Dental Features of an Irish Patient Cohort with Ectodermal Dysplasia and Resultant Impact on Their Quality of Life

A Mixed Methods Study

Submitted in accordance with the requirements for the degree of Clinical Doctorate in Dental Surgery (Prosthodontics)

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Declaration

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______________________________
Shkre Abdalla Ali Agkre
Summary

**Aims:** This study involved two principal aims. Firstly, to investigate and explore children’s’ and parents’ perception of life with Ectodermal Dysplasia (ED) and resultant impacts on Oral Health Related Quality of Life (OHRQoL). Secondly to record the thoughts of a number of ED patients who have undergone dental treatment as adolescents to obtain their insights into the adolescent phase of living with ED.

**Methods:** The quantitative section involved (N=22) children and their parents and a matched age and gender control group who were asked to complete identical questionnaires. All children and parents were asked to fill the short forms of the Child Perception Questionnaire (CPQ8-10), (CPQ11-14), and the Parental-Caregiver Perception Questionnaire (PPQ). Clinical assessments and examinations for all participants were completed by the main researcher.

The descriptive qualitative study involved a structured focus group discussion of a sample of adults affected by Ectodermal Dysplasia who had undergone dental rehabilitations. A topic guide was formulated by experienced clinicians and researchers to cover all related aspects of the condition in a series of open-ended questions. A cross sectional thematic analysis approach was followed in data analysis using MAXQDA software.

**Results:** In the final sample, (72.7%, N= 16) of the children were male in gender and the majority (86.3%, N= 19) were Caucasian in ethnicity. The mean age of children was 12.5 years. The control group had the same demographic data in terms of age and gender apart from the fact that they were all Caucasian. A total of 22 pairs of children and parents and matched controls completed all questionnaires for the age groups, 8-10 and 11-14 years. A Wilcoxon rank test was used to compare the values between participants within each
group \((P < 0.05)\). Children with Ectodermal Dysplasia CPQ total scores were higher than their controls for both age groups, which reflected worse (OHRQoL) \((CPQ = 40.1, P = .078,\) and \(CPQ = 75.3, P = .036)\) respectively. A statistically significant difference was also identified for the oral symptoms, functional limitation and emotional well-being domain scores in the older age group \((11-14\text{ years})\).

Parents of the children with ED reported higher scores than parents of healthy children. In addition, the younger ED group parents reported higher scores than their children \((8-10\text{ years})\) in the oral symptoms and functional limitation domains and the differences were statistically significant \((P = .04\) and \(P = .04\) respectively). In the older age group \((11-14\text{ years})\) the children reported higher scores than their parents, with no statistical difference observed between groups. A Spearman’s correlation test reported a weak to very weak relationship between childrens’ and parents’ perceptions for both age groups, with no statistically significant correlation reported in any domain.

The qualitative study analysis identified four main themes; the meaning of health, challenges of living with ED, impact of ED on Quality of Life (QoL) and managing the impacts of ED. Participants highlighted the life experience of living with ED from childhood to adulthood with emphasis on the emotional and psychosocial challenges experienced by them. Challenges from dental, dermatological and ophthalmological features of the condition had been reported, with greatest impacts reported from the dental features of the condition.

All contributors described the requirement for adaptation to the features of the condition by becoming “experts in self” in order to overcome challenges posed in the absence of formal medical guidance and peer support. The impact of their dental features and subsequent management were the most important aspects for all participants because of
their impact of function, appearance and self-esteem. Management of the dental challenges resulted in decreased levels of anxiety and improved their mental health. Long-term multidisciplinary treatment was deemed as being fundamental in the future management of dental, dermatological and ophthalmological features of the condition.

**Conclusions:** Ectodermal Dysplasia had a profound influence in the oral health-related quality of life (OHRQoL) of children and their families. Children as they developed reported higher scores than their parents, however, limited agreement was observed between childrens’ and parents’ perceptions in OHRQoL.

In addition, ED features had social, emotional, and physical impacts on this population from childhood to adulthood, and adaptation on a number of levels was required. Their dental features had the greatest overall impact on this cohort and it is management by healthcare professionals greatly improved their outlook. This was in contrast to other classic features of ED which were managed in a less cohesive manner.
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1 Literature Review

1.1 Ectodermal Dysplasia

1.1.1 Definition

Ectodermal Dysplasias (EDs) were defined by Freire-Maia as “congenital disorders characterised by alterations in two or more ectodermal structures, at least involving one in hair, teeth, nails, or sweat glands” (Freire-Maia, 1971, Freire-Maia, 1977). This definition was based on the four main classical signs of Ectodermal Dysplasia: trichodysplasia (abnormal hair), onychodysplasia (abnormal nails), dental defects, and dyshydrosis (abnormal perspiration / sweating) (Salinas et al., 2009). Proposals for a new consensus definition and classification of Ectodermal Dysplasias in 2008 concluded that the definition proposed by Freire-Maia was an appropriate definition, due to its clear execution of the condition which led to clear group and subgroup classifications (Visinoni et al., 2009).

1.1.2 Classification

As with any medical condition, the classification of different types of Ectodermal Dysplasia is important because it helps in the understanding of the condition and provides a method of communication with other medical professionals and families. In addition, it assists patients and their families in making well-informed decisions regarding prognosis, required treatment, services, and future planning (Salinas et al., 2009). Ectodermal Dysplasia classification is based on the clinical signs of the condition and identified by their number or Greek words initiators such as, “tricho-odonto-onycho-dyshidrotic”. Group A includes conditions with signs that affect at least two out of four classical structures of the condition: (1) hair (trichodysplasia), (2) dental defects, (3) nails (onychodysplasia), and (4) sweat
glands (dyshidrosis). Group B contains disorders involving one of the classical signs with another ectodermal defect (Freire-Maia, 1977).

Group A has eleven subdivisions related to the involved ectodermal structure; they are identified by their number combination or combination of Greek words initiators as mentioned in the first subgroup:

a) 1-2-3-4 (hair-teeth-nails-sweat gland) (tricho-odontononycho-dyshydrotic) subgroup

b) 1-2-3 (hair-teeth-nails) subgroup
c) 1-2-4 (hair-teeth-sweat glands) subgroup
d) 1-3-4 (hair-nails-sweat glands) subgroup
e) 2-3-4 (teeth-nails-sweat glands) subgroup
f) 1-2 (hair-teeth) subgroup
g) 1-3 (hair-nails) subgroup
h) 1-4 (hair-sweat glands) subgroup
i) 2-3 (teeth-nails) subgroup
j) 2-4 (teeth-sweat glands) subgroup
k) 3-4 (nails-sweat glands) subgroup (Freire-Maia, 1977)

Group B is classified using the same criteria as Group A, but number 5 is added to indicate an additional ectodermal tissue/organ defect. Examples of the ectodermal structures included are the mammary glands, adrenal medulla, thymus, anterior pituitary, thyroid gland, external ear, cornea, conjunctiva, lacrimal gland and lacrimal duct. Group B subgroup examples include 1-5, 2-5, 2-5, and 4-5 (Freire-Maia, 1977, Visinoni et al., 2009).

Another classification of Ectodermal Dysplasia is based on the molecular features of each type. This classification was described by Manuela Priolo and Peter Itin. They proposed a classification of Ectodermal Dysplasia on the basis of the causative genes which act through
two different pathogenetic mechanisms, and the observed clinical findings of the condition in affected patients are representative of either mechanism. Group 1 is characterised by defects “in the epithelial-mesenchymal interaction, with localisation of genes in the nucleus which are involved in differentiation and apoptosis” (Visinoni et al., 2009, Priolo et al., 2000, Priolo and Laganà, 2001). In this group, the affected individual has pure ED features, as ectodermal derivatives are affected, and major skeletal elements are involved. In Group 2 the disorders are categorised by defects in proteins of the ectodermal structure. “The genes involved in this group encode proteins which are localised and highly specialised in the plasma membrane domains and cytoplasm and are involved in the maintenance of the integrity and stability of cell membranes and underlying cytoskeleton”. From a clinical perspective, this group includes dermatologic ED where ectoderm and highly differentiated epithelium are involved. As a result of the involvement of primary ectodermal structures only, this classification is simple and has the advantage of being objective, in terms of the ED pathogenesis on a molecular basis, which contributes to the required knowledge for future molecular-based treatment (Salinas et al., 2009, Visinoni et al., 2009).

However, molecular information is not available for the majority of ED types, and clinical professionals (paediatricians, dermatologists, ophthalmologists, dentists, physiotherapists, and other professionals) who are involved in ED diagnosis and providing suitable therapy would benefit from a clear, convenient method to facilitate clinical diagnosis (Salinas et al., 2009, Freire-Maia, 1977).

The 2008 International Conference on Ectodermal Dysplasia classification reported that an accepted international classification of this condition should consider both molecular and clinical knowledge of Ectodermal Dysplasia and would:
1. “Support the accurate diagnosis of individuals with ED, offering patients an ‘answer’
to explain their condition and a greater understanding of its genetic implications, as
well as giving providers a path for counselling, testing, and treatment;
2. Move research forward, in particular, the overlay of molecular studies and clinical
knowledge may result in genotype-phenotype correlations and the discovery of
new mutations;
3. Lead to cures and hope for the future for patients and families;
4. Provide a common language around EDs for patients, families, researchers, and
different types of health-care providers, ultimately enhancing communication
among these diverse groups;
5. Help advocacy groups better serve their constituents and promote the effective use
of limited resources” (Salinas et al., 2009).

1.1.3 Hypohydrotic Ectodermal Dysplasia

There are almost 200 different types of ED and causative genes have been identified for 30
different types (Priolo et al., 2001; Clarke, 1987). The most common type of ED is the
hypohydrotic form which can be inherited as X-linked autosomal recessive or autosomal
dominant. The other names for this type are Anhydrotic Ectodermal Dysplasia and Christ-
Siemens-Touraine (CST) Syndrome. X-linked hypohydrotic ED was first described by
Thurnam in 1848 and 1875, Darwin described kindred men who had the same condition
(Irvine, 2009, Lexner et al., 2007a). In the early stages of the condition, the presence of a
collodion membrane in babies and scaling skin in children with hypohydrotic ED was
observed and these were considered clues to early diagnosis of the condition (Plantin et
al., 1992, Roselyn Anderson, 1989). In addition, the sudden death of infants with
hypohydrotic ED has been reported. This was linked to a lack of body temperature regulation due to decreased sweating ability (Salisbury and Stothers, 1981, Bernstein and Weakley-Jones, 1987, Bergendal, 2010).

Genetically, the pathogenesis of hypohydrotic ED at a molecular level has not been fully elucidated. A mutation in the Xq12–Xq13.1 region of an Ectodysplasin A (EDA) gene, which encodes for a transmembrane protein named ectodysplasin, results in X-linked hypohydrotic ED. The EDA (Xq13.1) mutation results in abnormal ectodysplasin-A production, which is important for ectodermal structural development for teeth, hair, and sweat glands. Evidence is accumulating that ectodermal-mesodermal interactions during embryogenesis require ectodysplasin-A, and that defects in its molecular structure might prevent the function of enzymes responsible for the development of ectoderm and/or its interaction with the underlying mesoderm (Bergendal, 2010, Wright et al., 2017, Mikkola, 2009). Another study reported that both autosomal dominant and recessive hypohydrotic ED are attributed to EDA signal receptor mutations in the signalling cascade which in turn, is responsible for ectodermal derivative development (Bergendal, 2010, Mikkola, 2009).

1.1.4 Incidence and Prevalence

Even though it is not specifically recorded, it is estimated to occur in 1 in 20,000 newborns worldwide, with a prediction that 5000-10,000 of newborns have hypohydrotic ED in the USA (GeneticsHomeReference, 2018). In addition, Clarke and co-workers reported that hypohydrotic ED birth frequency was 1:100,000, while Carter et al. in 1977 estimated the same frequency by presenting a range of 0.01 to 0.1 per 1000 (Stevenson et al., 1967, Carter, 1977, Wright et al., 2017, Clarke, 1987). On the other hand, a 1-7:10,000 occurrence
rate for hypohydrotic ED has been reported in an encyclopaedia on birth defects (Christine, 1992).

A Danish cross-sectional study aimed to estimate the prevalence of X-linked hypohydrotic ED in the Danish population before January 1, 2011 and define the common age and time of diagnosis. They reported on 90 X-linked hypohydrotic ED cases, 146 cases of clinically diagnosed hypohydrotic ED, and 988 cases that had the possibility of being hypohydrotic ED in the period between 1996 and 2010. This study reported an overall hypohydrotic ED prevalence of 21.9 per 100,000 and a genetically confirmed hypohydrotic ED prevalence of 1.6 per 100,000 (Nguyen-Nielsen et al., 2013).

The database of the US National Foundation for Ectodermal Dysplasia (NFED) had registered over 5,200 subjects with ED in the United States and 70 other countries. Hypohydrotic ED was reported for more than 1,900 (36.5%) of those individuals, while more than a half had no specific clinical diagnosis. In addition, only one-third of the registered subjects had a genetically defined diagnosis (Salinas et al., 2009, Bergendal, 2010). Following on from that report and to assist future national and international estimates of ED incidence, the NFED launched the Ectodermal Dysplasia International Registry (EDIR) in 2010. This database assists in capturing medical histories relevant to ED and recognising signs and symptoms that provide health-care professionals with information for ED diagnosis. From March 2010 to May 2011 information was collected from 835 Ectodermal Dysplasia patients; for this 429 (51%) of the subjects reported themselves as having hypohydrotic ED (170 females and 259 males) (Figure 1.1). Additional evaluation of the 429 subjects yielded 223 subjects who were self-identified as X-linked hypohydrotic ED, 82 females and 141 males (Figure 1.2). The majority of the registered subjects (71.3%) (159) were from the United States, with 4.93% (11) of the subjects resident in Ireland. Demographically, the mean age of the female subjects (82) was 32.1 years old,
and 99% of them had a positive family history of the condition. The mean age of male subjects (141) was 17.8 years old and 95% of them had a positive family history (Fete et al., 2014).

**Figure 1.1 Numbers for Ectodermal Dysplasia Registrants (2014)**

**Figure 1.2 Numbers for X-linked Ectodermal Dysplasia (XLHED) Registrants (2014)**
1.1.5 Clinical Features of Hypohydrotic Ectodermal Dysplasia

The clinical features of Hypohydrotic Ectodermal Dysplasia (HED) are representative of the affected ectodermal structures: teeth, hair, nails, and sweat glands. Individuals with this condition have similar distinguishing features which include: frontal bossing, a saddle nose, maxillary hypoplasia, and hyperkeratotic wrinkles around the eyes. Other common signs and symptoms include: “dry skin and eczema, nasal crusting, and thick ear wax”, particularly in small children (Mikkola, 2009, Bergendal, 2010). Wright et al. in a 2017 review provided further details about features related to Hypohydrotic ED (HED) in neonates including peeling skin and periorbital hyperpigmentation that might aid in diagnosis. In addition, during infancy, there is a possibility of the foetus being irritable as a result of elevated body temperature and subsequent heat intolerance (Wright et al., 2017).

Salisbury and Stothers reported that impaired body temperature control might result in hyperpyrexia and death (Salisbury and Stothers, 1981, Clarke, 1987). From a dental diagnostic perspective, Wright and co-workers stated that it was common for the eruption of teeth to be very delayed or failed or that malformed teeth may erupt at 6-9 months, which may be associated with eczema and wrinkled peri-orbital skin (Wright et al., 2017). Many young boys with X-linked hypohydrotic ED (XLHED) receive their diagnosis after a dental examination when no teeth erupt before 12–18 months of age, or when the first tooth appears late with an atypical form in an atypical place (Bergendal, 2014).

The features of HED are more defined in childhood where more signs and symptoms of hypotrichosis are distinct, the scalp skin is lightly pigmented with the thin and slowly growing scalp hair, which might result in excessive fragility of the shafts that break down easily with usual childhood wear and tear (Wright et al., 2017). In addition, skin and hair changes for all age groups were reported by Itin et al., where alopecia was considered as a feature of ED, and when the ED subject had hair it was reported to be fair, scanty, brittle.
and un-combable, while body hair maybe diminished (Itin et al., 1993, Itin and Fistarol, 2004).

During early childhood sweat gland functional impairment will become more obvious with subsequent hyperthermia that requires individuals and their families to apply environmental modifications in order to facilitate temperature control. A one-year cross-sectional survey study of parents of 100 children who had ectodermal dysplasia revealed that HED was the diagnosis for more than half (57%) (34 out of 60 cases) of the children whose parents returned the survey (63%, 57 out 91 of the total sample) (Blüschke et al., 2010). Seventeen infants achieved temperature control by being placed in an incubator after birth when temperature control was an issue. The majority of children 97.1% (33 out of 34 cases) with HED had unexpected episodes of fevers during their first year, and febrile seizures occurred in 5.9% (2 out of 34 cases) of cases. This one-year survey reported a 2.1% mortality rate in the first year of life for children with HED (Schneider et al., 2011, Blüschke et al., 2010).

In addition to the classical signs of Ectodermal Dysplasia, other embryonic ectoderm-derived structures may also be affected, including the thyroid gland, mammary gland, thymus, cornea, lacrimal gland, conjunctiva, lacrimal duct, and Meibomian gland (Kaercher and Ophthalmology, 2004). Common otolaryngeal signs and symptoms of ED are evident clinically but poorly reported in the dental literature. One case report highlighted a hypohydrotic ED patient who presented with both pneumonia and dysphagia which suggested laryngeal incompetence as the underlying cause of recurrent chest infections (Potter and Bowie, 1984). The dental community may not be familiar enough with other general medical issues that may affect this patient cohort. Another study that investigated otolaryngeal symptoms in 75 subjects who had a diagnosis or family history of ED found
that over half (51%) reported obstruction of the nose (37%) or had sinusitis (49%) and crusting respectively (Mehta et al., 2007, Bergendal, 2010).

An autopsy finding in an ED patient reported the absence of mucous glands in the pharynx, larynx, trachea, and oesophagus, while in the colon those mucous glands were hypoplastic (Reed et al., 1970). Reduction in aerodigestive tract mucous production was also reported elsewhere which resulted in chronic upper respiratory tract infections, otitis, dysphagia, hoarseness, bronchitis, and sometimes haemoptysis (Bergendal, 2010, Siegel and Potsic, 1990).

Ocular signs and symptoms were not considered as diagnostic inclusion criteria for HED, but they had been reported. Ophthalmological investigations of subjects with a confirmed diagnosis of ED revealed dry eye symptoms and volume reduction of eyebrows and lashes in 94% of patients. In addition, meibography demonstrated changes in Meibomian glands which are considered a sign of ED. The authors reported that this test can be performed at any age to confirm an ED diagnosis, particularly in children. Subjects with ED in older age groups experienced corneal changes with loss of visual acuity which may lead to amaurosis (Salinas et al., 2009, Dietz et al., 2013). Furthermore, Rodriguez and co-workers stated that cataracts, strabismus, and reduced tear production might occur in addition to bilateral panuveitis in children with HED (Rodriguez et al., 2002, Itin and Fistarol, 2004).

1.1.6 Dentofacial Signs and Symptoms of Ectodermal Dysplasia

Ectodermal Dysplasia is generally associated with dentofacial defects, and in the scope of this thesis, dentofacial defects are associated with the most common type of Ectodermal Dysplasia type (Hypohydrotic ED) will be discussed. These defects include craniofacial growth, hypodontia, anodontia, and malformed teeth with cone or peg-shaped crowns. In
addition, affected teeth are more susceptible to dental caries as a result of their malformed enamel and possible xerostomia (Bergendal, 2010).

The impact of hypohydrotic ED on craniofacial growth has been reported. A longitudinal study investigated the craniofacial growth of 61 individuals with ED, with a participant mean age of 133 months with a mean of 15.4 missing permanent teeth (severe hypodontia). Growth was observed incorporating a multilevel modelling technique using the MLwiN software with lateral cephalograms recorded at each visit. The visit occasions ranged from one to five visits with a mean of 2.66 cephalograms over periods of 0-252 months. The study continued until the mean participant age was 207 months. Trends in craniofacial growth were reported, and the most significant findings were changes in the sagittal relationship of the maxilla and mandible which resulted in class III skeletal relationships, in addition to increased rotational growth anteriorly as a consequence of growth discrepancies between the anterior and posterior face heights (Bondarets et al., 2002). Furthermore, at the National Foundation of Ectodermal Dysplasia (NFED) 2006 conference, growth rates were evaluated using 3D facial morphology analysis of individuals with Ectodermal Dysplasia from 37 families and compared to unaffected individuals (controls). The study reported that growth was not affected up to the age of five years and after that ED-affected boys had slower growth tendencies. This growth discrepancy should be considered to determine the appropriate age for intervention and selection of appropriate prosthetic treatments to restore missing teeth. There were no reported growth discrepancies observed from face-scanning of girls in that study (Peter Hammond, 2008, Bergendal, 2010). These facial analysis study findings were consistent with previous work by Goodwin who compared male facial morphology in 23 hypohydrotic ED patients (age range 5-29 years), with 59 controls (age range 4-31 years). In that study facial analysis was performed using the “3D Capturor II” camera system (InSpeck, Montreal, Canada) and
they reported that ED patients had smaller and shorter faces, proportionally longer chins and midface, midface hypoplasia, protruded mandibles, narrow mouths, and narrow pointed noses when compared to controls (Goodwin et al., 2014).

Another dentofacial finding that has been reported was a discrepancy in salivary flow rates between subjects with Hypohidrotic Ectodermal Dysplasia (HED) and non-affected subjects. This has been proposed as a possible future diagnostic tool for HED. Lexner and co-workers evaluated saliva flow in 11 affected males and 28 carrier females respectively against 15 controls from each gender. The test subjects had a confirmed molecular genetic diagnosis of HED with a 100% prevalence of hypodontia in affected males and 75% in female carriers. All participants were asked to refrain from drinking and eating one hour before saliva collection, which was collected using a draining method. All males and females in the test group reported daily mouth dryness in contrast to the control group. Saliva flow rates were reduced in both affected males and female carriers in contrast to the control group. Affected male mean saliva flow was almost a quarter (0.09 ml/min) of the control mean rate (0.34 ml/min) \( (P < 0.001) \), while female carrier saliva flow mean rate was about a half (0.14 ml/min) of the control female rate (0.34 ml/min) \( (P < 0.001) \) (Lexner et al., 2007b, Nordgarden et al., 2003).

Bergendal, in 2010, reported similar findings when saliva flow was evaluated for 116 individuals with hypodontia from 123 invited participants (71 females (57.7%) and 52 males (42.3%)). The saliva flow rate was measured twice. Unstimulated saliva was collected for 15 minutes while chewing stimulated saliva was collected for 5 minutes. The cut-off for low saliva secretion was 0.1 ml/min for unstimulated saliva and 0.7 ml/min for simulated saliva. Low saliva secretion was reported for one third (30.2%; 35/116) of the subjects (Bergendal, 2010).
1.2 Hypodontia and Ectodermal Dysplasia Subjects

1.2.1 Definition

Tooth agenesis had been defined as the developmental absence of teeth. Tooth agenesis is one of the most frequent human congenital dental anomalies (Kirac et al., 2016, Ritwik and Patterson, 2018b). It has three different clinical and congenital presentations: hypodontia, oligodontia and anodontia. Third molar teeth are excluded from this classification. Hypodontia is a congenital absence of five or fewer teeth. Oligodontia is a congenital absence of six or more teeth, while anodontia is the congenital absence of all teeth. Also, hypodontia severity had been classified according to the number of missing teeth, mild (with 1 or 2 missing teeth), moderate (with 3-5 missing teeth) and severe (with 6 or more missing teeth). Hypodontia had also been classified as isolated or syndromic depending on the presence of associated systemic conditions such as Ectodermal Dysplasia (Rakhshan, 2015, Al Shehrani and Al Qamn, 2013). Hypodontia prevalence among different populations had been reported. Prior reports showed that the prevalence of hypodontia among Mexicans was 2.7% and in a Turkish population was 4.3%-7.54% (Celikoglu et al., 2010, Silva Meza, 2003, Sisman et al., 2007). Meanwhile, the prevalence of hypodontia was in an Indian population was reported as 4.19% (Gupta et al., 2011), 4.5% in Norwegians (Nordgarden et al., 2002), a higher percentage (5.21%) among Iranians (Amini et al., 2012), and 6.3% in the Brazilian population (Gomes et al., 2009). The prevalence was almost one in ten in Japanese and Korean populations (Endo et al., 2006, Chung et al., 2008). Reports on hypodontia prevalence revealed that it was higher in the permanent dentition than in the primary dentition, with primary teeth hypodontia considered an indicator for hypodontia of the permanent teeth (Arte, 2001, Ritwik and Patterson, 2018a). Primary dentition hypodontia prevalence in Caucasian populations was less than 1 per cent, while
a higher incidence has been reported in a Japanese population (2.38%) (Yonezu et al., 1997, Nieminen, 2009). On the other hand, primary dentition hypodontia was found in less than 1 per cent (0.6%) of an Icelandic population (Magnússon, 1984). Reports found that primary maxillary lateral and mandibular central incisors were the most frequently missing teeth congenitally, accounting for 50 and 90% respectively (Nieminen, 2009). No gender differences were reported in hypodontia prevalence (Arte, 2001).

Further variations in hypodontia prevalence have been reported, with most suggesting that tooth agenesis is more common in black Americans than white Americans, and more common in Europe and Australia than in North America (Harris et al., 2008, Polder et al., 2004). The incidence of mild and moderate hypodontia was higher than oligodontia (severe hypodontia) and it varied between populations. Oligodontia prevalence in the Swedish population was 0.09%, 0.084% among Norwegians and 0.16% and 1.04% among Danish and Hungarian populations respectively (Bergendal et al., 2006, Gabris et al., 2006, Rølling and Poulsen, 2001, Nordgarden et al., 2002). Studies that reported oligodontia in those populations reported the prevalence of mild and moderate hypodontia as 4.5% and 16.6% in the Norwegian and Hungarian populations (Nordgarden et al., 2002, Gabris et al., 2006).

From a gender perspective, hypodontia prevalence was higher in females than males with a ratio of 3:2, while a meta-analysis reported the prevalence of dental agenesis is 1.37 times greater in females than males (Larmour et al., 2005, Polder et al., 2004). Prevalence of tooth agenesis in the maxilla is more common than in the mandible (Sisman et al., 2007, Celikoglu et al., 2010). Bilateral tooth agenesis is more common than unilateral agenesis in contrast to primary dentition (Sisman et al., 2007, Yonezu et al., 1997).

Third molar teeth are the most commonly absent permanent teeth and represent almost a quarter (24%) of congenitally missing teeth (Silva Meza, 2003). Excluding third molar teeth, studies have reported the following teeth to be the most commonly developmentally non-
syndromic missing teeth: mandibular second premolars, maxillary lateral incisors, maxillary second premolars, and mandibular central incisors (Celikoglu et al., 2010, Amini et al., 2012, Nordgarden et al., 2002, Sisman et al., 2007). Celikoglu and co-workers, in their 2010 study, reported that the most commonly missing teeth congenitally were mandibular central incisors, followed by mandibular second premolars and maxillary lateral incisors. On the other hand, Sisman and co-workers stated that the most frequently missing teeth were maxillary lateral incisors, mandibular second premolars and maxillary second premolars in descending order, while Nordgarden reported that tooth agenesis frequency was more common in the mandibular second premolars, maxillary lateral incisors and maxillary second premolars (Nordgarden et al., 2002).

1.2.2 Prevalence of Hypodontia in Ectodermal Dysplasia Subjects

In 2006, Bergendal and co-workers examined 123 participants who had signs and symptoms of Ectodermal Dysplasia and met their inclusion criteria (71 females (57.7%) and 52 males (42.3%)). Seven out of 123 individuals were excluded as a result of their inability to undertake the required tests. Half of the 116 subjects (58/116) had oligodontia in addition to one or more signs or symptoms of Ectodermal Dysplasia. The frequency of these signs and symptoms were ordered as follows: a third (35/116) of those individuals had low salivation rates and 10.3% had abnormalities in hair, nails, or sweat glands (Bergendal et al., 2006). Over a third (34.5%) of those subjects had one sign and symptom of Ectodermal Dysplasia, while 14.7% (N=17) had two signs and symptoms (abnormal hair and abnormal sweating). Oligodontia prevalence was reported from panoramic radiographs for all participants. The number of missing teeth ranged from 6 to 20 with a mean of 8.3. More than half of subjects (55.6%) had six or seven missing teeth, while one out of ten (9.9%)
was missing more than 12 teeth. More than half (51.3%) of the missing teeth were maxillary and mandibular second premolars, followed by lateral maxillary incisors (Bergendal et al., 2006).

Nordgarden, in 2001, investigated the frequency of Ectodermal Dysplasia signs and symptoms in Norwegian individuals with oligodontia. Sixty-eight participants with oligodontia and 39 healthy controls underwent clinical and dental examinations, interviews, and saliva secretion rate tests. Abnormalities in hair or nails and sweat production in combination with oligodontia was reported in 57% (N=39) of ED participants of whom 15 were females and 24 males with an average age of 12 years old. Only four of this ED group had an abnormality in all four areas of ED (Hair-Teeth-Nails-Sweat glands), while the remaining 35 participants with oligodontia had fewer features of Ectodermal Dysplasia. In addition, the results demonstrated a negative relationship between oligodontia prevalence and saliva secretory rates when ED and non-ED oligodontia groups saliva secretory rates were evaluated in both groups ($P < 0.001$). The overall median number of missing teeth in all oligodontia patients was 11 and was higher in the ED group in comparison to the non-ED group (13 and 10 respectively). Maxillary and mandibular second premolars were the most commonly missing teeth. Comparing the ED and non-ED oligodontia groups, the prevalence of missing maxillary and mandibular central incisors and canines and mandibular lateral incisors was identified more than twice as often in the ED group as in the non-ED group (Nordgarden et al., 2001).

A Finnish family cohort with Hypohydrotic Ectodermal Dysplasia (HED) was also identified, which included nine families comprising 15 males, of whom four had died. Among the 11 living males, the mean maxillary permanent teeth number present was 4.7 (range 0–8) and 2.8 (range 0–6) mandibular teeth. The study also reported that one individual had
anodontia, and that three subjects had anodontia of the mandible (Airenne, 1981, Bergendal, 2010). The prevalence of missing teeth and other dental anomalies were also examined in the Danish Ectodermal Dysplasia population. From a recognised Danish community dentistry database, males with hypohydrotic ED (23), heterozygous females (36) and control females (73) were included. The mean age of participants was 20, 33, and 29 years old respectively, and they underwent clinical and radiographic examination (Panoramic and Cephalometric). In addition, diagnostic dental study casts and blood samples for DNA and chromosomal analysis were obtained. Investigations revealed that the mean number of missing permanent teeth was five times higher in males (22 teeth) than females (4 teeth). The most stable teeth in the affected males were maxillary central incisors and canines, in addition to the first molar in the maxilla and mandible. For heterozygous females, the most frequently missing teeth were maxillary lateral incisors. Furthermore, this study reported that both affected males and females had a high prevalence of root morphology malformation or deviations (Lexner et al., 2007c).

1.2.3. Other Dental Anomalies in Patients with Hypodontia

An increased incidence of microdontia and conical teeth has been reported to be associated with hypodontia, and a positive relationship between hypodontia and the prevalence of microdontia had been reported. Lai and co-workers reported that 8.9% of patients with hypodontia had conical-shaped incisors (Lai and Seow, 1989). Another tooth anomaly associated with hypodontia was ankylosis of the primary molar teeth which was stated to be a feature of 65.7% of those with hypodontia, in contrast to 1.5% in the control children (Lai and Seow, 1989). Also, transposition of permanent maxillary canines and first premolars has been described for a fifth (20%) and a quarter (24%) of patients who were
missing lateral incisors and mandibular second premolars respectively (Camilleri, 2005). In addition, taurodontism in mandibular first permanent molars was observed with a higher prevalence in participants with hypodontia in contrast to those without hypodontia: 34.3% and 7.1% respectively (Lai and Seow, 1989). Kan et al., in 2010, reported no difference in the prevalence of taurodontism between boys with or without hypodontia (P=.83), while girls with hypodontia had a higher prevalence of taurodontism compared to controls (P=.003) (Kan et al., 2010). Lai and Seow also reported that 11.9% of patients with hypodontia also had enamel hypoplasia (Lai and Seow, 1989).

