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**SURVEILLANCE STUDY ON CHILDREN AND ADOLESCENTS
WITH DOWN SYNDROME
(3 MONTHS TO 18.9 YEARS)
IN THE
EASTERN REGIONAL HEALTH AUTHORITY**

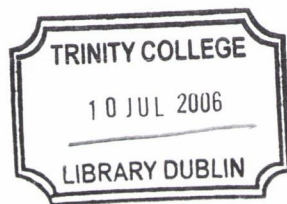
**THESIS SUBMITTED FOR THE DEGREE
DOCTOR OF PHILOSOPHY
BY RESEARCH**

By

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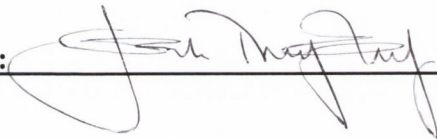


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DECLARATION

This thesis is the original work of the investigator with the exception of where acknowledged. It has not been submitted for any degree at any other university.

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DEDICATION

This thesis is dedicated to my husband Donal for his support and understanding, and
to my children Niamh Colin and Sonya who encouraged me all the way

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SUMMARY

Down syndrome is the most common cause of developmental disability in Ireland with a birth prevalence of 1 in 546 live births. Children with Down syndrome have a high incidence of associated treatable medical disorders. Studies have shown that early intervention carries a better medical outcome. A prerequisite for maximising medical care for those with Down syndrome is that information regarding the incidence and characteristics of their disorders together with agreed medical management guidelines are available to health professionals and policy makers.

In the absence of consensus protocols for the screening and management of children and adolescents with Down syndrome in Ireland, a study was undertaken on 394 children and adolescents with Down syndrome, aged 3 months to 18.9 years in the Eastern Regional Health Authority. The purpose was to: provide medical and developmental data on Irish children and adolescents with Down syndrome; outline existing screening and management practices; and provide an evidence base to develop best practice guidelines for assessment and management. It included medical assessment of growth and development, cardiovascular, thyroid status, vision, hearing, orthopaedic and cervical spine instability, speech and language. In addition, assessment of parental concerns and quality of life issues were explored by questionnaire.

A cross sectional study following national and international consultation was designed. Ethical approval for the study was granted by the Joint Ethics Committee of the Federated Dublin Voluntary Hospitals. The European Register of Congenital Anomalies & Twins (EUROCAT), which has monitored the birth prevalence of Down syndrome in Europe since 1980 acted as a source in identifying the number of children and adolescents with Down syndrome born between January 1981 and December 1997 in the Eastern Regional Health Authority. Down Syndrome Ireland, Medical Directors and Managers of the many Special Developmental Services, Paediatricians, and Area Medical Officers in the 10 community areas, following consultation, agreed to support this study and population identification was performed through these agencies.

Results showed that those with Down syndrome have an increased prevalence of medical problems and associated conditions. It was found that children with Down syndrome are shorter than the general population and have a smaller growth spurt at puberty. Their weight for height, and therefore body mass index (BMI), tends to be greater than that of the general population and increases with age. Considerable overweight and obesity was found in later childhood and adolescence. Obesity is associated with significant adverse health outcomes, is not inevitable in this group and requires early intervention with diet and child and family education. The average age of menarche in girls with Down syndrome was 12.6 years (range 9-15 years), which is one year earlier than the general Irish population.

Cardiac disease was diagnosed in 45% children with 29% having an atrioventricular septal defect. This study identified 4.1% children with hypothyroidism and 0.5% children with hyperthyroidism. Twenty-one percent of children were screened for thyroid dysfunction during the year of the study (mean age 6.9 years). However, a worrying finding was that 50% (age range 1 to 17 years) had not been screened since the newborn period. Undetected thyroid disease is associated with significant preventable secondary disability. Biochemical screening is necessary, however venepuncture may be traumatic in those with Down syndrome often causing long-term distress for children and their families. This study confirmed the feasibility of screening for hypothyroidism by the much less traumatic finger prick sampling of 305 children for Thyroid Stimulating Hormone (TSH) measurement using the national newborn screening card. Provision of this service for annual screening in Down syndrome patients would be most valuable in Ireland in order to promote early identification and treatment of thyroid dysfunction.

Vision disorders were reported in 50% of this cohort. The most common conditions included refractive errors, strabismus (squint), cataract and nystagmus. The mean age of first vision screening was at 2 years of age as reported by parents. These conditions, if undiagnosed with early intervention, may result in significant impairment of vision, reduced learning and poor quality of life.

Lifelong audiological surveillance is essential for people with Down syndrome because of the associated high incidence of hearing impairment, which increases with age. This study found over 80% of children had hearing abnormalities. These included abnormal outer ear findings on otoscopy (64%), middle ear abnormalities by tympanometry (58%) and inner ear abnormalities by Distortion Product Oto Acoustic Emissions (DPOAE) (11%). Even mild hearing impairment when associated with any intellectual disability can severely compromise speech development and learning. This may result in social isolation, behavioural difficulties and reduced opportunity in the working world leading to depression and reduced quality of life. Hearing assessment in children with intellectual impairment is difficult and time consuming. This study showed that hearing screening by otoscopy, tympanometry and DPOAE is rapid and non traumatic in children with intellectual disability, and in addition DPOAE compares well with the gold standard Pure Tone Audiometry, as a screening tool, but requires further studies to identify specificity and sensitivity. These objective tests are easy to perform, require no patient feedback and can be undertaken from infancy. A high proportion 94.5% of children had speech impairment.

The study showed that 71% of children had orthopaedic problems, the most common being pes planus (flat feet) in 63%, hip joint instability in 3%, knee and ankle joint problems in 3.5%, Perthes disease in 0.3% and atlanto axial joint instability was detected in 1% of children.

In addition a number of other medical problems were identified including gastro-intestinal disorders such as constipation, diarrhoea, coeliac disease and Hirschsprung disease. There was also an increased incidence of Type I Diabetes and epilepsy, which require identification and ongoing medical management.

Rearing a child with Down syndrome is not easy and a major challenge is to maximise Quality of Life (QOL). Assessment of QOL is important in determining whether additional multidisciplinary support is necessary. We found that QOL for parents caring for a child with Down syndrome was greatly disrupted due to the extra parental and medical care required, and particularly so when the child had more than one medical problem. Similarly, the majority of parents reported burden in disruption to their family routines. Very poor quality of life was reported when hearing and vision problems were associated. Early medical intervention, mainstream education, parent information, long term health needs, community acceptance and better integration were parent perceived areas in most need of urgent improvement.

This study shows a high incidence of associated medical disorders in this population and medical management guidelines for children and adolescents with Down syndrome in Ireland were developed with the support of the Down Syndrome Medical Interest Group UK. Many health care professionals with expertise in the management of children and adolescents with Down syndrome in Ireland have contributed to the Irish guidelines. These guidelines for Irish children have been approved by the Irish College of General Practitioners, The Faculty of Paediatrics of the Royal College of Physicians of Ireland and the Faculty of Public Health Medicine of the Royal College of Physicians of Ireland. Implementation of the guidelines and outcome audit is important and desirable.

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CHAPTER ONE

INTRODUCTION

CHAPTER 1 INTRODUCTION

Down syndrome is a chromosome disorder and one of the most common causes of developmental disability (Gath 1994, Marder and Dennis 2001). In Ireland Down syndrome accounts for a quarter of all persons aged over 14 years with moderate, severe and profound mental handicap (Mulcahy 1983). Children and adolescents with Down syndrome have a high incidence of associated treatable medical disorders for which early intervention is vital and is associated with a better outcome for the child and family.

In the absence of agreed protocols for the screening and management of children and adolescents with Down syndrome in Ireland this study of 394 children and adolescents (217 boys and 177 girls) in the Eastern Regional Health Authority (ERHA) was undertaken in order to assess their medical needs

1.1 Background

The first large-scale institution was built in London in 1860 for severely handicapped people. Dr John Langdon Down was the chief physician and in 1866 he was the first person to accurately describe the syndrome, which today bears his name. He named the syndrome “mongolism” because of the Mongoloid appearance of their eyes. In addition several ancient paintings and carvings depicting children with the typical facial characteristics of Down syndrome dating back to the 16th and 17th centuries have been identified, indicating that this condition has been

recognised since at least the middle ages (Selikowitz 1997a). It was not until 1959 that Professor Jerome Lejeune in Paris identified the extra chromosome as being the cause of Down syndrome and with this breakthrough came the possibility of diagnosing a whole range of genetic disorders. This disorder is now well understood at the chromosomal level, however, the factors responsible for generating chromosome abnormalities are largely unknown.

Ireland has one of the highest birth rates (1/546 live births) of children with Down syndrome in the developed world, the majority of whom are reared at home with the family giving an opportunity to study many of them in these natural conditions (home environment). Comparable birth incidences (1 in 700 live births) have been found in most populations and these figures compare with approximately 1 in 800 to 1 in 1,000 live births in developed countries Norway 9.8-11.3/10,000; South Africa 7.62/10,000; Australia 10.5-13.8/10,000 and New Zealand 8.8-9.6/10,000 (Baird & Sadovnick 1989, International Centre for Birth Defects Roma Annual Report 2001). Although some year-to-year fluctuation in birth rates of Down syndrome had been noted, the birth prevalence of this condition has been fairly stable over the last two decades. It is well documented that the incidence of Down syndrome increases with the increasing age of the mother (Selikowitz 1997b, Johnson 1996).

