiving and may reinforce traditional

gender roles. Finally, lived time captures the 24-hour, ongoing nature of caregiving and its emotional toll, including sleep deprivation and stress associated with fluctuating symptoms and behavioural challenges.

Together, these themes demonstrate that fathers' experiences are not static but require continuous adaptation as their child's CCN change over time. Despite these challenges, fathers report deriving significant meaning, joy, and purpose from caregiving.

This chapter is structured around the main themes, presented within the framework of van Manen's (1990, 2016) five existential lifeworld's. Each theme is explored individually and then considered collectively to understand the broader meaning of fathers' lived experiences. It is important to revisit the aims and objectives of this study to frame the analysis that follows:

- To explore fathers' experiences of how their child was diagnosed;
- To develop an understanding of how this diagnosis impacts fathers;
- To explore the roles and responsibilities fathers have in caring for their child with a chromosome abnormality and complex care needs;
- To identify what supports fathers have and how these supports impact their everyday life.

7.2 Receiving a diagnosis of a chromosome abnormality and the subsequent fight for services (lived body)

Findings from the first existential theme, "lived body" reveal that when their child receives a diagnosis of a CA it is a deeply multifaceted experience for fathers. This theme captures both the emotional and practical dimensions of their journey from the initial concerns about their child's development to the eventual receipt of a formal diagnosis. While the experience of learning that a child will have a disability is universally challenging for parents, this study offers a novel contribution by specifically highlighting fathers' perspectives and the often-rare nature of their children's CA. Beyond the emotional impact, the findings also highlight a critical concern: delays in accessing timely and appropriate diagnostic and support services. These delays within both the diagnostic process and broader disability support systems emerge as a central issue

affecting fathers' ability to navigate their roles and support their children effectively in the Irish context.

Three core aspects of the "lived body" theme are discussed in depth in the following section. First, the importance of how professionals deliver diagnostic information is examined. Second, the study explores the rare nature of many CAs and evaluates current practices in monitoring and recording such conditions, with attention to why this is important to fathers. Finally, the ongoing struggle to access disability services both within Ireland and internationally is addressed, highlighting systemic barriers and their implications for families.

The way the diagnosis is presented is vitally important, as research shows that when parents are given a diagnosis that their child will have a disability, situations such as poor diagnostic delivery and lack of information can lead to reduced parental coping (Kenny et al., 2022). A specific critique from fathers in this study was the overuse of medical terminology when professionals disclosed either a diagnosis of a CA or when explaining the genetic testing process. Doctors themselves have reported difficulties in understanding the complexities of genetic testing and find it challenging to keep up to date with constantly advancing methods of testing, including microarray, karyotyping and the next generation sequencing (Harding et al., 2019; Talwar et al., 2017). A recent study in Ireland explored paediatric trainees working within CHI, and found that 84% had recently obtained consent for a child and their family to have genetic testing, with 41% of the medical staff reporting that they had a poor understanding of genetic testing technologies and indications. Furthermore, 81% highlighted that they had no prior teaching on genetic testing, consent, or the implications (Quinn et al., 2023). This suggests that trainee medical professionals are not all adequately equipped to explain both these tests and/or the results to parents who have no medical training. The available best practice guidelines in Ireland highlight the need for healthcare professionals to communicate in simple understandable terms (HSE, 2015). People in Ireland have called for improved communication techniques in healthcare for some time. In a previous study, 39% of Irish people requested that healthcare professionals

use more understandable language and less “medical jargon” (National Adult Literacy Agency: Ipsos MRBI Survey, 2015).

The lack of information about their children’s developmental trajectory is a challenge for some fathers in this study. Research indicates that parents of children with abnormalities request more positive information that focuses on the quality of life and what the children can achieve rather than solely on the limitations of their conditions (Docherty & Dimond, 2018; Holm et al., 2021; How et al., 2019). Yet, findings of this study further indicate that this is difficult to achieve in some cases, as little is known about their children’s CA. This is significant as the recent advancements in genetic investigations, means more children with developmental delays are receiving a diagnosis of rare chromosome and genetic abnormalities (Gerik-Celebi et al., 2023; Kim et al., 2023; Zhang et al., 2024), which in turn means more fathers will potentially be subjected to this lonely journey. Fathers being told their children have a rare diagnosis is inevitable and support websites do exist, with registered charities such as rarechromo.org (Unique, 2025), act as an international group - supporting, informing, and networking people with rare CAs or professionals who may be interested. Yet, no fathers in this study mentioned them.

Monitoring and recording these newly identified and rare CAs helps healthcare professionals explore the health, developmental and educational outcomes of these children and share this information globally. In Europe, EUROCAT (European Surveillance of Congenital Anomalies) is a collaborative network of population-based registries that allows for ongoing surveillance and research of congenital anomalies including CAs. This network was established in 1979 and at present comprises 43 population-based congenital anomaly registries who submit their data to JRC-EUROCAT Central Registry once or twice per annum (EUROCAT, 2022). This allows for research such as the “EUROlinkCAT” study which uses data linkage to examine the outcomes of children born with congenital anomalies on 22 registries from 14 European countries. This was part of the European Union Horizon 2020 research and innovation programme that brought together different sources of information on the health, developmental and educational congenital anomalies including CAs (EUROCAT, 2020). These registers

are helping to add to the existing body of knowledge of some conditions (Morris et al., 2021). Yet, in the Republic of Ireland there are only three regional congenital anomaly registers that are members of EUROCAT, and of these only two are currently active (HIQA, 2025). This means that only 23% of all births in the Republic of Ireland provide Irish data on congenital anomalies to the EUROCAT database (HSE Congenital Anomaly Registers in Ireland, 2025). Each is only led by a Specialist in Public Health Medicine, with only a part-time research nurse and limited clerical support (HSE Congenital Anomaly Registers in Ireland, 2025). This highlights the need for Ireland to increase their surveillance on such anomalies to expand the body of knowledge.

Despite some of the challenges in relation to the rare nature of their children's conditions during the diagnosis stage, findings from this study highlight some positive experiences in their diagnostic journey, particularly in relation to early recognition by professionals. Fathers appreciated the speed in which professionals were able to link their children's features with their subsequent CA diagnosis particularly in the neonatal period. This was especially important for those of children with Prader-Willi syndrome as this condition is known for the rare and subtle dysmorphisms which can result in challenges with early identification in the neonatal period (Janssen et al., 2024). Research shows the importance of early and precise identification of genetic anomalies, as this is essential for improving child outcomes and offering effective genetic counselling (Choo et al., 2019; Gaadi et al., 2025).

One study exploring children and adults with a genetic condition, reported that parent's satisfaction during the diagnosis was significantly lower for parents of children diagnosed at birth, compared to parents of those diagnosed in childhood (Zampini et al., 2025). This does not correlate with the findings in this study, as fathers who were more disheartened were those who had a late detection by professionals. A lack of identification of some specific CA can have negative health outcomes. For example, one father recounted a "near miss" incident in which his child underwent a general anaesthetic before receiving a diagnosis. The child's condition significantly increases the risk of morbidity and mortality during sedation and anaesthesia (Twite et al., 2019), underscoring the critical importance of early identification.

Findings from this study highlight some gender specific challenges fathers experienced; challenges they felt were not shared by the child's mother during the diagnostic process. In several cases, fathers reported being excluded from crucial conversations, as they were unaware that diagnostic results would be shared and were therefore not present. A recent Irish study examining parents' experiences of receiving a diagnosis of a rare genetic abnormality in their child acknowledged a gender imbalance in its sample; predominantly mothers participated, and this is stated as a limitation (Fitzgerald et al., 2021). This study helps address that gap by giving voice to fathers whose perspectives have often been overlooked. Research has shown that parents frequently feel unprepared during and after conversations where bad news is delivered (Brouwer et al., 2021). Consistent with this, these findings suggest that preparation is particularly important for fathers, who expressed a need for advance notice so they could arrange to be present. This concern of wanting prior warning of when diagnoses will be given emerged as a uniquely male perspective in this context.

The frustration many fathers felt in relation to delays in identifying missed developmental milestones went hand in hand with the significant challenges in childhood disability services in Ireland. These challenges were felt both from a diagnostic standpoint and from delays in accessing disability services for interventions. Fathers opted to bring their children to private professional support, including private consultants to receive a genetic diagnosis, private psychologists to get their child a cognitive assessment and private therapies such as occupational therapy, SALT, physiotherapy and sleep therapists due to waitlist times or therapist service deficits in their area.

It is important to give context as to why fathers opted for private support in a country that has a public health system (The HSE). The HSE is the publicly funded healthcare system in Ireland, responsible for providing health and social care services. Every child in the Republic of Ireland, is entitled to free health and development checks undertaken by the HSE public health nurse administered from 0 to 5 years, which are often the first step in identification of developmental delays (HSE, 2022). Yet, many children are missing these vital assessments due to staffing issues where there is a lack of public

health nurses undertaking developmental assessments, with many children missing their milestone check-ups as a direct result (Houses of the Oireachtas, 2024). The 2018 “Health Service Capacity Review” anticipated a 46% increase in demand for public health nurse appointments by 2031 (Department of Health, 2018) yet, the latest census report shows there were 1,479 public health nurses in 2023, a decrease of 53 since the December 2019 (HSE, 2023). The HSE has identified a recruitment challenge for public health nursing as a cause for these delayed developmental checks. This lack of public health nurses has resulted in children missing their essential developmental assessments (Houses of the Oireachtas, 2024). This has in turn resulted in some families self-checking their children’s developmental milestones which may present missed opportunities to identify developmental delays within appropriate timeframes.

Once a developmental delay has been identified, children can then be referred to a paediatrician for a medical assessment, and to consider if medical tests including genetic testing, is required (CHI Ireland, 2024). However, in Ireland there are significant waiting lists for these children to see a paediatrician, thus resulting in delayed access to genetic testing. For example, in one catchment area (two of three sites across the capital city), there are currently 691 children waiting to be seen by a paediatric developmental consultant, many of whom need assessment for genetic testing (HSE, 2024). Rare Ireland indicates that families are waiting up to two years to see a genetic counsellor/or consultant (Rare Ireland, 2024). The past decade has led to significant advances in diagnostics, including the availability of next-generation sequencing which internationally has reduced the time for diagnosis, and increased the numbers of children receiving a CA diagnosis (Wigby et al., 2024). Yet, the findings in this study show that waiting lists to have these tests completed are still a significant challenge for families.

Aside from genetic testing, when a potential disability is identified children are referred for an ‘Assessment of Need’ (AON) which is a statutory process identifying children’s health and social care needs (HIQA, 2024). Families in Ireland face significant challenges in accessing assessments, despite legal protections provided by the “Education for Persons with Special Educational Needs” (EPSEN) Act 2004 and the “Disability Act 2005”.

The AON is experiencing prolonged delays, despite a legal requirement to initiate the assessment within three months of receiving an application (Ombudsman for Children, 2020). The demand for AON applications quadrupled between 2007 and 2018, yet in 2018 only 8.8% of assessments were completed within the required timeframe (Ombudsman for Children, 2020). The AON received 7,612 new applications from June 2022-2023 (HSE 2023). Moreover, 11,430 AONs were overdue or had to be redone, resulting in a waiting list of 19,042 applications in 2023 (HSE, 2023). The impact these delays have on families are vast, and these delays can have a serious negative impact on childhood (Barnardos, 2017). Multiple fathers in this study had to seek private assessment or relied on chromosome specific organisations to provide cognitive assessments.

These delays are not issues that are unique to Ireland; in England, children with a neurodevelopmental condition are waiting several years for a diagnosis, delaying access to essential support services. Within community health services, the median waiting time for a diagnosis is two years and three months. Nearly 41% of children waited over two years for their diagnosis, with 17% waiting more than four years (Children's Commissioner, 2024). In Australia, one study reported that caregivers typically identified developmental concerns at an average age of 3 years, yet the average age at which children received a developmental assessment was 6.6 years. Despite 88% of caregivers expressing concerns about their child's development by the age of five, only 46.4% of children had received a diagnostic assessment by that age (Boulton et al., 2023).

Fathers identified significant delays in accessing disability services once a child has been diagnosed and as their needs changed. In Ireland, children's disability services have been reorganised into Children's Disability Network Teams (CDNT) as part of the HSE's Progressing Disability Services for Children and Young People. The programme was initiated in 2010 in response to recommendations in the Report of the Reference Group on Multidisciplinary Services for Children Aged 5–18 Years (HSE, 2009) and included a reconfiguration of early intervention services for children aged less than five years. These teams were developed to provide services based on the needs of children with disabilities rather than their diagnoses and is based on the child's home address. The

aim of a CDNT is to provide specialised support and services for children who have CCN associated with their disability (HSE, 2025). Ninety-one CDNT's have been developed by the HSE and its partner Lead Agencies (HSE, 2023). However, there have been significant issues in the implementation of these teams due to staff shortages, with a national average vacancy rate of 34% and some teams experiencing over 60% staff vacancy rates. In one area the waitlist for children to be seen in the CDNT was up to 5 years (Wardick & Rabbitte, 2024). This is also not unique to Ireland. In England, some community health services have children waiting for community Down syndrome services for up to 2 years and 7 months (Children's Commissioner, 2024).

The importance of early intervention for children with developmental and intellectual disabilities is well documented within the literature, it is known to improve child development and adaptive skills, behavioural outcomes and improves sleep habits (Lai et al., 2014; Ryberg, 2015; Scherzer et al., 2017; Smith et al., 2015; Smythe et al., 2021). Specifically, all children in this study with a CA were accepted onto their local CDNT, yet most of them reported not receiving regular support or services (i.e. were waitlisted or therapists' roles were not filled). Children with a CACCN identified in this study need intervention through the CDNT for many stages of their lives. For instance, a child with PSW, would need a multidisciplinary approach to care (Naotunna et al., 2025). Children with this condition require SALT, OT and physiotherapists to address challenges in infancy, such as feeding difficulties that often require the use of special nipples/teats, external feeding such as NG or PEG feeding, and sensory integration (Duis et al., 2019; Metzler et al., 2025). They are at increased risk of gross and fine motor delays as a result of hypotonia, and regular assessments and support is needed to meet their educational requirements (Duis et al., 2019). Psychology and behavioural therapies are required due to their increased risk of mental health challenges (Skokauskas et al., 2012). One study in Ireland showed that hyperphagia occurred in 81% of participants of people with Prader-Willi syndrome over the age of 4, with the average wait of 22 months for an appointment with a psychologist (Feighan et al., 2020). In this study, fathers articulate the urgent need for better MDT approaches in caring for their children. The need for families to pay privately for therapeutic interventions is a significant concern, especially

when children in Ireland are entitled to a public healthcare system which means they are entitled to access free healthcare. In 2022, 22.1% of Irish children had private health insurance (Health Insurance Authority, 2023). Furthermore, this will inevitably result in inequitable access to interventions, based on socioeconomic disparities. In the UK, one study found that some children with an unmet need for early support was associated with economic deprivation (Sapiets et al., 2024). Another UK study showed that 65% of occupational therapists felt that the cost-of-living crisis is making it difficult for children to access occupational therapy through private means (Royal College of Occupational Therapists, 2023).

Despite the calls for integrated care systems for children with CCN, this study concludes that fathers still feel their care pathways are fragmented, particularly in relation to community services. In a recent study of parents of children in Ireland with rare diseases, 47% reported difficulties in maintaining a consistent healthcare team for their child (Somanadhan et al., 2025). Integrated care aims to link health and social care services, to deliver individual care that is safe, timely, efficient and as close to home as possible. There has been a significant growth over the past decade in literature to support the development of integrated care for children with CCN, but little guidance on what processes or elements are essential for successful implementation of integrated care programmes (Cassidy et al., 2023). A care pathway is a complex multidisciplinary plan of care that stipulates the necessary steps for managing children with particular clinical conditions, thus standardising care across settings, yet the challenges persist.

7.3 The complexity of relationships (lived relation)

Findings from the existential theme of "lived relation" underscores the enhanced complexities that having a child with a CA has on fathers' relationships. These findings establish the intricacies of their relationships with the entire family unit, including their children (both with a CA and without), the mother of the child and the grandparents. Without a doubt, the love and desire to do their best for their children is evident. Family members, such as mothers and grandparents were at times, a significant support system, but equally could be a source of distress for these fathers.

Within the existential theme of “lived relation” three core concepts emerge that illuminate the relational dynamics fathers experience in the context of parenting a child with a CACCN. Firstly, fathers describe the emotional complexity of balancing deep love and concern for their child with a CACCN alongside the need to remain present and supportive for their other children without a diagnosis highlighting the challenge of maintaining equity in their parenting roles. Secondly, the findings emphasise the importance of navigating the parental partnership, as fathers work to support their child while also sustaining their relationship with the child’s mother. This dual responsibility often requires careful negotiation between being a caregiver and a supportive partner. Finally, the role of grandparents is explored, with fathers acknowledging the benefits of their involvement, while also recognising the importance of setting boundaries to maintain manageable family dynamics.

Despite the initial fear and worry associated with having a child with a CA, fathers experience profound feelings of love, pride, and joy in their relationships with their children. The emotional journey for these fathers is complex, as the challenges of raising a child with a CACCN co-exist with the rewards of creating unique and meaningful connections with their children. This interlinked phenomenon aligns with other empirical literature in this field (Uribe-Morales et al., 2021). Research shows fathers often have anxiety prior to their baby’s birth, particularly in relation to fear for their baby’s health (Figueiredo et al., 2007; Furukawa et al., 2020). However, these fears are alleviated once the babies are born safe and well (Furukawa et al., 2020). Yet, for many of these the first few days of their baby’s lives were terrifying as their children’s CAs and associated CCN were being identified. Within the literature, typically, the excitement and positive responses from their “healthy” baby are deemed crucial in the bonding process between a father and his child (Brady et al., 2017). Yet for some, this was not the experience at birth due to the symptoms of their CA as they experienced emotional turmoil. This grief resulted in some fathers looking back on the early days with sorrow and expressing feelings of missing out on “normal” father-baby experiences. Literature shows that when fathers can carry out tasks for their new babies such as bottle feeding, this increases father-infant bonding, which validates why these fathers may still reflect

in sadness at not being able to do such activities with their babies (Avery et al., 2011; de Montigny et al., 2018), and this study concludes that fathers want and need to be included in initial care of their child.

Despite the somewhat challenging start, the traits associated with their child's conditions that were once feared, often evolved into a source of pride and admiration. Fathers' emotions shifted and they began to celebrate these traits as integral and positive aspects of their children's personalities. Similar research reports the pride that fathers often feel when their children with CCN connect with other people and their communities (Woodgate et al., 2024). Simultaneously, the very aspects of their children's personalities that the fathers love so much, are an equal source of worry and concern, particularly in relation to their children's friendships and safety. For example, many children with Williams Syndrome have a "hypersocial" personality and a propensity to seek out social interaction (Järvinen-Pasley et al., 2010; Jawaid et al., 2012). Yet, these children can also have significant challenges making and maintaining friendships, have little safety awareness which can make them vulnerable, thus highlighting the complex and interlinking nature of the children's condition (Fisher et al., 2014; Riby et al., 2017). Much of the existing literature on fathers caring for children with disabilities primarily focuses on the negative impact of care burden, often overlooking the positive effects that these children have on their families and communities (David et al., 2024). The positive experiences shared by these fathers suggest that their children's identities and capabilities are multifaceted, filled with potential, and not confined to their abnormalities. Fathers who experience less stigma on their children often have more positive relationships and experience less stress (Manor-Binyamini, 2025).

Furthermore, findings from this study show the distinct characteristics of these children have a broader positive impact, not only on their families but also on the communities in which they interact. A large amount of the existing literature surrounding the societal implications of children with CCN tends to focus on the pressure these children place on healthcare systems, including the financial burden to society, including healthcare costs and hospital stays, medications, equipment, respite and residential care (Berry et al.,

2015; Foster et al., 2019; Friedman & Kalichman, 2014; Knapp et al., 2008; Kuo et al., 2015; Report for the British Healthcare Trades Association, 2014). This often neglects the broader, more nuanced contributions these children and their families make to society, which fathers in this study have highlighted. This is important to note, as many fathers describe resorting to reading literature on their children's diagnosis and disabilities on google. In social science literature, interpretations of disability are changing, with a focus on how society frames disability (Carmel et al., 2025). This starts with research highlighting the positive experiences as much as the challenges and values the person as a whole.

The bond that fathers have with their children goes beyond just their child with CCN. They express feelings of personal grief related to missing out on the childhoods of both their children with and without a CACCN. Fathers articulate the challenge of wanting to always be present with their child with a CACCN, yet, simultaneously recognising the impact this has on their other children. Many fathers highlight the guilt they feel when the needs of their child without a CA are deprioritised, expressing concern over the potential long-term effects this may have. According to Wikle et al. (2025), parents who have a child with a disability spend more time with their disabled child and less time with their other children. Siblings of children with CCN report challenges in many aspects of daily life and have been shown to display more psychosocial problems than their peers (Haukeland et al., 2021; Linimayr et al., 2025). Specific to CACCN, siblings of children with Prader-Willi syndrome, have shown to exhibit symptoms of post-traumatic stress disorder, which often increases with severity in conjunction with their siblings' levels of emotional outbursts (Micallef & Hughes, 2025). Existing research highlights the crucial role fathers play in the care of their children, as their involvement has been shown to foster social competence, enhance educational outcomes, and improve overall health outcomes for children (Cho et al., 2024). The fathers in this study all showed a deep understanding of how crucial their role was as a father and expressed a desire to provide equal parenting to both children, which they find is not always possible as the needs of their children with a CACCN need to take priority.

Furthermore, fathers demonstrated a profound awareness of the complex and ever evolving nature of their children's conditions, which they speculated would bring challenges to their other children. The anticipatory grief of fathers/siblings, stemming from the potential loss of their child with a CACCN was a particular concern. While they often cherished witnessing the unique bonds forming between siblings, they also expressed fear that the longer their child lived, the more difficult it would be for the siblings to cope with the eventual loss. Some similar studies of parents of children with CP, report greater anticipatory grief the longer their children survive (Al-Gamal, 2013). Research shows that siblings of children receiving palliative care are at risk of negative health related quality of life (Dinkelbach et al., 2023).

Understanding how much information to give their other children about their sibling was a source of anxiety for fathers, as they hoped that keeping some information from them would protect them. Siblings of children with CCN themselves report feeling like the lack of information about their sibling's diagnosis can lead them to guessing and imagining, which they feel negatively impacts them (Avieli et al., 2019). Furthermore, fathers emphasise the significant source of anxiety related to the additional roles and responsibilities that siblings assume or will need to undertake in the future. Many siblings often undertake extra roles within the family to gain recognition from their parents or to help reduce family stress (Avieli et al., 2019; Park et al., 2021).

The desire of fathers in this study to have more children as a means of alleviating some of the sibling caregiving burden was notable. Mulroy et al. (2008) suggests that siblings of children with Trisomy 21 who grow up in larger families are more likely to report positive experiences with their siblings. Fathers acknowledge their children's unwavering affection for their siblings while also recognising the challenges that they face. This dynamic is consistent with findings from studies on siblings of children with CCN where siblings express mixed emotions, such as "I love my sibling, but..." (Woodgate et al., 2016). There is a necessity for professional support in managing these complex family dynamics. Fathers demonstrated awareness of the challenges, but no father mentioned receiving specific sibling support from the CDNT.

Another important relational aspect for fathers, is the relationship with the mother of their child. Findings of this study are in line with international literature on this phenomenon, in which many of the father's report that the mother is an important source of support (Cohen et al., 2016; Huang et al., 2011; Rivard & Mastel-Smith, 2014; Takataya et al., 2016). This goes both ways as some mothers also report high fatherly support in the care of their children with CCN (Mohamed et al., 2024). Equally, however, research shows the way parents interpret information, and deal with the diagnosis is often very different, which can be a source of relationship strain (Ridding & Williams, 2019; Takataya et al., 2016; Uribe-Morales et al., 2022). Some research suggests that mothers may experience a greater emotional impact from their child's disability compared to fathers (Balbo et al., 2024). Yet, fathers in this study and within other literature report experiencing levels of worry comparable to their partners, although their concerns often focused on different issues or aspects of the situation (Zahl et al., 2024). This divergence highlights the distinct ways in which different genders may process and respond to receiving a diagnosis.

As fathers got further along into their journey of their children's CACCN, noticeable differences emerged in how fathers coped with the complexities of everyday life. Many reported wanting to be part of associations and support groups, yet the mother did not. Support groups and CA specific organisations/social media websites allow fathers to meet parents in similar circumstances, and research shows they report these groups to be fundamental in their coping (Bentley et al., 2015; Fitzgerald et al., 2021; Lowit & Averill, 2014; Marshak et al., 2019; Ridding & Williams, 2019). Traditional masculine norms have been shown to significantly influence men's psychological help-seeking behaviours and their self-reporting of mental health status. Fathers of children with a CA reflect on the concept of "masculinity" in their caring roles (Cameron & Cooper, 2020). Üzümpçeker (2025) found that adherence to traditional masculine ideologies negatively impacts men's willingness to seek professional psychological support. Similarly, Sullivan (2015) stated that men who score higher on measures of traditional masculine ideology exhibit more negative attitudes toward seeking professional mental health services. One father in this study described an instance where he wished to access

mental health support during the diagnosis stage but ceased doing so after his wife disagreed with the necessity of such support. This example highlights how family dynamics and traditional gender roles may inhibit fathers of children with a CACCN from pursuing mental health support, despite experiencing significant stress and concern.