1.2.4. Management of Hypodontia in Ectodermal Dysplasia Patients

Bergendal, in 2010, illustrated the role of the mouth during infancy and identified that the mouth is the organ of the body first discovered by a small child. An American psychoanalyst René Spitz highlighted the importance of the mouth from birth saying “the mouth is the cradle of perception” (Spitz, 1965). Many oral functions are vital including feeding, swallowing, harmonisation of breathing, in addition to social communication that includes speaking and laughing (Nunn, 2000, Capra, 1995). The rich maps of the motor and sensory regions of the mouth increase the vulnerability of individuals with congenital diseases to oral function and structure disturbances in both foetal and after-birth periods (Bergendal, 2010).

Treatment strategies in individuals with hypodontia for oral rehabilitation require long-term patient and family commitment, involving many different dental and medical specialities. The goal of treatment planning for those young individuals should be within the concept of Hedegård’s recommendation at the 1965 European Orthodontic Society Congress which stated that “every effort should be taken to avoid prosthetic treatment at
the final stage” (Hedegärd, 1965). Treatment planning for individuals with missing teeth at an early age is the cornerstone of their treatment because this aids in minimising the number of missing teeth requiring replacement. Examples of treatments or interventions that can be provided at the growth stage are early extraction of primary teeth to facilitate more mesial eruption of permanent teeth into more favourable positions in the jaw and auto-transplantation of permanent teeth to a more favourable position. In addition, orthodontic treatment is one of the treatments that can be provided during the growth stage that may facilitate later prosthodontic treatment (Nunn et al., 2003, Jepson et al., 2003, Forgie et al., 2005).

The dental literature has reported different treatment modalities for individuals with missing teeth. Worsaae and co-workers, in 2007, reported on a cohort of 112 participant patients with severe hypodontia with an average of 10 missing teeth, among whom 10 participants had Ectodermal Dysplasia. The majority of all patients (97%) commenced orthodontic treatment and of the (112) patients, (51) completed orthodontic treatment. Ninety per cent (46) of them had implant-supported restoration, while two and three of those patients had conventional bridges and removable partial dentures respectively (Worsaae et al., 2007). Furthermore, Thofelt stated that multidisciplinary treatment planning for 61 Swedish patients with oligodontia (624 missing teeth) revealed that prosthodontic treatment replaced fewer than half of the missing teeth (N = 260) (42%) among those individuals, while other missing teeth were not replaced. Two-thirds (N=40) had implant placement as part of their treatment, which was the most frequent restorative treatment for all patients (Thofelt, 2013).

Oral rehabilitation for hypodontia in Ectodermal Dysplasia patients through conventional removable prosthodontic treatment has been reported in clinical case reports. This treatment may be indicated for young growing patients and where fixed prosthodontics or
implant treatment are contraindicated. These reports presented oral rehabilitations with maxillary and mandibular complete or removable partial dentures, and recommended that they start as early as possible to restore the patient’s missing function, mastication, speech, aesthetics, and prevent growth anomalies; in other words, it was not only teeth replacement or restoration (Hekmatfar et al., 2012, Omondi et al., 2015, Derbanne et al., 2010, Tarjan et al., 2005b). Complete or removable prostheses provide maintenance of healthy dietary intake for children with missing teeth, which is important given that lifelong dietary intake patterns are established during childhood (Tarjan et al., 2005a, Kravitz et al., 1983). Also, restoration of missing teeth improves speech-related problems relative to the pre-prosthetic treatment stage (Mitchell and Grant, 1976, Riekman and el Badrawy, 1985, Tarjan et al., 2005a). Management using removable prostheses at an early age is indicated to maintain vertical and sagittal skeletal relationships during craniofacial growth, and facilitate future treatment, which consequently contributes to improvements in the social integration of individuals with Ectodermal Dysplasia (Hekmatfar et al., 2012, Omondi et al., 2015, Derbanne et al., 2010, Tarjan et al., 2005b). Treatment stages and sequencing for young patients involving removable prostheses were the same as for non-syndromic edentulous or partially dentate patients, with ED patients presenting with severely deficient alveolar ridges as a result of the absence of teeth germs with consequent teeth agenesis (Imirzalioglu et al., 2002, Hekmatfar et al., 2012). However, as the patients develop and grow, adjustments to the prostheses are required at different stages and prostheses replacement should be considered when the occlusal vertical dimension has decreased or changes in the mandibular position become evident as the patient ages (Hekmatfar et al., 2012, Omondi et al., 2015, Derbanne et al., 2010, Tarjan et al., 2005b). In addition, recent advancement in digital dentistry and CAD/CAM technology should be employed in the fabrication of removable prostheses for these patients with the
advantages of improved strength, convenient laboratory steps, improved fit and easy duplication (Punj and Kattadiyil, 2019).

1.2.3 Treatment of Young Individuals with Dental Implants

With the reported success of dental implants in the replacement of missing teeth, their use in the treatment of young patients with ED has been considered and investigated. Replacement of a missing central incisor as a result of dental trauma with a dental implant in young individuals has been attempted. However, follow up revealed that the dental implants behaved like ankylosed teeth, preventing alveolar bone growth in the implant region (Johansson et al., 1994, Oesterle and Cronin Jr, 2000). This was confirmed by further experiments on growing pigs and long-term follow up in another child with an early placed implant (Odman et al., 1991, Rossi et al., 2003). Nevertheless, a 1995 Swedish consensus conference on dental implant treatment in young patients stated that “although implants should not, as a rule, be placed in healthy young individuals until growth is completed, implants may be placed in individuals with anodontia or severe oligodontia before the pubertal growth spurt” (Sharma and Vargervik, 2006, Koch, 1996).

Oesterle and co-workers (1993) discussed implant placement in a growing patient and key factors were the pattern of facial growth and appropriate age for implant placement. The mandible had a posteriorly directed V-shaped growth pattern in a posterior superior direction, and before the eruption of the primary teeth, the symphyseal suture growth ceased, which stabilised the mandibular anterior width which had further slight appositional growth. In addition, the width of the mandibular inter-canine area does not change significantly following the eruption of the mandibular lateral incisors (around the age of 8 years), while at the arch level, individual changes have been reported with a mean
3.5 mm increase in intercuspal width between the second premolars from age 4 to 8 years old. These changes do not provide a predictable estimation of the intercanine width for when the individual will be fifteen years of age. The authors recommended implant placement in the mandible when growth had slowed, at the age of 14-15 for girls and 17-18 years of age for boys.

On the other hand, the maxilla had a different growth pattern, which occurred in three planes: downward, forward, and remodelling, which makes the behaviour of osseointegrated implants unpredictable, particularly in the posterior area of the maxilla where more growth occurs. Nevertheless, it was reported that placing implants during the pubertal period involves a higher likelihood of success than at an earlier age, but the success rate is still less than post-growth implant placement (Cronin Jr et al., 1994, Oesterle et al., 1993).

Favourable reports of dental treatment involving dental implants in young males with Hypohydrotic ED treated between the ages of 1.5 to 6 years old have been presented. Bergendal described the oral rehabilitation of a boy who presented with maxillary hypodontia and mandibular anodontia and severe alveolar ridge resorption when aged 3 years. A maxillary removable partial denture was fabricated for the boy and at the age of 6 years and two Brånemark implants were placed in the mandibular canine sites with subsequent implant overdenture construction. Over the following four years, adjustments were completed as the patient grew and permanent teeth erupted.

In addition, Smith (1993) reported implant placement in a 5-year-old boy who had hypohydrotic ED. The child was partially dentate in both arches and had been subjected to teasing from other children. A single Brånemark implant was placed in the midline of the mandible at age 5 years. A resilient attachment was included in the mandibular removable partial denture and the patient was re-evaluated every six months and following 4.5 years
of denture wear a longer abutment was utilised as the implant had become submerged. For this patient, no implants were considered for the maxilla due to inadequate bone volume. The author reported the importance of precautions with this kind of treatment because of the possible nature of the implant as a foreign body when it is completely submerged. Bonin concluded that, following treatment of a 4-year-old ED patient with successful implant placement, this form of treatment for ED patients was endorsed, provided good multidisciplinary support was available (Bergendal et al., 1991, Smith et al., 1993, Bonin et al., 2001).

On the other hand, Kearns reported the loss of one implant among six patients with an average age of 7.8 years who were followed up for five years (Kearns et al., 1999). A large prospective study reported on 51 patients with Ectodermal Dysplasia with an age range of 6 to 68 years who had 243 implants placed in the anterior mandible and 21 implants in the anterior maxilla. High survival rates were reported in the mandible (91%) while in the maxilla, a lower 76% survival rate was reported. In the study, 14 patients lost an implant with two of these occurring before implant loading; the discrepancy between these survival rates was because it was easier to modify the bone and place graft material in the mandible than in the maxilla. In addition, previous surgeries for cleft palate management in some patients resulted in compromised bone volume and blood supply to the implants placed in the maxilla (Guckes et al., 2002).

A critical review of the survival rate of dental implants in patients with Ectodermal Dysplasia and tooth agenesis revealed survival rates of between 88.5% and 97.6% in patients with ED, while the failure rate of implants at the individual level ranged from 16.7% to 35.7%. It is noteworthy that 93-100% of implant failures occurred before prostheses insertion with a higher rate in the maxilla than the mandible (5-29% and 0-9% respectively). Seven studies that assessed implant survival in Ectodermal Dysplasia patients were included in that
review, with a range of participants from 4 to 52 years of age. A total of 264 implants were included in one reviewed study that had 52 participants with a mean follow-up time of 1.9 years, while the other reviewed studies had 10 to 87 implants with a mean follow-up time ranging from 2.3 to 3 years. The authors stated that the studies included in that critical review had Level 3 evidence, in other words, they were descriptive studies at the level of comparative and correlation studies or case series (Yap, 2008).

Furthermore, a retrospective study reported on the provision of dental implants in 14 young subjects with Ectodermal Dysplasia and stated that 88.5% of the sixty-one implants had successfully integrated, whereas 3/15 in the anterior maxilla and 4/46 in the anterior mandible were lost. Loss of at least one implant before loading had been reported among 5/15 patients in this study (36%). The failures were attributed to multiple reasons in different patients. Limited bone volume was one of these reasons; the second reason was a history of osteotomy and iliac bone grafting that was followed by sinusitis and implant loosening, while the last reason was placing implants on the site of extracted impacted mandibular canines (Sweeney et al., 2005).

Bergendal retrospectively investigated implant failure in children (less than 16 years old) with Ectodermal Dysplasia in Sweden between 1985 and 2005. A questionnaire was sent to specialist clinics in Oral and Maxillofacial Surgery and Prosthodontics to record implant-related information for the selected population. The results revealed that 14 implants had been placed for five children with Ectodermal Dysplasia (with an age range of 5 to 12 years), of whom four were male. Over half (64.3%) of these implants had been lost before loading. The rate of implant failure was higher in younger age children, and higher than implant failure in non-syndromic hypodontia patients. It was argued that the reasons for the reported failures were a low quality of alveolar bone and the limited dimension of the available bone. This study recommended a combination of CT scanning, thorough
treatment planning, use of small dimension implants and delaying treatment until jaw size was sufficient for implant placement (Bergendal et al., 2008).

Failure and complications of dental implants and prosthodontic treatment was also reported by Krieger who reported on individuals who had congenital birth defects including hypodontia. After over eight years, more than 60% of single crowns and 64% of complete dentures were free from complications. Complication with dental implants occurred earlier than complications with single crowns. Nevertheless, the author recommended dental implant treatment for patients with hypodontia as it decreased the biologic cost of the treatment with tooth preparation being avoided (Krieger et al., 2009).

1.2.6. Scientific Evidence for Diagnosis and Treatment of Ectodermal Dysplasia

Evidence-based medicine is the concept proposed for the determination of efficient cures for common diseases and public health problems (Haynes et al., 1997). The highest level of evidence-based medicine is “strong evidence from at least one published systematic review of multiple well-designed randomised controlled trials”, while the lowest level is “opinions of respected authorities based on clinical evidence and descriptive studies or reports of expert consensus committees” (Bergendal, 2010). In dentistry, the evidence-based medicine concept has had an impact on developing and defining preventative and treatment approaches for common dental diseases, in addition to outcome evaluation for common therapies (Bergendal, 2010). However, some clinical condition management and treatment does not fit into the evidence-based medicine or dentistry models as a consequence of low levels of evidence in rare disorders. In addition, clinicians and researchers do not frequently refer to case reports as evidence of research work (Bergendal, 2010).
Bergendal argued that in relation to small groups with rarer conditions, long-term follow-ups of a group of subjects with the same condition had a significant value as scientific evidence. Clinical understanding of some conditions requires an understanding of the patients rather than the disorder itself, which cannot be identified using a standard quantitative research paradigm (Malterud, 2001). Also, rare disorders and their treatment cannot be examined in randomised clinical studies for ethical reasons but examined by research designs with low levels of evidence, such as case reports in combination with qualitative search methods (Bergendal, 2010). This combination of clinical evidence will “provide the best possible evidence” with respect to the optimal evidence achieved from prospective monitoring of large cohorts in other (non-rare) clinical conditions (Bergendal, 2006, Bergendal, 2010).

Clinical trials for the diagnosis and treatment of Ectodermal Dysplasia have been documented. In 2013, Edimer Pharmaceuticals began the first clinical trial to develop therapeutic interventions for X-linked Hypohydrotic Ectodermal Dysplasia. The study tested protein therapy (EDI200) for signs and symptoms in 10 newborns affected by the condition. The trial reported no significant changes in sweat gland function after the use of the protein-derived therapy (NationalFoundationforEctodermalDysplasias, 2020). The investigators proposed that the treatment was not provided early enough to result in the expected outcomes. This was supported by later studies completed by Schneider and co-workers who examined the impact of protein therapy on animals and affected humans. In the human trial, treatment was provided for twin boys and another third infant in utero at different time points during their mother’s pregnancy. The three children had no problems in the summertime as they perspired normally and also had normal saliva production and more tooth germs formed (permanent teeth) in contrast to non-treated affected siblings (NationalFoundationforEctodermalDysplasias, 2020).
Diagnosis and treatment of X-linked Hypohydrotic Ectodermal Dysplasia at the prenatal stage is currently being researched in a project funded by the National Foundation for Ectodermal Dysplasia led by Professor Angus Clarke which aims to find a novel non-invasive method of diagnosing X-linked Hypohydrotic Ectodermal Dysplasia in utero (NationalFoundationforEctodermalDysplasias, 2020). The test method will be based on blood sample collection from pregnant women who are carriers of X-linked Hypohydrotic Ectodermal Dysplasia (XLHED). DNA will be extracted from the blood samples, and as this is a mixture from both mother and infant, it may be used to determine if the baby’s DNA includes the XLHED gene. The success of this diagnostic method is a key for further future trials in which EspeRare plans to examine protein replacement therapy (ER-004) as an in utero treatment for XLHED (NationalFoundationforEctodermalDysplasias, 2020).

1.3 Oral Health-related Quality of Life

1.3.1 Quality of Life

The impact of various medical conditions on quality of life has been a topic of interest for many researchers over the last three decades. Quality of Life (QoL) had been defined as “a person’s sense of well-being that stems from satisfaction or dissatisfaction with the areas of life that are important to him/her” (Becker et al., 1993). The World Health Organization (WHO) Quality of Life working group defined QoL as the “individual’s perception of their position in life in the context of the culture and value systems in which they live and in relation to their goals, expectations, standards and concerns” (Organization, 2004). Quality of Life (QoL) is a wide concept that covers a wide range of human experience, including living conditions, job satisfaction, poverty, etc. (Erickson and Patrick, 1993). Consequently, caution should be exercised when health-related quality of life (HRQoL) or oral health-
related quality of life (OHRQoL) is examined because the quality of life might be influenced by covariant factors other than health-related ones.

1.3.2 Oral Health

The WHO has defined oral health as “a state of being free from chronic mouth and facial pain, oral and throat cancer, oral infection and sores, periodontal (gum) disease, tooth decay, tooth loss, and other diseases and disorders that limit an individual’s capacity in biting, chewing, smiling, speaking, and psychosocial wellbeing.” (Petersen and Programme, 2003).

1.3.3 Health-related Quality of Life (HRQoL)

This refers to the psychological, physical, and social domains of health, which are seen as distinct areas that are influenced by a person’s experiences, beliefs, expectations and perceptions (Testa and Simonson, 1996).

This concept has developed in 1980 to include:

- “On the individual level, HRQoL includes physical and mental health perceptions and their correlates—including health risks and conditions, functional status, social support and socioeconomic status” (CDC, 2000, NationalCenterforChronicDiseasePrevention and Health Promotion, 2018).

  - “On the community level, HRQoL includes community-level resources, conditions, policies, and practices that influence a population’s health perceptions and functional status” (CDC, 2000, NationalCenterforChronicDiseasePrevention and Health Promotion, 2018).
On the basis of a synthesis of the scientific literature and advice from its public health partners, CDC has defined HRQoL as “an individual’s or group’s perceived physical and mental health over time” (NationalCenterforChronicDiseasePreventionandHealthPromotion, 2018, CDC, 2000).

1.3.4 Oral Health-related Quality of Life (OHQoL)

OHRQoL has been defined as the “cyclical and self-renewing interaction between the relevance and impact of oral health in everyday life” (Gregory et al., 2005). Locker defined OHRQoL as “the impact of oral disorders on aspects of everyday life that are important to patients and persons, with those impacts being of sufficient magnitude, whether in terms of severity, frequency or duration to affect an individual’s perception of their life overall.” Locker also stated that a detailed definition of OHRQoL was required to carefully and critically address the questionnaires/tools used in OHRQoL assessment. In addition, they recognised that not all the concepts which needed to be measured were addressed by the current OHRQoL assessment tools. While the tools measure the frequency of the impact, many fail to determine the value of that impact on the individual (Locker et al., 2007).

1.3.4.1 Oral Health Quality of Life Historic Overview

The WHO has defined the health concept taking into account mental, social and physical wellbeing (InternationalHealthConference, 2002). The definition of health, therefore, shifted from a biomedical concept of health to a biopsychological concept that measured the outcome of disease through its impact on the physical, emotional, and social wellbeing of the individual rather than a mere absence of the disease. This health definition or concept was followed by the emergence of quality of life (QoL) and health-related quality
of life (HRQoL) in the medical literature from 1960 (Alzoubi et al., 2017). Advancement in medical and health-care concepts led to a shift from focusing on survival to prevention of disease, while diagnosis and treatment of chronic disease and thus mortality and morbidity rate parameters could not be used as the only indicators of the population’s health status or the efficiency of the health-care system (Gift and Atchison, 1995).

The importance of quality of life and HRQoL measures has been well documented in the published literature. Healthy People 2020 was a creative start by the Department of Health and Human Service in the US which highlighted HRQoL measures’ importance for society. Public health sectors and clinicians in the US implemented HRQoL measures that facilitate a better understanding of the impact of chronic illness, treatment and short- and long-term disabilities. These measures are used to define and prioritise the health issues and enable clinical decision-making and communication between patients and clinicians, in addition to the response to treatment monitoring (Inglehart and Bagramian, 2002a). The late emergence of QoL and HRQoL concepts and their application could reflect the lack of awareness of the impact of oral health on quality of life in earlier decades (Bennadi and Reddy, 2013).

Davis stated that the oral cavity was considered an anatomical anomaly isolated from the body which, apart from oral cancers, had no impact on social life (Davis, 1976). Gift and Atchison, in 1995, challenged Davis’s notion, and they hypothesised that oral health is an essential part of general health. Also, they stated that the contribution of the oral cavity in oral health-related quality of life started at a basic biological level “through protection from systemic infection, chewing and swallowing and at a more social and psychological level through self-esteem, self-expression, communication and facial aesthetics.” In other words, overall health and the quality of life might be adversely affected when oral health
is compromised, which was reported in the late eighties in the literature which concluded that oral diseases can influence the emotional and psychological well-being of individuals (Cushing et al., 1986, Gift et al., 1992, Reisine et al., 1989).

1.3.4.2 Health Models

OHRQoL and HRQoL have multi-dimensional constructs and theoretical model frameworks that link the relationships between oral health/health and QoL as moderated by numerous domains. The two most popular conceptual health/oral health models will be illustrated.

1.3.4.2.1 Wilson and Cleary’s Health-related Quality of Life Conceptual Model

This model has a range of measures of health outcomes which are divided into five levels: biological and physiological factors, symptoms, functioning, general health perceptions and overall quality of life. The arrangement of these five levels reflects an increase in complexity with the biological and physiological factors at one end connected at the other end to quality of life through other levels influenced by personal and environmental factors (Figure 1.3). This model was designed in such a way that as one moves from the left to the right the different levels have increased influence with a greater number of integrated inputs which are outside of the scope of clinical professionals and health-care systems. The arrows do not essentially mean a reciprocal relationship and the absence of arrows between non-adjacent boxes/levels does not necessarily imply the lack of one (Wilson and Cleary, 1995).
1.3.4.2.2 Locker’s Conceptual Model of Oral Health

This model was created from the concept of the WHO international classification of impairment, disability and handicap (ICIDH) which was first proposed in 1980. This classification was designed to define the psychological and biological consequences of diseases and intellectualise them within a framework where the fundamental relationship is implied between the consequences (WorldHealthOrganization, 1980). The biomedical outcome of the disease at the organ level is the immediate result of impairment; disability represents a difficulty in carrying out daily activity at the individual level, while handicap represents impairment experienced by the individual at a social level (overall impact) as a consequence of disability/impairment as strengthened by the surrounding circumstances (Allen, 2003b, Locker, 1988).

This model was further adopted by Locker for oral disease to provide a prediction of the pathways in which an individual might be affected. Like the Wilson and Cleary model, there are five levels of oral disease experience hierarchy where the impacts extend from a biological level to the level of the individual and then ultimately have an impact
psychosocially (Locker, 1988). This model framework has been important in the development of several OHRQoL measures and verification of their reliability and validity (Leao and Sheiham, 1996, Slade and Spencer, 1994, Allen, 2003a). In addition, where Locker’s model provided oral disease consequences, Locker’s model provided an implicit link to the quality of life (Figure 1.4) (Locker and Allen, 2007).

![Lockers Conceptual Model of Oral Health](image)

**Figure 1.4 Locker’s Conceptual Model of Oral Health**

### 1.3.4.3 Assessment of Oral Health-related Quality of Life

The OHRQoL measurement process has three main stages: defining a conceptual framework, undergoing technical procedures to formulate the measure and the application of a measurement interpretation scope (Locker, 1996). Validity and reliability of the measurement are required, and the measurement should be able to differentiate between subjects across different levels of health, with sufficient sensitivity to detect changes over time. Three different categories of OHRQoL measure have been identified: firstly, social indicators - the impact of oral disease at the community level; secondly - global ratings; and lastly multi-item questionnaires (Inglehart and Bagramian, 2002b).
The global ratings and multi-item questionnaire are the most frequently used measures in OHRQoL. The multi-item questionnaire describes subjective oral health status by frequency and severity of oral disease measurement across the functional and psychological domains. The global ratings allocate the domain’s importance to the individual’s OHRQoL which reflects their overall oral disease impact perception (Locker and Allen, 2007). The global rating involves a broad generic question, providing answers, addressing concerns, opinions, and considering each domain’s importance to the individual. It is also considered a validity check tool when a new measure is being established (Benson et al., 2016).

In addition, the benefits of having multi-item questionnaires are that they can be generic, such as the Oral Health Impact Profile (OHIP) questionnaire, or specific for a particular condition, such as those generated for cancer patients or those developed for patients with dentofacial deformities (Cunningham et al., 2000, Terrell et al., 1997). Comparisons between different populations is facilitated using generic measures, while specific questionnaires are more sensitive to the measurement of changes over time of a specific disease or condition and provide a clinically relevant response. Measurement questionnaires can also be age- or culture-specific; OHRQoL (UK) is an example of a culture-specific measurement tool (McGrath and Bedi, 2002). The Geriatric Oral Health Assessment Index is an example of an age-specific questionnaire which applies to senior adults (Atchison and Dolan, 1990).

For children, several OHRQoL measurements tools have been developed. They include the Early Childhood Oral Health Impact Score (ECOHIS), Child Oral Impacts on Daily Performances (Child-OIDP) Child Oral Health Impact Profile (COHIP) and Child Perception Questionnaire (CPQ) (Genderson et al., 2013). For adolescents, the Malocclusion Impact Questionnaire (MIQ) had been developed recently as a result of questions raised in relation to the meaning and significance of other generic OHQoL measures (Patel et al., 2016).
critical appraisal of the common ORQoL questionnaires was conducted by Locker and Allen, applying criteria proposed by Guyatt and Cook, to evaluate if these questionnaires actually measured the intended outcomes (Locker et al., 2007). This appraisal revealed that these questionnaire tools covered important aspects for the patient, while linking the tools used to the quality of life measurement had not been relevant or successful. In addition, the appraisal recommended the incorporation of more global rating tools in order to provide meaningful and individualised data which can be grouped for analysis (Locker et al., 2007).

1.3.4.3.1 Child Oral Health-related Quality of Life

A literature review that evaluated oral health-related quality of life (OHRQoL) in children with Ectodermal Dysplasia revealed a paucity of studies. Previously, the source of quality of life assessment for children and adolescents was via their parents. However, validated questionnaires to assess the quality of life for children and adolescents based on their self-experience have been developed (Levi and Drotar, 1999).

The development of children as they grow involves continuous cognitive, physical, and emotional changes. Consequently, a child’s awareness of their general health and its physical and psychological impact is age-dependent, which reflects their ability to answer health-related questionnaire at different ages. Understanding complex emotions such as shame, worry, jealousy, and peer pressure effect are clear and understandable for children by the age of 11 to 12 years (Hetherington et al., 1999). Oral health-related quality of life (OHRQoL) research that used parental response as proxies found that parents’ knowledge of children’s health psychological impact was limited and did not reflect the children’s health-related quality of life (Levi and Drotar, 1999).

Barbosa and Gavião identified three oral health-related quality of life questionnaires that have been validated. These questionnaires were the Child Perception Questionnaire (CPQ)
for both age groups 8-10 and 11-14 years of age. The second one was the child-oral impacts on daily performances (Child-OIDP) and the third was the Child Oral Health Impact Profile (COHIP). The COHIP questionnaire has been developed from the same item pool as the CPQ, but it deviates from CPQ by including positive health instruct. The authors stated that reliable results for oral health-related quality of life can be achieved from children when the appropriate validated questionnaire is used (Barbosa and Gavião, 2008). The CPQ was selected because it is an age-specific tool that examines the quality of life from different four aspects, in addition to its inclusion of the family impact scale that considers the impact of oral health impairment on the children's family.

1.3.4.3.2 The Child Perception Questionnaire

The Child Perception Questionnaire CPQ is one of the validated instruments for the measurement of Child Oral Health Quality of Life (COHQoL) which has age-specific measures for children and their primary caregiver. This questionnaire was originally developed by Jokovic at the Faculty of Dentistry, the University of Toronto for paediatric and orthodontic patients. The purpose was to develop a tool to measure dental and oro-facial disorders in a clinical trial and other assessment studies (Jokovic et al., 2002). It consists of three versions by age: CPQ 6-7 years old, CPQ 8-10 years old, and CPQ 11-14 years old. In addition, it has a parent-caregiver perception questionnaire (P-CPQ) and Family Impact Scale to test the impact of children’s oral health on their family. The questionnaire is generic and can be used for the measurement of different oral and oro-facial conditions (Jokovic et al., 2003b).

Theories of measurement and development scales have been used to develop these child oral health quality of life questionnaires as outlined by Guyatt et al. (1986). A two-phase process was used to develop the items for the instrument. Firstly, an initial pool of 46
questions that covered a range of health domains was developed. Then the questions’ relativity, clarity, and comprehensiveness were assessed by experienced health professionals who provided treatment regularly to children with different oral-facial conditions. In addition, detailed interviews with eleven children were conducted leading to the development of a modified question list. Item study was used to assist in the selection of the final instrument questions which aimed to scale the items of higher importance for inclusion in the definitive questionnaire. Three groups of participants were involved in the development and evaluation of the child oral health quality of life questionnaires. The included groups had different clinical conditions including dental disease (dental caries), malocclusion and oro-facial disorders (primarily cleft lip/palate) (Guyatt et al., 1986).

The child perception questionnaire (CPQ) developed by Jokovic has three versions: 6-7, 8-10, and 11-14 years of age. The 11-14 version consists of 36 items that are divided into four main domains: oral symptoms (6 items), functional limitations (9 items), emotional wellbeing (9 items), and social wellbeing (peer interaction, schooling and leisure activities) (12 items). They aim to detail the child’s experience about the occurrence of an event over the last three months with the following options: never = 0; once or twice = 1; sometimes = 2; often = 3 and every day or almost every day = 4. Responses code sum creates each domain’s score and an overall CPQ questionnaire score. The questionnaire also includes a global rating of a child oral health and the level at which the oral conditions impact the child’s life. The first question is “Would you say that the health of your teeth, lips, jaws and mouth is...?” The answer options ranged from ‘poor’ to ‘excellent’. The second question is “How much does the condition of your teeth affect your life overall?” The answer options scaled from ‘not at all’ to ‘very much’ (Jokovic et al., 2002).

The Child Perception Questionnaire (CPQ) 8-10 consists of 25 questions that cover four domains: oral symptoms, functional limitations, emotional well-being and social well-
being. The study of CPQ 8-10 development included children with dental caries or oro-facial defects (cleft lip or cleft palate). The participants provided responses for each question as coded ‘never’ = 0; ‘once/twice’ =1; ‘sometimes’ = 2; ‘often’ = 3; and ‘everyday/almost every day’ = 4. The total response score for children with dental caries was (18.6, SD = 11.5), while the orofacial group CPQ scores were higher (19.1, SD = 14.4), and the difference was statistically significant. More oral symptoms had been reported by children with dental caries, while functional limitations and social well-being issues had been reported more frequently by those with oro-facial defects. A higher CPQ score (21.1) was reported by children who had caries in contrast to caries-free subjects (14.7), with a positive correlation between CPQ score and the number of decayed tooth surfaces. The authors confirmed the reliability and validity of the questionnaire (Jokovic et al., 2004b).

1.3.4.3.3 Validity and Reliability of CPQ

The validity and reliability of the Child Perception Questionnaire (CPQ) 11-14 years old have been evaluated by Jokovic and co-workers in children with dental caries in comparison to those who have orthodontic disorders and those with orofacial conditions (cleft lip and cleft palate). Higher scores were reported by children with orofacial conditions (31.4, SD = 17.1) in contrast to those with dental caries (23.3, SD = 18.4) and orthodontic treatment (24.3, SD = 14.5), with a significant correlation between CPQ score and global ratings of health and overall well-being. In addition to the reliability and validity of the questionnaire, the authors concluded that “the impact of child oral health on functional and psychological well-being are substantial, and the children were able to give psychometrically acceptable account of that impact” (Jokovic et al., 2002). Marshman tested the CPQ 11-14 in a UK sample of 89 children from Sheffield who had various oral conditions including dental caries, malocclusion and enamel opacities. The validity was assessed by contrasting the
global rating of oral health to the overall CPQ rates in addition to testing the association between CPQ rates/scores and the clinical data. Their findings were that the CPQ results corresponded well with global oral health ratings and impact on life overall, demonstrating acceptable criterion validity. The reported impacts were influenced by the number of teeth absent and those missing as a consequence of caries (Marshman et al., 2005a).

Furthermore, O’Brien et al. (2006) validated the CPQ as a measurement tool for measuring the impact of malocclusion on children. The authors conducted a longitudinal study of a sample of UK children from Manchester. This study found that CPQ scores for the participants agreed with those reported by the Index Orthodontic Treatment Need (IOTN) scores, with greater scores in children who assumed that their teeth needed to be straightened. However, no relationship was developed between CPQ scores and the IOTN aesthetic component (O’Brien et al., 2006).