Apart from Malta, Ireland is the only European country in which ante-natal screening for Down syndrome and induced terminations are not carried out (Johnson 1996). A survey from 1981-1991 by EUROCAT (European Register of

Congenital Anomalies and Twins) in the counties Dublin, Kildare, Wicklow and Galway, which included more than one-third of the population of the state, showed a crude prevalence rate of live Down syndrome babies born of 18.3 per 10,000 live births (1 in 546 live births) (Johnson 1996). Children born with Down syndrome represented 79% of births with chromosomal abnormalities during that period

1.2 Life Expectancy

Life expectancy for persons with Down syndrome has increased from 9 years in 1929 (Penrose 1949) to 15 years by 1947 (Penrose 1949) and 18 years by 1961 (Collmann & Stoller 1963); to 60 in 1979 (Mulcahy 1979) and 68 years by 1989 (Baird 1989). A number of studies demonstrate that over three decades the percentage survival to age 10 years rose from 36.8% in 1958, to 64.5% in 1970, to 86% in 1981 and to 90% in 1997 (Carter 1958, Fabia 1970, Masaki 1981, Hayes 1997). The dramatic improvement over the past 50 years is due primarily to increased survival in the first year of life. However, the life expectancy of a person with Down syndrome at any given age is still lower than the life expectancy for the background population (Dupont 1986)

The biggest factor influencing survival is the presence or absence of congenital heart disease (Hayes 1993). Since deaths from infections have declined in children with Down syndrome, deaths from heart disease have dominated, accounting for 65% of deaths in one study and 47.3% in another study (Hayes 1997, McGrother and Marshall 1990, Masaki 1981). Nevertheless the development of surgical techniques, and of greater willingness to use them where people with Down

syndrome are concerned, have resulted in an improvement in survival rates for those with congenital heart disease (McGrother and Marshall 1990). Although the sex ratio in Down syndrome is in favour of males no sex difference in longevity has been found (Baird 1988, Fryers 1984). The availability of accurate information on incidence and clinical outcome

is an important requirement for clinical management and health care services provision. Unfortunately, there is no national register of Down syndrome births in Ireland.

1.3 Chromosomal Karyotype in Down's Syndrome

1.3.1 Trisomy 21

Down syndrome is just one example of a birth-related disorder caused by a chromosome abnormality. Chromosomes are present in all cells of the body that carry the messages or genes, which guide the development of a foetus. Chromosomes are normally grouped together in 23 pairs i.e. 46 in total and these comprise of 22 pairs of autosomes and 1 pair of sex chromosomes. The diagnosis is made by taking a blood sample following which by special processes including leukocyte culture, arresting cells at metaphase, a karyotype is determined (Pappas 1994). A baby with Down syndrome usually has an additional chromosome 21, resulting in 47 chromosomes in every cell. This pattern is known as Trisomy 21

(47XX, 21+ or 47XY, 21+) and accounts for the majority (90-95%) of children with Down syndrome. Although Down syndrome is considered a chromosomal disorder, it is usually not inherited. In the majority 90-95% of children with Down syndrome the extra chromosome 21 is separate and unattached. The additional chromosome is the result of an error in cell division of the egg or the sperm. The chance of recurrence in further children is 1% in mothers under 35 years.

1.3.2 Translocation Trisomy 21

Approximately 3%-5% of cases will present with the extra chromosome 21 attached to another chromosome, which is called translocation trisomy 21. In this instance the additional chromosome is attached to another chromosome, most often chromosome numbers 13, 14, 15, 21 or 22 and the total chromosome count is 46. The most frequent seen is the 14, 21 translocation (Robertsonian). In this form the carrier has one chromosome 14, one chromosome 21 and one fused chromosome 14/21 giving a total of 46 chromosomes e.g. 46,XY, -14, +t (14q21q). In 50% of babies with translocation Down syndrome one parent is found to be a carrier. In this situation the chance of a recurrent birth with Down syndrome is 1 in 8 if the mother is the carrier and 1 in 40 if the father is the carrier. Further studies of the family should be undertaken because if one parent is a carrier, other members may also be carriers (Selikowitz 1997).

1.3.3 Mosaic trisomy 21

The remaining 1%- 2% of cases have a mosaic pattern. This occurs when some cells of an individual have the normal 46 chromosomes and whilst some have 47, due to an additional chromosome 21 e.g. 46,XY, /47,XY, +21. The chromosomal pattern of an individual with Down syndrome is not a predictor of mental functioning or physical characteristics, however, children with mosaic trisomy 21 may range from apparently normal individuals to those with typical Down syndrome. It is very difficult to predict the likelihood of recurrence since it will ultimately depend on the level of mosaicism. Chromosome testing may be offered in further pregnancies.

1.4 Clinical Features

Diagnosis of Down syndrome is usually made at birth on the basis of physical examination and is confirmed by chromosome analysis in order to detail the karyotype. Approximately 120 clinical features have been described in Down syndrome however many children may have no more than six or seven recognisable features at birth. The child with Down syndrome usually has a round face in frontal view and a flat profile from the side (brachycephaly) associated with a slightly flattened back of head. The neck is short and thick with excess skin at the back of the neck in 80% of children.

The child's eyes usually slant upwards and outwards appearing widely set across a short nose with a low bridge. Sometimes there is an extra fold of skin at the inner corner of the eye. This "epicanthic fold" tends to disappear in the teen years. Brushfield's spots (whitish spots scattered around the periphery of the iris) are found more frequently in Down syndrome than in the general population. Clouding of the lens of the eye (cataract) and/or glaucoma (raised pressure within the eye) may occur in infancy (Traboulsi 1988). The ears lobes are usually small and may have some malformation. The ear canals may be narrow and in some cases so narrow that visualization of the eardrum is impossible. Small specks of wax (cerumen) may block the canal and the air conduction pathways of hearing. The tongue appears large and may protrude because the lower jaw and palate may be relatively small. One study found that the palatal length and width were more than 2 standard deviations below the normal mean in 95.5% people with Down syndrome (Shapiro 1975).

Muscle tone is generally low (hypotonia) in up to 80% of children with Down syndrome. Therefore the baby may feel floppy which makes them difficult to handle but this feature often disappears with time. The head and neck may be particularly wobbly and need support at first. Joint laxity may also occur. The hands are often broad and stubby. Frequently the little finger is curved inwardly and there is often a single palmar crease. The feet are generally small and there may be a wide gap and deep crease between the first and second toes. Some toes may be "webbed" with skin.

There is an increased risk of malformations of the gastrointestinal tract (12%). They include tracheo-oesophageal fistula, pyloric stenosis, duodenal atresia, imperforate anus and Hirschsprung's disease (where some of the nerve cells (mesenteric plexus) that are normally present in the wall of the intestine do not form properly during fetal development). These may be associated with obstruction of the oesophagus, duodenum or further along the tract in the small or large bowel. Almost all these malformations can be corrected surgically. The genital organs are usually smaller than in other babies.

1.5 Medical Problems

1.5.1 Growth

Short stature is a recognised characteristic of those with Down syndrome. This is also associated with a reduced pubertal growth spurt the aetiology of which is not fully understood (Cronk 1988, McCoy 1992). Research has shown that in some children associated conditions, such as hypothyroidism, sleep related upper airway obstruction, coeliac disease or cardiac disease may contribute to this short stature and must be excluded promptly in order to optimise the already compromised growth potential (Karlsson 1998, Sharav 1988, Stebbens 1991, George 1996, Jansson 1995). Those with DS have an increased incidence of overweight and obesity (Chumlea 1981, Prasher 1995a). Weight gain in DS may reflect undetected thyroid disease. The onset of puberty requires anticipation with parental and child education and support. The timing of puberty varies in this population (Benda 1960, Tanner

1973, Takano 1999), requiring anticipation, parental and child education including support.

Specific growth charts for assessment of height and weight for children with Down syndrome reared at home were developed by Cronk and Palmer in America and Cremers in The Netherlands (Cronk 1988, Palmer 1992, Cremers 1996). These centile charts do not include the first 3 months of life.

1.5.2 Cardiac

Cardiac abnormalities are strongly associated with Down syndrome and these vary from a slight murmur to a severe abnormality previous studies have found had 40-50% of infants have congenital heart disease (Tubman 1991, Eurocat Report 6 1995). Cardiac anomalies include atrioventricular canal defect, atrial septal defect and ventricular septal defect, patent ductus arteriosus and Fallot's tetralogy. The most common defect is complete atrioventricular septal defects (AVSD) which occurs in 30 to 40% of those with congenital heart disease (Amark 1999). The majority of congenital heart lesions in Down syndrome can now be successfully treated either medically or surgically. The cardiac condition may not be obvious at birth but develop later. Early detection is essential as delay in treatment can result in irreversible pulmonary vascular disease (Yamaki 1993). Cardiac disease should be actively excluded in all infants with DS at or before their 6-week check (Frontera-Izquierdo 1990, Cullen 1990, Taylor 1990, Wren 1999, Committee Report 1995). Mitral valve prolapse may present in early adulthood (Goldhaber 1987). Down

syndrome is associated with an increased incidence of periodontal disease and as with all children with cardiac anomalies there may be a risk of infective endocarditis and prophylactic antibiotics may be required (Committee Report 1995).