The experiences of fathers in this study suggest that they want equal participation with the mother in the care of their children, and recognise their roles as equally important, which is in keeping with the findings of the scoping review (Docherty & Dimond, 2018; Huang et al., 2011; Huang et al., 2012; Ogourtsova et al., 2021; Ricci & Hodapp, 2003; Schippers et al., 2020; Uribe-Morales et al., 2022). Fathers felt that they worked in tandem in the allocation of responsibility, which included a mutual shared load of separating tasks both mentally and emotionally. These findings conclude that although at times these roles differed, their contribution to the family was equally important. This finding is different to Ridding & Williams (2019) who reported that some fathers felt that mothers were the main caregivers. Instead, the fathers in this study accepted their roles were just different, not less. The evolving nature of fatherhood is reflected in the increasing involvement of men in both the physical and emotional aspects of childcare (Baldwin et al., 2018; Volling & Palkovitz, 2021; Woodgate et al., 2024). In 2021, just under one-in-five mothers (17%) in Ireland stated their occupation as “homemaker”, meaning 83% reported to be in paid employment (Central Statistics Office, 2021). This highlights the increasingly important role that fathers play within the family unit, roles that are equally vital and deserving of recognition and respect.

Finally, this study highlights the importance of grandparents in the family unit. This dynamic is not unique to fathers of children with CCN, as, internationally, grandparents are assuming increasingly significant responsibilities in childcare. For example - In Ireland, 29.1% of individuals over the age of 70 provide care for their grandchildren, with an average of up to 16 hours per month (McGarrigle et al., 2022). In the UK, approximately 40% of grandparents provide regular care for their grandchildren (Age UK, 2017). In the United States, grandparents across all age groups are providing more support to their grandchildren, particularly grandparents aged 75 - 84 (Hyeonji, 2025). There are notable personal benefits to grandparents assisting with the care of their

grandchildren, with many reporting fewer limitations to their activities of daily living and improved mental health status (Danielsbacka et al., 2019; Eibich & Zai, 2024; Zeng et al., 2021).

Within the context of childhood disability literature, grandparents remain underrepresented as members of the family, with their experiences insufficiently explored (Novak-Pavlic et al., 2022). Much of the available literature in relation to grandparents is in the context of children with cancer (Moules et al., 2021; Wakefield, 2014). Nevertheless, many grandparents report providing a combination of financial, emotional, and physical support to their grandchildren with CCN (Novak-Pavlic et al., 2022). Some describe their roles as extending to the care of siblings of children with disabilities, effectively taking on "co-parenting" responsibilities, thereby enabling parents to focus more on the child with CCN (Yang et al., 2018). It is not surprising therefore that some grandparents report having a close relationship with all their grandchildren, yet the quality of the relationship is sometimes higher in grandchildren without developmental disabilities (Kornilaki & Kypriotaki, 2024).

The concern for fathers overburdening their own parents is a finding of this study. Alongside father's concern for the increased physical demands that their children with a CACNN will require, which they felt could impact their ageing parents' ability to care for them effectively. This concern is particularly pertinent given that, while more grandparents are contributing to childcare across Ireland, the average age of parents is also rising. In 2021, the average age of mothers giving birth was 33.3 years, the highest recorded since the data was first collected in 1955 (Central Statistics Office, 2022). As women are increasingly having children later in life, the age of grandparents is also rising, and the impact of this on caregiving dynamics is becoming more evident. In the UK, during 2017-2018, the average age at which 50% of individuals had a grandchild living outside their home was 63, which was three years older than in 2009-2010 (Kornilaki, 2024). This shift in intergenerational dynamics highlights the challenges fathers anticipate facing as their children's grandparents grow older, particularly as the children enter adolescence and grandparents' increasing age will prevent them from remaining part of the caregiving team. Findings in this study cement the increasing

concern for the void that ageing grandparents will leave in the family unit when they can no longer help to look after their grandchildren.

Although grandparents were an equal source of support, they can equally evoke emotions of anger, disappointment and resentment in fathers. This is particularly in relation to the lack of understanding and interest in their children's conditions. The absence of empathy regarding the symptoms associated with the CA such as behavioural challenges and fluctuating life expectancies was particularly pronounced. Research shows that older individuals tend to hold more negative attitudes towards people with disabilities than younger people, which is hypothesised to be because of greater exposure and integrative and inclusive education (Domagała-Zyśk, 2021). In Ireland, this lack of understanding about people with disabilities is evident, with a 2017 survey showing that only 30% of participants believed that people with disabilities are treated fairly in Irish society (National Survey of Public Attitudes to Disability in Ireland, 2017). There is a notable gap in the education of family members regarding their children's medical conditions, which can subsequently lead to insufficient support for both fathers and their children. A recent study conducted in Ireland, which examined families of children with rare diseases, including CA, revealed that 17.6% of respondents faced challenges when attempting to explain their child's condition to parents or other relatives (Somanadhan et al., 2025). Notably, most fathers in this study reported taking the responsibility of informing their own parents and the mothers' parents about their children's diagnosis and its implications.

7.4 The physical and psychological space occupied by fathers (lived space)

The third existential theme of "lived space" determines the simultaneous occupation of space, both physically and psychologically, that caring for a child who has a CACCN has on fathers. Complex decision making, long hospital admissions, transitioning home, receiving a homecare package, and the fight for respite and future residential care all take up both cognitive and physical space in fathers' lives. Individually, these environmental aspects of care are not a new phenomenon in the complex care arena, yet these issues are predominately from the experiences of the mother. Findings therefore give a unique viewpoint from the fathers' perspective.

Within the subtheme of “lived space” three core concepts are discussed. Firstly, the transition from hospital to home is examined, with particular focus on how national procedures shape and contextualise this experience for fathers. This includes their experiences within the hospital setting, the discharge process, and the navigation of home care packages. Secondly, this theme addresses fathers’ ongoing concerns about their children’s long-term living arrangements, situating these experiences within the broader contexts of respite care and future residential planning. Finally, this theme explores the complexity of decision making that fathers face, including considerations around procedures and decisions regarding their child’s educational placement.

Children with a CACCN are at an increased risk of requiring hospital admissions due to issues such as increased medical needs, intellectual disabilities, susceptibility to infections and have more serious subsequent illnesses than typically developing children (Baicher et al., 2019; Glover et al., 2019; Marçola et al., 2023; Takita et al., 2024; Wang et al., 2016). Survival rates of children with conditions that previously have been considered as incompatible with life such as Trisomy 13 and Trisomy 18, have now increased, because children with these conditions can receive medical interventions such as mechanical ventilation, feeding tubes, cardiac surgeries and tracheostomies that require long hospital admissions (Marçola et al., 2023). Despite the survival rates of children with Trisomy 21 across all European States, with a 92.7% survival rate at 1 year of age, 80% of these children need at least one hospital admission in the first year of life (Seaton et al., 2025). One English study showed that over a ten-year period, 175 out of 231 children with Trisomy 21 had attended the emergency department at least once (Baicher et al., 2019). Furthermore, a Brazilian study showed that of children with Trisomy 21 that are admitted to the acute setting with respiratory or cardiac issues, roughly one-third required admission to the intensive care unit (Takita et al., 2024). A study by Gallagher et al., (2017) showed that across a 12-month period in Ireland, 40% of children aged 0-4 and 47% of children aged 5-12 with Prader-Willi syndrome that were admitted to hospital required an acute hospital stay. Therefore, for many fathers of children with a CACCN, these hospital admissions are inevitable and understanding their experiences during these acute admissions is vital.

Fathers in this study reported experiencing fear of not wanting to leave their children in hospital in the care of hospital staff, due to a lack of trust in staff competencies. Evidence shows that when a child has CCN there is often parental mistrust in healthcare workers' ability to care for their complex children, resulting in fear for their children's safety while they are in hospital (Ong, 2024; Oulton et al., 2020; Sharkey et al., 2014). Hospital staff themselves have reported that they feel less competent in caring for children with intellectual disabilities (Oulton, 2018), with some research showing these children experience inequitable quality and safety outcomes in the acute setting (Mimmo et al., 2022). Healthcare professionals have reported difficulties when attempting to provide holistic care to children with CCN in the acute setting, highlighting issues such as time constraints, staffing levels, low confidence levels as well as challenges in organisation of multiple subspecialty interests (Mclorie et al., 2023). Issues with the children's nursing workforce has been a concern internationally. Although the ROI is currently undergoing a positive transformation in the delivery of care for children (Brenner & Begley, 2019), with the imminent opening of the new children's hospital (CHI), staffing levels remain a concern. Research has shown that patient safety is negatively affected by unsafe nursing ratios and skill mix (Aiken et al., 2002a, 2002b, 2018; Griffiths et al., 2019). In the UK, there is a widening gap between the number of children and the number of qualified nurses. Approximately 25% of the UK population are children, yet only around 5% of registered nurses are qualified in paediatric nursing (Viner et al., 2017).

This mistrust resulted in many fathers either taking on the full-time care role within the hospital or taking on additional roles within the household so that the mother could spend more time in hospital. However, the findings identify some gender specific challenges in the acute setting such as chairs being taken away or conversations happening when they were not present. At times fathers deemed this to be a barrier to their equal participation and left fathers feeling unwelcome. Previous research highlights that negative social factors, such as gender bias and restrictive gender norms can limit fathers' involvement in their children's hospital care (Watkins et al., 2024).

This study highlights the complexity of navigating life when their child was needing long term hospital admissions. Unlike other research, some fathers in this study reported

spending more time in the hospital with their child than the mother, citing factors such as financial considerations, where the mother was the primary breadwinner, or the mother's inability to bond with the child as a direct result of their child's diagnosis. Some research shows that during acute hospital admissions, fathers typically spend less time with their hospitalised child compared to mothers, yet there is no significant difference in emotional distress levels between fathers and mothers during such admissions (Mangelsdorf, 2019). Therefore, the emotional toll fathers face is noteworthy, given that many fathers in this study described assuming additional responsibilities, particularly in managing siblings and balancing work commitments which they deem necessary to keep the family unit going.

Most of the literature on the experiences of parents of children with CCN in hospital, is the mother's perspective, with many titles stating 'parents', yet findings from fathers are not included (Murdoch & Chang, 2022). Some of the issues faced by fathers are similar to that of the mother, and indeed all parents of children who are in hospital, this included issues such as the financial cost of hospital admissions and the lack of appropriate accommodation (Nassery & Landgren, 2018). This is more apparent for parents of children with longer admissions and given the evidence that children with CA have increased frequency and length of hospital stays, this is a significant aspect in their lives. Research carried out by Children in Hospital Ireland and Childhood Illness, Financial Stress (2020), highlights the range of non-medical financial costs which may be incurred by parents of children needing prolonged or frequent admissions (again only 9.7% of these respondents were fathers). As often fathers report that a large proportion of their worries are financial in nature, understanding this financial strain is vital, as often, fathers not being present in hospital at all times is not a choice, but a family necessity.

The need for better communication and the use of interpreters was a significant finding in this study. Research shows when parents give operative consent for children to undergo emergency surgery, there is poor parental retention of this information (Li et al., 2014). The importance of giving parents written information during healthcare related decision making is also highlighted, with professionals needing to ensure

individual parent's information needs are met (Wu et al., 2023). In 2022 in the ROI almost 14% of the population were non-Irish nationals, with the 2016 census reporting that 86,000 people did not consider themselves to be able to speak or understand English well (Central Statistics Office, 2017). An interpreter is a person who speaks more than one language and can assist communication with individuals or families (HSE, 2024) and is needed when delivering healthcare related information. Yet, one study found that in a paediatric emergency department, when verbal interpretation was needed, this was completed by untrained individuals such as relatives and friends, ad-hoc bilingual medical staff or not at all (Dungu et al., 2019; Health et al., 2023). Nurses themselves have reported using bilingual nurse colleagues and family members when healthcare interpreters were unavailable (Seremetkoska et al., 2025). In Ireland a HSE guideline "On Speaking Terms: Good Practice Guidelines for Staff in the Provision of Interpreting Services" (HSE, 2009) highlights the importance of competent interpretation, yet Ireland does not have any regulatory standards on use of healthcare interpreters (Martín & Phelan, 2009).

Once their child was discharged, fathers' spoke of a period of adjustment. Some spoke of the fear of their children's conditions, and being away from the hospital in the event that something went wrong. This is a common concern with families of children with CCN during this discharge process (Amar-Dolan et al., 2020; Condon, 2020; de Riet et al., 2023). However, these findings further elaborated on fathers' perceived roles in the practical challenges of discharge, such as adapting the home environment to accommodate their child's needs. Fathers documented the change in their physical space, mostly related to storage of equipment, which included items such as hoists, non-invasive ventilation, suction machines, and mobility aids. Furthermore, fathers spoke of having to rearrange their homes to accommodate their children's discharge or HCP, such as converting kitchens or sitting rooms into bedrooms. These findings are somewhat similar to Woodgate et al., (2024), who reported that fathers help make participation in everyday life possible for their children with CCN through organising accommodations and adjustments (e.g., modifying physical environments, adapting or buying devices and needed technology). This was not noted as a major finding in a systematic literature

review exploring parents' experiences of children with CCN being discharged from hospital to home (Ronan et al., 2019). Instead, this review highlighted that many parents report the emotional loss of leaving the hospital as a result of leaving the support and relationships built in the hospital (Ronan et al., 2019). Yet much of the literature around the experiences of "parents" when their child with CCN is discharged, are not inclusive of fathers, and often the mothers voice outweighed the fathers (Brenner et al., 2015; Callans et al., 2016; Desai et al., 2016; Lakshmanan et al., 2019; Leyenaar et al., 2017; Zanello et al., 2015). This highlights that mothers and fathers may have different priorities and deal with the challenges of having a child with CCN differently. Thus, understanding and supporting individual needs throughout these transitions is important.

Safely discharging children with CCN home can be a challenging experience for both families, and healthcare providers. National policies are in place to help support families of children with CCN live at home. The 'National Model of Care for Paediatric Healthcare Services in Ireland' (HSE, 2016) highlights the importance of enhanced community care for children (HSE, 2016), and the 'Slaintecare' policy which aims to ensure a transition from acute care to community-based supports (Department of Health & Health Service Executive, 2023). However, delays in implementation of these policies are still apparent, and fathers identified that waiting for HCP hours to be approved was often the cause of delayed discharges. Families of children with tracheostomies in Ireland highlighted their frustrations that despite their children being medically well, discharge from the acute setting was delayed due to a lack of HCP (Brenner et al., 2015). Similar prolonged admissions for children with CCN are seen internationally, with delayed discharges being primarily due to lack of home nursing (Amar-Dlan et al., 2020; Antoniou et al., 2022; Chatoos et al., 2020; Gong et al., 2019; Sobotka et al., 2019).

Conversely however, one father in this study recalled being discharged too soon, without adequate support or education on his child's condition, which resulted in a readmission. High readmission rates of children with CCN are evident with one study showing a readmission rate of 27% during the first 30 days of discharge (Musial et al., 2020). Research shows that nurses need to assess children and family's readiness for

discharge to reduce readmission rates (Zhou et al., 2021). It is imperative that the multidisciplinary care team liaise with parents in planning for discharge. The term 'complex discharge' is used when referring to children who are being discharged from hospital, but who will require ongoing specialised care to live at home (Wellchild, 2024). In the UK there are specialist nurses known as discharge coordinators who help co-ordinate this complex process (WellChild, 2024). These are a senior nurse who specialises in discharging children with CCN (van de Riet et al., 2023). In the ROI, this role is the complex needs co-ordinator, and these are pivotal in enabling children to be cared for in the community.

Children with CCN cannot be discharged home without a HCP, and this process in Ireland starts with an assessment assessing children's complexities to help identify the type of HCP needed. Nursing assessment is fundamental to the planning and delivery of care for children with CCN (Toney Butler et al., 2019). An assessment tool is required to assess the level of clinical needs and is an integral part of the discharge process (HSE, 2014). In the ROI, the tool that is used was adapted by the "NHS Northern Irish assessment tool and the Paediatric Community Assessment Tool" (P-CAT) (HIQA, 2021). The Irish P-CAT was introduced following an education program in 2019 (HIQA 2021), and it is supported by a guideline on the completion of the Paediatric Community Assessment Tool for Nursing (P-CAT) (2021). This is used by scoring 11 domains that give a score for the complexity, risk, frequency and severity of the child's condition (P-CAT Tool). Funding is then approved by the public health department, and the local public health team invites the providers to apply to provide services for the child. This process is underpinned by the national guidelines and by a national tender. There are private, voluntary, and not-for-profit homecare providers that are subject to monitoring that have a service agreement with the HSE. The successful provider then enters a service arrangement with the HSE (HIQA, 2021).

P-CAT assessments are then completed regularly by public health nurses, increasing or decreasing the hours as necessary. However, there continues to be some ambiguity around how to use the P-CAT with public health nurses requesting updates and further education on the P-CAT process (The Centre of Children's Nurse Education (CCNE),

2024). Since the commencement of the P-CAT tool in 2019, there have been two audits on its efficiency and revisions have been made in the hope of supporting these families with appropriate care. Yet, many fathers in this study highlighted that once the medical needs subsided, managing their child's behaviour was the most challenging aspect of care, and was the time they needed the HCP the most, yet hours were then reduced without the availability of out of home respite. Within the P- CAT challenging behaviours are part of the unscored domain (P-CAT., 2022). Meaning, that no matter how challenging this behaviour is, it does not count towards the children's overall score of complexity, which is used to assess their need for in-home support.

Another unique finding from this study is the experiences of fathers of children with a CACCN whose children have a PHCP. Many fathers reported positive support from providers, yet some challenges were identified. Some felt a significant lack of trust in staff competencies to the extent that one father reported cancelling the package. Current standards and requirements for general nurse education (NMBI, 2016) and across Europe have a limited focus on children and do not prepare nurses adequately to deliver the highly specialised care needed to care for these children with CCN (Clancy et al., 2021). Staff working with these children in the community do not need to be a Registered Children's Nurse (RCN) with HIQA (2022) suggesting that this care delivery of children's HCP in the community requires nurses with children's nursing experience. Although many registered general or intellectual disability nurses are providing excellent care to these children, there is a clear need for these nurses to have additional training opportunities when caring for children with CCN in the community (CCNE, 2024). Leading the Way: A National Strategy for the Future of Children's Nursing in Ireland 2021-2031 highlights the ongoing need for ongoing examination to ensure that nurses are reflexive and adaptive to the changing complexity of many children (HSE, 2021).

The CCNE in CHI (2024) completed a national learning needs analysis which included a survey of nurses caring for children with CCN in the community. Over a quarter of these nurses (26%) reported they had not engaged in continuing education specific to children with CCN, and 98% highlighted a desire for more educational opportunities (CCNE, 2024). This lack of confidence and competence is not just in the Irish context, with nurses

internationally reporting the additional support in their education and their emotional needs in working with this cohort of children (Chong & Abdullah, 2017; Connor & Corcoran, 2022; Desai et al., 2016). The CCNE identified recommendations with the hope of implementing a diverse education in multiple clinical skills, communication and working collaboratively with children and families (CCNE, 2024). This is an important step in ensuring fathers of children with a CACCN do not feel they need to cancel vital support such as HCP due to lack of staff skills.

Furthermore, evaluating governance and standards of care is vital within healthcare structures (Ghavamabad et al., 2021), as regulation can be a positive influence on quality maintenance and improvement in healthcare practices (Behan et al., 2024). This helps ensure best practice, and safe care is delivered. Yet, there is currently no independent regulation for children's HCP in Ireland, with the Ombudsman for Children reporting that it cannot be assured that children with CCN are being provided with consistent, high quality and safe care (Ombudsman for Children, 2025).

Not all children in this study had a HCP. However, many fathers spoke of the importance of both day and night respite with parental burnout also evident in the international literature of children with CCN (Patty et al., 2024). For some, this was deemed essential for their wellbeing, while for others, their children were waiting for appropriate respite facilities to be open or were on significant waitlists. Some fathers of younger children reported this availability of respite care was something they considered as vital for future planning and would want their child to attend as they got older. Evidence of the benefit of children with CCN accessing respite is well reported within the literature, with families expressing that respite, is not a choice, but something that is lifesaving and necessary (Murphy et al., 2021; Woodgate et al., 2023). Yet, HIQA (2022) highlighted that currently there is limited access to respite which impacts significantly on the quality of life of the child and their families. This is not an Ireland specific challenge, in Canada, service providers have described respite care as being in "crisis mode" (Woodgate et al., 2023) and families have significant challenges with accessing, navigating and sustaining this care (Woodgate et al., 2023). In the UK, 64% of divorced or separated parents cited having a child with CCN as a contributing factor in the breakdown of their relationship,

and of those, 75% had no access to short breaks at the time- highlighting the critical impact of limited respite services (Together for short lives, 2020).

Fathers in this study highlighted their continuous worry as to where their children will live in the future. This was both for when they themselves could no longer care for their child, but equally, they reported their own children's desire to live independently away from them. One survey of people with intellectual disabilities reported that 40% of people living with their parents wanted to move out from their family home (Inclusion Ireland, 2024). Yet, fathers felt their children's unique complexities make planning for this difficult. Fathers report needing to advocate for appropriate housing for people with their children's conditions. This was particularly prominent within their syndrome specific organisations, which included meeting with ministers for health about plans for residential care facilities within the community that are specifically designed in how to care for their children as they age.

The policy adopted in Ireland in 2012, "Time to Move on from Congregated Settings", aims to move people from living in isolated institutions and segregated campuses away from their communities (HSE, 2011). Historically, people with an intellectual disability were not regarded as citizens capable and valuable, but instead experienced daily restrictions on their lives (Murphy & Bantry-White, 2020). Currently, nearly four thousand people with disabilities are still living in congregated settings; including residential disability services and people under the age of 65 inappropriately living in nursing homes (HIQA, 2021; Ombudsman Report Wasted lives report, Inclusion Ireland 2021). Housing for all (2021) the Government's housing plan to 2030 aims that all people will have the opportunity to live in a home that is affordable. Yet, challenges for this population continue with the "Disability Capacity Review to 2032", highlighting that an average of about 90 new residential places will be needed every year from 2020 to 2032 to cater for changes in the size and age structure of the disability population, and currently the demand for residential care significantly exceeds supply (Department of Health, 2021) These waitlists include people living with very elderly parents, and younger people with CCN or behavioural issues who require intensive supports which exceed the abilities of individual family members (Disability Capacity Review, 2020).

Therefore, these findings of father's concern for their children's future housing is valid and can impact fathers' emotional wellbeing.

The inevitable challenges of their children's conditions were important to these fathers, and how, as they aged, they anticipated that they would not be able to provide this care. Some fathers felt that currently, residential options were not suitable for their children's conditions, particularly when the challenges of care were in relation to behaviours. Studies show that challenging behaviours coupled with a lack of respite are often the determining factors for parents making the decision to place their children into residential care, and often a choice that is made when families are in a state of crisis (Brown et al., 2011; Ellem et al., 2016). A recent scoping review that explored the prevention and management of crisis situations in children and young adults with a disability reported that a lack of support services alongside difficulties in navigating these services (if and when they are available), are challenges that can lead to the eventual crisis situations (Doyle et al., 2024). Findings from this study highlight that fathers are anticipating this as inevitable yet worry that residential care will also not be equipped to care for their children's complex behavioural needs.

Alongside the physical space discussed by fathers, the mental space involved in caring for a child with a CACCN is vast, and many fathers spoke of how they often needed to make difficult decisions quickly about their child. Research shows the importance of early conversations with family members that need to be flexible, rather than definitive decisions about end-of-life care and treatment. These need to be given by trusted staff in a private room, with time to think (Bennett et al., 2023; Carr et al., 2022). Yet for these fathers, as a result of sudden deterioration of their children, decisions had to be made quickly, which resulted in feelings of anxiety. One father directly attributed these experiences to his long-term mental health challenges and anxiety. Effective symptom management ensures comfort and improves quality of life for children with life-limiting conditions. Early, sensitive conversations about end-of-life care allow families to express wishes and make informed decisions. Advance care planning (ACP) supports children and families, guiding professionals to provide care aligned with their values and preferences. Research of children's ACP is a growing phenomenon, as healthcare

professionals internationally are encouraged to initiate ACP in paediatric complex care (Engel et al., 2024; Rapoport, 2024). Yet, empirical evidence on families' experiences is still limited (Lusney et al., 2023). Moreover, the experiences of fathers are even more scarce with much of the research either focusing on mothers, or fathers being less represented (Bennett et al., 2024; Carr et al., 2022).