A Danish version of CPQ was also developed and validated. Wogelius et al. developed a CPQ 8–10 that had 27 questions and a CPQ 11–14 that had 39 questions. CPQ 8–10 was tested on a sample of 120 children from three different groups: a healthy group, children with cleft lip and palate, and those with rare oral conditions such as Ectodermal Dysplasia and Amelogenesis Imperfecta. The CPQ 11–14 was tested on 225 children who were divided into four groups; the first three were the same as the CPQ 8–10 and the fourth group was an orthodontic patient group. The findings of the study were as follows: in the 8–10-year-old group from a total of 108, the overall median CPQ score was higher in children with rare oral conditions (15) (Ectodermal Dysplasia and Amelogenesis Imperfecta) than healthy children and children with cleft lip and palate (7 and 5 respectively). The median overall scores for the 11–14 group were higher in orthodontic groups (24.4, SD = 12.5) than rare oral condition children (17.8, SD = 8.8), which were both higher than healthy and those with cleft lip and palate (10.5, SD = 7.2) from the total possible score of 156. The authors
concluded that the CPQ 8-10 and CPQ 11-14 were valid tools for quality of life measurement in Danish children (Wogelius et al., 2009).

In North Ireland, Humphris and co-workers examined the validity and reliability of the CPQ 8-10 version by assessing oral health-related quality of life in a sample of 270 children using both CPQ 8-10 and the Coopersmith Self-Esteem Inventory-School Form (Coopersmith SEI-SF) for children aged 8-15. There was no significant difference statistically between both instruments’ scores with respect to both ages and gender, and the study confirmed the validity and reliability of the CPQ 8-10 (Humphris et al., 2005).

1.3.4.3.4 Parent-Caregiver Perception Questionnaire (PPQ)

Parental perception of their children’s quality of life is vital because of their role in treatment choice selection for their children. In addition, parents are considered to be the primary decision-makers for their child’s healthcare measures, which might influence the child’s treatment needs and possibly lead to treatment provided based on the parents’ view rather than the child’s needs. For instance, Stricker reported that parents often have a role in seeking orthodontic treatment for their children with 63-67% pressurising their children to seek orthodontic treatment (Stricker, 1970, Wędrychowska-Szulc and Syryńska, 2010).

The Parent Perception Questionnaire (PPQ) provides limited information about the individual child’s activity, particularly that occurring inside the home. Thus, the purpose of the PPQ is to be used as a proxy rather than as a secondary or complementary information source to information provided through the Children Perception Questionnaire (CPQ). Jokovic originally developed the PPQ as one of the child oral health-related quality of life questionnaire components for the parents of children aged 6-14 years (Jokovic et al., 2003b).
The 31 items included in the PPQ were selected from a pool of 47 items through an item impact study similar to the Children Perception Questionnaire 11-14. The validity and reliability of the questionnaire were evaluated by the same author in a group of 231 parents, where 79 of them completed the questionnaire twice at a two-week interval for reliability assessment. The 31 questions had a response with the following codes: ‘never’ = 0, ‘once or twice’ = 1, ‘sometimes’ = 2, often = 3, ‘every day or almost every day’ = 4, and ‘I don’t know’ responses. The last response (‘I don’t know’) led to information interpretation loss. This was managed by giving a zero score to this response and calculating the response by dividing the sum of all 31 responses with the number of items with valid responses and reducing the denominator according to the number of items with a response of ‘I don’t know’. In this validation study, the overall PPQ mean was 25.1 (out of a possible 124), with a higher perception in the orofacial group (31.83) in contrast to the orthodontic and paediatric groups (23.84 and 5.86 respectively). In three domains (the functional limitations, emotional well-being and social well-being domain) the orofacial group reported the highest scores, while in the paediatric group, it was higher in children with higher caries rates (Jokovic et al., 2003b).

Agreement between the Children Perception Questionnaire 11-14 and PPQ was examined in 42 pairs of mothers and their children who had orofacial conditions. The overall substantial agreement Intraclass Correlation Coefficient score was (ICC: 0.7), with an excellent agreement in the oral symptoms response (ICC: 0.81), while demonstrating substantial agreement for functional limitations (ICC: 0.71) and moderate agreement for social well-being (ICC: 0.58). At the level of global rating questions, there was moderate agreement between children and their mothers (ICC: 0.51). Also, variations in the level of agreement were influenced by various factors: lower levels of agreement were observed in older children, females, the orofacial group and children who reported the highest scores.
for QoL. The overall perception score for child CPQ was 26.2 (SD 15.8) in contrast to their mothers’ PPQ which was 22.6 (SD 17.1). The authors concluded that both Child Perception and Parent Perception Questionnaires could be used with children for oral health-related quality of life assessment (Jokovic et al., 2003a).

Another study assessed the agreement between the Brazilian version of the Parental Perception Questionnaire and the short form of the Children Perception Questionnaire (CPQ 11-14). A sample of 960 pairs of healthy children and their mothers completed the questionnaires for oral health-related quality of life assessment. Children reported higher scores than their mothers, particularly in the functional limitation and social well-being domains. The authors concluded that children and their mothers had only a moderate level of agreement in terms of oral health-related quality of life assessment (Ferreira et al., 2012). Furthermore, Kohli assessed oral health-related quality of life in 35 children and adolescents (11-19 years old) with Hypohydrotic Ectodermal Dysplasia (HED) and their parents/caregiver using the CPQ11-14 and PPQ respectively. They found that older children and adolescents (15-19) reported more functional limitations than younger children (11-14), while female participants reported more emotional limitations. Interestingly, parents reported higher scores than their children, but no significant differences were observed between both sets of perception scores (Kohli et al., 2011).

Jockovic and Ferreira reported a low level of agreement between parents and adolescents, while Abreu reported a poor level of the agreement particularly in social and emotional domains, where children reported higher scores. In addition, despite the findings earlier presented above, it was recommended that caution should be exercised when parents were used as a proxy for oral health-related quality of life assessment (Kohli et al., 2011). Children had a different opinion from their parents/caregivers, and it was argued that the explanations for these different views were inconsistent data in the studies which reported
these opinions and also that the children/parent dynamics might be influenced by cultural factors (Ferreira et al., 2012). While Abreu stated that whilst limited insight of parents might influence their perception of their children, it does not undermine the importance of parent participation in oral health-related quality of life assessment (Abreu et al., 2015).

1.3.4.4 Ectodermal Dysplasia and Hypodontia Impact on Quality of Life

In recent years, more emphasis had been given to oral health-related quality of life (OHRQoL) research. A review of the available literature has suggested that there is a relationship between oral health and OHRQoL. Some of the reviewed studies only found a weak relationship which was credited to low disease levels in the included samples with low impact on OHRQoL. The reviewed studies reported a strong correlation between dental caries and fluorosis with OHRQoL, while a negative impact on OHRQoL was reported by children with cleft lips and palates (Barbosa and Gavião, 2008).

Ectodermal Dysplasia (ED) is one of the genetic craniofacial conditions that affects subjects and impacts their quality of life. ED affects teeth, hair, sweat glands and nails and impacts the affected subjects in different ways depending on the severity of the condition. Dental anomalies are very common in ED patients, which can present as hypodontia or tooth malformation with conical or hypoplastic teeth. The relationship between Ectodermal Dysplasia, orofacial syndromes and quality of life in children and adolescents has been investigated in the literature. Geirdal et al., evaluated the psychological impact and quality of life of various craniofacial condition in adults aged over 18 years using the Short Form Health Survey 36 (SF-36) questionnaire that has a score range of (0-100). The ED group involved 49 participants of whom 33 of them had dental prostheses planned before the study. In contrast to levels of anxiety among Treacher Collins Syndrome (mean 3.18, SD = 2.71) and Cherubism groups (mean 2.46, SD = 2.11), the ED group reported significantly
higher levels of anxiety (mean 6.96, SD = 3.73) in addition to reporting the lowest mental health-related quality of life scores. It was argued that anxiety was attributed to the subjects’ worries about their future, in addition to the stress experienced by bullying during adolescence. On the other hand, the Treacher Collins Syndrome group reported higher levels of depression and lowest quality of life scores (Geirdal et al., 2015a). Another study by Saltines and co-workers using the same instrument (SF-36) among 47 adults (mean age 29.2) with hypodontia, of whom 35 had ED, reported higher levels of depression and anxiety in individuals with more than ten missing teeth, and poorer mental-health-related quality of life and HRQoL in those with removable dentures and those complaining of a dry mouth (Saltines et al., 2017a).

The German version of the Oral Health Impact Profile questionnaire (OHIP-14) was utilised in two studies for OHRQoL assessment in ED patients. The first study evaluated OHRQoL in 110 Ectodermal Dysplasia patient participants with an average age of 5.19 and 17.02 years for males and females respectively. In total, 99.09% (109) of participants reported oral symptoms as an influencing factor and difficulty finding a dentist to provide appropriate treatment and dissatisfaction with the health-care system. The highest OHIP score was 56 which indicates the worst OHRQoL; among participants, the reported OHIP scores were higher in females (12.23, SD = 12.39) than males (11.79, SD = 11.08), being higher than the mean normal German population OHIP score (4.09) (John and Micheulis, 2003). However, gender was not an influencing factor in that study (Hanisch et al., 2019a).

A second study assessed the OHRQoL of 484 individuals who had one of nine rare diseases, including ED (46). The participants were adults (mean age 44.6 years) with a higher percentage of females (64.7%). Following data collection using the German version of OHIP, a different cluster analysis was completed for participants who reported oral symptoms.
These symptoms included anomalies of the tooth formation, dysgnathia, changes in the number of teeth, and malocclusions. The reported OHIP scores were higher than the mean German OHIP score (4.09) with a range of scores between 15.1 and 19.9 for all participants, and 15.9 for the oral symptoms cluster group and 19.7 for the oral mucosal disease cluster group (Wiemann et al., 2018). The findings from both studies show how Ectodermal Dysplasia influenced the quality of life from the presented OHIP scores with no significant difference between genders, in particular for participants with oral symptoms.

Wong assessed the impact of isolated hypodontia on OHRQoL in 25 children with severe hypodontia. The most commonly absent teeth were maxillary lateral and mandibular incisors. All participants reported one or more social impacts, and 88% of subjects reported functional limitations and some impact on social well-being with a mean CPQ of 29.0, which was positively correlated with the number of missing teeth. This consistency or correlation between missing teeth number and CPQ was (0.54) and changed to (0.94) when retained primary teeth replaced the missing permanent teeth, which reflected the influence of having the teeth present on the quality of life (Wong et al., 2006).

Another study examined the impact of non-syndromic hypodontia on 36 children using the CPQ11-14. The mean number of missing teeth was 6.8 with a higher prevalence of premolar (58%), followed by anterior and molar teeth, (26%) and (16%) respectively. Almost two-thirds (61%) of the children reported some functional limitations, a fifth (19%) reported impacts on emotional well-being and 17% reported an impact on social well-being. The mean CPQ was 22.3 with a range from 4 to 69, with no gender impact observed. The position of the missing teeth (premolars and anterior) was the influencing factor of the reported CPQ scores in terms of both the emotional and social well-being domains (Locker et al., 2010).
A cross-sectional study investigated the psychological impact of hypodontia using the CPQ in a sample of 62 children (aged 11-16 years old) in contrast to 61 children without hypodontia but with a 4 or 5 IOTN score. With a mean of 4.52 missing teeth in the hypodontia group, they reported no significant difference between CPQ scores for the hypodontia and orthodontic groups (26.8 and 28.5). In addition, psychological status was not impacted by hypodontia with a mild positive association between the number of missing teeth and chewing difficulty (Laing et al., 2010).

Another study investigated the impact of hypodontia on OHRQoL, both quantitatively and qualitatively. Ten adult participants (aged 16-25 years) answered the Oral Health Impact Profile (OHIP) questionnaire. The participants presented with a range of hypodontia severity and were involved in semi-structured interviews. The OHIP response ranged from 24 to 143 with no gender effect observed. Qualitative data were analysed using the NVivo software and identified three themes including (1) the patient transition from childhood to adulthood influenced their understanding of the condition and hypodontia treatment decision-making, and in particular, their definitive treatment plan was started in adolescence when they could understand and intervene on the plan, however, two participants didn’t change what had been discussed and planned for them by their parents and their dentist; (2) participants indicated dissatisfaction with the lack of communication with dental services throughout early adolescence as they became more cognisant of their condition which, in turn, led to concerns about appearance, which impacted on their psychosocial well-being and (3) feelings of frustration as a consequence of delays in treatment initiation and transferring from one waiting list to another was a source of psychological stress and treatment dissatisfaction (Meaney et al., 2012).

However, participants stated that once their treatment started, the expectation of the treatment outcome changed and satisfaction with the treatment improved. From an
aesthetic perspective, they stated that feelings of self-consciousness decreased, and they became more comfortable socially. The authors concluded that hypodontia patients had difficulties in understanding the protocols for their treatment and, in addition, the time frame between diagnosis and commencement of treatment was a source of aesthetic concerns and psychological stress. These concerns reduced as soon as the treatment started (Meaney et al., 2012).

There a lack of peer-reviewed literature which examines the effect of other ED features on the quality of life. However, one study investigated the implications of ED cutaneous features in 28 children and 14 adults with a mean age of 6 years and 33 years respectively. They used the Children’s Dermatology Life Quality Index in children, and the Skindex-29 and RAND Short Form-36 in adults. All subjects reported increased impacts from hypohydrosis when compared to other cutaneous features (alopecia, fingernail deformity) with no significant impact of alopecia in their QoL burden. However, they also reported that quality of life was increasingly influenced by overall ED symptoms. Adult subjects reported that ED cutaneous features alone had no social or emotional impacts and did not influence daily activities. The authors concluded that collaboration between different disciplines was required for ED patients, including dermatology, to provide appropriate skincare and improve ED patients’ symptoms (Pavlis et al., 2010).

1.4 Summary

Following a review of the most relevant literature, it can be appreciated that there is a lack of understanding of the effects of Ectodermal Dysplasia on quality of life and OHRQoL in both adults and children. Further research and understanding of the OHRQoL and its interpretation of Ectodermal subjects is required. If children have sufficient cognitive ability
to understand and answer the relevant questionnaire, their reports are preferred to parental/caregiver reports. However, if that is not possible, parental/caregiver reports can be considered. Comparison of the children and parental/caregiver OHRQoL perspectives is important in comprehensive treatment planning for those subjects, in addition to the determination of factors that impact OHRQoL.

1.5 Statement of the Problem

Following synthesis of the most relevant literature, most of the literature assessed the quality of life or OHRQoL in children or adults with isolated hypodontia, while few examined Ectodermal Dysplasia patients. There is also a lack of data impact exploring if Ectodermal Dysplasia symptoms influence the quality of life of those individuals, and how much each of those symptoms or features influences the quality of life and OHRQoL. Hence, further research is required in this field to provide further evidence and help understand the impact of Ectodermal Dysplasia.

1.6 Aims and Objectives

Aims:

• To report the hypodontia and other dental features prevalence on children with ED.
• To investigate Oral Health Related Quality of Life (OHRQoL) of children with ED based on their self-report.
• To explore the relationship between children and parent (OHRQoL) perceptions.
• To determine the parent’s perception of children with ED.
• To elucidate the experiences of an ED cohort after dental rehabilitation.
• To determine the impact of their dental health on their overall quality of life.
Objectives:

- To quantitatively report the impact of ED on the OHRQoL through a personal perception questionnaire for children and their parents.
- Count the correlation between values of the children and parents’ perception at individual (CPQ)domains.
- To determine the priority and importance of the individual’s dental issues regarding their overall quality of life.
- To evaluate the optimal treatment approaches for this patient population.
- To conduct a structured focus group interview with a number of patients who have undergone dental treatment as adolescents and obtain their insights into the adolescent phase of living with ED.

1.7 Study Hypothesis:

- There is no significant impact of hypodontia and general features of Ectodermal Dysplasia on children’s’ Oral Health Related Quality of Life (OHRQoL).
- There is no difference in the (OHRQoL) scores reported by children with and without hypodontia and general features of Ectodermal Dysplasia.
- There are agreement between children and their parents reported (OHRQoL) scores.
2 Material and Methods

2.1 Introduction

There is increasing evidence that Ectodermal Dysplasia (ED) and hypodontia cause increased levels of anxiety resulting in a decreased Quality of Life (QoL) for children and adolescents who are affected by these conditions (Geirdal et al., 2015b). Oral health-related QoL (OHQoL) studies have also reported increased anxiety levels and limitations in daily physical activity in adults diagnosed with Ectodermal Dysplasia (Mehta et al., 2007, Geirdal et al., 2015b).

A recent study by Hanisch assessed the OHRQoL in 110 German participants who were aged 16 and older and affected by ED. They reported that participants’ OHIP questionnaire scores were 12.23 (SD = 12.39) and 11.79 (SD = 11.08) for females and males respectively. The main influences on their OHIP score were difficulty in finding a dentist to provide the appropriate treatment and dissatisfaction with the health system. These values were greater than the mean German adolescent score (4.09), highlighting the decreased QoL of those living with Ectodermal Dysplasia (Hanisch et al., 2019b).

The objective of a previous study was to contrast the psychological stress levels and QoL between adults with self-reported ED and hypodontia with normative control participants. This study identified increased psychological stress and anxiety levels in the study group which was linked to appearance, removable denture use and limitations of function. In addition, those with self-reported ED signs reported poorer OHRQoL than those with hypodontia, and the ED group reported increased levels of dysfunction and poorer physical abilities for coping with warmer environmental conditions (Saltnes et al., 2017b).

This study aimed to highlight the experience of children and adolescents affected with ED and its impact on their QoL compared to unaffected control participants. The study will also
help elucidate how orofacial anomalies rank among their health complaints and also contrast the adult experience before and after they received oral rehabilitations.

2.2 Ethical Approval

Ethical approval was granted by Tallaght Hospital /St. James’s Hospital Joint Research Ethics Committee (REC) in December 2018 (Appendix 1).

2.3 Research Stages and Planning

The research stages and planning are outlined in the following figures (Figure 2.1; Figure 2.2).

![Figure 2.1 Quantitative Data Collection Planning](image1)

![Figure 2.2 Qualitative Data Collection Planning](image2)
2.4 Quantitative Study

2.4.1 Sampling

2.4.1.1 Determining the Sample Size

Sample size determination is normally required to estimate the number of participants required to reach data saturation. Data saturation is reached when there is sufficient information to replicate the study, and when new information cannot be attained and when further coding is no longer feasible (Ness, 2015). Estimation of the number of participants required for this is dependent on multiple factors (Morse, 2000):

- The scope of the study: the broader the scope, the larger the sample size required to reach data saturation. The scope of this study focused on patients’ and parents’ experiences with Ectodermal Dysplasia.
- Nature of the topic: the topic covered in this study was clear, and information was easily obtained from research participants through questionnaires and semi-structured in-depth interviews according to the study group.
- Study design: including parents in this research provided a source of “shadowed data” (Morse, 2000), while control group involvement provides a source of contrasting data. The focus group provided information about the range of experiences of living with Ectodermal Dysplasia.
- Quality of data: if the quality of data obtained was rich, experiential and data saturation was expected to be achievable.

2.4.1.2 Sampling Method

This study was novel and based on information extracted from the published dental and medical literature; to date no study has explored the same condition with a sufficiently
large sample size that it can be used for sample power calculation. In addition, as Ectodermal Dysplasia is a relatively rare condition, this study aimed to collect as many patients as possible, combined with controls matched in age and gender. The control group was sampled randomly from the undergraduate Dental Science paediatric dentistry clinics according to strict inclusion and exclusion criteria.

In this research project, all participants were patients in the Dublin Dental University Hospital. In addition, the research supervisor knew many of the participants and suggested recruitment of patients with a range of experience, but most importantly, those who were articulate and expressive and willing to share their experience with the interviewer in the qualitative part of this research project.

2.4.2 Inclusion and Exclusion Criteria

**Inclusion criteria for the study group:**

- Confirmed diagnosis of Ectodermal Dysplasia by the referring physician and included in the referral letter to the DDUH.
- Patients aged between 8 and 18 years old and their caregiver.
- Patient or caregiver is able to provide informed consent.

**Exclusion criteria for the study group:**

- Patients who had additional congenital conditions or chronic illness.

**Inclusion criteria for the control group:**

- Medically fit children and adolescents.
- Children and adolescents aged between 8 and 18 years old and their caregiver.
- The patient or caregiver is able to provide informed consent.

**Exclusion criteria for the control group:**
- Children and adolescents who had chronic illnesses or other dental abnormalities.
- Children and adolescents who were undergoing orthodontic treatment.

### 2.4.3 Participant Recruitment

Research participants were recruited from patients attending the Dublin Dental University Hospital for paediatric and restorative treatment assessment/reassessment and from the Irish Ectodermal Dysplasia Society. This recently formed society was contacted in relation to the study and kindly offered to circulate information pertaining to the study to all its members. Members of the society who wished to participate in the study then contacted the dedicated gatekeeper in the DDUH.

Patients were also identified from the Special Dental Needs treatment list in the hospital. Each patient or his/her guardian received, from the gatekeeper, a pack with a Patient Information Leaflet to self-assess their eligibility and interest (Appendices 4 and 5). Each pack contained an Expression of Interest Form so that the invitee could contact the gatekeeper to ask any questions in relation to the study and indicate if they were willing to participate. Where the invitee expressed interest in participation at a later stage, they were asked to meet the investigator, at which time they were asked to provide Informed Consent. The invitees had seven days from receiving the study information before seeking Informed Consent. Any questions regarding the study at this assessment visit were answered by the researchers, and data collected.

If the invitee did not wish to participate in the study, they still received dental treatment at DDUH in the normal manner. Subjects were free to withdraw from the study at any time and this did not affect their clinical care. All patients (Number = 42) with a confirmed diagnosis of Ectodermal Dysplasia and aged under 18 years old, with their caregiver, were
invited to participate. Thirty subjects (71.4%) and their parents agreed to participate in this research. Therefore, the richness of the quality of the data obtained was ensured and the recruitment of caregivers confirmed that all dimensions of each patient’s experience could be explored.

2.4.4 Data Collection Tools

2.4.4.1 Child Oral Health Quality of life Questionnaire

The Child Oral Health Quality of Life (COHQoL) was first developed by Alexandra Jokovic and used to measure health outcomes in clinical and quality of life evaluation studies. The questionnaire comprises three sections: Parental-Caregiver Perceptions Questionnaire (P-CPQ), Child Perceptions Questionnaires (CPQ) (8-10 years) and Child Perceptions Questionnaires (CPQ) (11-14 years). The questionnaire measurement tools are broad, allowing researchers to use them to research various conditions that include dentofacial anomalies, dental caries, and malocclusion (Jokovic et al., 2002, Marshman et al., 2005b). The Child Perceptions Questionnaires (CPQ) for 11-14 years old is a self-administrated questionnaire which has 37 items which cover four health domains: oral symptoms, functional limitations, emotional well-being and social well-being (peer interaction, schooling, and leisure activities) during the previous three months. Each item of the questionnaire has a five-point scale including ‘never’=0; ‘once or twice’=1; ‘sometimes’=2; ‘often’=3; ‘every day or almost every day’=4. The collection of the response score for all items makes an overall CPQ 11-14 score (Jokovic et al., 2002, Marshman et al., 2005b).

The Child Perceptions Questionnaires (CPQ) for 8-10 years old has 25 items investigating impacts in four health domains: oral symptoms, functional limitations, emotional, and social wellbeing. Those items cover contemporary concepts of child health. In addition, the
questionnaire ensured assessment of the extent of the disease or disorder on an individual’s quality of life and was not restricted to the experience that related to the disease (Jokovic et al., 2004c).

Validity and Testing

The Child Perception Questionnaire (CPQ) validity has been tested against global ratings by many studies and found to be suitable for use (Ferreira et al., 2012, O’Brien et al., 2006). However, a study evaluating the CPQ responsiveness and the ability to detect changes over time found that it decreased in suitability when used as a tool for longitudinal studies. One of those studies specified that it was not suitable for longitudinal studies which investigated the effects of orthodontic treatment on quality of life (Do and Spencer, 2008, Marshman et al., 2005b, Olivieri et al., 2013, Abreu et al., 2018).

Limitation

Johal and co-workers (2007) reported that the main limitation of the questionnaire was that it was not condition-specific. The authors stated that it was not a specific tool for malocclusion assessment from an orthodontic perspective. In addition, other studies have showed that the participants’ responses to the questionnaire might be affected by other factors such as social and cultural background, the status of general health, household income, and life stress (Marshman et al., 2005b, Johal et al., 2007, Barbosa et al., 2009).

2.4.4.2 Parent Questionnaire (P-CPQ)

Assessment of child’s quality of life often requires supplemental information from parents or guardians which may be used as a proxy. Parental or guardian proxies have the advantage in terms of their ability to provide information that represents their social and
psychometric understanding of the child’s problems. The Child Oral Health Quality of Life (COHQoL) questionnaire includes the Parental-Caregiver Perceptions Questionnaire (PPQ), which was designed to complement the Child Perceptions Questionnaire (CPQ) and provide supplemental information to that collected via the CPQ. The use of a PPQ is also a tool for investigating levels of agreement between the CPQ and PPQ questionnaires. The Parental-Caregiver Perceptions Questionnaire (PPQ) has 31 questions which cover four domains: oral symptoms, functional limitations, emotional well-being, and social well-being. An additional 14 questions cover the impact of the condition on the family (Family Impact Scale (FIS)). The questionnaire collects the responses using a six-point Likert scale (‘never’=0, ‘once or twice’=1, ‘sometimes’=2, ‘often’=3, ‘every day or almost every day’=4, and ‘don’t know’) according to the frequency of the events that have affected the child over the previous three months. The ‘don’t know’ response has been included in the questionnaire by the author as a result of the awareness of the parent’s knowledge limit for their children feelings and activities (Jokovic et al., 2002, Eiser et al., 2001, Saigal et al., 1996, Jokovic et al., 2003b).

Validity and Testing

Jokovic et al. (2003) have validated this version (long) of the questionnaire, while its short version has been validated in studies that have assessed early childhood dental caries and its impact on quality of life (Jokovic et al., 2003b, Thomson et al., 2013).

2.4.4.3 Data Collection

An electronic version of all questionnaires was used for data collection using SurveyMonkey®. All questionnaires were transferred manually into the SurveyMonkey® database by the principal investigator. This survey method was used to enable the
researcher to handle the data electronically and export the findings to an Excel® sheet (Microsoft Office 365 ProPlus) for data collation, cleaning, and variable manipulation.

In the Prosthodontic Postgraduate Clinic, children and adolescents had a restorative clinical assessment which includes full teeth charting, defining missing and malformed teeth, assessment of current prosthesis if applicable, and radiographic assessment if no recent radiographs available. All of the dental findings had been saved in a password encrypted Salud software in Dublin Dental University Hospital. If patients required treatment or referral to other departments, arrangements were made to complete any required dental care.

During the clinical assessment time, parents/caregivers completed the questionnaire on a password protected iPad (Air 2 version). Subsequently, the children/adolescents were asked to complete the appropriate questionnaire. During the clinical assessment and questionnaire completion, children and their caregivers were kept separate to avoid any potential confounding influence they might have on each other’s responses. Gender and age for each participant were recorded as part of the questionnaire and a specific identification code was assigned by the main researcher to each participant to ensure anonymity and provide data security. A convenient sample of control participants had been recruited randomly from Paedodontic Clinics in Dublin Dental Hospital, each participant was randomly matched by age and gender to the participant from the control group, the randomisation was by a selection of each second patient from the provided list of eligible controls. When more than one participant of the same age and gender was identified, the computed random allocation was applied. Each controlled participant subjected to the same dental assessment and dental setting same as that for the study case subjects, the parents’ of those controlled subjected were asked to answer the relevant questionnaire while their children underwent dental assessment.
2.4.5 Statistical Analysis

Statistical analysis was carried out using SPSS Statistical Package for Social Sciences (SPSSS) version 25 (SPSS Inc., Chicago, USA). The distribution of the independent variables: age, gender, ethnicity, medical sign and symptoms, dental features, number and type of dental prosthesis were summarised by counts and percentages; meanwhile, the distribution of the continuous dependent variables: child perception questionnaire score, parent perception questionnaire score and individual domain score were summarised by means, standard deviations, medians, and interquartile ranges.

Comparisons between the questionnaire scores of participants and their controls, participants and their parents were carried out using the Wilcoxon signed rank test. The Wilcoxon signed rank test is appropriate for detecting differences between two dependent samples when the distribution of the difference between the samples’ means violates the assumption of normality for parametric tests (McDonald, 2014). Associations between questionnaire scores of both children and parents were tested using Spearman’s rank order correlation in two clusters, cases and controls. Spearman’s correlation is appropriate for testing the association between variables in monotonic but non-linear relationships (McDonald, 2014). In all hypothesis testing, the critical p-value for statistical significance was set to 0.05.

2.5 Qualitative Study

2.5.1 Qualitative Interview Design and Topic Guide

A qualitative study was proposed to further investigate the context of what it means to live with Ectodermal Dysplasia (ED) and to explore the effects of both hypodontia and other ED
features on quality of life. Qualitative data were collected through structured in-depth focus group interviews with participants with Ectodermal Dysplasia. Those participants were all adults who had completed full mouth rehabilitation treatment and undergone several phases of restorative treatment as they grew up.

An initial topic guide (Appendix 11) was formulated to elaborate on certain aspects related to the research question. However, participants were encouraged to talk about their experiences, and themes were extracted from transcripts of the interview. The topic guide was formulated based on a literature review of qualitative studies that included patients with congenital diseases. The following online databases were searched: Medline, PubMed, and the Cochrane Library. Research terms used were: qualitative, ectodermal dysplasia, quality of life, experiences, patient, structured interview and hypodontia. The process was supplemented using reference lists in Google Scholar. Selected articles were searched until July 2019. Only articles published in English were considered due to limitations in terms of time and resources. The topic guide was formulated and revised by the research supervisor and an experienced qualitative researcher, Dr Alison Dougall, a Consultant in Medically Complex Patients who conducted the interview and who had extensive experience in qualitative research.

The topic guide was used to elucidate upon certain aspects related to Ectodermal Dysplasia: (1) individual self-knowledge of the condition; (2) living with the condition; (3) social and lifestyle impact of the condition; (4) impact of the condition on the different parts of the body; (5) how much hypodontia impacted on quality of life; (6) comparing how the absence of teeth impacted relative to other ED features; and (7) the importance of hypodontia management for individuals with the condition.
2.5.2 Sampling

Sampling in qualitative research provides flexibility and convenience in terms of participant sampling. In addition, randomly selected sampling in qualitative research requires a purposeful sample, where participants recruited are reflective and willing to share their experiences with the interviewer (Coyne, 1997). Purposeful sampling aims at interviewing participants who have undergone the experience, and their experiences are considered typical in addition to their broad general knowledge of the topic. Moreover, participants with atypical experiences are also sought so that the entire range of experiences is covered, and the breadth of the concept is understood (Coyne, 1997, Morse, 1991, Morse, 2000).

In the current study, all participants were patients in the Dublin Dental University Hospital. In addition, the research supervisor knew the participants and suggested recruitment of patients with a range of experiences, but most importantly, those who were articulate and expressive and willing to share their experience with the interviewer in the qualitative part of this research project.

2.5.3 Inclusion and Exclusion Criteria

Inclusion Criteria:

- Confirmed diagnosis of Ectodermal Dysplasia by the referring physician and included in the referral letter to the DDUH.
- Patients who had completed full mouth rehabilitation.
- Over 18 years of age.
- Able to provide consent.

Exclusion Criteria:

- Participants who had another chronic illness or other dental abnormality.
2.5.4 Participant Recruitment

For the qualitative part, a total of seven patients were invited to participate in the research; four of them responded positively and were willing to attend a focus group session. Those who refused to take part had different reasons: the first had difficulty in commuting to the hospital from another city; the second refused to take part of this research without any reason mentioned, while the third reported that she was not willing to share her personal experience in group discussion-based research.

2.5.5 Data Collection

2.5.5.1 Trial Interview

Before the actual interview was undertaken, a mock interview was conducted with three colleagues and one consultant who had experience of qualitative research and treating patients with Ectodermal Dysplasia. The colleagues were allocated the role of patients during the trial interview while the consultant observed the interview to reflect on the interview topic guide in terms of its nature and fluency.

Interviews were arranged in the setting planned for the definitive interview to ensure that the room seating was suitable, and the recording equipment was sufficiently effective to record the interview. The trial interview was conducted in the exact manner planned for the definitive interview. At the end of the session, a detailed discussion was carried out to provide feedback on the topic guide and questions asked. A small number of items were changed in the topic guide to ensure that the questions were clear and open-ended and encouraged the participants to express their thoughts.
2.5.5.2 Conducting the Interview

Participants were invited to attend the DDUH on a day and time that suited them during the week. They were greeted in the main reception by the main investigator and the interviewer; however, one of the participants could not attend on that day. They were brought to the meeting room where the interview was to be completed. They were offered coffee, tea, and refreshments to ensure the informality of the interview. An introduction about the research project and its aims was provided. Then the interview process was explained to the three participants and informed consent was obtained. Any questions raised were answered before commencing the interview.