1.5.3 Thyroid

It is well recognised that thyroid dysfunction occurs more frequently in Down syndrome than in the general population and that the prevalence increases with age (Fort 1984, Loudon 1985, Sare 1978, Pueschel 1985, Selikowitz 1993, Karlsson 1998, Fisher 1979, Alm 1984, Baxter 1975, Pueschel 1991, Rooney 1997, Prasher 1995b, Mani 1988). Cross sectional studies of thyroid function in children with Down syndrome have shown an increased prevalence in both congenital hypothyroidism and acquired thyroid dysfunction.(Fort 1984, Prasher 1995, Zori 1990). In the latter group Hashimoto's thyroiditis is the most common disorder and consists of both compensated {raised plasma Thyroid Stimulating Hormone (TSH) together with a normal plasma thyroxine (free T4)}, and uncompensated (raised plasma TSH with a reduced free T4) hypothyroidism (Loudon 1985, Pueschel 1985, Zori 1990, Stewart 1994). When Type I diabetes is associated with Down syndrome there is a higher risk of developing hypothyroidism (Daniels 1968, Griffin 1997). The prevalence of hyperthyroidism is also increased in Down syndrome when compared to the general population (Sare 1978, Takahashi 1979, Vanderpump 1995).

The wide-ranging effects of thyroid hormone are demonstrated by the consequences of thyroid hormone deficiency and excess. The thyroid hormones affect the body's basal metabolic rate; growth; intermediary metabolism; neurological development, cardiovascular, skeletal, gastrointestinal and reproductive system activities. Undiagnosed and untreated thyroid disorders constitute a significant cause of preventable secondary handicap including the risk of permanent brain damage, poor height attainment and excessive weight gain. In most patients, major effects can be prevented or reversed by treatment with thyroxine. It is therefore vitally important that screening be carried out at appropriate intervals in order to diagnose and commence treatment as early as possible and thus prevent unnecessary secondary handicap (Glorieux 1983). The clinical features of children and adolescents with Down syndrome which include short stature, reduced activity and alertness, tendency to overweight, intellectual disability, hoarse voice and susceptibility can subtly mask the signs of hypothyroidism and may be attributed to the Down syndrome itself (Rooney 1997, Quinn 1980). Diagnosis on clinical grounds is therefore unreliable and biochemical screening is essential (Prasher 1995, Mani 1988, Lobo 1980).

Venepuncture in children and adolescents with Down syndrome can sometimes be traumatic and difficult to perform and may cause long-term emotional distress. Preliminary evaluation suggests that TSH assessment using the newborn screening (Guthrie) card can be performed in non-hospital settings and may prove to be a practical and feasible alternative to venepuncture (Noble 2000). Although capillary heel prick screening is carried out in Ireland in neonates, TSH screening by

fingerprick has not been undertaken. It is a simple, quick and non frightening way to take a blood sample and could be more acceptable in this population than venous sampling.

1.5.4 Hearing

Impaired hearing is one of the most prevalent disabilities in children and adolescents with Down syndrome and it is challenging to identify and manage (Cunningham 1981, Dahle 1986, Davies 1998, Evenhuis 1996, Roizen 1997). Previous studies show that 60% to 85% of children with Down syndrome have mild to moderate conductive hearing loss. There is an increased prevalence of conductive, sensorineural and mixed hearing loss in adolescents and adults, which is progressive in nature (Evenhuis 1992 Whiteman 1986). Children with Down syndrome are much less able than those of normal intelligence to use various strategies to compensate for hearing loss (Cunningham 1981).

The auditory neural pathway continues to develop by external stimulations after birth and the most sensitive time for this final development is in the first year of life (Maroudias 1994). Children with hearing loss of more than 25dB who receive intervention before 6 months of age develop near-normal language skills, whereas those whose treatment is begun after 6 months of age have significantly language-delayed (Yoshinaga-Itano 1998). Early language deficit can lead to poor speech, learning difficulties and associated behavioural and emotional problems (Wetherby 1989, Sigman 1999, Yoshinaga-Itano 2003).

Children with DS are particularly predisposed to accumulation of wax (due to narrow ear canals) and persistent otitis media with effusion (Schwartz 1978). Early detection and removal of wax is important to maximise hearing (Hall 1996). Where any degree of hearing abnormality is detected reasonably early access to specialist hearing support services is vital (National Deaf Children's Society 1994).

Hearing screening has traditionally been assessed by the distraction test (9–12 month developmental visit). This screening is “variable in efficiency and effectiveness, depending upon the personnel and training of the practitioners involved, the conditions under which testing is conducted and the ability of the child to respond at an appropriately early age” (Pitt T, 1999). It is time consuming, and may be difficult in children with Down's syndrome (McCormick 1997). A developmental maturity level of around seven months is optimum for assessment as prior to this age, sitting balance, head control, and sound localization ability are imperfect. Beyond ten months of age, the development of object permanence and increasing sociability make the test more difficult. Results may be unreliable if the child cannot co-operate and respond at an appropriately early age because of associated intellectual disability and this can lead to long delays in diagnosis (Hall 1996).

Pure Tone Audiometry (PTA) is the audiological assessment that has always been viewed as the “gold standard” for documenting hearing loss (Gorga 1995). The magnitude of hearing ability and hearing loss is quantified by the pure-tone

audiogram, ranging from normal hearing to mild / severe hearing loss (Lee 1999). Behavioural assessments require voluntary response from the child and depend on the child's ability to provide appropriate voluntary responses, however many children may be incapable of providing these responses, either to age or developmental level (Gorga 1995).

Auditory Brain Stem (ABR) screening is an electric response from various levels along the auditory pathway. Four standard electrodes are placed on the scalp (top of the head and behind the ear) and minute electrical changes occurring in response to an auditory signal are recorded (Mason 1988). In children with Down syndrome this test is usually carried out under anaesthetic, however, they have unique ABR findings, in particular short central conduction times regardless of hearing status, which may make identification of hearing loss particularly difficult (Widen 1987, Diaz 1995, Seidl 1997)).

Following the National Institute of Health (NIH) report in 1993 recommending universal newborn hearing screening many studies of hearing screening methods have been undertaken (NIH Consensus Statement 1993). David Kemp, the English biophysicist discovered otoacoustic emissions (OAE's) in 1978 (Kemp 1990). These are sounds that originate inside the cochlea and are generated only when the organ of Corti, which is located in the inner ear, is normal. OAE's are sounds that come from the ear naturally and they can also occur as a result of an external stimulus. The absence of an OAE response to stimulation indicates a hearing loss

of 30dB or more. It will not define the exact auditory threshold but indicates the need for further diagnostic assessment (Kemp 1990, Probst 1991).

Two types of responses can be assessed namely Transient Evoked Otoacoustic Emissions (TEOAE) and Distortion Product Oto Acoustic Emissions (DPOAE). TEOAE responses are generated when an external click or tone burst is introduced to the ear. DPOAE responses are produced when the ear is stimulated by two primary tones presented simultaneously. Screening the function of the inner ear by TEOAE allows assessment across the frequencies 1-4 KHz only whilst DPOAE tests the wider range of 1-8 KHz. This is important in those with Down syndrome, because of their increased incidence of high frequency sensorineural hearing loss between 4 and 8 KHz, which would be missed by the TEOAE (Lopponen 1992, Marcell 1992).

Screening by OAE's is now used for identifying hearing function in neonates in the United States, Canada, parts of Europe and Australia and appears promising for use in people with intellectual disability (NIH Consensus Statement 1993, Gorga 1996, European Consensus on Neonatal Hearing Screening 1998).

The DPOAE test is an objective, reliable, non-invasive method of hearing screening of inner ear function which requires no patient feedback and could thus be very beneficial in children with Down syndrome in early childhood and adolescence (Haggard 1990, Evenhuis 1996). Early identification and intervention with appropriate programmes of remediation are essential to minimize the period of language deprivation and give a better prognosis for language development, social

interactions and successful participation in society (Pappas 1994, Yoshinaga-Itano 1998, Wetherby 1989, American Academy of Audiology 1988, Bess 1993, McCormick 1997).

1.5.5 Orthopaedics

Relaxed ligaments which hold the joints in place, reduced muscle tone and abnormal skeletal development may lead to a number of orthopaedic problems in children with Down syndrome (Daimond 1981, Jacobson 2000). The areas of particular concern include the neck, elbows, knees, hips, spinal column and feet.

The hip and knee joints are the most common areas requiring treatment (Reis 1994, Shaw 1992, Mendez 1988). Those with Down syndrome may show abnormal movement at the knee joint and in approximately 20% the patella dislocates (slips out of position) due to increased laxity of the ligaments (Goldberg 1987). Hip disorders such as abnormal development and subluxation/dislocation occur in up to 10% of those with Down syndrome (Shaw 1992, Lawhorn 1990). Scoliosis develops in approximately 50% (Jacobsen 2000, Diamond 1981). It occurs more frequently in institutionalised patients than in those living at home in the community due to less physical activity (Lawhorn 1990). Pes planus (flat feet) due to ligament laxity and poor muscle tone is common and clubfoot may also occur (Lawhorn 1990).

The orthopaedic area that causes most concern in those with Down syndrome is the possibility of cervical spine instability at the atlantoaxial joint. This joint is used when the head rotates, flexes or extends. An inappropriately relaxed transverse ligament of the first cervical vertebrae (the atlas) or poor development of both the first and second cervical vertebrae (the atlas and the axis), may predispose to dislocation/subluxation of the atlantoaxial joint. This instability may occur with neck movement when the atlas slides forward over the axis i.e. subluxation of the atlantoaxial joint. Studies have reported that 1 to 2 % of children with atlantoaxial dislocation/subluxation present with symptoms whilst 10 to 15% have no symptoms (Jacobsen 2000, Davidson 1988, Pueschel 1984,).

The spinal cord carrying all the nerves from the brain to the entire body passes very close to the atlantoaxial joint. There is a small risk of neurological problems caused by cervical spine instability in those with Down syndrome (Davidson 1988, Pueschel 1987a,). This concern has led to restrictions in various contact sporting activities and insistence on x-ray screening prior to admittance to special Olympic sports. There is currently no screening procedure, which can predict those at risk. Cervical spine x-rays of the atlantoaxial joint have no predictive value for subsequent acute dislocation and or subluxation of the joint (Selby 1991, Cremers 1993, American Academy of Paediatrics Committee on Genetics 1994, Department of Health 1995, Morton 1995).