This study highlights the internal conflict fathers face in placing their children in mainstream education versus special education but the desire for inclusive education was paramount for most fathers. Many fathers had to fight for the rights of their children to be educated with their peers and within their local community. Since the UN Convention on the rights of the child (1989) and the Salamanca statement (UNESCO, 1994), most European countries have introduced policies and practices to support children with disabilities to have inclusive education (Arcidiacono & Baucal, 2020). Ireland ratified the UN Convention on the Rights of Persons with Disabilities in 2018, and Article 24 is the first legally binding instrument to mandate inclusive education (Broderick 2014, Broderick 2018). Yet, despite best intentions and significant advancements, there are also many challenges and barriers to inclusive education in Ireland and across Europe (Howe & Griffin, 2021). Special schools still function in the majority of European countries as mainstream schools are not equipped to manage their additional needs (European Agency, 2021). Although there have been some positive developments, such as the Education (Admissions) to School Act 2018 and the pilot School Inclusion Model scheme, which advance access and participation for children with disabilities in inclusive education (Murphy et al., 2023) - findings from this study conclude that families are still facing challenges accessing school nurses. One father recalled successfully advocating for his son to be part of the "National Nursing Pilot for Children with Complex Healthcare Needs" (NCSE & HSE, 2023), which was a pilot initiative, for children with CCN to attend mainstream school with their community or homecare nurses. Although this initiative is positive, it is not yet available for all children with CCN.

Although full inclusivity in education remains a global challenge, some Canadian provinces are regarded as leaders in advancing inclusive education practices. A recent

critical review of Canadian literature by Mann et al. (2024) sought to develop a conceptual framework outlining the drivers and barriers to the closure of special schools. Notably, the complex medical needs of children were not identified as a barrier within this framework. Fathers in this study highlighted that the children's medical complexities were a barrier to inclusive education and the struggle to secure placements in special schools emerged as a recurring theme in this study. This aligns with findings from Inclusion Ireland (2024), which reported that 45% of children requiring additional support are being denied equitable access to education. A key barrier is the lack of local availability, often forcing families to consider schools located significant distances from their homes. This shortage of special school places has posed an ongoing challenge in recent years, as highlighted in the "Plan for Places report" (2022). According to the Department of Education (2024), as of September last year, 126 children in Ireland remained without a school placement, underscoring the persistent gaps in educational provision for students with CCN.

When children did receive a special school place, fathers sometimes felt regret about their decision in where they decided to send their children, in both special and mainstream education. This was due to the perceived lack of school staff's understanding of their children's conditions. Research shows a lack of quality specialist training for both teachers and special needs assistance of children with special educational requirements (Murphy et al., 2023). Fathers felt this poor understanding caused negative health and social implications for their children. This was particularly prominent in relation to food related policy, in which some fathers believed that feeding challenges associated with their children's CACCN were not being met. Research on school staff teaching children with CAs often report being unprepared and perceive their training in specific conditions limited (Castro-Kemp & Rawlinson, 2024).

7.5 The material aspects of care (lived things)

Findings related to the fourth existential theme of "lived things," provide insight into how fathers experience the material dimensions of everyday life while caring for a child with a CACCN. This theme encompasses the physical items and financial realities of caregiving, including the economic burden and the difficulty of balancing employment

with the intensive demands of care. For many fathers, these material aspects significantly shape their caregiving experiences, as they navigate the pressures of sustaining a household while meeting their child's CCN. However, this existential theme appeared to hold less prominence in the narratives of fathers compared to other dimensions of their lived experience, suggesting that while material challenges are present, they may be overshadowed by emotional, relational, or temporal aspects of caregiving.

Within this theme, two key concepts emerged. First, fathers described the significant financial implications associated with meeting the CCN of their children, including out of pocket costs for medical equipment, therapies, and homecare packages. These additional expenses often placed considerable strain on family finances. Second, the theme highlights how fathers' caregiving roles are shaped by their employment contexts, with particular attention to issues surrounding parental and paternity leave. Challenges related to limited or inflexible leave entitlements both in Ireland and internationally are discussed in the context of how this impacts fathers' ability to participate fully in caregiving, often reinforcing traditional gendered divisions of labour and limiting their involvement in day-to-day care.

There was a noteworthy appreciation for the support offered by the Irish healthcare system including medical equipment, home respite and homecare packages. However, when these medical needs subsided, care was reduced - resulting in the need for private support. Some fathers of children with a CACCN are paying privately for HCP, yet data that reports the cost of private home care only includes care provided to adults, the equivalent data on services provided to children is unavailable (Ombudsman for Children, 2025). Fathers reported when they had a state funded HCP, they still needed to pay privately for a second trained individual to be in the home, due to the *loco parentis* clause. This clause requires that, while a qualified nurse is caring for a child with CCN there also needs to be 'a designated competent individual' to be in the home alongside with the trained nurse (Jack and Jill foundation, 2018). Therefore, findings from this study suggest that this clause is preventing adequate provision of in-home respite.

The findings from this study acknowledges the positive and vital support that the Irish government provides in relation to allowances and tax credits, as these alleviate some levels of stress experienced by fathers and families. Research shows the positive impact that benefit entitlements have in the quality of life of parents with children with a CACCN (Chen et al., 2023). The Irish government provides a range of allowances, tax credits and entitlements that all fathers in this study were entitled to, and for the most part in receipt of (See Appendix 15). These allowances are fundamental, as internationally, families of children with CCN are at an increased risk of living in poverty due to a lack of welfare support, or the cost of disability exceeds the amount of welfare payments (Choi, 2025; Schuelke et al., 2022). In 2021, for the first time, the Irish government identified that there are additional costs to having a disability (Inclusion Ireland, 2025). According to the Government's "Cost of Disability Report", the additional expenses associated with disability have risen significantly in recent years due to inflation (The Department of Social Protection, 2021). The Social and Metrics Commission (2020) indicates that 50% of those in income poverty are part of a family that supports at least one person with disabilities. In the UK, families with a disability had a poverty rate of 26% compared to 19% of families in which no disability was present (Joseph Rowntree Foundation, 2020). In Ireland, children are twice as likely to experience poverty if someone living in their home has a disability (Russell et al., 2025). For one out of five of these children, the person living with disability is a child (Expert Advisory Panel, 2020). In the United States families with a child with a disability are more likely to experience income to poverty ratios than those families without children with disabilities (Costanzo & Reilly, 2025) due to increased living costs, reduced parental employment, inadequate support, and systemic barriers.

Balancing work and care demands is a prominent finding, which is in keeping with other literature relating to fathers of children with CCN (Giallo et al., 2015; Marshak et al., 2019; Schippers et al., 2020; Uribe-Morales et al., 2022). Many of these concerns were during long hospital admissions, particularly during the neonatal period. Some fathers felt guilty at having to return to work, often while their infant was still in hospital when their paternity leave ended. While other fathers reported needing to take unpaid sick

leave after returning to work too soon. This study highlighted the challenges fathers faced in taking time off work, with concerns particularly related to paying their mortgage. In recent years, policies have been put in place to encourage fathers to use their paternity leave entitlements. Europe has introduced the “EU Work-Life Balance Directive” for parents and carers throughout the European Union (Directive EU, 2019). This strategy seeks to improve the balance between work and family life. This includes fathers' rights and paternity leave, these are among the directives' most important features (Directive EU, 2019). Around the birth of the child, fathers are encouraged to take at least 10 working days of paternity leave, that is compensated at least at the level of sick pay. Additionally, it recommends that both parents have four months (unpaid) parental leave, two of which are nontransferable between mothers and fathers. Furthermore, carers, who are workers providing personal care or support to a relative or person living in the same household, are entitled to carers' leave of five working days per annum (Directive EU, 2019). The aim of this policy is to make paternity leave financially viable, to encourage fathers to use it. Paternity leave in Ireland can be postponed for a maximum of six months if a child is hospitalised, but this does not take into consideration that for some babies with a CACCN, delayed discharges often occur (Citizen's information, 2025).

Research shows that paid paternity and parental leave can help to mitigate work- care conflict and is one of the most significant ways to aid fathers to achieve active parenthood (André et al., 2025; Pizarro & Gartzia, 2024). Yet challenges remain, with a recent study estimating that only 26% of fathers in Ireland in 2021 availed of paternity leave, compared to 69% of eligible mothers who took such leave (Keane et al., 2025). Compliance with the EU Work-Life Balance Directive has been uneven across many countries including Ireland (Zumbyte & Szelewa, 2024). While Ireland has made some progress in implementing this policy, full compliance is still pending. Key provisions, particularly regarding paid paternity and parental leave, have yet to be fully implemented. Ireland along with some other EU states such as Belgium and Spain could be referred to the Court of Justice of the European Union to impose financial sanctions (European Commission, 2025).

In the ROI, entitlements for mothers and fathers for the care of young children are generally on a par with other EU countries and the UK in terms of duration, however the value of benefits is amongst the lowest (Keane et al., 2025). The two-week paid paternity leave in Ireland is €289 (Citizen's information, 2025), leaving it to individual employers' discretion whether to top-up this pay. Yet in 2023, the median weekly earnings of males between the ages of 30-39 was €807.69 (Central Statistics Office, 2024). This is a significant pay reduction if fathers were to take parental leave without top-ups from individual employers. Recent research found that uptake of paternity leave increased with males that earned a higher salary, and for public sector workers whose employers topped up their parental leave (Keane et al., 2025), highlighting the challenges many fathers face in taking this entitled leave depending on their employer.

Findings from this study show that limited flexibility for fathers of children with a CACCN will impact how they use their paternity leave. In Ireland, if their child is in hospital during those two weeks, fathers can postpone their paternity leave (within a 26-week window). Some countries have made strides in recognising the financial burden of hospitalisations of sick children, adding additional provisions for fathers (as well as mothers) when a newborn is sick, hospitalised, or premature, by offering extended or flexible paternity or parental leave. Sweden for example has "temporary parental benefit", in which parents can take up to 120 days per child (until 12 years old), at 80% of their income (UNFPA, Georgia 2018). Some reforms have come into fruition for mothers in Ireland since 2017, such as extended maternity leave for a premature baby (The Department of Social Protection, 2019), yet this does not include fathers, despite the evidence of the importance to the whole family of fathers being present.

7.6 Adapting to their children's chromosome abnormality and complex care needs changing over time (lived time)

The final existential theme of "lived time" concludes how past, present and future experiences of caring for their child impact their current everyday lives. According to van Manen (2014) time is experienced subjectively, and it is vital to how people interpret their lives. Findings in this study capture the multifaceted nature of their daily lives, in how fathers manage their responsibilities, at times in a fragmented manner. This was

particularly prominent in addressing their children's mental health and behavioural challenges. Fathers spoke in the present tense, about what life was like for them now, but this experience was influenced by their children's conditions and their past experiences. They also spoke about how their journey has reached this point in time, and how they became competent in caring for their children. Furthermore, fathers spoke of the anticipatory challenges that were to come, concluding that their children's needs are dynamic, and their complexities change over time which in turn changes how fathers engage with their children.

Within the theme of "lived time", three key concepts emerge. First, the emotional impact of fluctuating symptoms associated with children's CACCN is explored, particularly as these changes often affect perceptions of their child's life expectancy, placing a hidden yet significant emotional burden on fathers. Second, the evolving nature of children's behavioural challenges is discussed, with specific emphasis on the lack of adequate mental health support to assist families in managing these changes over time. Finally, the relentlessness of the caregiving role is highlighted, as fathers describe the continuous nature of their responsibilities across a 24-hour period. Sleep deprivation emerges as a prominent and ongoing concern, underscoring how caregiving frequently extends into the night, further compounding the physical and emotional demands placed on fathers. Fathers' mindset often evolved over time as they adapted to the challenges of raising a child with a CACCN. While immediate medical crises often lessened as their children grew, an enduring sense of impermanence and anxiety remained. Research shows that negative experiences with their children's health can have negative paternal implications such as anxiety, PTSD and depression, with much of the literature relating to traumatic childbirths and poor neonatal outcomes or the NICU (Cross, 2024; Golubitsky et al., 2024; Inglis et al., 2016; Thomas & Anderton, 2021; White, 2007) or trauma related to complex medical events such as cardiac surgeries or cancer (Franich-Ray et al., 2013). Research on fathers who had experienced a pregnancy or childbirth complication including a significant congenital abnormality, found high rates of distress in fathers, which could persist for months after the event (Kothari et al., 2022). For fathers in this study the levels of anxiety persisted due to their children's

adverse events, despite in some cases many years having passed. Findings in this study present a new viewpoint, of how fathers felt traumatised by expecting their child to die, and how they adapted when their child exceeded their projected life expectancies at time of diagnosis, and their needs changed. Research shows that supporting the integration of trauma-informed care practices and advocating for parental mental health services could better support fathers to overcome some of these challenges (Dewan et al., 2023).

This study details how fathers report living in the now yet always anticipating a change. Parents of children with a life limiting diagnosis have reported the concept of time, sometimes referring to this being in limbo, borrowed time or mourning without a coffin, as they anticipate the death of their child (Côté-Arsenault & Denney-Koelsch, 2011; Najafi et al., 2022). The term “anticipatory grief” was coined to describe the emotions that a person experiences prior to losing someone they love (Lindemann, 1944). Rando (2000) explored this loss as living in the past, present and future. Research shows there is little difference between mothers and fathers’ anticipatory grief in children when children are born prematurely and may have complex disabilities as a result (Valizadeh et al., 2013). Findings of this study show that living with anticipatory grief extends to fathers of children caring for a child with a CACCN and highlights the need for healthcare professionals to remember and be mindful of the needs of fathers during this time.

However, this grief was not always in the context of potential death or even medical complexities, instead fathers in this study anticipated the symptoms of their children’s CA and grieved the current child they have or had. This was particularly prominent in relation to the complex behavioural challenges of some of the CA trajectories. Much of the literature that refers to parental grief in the complex needs arena, is related to either life limiting conditions in which life expectancy is shortened, or when their child is diagnosed originally (Barros Torquato et al., 2013; Bentley et al., 2016; How et al., 2019; Hurley et al., 2021; Marshak et al., 2019). Yet, the grief related to the changing behavioural needs are not typically included in the CCN umbrella, although emotional and behavioural conditions are factors that reduce caregiver quality of life (Huynh et al.,

2025). This study highlights that behavioural complexities associated with children who have a CACCN are particularly significant and contribute substantially to caregiver burden. Domaradzki et al. (2024), in a study of (mostly mothers) children with Williams Syndrome, found that 62.5% reported severe or very severe health problems in their children, with behavioural and mental health challenges being the most frequently cited concerns. Similarly, Kayadjanian et al. (2021), examined the interplay between hyperphagia, anxiety, behavioural difficulties, and caregiver burden in families of children with Prader-Willi syndrome, found that behavioural issues had the most profound impact on caregivers (again, primarily mothers). These findings underscore the central role that behavioural and psychological needs play in shaping the emotional strain of caregiving for children with a CACCN, this behavioural dimension was found to contribute not only to current experiences of grief but also to anticipatory grief, as they grappled with the ongoing and future implications of their child's behavioural challenges.

Children's self-injurious behaviours (SIB) caused a huge amount of stress for these fathers. Many children with a CA such as William syndrome, Prader-Willi syndrome, Trisomy 21, Cri de chat and Fragile X Syndrome can present with commonalities in behavioural traits including SIB (Buono et al., 2010; Crapper et al., 2015; Erickson et al., 2011; Huisman et al., 2018; Moskowitz et al., 2015). Crapper et al. (2015) defines SIB as behaviours performed with the intent of physical self-harm but without the intent to die. Fathers in this study noted how challenging SIB can be, for example fathers of children with multiple abnormalities noted how anxiety caused their children to pick their skin. Skin picking has been seen to be extensively reported in children with Prader-Willi syndrome and William syndrome and is a widely recognised feature of the behavioural phenotype of these conditions (Kozel et al., 2014; Woodruff-Borden et al., 2010), yet fathers noted access to mental health services in the CDNT for their children are limited.

In the ROI, recent publications indicate that disability and mental health supports are not being adequately provided by the State and have been insufficient for a sustained

period (Joint Committee on Children, Equality, Disability, Integration and Youth, 2024). For families of children with CACCN the service landscape is particularly difficult to navigate due to its fragmented and conditional nature. The HSE Child and Adolescent Mental Health Services (CAMHS) accepts referrals for children with moderate to severe mental health difficulties, including those with mild intellectual disabilities (HSE, 2025). However, recent reports by Families for Reform (2024) highlight that, in practice, children with intellectual disabilities are often not accepted into CAMHS unless they are already under the care of a CDNT. Given the current crisis in CDNT services, as previously discussed, many of these children remain without access to appropriate mental health support. For children with moderate to profound intellectual disabilities, referrals are directed to a separate service known as Child and Adolescent Mental Health Services-Intellectual disability (CAMHS-ID). However, Families for Reform (2024) also noted that CAMHS-ID operates only in select catchment areas, meaning that access to these specialised mental health services is largely dependent on geographic location, resulting in inequitable service provision across the country.

A lack of mental health services, internationally, have been attributed to an increase in the number of children with intellectual disabilities and/or autism presenting to the emergency department as a result of SIB in crisis. One study reported that 129 children with autism/intellectual disabilities presented to Melbourne emergency department due to an inability to manage their child's behaviour and/or by carer burn-out (Effron et al., 2023). Research shows the need for families of these children to have improved access to appropriately skilled community health professionals, allowing their behaviours to be addressed as they emerge, rather than at crisis point (Pillai et al., 2023). Yet, all fathers in this study reported the lack of respite and CDNT support.

Findings from this study present a unique perspective specifically around the sleep challenges children with CAs experience and the resultant impact on fathers and their wider family units. A CA predisposes children to chronic sleep challenges, such as hypersomnia, narcolepsy-like phenotypes, insomnia, sleep distorted breathing or sleep apnoea, which often need treatment such as medication or non-invasive ventilation

(Gwilliam et al., 2020; Kettler et al., 2020; Naime et al., 2021; Shankar et al., 2024; Sorina et al., 2020; Thottam et al., 2015). Fathers whose children did not have sleep challenges still reported interrupted sleep due to having to constantly remain vigilant, which is in keeping with the literature (Geuze et al., 2022; McCann et al., 2015). Sleep problems can be ongoing, and change over time and age (Abel et al., 2017; Bassell et al., 2015). With research showing that chronic sleep issues can impact long-term behaviours of children with CAs and can increase attention deficit and hyperactivity, and have poorer developmental outcomes (Soltani et al., 2025; Vaishakh et al., 2021). Furthermore, it can impact on family functioning and impact on siblings (Cooke, 2022; Esbensen et al., 2021).

Many fathers in this study reported to be the ones to manage nighttime sleep challenges more than the mother. One study exploring parental experiences of Smith-Magenis Syndrome (SMS) (deletion of chromosome 17p11.2), when compared to the fathers, mothers reported less interrupted night's sleep over the week and a slightly higher hours of sleep per night than fathers highlighting another difference between parental roles (Foster et al., 2010).

Fathers reported needing to go privately to sleep therapists and paediatricians for medications to help aid sleep as they felt this wasn't taken seriously by public health professionals. Research shows that when some parents attempt to seek treatment for their child's sleep difficulties, they experience insensitive comments and healthcare professionals normalise children's sleep difficulties among those with a clinical cause for sleep disturbance (Chawla et al., 2022). In one study sleep issues were spoken about more frequently in caregivers of people with Prader-Willi than issues related to hyperphagia (Patel et al., 2019). Yet, there is a significant lack of research on effective management and best practice guidelines. This is needed to enable a consensus for medical management of sleep challenges of people with CAs such as Prader-Willi syndrome, Williams syndrome and Trisomy 21 (Chawla et al., 2020; Duis et al., 2020; Gwilliam et al., 2021).

7.7 Fathers' experience as a whole

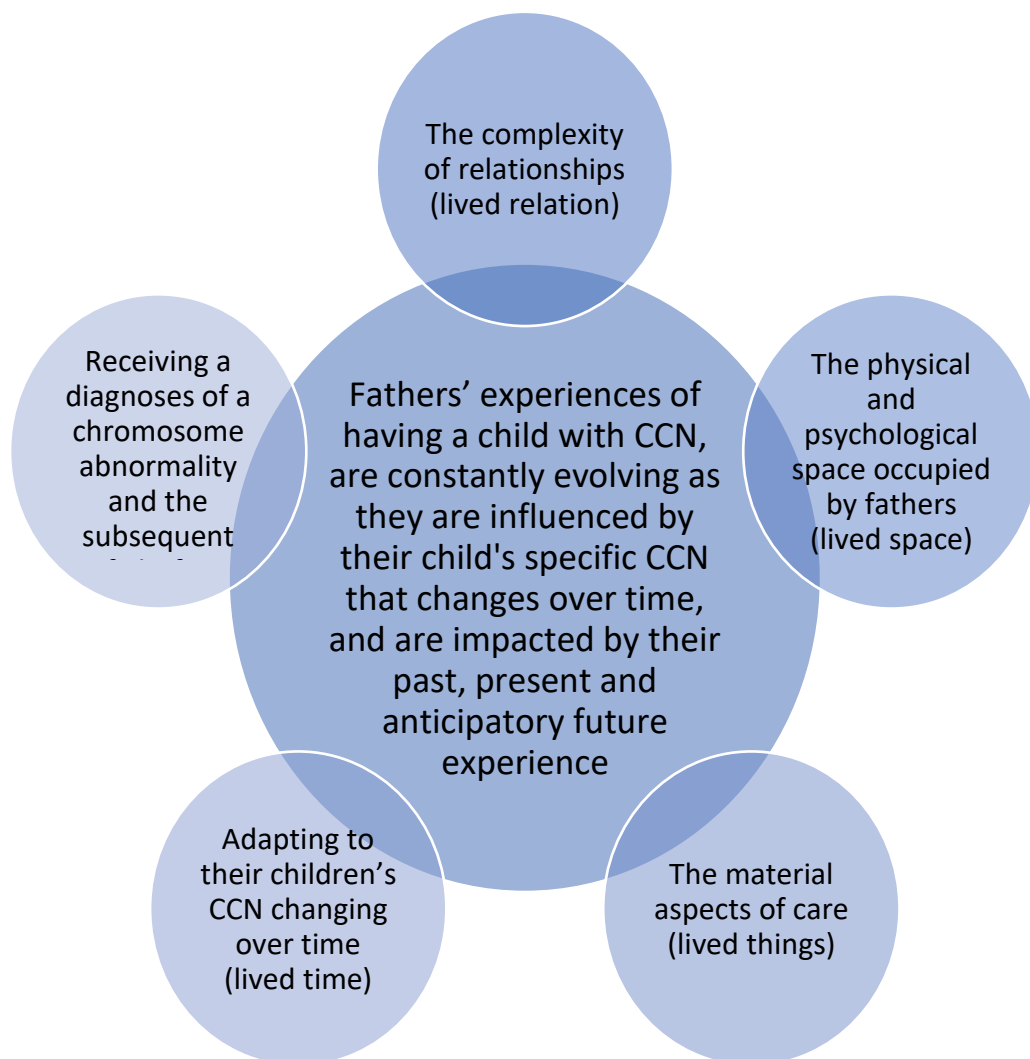
Although the five existential themes individually ascertain a certain degree of complexity, the hermeneutic phenomenology approach guided by van Manen (1990) argues the importance of maintaining a dynamic balance between the particular and the whole experience. This stresses the importance in attending to the nuanced details of the whole experience. While the existential themes prove useful and individually add to the existing body of knowledge on this phenomenon, they also need to be interpreted in relation to the totality they comprise. The experience of fathers within the existential themes often fluctuate and intertwine with one another throughout different stages and elements of their lives. When all five of these existential themes are brought together, a deep understanding of the father's experience emerges. This is what van Manen calls balancing of the parts and the whole. This *fusion of horizons* within the hermeneutic circle allows for the explication of meaning which is an uncovering of the dimensions of lived experience (van Manen, 1990 109-110). Through this process, meaning is not simply interpreted but embodied and evoked, allowing the essence of father's experiences to be felt as well as understood.

When examined in relation to the aims and objectives of this study, the existential themes show the multifaceted experiences that fathers face when receiving the diagnosis of a CACCN. Findings highlight how this diagnosis impacts their physical and emotional space, their relationships and how they experience time. Furthermore, findings identified a significant gap in service provision for children with a CACCN particularly in relation to ongoing therapeutic interventions and support to these fathers. Additionally, the findings conclude that fathers' have extremely important roles within the family unit, although at times these experiences are perceived differently to that of the mother.

Fathers' experiences are not confined to a specific event, and many reflect on situations that often-happened years ago, discussing them as though they occurred recently and clearly still impact their daily lives. While many fathers spoke about the present, this was often out of apprehension about the uncertainties of the future. Fathers experience significant delays in accessing disability services and the dynamics of their relationships

significantly shape how they perceive and cope with their children's diagnosis. Furthermore, their responses to their roles in caring for a child with a CACCN involves continuous adaptations and adjustments. Yet, the love and joy their children with a CACCN bring to their families and the wider communities outweigh these challenges. Therefore, this research concludes: "Fathers' experiences of having a child with a CACCN, are constantly evolving, as they are influenced by their child's specific CCN that changes over time, and are impacted by their past, present and anticipatory future experience" Figure 10 depicts how all the existential themes come together as the whole experience.