2.5.5.3 Transcription

The audiotapes were transcribed using a recognised professional transcription services provider and after a confidentiality agreement was signed in relation to the storage of digital and text information. Participants were given different names in the transcribed data to ensure full anonymity and confidentiality. Data were anonymised and stored on an encrypted and password-protected computer. Once digital recordings had been transcribed and the text documents received by the investigator, all recorded data were deleted.

2.5.6 Qualitative Data Analysis

Following the focus group interview, a transcript of the interview was formatted. The transcript was then converted to a text file and uploaded to qualitative data analysis software (MAXQDA). MAXQDA is the successor of winMAX and was developed and distributed by VERBI Software based in Berlin, Germany. The software is used for
qualitative and mixed methods data content analysis by systematically evaluating and interpreting textual data (Figure 2.3). An objective coding scheme was used to organise the data. Each code was assigned to selected segments of the text. Codes and sub-codes were ordered into a hierarchical structure as in (Figure 2.4).

The software has three main display windows that provide the following features:

1. Document System Window: it offers data import options (text documents, table documents, PDF files, video and audio files and images). The data files are accessed through this window.

2. Code System Window: this allows assignment of a code to the selected part of the document; both code and document part had the same colour code. The name of the code was given to the label attached to it. The codes then have a hierarchical structure and a main code might have several sub-codes.

3. Document Browser Window: this allows visualisation of the selected document where a text segment can be marked by the researcher or code can be attached or memos created.

Data were analysed using a thematic analysis approach as illustrated in (Figure 2.3). This was initiated by a careful reading of the transcript to have sufficient familiarity with the breadth of the topic transcript content. Patterns and meaning within the document text were identified and highlighted for future reference. Then, the initial codes were generated, and the most basic elements of the data defined by the key concept. The codes review carried out at two stages. The first stage included a review of the coding at the level of coded segments and in relation to the entire data set by the main researcher.

At the second stage, the data reviewed independently by the supervisor and an independent experienced qualitative researcher before agreement on the main themes, this ensured both consistency and comprehensiveness in coding and data analysis and
limit any researcher bias. The researcher and both reviewers met subsequently to discuss
the coding, new themes identified and some themes were collapsed into each other and
where controversies between themes or coding were resolved. Lastly, the subsequent
themes were analysed with the code’s organisation into higher-order groups. (Table 2.1)
illustrates the main themes and codes.
Figure 2.3 Focus Group Data Management Order

Figure 2.4 Codes and Sub-codes Systems Assignment
<table>
<thead>
<tr>
<th>Main Themes</th>
<th>Codes</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>The meaning of health</strong></td>
<td>• Mental health definition</td>
</tr>
<tr>
<td></td>
<td>• General health definition</td>
</tr>
<tr>
<td><strong>Challenges of living with Ectodermal Dysplasia</strong></td>
<td>• Undergoing prolonged treatment</td>
</tr>
<tr>
<td></td>
<td>• Limited support and understanding of the condition</td>
</tr>
<tr>
<td></td>
<td>• Small number of specialists in the country</td>
</tr>
<tr>
<td></td>
<td>• The cost of private treatment</td>
</tr>
<tr>
<td></td>
<td>• Medical professionals required for management of the condition</td>
</tr>
<tr>
<td><strong>Impact of ectodermal dysplasia on Quality of Life (QoL)</strong></td>
<td>• Impact on physical activities, sports and being out in general</td>
</tr>
<tr>
<td></td>
<td>• Self-esteem and self-confidence</td>
</tr>
<tr>
<td></td>
<td>• Impact on skin</td>
</tr>
<tr>
<td></td>
<td>• Impact on social relationships</td>
</tr>
<tr>
<td></td>
<td>• Anxiety</td>
</tr>
<tr>
<td></td>
<td>• Having to travel and make arrangements to get treated</td>
</tr>
<tr>
<td></td>
<td>• Small random activities</td>
</tr>
<tr>
<td></td>
<td>• No impact</td>
</tr>
<tr>
<td></td>
<td>• Emerging health issues (Ophthalmic)</td>
</tr>
<tr>
<td></td>
<td>• Impact on eating</td>
</tr>
<tr>
<td></td>
<td>• Impact on speech</td>
</tr>
<tr>
<td></td>
<td>• Impact on work schedule and arrangements</td>
</tr>
<tr>
<td></td>
<td>• Impact on breathing</td>
</tr>
<tr>
<td></td>
<td>• Dry mouth</td>
</tr>
<tr>
<td><strong>Managing the impacts of Ectodermal Dysplasia</strong></td>
<td>• Adaptation aids</td>
</tr>
<tr>
<td></td>
<td>• Managing self-perceptions</td>
</tr>
<tr>
<td></td>
<td>• &quot;Home remedies&quot; and knowing how to behave</td>
</tr>
<tr>
<td></td>
<td>• Networking, socialising and researching the condition</td>
</tr>
</tbody>
</table>
3 Results

3.1 Quantitative Study Results

3.1.1 Introduction

This study had two distinct investigations comprising both qualitative and quantitative sections. The quantitative part will be described firstly in this chapter. A total of 30 eligible children and their caregivers were asked to participate in the study. Seven participants and their parents refused to participate for different reasons. Two of those invited lived far from the study location, one was unable to arrange sufficient child care, whilst the others were not interested in taking part in the research investigation. In total, 23 children and their parents (76%) agreed to participate. Further participants were excluded at the data analysis stages. One participant and his caregiver were excluded because they did not save their related questionnaires in SurveyMonkey®. Another two participants were excluded following the parental perception questionnaire response analysis because their parents’ questionnaire was not complete (Figure 3.1).

3.1.2 Demographics

The demographic data of the participants by age, gender, and ethnicity are illustrated in (Table 3.1). In the final sample, (N = 16, 72.7%) of the children were male in gender and the majority (N = 19, 86.3%) were Caucasian in ethnicity. The mean age of participants was (12.5) years; the youngest participant was 8 years old while the oldest was 18 years old. The control group had the same demographic data in terms of age and gender apart from the fact that they were all Caucasian.
3.1.3 Prevalence of Ectodermal Dysplasia Signs and Symptoms

The main features of Ectodermal Dysplasia (ED) of the participating children were collected during the assessment and illustrated in (Tables 3.2, 3.3). Some of these features were obvious and were recognised by the investigator, for example, sparse hair, and abnormal nail shape, while other features were recognised by asking the participants and their caregivers. Sparse hair and dry skin were common extraoral features; abnormal sweating and eczema were both consistent with the dry skin presentation (Table 3.2). In total, (N = 15, 68.1%) of the participants had a positive family history.

Ectodermal Dysplasia dental features of the participants are illustrated in (Table 3.3). More than two-thirds had Class III facial profiles (N = 14, 63.6%), while in all instances, subjects had missing permanent teeth. This was confirmed clinically and radiographically with a total mean of 18.5 (± 8.69) missing teeth. Severe hypodontia (more than six missing teeth) was the most common dental feature of all participants (N = 15, 68.1%). It was present in three quarters of male participants and half of the female subjects. Moderate and mild hypodontia was reported in (N = 3, 13.6%) and (N = 1, 4.5%) of participants respectively.

Malformed teeth and anodontia were also charted, at a rate of (N = 17, 77.2%) and (N = 3, 13.6%) respectively, with a higher prevalence of mandibular anodontia (N = 6, 27.2%). Almost half of malformed teeth that were located in the incisor region (N = 10, 45.4%) were restored with composite resin restorative material for aesthetic reasons (Table 3.3). Two hypomineralised teeth were charted in one male participant, while a female subject had hypomineralisation in all her erupted permanent teeth (23).

The missing teeth were replaced by an acrylic denture in a third (N = 2, 33.3%) of the females and more than two thirds (N = 11, 68.7%) of male subjects. From the patient histories, in the region of (N = 12, 83.3%) of these dentures had undergone replacement more than three times throughout their childhood (Table 3.3). However, one female
participant who was missing all her permanent teeth radiographically did not have
dentures as all her primary teeth were retained.

*Child and Parent, **Child Perception Questionnaire, ***Parent Perception Questionnaire

![Study Recruitment Flow Chart]

*Child and Parent, **Child Perception Questionnaire, ***Parent Perception Questionnaire

**Figure 3.1 Study Recruitment Flow Chart**
Table 3.1 Sample Demographic

<table>
<thead>
<tr>
<th>Sample (N = 22) (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Age</strong></td>
</tr>
<tr>
<td>8 Years N = 2 (9.1%)</td>
</tr>
<tr>
<td>9 Years N = 2 (9.1%)</td>
</tr>
<tr>
<td>10 Years N = 2 (9.1%)</td>
</tr>
<tr>
<td>11 Years N = 2 (9.1%)</td>
</tr>
<tr>
<td>12 Years N = 4 (18.18%)</td>
</tr>
<tr>
<td>13 Years N = 1 (0%)</td>
</tr>
<tr>
<td>14 Years N = 3 (13.6%)</td>
</tr>
<tr>
<td>15 Years N = 3 (13.6%)</td>
</tr>
<tr>
<td>16 Years N = 1 (4.5%)</td>
</tr>
<tr>
<td>17 Years N = 1 (4.5%)</td>
</tr>
<tr>
<td>18 Years N = 1 (4.5%)</td>
</tr>
<tr>
<td><strong>Gender</strong></td>
</tr>
<tr>
<td>Male N = 16 (72.7%)</td>
</tr>
<tr>
<td>Female N = 6 (27.2%)</td>
</tr>
<tr>
<td><strong>Ethnicity</strong></td>
</tr>
<tr>
<td>Caucasian N = 19 (86.3%)</td>
</tr>
<tr>
<td>Other N = 3 (13.6%)</td>
</tr>
<tr>
<td><strong>Caregiver Respondents</strong></td>
</tr>
<tr>
<td>Father N = 6 (27%)</td>
</tr>
<tr>
<td>Mother N = 16 (72%)</td>
</tr>
<tr>
<td>Other None</td>
</tr>
<tr>
<td><strong>Familty History</strong></td>
</tr>
<tr>
<td>N = 15 (68.1%)</td>
</tr>
</tbody>
</table>
### Table 3.2 Medical Signs and Symptoms of ED Subjects

<table>
<thead>
<tr>
<th>ED Feature</th>
<th>Male (N = 16) (72.7%)</th>
<th>Female (N = 6) (27.2%)</th>
<th>Total (N =22) (100%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Asthma</td>
<td>N = 7 (43.7%)</td>
<td>N = 0 (0%)</td>
<td>N = 7 (43.7%)</td>
</tr>
<tr>
<td>Dry Skin</td>
<td>N = 11 (68.7%)</td>
<td>N = 5 (83.3%)</td>
<td>N = 16 (72.7%)</td>
</tr>
<tr>
<td>Eczema</td>
<td>N = 11 (68.7%)</td>
<td>N = 3 (50%)</td>
<td>N = 14 (63.6%)</td>
</tr>
<tr>
<td>Abnormal Nails</td>
<td>N = 6 (37.5%)</td>
<td>N = 3 (50%)</td>
<td>N = 9 (40.9%)</td>
</tr>
<tr>
<td>Abnormal Sweating</td>
<td>N = 11 (68.7%)</td>
<td>N = 5 (83.3%)</td>
<td>N = 16 (72.7%)</td>
</tr>
<tr>
<td>Sparse Hair</td>
<td>N = 13 (81.2%)</td>
<td>N = 3 (50%)</td>
<td>N = 16 (72.7%)</td>
</tr>
<tr>
<td>Dry Eyes</td>
<td>N = 10 (45.4%)</td>
<td>N = 3 (50%)</td>
<td>N = 13 (81.2%)</td>
</tr>
</tbody>
</table>

### Table 3.3 Dental Features of ED Subjects

<table>
<thead>
<tr>
<th>Dental Feature</th>
<th>Male (N = 16) (72.7%)</th>
<th>Female (N = 6) (27.2%)</th>
<th>Total 22 (100%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Missing Teeth</td>
<td>Mean = 19.19 (± 8.1)</td>
<td>Mean = 16.67 (± 10.6)</td>
<td>Mean = 18.5 (± 8.6)</td>
</tr>
<tr>
<td>Severe Hypodontia</td>
<td>N = 12 (75%)</td>
<td>N = 3 (50%)</td>
<td>N = 15 (68.1%)</td>
</tr>
<tr>
<td>Moderate Hypodontia</td>
<td>N = 1 (6.2%)</td>
<td>N = 2 (33.3%)</td>
<td>N = 3 (13.6%)</td>
</tr>
<tr>
<td>Mild Hypodontia</td>
<td>N = 1 (6.2%)</td>
<td>N = 0 (0%)</td>
<td>N = 1 (4.5%)</td>
</tr>
<tr>
<td>C/C Anodontia</td>
<td>N = 2 (12.5%)</td>
<td>N = 1 (16.6%)</td>
<td>N = 3 (13.6%)</td>
</tr>
<tr>
<td>C/- Anodontia</td>
<td>N = 2 (12.5%)</td>
<td>N = 1 (16.6%)</td>
<td>N = 3 (13.6%)</td>
</tr>
<tr>
<td>-C Anodontia</td>
<td>N = 5 (31.2%)</td>
<td>N = 1 (16.6%)</td>
<td>N = 6 (27.2%)</td>
</tr>
<tr>
<td>Class III Facial Profile</td>
<td>N = 9 (56%)</td>
<td>N = 5 (83.3%)</td>
<td>N = 14 (63.6%)</td>
</tr>
<tr>
<td>Class II Facial Profile</td>
<td>N = 5 (31.2%)</td>
<td>N = 1 (16.6%)</td>
<td>N = 6 (27.2%)</td>
</tr>
<tr>
<td>Class I Facial Profile</td>
<td>N = 2 (12.5%)</td>
<td>N = 2 (33.3%)</td>
<td>N = 4 (18.1%)</td>
</tr>
<tr>
<td>Malformed Teeth</td>
<td>N = 13 (81.2%)</td>
<td>N = 4 (66.6%)</td>
<td>N = 17 (77.2%)</td>
</tr>
<tr>
<td>Aesthetic Restoration</td>
<td>N = 8 (50%)</td>
<td>N = 2 (33.3%)</td>
<td>N = 10 (45.4%)</td>
</tr>
<tr>
<td>Acrylic Denture</td>
<td>N = 11 (68.7%)</td>
<td>N = 2 (33.3%)</td>
<td>N = 13 (59.1%)</td>
</tr>
</tbody>
</table>

C/C: Maxillary and mandibular complete denture, P/P: Maxillary and mandibular partial denture, P/C: Maxillary partial denture and mandibular complete denture, M: Male, F: Female.
Table 3.4 Type and Number of Dental Prostheses

<table>
<thead>
<tr>
<th>Participant</th>
<th>Age</th>
<th>Gender</th>
<th>Type of Denture</th>
<th>Number of Sets</th>
</tr>
</thead>
<tbody>
<tr>
<td>Participant 1</td>
<td>8</td>
<td>M</td>
<td>P/P</td>
<td>3</td>
</tr>
<tr>
<td>Participant 2</td>
<td>9</td>
<td>M</td>
<td>C/C</td>
<td>3</td>
</tr>
<tr>
<td>Participant 3</td>
<td>9</td>
<td>M</td>
<td>P/P</td>
<td>5</td>
</tr>
<tr>
<td>Participant 4</td>
<td>10</td>
<td>M</td>
<td>P/C</td>
<td>3</td>
</tr>
<tr>
<td>Participant 5</td>
<td>10</td>
<td>M</td>
<td>C/C</td>
<td>5</td>
</tr>
<tr>
<td>Participant 6</td>
<td>11</td>
<td>M</td>
<td>P/C</td>
<td>2</td>
</tr>
<tr>
<td>Participant 7</td>
<td>12</td>
<td>M</td>
<td>P/C</td>
<td>2</td>
</tr>
<tr>
<td>Participant 8</td>
<td>12</td>
<td>M</td>
<td>P/P</td>
<td>2</td>
</tr>
<tr>
<td>Participant 9</td>
<td>12</td>
<td>F</td>
<td>C/C</td>
<td>4</td>
</tr>
<tr>
<td>Participant 10</td>
<td>14</td>
<td>M</td>
<td>P/P</td>
<td>3</td>
</tr>
<tr>
<td>Participant 11</td>
<td>15</td>
<td>M</td>
<td>P/P</td>
<td>3</td>
</tr>
<tr>
<td>Participant 12</td>
<td>15</td>
<td>F</td>
<td>C/C</td>
<td>3</td>
</tr>
<tr>
<td>Participant 13</td>
<td>17</td>
<td>M</td>
<td>C/C</td>
<td>5</td>
</tr>
</tbody>
</table>

C/C: Maxillary and mandibular complete denture, P/P: Maxillary and mandibular partial denture, P/C: Maxillary partial denture and mandibular complete denture, M: Male, F: Female.

3.1.4 Comparison of CPQ Scores between ED Population and Controls in the 8-10-year-old Groups

As a result of the data being skewed and hence not normally distributed, Wilcoxon signed ranked tests were used to describe the differences between participant mean scores. The tests indicated that the mean value of the overall and subdomain scores of the ED population were higher than corresponding controls, with the exception of oral symptom domains where scores were higher for control subjects. The descriptive results demonstrated low levels of variation between domain means except in the functional limitation domain where ED population score values were double that of controls (10.1, SD = 4.2) and (5.8, SD = 0.9) respectively. However, the discrepancy in the values between
cases and control was not statistically significant ($Z = -1.8, P = .05$). In addition, functional
limitation domain variation did not influence the overall CPQ score, which was not
statistically significant ($Z = -1.7, P = .07$) (Table 3.5).

Table 3.5 Comparison of CPQ Scores (8-10) Age Group

<table>
<thead>
<tr>
<th>Domain</th>
<th>Controls</th>
<th>Cases</th>
<th>Statistics Values</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Mean (SD)</td>
<td>Median (IQR)</td>
<td>Mean (SD)</td>
</tr>
<tr>
<td>Oral Symptoms</td>
<td>10 (2.1)</td>
<td>9 (3)</td>
<td>9.6 (3.01)</td>
</tr>
<tr>
<td>Functional Limitation</td>
<td>5.8 (0.9)</td>
<td>5.5 (2)</td>
<td>10.1 (4.2)</td>
</tr>
<tr>
<td>Emotional Well-being</td>
<td>6 (0.8)</td>
<td>6 (2)</td>
<td>8 (3.6)</td>
</tr>
<tr>
<td>Social Well-being</td>
<td>11.5 (1.6)</td>
<td>11 (3)</td>
<td>12.3 (1.8)</td>
</tr>
<tr>
<td>CPQ Sum</td>
<td>33.3 (4.7)</td>
<td>33 (7)</td>
<td>40.1 (9.8)</td>
</tr>
</tbody>
</table>

b. Based on positive ranks. c. Based on negative ranks.

3.1.5 Comparison of CPQ Scores between ED Subjects and Controls in the 11-14-year-old Group

A Wilcoxon signed ranked test was used to report the differences between the participants’
scores which indicated that the values of the overall and subdomains scores of the ED
subjects were higher than the controls (Table 3.6). This difference was statistically
significant for overall CPQ score ($Z = -2.1, P = .03$), and for all other subdomains except the
social well-being domain where the differences were not statistically significant ($Z = -1.7, P
= .07$), which indicates less effect from this domain in the subjects with hypodontia and ED
(Table 3.6).
### Table 3.6 Comparison of CPQ Scores (11-14) Age Group

<table>
<thead>
<tr>
<th>Domain</th>
<th>Controls</th>
<th>Cases</th>
<th>Statistics Values</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Mean (SD)</td>
<td>Median (IQR)</td>
<td>Mean (SD)</td>
</tr>
<tr>
<td>Oral Symptoms</td>
<td>11.1 (2.3)</td>
<td>11.5 (4)</td>
<td>13.4 (3.5)</td>
</tr>
<tr>
<td>Functional Limitation</td>
<td>12.7 (4.07)</td>
<td>12 (6)</td>
<td>19.1 (7.1)</td>
</tr>
<tr>
<td>Emotional Well-being</td>
<td>12.5 (4.7)</td>
<td>10 (5)</td>
<td>21.1 (12.3)</td>
</tr>
<tr>
<td>Social Well-being</td>
<td>17.3 (6.9)</td>
<td>15 (5)</td>
<td>21.5 (7.3)</td>
</tr>
<tr>
<td>CPQ Sum</td>
<td>53.7 (16.1)</td>
<td>48 (11)</td>
<td>75.3 (27.3)</td>
</tr>
</tbody>
</table>

b. Based on positive ranks. c. Based on negative ranks.

#### 3.1.6 Comparison of P-CPQ Scores between Cases and Control in 8-10-year-old Group

The Wilcoxon signed ranked test identified that the mean values for the overall and subdomains scores of the parents of ED subjects were higher than the parents of controls (Table 3.7). These differences were statistically significant for the overall CPQ score ($Z = -2.03$, $P = .04$) and for the functional limitations and family impact scale domains ($Z = -2.03$, $P = .04$) and ($Z = -2.02$, $P = .04$) respectively. The discrepancy of the other three subdomain scores (social well-being, oral symptom domains, and emotional well-being) for the ED subject’s parents and control parents were not statistically significant. This might be an
indicator of less influence of these three domains in the ED subjects’ families from the parents’ perspective (Table 3.7).

Table 3.7 Comparison of P-CPQ Scores between ED Subjects and Controls in the 8-10-year-old Group

<table>
<thead>
<tr>
<th>Domain</th>
<th>Control</th>
<th>Cases</th>
<th>Statistics Values</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Mean (SD)</td>
<td>Median (IQR)</td>
<td>Mean (SD)</td>
</tr>
<tr>
<td>Oral Symptoms</td>
<td>9.64 (2.7)</td>
<td>8 (5.1)</td>
<td>11.2 (1.3)</td>
</tr>
<tr>
<td>Functional Limitation</td>
<td>11.9 (5.7)</td>
<td>10 (7.8)</td>
<td>19 (5.3)</td>
</tr>
<tr>
<td>Emotional Well-being</td>
<td>10.04 (2.1)</td>
<td>10.2 (4)</td>
<td>13.4 (3.9)</td>
</tr>
<tr>
<td>Social Well-being</td>
<td>11.6 (1.3)</td>
<td>11 (2)</td>
<td>17.4 (7.5)</td>
</tr>
<tr>
<td>Family Impact Scale</td>
<td>16.6 (3.2)</td>
<td>15 (6)</td>
<td>25.4 (5.3)</td>
</tr>
<tr>
<td>CPQ Sum</td>
<td>59.7 (12.9)</td>
<td>57 (20)</td>
<td>86.5 (16.2)</td>
</tr>
</tbody>
</table>

b. Based on positive ranks. c. Based on negative ranks.

3.1.7 Comparison of P-CPQ Scores between ED Subjects and Controls in the 11-14-year-old Group

A Wilcoxon signed ranked test was applied to describe the difference between the parents of the ED subjects and control values which indicated that the mean values for the overall domains were higher for the responses of ED subjects’ parents compared to controls (98.04, SD = 42.02 and 58.6, SD = 10.8 respectively). In addition, the subdomains scores of the parents of ED subjects were higher than the parents of controls (Table 3.8). The differences were not statistically significant in the oral symptoms domain, only (Z = -1.4, P = .15). The other subdomains and overall CPQ scores were statistically significant, which
indicated a greater influence of these domains in the ED children and their families from the caregiver point of view (Table 3.8).

Table 3.8 Comparison of P-CPQ Scores between ED Subjects and Control in the 11-14-year-old Group

<table>
<thead>
<tr>
<th>Domain</th>
<th>Control</th>
<th>Cases</th>
<th>Statistics Values</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Mean (SD)</td>
<td>Median (IQR)</td>
<td>Z</td>
</tr>
<tr>
<td>Oral Symptoms</td>
<td>9.4 (2.47)</td>
<td>12.2 (5.7)</td>
<td>-1.4b</td>
</tr>
<tr>
<td>Functional Limitation</td>
<td>10.5 (2.7)</td>
<td>20.8 (8.7)</td>
<td>-2.7b</td>
</tr>
<tr>
<td>Emotional Well-being</td>
<td>9.9 (1.8)</td>
<td>15.9 (8.8)</td>
<td>-2.4b</td>
</tr>
<tr>
<td>Social Well-being</td>
<td>11.7 (1.2)</td>
<td>21.6 (11.4)</td>
<td>-2.8b</td>
</tr>
<tr>
<td>Family Impact Scale</td>
<td>16.8 (5.1)</td>
<td>27.3 (10.8)</td>
<td>-2.6b</td>
</tr>
<tr>
<td>CPQ Sum</td>
<td>58.6 (10.8)</td>
<td>98.04 (42.02)</td>
<td>-2.7b</td>
</tr>
</tbody>
</table>

b. Based on positive ranks. c. Based on negative ranks.

3.1.8 Comparison of Children and their Parents’ Perception in the 8-10-year-old Age Group

Table 4.9 displays the descriptive values for each domain in the ED patient group. However, it does not include the domains’ total as the parent total response includes the Family Impact Scale domain which is not in the child version. Parental perception scores were higher in all domains than their children, and the differences were statistically significant in two of the four domains based on Wilcoxon signed ranked test values (Table 3.9).

A similar pattern for value discrepancy is displayed in (Table 3.10), where the parents of the control group of children reported higher values than their children in all domains.
Using the Wilcoxon test, the differences between children and parents were significant for both the emotional well-being ($Z = -2.02$, $P = .04$) and functional limitation domains ($Z = -2.02$, $P = .04$).

**Table 3.9 Comparison of ED Subjects and their Parents’ Perception (8-10 Years)**

<table>
<thead>
<tr>
<th>Domain</th>
<th>Child</th>
<th>Parent</th>
<th>Statistics Values</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Mean (SD)</td>
<td>Median (IQR)</td>
<td>Mean (SD)</td>
</tr>
<tr>
<td>Oral Symptoms</td>
<td>8.8 (2.3)</td>
<td>10 (4)</td>
<td>11.2 (1.3)</td>
</tr>
<tr>
<td>Functional Limitation</td>
<td>9.2 (3.8)</td>
<td>8 (7)</td>
<td>19 (5.3)</td>
</tr>
<tr>
<td>Emotional Well-being</td>
<td>6.6 (1.5)</td>
<td>7 (3)</td>
<td>13.4 (3.9)</td>
</tr>
<tr>
<td>Social Well-being</td>
<td>12.6 (1.9)</td>
<td>12 (4)</td>
<td>17.4 (7.5)</td>
</tr>
</tbody>
</table>

<sup>b</sup>. Based on negative ranks.

**Table 3.10 Comparison of Control Subjects and their Parents’ Perception (8-10 Years)**

<table>
<thead>
<tr>
<th>Domain</th>
<th>Child</th>
<th>Parent</th>
<th>Statistics Values</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Mean (SD)</td>
<td>Median (IQR)</td>
<td>Mean (SD)</td>
</tr>
<tr>
<td>Oral Symptoms</td>
<td>9.2 (1.09)</td>
<td>9 (2)</td>
<td>9.6 (2.7)</td>
</tr>
<tr>
<td>Functional Limitation</td>
<td>5.6 (0.8)</td>
<td>5 (2)</td>
<td>11.9 (5.7)</td>
</tr>
<tr>
<td>Emotional Well-being</td>
<td>5.8 (0.8)</td>
<td>6 (2)</td>
<td>10.04 (2.1)</td>
</tr>
<tr>
<td>Social Well-being</td>
<td>11 (1.2)</td>
<td>11 (2)</td>
<td>11.6 (1.3)</td>
</tr>
</tbody>
</table>

<sup>b</sup>. Based on positive ranks. <sup>c</sup>. Based on negative ranks.
3.1.9 Comparison of ED Children and their Parents’ Perception in the 11-14 Age Group

Differences between the descriptive values of ED children and their parents in the 11-14 group are displayed in (Table 3.11). In contrast to the 8-10 age group the children’s scores were higher than that of their parents’, except in the functional limitation domain. The parental score was 20.8 (SD = 8.7) and the ED children’s’ mean score was 19.6 (SD = 7.04). None of the four domain value differences between parents and their ED children were statistically different (Table 3.11).

<table>
<thead>
<tr>
<th>Domain</th>
<th>Child</th>
<th>Parent</th>
<th>Statistics Values</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Mean (SD)</td>
<td>Median (IQR)</td>
<td>Mean (SD)</td>
</tr>
<tr>
<td>Oral Symptoms</td>
<td>13.3 (3.6)</td>
<td>14 (5)</td>
<td>12.2 (5.7)</td>
</tr>
<tr>
<td>Functional Limitation</td>
<td>19.6 (7.04)</td>
<td>17 (13)</td>
<td>20.8 (8.7)</td>
</tr>
<tr>
<td>Emotional Well-being</td>
<td>21.6 (12.6)</td>
<td>18 (18)</td>
<td>15.9 (8.8)</td>
</tr>
<tr>
<td>Social Well-being</td>
<td>21.8 (7.5)</td>
<td>18 (10)</td>
<td>21.6 (11.4)</td>
</tr>
</tbody>
</table>

*Based on negative ranks.*

In the same age group, comparison between parents’ and children’s perception in the control group revealed the same pattern as for the ED group. In other words, the children reported higher scores than their parents (Table 3.12). The difference between the reported scores was significant only in two domains: social well-being ($Z = -3.4, P = .001$), and emotional well-being ($Z = -2.2, P = .025$). Interestingly, when compared to the 8-10 age group, children in both the ED group and controls reported higher scores than their
parents, which reflected the children’s increased awareness of each domain as they get older, in addition to the limited parental awareness of their children’s well-being in all domains.

<table>
<thead>
<tr>
<th>Domain</th>
<th>Child</th>
<th>Parent</th>
<th>Z</th>
<th>p-Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Oral Symptoms</td>
<td>11 (2.3)</td>
<td>9.4 (2.4)</td>
<td>-1.6b</td>
<td>.107</td>
</tr>
<tr>
<td>Functional Limitation</td>
<td>12.8 (4.1)</td>
<td>10.5 (2.7)</td>
<td>-1.6b</td>
<td>.102</td>
</tr>
<tr>
<td>Emotional Well-being</td>
<td>12.6 (4.8)</td>
<td>9.9 (1.8)</td>
<td>-2.2b</td>
<td>.025</td>
</tr>
<tr>
<td>Social Well-being</td>
<td>17.6 (7.08)</td>
<td>11.7 (1.2)</td>
<td>-3.4b</td>
<td>.001</td>
</tr>
</tbody>
</table>

b. Based on positive ranks.

3.1.10 Association between Child and Parental Perceptions

To explore correlations visually, parent scores were plotted against their child’s scores. In all domains, linearity between parent and child scores was poor. Amongst oral symptoms and social well-being domains, a weak positive association was noted. Among controls, no pattern was discerned (Figures 3.2, 3.5). The same weak positive association for the functional limitation domain were visualised between parents and children among the cases, while there was a negative association between parents and children in the control group (Figures 3.3). However, in the emotional well-being domain and in the total CPQ score plot, the association was weak and positive in both cases and controls, between parents’ and children’s scores (Figures 3.4, 3.6).

Based on that finding a Spearman’s test was run to examine the correlation values in two clusters: ED patients and controls. The correlation test for the control groups (n = 20)
demonstrated a very weak positive correlation between children’s’ and their parents’ response in the functional limitation domain ($r_s = 0.1, n = 20, p = 0.459$), while in the other domains, children’s’ responses had weak positive relations. None of these correlations was statistically significant (Table 3.13).

Table 3.13 Correlations between Control Participants and Parental Scores

<table>
<thead>
<tr>
<th>Age Group</th>
<th>Cases or Controls</th>
<th>Domain</th>
<th>Spearman’s rho</th>
<th>p-Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>8-10 and 11-14</td>
<td>Controls</td>
<td>Oral Symptoms</td>
<td>0.03</td>
<td>0.887</td>
</tr>
<tr>
<td>8-10 and 11-14</td>
<td>Controls</td>
<td>Functional Limitations</td>
<td>0.1</td>
<td>0.459</td>
</tr>
<tr>
<td>8-10 and 11-14</td>
<td>Controls</td>
<td>Emotional Well-being</td>
<td>0.2</td>
<td>0.365</td>
</tr>
<tr>
<td>8-10 and 11-14</td>
<td>Controls</td>
<td>Social Well-being</td>
<td>0.3</td>
<td>0.157</td>
</tr>
</tbody>
</table>

On the other hand, the Spearman’s correlation test run for the ED patient group (n = 20) showed a very weak negative relationship between the children and their parents’ response in the emotional well-being domain ($r_s = -0.07, n = 20, p = 0.743$) (Table 3.14). Other domain responses demonstrated a weak positive relationship between both children’s and parents’ responses. This indicated a positive influence of Ectodermal Dysplasia on the parents’ responses, which were primarily influenced by the children’s responses. However, these correlations were not statistically significant (Table 3.14).