1.5.6 Vision

There is a high incidence of eye disorders among people with Down's syndrome (Roizen 1994). Refractive errors of nearsightedness and far sightedness may present at an early age (Woodhouse 1993, Berk 1996, Hestnes 1991, Caputo 1989). Clouding of the lens (cataract), crossing (strabismus) and glaucoma (raised pressure within the eye) may occur alone or in combination in infancy (Caputo 1989, Traboulsi 1988). Keratoconus (a non inflammatory protrusion of the central part of the cornea) may occur at any age but is more common in adolescents and young adults (Caputo 1989, Cullen 1963, Shapiro 1999). As the child increases in age high myopia (near sightedness) may occur (Catalano 1990). There is also an increased incidence of naso-lacrimal duct obstruction (Hayes 1993). Early detection of these conditions is important, to allow early intervention as untreated eye problems may result in significant impairment.

1.5.7 Speech and Language

Communication skills are important and contribute to inclusion and integration. Communication includes not only speech, but also facial expressions, smiles, gestures, and alternative systems such as sign language and computer-based systems. The most sophisticated of human cognitive skills is development of language. Those with Down syndrome display many problems in language development and in particular, the development of speech (Launonen 1996, Bray 1988, Pueschel 1987b). While there is considerable individual variation in language development in children with Down syndrome the reasons for these problems is not yet known and cannot be explained by intellectual impairment

alone (Dykens 1994). However, this may be caused by abnormalities of early communication and auditory processing or impaired hearing. Recent studies of early language development have indicated the importance of the role of pre-speech communication with the development of spoken language (Launonen 1997, Mundy 1995). Early intervention services are needed to meet the developmental needs of an infant or toddler with a disability in any one or more of the following areas: physical, cognitive, communication, social or emotional and adaptive development. Communication skills are vital for education, integration, work acquisition, reduction in depression and quality of life.

1.6 Quality of Life

Caring for a child with special needs presents demands that may be difficult in different ways for parents and siblings and may adversely affect the family quality of life. In recent years literature has emerged on factors relating to stress and family functioning. Previous studies have often focused on identifying poor outcome for families (Byrne et al 1988). Other studies have emphasized the importance of resources and coping factors which may moderate or mediate the effects of stress (Donovan 1988, Friedrich et al 1985).

Within the field of special needs much focus has been placed on the characteristics of the person with special needs as the major stressors. However, the characteristics which are most likely to put families at risk are ill defined. The relationships between interrelated risk factors, such as care and supervisory demands, unexpected

physical care demands, marital status and the need for additional social support, all of which relate to stress, are likely to be interconnected (Barnett 1995, Sloper 1991). Adjusting to life with a child with a learning disability is often a long process marked by times of crisis, when extra professional intervention may be necessary (Maxwell 1993). Quality of life (QOL), together with early intervention of medical problems, is an important consideration for the best possible family centred management of children and adolescents with Down syndrome. QOL measures are also desirable for identifying areas of stress in order to allocate resources appropriately.

Aims of the Study

This study was conducted in order to assess the medical needs of children and adolescents with Down syndrome. The areas studied include growth and pubertal development, cardiovascular assessment, thyroid function and assessment of vision, hearing, orthopaedic and cervical spine instability and speech and language status. In addition, assessment of parental concerns and quality of life issues were explored by questionnaire.

The main aims of the study were to:

1. Provide medical and developmental data on Irish children and adolescents with Down syndrome.
2. Outline existing screening and management and provide an evidence base to develop best practice guidelines for assessment and management.

In addition the feasibility of:-

- screening for hypothyroidism by fingerprick sampling using the national newborn screening card was undertaken, and
- screening hearing impairment by otoscopy, tympanometry, distortions product oto acoustic emissions (DPOAE).
- Comparing reliability of DPOAE Screening with Pure tone audiometry (PTA)

CHAPTER TWO

METHODS

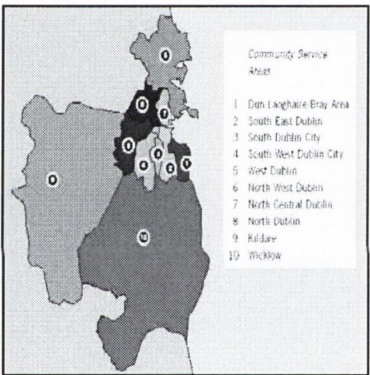
CHAPTER 2 METHODS

The plan and proposal for screening children and adolescents with Down syndrome (DS) in the Eastern Health Board Area (now the Eastern Regional Health Authority ERHA) was formulated following in-depth discussions and collaboration with many healthcare professionals both in Ireland and internationally together with extensive literature reviews. Ethical approval was obtained from the Joint Ethics Committee of the Federated Dublin Voluntary Hospitals.

2.1 Study Area

The study area included counties Dublin, Wicklow and Kildare, with a representative population of 1.3 million including a childhood population of 412,062 (31.7%; less than 19 years of age) 1996 Census. The three counties are divided into 10 Community Care Areas (Tables 2.1 – page 280).

Figure No 2.1 Study Area



2.2 Population Sampling

All children and adolescents with Down syndrome born between January 1981 and December 1997 in the Eastern Regional Health Authority were identified, using the database compiled by the European Register of Congenital Anomalies & Twins (EUROCAT), which has monitored the birth prevalence of Down syndrome in Ireland and across Europe since 1980. EUROCAT maximise case identification by exploring all possible avenues of patient presentation to the maternity hospitals in the catchment area studied. However, the accuracy of any incidence rates generated from this data

would depend on the degree of completeness of case identification. While it is believed that this methodology should capture the vast majority of new cases of chromosomal anomalies and twins in the population in this time period, the potential remains that incident cases could be missed. Derval Igoe (1996) undertook a study to determine the completeness of ascertainment and estimation of the true number of incident cases in the population studied by EUROCAT using the capture-recapture method. EUROCAT had a very favourable outcome was a 93% capture-recapture result.

EUROCAT criteria for case identification stipulated that the mothers be resident in the counties monitored. In the period studied 679 children were born with Down syndrome within the area studied and registered with EUROCAT. Of these 21 were stillborn and a further 91 died. In the absence of a live register the names of these children were not available to us from EUROCAT due to ethical reasons. The names of 40% of children were obtained from the Down Syndrome Association of Ireland register. Meetings with the Medical Directors and Managers of the Special Developmental Services and the Area Medical Officers from the 10 Community Areas, within the Eastern Regional Health Authority, were undertaken in order to identify the names of the remaining sixty percent of children with Down syndrome. They very willingly assisted us in identifying and contacting parent(s)/guardian(s) of children with Down syndrome attending their services. Seven parent(s)/guardian(s) contacted us themselves on hearing of the study by word of mouth. Thirteen additional children were included in the study at the request of the services (3 children born in 1997; 3 born in 1980; and 7 born 1998). Five hundred and seventy one (571) families were contacted by letter through their services and 415 parents responded to

our letters. Three hundred and ninety-four (394) parents agreed for their child to participate in the study.

2.3 Logistics of study

An extensive questionnaire was developed in consultation with healthcare specialists with expertise in the medical management of children and adolescents with Down syndrome both in Ireland and abroad. This was devised to elicit key clinical and demographic data. Letters to parent(s)/guardian(s) explaining the purpose of the study together with consent forms were constructed. (Appendix 1 – 3)

Auxology training was obtained in the Department of Paediatric Diabetes Growth and Endocrine at the National Children's Hospital by myself (JM) Research Nurse Specialist. Hearing screening training using tympanometry and distortion product otoacoustic emissions (DPOAEs) were undertaken at the Department of Audiology by myself (JM), at the National Children's Hospital. Consultation with the National Newborn Screening Centre in the Children's University Hospital, Temple Street was undertaken to establish the criteria for blood sampling, delivery, analysis and reporting. The questionnaire was pre-tested in a pilot study in December 1997 and January 1998. Final changes to the questionnaire were completed in early 1998.

Information posters relating to the study were designed and displayed in the Special Services Centres and were also included in the Down Syndrome Ireland monthly magazine. All identified children with Down syndrome were invited to participate in the study through their parent(s)/guardian(s). Letters were sent directly to parent(s)/guardian(s) from the special services inviting them to participate. Those

who did not respond to the invitation were crosschecked with their Special Services Centres to confirm that they were alive and resident in the community. Reminders were sent out with stamped addressed envelopes and in addition, fresh posters, giving information about the study and how to make contact with us, were placed in each of the services in a prominent place. Following this the number of parents responding increased to 415.

Families who consented were contacted directly by phone or letter. Individual appointments were arranged to suit parent(s)/guardian(s) and child at a choice of venues including nine Special Services Centres, one special school and the National Children's Hospital. Home visits were made where required. Parent(s)/guardian(s) were requested to complete a questionnaire and to bring it with them at the time of their appointment.

Each child was seen with parent(s)/guardian(s), usually at the centre they were attending, in order to have each candidate in their most comfortable and familiar setting. Interviews and medical examinations commenced in April 1998 at nine venues and continued throughout the school timetables through to June 1999. Patient information was collected on the data sheets, which included clinical assessment, growth measurements, hearing screening, capillary blood finger prick sample results and parental concerns. Clinical findings requiring follow up were referred back to the General Practitioner or Paediatrician caring for the child/adolescent for further management. Each Special Services Centre promoted and accommodated this project.