Figure 10: The existential themes come together as the whole experience



7.8 Conclusion

This chapter demonstrates that the study successfully achieved its main aims and objectives and contributes meaningfully to the existing body of knowledge on this phenomenon. It also illustrates the multifaceted experiences fathers face when receiving the diagnoses of a CACCN and presents a discussion as to the emotional impact this diagnosis has on them. Furthermore, the findings conclude that fathers' have extremely important roles within the family unit, although at times these experiences are perceived differently to that of the mother. Finally, findings identified significant gaps in service provision for children with CCN particularly in relation to ongoing therapeutic interventions and support to these fathers.

Findings from the first existential theme "lived Body" demonstrates that receiving a diagnosis of a CACCN is an emotional journey for fathers. Furthermore, they are facing significant challenges in accessing subsequent services. Findings from the existential theme of "lived relation" underscores the enhanced complexities that having a child with a CACCN has on fathers' relationships. These findings establish the intricacies of their relationships with the entire family unit, including all their children, the mother of the child and the grandparents. The third existential theme of "lived space" determines the simultaneous occupation of space, both physically and psychologically. Complex decision making, long hospital admissions, transitioning home, receiving a HCP and the fight for respite and future residential care all take up both cognitive and physical space in fathers' lives. The fourth existential theme of "lived things", depicts how fathers experience their everyday lives in caring for their children with CCN in relation to material aspects of life. The financial implications of caring for a child with a CACCN and the challenges that balancing work with their care demands highlight the additional complexity of father's experiences. The final existential theme of "lived time" concludes how past, present and future experiences of caring for their child impact their current everyday lives. When these five existential themes come together, it concludes that fathers' experiences are constantly evolving, and this is influenced by their children's specific CCN that change over time. These experiences are impacted by their past, present and anticipatory future experiences.

The next chapter summarises the findings, presents a conclusion, implications and recommendations of this study. Additionally, the limitations of the study will be presented.

8 Conclusion, Implications and Recommendations

8.1 Introduction

The previous chapter discussed the key findings related to fathers' experiences of caring for a child with a CACCN and related this to relevant national and international literature, policy and practice. This chapter examines the findings and presents the implications and recommendations from the results. A summary of the findings is presented. The implications highlight the overall significance of the results and offer an insight into how the experiences of fathers caring for children with a CACCN can inform policy, practice and service provision. These implications arise from the key themes that emerged from fathers' experiences that were depicted through their interviews and journals and aim to deepen the understanding of fathers' experiences. Following on from these implications, targeted recommendations are presented. This chapter considers these findings in relation to the implications for policy, practice and service provision, including CDNT, respite care, residential care, schools, hospitals and provides recommendations for healthcare professionals. It also provides recommendations for further research and education, and highlights both the limitations of this study, as well as the unique strengths. Furthermore, a plan for dissemination and a personal reflection of this PhD study is discussed. Finally, an overall conclusion is presented as well as a summary of the included chapters.

8.2 Summary of the findings

This study aimed to explore the experiences of fathers caring for a child with a CACCN. To the best of the author's knowledge, this is the first study specifically exploring this population. This study produces unique findings from the perspectives of nine fathers of children with a range of different CACCN. A total of sixteen qualitative interviews and two journals were analysed using van Manen's five existential themes (lived body, lived relation, lived space, lived things and lived time). Findings were structured around these five existential themes and then are brought together for a deep understanding of the father's experiences.

When their child receives a diagnosis of a CACCN, this is a profoundly complex experience for fathers, encompassing both emotional turmoil and practical challenges. Findings of this study traces their journey from initial concerns to the confirmation of a diagnosis, foregrounding the unique and often underrepresented perspectives of fathers, particularly in the context of rare CAs. Professionals play a critical role in communicating diagnostic information and must also recognise the complexity surrounding the rarity of many CACCN, including the significant gaps in how these conditions are monitored, recorded, and understood. Beyond the emotional weight, a central issue that emerged is the delay in accessing timely and appropriate diagnostic and support services - barriers that significantly impact fathers' capacity to navigate their roles and advocate for their children within the Irish healthcare and disability support systems.

The complex relational landscape fathers must navigate when parenting a child with a CACCN is evident in this study. Fathers strive to balance their emotions and caregiving responsibilities between their child with a CACCN and their other children, seeking to be present and equitable in their roles. They also face the challenge of sustaining their relationship with the mother of their child while managing the demands of caregiving, often needing to shift between being a supportive parent and a committed partner. While grandparents can offer valuable support, fathers also recognise the importance of maintaining boundaries to protect the emotional and practical stability of the family unit.

Fathers note that the transition from hospital to home is impacted by national policies and procedures, particularly within hospital settings, the discharge processes, and access to home care packages which all influence their sense of preparedness and control. Many fathers worry about the future, expressing anxiety around long-term living arrangements for their child, including access to respite services and residential care planning. Fathers face complex decisions regarding their child's care and education, often having to navigate emotionally and logistically challenging choices about medical procedures and school placement.

Structural and financial pressures shape fathers' experiences in caring for a child with a CACCN. Fathers report significant out-of-pocket expenses related to medical equipment, therapies, and homecare supports that place substantial strain on family finances. In addition, their ability to engage fully in caregiving is often constrained by inflexible or limited parental and paternity leave policies, both in Ireland and internationally. These employment related challenges not only restrict fathers' participation in daily care but also tend to at times reinforce traditional gender roles within the family.

Fluctuations in their children's symptoms particularly those with changing life expectancy-results in some fathers carrying a profound emotional toll. Fathers also face increasing behavioural challenges as their children grow, which are compounded by a lack of accessible mental health support. Moreover, caregiving is not confined to daytime hours; it is a continuous, 24-hour responsibility. Sleep deprivation is a common and deeply affecting feature of their experience, as night-time care routines further intensify some of the caregiving strain.

Responses to their roles in caring for a child with a CACCN involves continuous adaptations and adjustments. Yet, the love and joy their children bring to their families and the wider communities outweigh these challenges. Therefore, this research concludes: Fathers' experiences of having a child with a CACCN are constantly evolving as they are influenced by their child's specific CCN that changes over time, and are impacted by their past, present and anticipated future experience.

8.3 Implications and recommendations for policy and service provision

Policy in both government and healthcare structures refers to the governmental guidelines to translate decisions and plans into action to attain the national health goals, reflecting the population's health needs (Department of the Taoiseach, 2025). In the context of this study, the findings intersect with several key areas of healthcare policy, particularly in relation to diagnosing a CACCN, disability and mental health services, parental and paternity leave policies, and the behavioural element of children's care needs. Therefore, engaging critically with healthcare policy is essential for understanding the systemic enablers and barriers fathers of children with a CACCN

encounter, and to consider how current policies may need to evolve to better meet the needs of this population. This section examines the principal implications of the study across the diagnostic pathway; disability and mental health services; respite and residential care; homecare packages; educational settings; hospital care; and the recognition of fathers. Table 14 subsequently presents a structured summary of the key issues identified, corresponding recommendations, and suggested responsible stakeholders.

8.3.1 The diagnostic pathway

This study highlights several implications for healthcare policy and practice related to the diagnostic pathway for children with a CACCN. First, fathers highlight the urgent need for timely, relevant information in relation to their children's diagnosis, and highlight a gap in current health communication. Additionally, identifying dysmorphic features is key to early identification of a CACCN, ensuring all healthcare professionals including public health nurses, GP's and primary care teams are competent in identifying such features.

Secondly, the study suggests the need for clear guidance to help families navigate information sources related to rare diseases, which will become increasingly important with advances in genetic diagnosis. Finally, it emphasises the value of systematically recording the incidence and outcomes of rare conditions to support professional knowledge-sharing and in turn share this information with families of children with a CACCN. While international progress has been made, Ireland's limited contribution to Eurocat due to funding constraints restricts its ability to include all births in Ireland onto this platform.

8.3.2 Disability and mental health services

This study highlights several critical implications for healthcare policy and service provision in relation to disability services and mental health supports. Notably, the lack of appropriate access to disability and mental health support for children with a CACCN is placing significant strain on fathers. Despite the existence of a national policy that

outlines equitable access to services based on need, the findings reveal substantial delays and service gaps, indicating a disconnect between policy and practice.

The absence of a legally mandated timeframe for accessing essential care has led some fathers to pursue private therapeutic interventions (such as occupational therapy, speech and language therapy, physiotherapy) which is placing considerable financial pressure on families already coping with the complexities of raising a child with a CACCN. Fathers also report sourcing medications to aid with sleep privately and utilising private sleep therapists. This is a direct result of challenges with recruiting and retaining staff in the disability services. This underscores the need for stronger regulatory mechanisms to ensure timely, equitable access to publicly funded services.

Moreover, the findings suggest that children with a CACCN often require mental health support yet face barriers in accessing services directly related to their diagnosis. This points to the necessity for improved integration between disability and mental health services, as well as the development of clear, accessible care pathways tailored to the needs of children with rare and complex conditions. This is particularly prominent with regards to the complex behavioural element of their children's conditions.

8.3.3 Respite and residential care needs

The findings of this study indicate that fathers are deeply concerned about both current and future access to respite and residential care for their children, particularly regarding the availability of suitable placements. A key issue raised is the insufficient training of respite and residential care staff in managing the specific conditions and behavioural needs associated with certain CACCN. This highlights a critical gap in service preparedness and suggests that respite care services must be better resourced and trained to meet the complex, condition-specific needs of children with behavioural challenges. Addressing this gap is essential to ensuring that respite care is both accessible and appropriate, reducing stress for families and improving care continuity.

8.3.4 Homecare packages

Homecare packages are a critical support for fathers caring for a child with a CACCN, particularly in providing essential nighttime respite. However, the findings reveal some

limitations that reduce their overall effectiveness. Challenges such as frequent changes in staffing and rostering undermine continuity of care and hinder the development of trust between families and homecare providers. Most notably, the expectation that fathers remain in loco parentis while homecare staff are present prevents them from leaving the home, effectively limiting the autonomy and relief that such support is intended to provide. These findings suggest that the current homecare model that is available in Ireland needs to be reviewed to offer more consistent staffing and to clarify the boundaries of parental responsibility during care hours, ensuring homecare can function as true respite.

8.3.5 Schools

The findings highlight that fathers face significant stress in securing appropriate school placements for their children with a CACCN, despite the entitlement of all children to have a suitable educational setting. The study underscores the value of the National Nursing Pilot for Children with Complex Healthcare Needs, which enabled some children to access mainstream education with appropriate clinical support. This suggests a need for sustained investment in and expansion of such initiatives to ensure equitable access to inclusive education for all children with a CACCN, while also addressing gaps in school readiness to support children with a CACCN.

8.3.6 Hospitals

This study highlights the critical role fathers play during their child's hospitalisation, often spending substantial time in the hospital or taking on increased responsibilities at home. Children with a CACCN face a higher risk of hospital admissions, adding to the burden on families. Despite their involvement, fathers reported feeling that their roles were not fully acknowledged or respected within the acute care setting. These findings underscore the need for hospital systems (particularly with the opening of the new national children's hospital to recognise and accommodate the complex needs of families of children with a CACCN). This includes acknowledging fathers' caregiving roles, addressing family dynamics during prolonged admissions, and ensuring staff are trained to support families holistically.

8.3.7 Recognising fathers when their child with born with a chromosome abnormality and complex care needs

The findings of this study underscore the integral role that fathers play within the caregiving dynamic of the entire family unit. Fathers bring distinct perspectives and may assume different responsibilities as to those of the mothers, contributing uniquely to the caregiving process. However, existing policy frameworks both nationally and internationally may inadvertently limit fathers' capacity to engage in caregiving on an equal footing with mothers. This is particularly evident in the context of paternity and parental leave entitlements, when a child is born with a CACCN and requires additional support in the neonatal period. These practices reflect traditional gender norms and may not adequately support or encourage paternal involvement particularly when their child had a CACCN.

Table 14: Key issues, recommendations, and responsible stakeholders

Domain	Key issues identified	Recommendations	Level of responsibility	Responsible stakeholders
Diagnostic pathway	Delayed diagnosis; inadequate communication around the diagnosis; not being informed that a diagnosis will be taking place; lack of guidance for rare chromosome abnormalities; the importance of recording rare chromosome abnormalities	• Ensure both parents are asked to be present at diagnostic conversations with advance notice • Provide written, evidence-based information at diagnosis • Direct families to accurate syndrome-specific support groups and rare disease communities (e.g. <i>Unique</i>) • Improve guidance on navigating rare disease information sources • Continue and expand funding for rare disease research, including ROI participation in Eurocat • Regular training for healthcare professionals on recognising dysmorphic features	Individual clinician; Service level; National policy / system level	All healthcare professionals involved in diagnostic communication; HSE; Department of Health; Clinical genetics services; Public health nursing services; Professional regulatory bodies (e.g. NMBI/IMC); Health service funders
Disability & Mental Health Services	Service delays; workforce shortages; reliance on private care; limited integration of behavioural and mental health supports	• Develop and implement workforce recruitment and retention strategies for CDNT and CAMHS/CAMHS-ID • Integrate behavioural challenges associated with CACCN into assessment and care pathways • Develop countrywide CDNT-led individualised sleep programmes in collaboration with GPs/paediatricians • Conduct regular assessments of fathers' psychosocial support needs, including anticipatory grief • Develop grandparent education programmes • Provide fathers with education on consent, puberty, and interpersonal relationships	Service level; Regional (HSE); National workforce planning	HSE disability services; CDNTs; CAMHS/CAMHS-ID; Department of Health; Professional training bodies; Primary care services
Respite & Residential Care	Insufficient respite availability; lack of condition-specific	• Increase provision of timely and appropriate respite services	Service level; Regional planning;	HSE disability services; Disability service

	staff training; uncertainty around future residential placements	• Regularly review respite needs as children age and family circumstances change • Develop residential care options with staff trained in behavioural, mental health, and dietary needs of children with CACCN	National commissioning	providers; Department of Health; HIQA; Workforce education providers; Private residential care services
Homecare Packages	Staffing instability; lack of continuity; in loco parentis rule limiting meaningful respite	• Review the appropriateness of the in loco parentis rule in homecare settings • Improve staff rostering and continuity of care practices within homecare services	National policy level; Service delivery level	HSE homecare services; Department of health; Private homecare providers that are on the national tender; Regulatory bodies
Schools	Difficulty securing appropriate placements; limited staff knowledge of CACCN; challenges managing medical needs in schools	• Develop sufficient appropriate school placements for children with CACCN • Ensure flexible, inclusive educational planning • Provide education and training for school staff on CACCN-specific needs, including dietary management	School level; Regional (NCSE); National education policy	Department of Education; National Council for Special Education (NCSE); School management; HSE
Hospitals	Fathers' caregiving roles under-recognised; lack of coordinated care pathways; burden of repeated admissions	• Implement policies recognising fathers as key caregivers during hospitalisation • Provide staff training on inclusive family-centred communication • Develop integrated multidisciplinary care pathways, including complex care coordination	Hospital/service level; National clinical programmes	Hospital management; HSE acute services; Clinical Nurse Specialists; Multidisciplinary teams; National children's hospital governance
Recognising fathers	Limited paternity leave entitlements; gendered caregiving norms; insufficient recognition of fathers' roles	• Advocate for extended paternity leave in cases of prolonged neonatal admission • Formally recognise fathers' 24-hour caregiving roles, including nighttime care	National policy level; EU policy level	Department of Health; Department of social protection; EU policy stakeholders; Healthcare professionals

8.4 Recommendations for future research

The findings from these fathers' depictions of their experiences of caring for a child with a CACCN, coupled with the international literature, highlight gaps for further research. Identifying and highlighting gaps in research related identify to fathers of children with CCN is essential to help healthcare professionals improve health care practices and support strategies for these fathers. Below are some identified areas for further research related to the findings of this study:

- i. Fathers identified some areas of coping mechanisms that differs from those of the mother of the child with a CACCN. This suggests a need for evaluation of gender specific coping mechanisms.
- ii. Grandparents are a significant part of the family unit, and their support is often vital to help care for children with CACCN. Yet, there is little research on their experiences, therefore, further research is warranted with this cohort to help tailor supports and education pathways.
- iii. Fathers in this study noted delays in being discharged from hospital during acute admissions. Further exploration and auditing the role of the complex discharge teams in the Irish context is warranted, this includes auditing of the P-CAT assessment tool with an aim of improving delayed discharges.
- iv. Audits are warranted on staffing levels of nurses caring for children with a CACCN in the community, particularly for private homecare providers.
- v. Fathers' experiences of being involved in advanced care planning is significantly less reported than that of the mother and needs further exploration to ensure their voices are also included.

8.5 Limitations of the research

Findings of this study provide a rich description of the experiences of fathers caring for a child with a CACCN. However, Polit & Beck (2020), highlight that all research is subject to limitations and highlight the importance of identifying such limitations. There is an obligation for all researchers to depict an honest presentation of their research limitations (Ross, 2019). The limitations of this study are identified below, and the results of this study should be understood considering these limitations.

- i. One of the primary limitations of this study pertains to the recruitment process. Purposeful sampling was employed, which, while aligning with the study's aims and objectives and proving effective in recruiting fathers of children with a CACCN, does introduce certain limitations. A notable concern is related to the recruitment routes used. Not all children with a CACCN are receiving HCP, which may have resulted in a bias towards fathers with in-home nursing support. To mitigate this potential bias, a secondary recruitment route through support groups was also utilised. While this approach identified additional participants, the majority of the support groups were syndrome specific. Consequently, more prevalent conditions received greater exposure during the recruitment process, potentially overshadowing rarer conditions. In an effort to include fathers of children with less common CAs, support groups focused on rare conditions were also approached. Nevertheless, it is important to acknowledge that some fathers may not have access to social media or may not be inclined to attend support groups, which could have limited their opportunity to participate in the study.
- ii. Fathers self-volunteered to be included in this study which could present some potential bias. Fathers who were part of support group associations may have been more inclined to want to be included in the study, which could result in some self-selection bias.
- iii. The impact that Covid-19 had on families of children with CACCN was addressed in this study, and this cannot be understated as a potential limitation. As explained in section 5.7, the option for Zoom interviews were completed despite national restrictions being lifted. Eight out of nine fathers opted to have their interview over Zoom, which could have potentially changed the level of rapport, due to potential reduced nonverbal communication such as eye contact and facial expressions. However, some of this was alleviated by ensuring extra time was taken to allow for an adjustment period. Furthermore, it must be noted that some technical difficulties were evident. One father was interviewed during a storm and his internet connection dropped which momentarily interrupted the flow of the interview.

8.6 Unique Strengths of this Study

It is equally as important to identify the unique strengths of a study, as well as presenting the limitations (Ross et al., 2019). This is imperative to identify the original contributions this study presents to the nursing field, which outweigh the limitations that were previously discussed.

- i. To the authors' knowledge, this is the first study to specifically examine fathers' experiences of caring for a child with both a CA and CCN, presenting it as a unique contribution to the field. While CA can result in multidomain disabilities, the holistic experience of fathers in this context has not been comprehensively captured in existing literature. Research relating to fathers of children with CCN has typically focused either on broad diagnostic categories (i.e. CCN) or on specific medical interventions, such as tracheostomy care. When CAs have been explored, the experiences of fathers caring for children with Trisomy 21 have dominated, often overshadowing other conditions.
- ii. Within the context of CACCN, maternal perspectives have historically dominated the literature. This study addresses this imbalance by foregrounding the voices of fathers presenting a rich description of their experiences. It represents the first research in Ireland specifically dedicated to exploring fathers' experiences of caring for a child with a CACCN. This is particularly important for healthcare professionals to recognise the importance of father's roles in caring for children with CACCN.
- iii. The use of Zoom to complete the interviews was also a considerable strength to this study. It allowed a convenient method of interviewing for fathers who had a very busy schedule. Furthermore, given the risk that Covid-19 could present to children with CACCN, it allowed fathers who wanted to continue to reduce their social contact the option to participate. It afforded fathers the opportunity to choose a place where they were most comfortable to openly talk.
- iv. This study presents policy and practice recommendations which are especially important to inform policy makers in the imminent opening of the new national children's hospital.

8.7 Dissemination

Dissemination in healthcare is the process of distributing information to specific clinical audiences or relevant public health sectors (The United States Agency for Healthcare Research and Quality, 2022; Ravinetto & Singh, 2023). This study has evidenced the lived experiences of fathers caring for a child with a CACCN and it is important that these findings are accessible to a wide audience of both healthcare professionals caring for these children, policy makers and voluntary organisations, as these findings can have a positive influence, but only if the results reach a broad audience (Capili et al., 2024). Methods of dissemination vary to include publications in academic journals, presentations at conferences or with relevant stakeholders (Brownson et al., 2018; Capili et al., 2024). There is an ethical need to disseminate these results as fathers’ experiences in this study are vital to the contribution of knowledge.

Dissemination of this study is already underway through engagement with international academic audiences. The scoping review was first presented as a poster at “THE Conf, Trinity College,” held at Trinity College Dublin on 6–7 March 2024, where it was selected as a winning presentation. Subsequently, key findings from the study were shared in an oral presentation at “The Conf, Trinity College, 5/6 March 2025”, further contributing to scholarly dialogue in this field. An abstract has also been submitted to present the findings at the European Academy of Childhood-onset Disability international conference and has been accepted for discussion. Plans for dissemination also include submission of the findings to fathers and to syndrome specific groups and homecare providers. Table 15 provides the references to peer reviewed presentations.

Table 15: Peer reviewed presentations

Peer presentations
• Mahon, I., Doyle, C., Hughes, M.,, Fathers experiences of caring for a child with complex care needs and a chromosome abnormality: a scoping review, THE Conf, Trinity College, 6/7 March 2024, Poster. • Mahon, I., Doyle, C., Hughes, M.,, Fathers experiences of caring for a child with a chromosome abnormality and complex care needs., <i>THE Conf, Trinity College, 5/6 March 2025, Dublin, 5/6 March</i>, 2025 Conference Paper.

8.8 Personal reflection

Personal reflection has been a vital element in developing this PhD study; in keeping with van Manen's hermeneutic phenomenology, reflection is paramount in keeping "hermeneutic awareness" (van Manen 2007, p69). As a healthcare professional working in the paediatric acute setting, instead of bracketing my knowledge, my prior understanding was acknowledged throughout this entire research process to reduce potential bias. Personal reflection was completed regularly using a journal. Figure 7. displays some examples of how my journal entries developed and changed over the years and proved vital in the interpretation and analysis of the father's interviews. This was vital in ensuring that I was not placing my own interpretations of previous understandings onto the participant's experiences. Journaling also proved as a central element to gaining confidence as a researcher and not a clinical nurse. In the beginning of this research journey, I was worried that my own experiences as a nurse would influence my interview techniques, however when I listened back to the interview transcripts, by documenting my own feelings prior to the interviews and after I had listened to the transcripts, I was able to prove to myself that I am able to be a good researcher which allowed me to feel more confident as the interviews progressed. On reflection, this may have been a benefit of my previous knowledge, as I have developed good communication skills over the years working with fathers of children with a CACCN.

Aside from my own personal reflections, it is also important to reflect on the methodology used in completing this PhD. Firstly, using van Manen's hermeneutic phenomenology allowed me to give voice to fathers caring for a child with a CA to which previous research was limited. The semi-structured interviews being completed over Zoom was extremely beneficial in allowing fathers who may previously not have taken part as they may have wanted to reduce social distancing as a result of Covid 19. Furthermore, aside from when the internet connection dropped during one interview, I did not feel there was any disadvantage and rapport was established quickly.

Using the five existential themes and developing an understanding of “the art of writing and rewriting”, guided by van Manen was extremely helpful in aiding me develop a systematic approach to understanding the lived experience of these fathers. As shown in my journal entries in Table 15, when commencing data analysis, I found the volume of data difficult to comprehend, but by repeatedly going back to van Manen’s hermeneutic phenomenology approach enabled me to overcome this. Additionally, I have a diagnosis of dyslexia, and I found that by transcribing the transcripts myself I was able to fully hear the experiences as I was writing instead of only seeing them in written form.