Table 3.14 Correlations between ED Subjects and Parental Scores

<table>
<thead>
<tr>
<th>Age Group</th>
<th>Cases or Controls</th>
<th>Domain</th>
<th>Spearman’s rho</th>
<th>p-Value</th>
</tr>
</thead>
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<tr>
<td>8-10 and 11-14</td>
<td>Cases</td>
<td>Oral Symptoms</td>
<td>0.3</td>
<td>0.154</td>
</tr>
<tr>
<td>8-10 and 11-14</td>
<td>Cases</td>
<td>Functional Limitations</td>
<td>0.2</td>
<td>0.315</td>
</tr>
<tr>
<td>8-10 and 11-14</td>
<td>Cases</td>
<td>Emotional Well-being</td>
<td>-0.07</td>
<td>0.743</td>
</tr>
<tr>
<td>8-10 and 11-14</td>
<td>Cases</td>
<td>Social Well-being</td>
<td>0.1</td>
<td>0.469</td>
</tr>
</tbody>
</table>
Figure 3.2 Scatterplot of Parents’ Oral Symptom Domain

Children’s Oral Symptoms

Parents’ Oral Symptoms Score vs. Children’s Oral Symptoms Score

Figure 3.3 Scatter Plot of the Functional Limitation Domain

Parents’ Functional Limitation Score vs. Children’s Functional Limitation Score

Children’s Functional Limitation Score
Figure 3.4 Scatter Plot of the Emotional Well-being Domain

Figure 3.5 Scatter Plot of the Social Well-being Domain
Figure 3.6 Scatter Plot of the Sum of all Domains
3.2 Qualitative Study Results

3.2.1 Understanding General and Mental Health Concepts

At the initial stages of the discussion, participants discussed the general topic of health and what “health” meant to them. Although this was seemingly not directly relevant to the topic of Ectodermal Dysplasia, these discussions provided additional insight into their beliefs about what it means to be healthy and introduced the topic for later evaluation of the impact of the condition on their lives.

When providing a definition of “good health” Participant 2’s belief was that “good health” is something “you don’t notice”, and “it doesn’t affect your everyday life”, and Participant 1 further explained, “you go to open a door and if the door handle is where you expect it, you push the door and you don’t notice that”. Other participants agreed, with Participant 1 believing that if something has an impact on daily life and routine, “you’re not fit for much”, and Participant 3 noted that being in good health means “feeling comfortable in yourself, just being able to do whatever you want to do, and being fit and healthy”. Participant 2 added “having no health condition that doesn’t impact your daily life routine”.

The idea of “feeling comfortable in yourself” also led to the participants’ definition of “good mental health” which referred to resilience and was defined by them as “being able to deal with things like stress”. All participants believed that good mental health involved resilience and being able to cope well with stress and challenges. All participants also highlighted that a “balanced nutritional diet” is important for general health. As shown further below, both notions (good health and good mental health) are relevant to the participants’ beliefs about the impact of Ectodermal Dysplasia on their quality of life. They evaluated this impact according to the extent to which it affected their daily routines and mental “self-perceptions” were among their principle ways to manage this impact.
3.2.2 Diagnosis of Ectodermal Dysplasia

Participants discussed their different experiences in how their condition was diagnosed and the challenges of having the diagnosis. Diagnosis age and location were variable. Participant 1 was diagnosed “in the children’s hospital around the age of two or three”, while Participant 2 stated that he did not become aware of the condition until he was around 7 years of age as his parents identified him as being the same as his siblings “I was one of six kids and my parents reared me the same as anyone else, like I was never told”. In addition, he stated “I went to dentist in Dublin instead of the dentist at home”. Around the same age (6-7 years), the third participant received his diagnosis in a children’s hospital when he was seeking treatment for asthma. However, he stated “I didn’t see that unusual. I thought that was just like everybody did”, when he referred to the condition’s general features and appearance.

Unlike dental treatment, diagnosis or management of dry skin did not occur until a much later age “I didn’t go to dermatologist until I was twenty years old”. Participant 3 emphasised the importance of the teeth saying that his family sought dental intervention early in life in contrast to the skin features where they sought a dermatological diagnosis and management at a much older age. At a young age, the participant’s familiarity with the condition was vague “it comes out of the blue”, which reflected the lack of knowledge and support of participants’ families to achieve the diagnosis. However, Participant 1 acknowledged the family factors involved in the condition and that it is an inherited condition “I have a nephew that has it, yeah, my sister a carrier too”.

3.2.3 Challenges of Living with Ectodermal Dysplasia

The participants discussed various challenges of living with ectodermal dysplasia, some of which seemed to have an influence on their quality of life.
3.2.3.1 Undergoing Prolonged Treatment

The main challenge that they discussed was *undergoing prolonged treatment* (coded 22 times) which is inevitable for people with this condition. Although the participants understood that this is necessary, some of them found the process frustrating, with Participant 3, whose treatment lasted “thirteen to fourteen years”, mentioning that he “thought it would never end”. This treatment involved frequent travel and arrangements at inconvenient times and places, which is later discussed in the discussion on the impact of Ectodermal Dysplasia on participants’ lives.

3.2.3.2 Limited Support and Understanding of the Condition (Achieving Diagnosis)

Another challenge of living with ectodermal dysplasia, and arguably a factor that negatively influenced the prolonged treatment, was the *limited support and understanding of the condition*. Because the condition is “very uncommon” (Participant 1), “there are not many resources available”, and the participants reported having to rely on “trial and error” (Participant 3) and trying to figure out “what works here” (Participant 2). When reflecting on his experiences and how they may have been improved, Participant 3 believed that simply understanding the process of comprehensive treatment and “knowing where you’re going” and knowing “where exactly like the finished product would be” would have improved this experience.
3.2.3.3 Difficulty with Diagnosis

The problem with limited support and understanding of the condition referred to the doctors’ limited knowledge of Ectodermal Dysplasia that, again, stems from the rarity of the condition. Participant 3 explained, for example, that there are “twenty or thirty people in Ireland with it”, and because of this, the doctors to whom he was initially taken “didn’t know much about Ectodermal Dysplasia”. Participant 1, who believed he was “the third person in Ireland diagnosed with it”, also reported similar experiences (“It was like, you have what? Oh, hang on a second…”).

3.2.3.4 Small Number of Specialists in the Country

Another challenge of living with Ectodermal Dysplasia is the small number of specialists in the country, which resulted in people affected with Ectodermal Dysplasia having to travel in Ireland or to another country to access specialist treatment. There were also various medical professionals required for management of the condition, and the participants mentioned that there was a need for organised teams consisting of a number of professionals including dentists, ophthalmologists and dermatologists who could jointly successfully help the patients manage their condition. In addition, Participant 2 also raised concerns that because of how rare Ectodermal Dysplasia is, the cost of private treatment is high, and “it would probably cost an absolute fortune”.

3.2.4 The Impact of Ectodermal Dysplasia on Quality of Life

Interestingly, although all participants discussed various ways in which Ectodermal Dysplasia influenced their lives, they also all made statements that were coded as no impact. This is not to say, however, that Ectodermal Dysplasia did not, in fact, have any impact on their lives, but rather that some of the symptoms had less impact than others,
because at this age, they had lived with the condition for a minimum of 18 years and had been able to modify their daily life to feel comfortable with the condition’s symptoms. Another reason is that, as previously discussed, their definition of a “good” health generally involved “feeling comfortable in yourself, just being able to do whatever you want to do” (Participant 3), and it seems that, overall, they were, in fact, mostly able to do what they wanted to do, although they had to undertake planned precautions. As they were familiar and competent with self-management, they did not consider their ED to be affecting their lives “on a day to day anymore” (Participant 2). Participant 3, for example, explained that although he needed to wear dentures, “you can pretty much live your day-to-day life”. Similarly, Participant 1 explained that the only effect of his dentition was that he “went to the dentist in Dublin instead of the dentist at home” as a result of the previously mentioned lack of dental professionals experienced in the management of Ectodermal Dysplasia.

3.2.5 Impact of Physical Activities, Sports and Being Active

Although they may have acclimatised to certain limitations stemming from their condition, Ectodermal Dysplasia certainly did have an impact on their lives. Impact of physical activities, sport and being out in general were the most discussed topics, and these were most frequently linked to the skin condition resulting from Ectodermal Dysplasia. Inability to sweat and control body heat was a challenge that had a significant impact “the heat is a killer” (Participant 2). This involved not being able to participate in sports events and other outdoor activities due to becoming easily overheated even when trying to play and going against advice, which Participant 1 believed may have “cost me the chance to play sports professionally”. This was most evident during the summer where Participant 2 agreed with that, as he had bad experiences with heat control whilst on summer holidays if the
temperature was high: “I would just die”. Another problem with playing sports, as Participant 1 explained, was that his dry hands that made it difficult to hurl. It also affected other activities, such as going out to a public house or being on vacation, and often resulted in “getting agitated” and “a panic mode” (Participant 3) upon realising that they needed to cool down quickly. This, as discussed below, also linked to increased anxiety, and even to emerging health issues (Ophthalmic), such as in the case of Participant 3 who felt that frequent “stepping outside” during parties and gatherings encouraged him to smoke tobacco as a socially acceptable reason to go outdoors.

3.2.6 Impact on Self-esteem and Self-confidence

Unsurprisingly, the impacts on self-esteem and self-confidence were also discussed at length. Two comments related to the impact of sparse hair in that “you’re seventeen or eighteen all the boys are getting highlights in the hair and stuff like that and you’re saying ‘Jesus, I’d love to get that’” (Participant 2) and “when you’re growing up you’re pretty much half-bald, people are kind not bullying but you know what I mean it’s the usual teenage banter”. The reminder of the discussion where self-esteem and self-confidence were involved pertained to dental issues. For Participant 3, his comments related to the impact of his teeth as in the following exchange:

4: Okay. So, do you think teeth is important for self-esteem? Do you think?
3: Oh absolutely, yeah.
2: Of course.
4: Really?
2: Hundred per cent.
4: In what way?
2: It’s the first thing I see on everybody.
3: If you were to,
4: Really?
2: Yeah.
3: if you were to meet somebody and just
2: Yeah.
3: like look at somebody
4: Yeah.
3: and they’re with a smile,
2: Yeah.
3: you notice straight away if they’re missing a tooth or if they’re...
4: Okay.
1: People are bald, people are, big noses and small noses and whatever, but everyone
has teeth like, do you know,
4: Okay.
1: barring,
4: Yeah.
1: eh, if you were,
4: So, you think it...
4: Yeah, so you think it’s more than just eating.
1: [Oh, it so, of course. It’s appearance.]
2: [Oh yeah.]
4: Really?
2: Yeah.
1: I, I would eat no bother like, it’s tough but I,
2: Yeah.
1: if you told me I had go home and eat with no teeth every day I’d do it. Something
like that, but no, it’s pure appearance.

All participants’ recollections in relation to their teeth impacted on self-esteem, mostly
because of not being able to smile, or because of additional problems with the dentures,
such as caps that fell off or dentures moving, which “you can’t really put it to the back of
your mind because even when you’re talking, it’s slipping, you’re trying to put it to the back,
to the top of your mouth” (Participant 2). However, once that dental issue was sorted the participant reported a rapid return in self-esteem “walking out you would think you were a new person”. As participant 2 explained when referring to his previously mentioned definition of good mental health, “being comfortable with yourself” is crucial and “teeth play such a big issue”.

3.2.7 Impact on Social Relationships

Understandably, the impact on self-esteem and self-confidence also directly links to impact on social relationships, and here, again, mostly dental issues were discussed. The participants observed that teeth were “the first thing I see on everybody” (Participant 2), and that the condition affected self-confidence and resulted in withdrawal from socialising with others. Participant 1 emphasised that by relating his parents’ advice at a young age “Don’t tell anybody about your teeth” to avoid drawing attention to his dental issues.

3.2.8 Impact on Anxiety

Anxiety was another effect discussed that linked to the previously discussed topics, and it related to both dental and dermatological conditions associated with Ectodermal Dysplasia. The participants discussed the anxiety linked to the previously mentioned “panic” when needing to quickly cool down, as well as being worried about displacing a denture on a night out and the general anxiety which accompanied the previously discussed social communication. In addition, dental restorations discussed by a participant, including caps, were a source of stress as a result of his self-consciousness and constant efforts to avoid caps displacing in front of children. On further questioning “caps” appeared to refer to direct composite resin restorations placed on conical teeth to restore normal
tooth form. Adaptation to dentures and being comfortable while eating was discussed: “you are trying to make sure it doesn’t fall down and doesn’t really want other people to notice it” and this confidence reflected the mental health concept that was defined earlier as “feeling comfortable in yourself”.

Considering the previous discussion of the limited number of health professionals specialising in Ectodermal Dysplasia, it is not surprising that having to travel and make arrangements to get treated was another impact of the condition on the participants’ lives, and this was discussed in terms of the inconvenience of making alternative arrangements at work in order to facilitate travel for treatment.

3.2.9 The Impact on Skin

This topic was extensively discussed, and the participants raised concerns about having “really, really dry skin” and “the skin layer being quite thin as well” (Participant 3). They provided a number of examples of small, random activities that were affected, as in the following extract:

2: Or here, dealing cards. Nightmare.
4: Dealing cards?
3: Yeah.
2: Nightmare.
4: Really?
2: Yeah.
4: In what ways?
2: I don’t know why I thought of that just there. [laughter]
4: Yeah, really?
2: Yeah, because you’re so dry.
1: Sure, there’s no moisture.
2: Yeah, there’s no grip at all, it’s like picking up a page here.
4: Yeah, so picking things up and dropping things.
2: Emm, it’s more like that.
2: [It’s more like that, like] if something’s shiny and you’re trying to pick it and you’re trying to just move it, like.
4: Ah.
3: Opening, like with plastic bags, like
2: Yeah.
3: trying to...
4: Oh, really? So that’s, [that’s tricky? Okay, interesting.]
1: Or at the airport actually, standing at the airport,
1: Yeah.
1: I don’t think I have fingerprints, like.
3: Oh yeah.
2: Oh yeah, fingerprints are really, like, that actually, like when I was going over to Japan, they were like, they had to do all the visa stuff
1: Yeah.
2: when you get there,
3: Yeah.
2: and the guy, after like fifteen minutes just like,
1: Go on.
2: go on like.
4: Yeah
4: Really?
2: I remember at the American embassy too,
4: So, can you...
2: it was like harder sir, harder.
3: Yeah.
2: And like, I’m going as hard as I can, pal, I’m gonna go through the plate.

In this category, the impact of thin hair and hair loss (baldness) and its resultant anxiety and bullying at a young age were discussed under the social relationship context “you’re seventeen or eighteen all the boys are getting highlights in the hair and stuff like that and
you’re saying ‘Jesus, I’d love to get that’” (Participant 2). “My hair was always like super-thin since I was young and I started getting a bald patch in the middle of my head” another participant reported.

3.2.10 Emerging Health Issues

Emerging health issues were also discussed. As previously noted, for example, Participant 2 believed that his skin condition, as a result of heat intolerance, resulted in him often stepping outside with smokers during various parties which, as a result, “encouraged me to be a smoker which is awful”. Participant 2, in turn, described problems with breathing linked to his asthma or having a blocked airway that participants experienced at young ages. Participant 3 described an eye infection resulting from dry eye and “ingrown eyelashes” adding that “dry eye and an ingrown eyelash almost resulted in loss of one of my eyes”. He described how a small infection in his eye, caused by dryness, resulted in a serious infection that has resulted in near total sight loss in that eye. The other participants were unaware of the risks of decreased lacrimal gland function. They questioned why this had never happened to them and why they were never informed by medical practitioners about this potential risk.

3.2.11 Impact on Eating and Speech

Two effects of Ectodermal Dysplasia that were directly related to the dental condition were the impact on eating and impact on speech. Regarding eating, it was more difficult to chew, which they reported led to indigestion and was generally inconvenient. This linked to the participants’ earlier definitions of good health, as they believed their dental issues prevented them from “normal” functioning:
2: I think dental is something that’s like very like forward-facing, it’s very, you know, and it’s, it’s something that like, if you get a, you know, you’re chewing a piece of food, you know,

3: Yeah.

2: if your teeth, if you hit like, like it literally stops you, like you were saying quality of life earlier on,

4: Yeah.

2: that’s something that would affect, you know,

3: Yeah.

2: if you can’t, you can’t eat food properly then, that’s poor.

Participant 1 agreed with that sentiment “well obviously mastication and chewing food is like important for breaking down, before it gets into your stomach, and therefore it I wasn’t wearing my dentures for a day”. Participant 3 stated “since I had a denture made, I would always have the bottom one now for eating” which highlighted the impact of missing teeth on chewing function, and how using the denture influenced the ability to eat and chew. The condition also affected speech, “because a lot of the time you’re like spitting if you don’t have the denture in your mouth” (Participant 3), “because there’s obviously air gaps” (Participant 1) that affected clear speech, and decreased confidence in being able to hold a conversation. In addition, the ability to talk was influenced by the fit of the denture at certain ages as the participants were actively growing in the adolescent years “my caps kept falling off, which meant that the denture wasn’t fitting right” which overlapped with the condition’s impact on social relationships and anxiety.
3.2.12 Impact on Dental Aesthetics and General Appearance

The impact of Ectodermal Dysplasia on general appearance was discussed in relation to facial appearance, hair loss, and dental issues. The face was highlighted only by Participant 2 “the nose (...) it doesn’t give the symmetry to my face” which reflected the importance of full-face features while managing and treating this patient cohort. The impact of the hair was discussed earlier under the self-esteem category, but it impacted more in younger years as a source of teenage bullying.

The impact of the dentition overlapped from different perspectives including socialising and function. But in these contexts the importance of teeth for general appearance was emphasised by Participant 2 “dental is something that’s like very like forward-facing” which represented the dental factors in the whole face picture or appearance of each person: “if your teeth, if you hit like, like it literally stops you” (Participant 2). The dental impact on general appearance in contrast to other facial features was discussed “people are bald, people are, big noses and small noses and whatever, but everyone has teeth like, do you know” (Participant 2), and the dentition was a major factor for normal appearance “you notice straight away if they’re missing a tooth” (Participant 3), and on normal subject presentation “have a set of teeth that approximate normality”, “that is what’s seen as normal”.

The aesthetic impact of teeth was not fully defined or discussed separately from the general facial appearance: “they’re with a smile” (Participant 2), which raised the aesthetic relevance of teeth in self-esteem “I noticed recently, I’m like trying to get more confidence like smiling with my teeth”. The smile was reported to be affected when teeth or prostheses were being problematic, as a result of an ill-fitting denture or fractured restorations “don’t want to smile, barely open your mouth”. In addition, teeth are forward facing as discussed earlier because “It’s the first thing I see on everybody” (Participant 3).
Dental aesthetics were reported as being more important than the ability to eat “if you told me I had go home and eat with no teeth every day I’d do it. Something like that, but no, it’s pure appearance” (Participant 1). The importance of the dentition was projected to other people who did not take care of their teeth by saying “you have no idea what you have got” (Participant 2). This highlighted how central the dentition was to their outlook, as these group constantly referred to other regular appearances as a benchmark for their own dental situation, “I think dental is something that’s like very like forward-facing, it’s very, you know, and it’s, it’s something that like, if you get a, you know, you’re chewing a piece of food, you know”

4: Okay. So, do you think teeth is important for self-esteem? Do you think?
3: Yeah.
2: if your teeth, if you hit like, like it literally stops you, like you were saying quality of life earlier on,
3: Of course.
4: Really?
2: Hundred per cent.
4: In what way?
2: It’s the first thing I see on everybody.
3: If you were to,
4: Really?
2: Yeah.
3: if you were to meet somebody and just
2: Yeah.
3: like look at somebody
4: Yeah.
3: and they’re with a smile,
2: Yeah.
3: you notice straight away if they’re missing a tooth or if they’re...
4: Okay.
1: People are bald, people are, big noses and small noses and whatever, but everyone has teeth like, do you know,
4: Okay.
1: barring,
4: Yeah.
1: eh, if you were,
4: So, you think it’...
4: Yeah, so you think it’s more than just eating.
1: [Oh, it so, of course. It’s appearance.]
2: [Oh yeah.]
4: Really?
2: Yeah.
1: I, I would eat no bother like, it’s tough but I,
2: Yeah.
1: if you told me I had go home and eat with no teeth every day I’d do it. Something like that, but no, it’s pure appearance

3.2.13 The Impact on Breathing

The impact of breathing was also discussed by two participants in relation to having asthma whilst younger and how this affected their ability to participate in sports: “you are not getting enough oxygen”, while this participant still used an inhaler when he had a cold or had a chest infection. The other participants discussed ways to minimise the impact of asthma, such as using inhalers. Finally, having a dry mouth was mentioned only by Participant 3 who explained that “your mouth just feeling like, you know, it’s been open all night” which linked with the dry skin and eye issues that were discussed previously.

In summary, when talking about the impact of various aspects of Ectodermal Dysplasia on their quality of life, what all participants agreed was that “definitely teeth [are] number one” (Participant 1) because teeth affect a number of other aspects of daily life, including “eating (...) confidence (...) talking to people (...) ability to smile” (Participant 3). Dental
issues was followed by the impact of skin on participants in terms of dry skin, followed by temperature control and hair loss factors, and lastly, breathing difficulty which wasn’t considered by all participants as an issue generally: “it is never been an issue”.

Appreciation of the dental treatment and referral arrangements for treatment was discussed by all participants. They detailed their understanding of the treatment steps and types of prostheses that they have, and how these prostheses were different from other each other. This gave the impression of how central their dentition was to each of them and the importance in early and adolescent years. In addition, with respect to long treatment plans that they had had since they were young, they reported acceptance and happiness with their end treatment in the Children Crumlin Hospital and Dublin Dental University Hospital.

3.2.14 Managing the Impact of Ectodermal Dysplasia

An emergent finding in this study was the various ways of managing the impact of Ectodermal Dysplasia “just cope yourself” (Participant 2). The first of the two most commonly discussed ways was, not surprisingly, using certain adaptation aids. When discussing the effects of ectodermal dysplasia on the skin, for example, the participants discussed custom-designed vests used to regulate the body temperature:

2: And they had things like eh, vests, school vests that you could get
4: Yeah.
2: that like, obviously you, you’d put them in the freezer or something and they had ice packs.
1: I had to buy that, em, for sport.
2: Oh yeah, I remember.
1: I used it I’d say twice since then.
2: Oh really? Yeah, yeah.
4: So that’s a vest with ice in it. Interesting.
1: Well you
2: I don’t know.
1: dip it in water like,
4: Yeah.
1: and then there’s crystals in it or something and they expand, and then you freeze it.

Other aids that were discussed were: (a) lubricants for dry eyes “I’m having to like lubricate my eyes like maybe five or six times a day” (Participant 3), (b) sun cream for the skin condition, (c) hair transplants: “I might now look at hair transplants” (Participant 3), (d) inhalers and a nebuliser which is “a mask” which has “some steroid pumping through it” (Participant 1). As previously mentioned, the clear opinion was that the impact of teeth was “definitely (...) number one” (Participant 1). However, the participants mostly discussed the details of wearing dentures as part of earlier management treatment of the condition: "I have had dentures since I gave can remember” (Participant 1), and how these dentures are replaced over years: “I remember the year I was doing my Leaving Certificate, I started having issues with, it’s kind of that stage during puberty when you kind of need to advance to the adult dentures” (Participant 3). Also, denture treatment at different stages of life reflected the importance that was emphasised by Participant 2 “Jesus, I wouldn’t leave my bedroom without it now”. Thus, having dentures fabricated for different ages, the participants reflected upon their experiences and having a sense of how their dentures became part of their life adaptation tools, which required extensive work and required ongoing adjustment or replacement. At this stage, all of the participants had been definitely restored for more than five years and were reflecting back on their dental journey and how their growth and development required constant dental treatment including making of new dentures to accommodate changes in their mouths. However, it appeared that whilst
undergoing treatment, they did not have that perspective. This may have resulted in earlier reported comments such as the problems with long treatment journeys.

An interesting finding was that, according to the participants, the impact of ectodermal dysplasia on their lives could be minimised by managing their identity. The participants reported that changing the way they perceived their condition, and themselves, essentially helped them lead a better life. As Participant 3 summed up, "I felt a lot more confident once I was like, screw it". Participant 1 also explained how he “always said I’m the same as anyone else”, which clearly increased his self-esteem and self-confidence. Sometimes it was the way people around them behaved that helped them manage these self-perceptions, such as families and colleagues. In the following extract, for example, Participant 3 described how other people’s accepting and open-minded attitudes towards his condition influenced the way he perceived it:

3: it was only really when we went to college that I was kind of like okay, this is what I have, and people
4: Yeah.
3: were like,
4: Okay.
3: I mean everybody has their own.
4: Yeah.
2: Everybody’s got something, so.
4: Yeah.
3: Yeah, exactly.
2: At that point it’s...
3: And then your confidence kind of shoots up, because like okay, you don’t have this thing that you have to keep hidden.

As Participant 3 further explained, “everybody has their own, I guess, messed-up thing about them, so don’t feel isolated”. All of them agreed that “getting out there”,
participating and getting involved in various activities was crucial for managing their identity and, thus, minimising the impact of the condition on their lives.

Another theme discussed in relation to managing the impact of Ectodermal Dysplasia on life was becoming an **expert in self** and knowing how to behave. This theme was essentially about making use of available resources to address some symptoms of the condition through “**trial and error**” (Participant 1), as well as about understanding one’s condition and being reasonable about what one could and could not do. The participants discussed, for example, “**drinking protein shakes because I couldn’t really chew**” (Participant 3), “**using moisturising creams for the dry skin**”, “**having a bottle of water next to a bed because of dry mouth**”, or “**putting one’s feet or hands into a sink with cold water to cool down**”.

**Expert in self** by trial and error was a method of adaptation as a consequence of a lack of peer support for information exchange, and absence of organisations that provide support and guidance.

In addition, lack of general medical and dental support, “**no medical home**” in their home area, absence of a central referral point in association with limited numbers of general medical and dental practitioners experienced in managing the condition led to frustration among the participants. In terms of knowing how to behave, what to do and what not to do, again, participants discussed the need to remember their own limits and select places that have access to controlled temperature, such as work places and even holiday hotels.

Careful planning ahead was a constant feature of life with ED, including wearing a hat or carrying water or creams to avoid overheating if they visited foreign countries or planned to be outside for a long time. They also advised caution when planning to seek employment overseas and in terms of a choice of holiday destination in general.

Finally, **information gathering, socialising** and **researching the condition** all helped to minimise the impact of the condition on the participants’ lives. The participants discussed
special support networks and events where they could talk to others who had similar experiences. In addition, one participant stated that his sister tried to make connections with other families with children with the same condition and they had hosted some meetings where they shared the experience and provided peer support. He mentioned the role of these networks and societies in England and the USA where they have dental expertise at the event who will arrange care for the affected subjects, which reflected the priority of dental issues. They reported that further information was gathered through the use of social media platforms from the experiences of other ED patients in other countries such as America and England. The participants suggested and emphasised the positive benefits of having a society to join peers with the same condition which could provide support and assistance to others. Also, parents would be in a position to share their experience with those who have younger children with the same condition.

Participants stated that in their opinion, the ideal team should be multidisciplinary and include specialist medical practitioners, specialist dentists (depending on the age of the ED patient), dermatologists, and ophthalmologists. This represented the main impacts of the condition. However, the ophthalmologist was mentioned by only one participant, but it raised awareness among the other participants who then discussed the importance of seeking ophthalmology advice. In addition, it was felt that general health conditions including asthma could be managed by a general practitioner.

When asked for their advice for a child who has Ectodermal Dysplasia, they gave only positive answers “we are all the same, do it”, “Do whatever you want to do”, “it is a hundred percent perspective”, “there is nothing holding me back”. However, the participant who had difficulty in playing sport suggested being careful while playing sport and retaining control of temperature management and skin moisturising. Participant 2 encouraged socialising and being involved: “you have to get involved with something”,
“don’t feel isolated”, “everybody had their own I guess messed-up thing about them”, “you can talk to your friend about it at mature age not at teenager age” and “have a dermatology consultation at younger age”.

4 Discussion

4.1 Discussion of the Quantitative Study Findings:

A number of studies have been completed on the prevalence and aetiology of isolated hypodontia and its effects on Oral Health-Related Quality of Life (OHRQoL). However, a paucity of studies have been undertaken on the impact of Ectodermal Dysplasia and its associated features on OHRQoL and resultant psychological impact on children and adolescents (Bergendal et al., 2006, Nordgarden et al., 2002). Previously, parents or caregivers were used as a source of information to indirectly assess the quality of life of their children. However, the development of validated questionnaires capable of ascertaining health-related quality of life information has changed the landscape and facilitated information gathering directly from children, which can later be utilised in combination with the parent/caregiver views.

This cross-sectional case-control study was conducted to investigate the impact of Ectodermal Dysplasia on Oral Health-Related Quality of Life in a sample of Irish children and adolescents. The total of 22 pairs were deemed eligible to take part in this study. All accepted participants were then matched by age and gender with control subjects. This research was novel and to date, no study had explored the same condition with a sufficiently large sample size to complete a power calculation. In addition, as Ectodermal Dysplasia is a relatively rare condition, this study aimed to collect as many participants as possible, in combination with controls matched in terms of age and gender. Sufficient
consideration also applied to the malocclusion of the control participants, and only children and adolescents without a significant malocclusion were recruited. This is as a result of previously reported impacts of malocclusion on OHRQoL (Barbosa and Gavião, 2008).

The majority of the participants recruited were male (72.7%) which is consistent with the participants in Kohli’s (2011) and Bergendal’s (2006) reports where males constituted (62.9%) and (65.2%) respectively (Bergendal et al., 2006, Kohli et al., 2011). The mean age of participants was (12.5) years, with a range from 8 to 18 years. This age group was comparable to the cited literature; a mean age of 14.5 years with a range from 3 to 55 years was reported by Bergendal in 2006, while Kohli (2011) reported the impact of Ectodermal Dysplasia in children and adolescents aged 11-19 years. The age of participants in those studies provided a valid comparison in terms of their study findings to the current study.

As with previous studies, the current study subjects had to present with one or more classical features of ED, such as missing or malformed teeth, dry skin, and sparse hair (Bergendal et al., 2006, Nordgarden et al., 2002). The ED subjects presented with significant hypodontia of their permanent teeth with the mean number of missing teeth being 18.5 with a range from 2 to 28. This was greater than reported in other studies such as that by Bergendal et al. (2006) who reported a median of 7 teeth absent (range 6-20: 46 participants). This study found that females had fewer missing teeth than males with means of 16.57 (SD = ± 10.67) and 19.19 (SD = ± 8.12) respectively. These figures were in agreement with previous findings by Lexner and co-workers who reported higher mean numbers of missing teeth (Mean = 22, Range 14-28) in males in contrast to females (Mean difference = 4, Range 0-22) (Nordgarden et al., 2002, Lexner et al., 2007c).

In addition to the prevalence of missing teeth, the erupted teeth malformation prevalence in this study was 77.2%. These malformations included conical shaped teeth, taurodontism and ankylosis of primary teeth. These malformations had been reported in the published...
literature where Lai and co-workers reported that 8.9% of patients with hypodontia had conical shaped incisors, with a prevalence of ankylosis of the primary molar teeth stated to be 65.7% in those with hypodontia in contrast to 1.5% in the control children. Meanwhile, taurodontism in mandibular first permanent molars had been observed with a higher prevalence in participants with hypodontia in contrast to those without hypodontia, 34.3% and 7.1% respectively (Lai and Seow, 1989).