2.4 Clinical assessment schedule

1. The Questionnaire data collection sheet was completed by interview with parent(s)/guardian(s) by myself (JM).
2. The Lecturer in Paediatrics, Dr Magued Philip (MP), performed all clinical examinations, with particular reference to general health, puberty, cardiac status, clinical signs of thyroid dysfunction and visual abnormalities.
3. The Senior Clinical Audiologist (Ms Aoife Walsh) performed the outer ear examinations by otoscopic review and Pure Tone Audiometry in comparative groups.
4. Growth measurements, hearing screening and capillary blood samples by simple finger prick were performed by myself (JM).

2.5 Data Collection Instruments

The questionnaire included sections on:-

- Demographic and socio-economic characteristics of family and child
- Delivery of diagnosis
- Early intervention
- Uptake of childhood immunisations
- Clinical history relating to medical problems including diagnosis, treatment and parental concerns were recorded in the following areas; growth, cardiac, thyroid, hearing, vision, orthopaedic, cervical spine instability and speech and language problems.
- Primary school education uptake and satisfaction
- Secondary school education uptake and satisfaction
- Special education uptake and satisfaction

- Parent(s)/guardian(s) satisfaction with services provided
- Quality of life of parent(s)/guardian(s) rearing a child with special needs

2.5.1 Growth measurements

- Height was assessed using the portable Leicester height measurer and recording to the last completed millimetre. Children over 2 years of age were measured standing up straight in bare feet, head in the Frankfurt plane with a gentle upward pressure applied under the mastoid processes by the measurer. Children under 2 years of age were measured in the supine position using the portable Holtain Infantometer.
- Weight was assessed using a Seca electronic self-calibrating scales, which was checked regularly. Children were weighed to the last completed 0.1kgs in bare feet and wearing light clothing only.
- Head circumference was measured using a steel Holtain Harpenden anthropometric tape of 0.6cm width.
- Pubertal development was recorded by the Lecturer in Paediatrics (MP) using the methodology described by Tanner (1973)
- Age of menarche was ascertained by questionnaire.
- All growth measurements followed the methodology as recommended by Cameron and were performed by the Research Nurse Specialist (JM) (Cameron 1978)

2.5.2 Cardiac anomalies

A cardiovascular examination was performed on each patient by MP. Cardiac anomalies and screening tests, age at diagnosis and treatment were recorded by

parent(s)/guardian(s) on the questionnaire and verified with the assistance of the EUROCAT register and also by reviewing the children's medical records where possible with parental/guardian consent.

2.5.3 Thyroid Dysfunction

Capillary whole blood thyroid stimulating hormone (TSH) samples were obtained by finger prick using a Softclix II autolet and filling one circle on the newborn screening (Guthrie) card. The cards were sent to the National Newborn Screening Laboratory at the Children's University Hospital where thyroid stimulating hormone (TSH) was measured using a monoclonal antibody fluoro immuno assay. All results were returned by post to the National Children's Hospital for recording and dissemination.

The criterion for diagnosing hypothyroidism was a blood TSH >10 mU/l. Children with results above this range had a venous sample taken for plasma TSH and free T4, which were measured using Elecsys 2010 system. Microsomal and thyroglobulin antibodies were measured by murex haemagglutination assay. Towards the end of the study the laboratory changed their analysis of thyroid autoantibodies to thyroid peroxidase (TPO) antibodies using an enzyme linked immuno sorbent assay (ELISA) with Varelisa kit (Pharmacia Upjohn). Children requiring further investigation were referred back to their General Practitioner or Paediatrician.

2.5.4 Vision

Clinical history of vision impairment, previous screening and parental concerns were recorded in the questionnaire by parent(s)/guardian(s) and each patient had a clinical examination performed consisting of inspection of strabismus, nystagmus, cataracts,

Keratoconus, light reflex and eye movement using an ophthalmoscope by the Lecturer in Paediatrics (MP).

2.5.5 Hearing Impairment

Two studies of hearing screening were assessment was undertaken under the following two aspects.

First study of hearing screening – was performed on 394 children participating in the study using three screening techniques (otoscopy, tympanometry and distortion product oto acoustic emissions (DPOAE)) as detailed below.

Second study of hearing screening:- assessed 20 children with Down syndrome and an age matched control group of 20 without Down syndrome, using four screening techniques (otoscopy, tympanometry, DPOAE and Pure Tone Audiometry) as detailed below.

First study of hearing screening - Hearing screening was performed at nine development and medical educational centres, one special school, the National Children's Hospital and home visits were also undertaken. In addition each parent/guardian completed a section in the questionnaire that included information about the child's clinical history of hearing assessment, impairment and treatment.

Hearing screening was performed by the following techniques: -

- External ear and eardrum were screened by otoscopy by the Lecturer in Paediatrics, Dr Magued Philip (MP). Abnormalities of the external ear which

could contribute to loss of auditory acuity, including middle ear effusion, tympanic perforation and cerumen (wax) impaction were recorded

- Middle ear function was screened by tympanometry using the Grason-Stadler, Inc. GSI 38 Auto Tympanometer and a probe pitch with a tone of 226 Hz. The results were graded according to guidelines of Jerger et al 1970 (Jerger 1970) Results were graded as illustrated in Table 2.2 (page 282)

Distortion Product Oto Acoustic Emissions (DPOAE) screening was performed to assess inner ear function and identify any possible abnormalities i.e. nerve or possible mixed (conductive and nerve) hearing loss. DPOAE screening was carried out using the GSI 60 unit attached to a laptop computer. The mode of stimulation used was two primary tones L1 and L2 set at specific dB sound pressure level (SPL). These give rise to a prominent responding distortion produce at $F2/F1$ (ratio of 1.22) frequency. Background noise (noise floor amplitude) is defined as the amplitude (sound level) of background noise that occurs at or near the test frequency at time of recording. Two DPOAE tests were performed:- 1) L1 = 65 dB SPL; L2 = 55 dB SPL; 2) L1=65 dB SPL; L2=50 dB (SPL) (Table 2.3).(page 282) These levels of screening had been recommended in previous studies which showed that moderate level primary stimulating tones elicited responses that separated normal from impaired hearing better than either lower level or higher-level stimulating tones (Stover 1996, Lonsbury-Martin 1990, Gorga 1996).

Two harmonic sounds were introduced into the ear in order to assess inner ear function over the frequencies from 1-8 kHz. The outer hair cells in the inner ear (cochlear) responded to the stimulation through the microphone in the ear probe. The

results were automatically plotted against frequencies (kHz 1-8) on an x y graph (called DP Gram) on the attached computer giving values for the response value (amplitude) together with the noise floor level (natural body sound) surrounding the response during the measurement. GSI 60 criterion for Pass/Refer was used i.e. two DPOAE product emissions needed to score ≥ 3 dB above noise floor amplitude level at two points across all 3 octaves to Pass (GSI User Manual 1997, Lonsbury-Martin 1997) (Table 2.4) (page 283)

The DPOAE test was performed by inserting a small probe with an appropriately sized soft rubber tip into the ear canal, followed by a gentle twist in order to create a firm seal, which is crucial to the success of screening by DPOAE. This was achieved by gently but firmly lifting the pinna upwards and backwards. It was essential that the child sit or lie quietly during screening but no subjective response was required from the child. All results were crosschecked by the Senior Clinical Audiologist (AW) for accuracy by checking the tympanogram and DPOAE printed results with the reported findings before input on the computer for statistical analysis.

Statistical analysis: Children were divided into yearly groups for age and gender. Results include the number of patients participating in the study and the number of patients tested by each screening tool during the study. The relationship between otoscopy, tympanometry and OAE is ear-specific rather than patient-specific. Percentages are expressed as per cent of total ears actually tested rather than percent of total available ears. Mean ear canal volumes were calculated for each age group. DPOAE amplitudes level results and noise floor amplitude levels were input onto the

computer and their mean amplitude levels were calculated across frequencies 1000 to 8000 kHz.

Second study of hearing screening In order to compare reliability of hearing screening by DPOAE with Pure Tone Audiometry a group of 20 children with Down syndrome. Two children from each yearly age group from the full study (394 children) were randomly selected to make up a sample of 20 children. This was undertaken using the random selection facility on the SPSS computer software package. The age matched control group included 20 children without Down syndrome with no previous history of hearing problems. These were recruited from children of staff and neighbours.

Hearing screening was performed at nine venues, at the National Children's Hospital and home visits were also undertaken. The following techniques were used: otoscopy, tympanometry as outlined above. Distortion Product Otoacoustic Emissions (DPOAEs) using the GSI 60 unit attached to a laptop computer using stimulating tones 65/55 dB for this aspect of the study.

Pure Tone Audiometry to assess hearing threshold levels using Clinical Audiometer Model GSI 61 and screening was carried out by Senior Clinical Audiologist, Ms Aoife Walsh (AW).

Pure Tone Audiometry (PTA) is the procedure most commonly used for the measurement of hearing impairment. Pure tones (sounds with a single frequency of vibration) are presented via headphones (air conduction) or via a bone vibrator (bone

conduction) and the child's sensitivity at discrete frequencies is measured. Screening by PTA may take as long as 30 minutes to perform and children with DS may be able to cooperate at approximately 8 years of age.

The results were recorded on a data sheet and categorised in the following order; normal hearing, mild hearing loss, mild/moderate hearing loss, moderate hearing loss, moderate/severe hearing loss, severe hearing loss as described by Lee (1999) 'Hearing Thresholds' and illustrated in Table 2.5 (page 283).