Table 16: Example of personal reflection from researcher's journal

Self-reflections
Year 1 reflections (2020-2021) • "First year of this PHD journey...six years seems like a lifetime away..." • "Researching the background is difficult...there is a lot of medical research on chromosome abnormalities, lots of case studies".
Year 2 reflections (2021-2022) • "This scoping review is much harder than I anticipated." • "Ethical approval has finally been granted...woohoo...now for the exciting part..." • "Disappointed that one organisation felt fathers are saturated with research so have turned down being a gatekeeper..."
Year 3 reflections (2022-2023) • "First interview...here goes...What happens if there is an awkward silence..." • "Transcribing my first transcript... Funny that I thought there would be an awkward silence, I hardly ask any questions..." • "I found that interview personally hard to listen to, particularly how that father was treated by a professional...I had to be a researcher and not a nurse...what did I respond to him..." • "(listening to the transcript)...I was sympathetic and asked appropriate follow on questions..."
Year 4 reflections (2023-2024) • "That interview was interesting...I heard the same story as the last interview...I wonder if this means I'm reaching saturation..." • "Wifi connection went...why do I feel so panicked about this?" • "Final interview...how will I manage to put all this data into one chapter...these fathers have so much to say?"
Year 5 reflections (2024-2025) • "Am I doing justice to these fathers...I feel I could write a book on each interview...?" • "The art of writing and rewriting...still finding meaning after all this time..." • "Am I doing these fathers justice?" • "I finally think this study gives meaning to fathers experiences of caring for a child with CCN...I'm so thankful to the fathers that took part" • "first time presenting this data at a conference...I was nervous, but could also see that people were interested in hearing the experiences of fathers..."

8.9 Conclusion

This final chapter provided a comprehensive summary of the key findings and examined their implications and associated recommendations, with particular attention given to critical areas including the diagnostic pathway, disability and mental health supports, respite services, residential and HCP, as well as interactions with schools and hospitals. The chapter also underscored the importance of recognising the roles and experiences of fathers, which was previously an under searched aspect in caregiving research. Furthermore, it addressed the study's limitations and highlighted its unique methodological and conceptual contributions, concluding with proposed strategies for the dissemination of findings.

This PhD thesis explored the lived experiences of nine fathers of children with a CACCN, addressing a significant gap in the existing literature where paternal perspectives have been largely overlooked. Employing a hermeneutic phenomenological methodology informed by van Manen's approach, the study presents fathers' narratives elicited through in-depth interviews and reflective journal entries through the lens of existential themes. This approach enabled a nuanced interpretation of how fathers make meaning of their caregiving roles within the context of raising a child with a CACCN.

In conclusion, when a child receives a diagnosis of a CA it is an emotionally complex journey for fathers, yet the love for their children brings immense joy to these fathers, their families and the wider community. However, fathers note there are significant delays in both diagnostic and support services impacting on both their own wellbeing and that of their children. The evolving nature of their children's CACCN often results in shifting support needs for fathers over time. Notably, as the medical aspects of care may lessen in intensity, behavioural and psychosocial demands frequently increase, underscoring the multifaceted and dynamic nature of CACCN and the corresponding need for responsive, ongoing support for fathers in their caregiving roles. The term CCN is often synonymous with medical complexities, yet this study highlights that complexities are often vast, that stretch across both health, social and educational landscapes. Findings of this study show that Fathers' experiences of having a child with a CACCN, are constantly evolving as they are influenced by their child's specific CACCN

which change over time, and are impacted by their past, present and anticipatory future experience. Stakeholders need to recognise the complex nature of CAs, and service provision needs to incorporate all elements of children's support needs to support fathers caring for children with a CACCN.

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Appendix 1: Key search term derivatives and search string

(A) Search term derivatives

Concept 1: Fathers Experiences of complex care needs

Medline: (MH "Chromosome Disorders+") OR (MH "Sex Chromosome Disorders+") OR (MH "Chromosome Aberrations+") OR (MH "Sex Chromosome Aberrations+") OR (MH "Patient Care Team+") OR (MH "Chromosomes") OR (MH "Sex Chromosomes+") OR (MH "Ring Chromosomes") OR (MH "Chromosomes, Human+") OR (MH "Mentally Disabled Persons") OR (MH "Intellectual Disability+") OR (MH "Learning Disorders+") OR (MH "Mental Retardation, X-Linked+") OR (MH "Developmental Disabilities")

CINAHL: (MH "Developmental Disabilities") OR (MH "Intellectual Disability") OR (MH "Mentally Disabled Persons") OR (MH "Mental Retardation, X-Linked+") OR (MH "Chromosome Disorders+") OR (MH "Chromosome Aberrations+") OR (MH "Sex Chromosome Disorders") OR (MH "Sex Chromosome Aberrations") OR (MH "Chromosomes+") OR (MH "Sex Chromosomes+")

EMBASE: 'genetic disorder'/exp OR 'Down syndrome'/exp OR 'Williams Beuren syndrome'/exp OR 'mentally disabled person'/exp OR 'mental deficiency'/exp OR 'intellectual impairment'/exp OR 'cognitive defect'/exp OR 'chromosome disorder'/exp OR 'non-hereditary chromosome disorders'/exp OR 'multidisciplinary team'/exp

PsycINFO: DE "Intellectual Development Disorder" OR DE "Anencephaly" OR DE "Down's Syndrome" OR DE "Tay Sachs Disease" OR DE "Autism Spectrum Disorders" OR DE "Interdisciplinary Treatment Approach" OR DE "Chromosome Disorders" OR DE "Autosome Disorders" OR DE "Deletion (Chromosome)" OR DE "Sex Chromosome Disorders" OR DE "Translocation (Chromosome)" OR DE "Trisomy" OR DE "Williams Syndrome" OR DE "Deletion (Chromosome)" OR DE "Sex Chromosome Disorders" OR DE "Klinefelters Syndrome" OR DE "Chromosomes" OR DE "Autosomes" OR DE "Sex Chromosomes" OR DE "Telomeres" OR DE "Mutations" OR DE "Genetics" OR DE "Behavioral Genetics" OR DE "Epigenetics" OR DE "Eugenics" OR DE "Genetic Engineering" OR DE "Genomics" OR DE "Optogenetics" OR DE "Pharmacogenetics" OR DE "Population Genetics" OR DE "Fragile X Syndrome" OR DE "Sex Chromosomes" OR DE "Genetic Linkage"

Keywords on title and abstract: XB (chromosom* OR karyotyp* OR Trisom* OR syndrom* OR downs OR william* OR prader-willi OR "prader willi" OR "Intellectual Disabil*" OR "Intellectually disabled" OR "mentally disabled" OR "mental disabil*" OR "Intellectual Development Disorder*" OR "mental handicap*" OR "mentally handicapped" OR "mentally impaired" OR "mental impairment*" OR "intellectual impairment*" OR "mental defici*" OR "intellectual retard*" OR "mental retard*" OR "mentally retarded" OR "intellectually challenged" OR "intellectually deficient*" OR "intellectually handicapped" OR "intellectually impaired" OR "intellectually retarded" OR "mentally challenged" OR "mentally defici*" OR "cognitive disab*") OR

((Complex* OR critical* OR chronic* OR multifacet* OR intradisciplin* OR interdisciplin* OR "multi facet*" OR multiple* OR inter-team OR "inter team*" OR interprofess* OR integrate* OR life-limit* OR "life limit*" OR EOL OR end-of life* OR terminal* OR "life sustain*") N4 (need* OR care OR nurs* OR intervent* OR medic* OR caring OR assist*))

EMBASE: 'intellectual disabil*':ab,ti OR 'intellectually disabled':ab,ti OR 'mentally disabled':ab,ti OR 'mental disabil*':ab,ti OR 'intellectual development disorder*':ab,ti OR 'mental handicap*':ab,ti OR 'mentally handicapped':ab,ti OR 'mentally impaired':ab,ti OR 'mental impairment*':ab,ti OR 'intellectual impairment*':ab,ti OR 'mental defici*':ab,ti OR 'intellectual retard*':ab,ti OR 'mental retard*':ab,ti OR 'mentally retarded':ab,ti OR 'intellectually challenged':ab,ti OR 'intellectually deficient*':ab,ti OR 'intellectually handicapped':ab,ti OR 'intellectually impaired':ab,ti OR 'intellectually retarded':ab,ti OR 'mentally challenged':ab,ti OR 'mentally defici*':ab,ti OR 'cognitive disab*':ab,ti

OR ((Complex* OR critical* OR chronic* OR multifacet* OR intradisciplin* OR interdisciplin* OR "multi facet*" OR multiple* OR inter-team OR "inter team*" OR interprofess* OR integrate* OR life-limit* OR "life limit*" OR EOL OR end-of life* OR terminal* OR "life sustain*") NEAR/4 (need* OR care OR nurs* OR intervent* OR medic* OR caring OR assist*))

Concept 2: Fathers experience & Qualitative Research

Medline; ((MH "Parents") OR (MH "Fathers+") OR (MH "Adolescent Fathers")) AND ((MH "Qualitative Research") OR (MH "Hermeneutics") OR (MH "Grounded Theory"))

CINAHL ((MH "Fathers+") OR (MH "Parents") OR (MH "Adolescent Fathers") OR (MH "Parents of Children with Disabilities")) AND ((MH "Thematic Analysis") OR (MH "Meta Synthesis") OR (MH "Content Analysis") OR (MH "Qualitative Studies+") OR (MH "Phenomenology") OR (MH "Grounded Theory") OR (MH "Focus Groups") OR (MH "Interviews+") OR (MH "Narratives") OR (MH "Observational Methods") OR (MH "Naturalistic Inquiry"))

EMBASE (('father'/exp OR 'parent'/exp OR 'father child relation'/exp) AND ('action research'/exp OR 'comparative study'/exp OR 'observational study'/exp OR 'grounded theory'/exp OR 'narratives'/exp OR 'phenomenology'/exp OR 'focus group'/exp OR 'interview'/exp OR 'descriptive research'/exp))

PsycINFO (DE "Fathers" OR DE "Adolescent Fathers" OR DE "Single Fathers" OR DE "Expectant Fathers") AND (DE "Qualitative Research" OR DE "Grounded Theory" OR DE "Content Analysis" OR DE "Interviews" OR DE "Phenomenology" OR DE "Case Report" OR DE "Narratives" OR DE "Cohort Analysis")

Keywords on title and abstract:

XB (father* OR dad OR paternal* OR "male parent*" OR paternity* OR homoparent*) N3 (experien* or percept* or attitud* or view* or feel* or perspectiv* OR qualitativ* OR hermeneutic* OR "action research*" OR "empirical research*" OR ethnograph* OR phenomenolog* OR "grounded theor*" OR "grounded research*" OR "feminist research*" OR "explanatory research*" OR "exploratory research" OR "participatory research*" OR interview* OR "One-to-one" OR "interpretive research*" OR "emancipatory research*" OR "participant observation*" OR "observational stud*" OR "observational research*" OR "comparative stud*" OR "comparative research*" OR "narrative research*" OR "narrative stud*" OR "narrative analys*" OR "descriptive stud*" OR "descriptive research*" OR "exploratory stud*" OR "literature review" OR "interpretative analys*" OR "case stud*" OR "case report*" OR survey* OR "group*")

OR "content analys*" OR "thematic analys*" OR "thematic cod*" OR "observation* method*" OR "naturalistic*" OR "group discussion*" OR "Ethnonursing Research*" OR "participant stud*")

EMBASE: ((father* OR dad OR paternal* OR 'male parent*' OR paternity* OR homoparent*) NEAR/3 (experien* OR percept* OR attitud* OR view* OR feel* OR perspectiv* OR qualitativ* OR hermeneutic* OR 'action research*' OR 'empirical research*' OR ethnograph* OR phenomenolog* OR 'grounded theor*' OR 'grounded research*' OR 'feminist research*' OR 'explanatory research*' OR 'exploratory research' OR 'participatory research*' OR interview* OR 'one-to-one' OR 'interpretive research*' OR 'emancipatory research*' OR 'participant observation*' OR 'observational stud*' OR 'observational research*' OR 'comparative stud*' OR 'comparative research*' OR 'narrative research*' OR 'narrative stud*' OR 'narrative analys*' OR 'descriptive stud*' OR 'descriptive research*' OR 'exploratory stud*' OR 'literature review' OR 'interpretative analys*' OR 'case stud*' OR 'case report*' OR survey* OR 'group*' OR 'content analys*' OR 'thematic analys*' OR 'thematic cod*' OR 'observation* method*' OR 'naturalistic*' OR 'group discussion*' OR 'ethnonursing research*' OR 'participant stud*')):ab,ti

(B) Search String

Medline	CINAHL ULTIMATE	EMBASE	PSYCINFO
1365	2278	2971	624

#	Query	Limiters/Expanders	Last Run Via	Results
S9	S3 AND S8	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - MEDLINE	1,365
S8	S4 OR S5 OR S6 OR S7	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - MEDLINE	2,669,976

S7	XB ((Complex* OR critical* OR chronic* OR multifacet* OR intradisciplin* OR interdisciplin* OR "multi facet*" OR multiple* OR inter-team OR "inter team*" OR interprofess* OR integrate* OR life-limit* OR "life limit*" OR EOL OR end-of life* OR terminal* OR "life sustain*")) N4 (need* OR care OR nurs* OR intervent* OR medic* OR caring OR assist*))	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - MEDLINE	364,656
S6	(MH "Chromosome Disorders+") OR (MH "Sex Chromosome Disorders+") OR (MH "Chromosome Aberrations+") OR (MH "Sex Chromosome Aberrations+") OR (MH "Patient Care Team+")	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - MEDLINE	291,969

S5	(MH "Chromosomes") OR (MH "Sex Chromosomes+") OR (MH "Ring Chromosomes") OR (MH "Chromosomes, Human+") OR (MH "Mentally Disabled Persons") OR (MH "Intellectual Disability+") OR (MH "Learning Disorders+") OR (MH "Mental Retardation, X-Linked+") OR (MH "Developmental Disabilities")	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - MEDLINE	298,493
S4	XB (chromosom* OR karyotyp* OR Trisom* OR syndrom* OR downs OR william* OR prader-willi OR "prader willi" OR "Intellectual Disabil*" OR "Intellectually disabled" OR "mentally disabled" OR "mental disabil*" OR "Intellectual Development Disorder*" OR "mental	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - MEDLINE	2,126,928

	handicap*" OR "mentally handicapped" OR "mentally impaired" OR "mental impairment*" OR "intellectual impairment*" OR "mental defici*" OR "intellectual retard*" OR "mental retard*" OR "mentally retarded" OR "intellectually challenged" OR "intellectually deficient*" OR "intellectually handicapped" OR "intellectually impaired" OR "intellectually retarded" OR "mentally challenged" OR "mentally defici*" OR "cognitive disab*")			
S3	S1 OR S2	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - MEDLINE	10,572

S2	XB (father* OR dad OR paternal* OR "male parent*" OR paternity* OR homoparent*) N3 (experien* or percept* or attitud* or view* or feel* or perspectiv* OR qualitativ* OR hermeneutic* OR "action research*" OR "empirical research*" OR ethnograph* OR phenomenolog* OR "grounded theor*" OR "grounded research*" OR "feminist research*" OR "explanatory research*" OR "exploratory research" OR "participatory research*" OR interview* OR "One-to-one" OR "interpretive research*" OR "emancipatory research*" OR "participant observation*" OR "observational stud*" OR	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - MEDLINE	5,788

“observational research*” OR “comparative stud*” OR “comparative research*” OR “narrative research*” OR “narrative stud*” OR “narrative analys*” OR “descriptive stud*” OR “descriptive research*” OR “exploratory stud*” OR “literature review” OR “interpretative analys*” OR “case stud*” OR “case report*” OR survey* OR “group*” OR “content analys*” OR “thematic analys*” OR “thematic cod*” OR “observation* method*” OR “naturalistic*” OR “group discussion*” OR “Ethnonursing Research*” OR “participant stud*”)			

S1	((MH "Parents") OR (MH "Fathers+") OR (MH "Adolescent Fathers")) AND ((MH "Qualitative Research") OR (MH "Hermeneutics") OR (MH "Grounded Theory"))	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - MEDLINE	5,192

#	Query	Limiters/Expanders	Last Run Via	Results
S9	S3 AND S8	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - CINAHL Ultimate	2,278
S8	S4 OR S5 OR S6 OR S7	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases	593,209

			Search Screen - Advanced Search Database - CINAHL Ultimate	
S7	XB ((Complex* OR critical* OR chronic* OR multifacet* OR intradisciplin* OR interdisciplin* OR "multi facet*" OR multiple* OR inter-team OR "inter team*" OR interprofess* OR integrate* OR life-limit* OR "life limit*" OR EOL OR end-of life* OR terminal* OR "life sustain*") N4 (need* OR care OR nurs* OR intervent* OR medic* OR caring OR assist*))	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - CINAHL Ultimate	185,767
S6	(MH "Chromosome Disorders+") OR (MH "Chromosome Aberrations+") OR (MH "Sex Chromosome Disorders") OR (MH "Sex Chromosome Aberrations") OR (MH "Chromosomes+") OR (MH "Sex Chromosomes+") OR (MH	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - CINAHL Ultimate	28,573

	"Chromosome Disorders+") OR (MH "Chromosome Aberrations+") OR (MH "Sex Chromosome Disorders") OR (MH "Sex Chromosome Aberrations") OR (MH "Chromosomes+") OR (MH "Sex Chromosomes+")			
S5	(MH "Developmental Disabilities") OR (MH "Intellectual Disability") OR (MH "Mentally Disabled Persons") OR (MH "Mental Retardation, X-Linked+") OR (MH "Multidisciplinary Care Team+") OR (MH "Team Nursing")	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - CINAHL Ultimate	93,495
S4	XB (chromosom* OR karyotyp* OR Trisom* OR syndrom* OR downs OR william* OR prader-willi OR "prader willi" OR "Intellectual Disabil*" OR "Intellectually disabled" OR "mentally disabled" OR "mental	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - CINAHL Ultimate	337,219

	disabil*" OR "Intellectual Development Disorder*" OR "mental handicap*" OR "mentally handicapped" OR "mentally impaired" OR "mental impairment*" OR "intellectual impairment*" OR "mental defici*" OR "intellectual retard*" OR "mental retard*" OR "mentally retarded" OR "intellectually challenged" OR "intellectually deficient*" OR "intellectually handicapped" OR "intellectually impaired" OR "intellectually retarded" OR "mentally challenged" OR "mentally defici*" OR "cognitive disab*")			
S3	S1 OR S2	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - CINAHL Ultimate	18,147

S2	XB (father* OR dad OR paternal* OR "male parent*" OR paternity* OR homoparent*) N3 (experien* or percept* or attitud* or view* or feel* or perspectiv* OR qualitativ* OR hermeneutic* OR "action research*" OR "empirical research*" OR ethnograph* OR phenomenolog* OR "grounded theor*" OR "grounded research*" OR "feminist research*" OR "explanatory research*" OR "exploratory research" OR "participatory research*" OR interview* OR "One- to-one" OR "interpretive research*" OR "emancipatory research*" OR "participant observation*" OR "observational stud*" OR "observational research*" OR "comparative stud*" OR "comparative research*" OR "narrative research*" OR "narrative	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - CINAHL Ultimate	3,821

	stud*" OR "narrative analys*" OR "descriptive stud*" OR "descriptive research*" OR "exploratory stud*" OR "literature review" OR "interpretative analys*" OR "case stud*" OR "case report*" OR survey* OR "group*" OR "content analys*" OR "thematic analys*" OR "thematic cod*" OR "observation* method*" OR "naturalistic*" OR "group discussion*" OR "Ethnonursing Research*" OR "participant stud*")			
S1	((MH "Fathers+") OR (MH "Parents") OR (MH "Adolescent Fathers") OR (MH "Parents of Children with Disabilities")) AND ((MH "Thematic Analysis") OR (MH "Meta Synthesis") OR (MH "Content Analysis") OR (MH "Qualitative Studies+") OR (MH "Phenomenology") OR (MH "Grounded Theory") OR (MH "Focus	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - CINAHL Ultimate	15,413

	Groups") OR (MH "Interviews+") OR (MH "Narratives") OR (MH "Observational Methods") OR (MH "Naturalistic Inquiry"))			

#	Query	Limiters/Expanders	Last Run Via	Results
S10	S3 AND S8	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - APA PsycInfo	624
S9	S3 AND S8	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - APA PsycInfo	624
S8	S4 OR S5 OR S6 OR S7	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - APA PsycInfo	473,426

S7	XB ((Complex* OR critical* OR chronic* OR multifacet* OR intradisciplin* OR interdisciplin* OR "multi facet*" OR multiple* OR inter-team OR "inter team*" OR interprofess* OR integrate* OR life-limit* OR "life limit*" OR EOL OR end-of life* OR terminal* OR "life sustain*") N4 (need* OR care OR nurs* OR intervent* OR medic* OR caring OR assist*))	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - APA PsycInfo	96,057
S6	DE "Chromosome Disorders" OR DE "Autosome Disorders" OR DE "Deletion (Chromosome)" OR DE "Sex Chromosome Disorders" OR DE "Translocation (Chromosome)" OR DE "Trisomy" OR DE "Williams Syndrome" OR DE	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - APA PsycInfo	82,884

	"Deletion (Chromosome)" OR DE "Sex Chromosome Disorders" OR DE "Klinefelters Syndrome" OR DE "Chromosomes" OR DE "Autosomes" OR DE "Sex Chromosomes" OR DE "Telomeres" OR DE "Mutations" OR DE "Genetics" OR DE "Behavioral Genetics" OR DE "Epigenetics" OR DE "Eugenics" OR DE "Genetic Engineering" OR DE "Genomics" OR DE "Optogenetics" OR DE "Pharmacogenetics" OR DE "Population Genetics" OR DE "Fragile X Syndrome" OR DE "Sex Chromosomes" OR DE "Genetic Linkage"			

S5	DE "Intellectual Development Disorder" OR DE "Anencephaly" OR DE "Down's Syndrome" OR DE "Tay Sachs Disease" OR DE "Autism Spectrum Disorders" OR DE "Interdisciplinary Treatment Approach"	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - APA PsycInfo	118,611
S4	XB (chromosom* OR karyotyp* OR Trisom* OR syndrom* OR downs OR william* OR prader-willi OR "prader willi" OR "Intellectual Disabil*" OR "Intellectually disabled" OR "mentally disabled" OR "mental disabil*" OR "Intellectual Development Disorder*" OR "mental handicap*" OR "mentally handicapped" OR "mentally impaired" OR "mental	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - APA PsycInfo	252,508

	impairment*" OR "intellectual impairment*" OR "mental defici*" OR "intellectual retard*" OR "mental retard*" OR "mentally retarded" OR "intellectually challenged" OR "intellectually deficient*" OR "intellectually handicapped" OR "intellectually impaired" OR "intellectually retarded" OR "mentally challenged" OR "mentally defici*" OR "cognitive disab*")			
S3	S1 OR S2	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - APA PsycInfo	8,891
S2	XB (father* OR dad OR paternal* OR "male parent*" OR paternity* OR	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases	8,648

	homoparent*) N3 (experien* or percept* or attitud* or view* or feel* or perspectiv* OR qualitativ* OR hermeneutic* OR "action research*" OR "empirical research*" OR ethnograph* OR phenomenolog* OR "grounded theor*" OR "grounded research*" OR "feminist research*" OR "explanatory research*" OR "exploratory research" OR "participatory research*" OR interview* OR "One-to-one" OR "interpretive research*" OR "emancipatory research*" OR "participant observation*" OR "observational stud*" OR "observational research*" OR "comparative stud*" OR "comparative research*" OR		Search Screen - Advanced Search Database - APA PsycInfo	

	"narrative research*" OR "narrative stud*" OR "narrative analys*" OR "descriptive stud*" OR "descriptive research*" OR "exploratory stud*" OR "literature review" OR "interpretative analys*" OR "case stud*" OR "case report*" OR survey* OR "group*" OR "content analys*" OR "thematic analys*" OR "thematic cod*" OR "observation* method*" OR "naturalistic*" OR "group discussion*" OR "Ethnonursing Research*" OR "participant stud*")			
S1	PsycINFO (DE "Fathers" OR DE "Adolescent Fathers" OR DE "Single Fathers" OR DE "Expectant Fathers") AND (DE	Expanders - Apply equivalent subjects Search modes - Proximity	Interface - EBSCOhost Research Databases Search Screen - Advanced Search Database - APA PsycInfo	483

	"Qualitative Research" OR DE "Grounded Theory" OR DE "Content Analysis" OR DE "Interviews" OR DE "Phenomenology" OR DE "Case Report" OR DE "Narratives" OR DE "Cohort Analysis")			