Concerns had been expressed previously in relation to quality of life data acquisition from young people due to limited cognitive development and problems with communication. Proposals were made that parents or caregivers could instead be utilised as effective proxies (Theunissen et al., 1998). However, the Parent Perception Questionnaire (PPQ) provides limited information about the individual child’s activities, particularly those occurring outside the home. On the other hand, even incomplete parental reports about their children are still a source of valid information (Barbosa and Gavião, 2008). Thus, the purpose of the PPQ is as a proxy rather than as a secondary or complementary information source to information provided through the Children Perception Questionnaire (CPQ) (Jokovic et al., 2002).

In the current study, the impact of Ectodermal Dysplasia on children’s’ OHRQoL and their parents or caregivers was obtained using the Child Perception Questionnaire (CPQ). McGrath and co-workers had previously reported that no gold standard existed for OHRQoL measurement in children, but the long version of CPQ used in this study has now been validated in a variety of cohorts in different countries including Canada, Hong Kong, and the United Kingdom (Wong et al., 2006, Jokovic et al., 2002, O’Brien et al., 2006). It has been validated for assessment of the impact of malocclusion and hypodontia in children, which facilitated comparison of the current study findings with other cohorts (Wong et al., 2006, Laing et al., 2010). The CPQ has limitations in that it is not a condition-specific
questionnaire and it lacks open-ended questions to evaluate further experiences not covered in the closed questions. Lastly, it does not reflect the main cause of OHRQoL impact, which might be due to other reasons, such as social and cultural background, the status of general health, household income and life stress (Mandall et al., 2000).

4.1.1 Impact of Ectodermal Dysplasia (ED) on Children and Adolescents

The current study adds additional information on social, functional, and psychological impacts of ED on Irish children and adolescents with the condition compared to matched subjects in two age groups, 8-10 and 11-14 years old. The null hypothesis of this study was rejected. In the 8-10 age group the mean of the scores were higher in ED patients than control subjects. However, the oral symptoms domain mean scores were higher in the control subjects. The reported higher oral symptom domain scores in the control group might be related to natural physiological processes, such as primary teeth exfoliation, the eruption of permanent teeth, or spacing between the teeth at the jaw development age (Barbosa et al., 2009). However, in the current study, the differences between these scores were not statistically significant, which may be attributed to the dependence of children on their parents at this age (8-10) for decision-making and family support to overcome the condition symptoms, which consequently masked the real impact of the condition on the children. In addition, the children at this stage have less mature mental development which might be a difficulty for some of them when interpreting or understanding the questions in the questionnaire. For that reason, Jocovic included some drawings in the questionnaire that enable the children to understand the questions (Jokovic et al., 2004b). In addition, limited development at an emotional level resulted in less expression of the emotional factors or emotional perception in relation to oral and dental health. Furthermore, at this
age (8-10) the presence of primary teeth might mask the influence of permanent teeth hypodontia and so reduce the impact of functional, emotional, and psychological influence on their perception. Lastly, from the methodology point of view, the current study has a limited number of participants in the age group (N = 6) which might have influenced the study findings.

In contrast, among the older participants (11-14 years) there were statistically significant differences between ED subjects and their controls (P<0.05) in all areas with the exception of the social well-being domain (Mean = 21.5, SD = ± 7.3). Possible explanations for the reported higher scores in the case subjects in the oral symptoms and functional limitation domains include dental features associated with Ectodermal Dysplasia, including missing teeth, dry mouth, malocclusion, difficulty in chewing food and speech. In addition, at this age (11-14 years), participants tended to notice spaces between their teeth as a consequence of missing teeth (Barbosa et al., 2009). The oral symptoms domain findings were consistent with the results of Wiemann and co-workers in their OHRQoL investigation among adult subjects with rare diseases including Ectodermal Dysplasia (ED) (N = 46). The most commonly reported oral symptoms by Wiemann were dental anomalies, malformation, missing teeth, and malocclusion. The participants reported OHIP oral symptoms domain scores with a mean of 15.9 (SD = ± 13.1), which reflected the impact of oral symptoms on all participants including ED subjects, in addition to the high overall OHIP score range (15.9 - 19.7). This was in marked contrast to the average German population mean OHIP score of 4.09 (John et al., 2004, Wiemann et al., 2018).

In the same age group (11-14 years), the ED subjects reported higher scores in emotional well-being domain than those of the control group. This might be attributed to the influence of malformed and missing teeth on appearance and emotional impact related to the condition in terms of self-esteem and social interaction. In addition, teenage years were
associated with further interaction of the participants with other children in school and the surrounding community which might be accompanied by embarrassment in relation to their dental appearance or result in unfavourable comments being directed towards them. Furthermore, limited peer support and guidance resulted in limited adaptation to the condition’s general symptoms such as overheating. In addition, the 11-14 years of age group included more females in contrast to the younger age group, and females are reported to be more sensitive and anxious about their health and appearance which might explain the increased emotional well-being domain score (Kohli et al., 2011).

The differences observed between the two age groups were interesting and may have been due to a number of factors. Firstly, the increased self-awareness of the 11-14 years age group was consistent with the cross-sectional study of Kohli where older participants (N = 21; 15-19 years) reported higher OHRQoL scores (Mean CPQ = 35.9, SD = ± 21) in contrast to the younger group (N = 14; 11-14 years; Mean CPQ = 25.1, SD = ± 13.8). This awareness included the predicted difficulty in chewing certain foodstuffs, as more missing permanent teeth were noticed at this age, and limited physical activity because of overheating as reported by the current study ED subjects (N = 16, 72.7%), in addition to the probability of bullying by other children in schools as a consequence of their dental and general appearance. Secondly, the physiological cognitive development of the children from one stage to another might have been an influencing factor. The children’s’ knowledge and their awareness of their general and dental health increased, which was the interpretation reported by Kohil when an older participants reported higher scores, with a mean CPQ of 35.9 (SD = ± 21), than a younger age group with a mean CPQ score of 25.1 (SD = ± 13.8) respectively (Kohli et al., 2011). A third explanation may be that at the older age, children were more cognisant of their missing teeth as a result of being more engaged with other
children and surrounding communities which influenced their responses, particularly in the areas of functional limitation, social, and emotional well-being (Barbosa et al., 2009).

Furthermore, at the age of 11-14 years, 72% of the current study sample were already on board in terms of their dental restorative treatment, or they had planned treatment with other disciplines, such as orthodontics. These figures were not provided in most previous studies so comparison was not possible. The degree of restorative treatment being undertaken in this study was variable, with treatment received ranging from aesthetic tooth colour restoration of malformed teeth to a removable partial or complete acrylic denture. Fifty per cent of the 11-14 year age group had an average of four sets of dentures fabricated prior to the date of the study, while the remainder did not wish to wear dentures or they had missing teeth in areas that did not require dentures.

Provision of dentures was associated with multiple dental treatment appointments for fabrication of prostheses. These prostheses required maintenance and re-fabrication every 1-2 years to accommodate growth and development. In addition, these appointments involved loss of school time and may also have limited some social activities. One other possible reason may have been the greater number of participants in the older group which may have provided sufficient data to detect differences between groups. A comparable study by Kohil on the impact of ED on the OHRQoL of children aged 11-19 years shows that 61% (20 out of 33) of the participants used removable dental prostheses at the time of the study with 19 of those prostheses being replacement prostheses (Kohli et al., 2011).

Both age group participants in the current study had the benefit of dental treatment at a young age to manage their Ectodermal Dysplasia dental-related features at both the Dublin Dental University Hospital and Children's Health Ireland at Crumlin from where the participants had been recruited. All of these confounding factors were considered at the data collection stage by defining all dental features and their management when applicable.
and considered also in the participants’ perception analysis and data interpretation. This consideration, in addition to the recruitment of the control group from the Undergraduate Dental Science Paediatric Dentistry clinics, provided additional validity for the study findings and a control for dental treatment confounding factors. Consequently, the reported OHRQoL perception for both study and control groups benefited from the dental experience of those participants in all CPQ domains, including the oral symptoms, functional limitation, social well-being, and emotional well-being domains.

The current study findings are consistent with those of Kohli where ED subjects reported a high OHRQoL score with a mean CPQ score of 31.6 (SD = ± 19) among 35 participants aged 11-19 years; however, no control subjects were included in that study (Kohli et al., 2011). The inclusion of a control group in the current study was considered at the design stage to eliminate external biases and influences that might alter the experiment results. In addition, the control group increased the focus on the study variables (Ectodermal Dysplasia signs and symptoms) and provided validity for the study results (Pithon, 2013).

The current study results were in agreement with those reported in a German adult population with rare diseases including Ectodermal Dysplasia. The average reported OHIP scores of 12.2 (SD = ± 12.3) and 11.7 (SD = ± 11.07) for males and females respectively, was significantly higher than that of the average German population of 4.09, which reflected the impact of rare diseases including Ectodermal Dysplasia on OHRQoL (John and Micheelis, 2003, Hanisch et al., 2019a).

The reported effects of Ectodermal Dysplasia on the older age group were compared to a previous study in adults (Mean age 29.6 years, SD = ±14.9) who were assessed using the Short Form Health Survey 36 (SF-36) questionnaire. That study by Saltines reported lowest mental health-related quality of life (Mean 74.4, SD = ±20.1) for ED patients in contrast to the mental health-related quality of life of participants with Treacher Collins Syndrome.
(Mean 81.1, SD = ±14.4) and Cherubism groups (Mean 82.2, SD = ±14.6), while at anexity the ED group reported higher than other two groups (Mean 6.9, SD = ±3.7), who reported (Mean 3.1, SD = ±2.7) and (mean 2.4, SD = ±2.1) for Treacher Collins Syndrome and Cherubism groups respectively (Geirdal et al., 2015b, Saltnes et al., 2017a). These reported values indicated the increased level of stress and anxiety associated with the dental and general features of Ectodermal Dysplasia (ED). In addition, the reported levels of anxiety among the adult ED population might have been an indication of the long-term impact of ED from childhood to adulthood.

Hypodontia is one of the main four features of Ectodermal Dysplasia, yet few studies have used the CPQ to evaluate the impact of hypodontia and other features of ED. For that reason, the current study findings were compared with studies reporting the impact of isolated hypodontia on OHRQoL. As a result, the findings from the current studies would have been anticipated to be greater for the ED population given the multiple features of the condition. The mean CPQ overall score of ED in this study was 75.3 (SD = ± 27.3) for the 11-14 year age group, which was higher than CPQ scores reported by both Wong and co-workers and Locker (Mean = 29, SD = ±16.4 and Mean = 22.3, SD = ±14 respectively). This higher score among the ED subjects was attributed to the influence of the other features of the condition, such as skin and eye dryness and overheating for general and oral health related quality of life.

4.1.2 Impact of Ectodermal Dysplasia on Parents and Caregivers

Accurate parental perceptions of their child’s quality of life are vital given their role in treatment choice selection for their children. The Parent Perception Questionnaire (PPQ), developed by Jocovic, is valid and has good internal consistency and reliability, and
excellent test-retest reliability (Jokovic et al., 2003b). The parental reports provide valid information for the assessment of oral health related quality of life in children or adolescents, even though this information is limited as a result of the limited awareness of the individual child’s activities, particularly those occurring outside the home (Barbosa and Gavião, 2008).

Assessment of child and parental agreement in a total of 42 pairs of children and parents by Jokovic found that the levels of agreement between children and mothers was good with some evidence of bias in mothers’ perceptions in contrast to their children. There was a substantial agreement between mothers’ and childrens’ total scores (ICC = 0.70); however, there was only moderate agreement for the emotional and social well-being subscales. The differences in the reports reflected the differences in their perspectives which may have been related to limited parental insight into their child’s feelings and activities away from home. Jokovic concluded that “although mothers may be used as proxies for their children in some circumstances and some purposes, the views of both should be obtained to fully represent child oral health-related quality of life” (Jokovic et al., 2004a, Jokovic et al., 2003a). Thus, the PPQ should be used as a proxy rather than as a secondary or complementary information source provided through the CPQ (Jokovic et al., 2004a). The present study reported parental perceptions about their child’s OHRQoL in two age groups for both Ectodermal Dysplasia patients and control subjects. In both age groups, the ED subjects’ overall PPQ and domain scores were higher than those of controls, with a statistically significant differences evident ($p = .05$). The 8-10-year age group PPQ mean score was 86.5 (SD = ±16.2) for ED subjects compared to 59.7 (SD = ±12.9) for controls ($p = .042$), with statically significant variances in functional limitation and family impact scale domains ($p = .042$) and ($p = .043$) respectively.
The mean for both overall and subdomain PPQ scores in the 11-14-year age group ED subjects was higher than those for controls. The overall PPQ of ED subjects was almost double that of control subjects with statistically significant differences between groups ($p = .006$) with scores of 98.04 (SD = ±42.2) and 58.6 (SD = ±10.8) respectively. However, the difference was not statistically significant for the oral symptoms domain ($p = .1$), where the ED subjects reported a mean score of 12.2 (SD = ±5.7), while the control reported a mean of 9.4 (SD = ±2.4).

The scores of parents of ED subjects reflected the influence of the condition on their families. Limited published literature was available on parental perceptions of ED for both age groups so limited comparison with previous studies was possible. The functional limitation domain highlighted parental awareness of difficulties with either food chewing or speaking as a result of limited dentition. Also, reported scores in this domain might be attributed to the additional efforts required for food preparation for their children who have missing teeth. In addition, this domain score interpretation may also have reflected parents’ awareness of the time and effort required for the replacement of missing teeth.

The influence of the condition on parental perception was greater than that of the control group parents who reported low perceptions in contrast to the ED subjects’ parents. The low perceptions reported by the control group parents reflected the low impact of routine dental treatment on their children in contrast to the ED subjects.

The Family Impact Scale scores delineated how the child’s condition impacted upon the whole family. In the 8-10-year age group, the parents of the ED subjects reported poorer scores than those of the control group (25.4, SD = ±5.3 versus 16.8, SD = ±5.1). Similar trends were observed in the older age group where the mean scores for cases’ and controls’ parents were 27.3 (SD = ±10.8) and 16.6 (SD = ±3.2) respectively. The higher scores in the case subjects may be as a result of the particular arrangements required indoors for
temperature control, food type choice for the whole family, travel destinations and holiday choices which may have financial and social impacts, where other family members might have to accept all of these adaptations in favour of the affected family member. The cost of treatment, the time a parent may be absent from home fulfilling dental and medical appointments and provision of increased time to the family member with ED may have wider impacts on the whole family unit.

4.1.3 Comparison of the Child and Parent Perceptions

The current study facilitated a comparison of the perceptions of parents and their children. This varied markedly with the age of the child. There were observed differences for the 8-10-year-old group between parents and children with ED in terms of the oral symptoms and functional limitation domains ($p = 0.04$ and $p = 0.04$ respectively). This could be possibly explained by three issues. Firstly, children at this age have limited cognitive development which may have limited their awareness of the condition’s impact on their general and oral health. Secondly, limited parental awareness of their child’s perception of feelings at certain times, such as in school as reported previously, may have resulted in the reported differences in these domains. Lastly, children tend to depend on their parents at this age for decision-making and treatment-related perception, which can be supported by the reported differences in the functional limitation and emotional well-being domains ($p = 0.04$) in the control group.

In contrast, the older group of children reported a higher mean score than their parents, but there was no statistical difference reported between the perceptions in all domains for both ED subjects and controls. The higher score, albeit small, could be related to the children maturing where they might have reported fewer problems to their parents such as school bullying or embarrassment as a result of their dental and facial appearance.
Parents may also be unaware of the emotions of their children and the social issues arising from the condition. Secondly, the difference between parents and children might be related to the nature of parental proudness in their children, their social relationships, and activities. Furthermore, reports on parental perception may have been influenced by the parents’ sympathetic feelings towards their child’s condition or disability, which results in an overestimation of the condition’s impacts, particularly in the social and emotional well-being domains. This parental perception interpretation had been reported previously in an interpretation of the comparison of parents’ perceptions of their autistic children in contrast to the parents of non-autistic children (Pani et al., 2013).

In addition, the older children and their parents may have had a greater awareness of missing teeth as the permanent teeth would have been expected to erupt at this age. As the children get older, they would also have been exposed to more dental treatment and the plans for future treatment would have been discussed with them, resulting in greater dental awareness and knowledge. This may have included discussions on orthodontic therapy and future dental implant provision. This may have had an unfortunate and unintended negative impact on both children and their parents. These missing teeth may also contribute to difficulty chewing food and speech, particularly in those who do not have retained primary teeth, which can mask some of the functional and aesthetic limitations.

The higher impact on OHRQoL reported by children than their parents was consistent with the study by Jokovic where children aged 11-14 years reported more OHRQoL impact scores than their parents. That study investigated orofacial problems and orthodontic treatment (Jokovic et al., 2002). In another previous study, parents reported worse OHRQoL than their children with ED in groups aged 11-14 and 15-19 years. The differences between parental and children’s perceptions were only statistically significant in the functional limitation domain in the older age group (Kohli et al., 2011). The author
explained the higher parental scores in the older group through family awareness of the child’s functional problems, such as speech and chewing difficulties over years which resulted in restricted dietary choices. Meanwhile, over the same period of years, adjustment of the emotional and social problems might be achieved, which justified the lack of differences between parents’ and children’s perceptions, even though parents’ perceptions were worse (Kohli et al., 2011).

4.1.4 Discussion of the Level of Agreement

The level of agreement between children and parents OHRQoL perception can be assessed at questionnaire individual domains level or by comparing the total scores of participant’s perception at group levels interim of gender or age, or at the individual level, by comparison, each child to his parent or caregiver. Intraclass Correlation Coefficient (ICC) is the test that had been adopted by Jokovic et. al., (2003) in the examination of the level of agreement between children and their mothers, also, a Bland-Altman plot is a useful tool defining the extent of disagreement between children and their parents. In the present study, levels of agreement between parental and child perception were investigated using the Spearman test and found not to be statistically significant with a weak level of agreement reported, but demonstrated a negative low level in the emotional well-being domain (-0.07, p = 0.7). This indicated parental overestimation of the emotional impact of Ectodermal Dysplasia on their children in both age groups. These reported values, in addition to the differences in the emotional well-being and social well-being domains reported, are interesting. Questions addressed to the parents in the emotional well-being domain included “How often has your child avoided smiling or laughing when around other children?” “...because of his or her teeth, lips, mouth, or jaws; how often has your child
been irritable or frustrated?“ “...shy or embarrassed?”. These questions were related to activities that took place outside the home environment and as such, the insight of the parents may have been limited.

On the other hand, questions related to functional limitations, such as difficulty with phonetics or with chewing certain food, could be directly observed at home. Some studies reported a high level of agreement in the functional limitation domain as parents tended to recognise their child’s difficulty in chewing certain types of food as an example. However, the current study results showed poor agreement between parental and child perceptions in the functional limitation domain (0.2, p = 0.3). This could be attributed to increased parental awareness of the child’s limited function, such as speech and chewing difficulties, which resulted in restricted diet choices to accompany missing teeth function and parental overestimation of the treatment required to restore function (Kohli et al., 2011). In addition, the children in both age groups might still have had limited awareness of the treatment and effort required to restore the missing teeth function.

Correlation values in the current study were not consistent with those reported by Jokovic and co-workers in the assessment of OHRQoL using CPQ in a paired sample (n = 42) of parents and children aged 11-14 years who had an oro-facial condition (N = 15), were undergoing dental paediatric treatment (5), or orthodontic treatment (N = 22). They reported substantial level of agreement between mothers and children with total intraclass correlation coefficient scores of 0.7, high agreement in relation to oral symptoms (0.8), substantial agreement for functional limitations (0.7), and moderate agreement for emotional and social well-being domains (0.5) (Jokovic et al., 2003a). In addition, Kohli assessed OHQoL in 35 children and adolescents aged 11-19 years old with Ectodermal Dysplasia and among their parents/caregivers using the same questionnaires as the current
study (CPQ). Interestingly, parents reported higher scores than their children, but there was a positive correlation between parents and their children in all domains except in the social well-being domain (Kohli et al., 2011).

The lack of agreement and non-significant correlations in the current study may have been for a number of different reasons. Family and parental adaptation to the social and emotional impacts over years, as reported by Kohli, might result in the same perceptions among parents, but the children might have reported different perceptions as they may not have adapted yet to the social and emotional impact of the condition, resulting in the reported differences (Kohli et al., 2011). In addition, the reported long-term experience of functional limitations by the parents in previous studies and the current study may be as a result of parents thinking that the child had adapted well as a result of available tools and treatments, such as special type of socks and vests for overheating control, or restoration of the missing teeth function by having dental prostheses which overcome the difficulty in food chewing and speech. Therefore, the parents may have formed the opinion that, through an intervention, the impact of functional limitations may have been mitigated. However, from the child’s point of view, this might not have been sufficient to adapt to the functional limitation associated with the condition. The children may not have had the maturity to see how interventions may have improved their function rather than focusing on existing deficits and that may have influenced their responses.

A previous study examining functional limitation in 11-14-year-old children and their parents (n = 46) reported high levels of agreement in the perception of the domain of functional limitation following orthodontic treatment (Khraishi, 2019). These findings were not surprising because the referred study participants had neither congenitally missing teeth nor ED features and the results were unsurprisingly at variance with the current study. The lack of agreement and non-significant correlation in the current study may have
been related to a small sample size in the younger group involved in the study, which might have influenced both reported perceptions. However, even with the larger number of participants (N = 16) in the older age group, disagreement was reported between parents’ and children’s perceptions in the same way as for the smaller sample size in the younger group (N = 6).

4.1.5 Strengths, Limitations, and Future Research

This study had significant strengths as it was the first study on the QoL of Irish ED children and provided more information on dental and other health issues facing this cohort. It also highlighted the significant need for dental intervention and may assist policymakers in recognising the treatment needs of the ED population. In addition, this novel study added valuable information in terms of children’s and adolescents’ experiences of living with ED, and how this condition influenced their dental function and routine daily function as well as social and emotional impacts. New insights have been achieved from this study in terms of parental perceptions regarding the impact of ED on their children. This information can be used as a resource for future education and to increase awareness of the condition and its impacts. It might also inform parents how their children might be feeling and recognise the different perspectives that may exist when living with ED. This increased parental awareness, in combination with dental and medical professional interventions, should help identify and assist in overcoming the challenges and compromised OHRQoL of this cohort. The value of the research was also increased by including a control group. In comparison to previous studies, this provided the current study with a point of comparison for children and their parents thereby increasing the validity of the presented results (Kohli et al., 2011). In addition, this study provided significant additional information regarding increased
awareness of subjects with ED of their condition at two stages of development. It also highlighted the caution needed in this population when using parents or caregivers as proxies for their children. This study confirmed the limited awareness of parents of their child’s perception at different stages identified in previous studies, and the limited exposure of parents to all of their child’s feelings and experiences. The parental reports should be used to support the child’s reported perception which should be considered the primary source for information on the condition or variable impact perception.

There are weaknesses inherent within this study such as a small sample size. This was unavoidable due to the low incidence of ED nationally. Every effort was made to recruit as many subjects as possible including contacting the ED Society in Ireland and health professionals who were providing medical and dental treatment for this population, whilst adhering to GDPR guidelines. This study did not facilitate a detailed comparison between genders because of the limited number of female participants. However, the condition has a higher prevalence in males and more ED features are generally present in males. Consequently, this study’s participants' perceptions are valid and can be used as an indicator of the impact of ED on children and their parents.

The other limitation of this study is the fact that the majority of the participants in both age groups were able to avail of comprehensive dental treatment which might have influenced the participants’ perception positively when the treatment was at a stable stage and negatively when the treatment was complicated by changes associated with growth. Furthermore, the questionnaire used in the study, and its parent version, is not condition-specific and cannot be used to examine individual Ectodermal Dysplasia physical features and the impact of psychological features, which limit the impact of the condition reported by this cohort and limited resultant interpretations and perceptions.
In relation to the control group, there was the possibility of bias because of the site of collection. The convenience sampling method used patients attending the Paediatric Dental Clinics at the Dublin Dental University Hospital and they may have presented with greater dental problems and needs than the general population. It was felt, however, that selecting a control group from the hospital should have some balance for this potential bias, as it provided a point of comparison from a dental treatment impact point of view, as the control group participants were undergoing routine dental treatment at the time of the study.

The other weakness of the study is no consideration was taken place to match the case and control participants from the socio-economic point of view, which was consequently might undermine the results presented in this study. The questionnaire used in this study has social well-being domain, answer to this domain questions depend on the socio-economic background of the children, parents, and the whole family. The presented study reported poor social well-being domain of the cases in contrast to the controls, but adjustment on sample matching might reflect otherwise, which should be considered in future researches.

As the study was a case-control type, it was not possible to follow the participants and capture the age-related differences in oral health related quality of life over time. Thus, the prevalence and perceptions reported in this study can only be applied to those who participated in it.

This study detailed additional information that should assist clinicians in understanding the nature of this condition and its functional and emotional impacts on the affected population and their families. The clinician should be more aware of the issues pertaining to this group, giving priority to, and considering the importance of restoring missing teeth in terms of speech production, chewing function, appearance, and self-esteem. In addition, the reported insights will aid in providing a suitable environment for those children in terms
of overheating and mouth dryness management as reported in the focus group discussion for this study.

Further research is recommended to provide further comparisons between the children and controls at an individual level and between parents of both groups. This will provide further information about the frequency level of the reported impacts in different domains. Furthermore, investigation of the impacts of the condition in contrast to other rare congenital conditions would provide further details on relative impacts. And finally, a larger sample size might help in elucidating the nature of the reported disagreement between participants and their parents at individual levels. The information from this study in combination with the focus group analysis report can be used for condition-specific questionnaire development.

In addition, it would be interesting to follow a group of children with ED longitudinally to see how their approach and management of the condition might change over time. This could be further re-evaluated alongside their parents’ perceptions. The current study looked at two age groups at a distinct moment in time, limiting comparison between them. The effects of maturation, development, and understanding would be best elucidated through a longitudinal research approach to add further to the work of this study.

4.2 Discussion of the Qualitative Study Findings

This study explored the experiences and perspectives of adults affected by Ectodermal Dysplasia, who have undergone dental rehabilitation as adolescents, in order to obtain their insights into living with ED. In addition, the study explored their experience of living with this condition from childhood to adulthood and how this impacted on their quality of life. Qualitative analysis helps expand understanding of the relative impact of ED, which
may influence options for future treatment planning and influence policies that dictate access to dental care. In addition, a qualitative methodology provides a vision for patient attitudes and opinions on having and living with this condition. In addition, qualitative analysis themes identified can be implemented in the future into condition-specific questionnaire development (Neergaard et al., 2009, Meaney et al., 2012). This quantitative study was not preceded by any published work on the Irish ED cohort.

In this study, participants discussed when they and their families were first informed of the **ED Diagnosis**. One participant had the diagnosis at 3 years of age, while the other two were aged 6-7 years. The age of diagnosis reflected the limited availability of diagnostic resources and knowledge among medical practitioners who might be expected to have encountered the patients at a younger age during routine medical appointments. The delay in having a diagnosis of ED and absence of a subsequent referral, in particular to dermatology and ophthalmology, were repeatedly raised by the participants as opportunities lost. This was highlighted by one participant who first attended a dermatologist when he was 21 years old and also the fact that two participants were unaware of the problems associated with their reduced lacrimal gland function and the risks to ophthalmological health. In contrast to medical management, the focus group discussions showed that all participants had a dental diagnosis and had commenced dental treatment at young age. They were unable to recall details of the first dental visits themselves as they stated that they were too young. In addition, one participant reported that when he was a child his family treated him the same as his siblings which masked his awareness of the condition at that age, in spite of having severe hypodontia and an overheating problem. On the one hand, he was happy to have been treated the same as his siblings but on the other hand felt that his condition should have been acknowledged in order to support him. He felt that effective symptom management would have been easier for him and could have reduced the personal distress
he felt in his teenage years. This behaviour reflected the importance of family support, and how this family behaviour may have contributed to stress and undermined mental health at a young age.

Ectodermal Dysplasia is a rare condition that requires multidisciplinary professional intervention at a young age to provide the patient and his/her family with a diagnosis and a programme for management of the condition. This was identified by the participants as being most crucial in the areas of dentistry, dermatology, and ophthalmology. These interventions could address the participants’ dental concerns about vague treatment plans and delays in treatment initiation, which were consistent with non-syndromic hypodontia patients who complained in another study, where participants aged 16-25 years stated that “participants outlined very strong feelings of frustration with the delay between initial diagnosis and initial treatment. Participants clearly stated that throughout that time they felt forgotten” (Meaney et al., 2012). Whilst the participants’ dental team effectively managed their dental condition, there seemed to have been little communication in relation to the “big picture” of their dental rehabilitation. This may have led to frustration at the lack of perceived progress during certain phases when no dental intervention was indicated. This may have been particularly prevalent during the teenage years whilst awaiting completion of growth in order to progress to definitive implant-based treatments (Cronin Jr et al., 1994). It would be beneficial for future treatment to discuss and have printed material and resources available explaining the timelines involved in the treatment of young people with ED. This might engage the patient and their families further and also explain the progress of their treatment. It would also highlight that the phasing of treatment is condition specific and provide reassurance that the patients are being managed effectively.
The participants in this study were adults and their definitive dental treatment had been completed many years previously. They had adapted to the symptoms of the condition over many years, which led the participants to report “no impact” of ED on their quality of life at the time of the focus group meeting. This was interesting as their dental symptoms were ranked by them as their foremost complaint when they were growing up in contrast to all the other problems they encountered including skin, eye and temperature control issues. They reported that completion of dental treatment had improved their social confidence, appearance, function, and decreased their levels of anxiety. The influence of dental treatment on their quality of life was consistent with qualitative study findings where restoration of missing teeth in non-syndromic hypodontia patients improved the participants’ anxiety levels and satisfaction with their dental appearance and durability of the treatment in adulthood (Meaney et al., 2012).

As their dental concerns reduced, there was evidence of a response shift where they changed focus to other aspects of their ED to further improve their quality of life. A response shift is where “the individual may change his or her internal standards, values, and/or conceptualisation on the target construct as a result of external factors such as a treatment or a change in health status” (Ring et al., 2005). In this study, the participants’ responses shifted from dental features of the condition to other features such as their eyes and temperature control, as the participants’ assessment of their own quality of life changed. That shift was attributed to the participants’ appreciation of dental treatment which consequently resulted in reporting less impacts of the dental features. The current study was quantitative in nature and response shift cannot be directly measured. However, a previous dental quantitative study reported quality of life improvements, following comprehensive treatment of edentulous patients, showed that a significant improvement in the QoL had been identified and higher satisfaction of the dental treatment had been
identified. When they reflected retrospectively to perceive their pre-treatment levels, they were scored worse on reflection (Ring et al., 2005). The participants in the current study reported satisfaction with the dental treatment that they received, in particular, the definitive treatment that they had after twenty years of age. As a result, their reported impacts focused more on the other features of the condition, and their satisfaction with the dental treatment might result in forgotten or reduced awareness of that impact. The participants in this study tended to consider at this stage that ED had no impact on their teeth, and they reported more discomfort or complaints regarding other ED features, such as overheating, skin and eye dryness.

Regarding **Advance Planning** significant preparation is required by people living with ED, where travel is planned to warm countries or environments. This has led to having to amend holiday destinations and having to pass up possible employment opportunities. The amount of preparation required when travelling to a warm destination or for outdoor sports activities frequently resulted in them cancelling the planned event. Planning for such events included detailed weather forecasting, ensuring access to water at all times, wearing a hat and other protective coverings, having sufficient quantities of skin and dry lubricants, and most interestingly the assurance of facilities in which to cool down body temperature. One participant reported his personal method of temperature regulation which involved frequently placing his hands and arms in cold water to avoid overheating in warm weather destinations.

Interestingly, whilst the focus group interview took place in the winter months, it was observed that all of the participants were wearing lightweight clothes such as T-shirts. When asked by the interviewer if they would like the heater to be turned off, they all answered in the affirmative, which reflected the importance of preparation of their indoor and outdoor environments. All of these preparation tools are restrictions from another
point of view because without effective management, travelling or outdoor activity are restricted.