The hearing procedures were explained to parent(s) / guardian(s) and child. The test was demonstrated and it was explained that a musical sound would be heard, which would not hurt and that their ears would work very hard to 'draw a picture on the two computers'. It was also explained that they must sit or lie very quietly, not talk or move their head. Each child was usually rewarded with a carton of juice for sitting still and allowing their 'ears draw pictures'.

2.5.6 Orthopaedic Problems

Clinical history of orthopaedic problems and parental concerns were recorded and each patient had a physical examination performed by the Lecturer in Paediatrics (MP).

2.5.7 Speech and Language

History of speech problems, previous screening and parental concerns were recorded by parent(s)/guardian(s). Each patient was observed clinically for speech and

language problems and a history was taken from the parent by the Lecturer in Paediatrics (MP).

2.5.8 Quality of Life

Parent(s)/guardian(s) perceptions of family perceived difficulty or burden on parents and/ or siblings in caring for a child with Down syndrome at home were assessed using an adapted questionnaire previously developed for those with diabetes (Hoey 2001) (Appendix 1, section 17). It comprised 7 questions to evaluate family burden (difficulty) relating to caring for a child with Down syndrome. The areas included:-

- Parent(s)/guardian(s) perception of child's general health and quality of life.
- Disruption to parent(s)/guardian(s), family routines.
- Physical problems requiring extra parental care.
- Concerns relating to a child's long-term health were also explored in relation to gender, age of child and health problems. In addition parents were asked for their views on how they were informed of the diagnosis of DS. They were also questioned on employment and family structure.

Questions were scored from 1 to 4; a lower score indicated less burden or better QOL

Cardiac Anomalies

Cardiac anomalies were coded into categories as designed by Pueschel 1980 for ease of statistical analysis as illustrated.

Cardiac Categories as per Pueschel 1980

- 1) No cardiac anomaly.
- 2) Single cardiac defects, such as atrial or ventricular septal defect without pulmonary vascular involvement. These children generally did not require cardiac medications nor cardiac surgery and had a normal growth rate.
- 3) More complex cardiac problems, such as endocardial cushion defects. These children often required diuretics. Their growth was generally sub optimal.
- 4) Very serious anatomical heart lesions, such as complex endocardial cushion defects, Fallot's tetralogy, etc. These children were often in cardiac failure, and most had pulmonary complications and generally reduced growth.

2.6 Data Analysis

a) Editing the data

The information was collected by questionnaire, clinical examination and screening procedures and the data were then coded by me personally. During this process any incorrectly completed questionnaires were detected and parents contacted to obtain the correct information. The coded data were then entered onto the computer using the Microsoft Excel programme and transferred to the statistical software package (SPSS version 12.01).

b) Computation

Data analysis was carried out using a personal computer DELL Pentium and the Statistical Programme for Social Science SPSS version 12.0.1

2.7 Statistical Analysis

Statistical analysis was carried out with the assistance of Dr. Myra O'Regan, Senior Statistician, Department of Statistics, University of Dublin, Trinity College.

i) Age and sex

Decimal ages were used throughout. The children were divided into half yearly and yearly age groups, for example, 4.0 years included 3.75- 4.24 years and so on. All measurements were plotted graphically by age and sex groups.

ii) Distribution

Means and standard deviations (SD), standard error of the mean, scatter plots, maximum and minimum values were determined.

The standard deviation (SD) of a series of observations was calculated as follows:

$$SD = \sqrt{\frac{x^2 - \frac{(\sum x)^2}{n}}{n-1}}$$

with x representing the values of the observations and n the number.

The standard error of the mean (SEM) was calculated by the formula:

$$SEM = \frac{SD}{\sqrt{n}}$$

Where n = number of observations.

95% confidence limits were determined by the formula:

$$\bar{x} \pm (1.96 \times SE)$$

where x-bar was the mean.

Means and standard deviations (SD) were calculated for height and head circumference and median values for weight. These were determined for age and sex and comparisons with present Irish national standards were undertaken as follows: - height and weight (Hoey 1987); body mass index (Cole 1996); head circumference (birth to 5 years) (Nelhaus 1968) and head circumference (5-18 years) (Hoey 1990). In addition growth comparisons were undertaken with Down syndrome specific growth standards as follows:- American height and weight (Cronk 1988), American head circumference (Palmer 1992) and Dutch height and weight (Cremers 1996). Standard scores (z scores) were used to calculate the SDS difference in the mean heights of the cohort of the general Irish population, by yearly age groups by gender, using the following equation: -

$$Z \text{ score} = \frac{x_1 - x_2}{SD_1}$$

where $\underline{x_1}$ represents the mean values of the height of the Irish general population, $\underline{x_2}$ represents the mean value of the height of the children with Down syndrome and SD_1 represents the standard deviation of the Irish general population at each age group. These were calculated for yearly age groups.

iii) Estimates of the Probability

a) Completeness of Ascertainment of population

The determination of completeness of ascertainment and estimation of the true number of incident cases in the population was made using the capture-recapture method. The estimate of the total number of incident cases of children with Down

Syndrome in the area studied was made using the following formula (LaPorte et al, 1993)

$$N = \frac{(M+1)(n+1)}{(m+1)} - 1$$

Where N is the total number of incident cases of Down Syndrome, M is the number of cases identified in the primary source (the capture), n is the size of the secondary source (the recapture) and m is the number of cases included in both sources.

The variance of N is estimated by:

$$\text{Var (N)} = \frac{(M+1)(n+1)(M-m)(n-m)}{(m+1)^2(m+2)}$$

The 95% confidence interval for the estimate of the total number of cases is:

$$95\% \text{ C.I.} = \pm 1.96\sqrt{\text{Var (N)}}$$

The degree of completeness of case ascertainment for each source is identified using the following equations (WHO Multinational Project for Childhood DIABETES< DIABETES Mondial – Diamond, 1991)

Primary Source, percentage ascertainment	= M ÷ N * 100
Secondary Source, percentage ascertainment	= n ÷ N * 100
Total estimated ascertainment	= $\frac{[M +(n-m)]}{N}$ * 100

b) Menarche

The mean age of menarche and the standard deviation were determined by probit analysis (Finney 1952). The percentage of girls who were post-menarchal was plotted for successive chronological age groups and a probit curve was fitted, from which median age, standard error and confidence limits were compiled to give an s-shaped curve.

The probit procedure provides the maximum likelihood estimates of the intercept,

slope, and natural (threshold) response rate for biological assay data. The probit equation was used:

$$\Phi^{-1}(y) + 5 = (A+Bx)$$

Where Φ is the cumulative distribution function of the standard normal distribution

X is the age of menarche

Y is the probability of a response

The Chi square (χ^2) test was used for goodness of fit of data to the underlying model of the probit curve

$$\chi^2 = \sum \frac{(O - E)^2}{E}$$

Where O = observed value

E = estimated value

Statistical significance was accepted with a p value of <0.05 .

Descriptive summary statistics were generated and levels of association between variables were explored using Chi-Square test of probability. Results include the number of patients participating in the study and the number of patients tested by each hearing screening tool used during the study.

Hearing screening analysis was performed on ear-specific and patient-specific findings. Percentages are expressed as per cent of total ears actually tested rather than percent of total available ears.

Confirmation of internal consistency of completing the Quality of Life section of the questionnaire was assessed using Cronbach's α coefficient as follows: -

$$\alpha = \frac{N \cdot \bar{r}}{1 + (N-1) \cdot \bar{r}}$$

Where N is equal to the number of items and $\bar{r}$ is the average inter-item correlation among the items.

Log transformation (Armitage 1998) was used when the total scores of the QOL questionnaire scale were calculated and the distribution was found to be positively skewed. The formula used was: $-100 \log_{10}$. This resulted in a more normal distribution and allowed the use of parametric statistical analysis.

Kappa (Cohen's 1996) measurement of agreement was used between the evaluations of the two hearing screening tests Pure Tone Audiometry (PTA) and DPOAE results. A value of 1 indicates perfect agreement. A value of 0 indicates that agreement is no better than chance. The equation for kappa was: -

$$k = \frac{\text{Pr}(a) - \text{Pr}(e)}{1 - \text{Pr}(e)}$$

Where $\text{Pr}(a)$ was the relative observed annotator agreement and $\text{Pr}(e)$ was the probability that agreement was due to chance.

The independent samples T-test procedure was used to test the significance of the difference between the two sample mean amplitudes of the children with and without

Down syndrome screened by DPOAE for inner ear function. The formula used was as follows: -

$$t = (\bar{x}_1 - \bar{x}_2) / \sqrt{A * B}$$

$$\text{Where } A = (n_1 + n_2) / n_1 n_2$$

$$\text{and } B = [(n_1 - 1)s_1^2 + (n_2 - 1)s_2^2] / [n_1 + n_2 - 2]$$

X-bar is the sample mean for each group and s is the sample standard deviation for each group.

Patient confidentiality was maintained by ensuring that their names were not included on the database and by giving each patient a unique identification number. The computer was password-protected and remained in a locked room in the Department of Paediatrics (Data Protection Act 1988).

CHAPTER THREE

RESULTS

CHAPTER 3 RESULTS

3.1 CLINICAL QUESTIONNAIRE

3.1.1 Response Rate

Five hundred and seventy one (571) families were contacted by letter through their services and 415 parents responded to our letters. Three hundred and ninety-four (394) parents/guardians (69%) agreed for their child to participate in the study and this included 13 additional children, born outside the set age frame, at the request of the Medical Development Services (3 children born in 1997, 3 born in 1980 and 7 born in 1998).

Three hundred and fifty two (352) of the children seen were registered with EUROCAT and 42 were not on the EUROCAT register (15 born outside the register catchment area and 27 born in maternity hospitals within the register catchment area but to parents resident outside the catchment area at the time of the child's birth).. 33% of parents seen were members of DSI and present on register at the time of the study.