☐ History	Save Delete Print view Export Email	Combine >	using ☒ And ☐ Or	Collapse
☐ #9	#8 AND [embase]/lim NOT ([embase]/lim AND [medline]/lim)			2,971
☐ #8	#4 AND #7			6,309
☐ #7	#5 OR #6			40,917
☐ #6	((father* OR dad OR paternal* OR 'male parent*' OR paternity* OR homoparent*) NEAR/3 (experien* OR percept* OR attitud* OR view* OR feel* OR perspectiv* OR qualitativ* OR hermeneutic* OR 'action research*' OR 'empirical research*' OR ethnograph* OR phenomenolog* OR 'grounded theor*' OR 'grounded research*' OR 'feminist research*' OR 'explanatory research*' OR 'exploratory research*' OR 'participatory research*' OR interview* OR 'one-to-one' OR 'interpretive research*' OR 'emancipatory research*' OR 'participant observation*' OR 'observational stud*' OR 'observational research*' OR 'comparative stud*' OR 'comparative research*' OR 'narrative research*' OR 'narrative stud*' OR 'narrative analys*' OR 'descriptive stud*' OR 'descriptive research*' OR 'exploratory stud*' OR 'literature review' OR 'interpretative analys*' OR 'case stud*' OR 'case report*' OR survey* OR 'group*' OR 'content analys*' OR 'thematic analys*' OR 'thematic cod*' OR 'observation* method*' OR 'naturalistic*' OR 'group discussion*' OR 'ethnonursing research*' OR 'participant stud*')):ab,ti			5,531
☐ #5	('father'/exp OR 'parent'/exp OR 'father child relation'/exp) AND ('action research'/exp OR 'comparative study'/exp OR 'observational study'/exp OR 'grounded theory'/exp OR 'narratives'/exp OR 'phenomenology'/exp OR 'focus group'/exp OR 'interview'/exp OR 'descriptive research'/exp)			36,106
☐ #4	#1 OR #2 OR #3			4,706,941
☐ #3	((complex* OR critical* OR chronic* OR multifacet* OR intradisciplin* OR interdisciplin* OR 'multi facet*' OR multiple* OR 'inter team' OR 'inter team*' OR interprofess* OR integrate* OR 'life limit*' OR 'life limit*' OR eol OR 'end-of life*' OR terminal* OR 'life sustain*') NEAR/4 (need* OR care OR nurs* OR intervent* OR medic* OR caring OR assist*)):ab,ti			453,587
☐ #2	chromosom*:ab,ti OR karyotyp*:ab,ti OR trisom*:ab,ti OR syndrom*:ab,ti OR downs:ab,ti OR william*:ab,ti OR 'prader willi':ab,ti OR 'intellectual disabil*':ab,ti OR 'intellectually disabled':ab,ti OR 'mentally disabled':ab,ti OR 'mental disabil*':ab,ti OR 'intellectual development disorder*':ab,ti OR 'mental handicap*':ab,ti OR 'mentally handicapped':ab,ti OR 'mentally impaired':ab,ti OR 'mental impairment*':ab,ti OR 'intellectual impairment*':ab,ti OR 'mental defici*':ab,ti OR 'intellectual retard*':ab,ti OR 'mental retard*':ab,ti OR 'mentally retarded':ab,ti OR 'intellectually challenged':ab,ti OR 'intellectually deficient*':ab,ti OR 'intellectually handicapped':ab,ti OR 'intellectually impaired':ab,ti OR 'intellectually retarded':ab,ti OR 'mentally challenged':ab,ti OR 'mentally defici*':ab,ti OR 'cognitive disab*':ab,ti			2,494,093
☐ #1	'genetic disorder'/exp OR 'down syndrome'/exp OR 'williams beuren syndrome'/exp OR 'mentally disabled person'/exp OR 'mental deficiency'/exp OR 'intellectual impairment'/exp OR 'cognitive defect'/exp OR 'chromosome disorder'/exp OR 'non-hereditary chromosome disorders'/exp OR 'multidisciplinary team'/exp			2,386,726

Appendix 2: The PRISMA Extension for Scoping Reviews (PRISMA-ScR): Checklist and Explanation

SECTION	ITEM	PRISMA-ScR CHECKLIST ITEM	REPORTED ON PAGE #
TITLE			
Title	1	Identify the report as a scoping review.	31
ABSTRACT			
Structured summary	2	Provide a structured summary that includes (as applicable): background, objectives, eligibility criteria, sources of evidence, charting methods, results, and conclusions that relate to the review questions and objectives.	Click here to enter text.
INTRODUCTION			
Rationale	3	Describe the rationale for the review in the context of what is already known. Explain why the review questions/objectives lend themselves to a scoping review approach.	Click here to enter text.
Objectives	4	Provide an explicit statement of the questions and objectives being addressed with reference to their key elements (e.g., population or participants, concepts, and context) or other relevant key elements used to conceptualize the review questions and/or objectives.	Click here to enter text.
METHODS			
Protocol and registration	5	Indicate whether a review protocol exists; state if and where it can be accessed (e.g., a Web address); and if available, provide registration information, including the registration number.	-

SECTION	ITEM	PRISMA-ScR CHECKLIST ITEM	REPORTED ON PAGE #
Eligibility criteria	6	Specify characteristics of the sources of evidence used as eligibility criteria (e.g., years considered, language, and publication status), and provide a rationale.	
Information sources*	7	Describe all information sources in the search (e.g., databases with dates of coverage and contact with authors to identify additional sources), as well as the date the most recent search was executed.	Click here to enter text.
Search	8	Present the full electronic search strategy for at least 1 database, including any limits used, such that it could be repeated.	Table 2
Selection of sources of evidence	9	State the process for selecting sources of evidence (i.e., screening and eligibility) included in the scoping review.	Click here to enter text.
Data charting process	10	Describe the methods of charting data from the included sources of evidence (e.g., calibrated forms or forms that have been tested by the team before their use, and whether data charting was done independently or in duplicate) and any processes for obtaining and confirming data from investigators.	Click here to enter text.
Data items	11	List and define all variables for which data were sought and any assumptions and simplifications made.	Click here to enter text.
Critical appraisal of individual sources of evidence§	12	If done, provide a rationale for conducting a critical appraisal of included sources of evidence; describe the methods used and how this information was used in any data synthesis (if appropriate).	-
Synthesis of results	13	Describe the methods of handling and summarizing the data that were charted.	Click here to enter text.
RESULTS			

SECTION	ITEM	PRISMA-ScR CHECKLIST ITEM	REPORTED ON PAGE #
Selection of sources of evidence	14	Give numbers of sources of evidence screened, assessed for eligibility, and included in the review, with reasons for exclusions at each stage, ideally using a flow diagram.	Figure 1
Characteristics of sources of evidence	15	For each source of evidence, present characteristics for which data were charted and provide the citations.	
Critical appraisal within sources of evidence	16	If done, present data on critical appraisal of included sources of evidence (see item 12).	-
Results of individual sources of evidence	17	For each included source of evidence, present the relevant data that were charted that relate to the review questions and objectives.	Table 4
Synthesis of results	18	Summarize and/or present the charting results as they relate to the review questions and objectives.	Click here to enter text.
DISCUSSION			
Summary of evidence	19	Summarize the main results (including an overview of concepts, themes, and types of evidence available), link to the review questions and objectives, and consider the relevance to key groups.	Click here to enter text.
Limitations	20	Discuss the limitations of the scoping review process.	
Conclusions	21	Provide a general interpretation of the results with respect to the review questions and objectives, as well as potential implications and/or next steps.	Click here to enter text.
FUNDING			
Funding	22	Describe sources of funding for the included sources of evidence, as well as sources of funding for the scoping review. Describe the role of the funders of the scoping review.	

JB I = Joanna Briggs Institute; PRISMA-ScR = Preferred Reporting Items for Systematic reviews and Meta-Analyses extension for Scoping Reviews.

* Where *sources of evidence* (see second footnote) are compiled from, such as bibliographic databases, social media platforms, and Web sites.

† A more inclusive/heterogeneous term used to account for the different types of evidence or data sources (e.g., quantitative and/or qualitative research, expert opinion, and policy documents) that may be eligible in a scoping review as opposed to only studies. This is not to be confused with *information sources* (see first footnote).

‡ The frameworks by Arksey and O'Malley (6) and Levac and colleagues (7) and the JB I guidance (4, 5) refer to the process of data extraction in a scoping review as data charting.

§ The process of systematically examining research evidence to assess its validity, results, and relevance before using it to inform a decision. This term is used for items 12 and 19 instead of "risk of bias" (which is more applicable to systematic reviews of interventions) to include and acknowledge the various sources of evidence that may be used in a scoping review (e.g., quantitative and/or qualitative research, expert opinion, and policy document).

From: Tricco AC, Lillie E, Zarin W, O'Brien KK, Colquhoun H, Levac D, et al. PRISMA Extension for Scoping Reviews (PRISMA-ScR): Checklist and Explanation. *Ann Intern Med.* 2018;169:467–473.

Appendix 3: Description of results

Author's, Year, Title, Country	Participants	Aims and Objectives	Methodology	Key Findings in relation to the research question	Methodological Limitations
Marsh et al., (2018) "Something was wrong": A narrative inquiry of becoming a father of a child with an intellectual disability in Ireland. Ireland	Ten fathers of children with an intellectual disability Including: Global developmental delay, unknown cause and Trisomy 21 CA: Trisomy 21	To present Irish fathers' narratives within the context of leading up to the diagnosis and the actual diagnosis of a child's intellectual disability.	A qualitative narrative inquiry Semi structured interviews Thematic analysis	• The visible and invisible nature of an intellectual disability • Emotional downturn • Denial and blame • Shock and disappointment - Grief and loss	The findings of this study are based on 10 fathers' experiences. Fathers were White, educated and had the continuous support of a long-term partner/wife. The findings do not claim to represent the universal perspectives of all fathers' who have been informed of their child's disability so generalisations based on this

					study cannot be made.
Marsh et al., (2020) The views and experiences of fathers regarding their young child's intellectual and developmental disability diagnosis: Findings from a qualitative study. Ireland	Ten fathers of children with an intellectual and developmental disability Including: Global developmental delay, Unknown cause and Trisomy 21 CA: Trisomy 21	To report the views and experiences of fathers following their child's diagnosis of an intellectual and developmental disability.	A secondary analysis of a qualitative data from Marsh et al. 2018 (see above). Qualitative descriptive study Thematic analysis	• Confirmation of their child's diagnosis • The impact of the diagnosis • Motivation to participate in disability research	Restriction to one country and limited to one service provider; therefore, the findings may not be reflective of father's experiences elsewhere.
Barros Torquo et al., (2013) Paternal Participation in Caring for the Child with Down Syndrome. Brazil	Eight fathers of children with down syndrome CA: Trisomy 21	To understand the paternal involvement in caring for the child with Down Syndrome.	Descriptive and exploratory study with a qualitative approach Semi structured interviews Thematic analysis	• Repercussion of the diagnosis of Down syndrome for the father • Involvement of father in the care for the child with Down syndrome • Multidisciplinary team in the care for children with Down syndrome	There is a lack of clarity surrounding the methodological procedures, which limits insight into how findings were generated and understood.

Marshak et al., (2019) Listening to fathers: Personal impacts of raising children with Down syndrome. USA	Three hundred and eleven fathers of children with Down syndrome CA: Trisomy 21	To contribute to an understanding of how biological fathers believe having a child with Down syndrome has impacted them personally.	Large scale Qualitative study Open-ended survey questions Thematic analysis	• Personal growth • Joy and gratitude • Stress and anxiety • Sadness, loss and depression.	Since demographic data was not collected, this limits the generalisability regarding the proportions of fathers who appear to cope well rather than fixate on the perceived struggles of having a child with Down syndrome.
Takataya et al., (2016) Perceptions and Feelings of Fathers of Children with Down Syndrome. Japan	Nine fathers of children with Down syndrome CA: Trisomy 21	To obtain information about the lives of families with children with Down syndrome through interviews with the fathers.	Descriptive exploratory design utilizing a focus group Qualitative Thematic analysis	• Crisis of life • Emotional turmoil • Attitudes toward the disability • Parental responsibility • Realization of fatherhood • Difference of feelings between mother and father	The sample size of 9 participants may not fully reflect the process of becoming a father of a disabled child. Recruited only through the

				• Contact with people	Down syndrome society.
Rivard & Mastel-Smith (2014) The Lived experience of Fathers whose children are diagnosed with a genetic disorder. USA	Six fathers of children with Genetic disorders CA: Trisomy 21, Prader Willi syndrome	To describe the father's experience of having a child diagnosed with a genetic disorder.	Qualitative Interpretive phenomenological analysis In-person interviews for participants living within 100 miles of the researcher; phone interviews with participants living more than 100 miles from the researcher	• The impact of diagnosis • Seeking understanding, coping with effects of the disorder • Looking to the future • Addressing father's needs	Small sample size. Different methods of data collection, some over the phone without video call options.

Boström & Broberg (2014) Openness and avoidance -- a longitudinal study of fathers of children with intellectual disability. Sweden	Seven fathers of children with intellectual disability Including: Awaiting Autism diagnosis CA: Trisomy 21, Deletion chromosome 14, Mosaic trisomy 14	To explore fathers' experiences of parenthood in relation to a child with intellectual disabilities from the initial discovery of the disability to 5 years later.	Qualitative longitudinal design Interpretative phenomenological analysis A series of semi-structured interviews Thematic analysis	• An interrupted path – no longer taking things for granted, which describes the fathers' reactions to their children's diagnosis • Being a good father, which describes the fathers' overall perceptions of their parenting of a child with intellectual disabilities • Dealing with the unexpected, which describes fathers' individual ways of integrating, managing, and living with the knowledge of their child's disability	The relatively heterogeneous sample. The themes of the present study were identified through various levels of interpretation as the aim of IPA is to make sense of the participants' experiences. Hence the formulation of these themes needs to be considered as one of several possible ways of interpreting the relationships between fathers' internal experiences and external factors.
Ridding & Williams (2019)	Fifteen fathers of children	To investigate fathers'	Grounded Theory	• Accommodating the child	Theoretical sampling may

Being a dad to a child with Down's syndrome: Overcoming the challenges to adjustment. UK	with Down syndrome CA: Trisomy 21	experiences of parenting a child with Down's syndrome, their contributions, influences on family functioning and inclusion in their child's support provision.	Semi-structured interviews were conducted Qualitative Thematic analysis	- Adapting to parental/ spouse roles - Adapting to society	limit the generalizing of findings to other populations. Self-selecting sample consisting of mainly White British fathers in financially stable jobs.
Lowit & Averill (2014) Fathers Reflect on Their Experiences of the Receipt of a Postnatal Diagnosis of Down Syndrome or Trisomy 2. USA	Thirteen fathers of children with Down syndrome CA: Trisomy 21	To investigate the experiences of fathers upon the postnatal news that their new-born babies had been given diagnoses of Down syndrome.	Qualitative exploratory study Semi- structured interviews Fathers known to the researcher and to various human service professionals and colleagues.	- Receiving the diagnosis - Messengers and experiences. - The importance of congratulations, Information, resources, and referrals Emotions - Insensitive comments - Additional findings were related to heart surgery for the babies, superstar stereotyping, and fathers' reactions to	Limited to one geographical region. Another limitation was the lack of diversity in this sample because the majority of research participants learned of this study through an organization for people with Down syndrome

				the interview process	and their families. Only included postnatal diagnosis of Down syndrome. Lacks sufficient detail about its methodological processes.
Cohen et al., (2016) Fathers of children with or without ID: Understanding long-term psychological symptoms. USA	One hundred and eighty-two fathers of children with and without an intellectual disability Including: Cerebral palsy, Unknown developmental disability, Trisomy 21 CA: Trisomy 21	To understand how one source of stress-reports of child behaviour problems, along with spousal support related to parenting tasks, associated with fathers' reports of psychological symptoms over time.	Quantitative Completed measures of child behaviour problems, spousal support and psychological symptoms.	- Fathers' reports of child behaviour problems predicted initial levels of paternal psychological symptoms but did not predict change in paternal psychological symptoms over time - Spousal support further reduced initial levels of paternal psychological symptoms	This study did not examine positive factors that may moderate psychological symptoms, such as optimism or positive perceptions.

Cameron & Cooper (2020) Fathers' Experiences as Carers for Autistic Children with Learning Disabilities. UK	Four fathers of children with Autism spectrum condition and learning disability (Dual Diagnosis) CA: Trisomy 21	To get as close as possible to understanding the experience of these fathers in their role as carers.	Qualitative Interpretative phenomenological analysis Interviews Analysis: 3-layer coding process	• Fatherhood: not doing enough, not doing it right • Crossing worlds: relearning how to communicate and reclaiming fatherhood • Uncertain futures	Small sample size of 4 fathers. Whilst "generalisation" in the tradition scientific sense is not appropriate for such a study, the conclusions drawn can be used to illuminate the experiences of those in similar positions.
Bentley et al., (2015) Down Syndrome and Fathering: An Exploration of Ambiguous Loss. USA	Fifty fathers of children with Down syndrome CA: Trisomy 21	To describe father's attitudes and coping behaviours, responses were evaluated in the context of Ambiguous Loss theory. This was part of a broader study (Springer & Goff, 2011).	Mixed method approach Hope, satisfaction, and coping were assessed by statistical analysis With three open ended questions	• The fathers expressed the ambiguity of loving the child they had while mourning a life they may have anticipated • Three clusters of fathers (Mastering, Connecting, and Thriving) were identified and described • Mastering cluster fathers used action-	Small sample size, which for qualitative limits generalizability. The study provides limited detail regarding its qualitative methodological approach, making it difficult to assess the rigor

				oriented coping strategies to manage their life	of the research process.
Vitiritti et al., (2015) Fathers' Perceptions of Their Expectations about the Future of Children with Mental Disability. Brazil	Twelve fathers of children with a mental disability Including: Unknown cause, Moebius syndrome, Cerebral Palsy, Metabolic error, degenerative disease and Trisomy 21 CA: Trisomy 21	To reveal the paternal perceptions about their expectations for the future of children with mental Disability.	Qualitative Phenomenology, in the hermeneutic perspective of Heidegger Semi structured interviews Thematic analysis	• Designing the future of the child from the news of disability • Reflecting about the future in daily life • Realizing that the child with disability has his own development time • Realizing the child's dependency • Having hope and faith as the future	Phenomenology , in the hermeneutic perspective of Heidegger, was used but little information on how this was applied. Small sample size was rationalised.
Docherty & Dimond (2018) " Yeah, that Made a Big Difference!": Importance of the Relationship between Health Professionals and Fathers Who Have a Child with Down Syndrome. UK	Seven fathers of children with Down syndrome CA: Trisomy 21	To explore the experiences of seven fathers who have a child with Down syndrome, focusing on their interactions	Qualitative interpretative phenomenological analysis (IPA) Semi structured interviews	• Evaluations of Health Care Experiences: Feelings of Exclusion, A Sense of Inequality • Expectations of care: Continuity of Care, Clear and Direct Communication	This is a small study, of seven fathers, which might not be generalizable to a much larger population. As specific demographic

		with health professionals.	Interoperative phenomenological analysis	Health professionals' attitudes towards Down syndrome: How Information Is Presented, Reactions of Health Professionals to their Child	material is not provided, to protect the anonymity of this small sample, how the results may have been impacted by a number of variables including parenting history and experience, the parents' occupation, the age and health of child, is not explored. All fathers utilized the support group.
Huang et al., (2012) (B) Father's Experiences of Involvement in the Daily Care of Their Child with Developmental Disability in a Chinese Context. Journal of Clinical Nursing 21(21–22), 3287–96.	Sixteen fathers of children with a developmental disability Including: Cerebral palsy,	To explore fathers' experience of involvement in the daily care of a child with developmental	Qualitative A hermeneutic phenomenological approach was applied Interviews	- Keeping hope alive - Concerns about quality of medical care - Maximising family function	This study did not recruit fathers of children with a specific diagnosis, certain type of

China	Chromosomal abnormality (included Trisomy 21 in this), Autism 1 Tetralogy of Fallot, Hydrocephaly , Stroke 1 , Neurological sequelae of infection with enterovirus 2 CA: Chromosomal abnormality (included Trisomy 21 in this)	disability within Chinese culture	Thematic analysis		developmental disability, or age, which may have influenced the fathers' experience. The small sample size prevents generalisation of the findings to other fathers of disabled children. However, small samples are needed in qualitative research to provide thick descriptions and saturation of data to richly describe fathers' experiences. Lack of clarity in the

					hermeneutic approach
How et al., (2019) ‘We would have missed out so much had we terminated’: What fathers of a child with Down syndrome think about current non-invasive prenatal testing for Down syndrome. Australia	Five fathers of children with Down syndrome CA: Trisomy 21	To explore fathers of children with Down syndrome about their views on the availability of Non-Invasive Prenatal Testing.	Qualitative Symbolic interactionist framework described by Blumer (1986) In depth interviews Thematic analysis	- Fathers appreciated an early diagnosis with NIPT, they saw the test as being a predetermined pathway to termination - Fathers felt that expectation to terminate reflects negative societal attitudes towards those with Down syndrome and disability, fearing that NIPT may become a form of eugenics - Fathers retrospectively contrasted these attitudes with the actual reality of raising their children, which they described as bringing joy to their lives.	A majority of participants were recruited from an online support group that advocates for disability rights. All participants had a relatively young child with Down syndrome and were mostly Caucasian and well-educated. The fathers in this study all had direct contact with health professionals during the prenatal stage. This may be unusual given

					that in many cases, the information is given to the mother who then passes it on to the fathers.
Huang et al., (2012) Fathers of Children with Disabilities: Encounters with Health Professionals in a Chinese Context. China	Sixteen fathers of children with a developmental disability. including: Cerebral palsy, Autism, Trisomy 21, Congenital heart disease, Joubert syndrome, Hydrocephaly, Spinocerebellar ataxia, Stroke, Chromosomal abnormality, Neurological sequelae of	To examine the experiences of fathers of developmentally disabled children during interactions with health professionals in Taiwan.	Hermeneutic phenomenological design based on the philosophical world views of Heidegger (1962) and Gadamer (1975) Qualitative 2 semi-structured interviews Thick description using the hermeneutic circle Thematic analysis	• Experiencing no supportive communication • Missing the critical time for disability management • Being excluded from medical decision making	This study did not recruit fathers of children with a specific diagnosis, certain type of developmental disability, or age, which may have influenced the fathers' experience. The small sample size prevents generalisation of the findings to other fathers of disabled children. However, small samples are

	infection with enterovirus 71 CA: Unnamed CA and included Trisomy 21				needed in qualitative research to provide thick descriptions and saturation of data to richly describe fathers' experiences. Lack of clarity in the hermeneutic approach .
Huang et al., (2011) Taiwanese fathers' experiences of having their child diagnosed with a developmental disability. China	Sixteen fathers of children with a developmental disability. Including: Cerebral palsy, Autism Trisomy 21. Congenital heart disease, Joubert syndrome, Hydrocephaly, Spinocerebellar ataxia,	To explore fathers' experiences of having a child diagnosed with a developmental disability in a Chinese cultural context	Hermeneutic phenomenological approach informed by the philosophical world views of Heidegger In-depth and semi structured interviews and were analysed using hermeneutic analysis Thematic analysis	• Losing hope • Feelings of failure • Being frustrated with family conflicts • Searching for positive coping strategies	This study did not recruit fathers of children with a specific diagnosis, certain type of developmental disability, or age, which may have influenced the fathers' experience. The small sample size prevents

	Stroke, Neurological sequelae of infection with enterovirus 71 CA: Unnamed CA and included Trisomy 21				generalisation of the findings to other fathers of disabled children. However, small samples are needed in qualitative research to provide thick descriptions and saturation of data to richly describe fathers' experiences. Lack of clarity in the hermeneutic approach.

Baumann & Braddick (1999) Out of their element: fathers of children who are “not the same.” USA	Eleven fathers of children with: Congenital anomalies, Trisomy 21, Significant intellectual disabilities, Heart defects Cleft lip and pallet, Severe cranial facial abnormalities CA: Unnamed CA and included Trisomy 21	To ascertain the meaning of fathering for fathers of children with congenital anomalies	Qualitative descriptive-exploratory study: 1.1 semi-structured interviews Thematic analysis	• Participants is a desire to grasp the situation, mingled with disturbing feelings, while facing one's limits and gives rise to comforting views • Comfortable-uncomfortable ways, of being with others emerge as fathering shifts one from the familiar to the unfamiliar, surfacing the joy-sorrow of the always-changing 'now' and the unpredictable 'not yet.'	Range of disabilities.
Giallo et al., (2015) Risk factors associated with the mental health of fathers of children with an intellectual disability in Australia. Australia	Three hundred and fifteen fathers of children with Intellectual disabilities including: Autism, Vision impairment, Hearing	To redress these gaps by reporting on the mental health of a large sample of fathers of children with an intellectual disability (ID)	Quantitative Large-scale evaluation of the Signposts for building better behaviour program Fathers completed a	• Fathers in the present sample reported significantly more symptoms of depression and stress than the Australian normative data, with approximately	The findings are based on a sample of fathers enrolled to participate in a parenting programme to manage their children's behaviour.

	impairment, Trisomy 21 Cerebral palsy, Epilepsy or other seizure disorder, Psychiatric disorder, Attention deficit and hyperactivity disorder, Acquired brain injury, Health problems (i.e., asthma, food allergies) CA: Trisomy 21	(aged 3-15 years), comparing this to published Australian norms and mothers of children with ID. The second aim of the study was to explore risk factors associated with fathers' mental health.	range of self-report questionnaires at baseline including the Depression Anxiety Stress Scale (DASS). Statistical analysis	6–8% reporting symptoms in the severe to extremely severe range • The strongest predictors of fathers' mental health difficulties were children's behaviour problems, daily stress arising from fathers' own needs and children's care needs, and low parenting satisfaction • Socio-economic factors did not predict mental health difficulties	Therefore, these findings may reflect the specific experiences of fathers who are very concerned about their children's behaviour, and those who are open to help-seeking and professional support. Fathers were predominantly partnered and employed, and thus the results may not be generalisable to fathers heading one-parent households or those not living with their children. The findings are based on cross-

					sectional data and causality cannot be inferred. It is likely that complex transactional and mediating relationships exist among the risk factors and mental health variables of interest.
Ricci & Hodapp (2003) Fathers of children with Down's syndrome versus other types of intellectual disability: perceptions, stress and involvement. USA	Fifty fathers of children with down syndrome versus those with other types of disability intellectual disability including: non-specific intellectual disabilities, Williams syndrome, Prader-Willi	Examined fathers' perceptions of stress relating to and involvement with children with Down's syndrome versus those with other types of intellectual disability.	Quantitative Personality Checklist Child Behaviour Checklist (CBCL). Parenting Stress Index (PSI)	- Fathers rated their children with Trisomy 21 as having more positive personality traits and fewer maladaptive behaviours - The two groups of fathers were very similarly involved in child rearing - The personality, age and maladaptive behaviours of the children related to stress levels in the	All the measures of fathers' perceptions, stress and involvement in this study were questionnaires. Although fathers' answers were similar to those of mothers, observing fathers in their interactions with their

	syndrome, cerebral palsy, Autism, CHARGE syndrome). CA: Trisomy 21, Williams syndrome, Prader–Willi syndrome			fathers of children with Trisomy 21, while maladaptive behaviours, gender and the fathers' education levels related to stress levels in the fathers of children with other types of intellectual disability	children may better distinguish paternal involvement in the two groups. The study did not include measures of other variables which could influence the fathers' participation with their children, such as motivation, social support and work-related limitations. The study does not include any measures of physical health and physical disabilities as potential stressors for parents.