The focus group discussion identified similar experiences among participants in relation to the **Embarrassment and Bullying** that individuals with ED were subjected to at young ages because of their dental and general appearance. They reported that the teeth were a focus of criticism from others because teeth were “forward-facing” and immediately visible upon smiling and had a significant influence on the whole face appearance and self-esteem. In addition, it was mentioned that other people may be bald or have dry skin, but everyone has teeth that are displayed in a smile. This self-consciousness with dental appearance was consistent with a study by Meaney and co-workers involving participants aged 16-25 years of age who had non-syndromic hypodontia where participants reported the negative impact of their smile and appearance as they grew up and that improving their smile was a motivational factor for having the dental treatment to improve their appearance and social confidence (Meaney et al., 2012). In the current study, overcoming bullying was not reported *per se*, but the participants’ advice to children with the same condition was “live your life” and all “people are the same”. These pieces of advice might reflect the social trauma from bullying that they have experienced, and that they want children with ED to overcome the bullying by being confident and ignoring the condition’s impact on their teeth and general appearance.

**Hypodontia** and the presence of malformed teeth were identified by participants as the main issue associated with the condition and their overriding concern through childhood and adolescence. This was because dental issues had more than one impact, as discussed earlier. They influenced self-esteem, appearance, and restorations themselves also caused some anxiety (Davis et al., 1998). From a daily routine perspective, missing teeth had other influences in relation to difficulty chewing food and difficulties with speech. The
consequences of having these dental issues influenced their food choices from two perspectives. Firstly, it limited the type of food that they chose and secondly, it required that additional efforts be made to source the best alternative to maintain a balanced nutrient intake. One participant who was playing sport mentioned the use of protein shakes as an alternative to overcome the limitations of a reduced dentition and ensure sufficient protein intake. This reflected the impact of the missing teeth function beyond food chewing and speech, which was the influence of the missing teeth on general nutrient intake. The subject recognised that his intake of food was insufficient for the amount of physical exercise he was undertaking so he took additional nutrients in a manner that did not require mastication. In addition, missing and malformed teeth negatively impacted their ability to communicate with other people for two reasons. The first was that when their front teeth were missing it influenced speech quality and was associated with lisping during speech. This was reported to be most concerning when their maxillary prosthesis no longer fitted as well as when it was initially fabricated. It was appreciated by the group retrospectively that this was due to their own facial growth which resulted in the dentures becoming ill-fitting. For future dental treatments, it might be advisable to explain to ED patients the reasons why dental prostheses may become loose and also consider interventions that may reduce the impact of same, such as the timing of dental recalls at periods of increased growth rates or the use of denture adhesives. The second reason was that malformed and missing teeth negatively influenced their appearance and lowered their self-esteem. This, in combination with the speech issues, led to self-consciousness and being introverted which was considered by the group as a significant social burden of the condition.

Missing and malformed teeth influenced their dental and general appearance and was interpreted by the group from different perspectives including aesthetics, self-esteem, and
social communication. Aesthetically, they reported that the teeth were “forward-facing” and immediately visible upon smiling and had a significant influence on the dental and whole facial appearance. They concluded that dental rehabilitations were fundamental to reinstate both aesthetically acceptable and “normal” dental and facial appearances. This consequently restored their self-esteem and ability to smile, providing confidence during social life interactions. This was consistent with findings from the study by Davis and co-workers who stated that “it also indicated that patients tended to feel, not that their appearance attained the ideal of beauty, but rather it was normalised and no longer needed to be a source of concern” (Davis et al., 1998).

Management of Missing and Malformed Teeth was the first line of treatment because of their multiple impacts, functionally, aesthetically, and socially. All participants had severe hypodontia which had been managed using removable dentures from young ages: “I have dentures since I remember”, until their growth had been completed at the end of their teenage years. Each participant had new dentures fabricated approximately every 18-24 months to accommodate the growth pattern.

The participants relayed the importance and positive impact of denture provision on them in their teenage years. They were very positive about having front teeth and looking like their peers. However, the dentures were also a source of anxiety at certain periods. During childhood there are periods of rapid growth and changes in intraoral anatomical structure occur, which consequently resulted in the dentures not being as well adapted, leading to soft tissue irritation and compromised fit, resulting in a less secure denture. Treatment appointments required for adjustment and the provision of new dentures were reported to have resulted in increased initial discomfort, but more importantly, were a source of stress. This stress was exacerbated as the need for multiple visits resulted in loss of time at school, college and free time. It may be worth considering future dental care at times when
they are least inconvenient such as during holiday periods where possible (Saltnes et al., 2017a).

On reflection, the participants also reported increased levels of anxiety and stress in relation to having to have their malformed teeth restored with composite resin restorations, which they referred to as “caps”. It appeared that there was a significant ongoing fear of doing something that would result in one of these restorations debonding or fracturing. The participants all agreed that they were prone to overthinking about “cap” failure and that it was constantly at the back of their minds. Malformed teeth restored for aesthetic reasons with tooth coloured restorations are subject to chipping and staining over time and may have to be altered to accommodate new dentures. They may also be placed in areas of increased force if placed adjacent to a denture and this may increase the possibility of failure. These restorations improved self-esteem and aesthetics; however, the maintenance and adjustment procedures were a source of anxiety in the same way as denture treatment during development. However, once growth was completed and definitive dental prostheses fabricated, with a clear understanding of the future maintenance required, the participants reported that their anxiety reduced, or even in their eyes, eliminated. The participants had been living with their current dental rehabilitations for some time and thankfully, each required minimal maintenance. This may, in turn, have resulted in them reflecting less favourably on their childhood where their restorations had increased risks of failure as the materials used were inherently weaker or required frequent change due to their growth (Meaney et al., 2012).

The reported impacts of ED from childhood through adulthood demonstrated the challenges that those participants faced with the condition in terms of body temperature regulation, dental issues, skin and eye dryness which resulted in a loss of self-esteem and increased anxiety associated with the condition. Adaptation and management of their own
identity was practised by each participant who each, over time, became “experts in self” to empower them to deal with overheating, dry eyes and skin and facilitated their own adaptation to the condition. This “expert in self” behaviour was consistent with other populations who have a chronic condition such as Diabetes Mellitus where children’s’ self-knowledge of the symptoms led to them developing self-management strategies that suited their lifestyle, a concept called “body-listening”. This concept is developed through empirical knowledge and their own experience to gain an understanding of the condition and place themselves within the disease context. In addition to “trial and error” to identify a comfortable and safe status, a combination of the “body-listening” and basic knowledge provided a greater understanding of the condition which facilitated improved adaptation (Ingadottir and Halldorsdottir, 2008). For this Ectodermal Dysplasia group, in the absence of basic knowledge, peer support and a cure for the condition, they reported that they worked primarily through “trial and error” in order to deal with the condition. They were mostly successful in completing these aims.

However, given the absence of professional advice, the affected population sometimes could not manage higher levels of adaptation. This was a source of frustration for the group where one of the participants had to decline the opportunity to play support professionally, while another participant lost most of the sight in one eye as a result of complications with dry eyes.

It was interesting to note that whilst all the group had individually adapted well to living with ED, in the short time they were together they exchanged many solutions that they had found to their non-dental problems. Information was exchanged in relation to temperature regulation, appropriate eye care and skin management. The group reported that they are still in contact via a WhatsApp group and are very keen to reach out to younger ED patients and their families. The main driver for this was to equip others with effective tools to
manage non-dental problems that they themselves had to learn through trial and error and becoming “experts in self”.

It was clear that the group used significant creativity to manage their own identities as reflected by their extensive efforts to identify what worked for them to adapt to the surrounding environment. This was achieved through trial and error and over the years, they identified what kept them comfortable which empowered them to live what they regarded as “normal lives” (Ingadottir and Halldorsdottir, 2008).

Examples included:

(1) The use of specific lubricants for skin and eye dryness as preventative measures.

(2) Body temperature regulation through placing just their arms in a sink of cold water to cool down. Significant discussion also took place in relation to new developments in clothing to regulate body temperature. Products such as E.COOLINE® personal cooling clothing offer huge future potential for ED patients in managing temperature control during sport. However only one of the group had been aware of the products and information was exchanged at the time and subsequently

(3) Intake of nutritional supplements such as protein shakes when their dental prosthesis did not fit or while new prostheses were being fabricated.

This self-adaptation reflected a lack of professional support and guidance to provide the ED population with suitable medical information and practical adaptation techniques. Also, each of the participant’s behaviour identified a lack of peer support for information exchange and the absence of organisations to provide support and guidance for affected individuals and their caregivers.

Management of the Dental Features of Ectodermal Dysplasia was different from that of other features as a result of the early intervention of dental professionals and early commencement of their dental treatment. Dental treatment provided included bonded
composite resin restorations, removable dentures (partial or complete) and fixed prostheses that underwent regular maintenance and replacement as the participants developed until they received definitive final treatment in their late teenage years. These prostheses were considered by the participants as tools for adaptation to having missing teeth and these prostheses had significant importance in restoring appearance, function, and self-esteem. In addition, all adaptation tools together played an important role in maintaining mental health and coping with stress which reflected the stress generated by ED and how important it is for the population affected with the condition to cope in the absence of medical support such as dermatological and ophthalmology intervention and provision of suitable advice and medication as required. It was noteworthy that the most significant issue for all the participants was their dental problems and that these were greater than problems with hair, skin and temperature regulation at all stages during their childhood. It is important that this fact be recognised by both the dental profession and in future health policy development.

The results of this study highlighted the need to introduce a national programme to facilitate the early diagnosis and appropriate referral of the affected populations by medical and dental practitioners. Multidisciplinary teams that involve both medical and dental specialists should be established in the public health service in Ireland. This would facilitate early diagnosis and the commencement of treatment at the earliest possible stage (Hobkirk et al., 1994). Multidisciplinary clinics for ED patients would increase the provision of coordinated care in a single location and increase cooperation across disciplines.

The provision of multidisciplinary care has been recommended by the National Foundation of Ectodermal Dysplasia (NFED) in the United States, through the foundation’s website. It offers the affected population and their families valuable guidance and information and facilitates appropriate medical and dental care with specialist support and access. This, in
turn, provides a “home” for ED patients and in addition, improves access to care to address the reported anxiety and frustration levels of this cohort and their families. In the UK, the Ectodermal Dysplasia Society works together with the ED population, their families, researchers, and health professionals to provide advice and assistance for condition management. The charitable organisation also works to increase the ED population’s awareness of the condition, experience sharing, and provide political lobbying to support ED patients.

In Ireland, the nucleus of an ED Society had been formed by the family of a patient with the condition. The society aims to provide the same support and guidance to the ED population as UK ED Society and NFED in the United States currently undertake. The focus group participants reported limited events and benefit from the society so far, but their meeting in this focus group discussion provided them personally with the motivation to further activate the society and improve its service to the ED population.

Although context-specific, the community medical and dental screening programmes should include detection of ED signs and symptoms in order to identify possible cases and accelerate access to appropriate health-care facilities and avoid the complications of a delayed diagnosis (Hobkirk et al., 1994, Meaney et al., 2012). Furthermore, support for the multidisciplinary team could be provided through charitable organisations such as the National Foundation of Ectodermal Dysplasia (NFED) in the US.

One of the other most important elements was the Lack of Peer Support in person or via social media networks allowing people to share experiences and guide the affected population and their caregivers. In this study, the participants were highly educated and yet their first meeting was at this focus group discussion. They found that being in contact for information and experience sharing was important as each of them shared an adaptation method or medical advice that was not known by the other two participants.
Examples included putting hands in a sink of cold water to overcome overheating, the importance of an ophthalmologist consultation at a young age to avoid the side effects of eye dryness, and lastly the use of cooling vests during sports.

The group were very positive about what they could offer other affected individuals and were keen to pursue avenues that would facilitate information exchange. Based on their personal experiences with the condition they felt that they were ideally placed to offer appropriate and strategic advice to children with the condition in a peer-to-peer fashion. They felt that the principal message that they wished to convey was in relation to self-confidence and that affected individuals should behave like any other child and not consider themselves different in any way. This advice reflected that they were themselves confident and in control of their condition. They each acknowledged the negative impact of ED on their own self-esteem while growing up, which they reflected may have been different if they had received the information that they were prepared to share with younger ED individuals. The participants offered to volunteer providing advice and guidance to children and their parents, which they could present based on their own experiences. They were also keen to discuss tools and techniques for how one can adapt to the challenges of living with ED. In addition, they felt that it was vitally important to emphasise the benefits of early referral to dental and medical specialists.

4.2.1 Strengths, Limitations, and Future Research

This study section was a qualitative study which adopted a descriptive approach in order to collect detailed and rich data in relation to attitudes among ED adults regarding the impact of Ectodermal Dysplasia on their own quality of life and elucidate their experience from childhood to adulthood. A focus group methodology was adopted to collect the data, which
was very useful for a variety of reasons. It allowed for in-depth group discussion of perception and a unique opportunity for experience sharing. The focus group interactions allowed for the emergence of new data, for example, innovative adaptation methods to cope with the condition, and the importance of peer interaction and information sharing. Furthermore, this study provided rapid, immediate access to the information, perceptions, and thoughts of the participants. The dynamic nature of the focus group resulted in further in-depth discussion, which consequently allowed the participants to easily share their insights and further develop the thematic areas. This study also had the strength of involving educated professional adults who had been through dental treatment since they were young, which reflected accurate perceptions of the condition at different ages and included physical, mental, and psychological aspects. The discussed thematic analysis can, in the future, be incorporated into a condition-specific questionnaire, which will allow further examination of the realities and perceptions of the condition.

This study also resulted in the creation of new connections between individuals with the condition which resulted in peer support for the first time for them all. Furthermore, they offered to volunteer their services to provide others with knowledge regarding the methods of adaptation to and management of the condition. These were areas that they felt were missing from their own development and they wished to redress the balance.

As with any study, this study had limitations. Qualitative studies’ results are not generalisable to an overall population and they are context specific. However, this methodology is valid in chronicling the attitudes and opinions of those participants. In this study, there was a potential for information recall bias as the participants were asked about their experiences when they were young which may not be answered accurately as a result of challenges with recall or limited cognitive development at that age. This study had the weaknesses that it only included a limited number of participants. Sample sizes and
selection procedures in qualitative research do not permit statistical estimates to be made with any calculable degree of accuracy. Furthermore, all participants were of the same gender. However, Ectodermal Dysplasia prevalence is higher in males, and females are less symptomatic so involving males may highlight the more extreme manifestations of living with ED. Nevertheless, this research has provided a unique insight into the perspectives of the ED population and its findings can inform future longitudinal quantitative research to ascertain the views of more children and parental perceptions before and after both dental and medical treatment.

This study was also valuable as it will increase dentists’ and medical professionals’ knowledge and understanding of this population. From the findings of this focus group, some recommendations can be made to those who provide holistic care to this cohort. From a dental features management point of view, early commencement of treatment which includes, in the majority of cases, removable dentures and tooth colour restorations are beneficial in restoring dental function in terms of speech, food chewing, and appearance. These tooth coloured restorations should be maintained regularly to repair any chipping and to remove any stains, together with regular adjustments of denture prostheses at ages of development to provide ED patient with comfortable restorative treatment that will increase self-esteem, improve dietary intake, improve appearance, and decrease levels of dental restoration related anxiety. Furthermore, detailed awareness of short-term and long-term treatment plans is important for dental practitioners to have and also share with ED patients and their families. In the majority of cases, short-term treatment includes removable dentures with aesthetic tooth colour restorations (composite resin), while the long-term treatment requires multidisciplinary dental intervention, commonly including orthodontic treatment, pre-prosthetic surgery, implant placement, and provision of implant supported prostheses. The key to patient satisfaction
and achieving the psychological and physical function benefits of dental treatment is dental prosthesis maintenance, particularly when patients are actively growing.

In addition, general dental and medical practitioners should refer their patients for dermatological and ophthalmological consultations as part of ED management. This is due to limited public awareness of the complications of skin and eyes dryness and the importance of early intervention when complications occur. In addition, early consultations will result in providing ED subjects with appropriate medical advice, management and information related to their ED-related skin and optical features. Currently, there is no central referral multidisciplinary centre for this population where they can have their combined dental and medical treatment provided. Such a centre would provide treatment for the population with interventions from different disciplines at the same time. In addition, it would provide a home for their collaborated treatment in a single site that would address the challenges associated with travelling between different treatment hospitals.

Secondly, it is incumbent upon the dentist providing care to explain in detail the short-term and long-term treatment plans. The focus group clearly reported that the participants’ limited awareness of their treatment stages and timelines were a source of anxiety and stress and that increased knowledge of planned interventions would have positively influenced their experience and decreased the anxiety associated with treatment.

Furthermore, the same consideration should be applied when the treatment plan is being discussed with parents at the start of the treatment. This would provide useful guidance on how the treatment is staged based on their child’s growth and development.

Thirdly, it is important to be aware of how the dental environment can be modified to increase the comfort of patients with ED. Effective temperature regulation with sufficient lubricants and water being offered during treatment procedures would acknowledge the
issues facing these patients and demonstrate empathy. This setting would provide greater comfort during dental treatment and reduce the risk of overheating and complications of a dry mouth such as from cotton roll abrasions. In addition, this would provide a suitable environment during treatment and improve the patients’ and parents’ perceptions of the treatment provided.

It is also recommended for future research that the perception of those who are under treatment is compared with that of those who have finished dental treatment. This might assist in providing further thoughts, visions, and perceptions from the participants who are under treatment that could be missed or considered of less importance once treatment has been completed. In addition, inviting parents or caregivers of those currently under treatment or have just completed treatment to focus group discussions would provide further understanding of perceptions and information about the condition’s impact on the whole family unit. Furthermore, parental inclusion may overcome this study’s childhood information recall bias.
5 Conclusions

Ectodermal Dysplasia is a rare congenital condition and, in this study, dental features and impacts on the Oral Health Related Quality of Life (OHRQoL) in children and adolescents have been reported. Sever hypodontia had been reported in more than two thirds of this study subjects, with a mean number of (18.5) missing teeth. In comparison to the control group, the children and adolescents with ED reported poorer OHRQoL scores. The children’s increased cognitive development and awareness of the condition resulted in higher scores in the older age group (11-14 years).

Parents were clearly concerned about their children and a combination of their feelings and the limited knowledge of the impacts of the condition on their children resulted in higher scores reported by parents in the younger age group (8-10 years). However, the older age group participants, as they were more competent in expressing themselves, reported higher scores than their parents which can be related to their awareness of the condition and how the dental features of ED impacted them.

With respect to the parental reports, a very weak relationship was identified between child and parental perception for both age groups. This highlights the fact that parents, whilst an essential source of information, should not be used as proxies when determining the OHRQoL of their children.

The focus group report reflected life experiences of living with ED from childhood to adulthood with particular emphasis on emotional and psychosocial challenges. Challenges from dental, dermatological, and ophthalmological features of the condition were reported, with greatest impacts reported from the dental features of the condition. In spite of all the challenges faced by ED patients, dental issues dominated their childhood and adolescent years. Dental features were the main source of their everyday challenges and
highlighted that they should be a priority for management of ED patients above all other features of the condition in the future. Dental management over the years from young ages by dental professionals resulted in an improvement in their self-esteem, appearance, mental health, and dental function, which consequently improved their dental health.

Failure to achieve a diagnosis of ED at an early age surprisingly was found for all focus group participants. This resulted in lost opportunities for early medical intervention to manage the medical features of the condition, such as eye dryness and temperature control. Diagnosis at a young age and multidisciplinary intervention with clearly defined long and short-term treatment plans should, in the future, result in decreased anxiety and improvement in life experiences for those with the condition. Absence of medical management resulted in participants having to seek and identify adaptation tools which resulted in the patients becoming “experts in self”. All participants were keen to find methods of transferring their experiences to the next generation of ED patients and their parents. The prior knowledge and experience of the focus group participants should serve to ameliorate some of the challenging aspects of the condition.

Both sections of the current study delineated the impact of the condition in both youth and adulthood. The reported higher impact by parents in contrast to their children at age 8-10 years was also identified in the focus group as all participants had difficulty remembering the age of diagnosis and when dental treatment commenced, which represented an early start of dental treatment in children and reflected again the limited cognitive development at that age. The children aged 11-14 years reported higher scores than those in the younger age group which confirmed the findings of the focus group who recalled difficulty in the adaptation to dentures as a consequence of mandibular growth and development. In addition, the limited knowledge of the children about possible adaptation tools and home remedies that experienced adults used increased the impacts of ED on them. Furthermore,
reported higher scores in the functional limitation and oral symptoms domains were consistent with the difficulties in chewing food and speech reported by adults with ED that led them to find food alternatives to balance their dietary intake, such as having protein shakes when they were uncomfortable chewing food.

The focus group discussion reported on the emotional impact of the condition in terms of dental and general features at a young age and their experiences of bullying, which were also reflected in the quantitative group results through increased scores in emotional well-being domains. In both groups, this was also influenced by hypodontia and malformed teeth and the resultant impacts on appearance and self-esteem. These reported features can be used as a guide to building an approach that can assist in providing targeted dental and medical holistic care that can help in the management of the dental and general features of the condition. In addition to early diagnosis, support networks will be key for the future in ensuring appropriate treatment for medical features and the provision of tools to facilitate adaptation to living with ED.
6 References


NESS, L. R. 2015. Are we there yet? Data saturation in qualitative research.


THOFELT, A. 2013. Young Individuals with Oligodontia: Outcome of Prosthetic Treatment Following Multidisciplinary Treatment Planning.


Appendix 1: Ethical Approval letter

Dr Shikre Aghaie,
Postgraduate Student,
Restorative Dentistry and Periodontology,
Dublin Dental University Hospital,
Lincoln Place,
Dublin 2

20th December 2018

REF: Dental Features of an Irish Cohort with Ectodermal Dysplasia and Resultant Impact on their Quality of Life

REC: 2018-12 Chairman’s Action (8)
(Please quote reference on all correspondence)

Date of Valid Submission to REC: 23.11.2018
Date of Ethical Review: 20.12.2018

Dear Dr Aghaie,

The REC is in receipt of your recent request to TUH/SJH Research Ethics Committee in which you queried ethical approval for the above named study.

The Chairman, Prof. Richard Dean, on behalf of the Research Ethics Committee, has reviewed your correspondence and has approved this study. The following documents were reviewed:

- Standard Application Form, dated 23.11.2018
- Focus Group/Study participant/Control participant PIL (please add version and date)
- Consent Form (please add version and date)

Applicant must submit an annual report for ongoing projects and an end of project report upon completion of the study. It is the responsibility of the researcher/research team to ensure all aspects of the study are executed in compliance with the General Data Protection Regulation (GDPR), Health Research Regulations and the Data Protection Act 2018.

Yours sincerely,

REC Officer – Dr Shardih O’Neill
SJH/TUH Research Ethics Committee

The SJH/TUH Joint Research and Ethics Committee operates in compliance with and is constituted in accordance with the European Communities (Clinical Trials on Medicinal Products for Human Use) Regulations 2004 & ICH GCP guidelines.
Appendix 2: Introduction Letter to Patients

Dear Sir/Madam

**Title of research study:** Dental features of an Irish patient cohort with ectodermal dysplasia and resultant impact on their quality of life.

We would like to invite you to consider taking part in this study being undertaken at the Dublin Dental University Hospital (DDUH). We wish to learn first-hand about people’s experience of living with ectodermal dysplasia (ED). We are looking to talk to children/their parents and adolescents to find out the impact of living with ED, and the effects of missing teeth and other oral symptoms through completion of a small number of questionnaires that seek your views and experiences.

We will also complete a dental examination and assessment and offer advice as to future treatment needs. The purpose of this is to establish the baseline dental health of patients living with ED and determine their treatment needs into the future.

You can read more about the study in the information sheet provided and if you wish to ask any questions about the study contact details are supplied below. If you are interested in taking part then we can meet you in DDUH on a day and time that is most convenient for you.

The administrative contact person for the study is Ms. Cathy Dillon and she can be contacted by:

(1) Telephone: 01 612 7603
(2) Email: Cathy.dillon@dental.tcd.ie

(3) By post using the slip provided below, in the envelope supplied.

Thank you for your interest in this project.

____________________  ____________________
Dr. Michael O’Sullivan          Dr. Shkre Agkhre
Associate Professor /Consultant                Prosthodontics Postgraduate
Programme Director

**Title of research study:** Dental features of an Irish patient cohort with ectodermal dysplasia and resultant impact on their quality of life.

____________________
Signed:

I am interested in taking part or learning more about the study with Drs. Michael O’Sullivan and Shkre Agkhre and am willing for them to contact me:

NAME:

Telephone Number:

Email:

Signed:

Date:
Appendix 3: Introduction Letter to Controls

Dear Sir/Madam

Title of research study: Dental features of an Irish patient cohort with ectodermal dysplasia and resultant impact on their quality of life.

We would like to invite you to consider taking part in this study being undertaken at the Dublin Dental University Hospital (DDUH). We wish to learn first-hand about people’s experience of living with a condition called ectodermal dysplasia (ED) and compare them to patients without the condition.

As part of the study we need to match patients with ED with patients without the condition. This allows comparison of the effects of the condition with the general population. You and your parents will be asked to complete some questionnaire about your daily life and your dental health.

You can read more about the study in the information sheet provided and if you wish to ask any questions about the study contact details are supplied below. If you are interested in taking part then we can meet you in DDUH on a day and time that is must convenient for you.

The administrative contact person for the study is Ms Cathy Dillon and she can be contacted by:
(1) Telephone: 01 612 7603
(2) Email: Cathy.dillon@dental.tcd.ie
(3) By post using the slip provided below, in the envelope supplied

Thank you for your interest in this project.

____________________  ____________________
Dr. Michael O’Sullivan  Dr. Shkre Agkhre
Associate Professor /Consultant  Prosthodontics Postgraduate
Programme Director

**Title of research study:** Dental features of an Irish patient cohort with ectodermal dysplasia and resultant impact on their quality of life.

____________________
I am interested in taking part or learning more about the study with Drs. Michael O’Sullivan and Shkre Agkhre and am willing for them to contact me to act as a control patient:

NAME:
Telephone Number:
Email:
Signed:
Date:
Appendix 4: Patient Information Leaflet

1. **Title of study:** Title of research study: Dental features of an Irish patient cohort with ectodermal dysplasia and resultant impact on their quality of life.

2. **Introduction:** We would like to invite you to participate in this postgraduate research project. You should only participate if you want to; choosing not to take part will not disadvantage you in any way. Before you decide whether you want to take part, it is important for you to understand why the research is being completed and what your participation will involve. Please take time to read the following information carefully and discuss it with others if you wish. Ask us if there is anything that is not clear or if you would like more information.

We are Drs Michael O’Sullivan and Shkre Agkhre from the Dublin Dental University Hospital and we are conducting a project about the dental aspects of ectodermal dysplasia (ED). We would like to discover more about your personal experience of living with ectodermal dysplasia and invite you to join this research study.

3. **Procedures:** If you agree to participate we will complete a dental assessment of all your teeth and oral health status in the Dublin Dental University Hospital on a day and a time that suits you. In addition this assessment will also help us determine your future treatment needs.

We would also like to ask a few questions about your views and experiences of living with ectodermal dysplasia and its impact on your quality of life. We will post you questionnaires to be completed by you and your parents. All information provided will be confidential and used as part of the study only.

4. **Benefits:** You will receive a full dental assessment in Dublin Dental University Hospital without being included in the assessment waiting list. After assessment you will be included in the treatment waiting list for dental treatment, if required. Also, by obtaining information from you we will understand better how ectodermal dysplasia affects the quality of life of patients. It will also help us, as clinicians, to predict the quality and benefit of treatment we provide to our patients. The results of the study will also be published in journals or presented at conferences.

5. **Risks:** There is no risk involved in this study except of course the taking up of your valuable time. You may of course, refuse to answer some or all the questions if you don’t feel comfortable with any of them.
6. **Exclusion from participation:** You have been told that you cannot be in this study if any of the following are true: (1) you are older than 20 years, and (2) if you do not consent to being involved in the project.

7. **Confidentiality:** Your identity will remain confidential. Your name will not be published and will not be disclosed to anyone outside the research study group. All data will be anonymised in any reports we write and no individual will be identified.

8. **Compensation:** Your dentists are covered by standard dental malpractice insurance. Nothing in this document restricts or curtails your rights.

9. **Voluntary Participation:** It is up to you to decide whether to take part or not. If you decide to volunteer to participate in this study, you may withdraw at any time without giving a reason. If you decide not to participate, or if you withdraw, you will not be penalised and it will not affect the standard of care you receive. Should you decide to withdraw from the study you need to do so before the 28th of February 2020 when the final results will be written up.

10. **Stopping the study:** You understand that your dentist may stop your participation in the study at any time without your consent.

11. **Permission:** This study has Joint Research Ethics Committee approval.

12. **Further information:** You can get more information or answers to your questions about the study, your participation in the study, and your rights, from Ms Cathy Dillon who can be telephoned at 01-6127200.

Many Thanks,

Dr. Shkre Agkhre

Postgraduate student in Prosthodontics,

Dublin Dental University Hospital,

Lincoln Place,

Dublin 2
Appendix 5: Control Information Leaflet

Patient Information Leaflet

1. **Title of study:** Title of research study: Dental features of an Irish patient cohort with ectodermal dysplasia and resultant impact on their quality of life.

2. **Introduction:** We would like to invite you to participate in this postgraduate research project. You should only participate if you want to; choosing not to take part will not disadvantage you in any way. Before you decide whether you want to take part, it is important for you to understand why the research is being completed and what your participation will involve. Please take time to read the following information carefully and discuss it with others if you wish. Ask us if there is anything that is not clear or if you would like more information.

We are Drs Michael O’Sullivan and Shkre Agkhre from the Dublin Dental University Hospital and we are conducting a project about the dental aspects of ectodermal dysplasia (ED). You are being asked to participate as you do not have the condition.

Ectodermal dysplasia is a group of inherited disorders that involve defects in the hair, nails, sweat glands and teeth. The condition most frequently results in the person having multiple missing teeth and this study is seeking to examine the impact of this feature and compare it to people without the condition (controls).

3. **Procedures:** If you agree to participate we would also like to ask a few questions about your, and your parents, views of your current dental health. We will post you questionnaires to be completed by you and your parents. All information provided will be confidential and used as part of the study only.

4. **Benefits:** There are no direct benefits to you in participating in the study. You will, however, be providing us with invaluable information about your dental health and allow us to compare it to people living with ectodermal dysplasia. We can then pinpoint the dental effects that the condition beings. This will allow us to develop a better understanding of patients living with ED and address their dental health needs.

5. **Risks:** There is no risk involved in this study except of course the taking up of your valuable time. You may of course, refuse to answer some or all the questions if you don’t feel comfortable with any of them.
6. **Exclusion from participation:** You have been told that you cannot be in this study if any of the following are true: (1) you are older than 20 years, and (2) if you do not consent to being involved in the project.

7. **Confidentiality:** Your identity will remain confidential. Your name will not be published and will not be disclosed to anyone outside the research study group. All data will be anonymised in any reports we write and no individual will be identified.

8. **Compensation:** Your dentists are covered by standard dental malpractice insurance. Nothing in this document restricts or curtails your rights.

9. **Voluntary Participation:** It is up to you to decide whether to take part or not. If you decide to volunteer to participate in this study, you may withdraw at any time without giving a reason. If you decide not to participate, or if you withdraw, you will not be penalised and it will not affect the standard of care you receive. Should you decide to withdraw from the study you need to do so before the 28th of February 2020 when the final results will be written up.

10. **Stopping the study:** You understand that your dentist may stop your participation in the study at any time without your consent.

11. **Permission:** This study has Joint Research Ethics Committee approval.

12. **Further information:** You can get more information or answers to your questions about the study, your participation in the study, and your rights, from Cathy Dillon who can be telephoned at 01-6127200.

Many Thanks,

Dr. Shkre Agkhre

Postgraduate student in Prosthodontics,

Dublin Dental University Hospital,

Lincoln Place,

Dublin 2.
Appendix 6: Consent Form

CONSENT FORM

**Title of research study:** Dental features of an Irish patient cohort with ectodermal dysplasia and resultant impact on their quality of life

This purpose of this study and this consent form have been explained to me. My dentist has answered all my questions to my satisfaction. I believe that I understand what will happen if I agree to be part of this study.

I have read, or had read to me, this consent form. I have had the opportunity to ask questions and all my questions have been answered to my satisfaction. I freely and voluntarily agree to be part of this research study, though without prejudice to my legal and ethical rights. I have received a copy of this agreement.

**PARTICIPANT’S NAME:**

**PARTICIPANT’S SIGNATURE:**

Date:

**Date on which the participant was first furnished with this form:**

**Statement of investigator’s responsibility:**
I have explained the nature, purpose, procedures, benefits, risks of, or alternatives to, this research study. 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.