3.1.1.1 Estimate of Ascertainment

The secondary source of case ascertainment yielded 394 cases in the study. Of the cases seen 352 were common to both the primary and secondary source. The remaining 42 were not appropriate for registration on the EUROCAT register for the reasons stated above. 224 children on the register were not seen and as they are

unknown to us we cannot comment on them. Using the capture-recapture formula shown in Chapter 2, the estimate of the total number of cases of children and adolescents with Down syndrome in the area and timeframe studied is 919 (N). The variance of N is 346.8 and 95% confidence interval of 955.9 – 882.

The estimated ascertainment is thus 61.7% for the primary source; 62.1% for the secondary source; and 85.5% for both sources combined.

3.1.2 Demographic/Socio-economic Characteristics of Family

The majority of families included the biological parent(s) of the children with Down syndrome, one child was adopted, and one child was being cared for by her sister and brother in law.

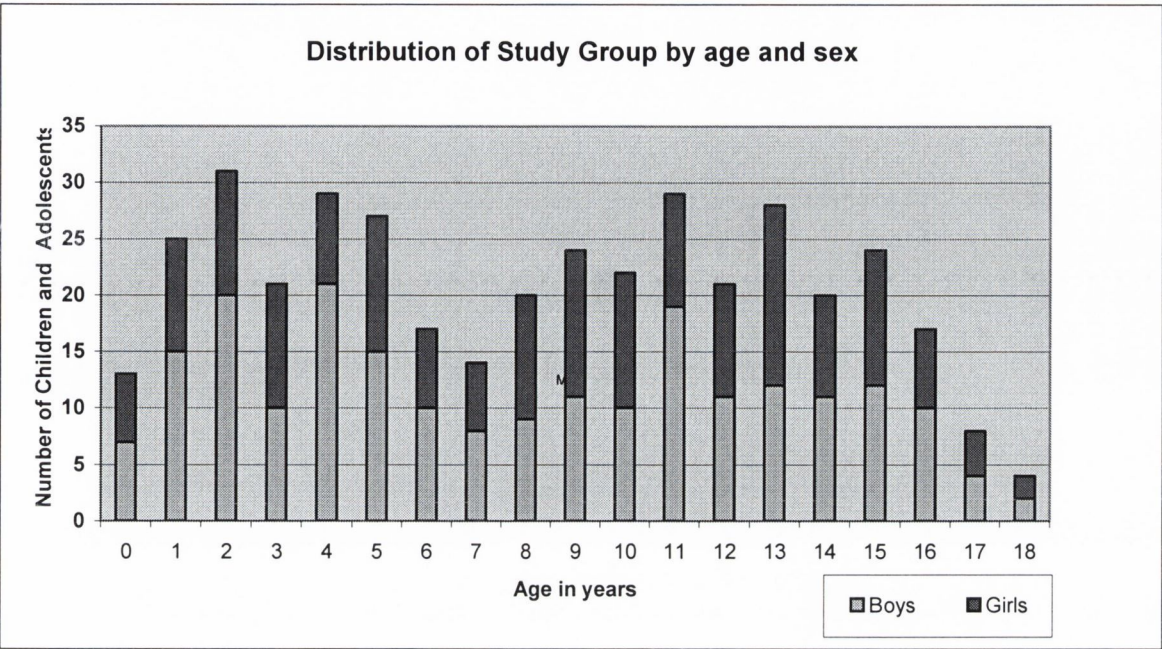
3.1.2.1 Age group and sex

There were 217 boys and 177 girls. The age profile of the children and adolescents who participated in the study ranged from 3 months to 18.9 years with smaller numbers in the older age groups. The mean age was 9.1 years (median 9.0 years)

for boys and 9.6 years (median 10.0 years) for girls (Table 3.1 and Figure 3.1).

Table 3.1 Age group and sex of children in the study			
Age group (yrs)	Boys N = 217	Girls N = 177	Total = 394
<1	7	6	13
<2	15	10	25
<3	20	11	31
<4	10	11	21
<5	21	8	29
<6	15	12	27
<7	10	7	17
<8	8	6	14
<9	9	11	20
<10	11	13	24
<11	10	12	22
<12	19	10	29
<13	11	10	21
<14	12	16	28
<15	11	9	20
<16	12	12	24
<17	10	6	16
<18	4	5	9
<19	2	2	4

Figure 3.1 Age group and sex of children in the study



3.1.2.2 Family size

Parents reported the number of children in the family. The average family size was 3.6 children (*median 3 children, range 1 to 13 children*). (Table 3.2.2)

Table 3.2 Family size				
Number of children	1 (%)	2 (%)	3 (%)	4+ (%)
Families	37 (9)	85 (22)	99 (25)	172 (44)

3.1.2.3 Birth Order

Parent(s) were asked the birth order of their child with Down syndrome within the family. 18% of children with Down syndrome were first-born, 24% were second

born, 22% were third born, 15% were fourth born and 10% were fifth born. One child was born as the thirteenth baby in the family (Table 3.3).

Table 3.3 Birth order of child with Down syndrome		
Child	N = 390	99%
1	72	18
2	95	24
3	87	22
4	60	15
5	39	10
6	15	4
7	11	3
8	4	1
9	4	1
11	2	1
13	1	0

3.1.2.4 Siblings

The number of male siblings (brothers) ranged from none to 9 with an average being one brother in 34% of the families (Table 3.4).

Table 3.4 Male siblings in family		
Male siblings	N = 392	99%
None	120	31
1	134	34
2	76	19
3	37	9
4	14	4
5	6	2
6	2	0.5
7	2	0.5
9	1	0.3

The number of female siblings (sisters) ranged from none to 6 with an average of one sister in 37% of the families (Table 3.5).

Table 3.5 Female siblings in the family		
Female siblings	N = 392	99%
None	113	29
1	146	37
2	81	21
3	36	9
4	13	3
5	2	0.5
6	1	0.3

3.1.2.5 Survey population

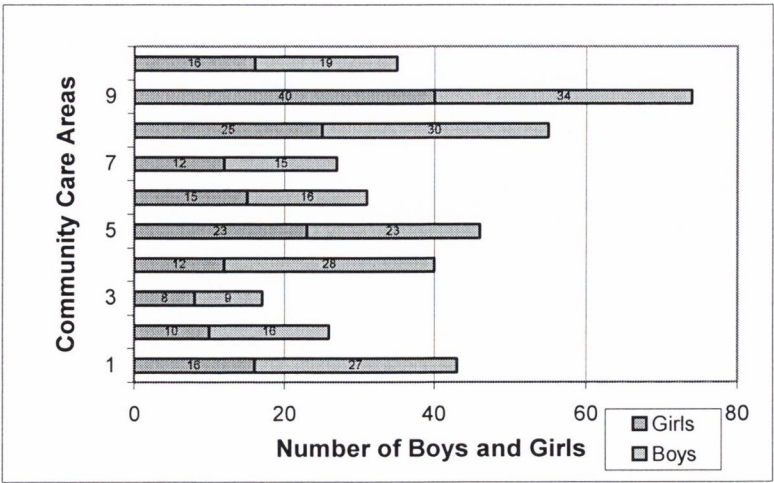
The number of families with 4 and/or more children represented 44% in this study, which was greater than the general population of 11% in 2002 which had fallen from 27% in 1981. In a previous study by Mercedes Egan (1993) of children with Down syndrome the larger family size was reported in 51% of families of children with Down syndrome in contrast to our findings indicating a similar downward shift in family size to the general population (Table 3.6).

Table 3.6 Survey population				
	Children in family			
	1 (%)	2 (%)	3 (%)	4+(%)
Present Down syndrome study group	9	22	25	44
Down syndrome study (Egan M 1993)	8	13	25	51
Irish Census 1981	27	26	20	27
Irish Census 2002	35	34	19	11

3.1.2.6 Geographical distribution

The distribution of boys and girls in each of the ten Community Care Areas in the Eastern Health Board (EHB) is presented in Figure 3.2 and Table 3.7.

Figure 3.2 Distribution of boys and girls in each Community Care Area in EHB



Dublin County with eight Community Care Areas had the highest proportion 72% of children with Down syndrome participating in the study whilst 28% of children with Down syndrome in Kildare and Wicklow counties participated in the study.

Table 3.7 Distribution of boys & girls in each Community Care Area EHB							
	Community Care Area	Girls (N=177)	%	Boys= (N= 217	%	Total (N= 394)	%
1	Dun Laoghaire	16	9.0	27	12.4	43	10.9
2	Dublin South East	10	5.6	16	7.4	26	6.6
3	Dublin South Central	8	4.5	9	4.1	17	4.3
4	Dublin South West	12	6.8	28	12.9	40	10.2
5	Dublin West	23	13.0	23	10.6	46	11.7
6	Dublin North West	15	8.5	16	7.4	31	7.9
7	Dublin North Central	12	6.8	15	6.9	27	6.9
8	Dublin North	25	14.1	30	13.8	55	14.0
9	Co. Kildare	40	22.6	34	15.7	74	18.8
10	Co Wicklow	16	9.0	19	8.8	35	8.9

3.1.2.7 Place of birth

The majority of children (94%) who participated in the study were born in Dublin maternity hospitals, one child was born in a general hospital, 2% were born at home,

3% were born outside the ERHA; 1% in the United Kingdom and 1 child in Bangladesh, Denmark, Norway, Saudi Arabia and Sweden (Table 3.8).