Schippers et al., (2020) The experiences of Dutch fathers on fathering children with disabilities: 'Hey, that is a father and his daughter that is it'. Netherlands	Twelve Fathers of children with disabilities including: Unspecific disabilities but mentioned Trisomy 21 CA: Trisomy 21	To gain insight into the perceived experiences of fathers of children with disabilities.	A qualitative study Semi Structure Thematic analysis	• Role identification, commitment, employment characteristics, cultural expectations and social support • As well as this new life perspective included a positive attitude, living in the moment, appreciation of the little things and transformation of expectations	This study is qualitative but does not explicitly state a guiding methodology.
Ly & Goldberg (2014) New measure for fathers of children with developmental challenges. USA	One hundred and one fathers of children with an intellectual disability including: Autism, Trisomy 21, Other intellectual disabilities, Attention deficit	To create and validate a brief measure designed to capture the perceptions and experiences of these fathers.	Mixed methods Semi- structured interviews Children with Developmental Challenges questionnaire	• Fathers feeling that professionals considered them secondary to mothers • Fathers felt isolated, the FCDC tool was straightforward to use and represented their experiences • Reasons for not attending child meetings included maternal role and work issues	The majority of the fathers had children with Autism. Reliance on a self-report methodology.

	hyperactivity disorder CA: Trisomy 21			The tool was shown to be reliable demonstrated content and construct validity and measured father's perceptions of support and barriers to their involvement with a child with an intellectual disability	
Mac Donold & Hastings (2010) Mindful Parenting and Care Involvement of Fathers of Children with Intellectual Disabilities. Ireland	One hundred and five fathers of children with intellectual disabilities including: Trisomy 21, Autism, Cerebral palsy CA: Trisomy 21	To determine if fathers who were more mindful in their parenting role (specifically, fathers who reported more present-centered attention in their relationship with their child) would use less avoidance in relation to their child with intellectual	Quantitative questionnaire survey (a mindful parenting measure and a measure of parental involvement)	- Fathers who did not work outside of the home and fathers whose child was younger were more involved in daily care tasks in caring for their children - Fathers who did not work outside of the home also carried out more child related parenting tasks - Fathers were more involved with socialisation related parenting tasks with their child with an	The measurement of mindful parenting was also limited. The Duncan (2007; Duncan et al. 2006) mindful parenting scale has further dimensions including non-reactivity and non-judgemental acceptance and other measures of mindfulness also include a

		disability and that this would be reflected in increased father involvement in childcare. In a questionnaire survey 105 fathers completed a mindful parenting measure and a measure of parental involvement.		intellectual disability when: their partner worked outside of the home, they had a male child, their child did not have Down syndrome, and their child had autism	number of different facets (e.g., Baer et al. 2006). Thus, future research should include further facets of mindfulness as relevant to the parenting role. The measurement of father involvement in childcare. Specifically, we have no more objective measure of father involvement (e.g., observed parenting behaviors) or reports from the other parent. The response rate obtained is unlikely to

					mean that the sample was representative of fathers of children with intellectual disabilities even in an Irish culture.
Ogourtsova et al., (2021) Fathers Matter: Enhancing Healthcare Experiences Among Fathers of Children with Developmental Disabilities. Canada	Seven fathers with children with developmental disabilities including: Epileps, Anxiety, Cognitive and language delay, Trisomy 21, Cerebral palsy, Autism, Attention deficit and hyperactivity disorder, Developmental coordination disorder, Behavioural challenges,	To explore barriers to and facilitators of positive and empowering healthcare experiences, from the perspectives of fathers of children with DD and Healthcare professionals.	A mixed-method approach, such as quantitative (survey) and qualitative (semi-structured interview) strategies, was used. Data analysis consisted of using descriptive statistics and an inductive-thematic analysis of emergent themes	- Fathers indicated that while they were moderate to very much satisfied with their interactions with healthcare professionals, they reported that Healthcare professionals were only sometimes attentive to them during interactions - Fathers also revealed that positive interactions with HCPs in relation to their children had multiple benefits - Several themes related to barriers and facilitators of	Small sample size of father-participants, potentially limiting the generalizability of the findings. The sample also included fathers of children from a wide age range and variability in diagnoses, single and married individuals, and of different educational backgrounds.

	Globoid cell leukodystrophy. 13 health care professionals CA: Trisomy 21			optimal interactions and parent–professional relationships emerged. These included: Session factors (time, attention), personal factors (knowledge of the condition, child and healthcare system, acceptance vs. denial, previous experiences, culture, stereotypes, pre-existing beliefs, stress levels, working schedule), and family dynamics	
Sheldon et al., (2021) Rewards and challenges of parenting a child with Down syndrome: a qualitative study of fathers' perceptions. USA	One hundred and seventy-five fathers of children with Down syndrome CA= Trisomy 21	To explore what was rewarding and challenging about parenting a child with Down syndrome. To take steps toward filling this gap in the research, the	Qualitative: Survey of open-ended questions Context analysis	• Child's Positive Attributes and Behaviours • Rewarding Father-Child Relationships • Child Has Positively Changed Father • Father's Positive Relationship with Others • Key themes for challenges included:	Self-selected, support-group sample is likely not representative of fathers from all families with a child with Down syndrome.

		purpose of our study is to broaden perspectives on fathers' experiences of parenting children with Down syndrome.		Father's Thoughts, Emotions, and Behaviours, Child's Challenging Behaviour and Development, System and Institutional Problems, Other people's Negative Attitudes/Behaviours	There was also a lack of diversity in fathers' children with Down syndrome.
Uribe-Morales, et al., (2022) Fathers in the Care of Children with Disabilities: An Exploratory Qualitative Study. Healthcare Spain	Seven fathers of children with disabilities including: Hearing impairment, 1P36 deletion syndrome, Trisomy 21, Mowat Wilson syndrome, ADHD accompanied by high abilities and oppositional defiant	To explore the perception and experience of fathers of children with disabilities in caring for their children; to know their role and how these tasks impact their daily life, health and physical, mental and/or emotional well-being.	Qualitative: phenomenological design Semi-structured interviews Discourse analysis through open and axial coding processes	- Shared responsibilities - Somewhat difficult to fit in either you join, or you split	The study reports enormous difficulties in finding a sufficiently large sample of fathers with minor children with disabilities who wanted to participate. The sample is also made up of fathers of children of

	disorder, and Autism CA: 1P36 deletion syndrome, Trisomy 21				different ages and disabilities. The interviews were all telephone interviews. Although this technique allowed for the inclusion of fathers from other areas of the country, it may have reduced the richness of the interviews due to the absence of visual observation. It is also important to bear in mind that, given the object of the study as they may understand it as “an examination” about their

					involvement in the care of their children, there was a possibility that participants may have been reluctant to give socially unacceptable answers, for fear of being judged.
Pisula & Banasiak (2020) Empowerment in Polish fathers of children with autism and Down syndrome: the role of social support and coping with stress – a preliminary report. Poland	One hundred and twelve fathers of children with Childhood autism with intellectual disability and children with Trisomy 21 and “Typical children” CA: Trisomy 21	To explore (1) Whether fathers of children with autism and DS and typically developing (TD) children differ in terms of empowerment; (2) if social support and coping styles correlate with fathers' empowerment; and (3) if there are differences between those	Quantitative Fathers completed three questionnaires: Family Empowerment Scale, Ways of Coping Questionnaire and Social Support Questionnaire – Short Form. Results: Compared with other groups, family empowerment	• Compared with other groups, family empowerment was lower in fathers of children with autism • No group differences were found for the service system and community dimensions of empowerment • In the fathers of children with autism, social support was not related to empowerment, but	Small size and internal heterogeneity of samples in terms of fathers' (e.g. education) and children's (e.g. ID severity, child gender and age range) characteristics. The use of instruments that were not validated on

		groups in these areas.	was lower in fathers of children with autism.	there was a negative correlation between emotional coping and service system • Satisfaction with social support in fathers of children with Down syndrome correlated positively with the community dimension of empowerment	large Polish samples.
Dabrowska & Pisula (2010) Parenting stress and coping styles in mothers and fathers of pre-school children with autism and Down syndrome. Poland	Twenty-six fathers and Twenty six mothers) of children with Down syndrome or with Autism and fifty-seven parents of typically developing children. CA: Trisomy 21	To explore (1) the profile of stress in mothers and fathers of preschool children with autism, Down syndrome and typically developing children. (2) to assess the association between parenting stress	Quantitative: Holroyd's 66-item short form of Questionnaire of Resources and Stress for Families with Chronically Ill or Handicapped Members and the Coping Inventory for Stressful Situations by	• Higher level of stress in fathers of children with Autism • Additionally, an interaction effect was revealed between child diagnostic group and parent's gender for two scales of parenting stress, dependency and management and limits of family • Mothers of children with autism scored	The groups of parents included in the analysis were relatively small, with significant internal variation. Only the parents of children who used professional assistance in therapeutic centres and

		and coping style.	Endler and Parker.	higher than fathers in parental stress No such differences were found in the group of parents of children with Down syndrome and typically developing children	kindergartens were recruited. The level of development of children with autism and children with Down syndrome varied. The severity of children's behavioural problems, which obviously affected parental stress, was not accounted for in the present study.
Rose (2021) Narrative Experiences of Fathers of Children with Down syndrome. USA	Twenty-two fathers of children with Trisomy 21 CA: Trisomy 21	To explore fathers' experiences of caring for a child with down syndrome	Thesis dissertation Narrative methodology Two-question, semi-structured interview with a	During diagnosis, fathers experienced adverse encounters with medical personnel, intense negative emotions about the diagnosis, distressing episodes of medical complexity in their	Author was a father of a child with Trisomy 21.

			follow-up conversation.	children, and fruitless and frustrating trials of information-gathering Fathers experienced changes to their roles, responsibilities, and relationships within their families, but were happy with their lives and proud of their children	
Armijos-Yambay (2020) Fathers matter too! Lived experience of pedagogical hope in raising children with Down syndrome. Spain, Ecuador and Scotland	Ten fathers of children with Down syndrome <i>Note: 2 adult children were included but main focus was children</i> CA: Trisomy 21	To explore fathers' lived experiences and to reflect on what it means to be a pedagogically hopeful father of a child with Down syndrome	Max van Manen's hermeneutic–phenomenological method Face-to-face phenomenological interviews Thematic analysis	- A pedagogically hopeful father is realistic - Believes in the strengths of his child with down syndrome - Trusts in his positive influence is open to unexpected surprises	The findings of the study cannot be empirically generalised. The excerpt provides no indication that the researcher reflected on their own positionality
MacDonald et al., (2010)	Ninety-nine fathers of children with	To explore the potential for psychological	Quantitative assessment measuring	Psychological acceptance was found to partially	Small sample size; response rate suggested

Psychological acceptance mediates the impact of the behaviour problems of children with intellectual disability on fathers' psychological adjustment. Ireland	developmental difficulties including: Down syndrome, Autism Cerebral palsy and the remainder with a variety of different aetiologies or no reported additional diagnosis/ aetiology CA: Trisomy 21	acceptance to mediate the impact of children's behaviour problems on paternal well-being	paternal adjustment Five measures were used	mediate the impact of child behaviour problems • Paternal stress, anxiety, and depression • Acceptance was also a positive predictor of fathers' perceptions of positive gain associated with raising their child with intellectual disability	that the sample was unlikely to have resulted in a representative sample of fathers. The measure of psychological acceptance developed for this research is new and there is a lack of information about its psychometric properties. A third limitation is that of source variance. Fathers provided data on their own psychological functioning and on their child's behaviour problems.

Manor-Binyamini (2025) Bedouin fathers of children with developmental disabilities-Stress, stigma and collaboration with professionals. Bedouin	Eighty-eight fathers of children diagnosed with developmental disabilities including: Down syndrome, chromosomal conditions, Intellectual disability, Intellectual disability, Autism CA: Trisomy 21 and unnamed CA	To explore 1) to investigate whether stigma and stress affect fathers' collaboration with professionals 2) to examine the relationship between stigma, stress and collaboration 3) to examine which variables, including sociodemographic variables, impact collaboration and stress as mediators	Quantitative Statistical tests, such as Pearson's correlation and multiple regression, were carried out to aid in the analyses of data	- The findings revealed a significant positive relationship between stigma and stress, a significant negative relationship between stigma and collaboration and a significant negative relationship between stress and collaboration. Three demographic characteristics were associated with greater influence on fathers' collaboration with professionals: - Comorbidity in child's diagnosis, - Age of DD diagnosis - Father's age	Small sample size. The focus on this Bedouin fathers, this study doesn't deeply engage with the cultural nuances and may not be relevant to Ireland.
Randolph (2023)	Eight fathers of children	To explore perceptions	Generic qualitative study	Experienced stress when raising their	Relatively small sample size .

Fathers' perceptions of Stress and Resiliency in Raising Children with Down Syndrome: A Qualitative Study USA	with Down syndrome CA: Trisomy 21	involving stress and resiliency among fathers whose children had been diagnosed with Down syndrome and understand how stress and resiliency affected the parenting skills of those fathers	qualitative thematic analysis, was guided by the family resilience theory	children who were diagnosed with Down syndrome • The main stressor that fathers experienced was uncertainty involving their children's future • They reported that resilience assisted them with strength and the ability to persevere through challenging and difficult situations • The main barriers they experienced when raising their children with Down syndrome were their children's behaviour, communication, and health	Fathers had children between the ages of 4 and 18 years and lived in one of three counties in Maryland, the results may not be generalizable to other populations and geographical regions. Mother of a child with Down syndrome.
Woodgate et al., (2024) Exploring fathers' experiences of caring for a child with complex care needs through ethnography and arts-based methodologies	Twenty-nine fathers (the findings emerged from a larger on parents) with	To examine how Canadian families of children with	Qualitative research approach of ethnography and arts-based methodologies	• Striving to be there for the child with CCN, was identified • Contributing to the parental team	Most fathers in our sample were of European descent and had an annual

Canada	CCN. Diagnoses included: Cerebral palsy, developmental disorders (e.g., global developmental delay), terminal cancer, seizure disorders, chronic lung disease, congenital disorders, and genetic disorders (e.g., trisomy 21). CA: Trisomy 21	CCN participate in society	(ecomaps and photovoice)	through various roles • Building accessibility through adaptation • Engaging in activities with the child • Expressing admiration and pride in their children • Meaning making	income of ≥\$60,000. Findings emerged from a larger study that sought to understand how parents of children with CCN participate in society. As such, parents spoke about their fathering experiences in that context.
Walker (2012) Expectations and experiences of fathers who have parented children with and without intellectual disabilities USA	Eight fathers of children with and without intellectual disabilities. Disabilities including Asperger's	To explore expectations and subsequent experiences of men who have fathered children with and without	Qualitative descriptive design Face to face interviews Thematic analysis	• Learning to dance in the rain • Just do what needs doing • The power of patience • Nurturing uniqueness	Change of inclusion/exclusion criteria thought the study. Reflective journal not kept up to date.

	syndrome, Autism, rare genetic disorders, Fetal alcohol syndrome, Down syndrome CA: Trisomy 21	intellectual disabilities			Varied in age.

Appendix 4: Letter to gate keeper

Gate Keeper letter to service providers

School of Nursing and Midwifery,
Trinity College Dublin,
24 D'Olier Street,
Dublin 2.

To whom it may concern,

My name is Isobel Mahon. I am a second year PhD candidate in Trinity College Dublin.

I am conducting a study titled 'Fathers experiences of caring for a child with Complex Care Needs and a Chromosome Abnormality' subject to ethical approval by Trinity College Dublin Research Ethics Committee. I am writing to ask your permission for your organisation to be a gatekeeper for this study.

I have attached a copy of my research proposal and associated documentation including, letter of ethical approval, letter to participants, participation information leaflet and informed consent form for your reference.

If this is possible, please could you e-mail me at ismahon@tcd.ie to confirm whether you are willing to allow access and to be a gatekeeper to these fathers providing they agree and are happy to take part.

Thank you for your time and I hope to hear from you soon.

Thank you.

Yours sincerely,

Isobel Mahon

0833420521

PhD Candidate Trinity College Dublin

Letter to online support groups and forums

School of Nursing and Midwifery,
Trinity College Dublin,
24 D'Olier Street,
Dublin 2.

To whom it may concern,

My name is Isobel Mahon. I am a second year PhD candidate in Trinity College Dublin.

I am conducting a study titled 'Fathers experiences of caring for a child with Complex Care Needs and a Chromosome Abnormality' subject to approval by Trinity College Dublin Research Ethics Committee. I am writing to ask your permission for your organisation to be a gatekeeper and to place a poster advertisement on your website for this study.

I have attached a copy of my participation information leaflet and the poster advertisement for your reference.

If this is possible, please could you e-mail me at ismahon@tcd.ie to confirm whether you are willing to allow access and to be a gatekeeper to these fathers providing they agree and are happy to take part.

Thank you for your time and I hope to hear from you soon.

Thank you.

Yours sincerely,

Isobel Mahon

PHD Candidate Trinity College Dublin

Appendix 5: Responses from providers

Please note, respondents' personal information has been redacted: emails, names

Site Number	Service provider	Response
1.	Homecare Provider for children with complex care needs	Email response agreeing to be a gatekeeper
2.	Homecare Provider for children with complex care needs	Email response agreeing to be a gatekeeper
3.	Support to families with rare conditions including chromosome abnormalities	Email response agreeing to be a gatekeeper
4.	Support to families with rare conditions including chromosome abnormalities	Email response agreeing to be a gatekeeper
5.	Supports families of children with [Named syndrome]	Email response agreeing to be a gatekeeper
6.	Supports families of children with [named syndrome]	Email response agreeing to be a gatekeeper
7.	Supports families of children with Support Organisation for [named syndrome]	Email Response agreeing to be a gatekeeper

Site number	Service provider	Response
8.	Homecare Provider for children with complex care needs	Declined due to too many staff nurses completing research and managers capacity to be a gatekeeper
9.	Homecare Provider for children with complex care needs	No response (reminder email sent)
10.	Homecare Provider for children with complex care needs	Did not have fathers who met the inclusion criteria
11.	Voluntary organisation for children with complex care needs	Declined due to families being saturated with research
12.	Organisation to support the inclusion of children with disabilities in society	No response
13.	[named syndrome] organisation	No response
14.	[named syndrome] local organisation	No response
15.	Rare syndromes organisation	No response

16.	[named syndrome] local organisation	No response
17.	[named syndrome] local organisation	No response

Site 1.



Isobel Mahon <ismahon@tcd.ie>

PHD Research

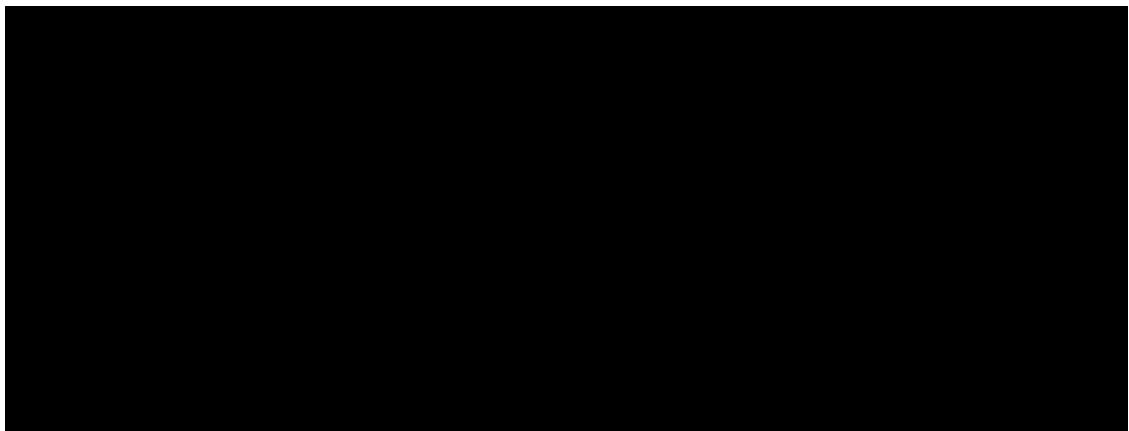
[REDACTED] 10 May 2022 at 14:26

To: "ismahon@tcd.ie" <ismahon@tcd.ie>

Good afternoon Isobel,

This sounds like a worthwhile research topic. I can confirm we will be a gatekeeper for these fathers. Once you have received ethical approval from Trinity Coll Dublin.

I look forward to hearing from you



Site 2.



Isobel Mahon <ismahon@tcd.ie>

**PHD Research on Fathers of Children with Complex Care Needs
and a Chromosome Abnormality**

4 May 2022 at 16:36

To: Isobel Mahon <ismahon@tcd.ie>

Hi Isobel,

Thank you for your email. I am confirming that I am willing to allow access and to be the gatekeeper to these fathers. I am happy to assist in any way possible.

Kind regards,

[Quoted text hidden]

The information contained in this email and in any attachments is confidential and is design solely for the attention and use of the intended recipient(s).

Site 3.



Isobel Mahon <ismahon@tcd.ie>

Research Study on Fathers of Children with Complex Care Needs and a Chromosome Abnormality

William's Syndrome Ireland <info@williamssyndrome.ie>

3 May 2022 at 16:33

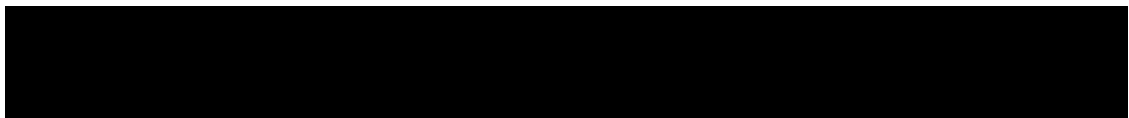
To: Isobel Mahon <ismahon@tcd.ie>

Hi Isobel,

This sounds like a fabulous research project. We would be more than happy to facilitate this or website and Facebook page.

Let me know how I can help in any way!

Kind regards



Site 4.



Isobel Mahon <ismahon@tcd.ie>

Research Study on Fathers of Children with Complex Care Needs and a Chromosome Abnormality

Rare Ireland <rareireland@gmail.com>

3 May 2022 at 22:08

To: Isobel Mahon <ismahon@tcd.ie>

Hi Isobel

Thank you for your email. It's great to see that you are considering your PHD study on chromosome abnormalities. We are happy to help you in any way we can. We have a group of 1400 members most of our children have chromosome abnormalities but most of our members are mothers. We have a handful of Dads so I will put it out there and see if any of them would have the time to part in your study.

Our social media platforms are mainly viewed by members of the general public, so I think the place to share this is within our private group. Unfortunately, as you are not the parent of a child with a rare condition I can't invite you to join but I will gladly share the information you sent and see if we get any takers. If anyone is interested, I'll then pass on your email address if that's ok with you.

Does this sound ok to you?

Kindest regards

Site 5.



Isobel Mahon <ismahon@tcd.ie>

Research Study on Fathers of Children with Complex Care Needs and a Chromosome Abnormality

Info Account <Info@rarechromo.org>

10 May 2022 at 11:57

To: Isobel Mahon ismahon@tcd.ie

Dear Isobel

Many thanks for sharing this study with us. Our internal ethics committee is really happy to support this project as it is of real interest and benefit to our families. And this is a topic that urgently needs more research!

A couple of small points.