**Dentist’s signature:**

Date:

(Keep the original of this form in the investigator’s records and give one copy to the participant.)
Appendix 7: Focus Group Consent Form

CONSENT FORM

Title of research study: Dental features of an Irish patient cohort with ectodermal dysplasia and resultant impact on their quality of life

This purpose of this study and this consent form has been explained to me. The researcher has answered all my questions to my satisfaction. I believe that I understand what will happen if I agree to be part of this study.

I have read, or had read to me, this consent form. I have had the opportunity to ask questions and all my questions have been answered to my satisfaction. I freely and voluntarily agree to be part of this research study, though without prejudice to my legal and ethical rights. I have received a copy of this agreement.

PARTICIPANT’S NAME:

PARTICIPANT’S SIGNATURE:

Date:

Statement of investigator’s responsibility:
I have explained the nature, purpose, procedures, benefits, risks of, or alternatives to, this research study. 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.

Dentist’s signature:

Date:

(Keep the original of this form in the investigator’s records and give one copy to the participant.)
Appendix 8: Focus Group Information Leaflet

Patient Information Leaflet

13. **Title of study:** Title of research study: Dental features of an Irish patient cohort with ectodermal dysplasia and resultant impact on their quality of life.

14. **Introduction:** We would like to invite you to participate in this postgraduate research project. You should only participate if you want to; choosing not to take part will not disadvantage you in any way. Before you decide whether you want to take part, it is important for you to understand why the research is being completed and what your participation will involve. Please take time to read the following information carefully and discuss it with others if you wish. Ask us if there is anything that is not clear or if you would like more information.

We are Drs Michael O’Sullivan and Shkre Agkhre from the Dublin Dental University Hospital and we are conducting a project about the dental aspects of ectodermal dysplasia (ED). We would like to discover more about your personal experience of living with ectodermal dysplasia and invite you to join this research study.

15. **Procedures:** If you agree to participate we will complete a dental assessment of all your teeth and oral health status in the Dublin Dental University Hospital on a day and a time that suits you. In addition this assessment will also help us determine your future treatment needs.

We would also like to ask a few questions about your views and experiences of living with ectodermal dysplasia and its impact on your quality of life. We will post you questionnaires to be completed by you and your parents. All information provided will be confidential and used as part of the study only.

16. **Benefits:** You will receive a full dental assessment in Dublin Dental University Hospital without being included in the assessment waiting list. After assessment you will be included in the treatment waiting list for dental treatment, if required. Also, by obtaining information from you we will understand better how ectodermal dysplasia affects the quality of life of patients. It will also help us, as clinicians, to predict the quality and benefit of treatment we provide to our patients. The results of the study will also be published in journals or presented at conferences.

17. **Risks:** There is no risk involved in this study except of course the taking up of your valuable time. You may of course, refuse to answer some or all the questions if you don’t feel comfortable with any of them.
18. **Exclusion from participation:** You have been told that you cannot be in this study if any of the following are true: (1) you are older than 20 years, and (2) if you do not consent to being involved in the project.

19. **Confidentiality:** Your identity will remain confidential. Your name will not be published and will not be disclosed to anyone outside the research study group. All data will be anonymised in any reports we write and no individual will be identified.

20. **Compensation:** Your dentists are covered by standard dental malpractice insurance. Nothing in this document restricts or curtails your rights.

21. **Voluntary Participation:** It is up to you to decide whether to take part or not. If you decide to volunteer to participate in this study, you may withdraw at any time without giving a reason. If you decide not to participate, or if you withdraw, you will not be penalised and it will not affect the standard of care you receive. Should you decide to withdraw from the study you need to do so before the 28th of February 2020 when the final results will be written up.

22. **Stopping the study:** You understand that your dentist may stop your participation in the study at any time without your consent.

23. **Permission:** This study has Joint Research Ethics Committee approval.

24. **Further information:** You can get more information or answers to your questions about the study, your participation in the study, and your rights, from Ms Cathy Dillon who can be telephoned at 01-6127305.
Appendix 9: Focus Group Topic Guide

Adult Patient Experiences with Ectodermal Dysplasia (ED)

Following Full Mouth Rehabilitation Treatment: A Qualitative Analysis

Focus Group Topic Guide

1. Introduction

Good morning and welcome to our session. Thanks for taking the time to join us to talk about Ectodermal Dysplasia. My name is Shkre Agkhre and I’ll be assisting Dr Dougall. We are here in Dublin dental hospital conducting this research that investigates the impact of Ectodermal Dysplasia on patient’s quality of life. This research is supervised by Dr Michael O’Sullivan.

You were invited because you have completed restorative treatment for any missing or malformed teeth here in DDUH, and have life experience with Ectodermal Dysplasia.

The aims and objective of this study is:

- To know your experience with this condition.
- To Rank your abnormal tooth condition in relation to other ED features.
- To determine the impact of receiving dental treatment on your quality of life.

There are no wrong answers but rather different points of view. Please feel free to share your point of view even if it differs from what others have said. Keep in mind that we're just as interested in negative comments as well as positive comments.

You’ve probably noticed the iPad. We're tape recording the session for 1 Hr because we don't want to miss any of your comments. People often say very helpful things in these discussions and we can't write fast enough to get them all down. We will be on a first name basis, and we won't use any names in our reports. You may be assured of complete confidentiality.
• Now I’ll handle you the consent form to read and sign it.
• Mobile phones should be turned off/silent
• Do you have any questions before we proceed?

Well, let's begin. We've placed name cards on the table in front of you to help us remember each other's names. Let's find out some more about each other by going around the table. Tell us your name and where you live.

2. **General Health**

   • Meaning of general health and its criteria

3. **The condition**

   • Think back to when you first became aware of the condition. What were your first impression?
     ➢ How did you develop your knowledge in the area to inform yourself?

   • Further information source about the condition
     ➢ ED society
     ➢ Social media group
     ➢ Internet
     ➢ Family or friends

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<thead>
<tr>
<th>Prompt These</th>
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<tr>
<td>Food types</td>
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<td>Exercise</td>
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<tr>
<td>Sun exposure</td>
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<tr>
<td>Stress management</td>
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</tbody>
</table>

| Genetics |
| Internet (inf.) |
| Types of ED |
• Feeling about the condition and its influence on you
  ➢ The reasons ……
  ➢ In which way/s …..

• Which part of your body do you think is most affected by the condition
  ➢ General appearance, mouth/ teeth, skin & hair or perspiring
  ➢ Order them according from least affected to most affected
  ➢ Which one affected you more in your early years, your teenage years and since then?
  ➢ How important are your teeth to you? Why?

4. Hypodontia and malformed teeth:

• As teeth were one of the main issues, was the reason functional or appearance /aesthetic?
  ➢ Satisfaction with treatment that received as a child /teenager /adult?
  ➢ Did it meet your needs (Function/eating)?
  ➢ Dentures experience (Number of sets, their impact on childhood and adolescence ages).
  ➢ How pleased are you with the appearance of your teeth now?
• Treatment using dentures or aesthetic restorations influence on quality of life (QoL).
  ➢ Did it impact your QoL? Social, Function/ eating (food types), Study/ Work, Self-esteem/ stress control

• Treatment or controlling of other ED related features on QoL improvement.
  Which one after teeth factor that helped on that? (Skin, Hair, Perspiring Control)

• Economically, was the condition causing extra life expenses for you?
  ➢ What sort of extra things that you pay for? And why
  ➢ Refund or government support for that?
  ➢ Indirect costs such as time off school, travel, effect on parents and other family

5. **Closing the Discussion**

• Planning any other non-dental treatment for your condition
• Do you have any recommendation/s for GDP or to general health care professionals?
• If you meet young person who have ED, what sort of advice you will give to herself/himself and their parents?
• As you are adult now, do you recommend anything that improve the quality or type of treatment that we provide in DDUH for those who have the same condition and younger than you?

6. **Conclusion**
  • Thank You for your time
  • You are welcome to contact members of the study team to ask questions later if you wish

END RECORDING
Appendix 10: Child Perception Questionnaire (8-10)

CHILD ORAL HEALTH QUESTIONNAIRE

8-10 years

Hello,

Thanks for helping us with our study!

We are doing this study to understand better things that may happen to children because of their teeth and mouth.

PLEASE REMEMBER:

• Don’t write your name on the questionnaire.
• This is not a test and there are no right or wrong answers.
• Answer as honestly as you can.
• Don’t talk to anyone about the questions when you are answering them.
• No one you know will see your answers.
• Read each question carefully and think about the things that have happened to you in the past 4 weeks.
• Before you answer, ask yourself: “Does this happen to me because of my teeth or mouth?”
• Put an ☒ in the box beside the answer that is best for you.

Community Dental Health Services Research Unit
Faculty of Dentistry, University of Toronto
124 Edward Street, Toronto ON, M5G 1G6

Supported by: The Hospital for Sick Children Foundation
FIRST, A FEW QUESTIONS ABOUT YOU

Today’s date: ______/______/______
DAY MONTH YEAR

1. Are you a boy or a girl?
   □ Boy
   □ Girl

2. When were you born? ______/______/______
   Age ______
   DAY MONTH YEAR

3. When you think about your teeth or mouth, would you say that they are:
   □ Very good
   □ Good
   □ O.K.
   □ Poor

4. How much do your teeth or mouth bother you in your everyday life?
   □ Not at all
   □ A little bit
   □ Some
   □ A lot
NOW A FEW QUESTIONS ABOUT YOUR TEETH AND MOUTH

How often have you had:

5. Pain in your teeth or mouth in the past 4 weeks?
   - □ Never
   - □ Once or twice
   - □ Sometimes
   - □ Often
   - □ Everyday or almost every day

6. Sore spots in your mouth in the past 4 weeks?
   - □ Never
   - □ Once or twice
   - □ Sometimes
   - □ Often
   - □ Everyday or almost every day

7. Pain in your teeth when you drink cold drinks or eat hot foods in the past 4 weeks?
   - □ Never
   - □ Once or twice
   - □ Sometimes
   - □ Often
   - □ Everyday or almost every day

8. Food stuck in your teeth in the past 4 weeks?
   - □ Never
   - □ Once or twice
   - □ Sometimes
   - □ Often
   - □ Everyday or almost every day

9. Bad breath in the past 4 weeks?
   - □ Never
   - □ Once or twice
   - □ Sometimes
   - □ Often
   - □ Everyday or almost every day
How often have you:

10. Needed longer time than others to eat your meal because of your teeth or mouth in the past 4 weeks?

☐ Never  ☐ Once or twice  ☐ Sometimes  ☐ Often  ☐ Everyday or almost every day

11. Had a hard time biting or chewing food like apples, corn on the cob or steak because of your teeth or mouth in the past 4 weeks?

☐ Never  ☐ Once or twice  ☐ Sometimes  ☐ Often  ☐ Everyday or almost every day

12. Had trouble eating foods you would like to eat because of your teeth or mouth in the past 4 weeks?

☐ Never  ☐ Once or twice  ☐ Sometimes  ☐ Often  ☐ Everyday or almost every day

13. Had trouble saying some words because of your teeth or mouth in the past 4 weeks?

☐ Never  ☐ Once or twice  ☐ Sometimes  ☐ Often  ☐ Everyday or almost every day

14. Had a problem sleeping at night because of your teeth or mouth in the past 4 weeks?

☐ Never  ☐ Once or twice  ☐ Sometimes  ☐ Often  ☐ Everyday or almost every day
SOME QUESTIONS ABOUT YOUR FEELINGS

How often have you:

15. Been upset because of your teeth or mouth in the past 4 weeks?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day

16. Felt frustrated because of your teeth or mouth in the past 4 weeks?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day

17. Been shy because of your teeth or mouth in the past 4 weeks?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day

18. Been concerned what other people think about your teeth or mouth in the past 4 weeks?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day

19. Worried that you are not as good-looking as others because of your teeth or mouth in the past 4 weeks?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day
QUESTIONS ABOUT YOUR SCHOOL

How often have you:

20. Missed school because of your teeth or mouth in the past 4 weeks?
   □ Never
   □ Once or twice
   □ Sometimes
   □ Often
   □ Everyday or almost every day

21. Had a hard time doing your homework because of your teeth or mouth in the past 4 weeks?
   □ Never
   □ Once or twice
   □ Sometimes
   □ Often
   □ Everyday or almost every day

22. Had a hard time paying attention in school because of your teeth or mouth in the past 4 weeks?
   □ Never
   □ Once or twice
   □ Sometimes
   □ Often
   □ Everyday or almost every day

23. Not wanted to speak or read out loud in class because of your teeth or mouth in the past 4 weeks?
   □ Never
   □ Once or twice
   □ Sometimes
   □ Often
   □ Everyday or almost every day
QUESTIONS ABOUT YOU BEING WITH OTHER PEOPLE

How often have you:

24. Tried not to smile or laugh when with other children because of your teeth or mouth in the past 4 weeks?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day

25. Not wanted to talk to other children because of your teeth or mouth in the past 4 weeks?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day

26. Not wanted to be with other children because of your teeth or mouth in the past 4 weeks?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day

27. Stayed away from activities like sports and clubs because of your teeth or mouth in the past 4 weeks?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day
28. Other children teased you or called you names because of your teeth or mouth in the past 4 weeks?

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day

29. Other children asked you questions about your teeth or mouth in the past 4 weeks?

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day
THERE, IT’S FINISHED!

One last thing! To see how good these questions are we need a group of children to answer questions again.

Would you like to help?
We would mail you the questions in the next 2 weeks.

YES

THANK YOU FOR YOUR HELP
Appendix 11: Child Perception Questionnaire (11-14)

CHILD ORAL HEALTH QUESTIONNAIRE

11-14 years

Hello,

Thanks for agreeing to help us with our study!

This study is being done so that there will be more understanding about problems children may have because of their teeth, mouth, lips and jaws. By answering the questions, you will help us learn more about young people’s experiences.

PLEASE REMEMBER:

• Don’t write your name on the questionnaire
• This is not a test and there are no right or wrong answers
• Answer as honestly as you can. Don’t talk to anyone about the questions when you are answering them. Your answers are private; no one you know will see them
• Read each question carefully and think about your experiences in the past 3 months when you answer
• Before you answer, ask yourself: “Does this happen to me because of problems with my teeth, lips, mouth or jaws?”
• Put an ☒ in the box for the answer that is best for you

Community Dental Health Services Research Unit
Faculty of Dentistry, University of Toronto
124 Edward Street, Toronto ON, M5G 1G6

Supported by: The Hospital for Sick Children Foundation
FIRST, A FEW QUESTIONS ABOUT YOU

1. Are you a boy or a girl?
   - Boy
   - Girl

2. When were you born? _____/_____/_____
   - DAY
   - MONTH
   - YEAR

3. Would you say the **health** of your teeth, lips, jaws and mouth is:
   - Excellent
   - Very good
   - Good
   - Fair
   - Poor

4. How much does the condition of your teeth, lips, jaws or mouth affect your **life overall**?
   - Not at all
   - Very little
   - Some
   - A lot
   - Very much
QUESTIONS ABOUT ORAL PROBLEMS

In the past 3 months, how often have you had:

5. Pain in your teeth, lips, jaws or mouth?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day

6. Bleeding gums?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day

7. Sores in your mouth?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day

8. Bad breath?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day

9. Food stuck in or between your teeth?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day
10. **Food stuck in the top of your mouth?**

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day

*For the next questions…*
*Has this happened because of your teeth, lips, jaws or mouth?*

<table>
<thead>
<tr>
<th>In the past 3 months, how often have you:</th>
</tr>
</thead>
</table>

11. **Breathed through your mouth?**

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day

12. **Taken longer than others to eat a meal?**

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day

13. **Had trouble sleeping?**

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day
<table>
<thead>
<tr>
<th>Question</th>
<th>Options</th>
</tr>
</thead>
<tbody>
<tr>
<td>In the past 3 months, because of your teeth, lips, mouth or jaws, how often has it been:</td>
<td></td>
</tr>
<tr>
<td>14. Difficult to bite or chew food like apples, corn on the cob or steak?</td>
<td>Never, Once or twice, Sometimes, Often, Everyday or almost every day</td>
</tr>
<tr>
<td>15. Difficult to open your mouth wide?</td>
<td>Never, Once or twice, Sometimes, Often, Everyday or almost every day</td>
</tr>
<tr>
<td>16. Difficult to say any words?</td>
<td>Never, Once or twice, Sometimes, Often, Everyday or almost every day</td>
</tr>
<tr>
<td>17. Difficult to eat foods you would like to eat?</td>
<td>Never, Once or twice, Sometimes, Often, Everyday or almost every day</td>
</tr>
<tr>
<td>18. Difficult to drink with a straw?</td>
<td>Never, Once or twice, Sometimes, Often, Everyday or almost every day</td>
</tr>
</tbody>
</table>
19. Difficult to drink or eat hot or cold foods?

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day

QUESTIONS ABOUT FEELINGS

Have you had the feeling because of your teeth, lips, jaws or mouth? If you felt this way for another reason, answer ‘Never’.

In the past 3 months, how often have you:

20. Felt irritable or frustrated?

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day

21. Felt unsure of yourself?

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day

22. Felt shy or embarrassed?

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day
In the past 3 months, because of your teeth, lips, mouth or jaws, how often have you:

23. Been concerned what other people think about your teeth, lips, mouth or jaws?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day

24. Worried that you are not as good-looking as others?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day

25. Been upset?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day

26. Felt nervous or afraid?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day

27. Worried that you are not as healthy as others?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day
28. Worried that you are different than other people?
- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day

QUESTIONS ABOUT SCHOOL

Have you had these experiences because of your teeth, lips, jaws or mouth? If it was for another reason, answer ‘Never’.

In the past 3 months, how often have you:

29. Missed school because of pain, appointments, or surgery?
- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day

30. Had a hard time paying attention in school?
- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day

31. Had difficulty doing your homework?
- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day

32. Not wanted to speak or read out loud in class?
- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day
QUESTIONs ABOUT YOUR SPARE-TIME ACTIVITIES & BEING WITH OTHER PEOPLE

*Have you had these experiences because of your teeth, lips, jaws or mouth? If it was for another reason, answer ‘Never’.*

In the past 3 months, how often have you:

33. Avoided taking part in activities like sports, clubs, drama, music, school trips?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day

34. Not wanted to talk to other children?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day

35. Avoided smiling or laughing when around other children?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day

36. Had difficulty playing a musical instrument such as a recorder, flute, clarinet, trumpet?
   - Never
   - Once or twice
   - Sometimes
   - Often
   - Everyday or almost every day
37. Not wanted to spend time with other children?

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day

38. Argued with other children or your family?

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day

In the past 3 months, because of your **teeth, lips, mouth or jaws**, how often have:

39. Other children teased you or called you names?

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day

40. Other children made you feel left out?

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day

41. Other children asked you questions about your teeth, lips, jaws or mouth?

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost every day
THERE, IT’S FINISHED!

Just one more thing. To test how good this questionnaire is at giving us the information we need, we would like a group of children to complete it again.

Would you be willing to help us by completing another copy of the questionnaire soon? We would mail it to you in the next 2 weeks.

YES ☐

THANK YOU FOR HELPING US
Appendix 12: Parent Perception Questionnaire (6-14)

COMMUNITY DENTAL HEALTH SERVICES RESEARCH UNIT
FACULTY OF DENTISTRY
UNIVERSITY OF TORONTO
124 Edward Street
Toronto, Ontario
M5G 1G6

CHILD ORAL HEALTH QUESTIONNAIRE
Parental report
6-14 years

SUPPORTED BY
THE HOSPITAL FOR SICK CHILDREN FOUNDATION
INSTRUCTIONS TO PARENTS

1. This questionnaire is about the effects of oral conditions on children’s well-being and everyday life, and the effects on their families. We are interested in any condition that involves teeth, lips, mouth or jaws. Please answer each question.

   - To answer the question please put an ☒ in the box by the response.

   - Please give the response that best describes your child’s experience. If the question does not apply to your child, please answer with “Never”.

     Example: How often has your child had a hard time paying attention in school?

     If your child has had a hard time paying attention in school because of problems with his/her teeth, lips, mouth or jaws, choose the appropriate response. If it has happened for other reasons, choose “Never”.

     ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐
     Never Once or twice Sometimes Often Everyday or almost everyday

     Please do not discuss the questions with your child, as we are interested only in the parents’ perspective in this questionnaire.
SECTION 1:  Child’s oral health and wellbeing

1. How would you rate the health of your child’s teeth, lips, jaws and mouth?
   - □ Excellent
   - □ Very good
   - □ Good
   - □ Fair
   - □ Poor

2. How much is your child’s overall wellbeing affected by the condition of his/her teeth, lips, jaws or mouth?
   - □ Not at all
   - □ Very little
   - □ Some
   - □ A lot
   - □ Very much

SECTION 2:  The following questions ask about symptoms and discomfort that children may experience due to the condition of their teeth, lips, mouth and jaws

During the last 3 months, how often has your child had:

3. Pain in the teeth, lips, jaws or mouth?
   - □ Never
   - □ Once or twice
   - □ Sometimes
   - □ Often
   - □ Everyday or almost everyday
   - □ Don’t know
4. **Bleeding gums?**

<table>
<thead>
<tr>
<th></th>
<th>Never</th>
<th>Once or twice</th>
<th>Sometimes</th>
<th>Often</th>
<th>Everyday or</th>
<th>Don’t</th>
<th>almost everyday</th>
</tr>
</thead>
</table>

5. **Sores in the mouth?**

<table>
<thead>
<tr>
<th></th>
<th>Never</th>
<th>Once or twice</th>
<th>Sometimes</th>
<th>Often</th>
<th>Everyday or</th>
<th>Don’t</th>
<th>almost everyday</th>
</tr>
</thead>
</table>

6. **Bad breath?**

<table>
<thead>
<tr>
<th></th>
<th>Never</th>
<th>Once or twice</th>
<th>Sometimes</th>
<th>Often</th>
<th>Everyday or</th>
<th>Don’t</th>
<th>almost everyday</th>
</tr>
</thead>
</table>

7. **Food stuck in the roof of the mouth?**

<table>
<thead>
<tr>
<th></th>
<th>Never</th>
<th>Once or twice</th>
<th>Sometimes</th>
<th>Often</th>
<th>Everyday or</th>
<th>Don’t</th>
<th>almost everyday</th>
</tr>
</thead>
</table>

8. **Food caught in or between the teeth?**

<table>
<thead>
<tr>
<th></th>
<th>Never</th>
<th>Once or twice</th>
<th>Sometimes</th>
<th>Often</th>
<th>Everyday or</th>
<th>Don’t</th>
<th>almost everyday</th>
</tr>
</thead>
</table>

9. **Difficulty biting or chewing foods such as fresh apple, corn on the cob or firm meat?**

<table>
<thead>
<tr>
<th></th>
<th>Never</th>
<th>Once or twice</th>
<th>Sometimes</th>
<th>Often</th>
<th>Everyday or</th>
<th>Don’t</th>
<th>almost everyday</th>
</tr>
</thead>
</table>
During the last 3 months, because of his/her teeth, lips, mouth, or jaws, how often has your child:

10. Breathed through the mouth?

❑ ❑ ❑ ❑ ❑ ❑ ❑ ❑

Never  Once or twice  Sometimes  Often  Everyday or Don’t almost everyday know

11. Had trouble sleeping?

❑ ❑ ❑ ❑ ❑ ❑ ❑ ❑

Never  Once or twice  Sometimes  Often  Everyday or Don’t almost everyday know

12. Had difficulty saying any words?

❑ ❑ ❑ ❑ ❑ ❑ ❑ ❑

Never  Once or twice  Sometimes  Often  Everyday or Don’t almost everyday know

13. Taken longer than others to eat a meal?

❑ ❑ ❑ ❑ ❑ ❑ ❑ ❑

Never  Once or twice  Sometimes  Often  Everyday or Don’t almost everyday know

14. Had difficulty drinking or eating hot or cold foods?

❑ ❑ ❑ ❑ ❑ ❑ ❑ ❑

Never  Once or twice  Sometimes  Often  Everyday or Don’t almost everyday know
15. Had difficulty eating foods he/she would like to eat?

Never ❑ Once or twice ❑ Sometimes ❑ Often ❑ Everyday or almost everyday ❑ Don’t know ❑

16. Had diet restricted to certain types of food (e.g. soft food)?

Never ❑ Once or twice ❑ Sometimes ❑ Often ❑ Everyday or almost everyday ❑ Don’t know ❑

SECTION 3: The following questions ask about the effects that the condition of children’s teeth, lips, mouth and jaws may have on their feelings and everyday activities.

During the last 3 months, because of his/her teeth, lips, mouth or jaws, how often has your child been:

17. Upset?

Never ❑ Once or twice ❑ Sometimes ❑ Often ❑ Everyday or almost everyday ❑ Don’t know ❑

18. Irritable or frustrated?

Never ❑ Once or twice ❑ Sometimes ❑ Often ❑ Everyday or almost everyday ❑ Don’t know ❑
19. **Anxious or fearful?**

Never  Once or twice  Sometimes  Often  Everyday or  Don’t know

During the last 3 months, because of his/her teeth, lips, mouth or jaws, how often has your child:

20. **Missed school (e.g. pain, appointments, surgery)?**

Never  Once or twice  Sometimes  Often  Everyday or  Don’t know

21. **Had a hard time paying attention in school?**

Never  Once or twice  Sometimes  Often  Everyday or  Don’t know

22. **Not wanted to speak or read out loud in class?**

Never  Once or twice  Sometimes  Often  Everyday or  Don’t know

23. **Not wanted to talk to other children?**

Never  Once or twice  Sometimes  Often  Everyday or  Don’t know
24. Avoided smiling or laughing when around other children?

Never  Once or twice  Sometimes  Often  Everyday or  Don’t know
almost everyday

_During the last 3 months, because of his/her teeth, lips, mouth or jaws, how often has your child:_

25. Worried that he/she is not as healthy as other people?

Never  Once or twice  Sometimes  Often  Everyday or  Don’t know
almost everyday

26. Worried that he/she is different than other people?

Never  Once or twice  Sometimes  Often  Everyday or  Don’t know
almost everyday

27. Worried that he/she is not as good-looking as other people?

Never  Once or twice  Sometimes  Often  Everyday or  Don’t know
almost everyday

28. Acted shy or embarrassed?

Never  Once or twice  Sometimes  Often  Everyday or  Don’t know
29. Been teased or called names by other children?

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost everyday
- Don’t know

30. Been left out by other children?

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost everyday
- Don’t know

31. Not wanted or been unable to spend time with other children?

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost everyday
- Don’t know

32. Not wanted or been unable to participate in activities such as sports, clubs, drama, music, school trips?

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost everyday
- Don’t know

33. Worried that he/she has fewer friends?

- Never
- Once or twice
- Sometimes
- Often
- Everyday or almost everyday
- Don’t know

During the last 3 months, how often has your child been:
15. Concerned what other people think about his/her teeth, lips, mouth or jaws?

Never  Once or twice  Sometimes  Often  Everyday or  Don’t know almost everyday

20. Asked questions by other children about his/her teeth, lips, mouth or jaws?

Never  Once or twice  Sometimes  Often  Everyday or  Don’t know


### SECTION 4: Effects on Parents and Other Family Members

During the last 3 months, because of your child’s teeth, lips, mouth or jaws, how often have you or another family member:

<p>| | | | | | | | |</p>
<table>
<thead>
<tr>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>36. Been upset?</td>
<td>Never</td>
<td>Once or twice</td>
<td>Sometimes</td>
<td>Often</td>
<td>Everyday or almost everyday</td>
<td>Don’t know</td>
<td></td>
</tr>
<tr>
<td>37. Had sleep disrupted?</td>
<td>Never</td>
<td>Once or twice</td>
<td>Sometimes</td>
<td>Often</td>
<td>Everyday or almost everyday</td>
<td>Don’t know</td>
<td></td>
</tr>
<tr>
<td>38. Felt guilty?</td>
<td>Never</td>
<td>Once or twice</td>
<td>Sometimes</td>
<td>Often</td>
<td>Everyday or almost everyday</td>
<td>Don’t know</td>
<td></td>
</tr>
<tr>
<td>39. Taken time off work (e.g. pain, appointments, surgery)?</td>
<td>Never</td>
<td>Once or twice</td>
<td>Sometimes</td>
<td>Often</td>
<td>Everyday or almost everyday</td>
<td>Don’t know</td>
<td></td>
</tr>
</tbody>
</table>
40. Had less time for yourself or the family?

<table>
<thead>
<tr>
<th>Choice</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>Never</td>
<td></td>
</tr>
<tr>
<td>Once or twice</td>
<td></td>
</tr>
<tr>
<td>Sometimes</td>
<td></td>
</tr>
<tr>
<td>Often</td>
<td></td>
</tr>
<tr>
<td>Everyday or</td>
<td></td>
</tr>
<tr>
<td>Don’t know</td>
<td></td>
</tr>
<tr>
<td>almost everyday</td>
<td></td>
</tr>
</tbody>
</table>

41. Worried that your child will have fewer life opportunities (e.g. for dating, getting married, having children, getting a job he/she will like)?

<table>
<thead>
<tr>
<th>Choice</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>Never</td>
<td></td>
</tr>
<tr>
<td>Once or twice</td>
<td></td>
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<tr>
<td>Sometimes</td>
<td></td>
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<tr>
<td>Often</td>
<td></td>
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<tr>
<td>Everyday or</td>
<td></td>
</tr>
<tr>
<td>Don’t know</td>
<td></td>
</tr>
<tr>
<td>almost everyday</td>
<td></td>
</tr>
</tbody>
</table>

42. Felt uncomfortable in public places (e.g. stores, restaurants) with your child?

<table>
<thead>
<tr>
<th>Choice</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>Never</td>
<td></td>
</tr>
<tr>
<td>Once or twice</td>
<td></td>
</tr>
<tr>
<td>Sometimes</td>
<td></td>
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<tr>
<td>Often</td>
<td></td>
</tr>
<tr>
<td>Everyday or</td>
<td></td>
</tr>
<tr>
<td>Don’t know</td>
<td></td>
</tr>
<tr>
<td>almost everyday</td>
<td></td>
</tr>
</tbody>
</table>

During the last 3 months, because of his/her teeth, lips, mouth, or jaws, how often has your child:

43. Been jealous of you or others in the family?

<table>
<thead>
<tr>
<th>Choice</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>Never</td>
<td></td>
</tr>
<tr>
<td>Once or twice</td>
<td></td>
</tr>
<tr>
<td>Sometimes</td>
<td></td>
</tr>
<tr>
<td>Often</td>
<td></td>
</tr>
<tr>
<td>Everyday or</td>
<td></td>
</tr>
<tr>
<td>Don’t know</td>
<td></td>
</tr>
<tr>
<td>almost everyday</td>
<td></td>
</tr>
</tbody>
</table>
44. Blamed you or another person in the family?

- [ ] Never
- [ ] Once or twice
- [ ] Sometimes
- [ ] Often
- [ ] Everyday or almost everyday

45. Argued with you or others in the family?

- [ ] Never
- [ ] Once or twice
- [ ] Sometimes
- [ ] Often
- [ ] Everyday or almost everyday

46. Required more attention from you or others in the family?

- [ ] Never
- [ ] Once or twice
- [ ] Sometimes
- [ ] Often
- [ ] Everyday or almost everyday

47. Interfered with family activities at home or elsewhere?

- [ ] Never
- [ ] Once or twice
- [ ] Sometimes
- [ ] Often
- [ ] Everyday or almost everyday

48. Caused disagreement or conflict in your family?

- [ ] Never
- [ ] Once or twice
- [ ] Sometimes
- [ ] Often
- [ ] Everyday or almost everyday

During the last 3 months, how often has the condition of your child’s teeth, lips, mouth or jaws:
49. Caused financial difficulties for your family?

☐ Never  ☐ Once or twice  ☐ Sometimes  ☐ Often  ☐ Everyday or almost everyday  ☐ Don’t know

SECTION 5: Child’s gender and age

a. Your child is:

☒ MALE
☒ FEMALE

30. Your child’s age is: ______ YEARS

Questionnaire completed by:

• MOTHER
• FATHER
• OTHER ______________

Date completed: ______ / ______ / ______

To test how good this questionnaire is at giving us the information we need, we would like a group of parents to complete it again.

Would you be willing to complete another copy of the questionnaire in the next 2 weeks?

☐ Yes

THANK YOU FOR YOUR PARTICIPATION