- There's mention of meeting in person but also 'no cost' .. so they may wish to men travel costs will not be provided, assuming they won't be

I look forward to hearing from you with an updated, set of documents.

With very best wishes



Site 6.



Isobel Mahon <ismahon@tcd.ie>

PhD Fathers experiences of caring for a child with complex care needs and a chromosome abnormality

10 May 2022 at 17:19

To: Isobel Mahon <ismahon@tcd.ie>

Hi Isobel,

Thanks for your call today.

I am happy to act a gatekeeper for any fathers of children with PWS that wish to participate in your research,

Kind Regards,

Site 7.



Isobel Mahon <ismahon@tcd.ie>

PHD Research

12 May 2022 at 15:50

To: ismahon@tcd.ie

Hi Isobel,

My name is [REDACTED] and I am currently Chair of SOFT Ireland. At our recent commi meeting your email requesting our assistance with your research was discussed and as organisation, we are happy to help out in any way we can.

We are a small charity, but will do our best to assist you in any way that we can.

If you wish to direct all further communication to myself, I will ensure that we will promote it on website and other platforms.

Wishing you the best of luck with this research.

Kind Regards

[REDACTED]

Appendix 6: Online poster advertisement



Coláiste na Tríonóide, Baile Átha Cliath
Trinity College Dublin
Ollscoil Átha Cliath | The University of Dublin

Fathers' do you have a child with complex care needs and a chromosome abnormality?

I would like to invite you to take part in my research study, which is being completed as part of my PhD, which is about exploring the experiences of fathers' who are caring for children with complex care needs and a chromosome abnormality.

This study is going to explore fathers' lived experience of caring to children with complex care needs and a chromosome abnormality. I want to find out:

- Your experiences of caring for your child with complex care needs and a chromosome abnormality
- Your experiences of how your child was diagnosed
- How this diagnosis impacts you
- Your responsibilities in caring for your child with complex care needs and a chromosome abnormality
- What supports you have and how these supports impact your everyday life
- Your recommendations for enhancing care for fathers of children with complex care needs and a chromosome abnormality.

Want to ask more questions or to take part?

You can get more information or answers to your questions about the study, your participation in the study, and your rights, from me, Isobel Mahon who can be telephoned at (0831185105).

Isobel Mahon: PHD Student Trinity College Dublin: ismahon@tcd.ie

Appendix 7: Letter of invitation

School of Nursing and Midwifery,
Trinity College Dublin,
24 D'Olier Street,
Dublin 2.

Dear Sir,

I would like to invite you to take part in a research study entitled "Fathers experiences of caring for their child with a chromosome abnormality and complex care needs".

I would like to learn more about your experience of caring for your child with a chromosome abnormality and complex care needs. This is important as by understanding your experiences, I hope to identify recommendations for enhancing care for fathers of children with complex care needs and a chromosome abnormality.

Before you decide to take part you need to understand why the research is being done, and what it would involve for you. If you would like to participate in the study, please take time to read the information contained in the Participant Information Sheet carefully. Please feel free to contact me if there is anything that is not clear, or if you would like more information.

Thank you.

Yours sincerely,

Isobel Mahon

PHD Candidate Trinity College Dublin

Appendix 8: Participation information leaflet

Study Title: Fathers experiences of caring for their child with complex care needs and a chromosome abnormality

Introduction: I would like to invite you to take part in my research study, which is being completed as part of my PhD, which is about exploring the experiences of fathers' who are caring for children with complex care needs and a chromosome abnormality. I am a dual qualified Intellectual disability and Children's nurse. I currently work in the area of children with Complex Care Needs. I feel that this study will impact positively for fathers' caring for children with Complex needs. I would be extremely grateful if I could hear about your experiences in caring for your child. This information sheet aims to enable you to fully understand this study prior to you agreeing to participate. Please ensure to ask any questions you have about this research project or what your role would be in it.

Principal investigator's name: Isobel Mahon

Principal investigator's Information: My name is Isobel Mahon, and I am a Registered Nurse in Intellectual Disability (RNID) and a Registered Children's Nurse (RCN). I have an MSc in Children and Young People with Complex Care Needs. At present, I am completing my PHD in Trinity College Dublin.

Telephone number of principal investigator: 083 *****

Data Controller: Trinity College Dublin

Data Controller details: College Green, Dublin 2.

Data Protection Officer's Identity: Mr John Eustace

Data Protection Officer's Contact Details: Data Protection Officer, Secretary's Office, Trinity College Dublin, Dublin 2, Ireland. Email: dataprotection@tcd.ie. Tel. 01-8961892.

Before you decide whether or not you wish to take part, you should read the information provided below carefully. Take time to ask questions – don't feel rushed and don't feel under pressure to make a quick decision.

You should clearly understand the risks and benefits of taking part in this study so that you can make a decision that is right for you. This process is known as 'Informed Consent'.

You don't have to take part in this study. You can change your mind about taking part in the study. Even if the study has started, you can still opt out up until the point of data analysis. You don't have to give me a reason.

Part 1 – The Study

Why is this study being done?

This study is going to explore fathers' lived experience of caring for children with complex care needs and a chromosome abnormality. I want to find out:

- Your experiences of caring for a child with complex care needs and a chromosome abnormality
- Your experiences of how your child was diagnosed with a chromosome abnormality and how this has impacted you

What supports you experience and what could be done to support you better

Why have I been invited to take part? • As a father of a child with Complex Care Needs and a chromosome abnormality your experiences of caring for your child are relevant and worth exploring. • Other fathers who have a child with complex care needs and a chromosome abnormality have also been asked to participate.
Do I have to take part? Can I withdraw? • The decision to take part in the study is completely voluntary. If you decide to take part you are free to withdraw without giving a reason up until the point of data analysis. You will complete a written informed consent form if you decide to participate. If you take part over Zoom, your consent will be sent via an online link for completion.
What happens if I change my mind? • You can change your mind or withdraw from the study until the point of data analysis without any implication. You can contact me on (ismahon@tcd.ie).
What will happen to me if I decide to take part? • When you contact me via email (ismahon@tcd.ie), I will arrange a time and place to interview you. • Your responses to interview questions will be audio recorded and transcribed for analysis. • You will be given a journal that you can fill out if you wish. This is voluntary and you do not have to complete the journal to take part.
What will happen to my data? • All information, which is collected about you during the course of the research, will be kept strictly confidential. Your name will not be published and will not be disclosed to anyone. All interview information will be made anonymous using codes and all information included in the report or future publications arising from the study will not identify you. • The final report of the study will not contain information about your identity so that you cannot be recognised from it. A code will be used instead of your name and it will not be possible to identify you in the final results. • The results will be compiled in a report as part of my PHD. The results will also be written up and published in journals. Data will only be retained until such time as all findings are published in peer reviewed journals. Study data will be retained for a maximum of 7 years after which time it will be destroyed as per the General Data Protection Regulations, 2018. Data will not be kept for any future research.
Are there any benefits to taking part in this research? • There may be no direct benefits of you participating in the study although many people do find it helpful to talk about their experiences. Furthermore, the aim of this study is that your experiences will inform how health professionals could better support fathers of children with complex care needs and chromosome abnormalities.
Are there any risks to me or others if I take part? • There are no foreseeable risks to you associated with participating in this study. However, if you wish to stop any of our conversations because you are feeling discomfort you are free to do so at any time. I will stop the interview or alternatively take a break. The interview will only be recommenced if you indicate that you would like to continue.

- If I am concerned for your welfare, I will advise you to contact a relevant person to seek advice/support (i.e. key worker, liaison nurse, GP, pharmacist or relevant other).
- If for any reason or your child's condition deteriorates during the study and you feel you cannot continue, you may withdraw without any penalties.
- This study has received ethical approval and permission to undertake the study has been obtained from Trinity College Dublin.

What happens if something goes wrong when I'm taking part in the study?

- I have years of experience in nursing and are skilled in supporting families of children with complex care needs. I will carefully guide you in discussing your experience and suggest suitable support services if needed. You may stop the interview at any point and withdraw from the study if you wish.

Part 2 – Data Protection

What information about me (personal data) and my child will be used as part of this study?

- Your child's medical records will not be accessed. However, we will ask you what your child's diagnosis is. This will allow us to place your child in the context of the study. This data will be pseudonymised. Your child will not be identifiable.

What will happen to my personal data?

- To ensure privacy and confidentiality in the study, I will be using a pseudonym for you or your child. No names or other personal identifiers will be included.
- Data storage and data analysis will be based on these pseudonyms. The coding for the numerical data identified will be held by myself in a locked cabinet in the school of nursing and will be known only to me.
- The final report of the study will not contain information about your identity so that you cannot be recognised from it. A code will be used instead of your name and it will not be possible to identify you in the final results.
- The results will be compiled in a report as part of my PHD. The results will also be written up and published in journals. Data will only be retained until such time as all findings are published in peer reviewed journals. Study data will be retained for a maximum of 7 years after which time it will be destroyed as per the General Data Protection Regulations, 2018.
- The results will be compiled in a report as part of my PHD. The results will also be written up and published in journals. All data from the study will be destroyed as soon as we have completed this process. No data will be kept for any future research.
- Your consent form will be stored in a locked filing cabinet in the school of nursing in TCD. This will be retained for a maximum of 7 years after which time it will be destroyed as per the General Data Protection Regulations, 2018.

Will my personal data be kept confidential? How will my data be kept safe?

- All information, which is collected about you during the course of the research, will be kept strictly confidential. Your name will not be published and will not be disclosed to anyone. All interview information will be made anonymous using codes and all information included in the report or future publications arising from the study will not identify you.
- If during the interview something is raised that leads me to think a child is at risk, I will discuss this with you. As a mandated person, under the Children First Act 2015, the researcher Isobel Mahon is required by Law to report any knowledge, belief or reasonable suspicion that a child has been harmed, is being harmed, or is at risk of being harmed. In the event that this occurs, a Tusla referral will be completed.
- Data Protection Impact Assessment has been carried out and approved by the data protection officer in TCD.

Training in data protection law and practice has been provided to myself and my supervisors.

What is the lawful basis to use my personal data?

- By law, we can use your personal information for scientific research (in the public interest). We will also ask for your explicit consent to use your data as a requirement of the Irish Health Research Regulations.

What are my rights?

You are entitled to:

- The right to access to your data and receive a copy of it
- The right to restrict or object to processing of your data
- The right to object to any further processing of the information we hold about you (except where it is de-identified)
- The right to have inaccurate information about you corrected or deleted
- The right to receive your data in a portable format and to have it transferred to another data controller
- The right to request deletion of your data

Part 3 – Costs, Funding and Approval

Has this study been approved by a research ethics committee?

- Yes, this study has been approved by Trinity college Dublin Research Ethics Committee. Approval was granted on (8th of August 2022)

Who is organising and funding this study? Will the results be used for commercial purposes?

- I am doing this study as part of my PHD in Trinity College Dublin. This is not being funded for commercial purposes.

Will my personal data be used in future studies?

- Your data will not be used for further studies

Part 4 – Further Information

Who should I contact for information or complaints?

If you have any concerns or questions, you can contact:

- Principal Investigator: Isobel Mahon (ismahon@tcd.ie)
- Data Protection Officer, Trinity College Dublin: Data Protection Officer, Secretary's Office, Trinity College Dublin, Dublin 2, Ireland. Email: dataprotection@tcd.ie. Website: www.tcd.ie/privacy.
- Under GDPR, if you are not satisfied with how your data is being processed, you have the right to lodge a complaint with the Office of the Data Protection Commission, 21 Fitzwilliam Square South, Dublin 2, Ireland. Website: www.dataprotection.ie.

Will I be contacted again?

If you would like to take part in this study, you will be asked to sign the Consent Form either online or in person.

Support Links

- Link to HSE support for children with disabilities: <https://www2.hse.ie/services/disability-support-and-services/if-you-have-just-found-out-your-child-has-a-disability/useful-childhood-disability-websites.html>
- Rare Diseases Ireland: <https://rdi.ie/>
- Unique: <https://rarechromo.org/>

Appendix 9: Informed consent form (in person)

Please indicate your informed consent to take part in this research study by completing this form. Please ask any questions you may have when reading each of the statements.

Thank you for participating. Please feel free to ask questions if there is something you do not understand. I ask that you read each statement and place your initials in the appropriate box.

Title: Fathers experiences of caring for their child with and complex care needs and a chromosome abnormality.

Statement	Initials
I confirm I have read and understood the Information Leaflet for this study. The information has been fully explained to me and I have been able to ask questions, all of which have been answered to my satisfaction.	
I understand that the interview will be audio recorded.	
I understand that if I keep a written account in the journal and give this to the researcher, I am willing to share this Journal. I understand that once this is given to the researcher, this will be anonymised and then destroyed.	
I understand that this study is entirely voluntary, and if I decide that I do not want to take part, I can stop taking part in this study at any time without giving a reason, and without sanction up until the point of data analysis.	
I understand that the researcher will collect data which will be anonymised and I agree that this data can be used for scientific purposes without identifying me.	
I understand that my contact information will be retained until such a time as all findings are published in peer reviewed journals. Study data will be retained for a maximum of 7 years after which time it will be destroyed as per the General Data Protection Regulations, 2018.	
I understand that all data collected will be managed and processed according to European and international law and best practice.	
I understand that I will not be paid for taking part in this study.	
I understand that there is the potential for unexpected findings and that if this occurs the research team will examine the significance of these findings and will report them based on local legal and ethical frameworks.	
I know how to contact the researcher if I need to.	
I am over 18 years of age, and I am competent to supply consent.	
I agree to take part in this research study having been fully informed of the risks, benefits and alternatives which are set out in full in the information leaflet which I have been provided with.	
I agree to being contacted by researchers by email/phone as part of this research study.	

I freely and voluntarily consent to be a part of this research study, though without prejudice to my legal and ethical rights.	
I understand that personal information about me will be protected in accordance with the General Data Protection Regulation.	
I have been asked if I would like a copy of the interview transcript.	
I understand that there are no direct benefits to me from participating in this study. I understand that results from analysis of my personal information will not be given to me.	
I understand that Isobel is a mandated person, under the Children First Act 2015, she is required by Law to report any knowledge, belief or reasonable suspicion that a child has been harmed, is being harmed, or is at risk of being harmed. In the event that this occurs, a Tusla referral will be completed.	
If there is a concern for my welfare, I consent to the researcher (Isobel Mahon) advising me to make contact with a relevant professional (i.e. key worker, liaison nurse, GP, pharmacist or relevant other).	

PARTICIPANT'S NAME: _____

CONTACT DETAILS: _____

PARTICIPANT'S SIGNATURE: _____

DATE: _____

I have explained the nature and purpose of this research study, the procedures to be undertaken and any risks that may be involved. I have offered to answer any questions and fully answered such questions. I believe that the participant understands my explanation and has freely given informed consent.

RESEARCHER'S SIGNATURE: _____ Date: _____

Appendix 10: Informed consent form (Zoom)

Instruction. Please indicate your informed consent to take part in this research study by completing this form.

Please ask any questions you may have when reading each of the statements. Thank you for participating. Please feel free to ask questions if there is something you do not understand.

Q1. Name:

Q2. Email:

Q3: I confirm I have read and understood the Information Leaflet for this study. The information has been fully explained to me and I have been able to ask questions, all of which have been answered to my satisfaction.

☒ I consent, begin the study

Q4: I understand that the interview will be audio tape recorded. I understand that if I keep a written account in the journal and give this to the researcher, I am willing to share this diary.

☒ I consent, begin the study

Q5: I understand that this study is entirely voluntary, and if I decide that I do not want to take part, I can stop taking part in this study at any time without giving a reason, and without sanction.

☒ I consent, begin the study

Q6: I understand that the researcher will collect data which will be anonymised and I agree that this data can be used for scientific purposes without identifying me.

☒ I consent, begin the study

Q7: I understand that all data collected will be managed and processed according to European and international law and best practice.

☒ I consent, begin the study

Q8: I understand that I will not be paid for taking part in this study. I understand that there are no direct benefits to me from participating in this study. I understand that results from analysis of my personal information will not be given to me.

☒ I consent, begin the study

Q9: I understand that there is the potential for unexpected findings and that if this occurs the research team will examine the significance of these findings and will report them based on local legal and ethical frameworks.

☒ I consent, begin the study

Q10: I understand that if I disclose any information that suggests unsafe practice or gives reasonable ground for concern I will contact a relevant person to seek advice/support or with my permission the researcher will make contact with a relevant person.

☒ I consent, begin the study

Q11: I agree to being contacted by researchers by email/phone as part of this research. I know how to contact the researcher if I need to.

☒ I consent, begin the study

Q12: I freely and voluntarily consent to be a part of this research study, though without prejudice to my legal and ethical rights.

I am over 18 years of age and I am competent to supply consent

☒ I consent, begin the study

Q13: I agree to take part in this research study having been fully informed of the risks, benefits and alternatives which are set out in full in the information leaflet which I have been provided with.

☒ I consent, begin the study

Q14: I understand that personal information about me will be protected in accordance with the General Data Protection Regulation.

☒ I consent, begin the study

Q15: I understand that there are no direct benefits to me from participating in this study. I understand that results from analysis of my personal information will not be given to me.

☒ I consent, begin the study

Q16: I understand that Isobel is a mandated person, under the Children First Act 2015, she is required by Law to report any knowledge, belief or reasonable suspicion that a child has been harmed, is being harmed, or is at risk of being harmed. In the event that this occurs, a Tusla referral will be completed.

☒ I consent, begin the study

Q17: If there is a concern for my welfare, I consent to the researcher (Isobel Mahon) advising me to make contact with a relevant professional (i.e. key worker, liaison nurse, GP, pharmacist or relevant other).

☒ I consent, begin the study

Appendix 11: Sample interview guide

Project Title: Fathers' experiences of caring for their child with a chromosome abnormality and complex care needs.

Introduction: Discuss participation, sign consent forms, opening conversation.

Question samples:

Please can you tell me about your experience of caring for a child with a chromosome abnormality and CCN.

What was it like when your child was diagnosed with a chromosome abnormality?

Probing and Interpreting Questions

Can you elaborate on what you mean by ...?

Could you give more detail on that?

How did this make you feel..?

Do you have any examples of that?

Do you mean that...?

Is it correct that...?

How does that support you...?

Appendix 12: Sample of the journal

Journal

How do I complete this Journal?

This journal is to help me understand what it is like to care for your child with complex care needs and a chromosome abnormality. This is optional, and you can choose what/when to write in it.

- Do not worry about what you write it; you are free to write whatever you wish.
- Please write an entry number as this will help me to determine how many entries are in the journal. You do not need to write the date.
- If you mention names of persons or places these will be removed, and no identifiers will remain.

In about 8 weeks, I will get back in contact with you and can arrange to have the Journal collected/posted with a sampled envelope.

|

to [name of regional hospital] for and xray (chest). Got to [name of regional hospital] for 2.40pm. results came back with nothing. Got back at 5pm and have to give Toby C-PAP and Calpol.

- (2) Collected Toby from [mothers] house today and dropped him to school with [name of homecare nurse]. I got a call at 11am to say that Toby had spiked a temperature so I have to collect him from school. This means that I have to cancel the rest of my shifts for today and tomorrow!!! [name of work]. I collected him at 11.30am and took him home. He is in ok form and his temp is only 37.2! so not a high temperature and im down two days work!
- (3) Today we have been on a funday with [name of association] about 15 members and their families turned up...[siblings names] also came along. Because Toby is the only one with a tracheostomy and in a wheelchair, all the other kids pay him loads of attention which he loves, but then he gets frustrated when he sees them running around and he cant join in. this makes me sad I must admit.
- (4) Its just another day of fighting the system today. Ive spoken to disability services (or lack there of) about finding out why we have NO speech and language therapy, NO physio, NO play therapy and NO music therapy for at least 18 months in [name of CDNT]. I understand that asking for music therapy is a bit of a push, but SLT and pysio is a necessity! I am trying my best to do physio, but seem to have just got to a certain level and cant go anymore! I used to ger so frustrated working in [name of company] but this is on another level.
- (5) If I hear one more song by the wiggles I am going to lose my mind!! Today I have to find some new TV grogram's to get Toby interested in.
- (6) What was supposed to be a nice relaxing day at home today ended up being the complete opposite! This morning at 5.30am the nurse noticed that PEJ line was leaking internally...I checked the balloon which was intact but there seems to be an infection in the line, contacted [CHI] radiology at 9.30am to let them know that we would be coming up. When we got there [name of doctor] who is the only doctor in the hospital that is trained to put on the PEJ line is on holidays

today- MARVELOUS! We waited for a very long 2hours to find that [name of doctor] came in to do the procedure in a dinner gown. It goes to show that if your good to your doctors and nurses, they will be good to you. What a hero. Eventually managed to get home for 7.30pm.

Appendix 13: Letter of ethical approval



Coláiste na Tríonóide, Baile Átha Cliath
Trinity College Dublin
Ollscoil Átha Cliath | The University of Dublin

Isobel Mahon
School of Nursing and Midwifery
Trinity College Dublin
24 D'Olier Street
Dublin 2

8th August 2022

COM_14_21.22

Study title: Fathers' experiences of caring for their child with complex care needs and a chromosome abnormality

Dear Isobel,

Your application for ethical approval has been approved.

Yours sincerely,

A handwritten signature in blue ink, appearing to read 'S. O'Donnell', with a long horizontal flourish underneath.

Prof Sharon O'Donnell
Chair of SNMREC

Scoil an Altranais agus an Chnámhseachais
Dámh na nEolaiochtáil Sláinte,
Coláiste na Tríonóide, Baile Átha Cliath,
Ollscoil Átha Cliath,
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Appendix 14: The complex care needs of the children involved in this study

*Participant number/pseudonym not identified to ensure confidentiality is maintained

Chromosome abnormality and associated syndrome	Complex care needs identified by fathers
Williams Syndrome	• Intellectual disability • Challenging behaviors • Cardiac abnormalities • Multiple medical appointments • Challenges with change and transitions • Anxiety • Sleep challenges requiring medications
Trisomy 21 Down syndrome	• Intellectual disability • Tracheostomy requiring: Nebulizers, suctioning to maintain a safe airway • Ventilation (C-PAP) • PEG feeding • Wheelchair user • Polypharmacy • Hypotonia • Communication difficulties • leukaemia
Trisomy 21 Down syndrome	• Stroke causing an acquired brain injury and quadriplegia • PEG feeding • Polypharmacy • Sleep challenges • Increased pain • Altered breathing and circulation • Communication difficulties
Williams syndrome	• Intellectual disability • Scoliosis requiring physiotherapy • Anxiety • Premature puberty

	• Constipation Complex social and emotional needs
Prader-Willi syndrome	• Intellectual disability • Behaviours • Anxiety • Hormone injections • Needed NG feeding in infancy • Complex social and emotional needs
Trisomy 13: Patau syndrome	• Profound intellectual disability • PEG feeding • Requires suctioning • Requires assistance with all activities of daily living • Epilepsy • Sleep challenges • Requires specialised seating and sleep systems • Regular chest infections • Palliative care needs • Wheelchair user • Polypharmacy
1.1mb missing on 14q.33 and slight loss on 22qxb22.33.12 and 12q22.1). No named syndrome	• Intellectual disability • Wheelchair user • Hypotonia • PED feeding • Previous use of suction machine • Febrile convulsions • Lagophthalmos • Polypharmacy
Turner syndrome	• Complex heart condition (hyperplastic ventricular) • Intellectual disability (mild) • Oxygen therapy • Hourly observations • 3 open heart surgeries • 3 cardiac arrests in 8 years • Polypharmacy

Appendix 15: Government supports for families of children with complex care needs

Child Benefit	Domiciliary care allowance (DCA)	Carers Benefit	Carer's Allowance	Respite Care Grant
€140 per month	€360 per month	€625 per week	€260 per week	€2,000 annually
This is a payment to the parents of children under 18-years of age	Eligibility is not based on the type of disability Not Means Test payment	A parent who gives up work to look after their child with CCN can receive this payment. It is not Means Tested but is based on PRSI classification and contributions. A parent can get Carer's Benefit for 2 years (104 weeks) for each child they care for	This is a Means Tested payment for those who are looking after a child with additional needs.	The Respite Care Grant is a tax-free payment made in June of each year for each child. This is not a Means Tested payment or a taxable source of income

Incapacitated child tax credit	Home Carer's Tax Credit	Homemaker's Scheme	Disability Passenger Scheme	Medical Expenses	Local Property Tax	VAT Relief
Tax credit claimed on Revenue for working parents of children with CCN	Tax credit claimed on revenue for people who care for one, or more, dependent persons.	The Homemaker's Scheme can help people qualify for the state Pension for people who care full-time for: A child under age 12.	The Disabled Drivers and Disabled Passengers Scheme provides a range of tax reliefs linked to the purchase and use of specially constructed or adapted vehicles.	Income tax back on some types of healthcare expenses	Properties purchased, adapted or built for use by incapacitated person may be exempt from Local Property Tax (LPT).	Refund of Value-Added Tax (VAT) on certain aids and appliances