Table 3.8 Place of birth of child		
Birthplace	N=392	99%
Dublin Maternity Hospitals	366	94
Home Birth in ERHA	6	1.5
General Hospital	1	0.3
Cavan	1	0.3
Galway	1	0.3
Laois	1	0.3
Limerick	1	0.3
Louth	2	0.5
Sligo	1	0.3
Wexford	3	0.8
Bangladesh	1	0.3
Denmark	1	0.3
Norway	1	0.3
Saudi Arabia	1	0.3
Sweden	1	0.3
United Kingdom	4	1.0

3.1.2.8 Type of birth delivery

Parents were asked how their child was born. Natural birth occurred either in hospital or at home in 78% whilst 23% had their babies delivered by Caesarean Section (Table 3.9)

Table 3.9 Type of birth delivery		
Delivery	N =394	100%
Natural	305	77.5
Caesarean	89	22.5

3.1.2.9 Gestation

Parent(s) were asked about the duration of their pregnancy. There was a small proportion (9%) of mothers with pregnancy duration of between 28 to 35 weeks, while the majority (91%) of mothers had pregnancy duration of between 36-43 weeks (Table 3.10).

Table 3.10 Pregnancy gestation		
Weeks	N =390	98%
28-31	6	2
32-35	27	7
36-39	196	50
40-43	161	41

3.1.2.10 Marital status

The majority of parents were married (83%), whilst 8% were separated, 6% were single parents and 4% were widowed. One family had adopted their child with Down syndrome (Table 3.11).

Table 3.11 Marital status		
Status	N=392	98%
Married	326	83
Separated	30	8
Widowed	14	4
Single Parent	22	6

3.1.2.11 Parental age

The distribution of maternal and paternal age at time of birth of baby with Down syndrome is shown in Figures 3.3 and 3.4.

Figure 3.3 Mother’s age at time of birth of baby with Down syndrome
The Maternal median age was 35.0 years (Range 16 to 49 years of age) at birth of child.

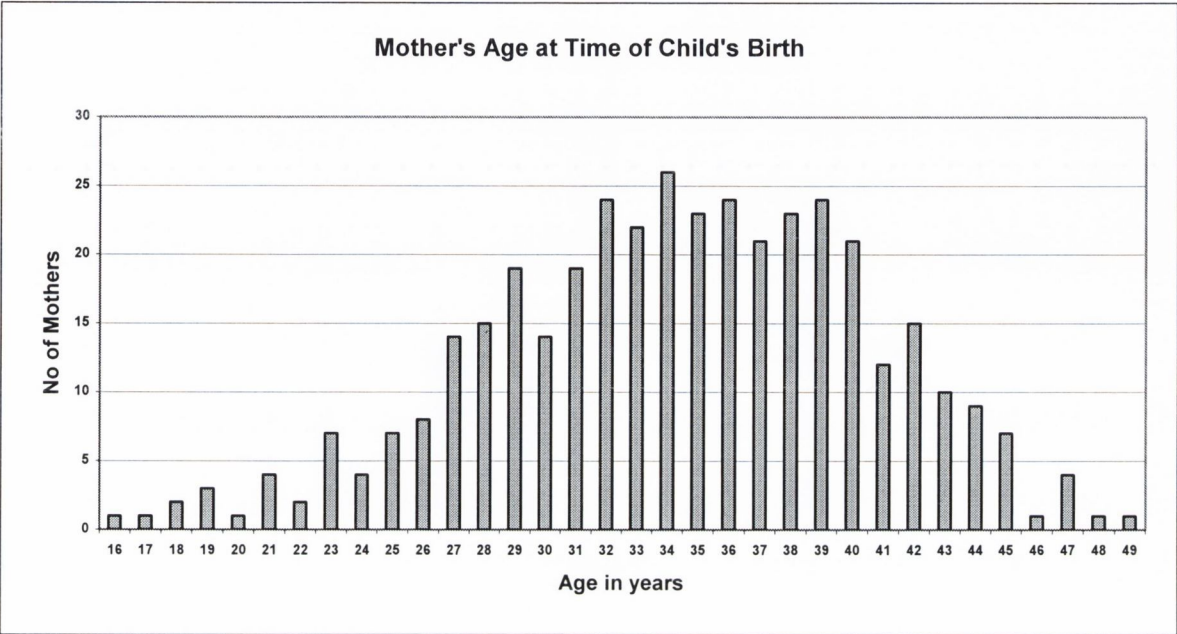
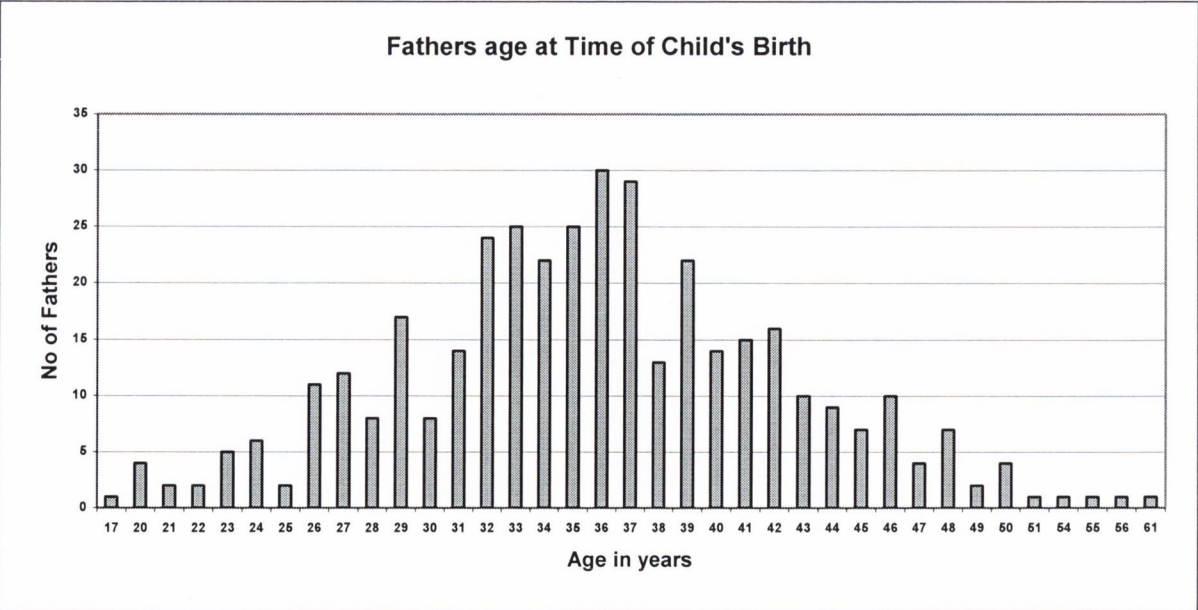


Figure 3.4 Father’s age at time of birth of baby with Down syndrome
The Paternal median age was 36.0 years (Range 17 to 61 years) at birth of child.



3.1.2.12 Parent employment status

Fathers were asked if they were working, 95% responded and of these 93% were working and 7% were not working at the time of the study. Five fathers were deceased (Table 3.12).

Table 3.12 Father Employed		
Working	N = 368	93%
Yes	341	93
No	27	7

Mothers were asked if they were working outside the home and 98% responded, 37% were working full time, 2% were working part-time and 61% were not working outside the home. Two mothers were deceased (Table 3.13). These figures suggest that the mother was the primary person caring for the child with special needs at home.

Table 3.13 Mother Employed		
Working	N = 385	97%
Yes	141	37
Part time	6	2
No	238	61

3.1.3 Diagnosis / Information / Education and Services Received

Services received and parent satisfaction relating to diagnosis of Down syndrome, information and education are described.

3.1.3.1 When told diagnosis of Down syndrome

Parents reported that 88% were told their child’s diagnosis straight away and a further 7% were told within two to seven days after the baby’s birth whilst 3% reported being told over a period of two weeks to four months after the birth of their baby (Table 3.14).

Table 3.14 When told diagnosis of Down syndrome		
When told	N = 394	100%
Straight away	346	88
Day 2	6	2
Day 3	12	3.0
Week 1	10	3
Week 2	1	0.3
Week 3	1	0.3
Week 6	5	1.3
Month 3	2	0.5
Month 4	1	0.3
Did not remember	9	2.2
During pregnancy	1	0.3

When asked who was with the mother when given the diagnosis 96% of parents responded indicating that 63% had support and 37% were either alone or with their baby (Table 3.15).

Table 3.15 Who was with mother when informed of diagnosis		
Person with mother	N=377	96%
Mother alone	105	28
Mother and baby	34	9
Mother and father and baby	64	17
Mother and father	171	45
Mother and friend	3	1

3.1.3.2 Location where informed of diagnosis and parent satisfaction

Parent(s) reported that 66% had been given their child’s diagnosis in a private location, 29% were in a non-private location and 4% did not respond

Table 3.16 Location where informed of diagnosis		
Location	N =394	100%
Private	262	66
Non private	115	29
Did not respond	17	4

Parent(s) were asked if they were satisfied with the way they were told their child’s diagnosis. 64% of parents were satisfied whilst 36% were not satisfied.

The majority of parents (91%) responded when asked whether they perceived that enough time had been given when communicating the diagnosis relating to their child with 55% stating that enough time had been given to them whilst 45% perceived that insufficient time had been given to them during this crucial time (Table 3.17).

Table 3.17 Time spent communicating diagnosis		
Enough time	N =361	92%
Yes	197	55
No	164	45

When parent(s) were asked if they had been offered the opportunity for a follow up discussion 93% of parent(s) responded, 54% said yes, 46% said no and 7% did not wish to respond to this question (Table 3.18).

Table 3.18 Follow up discussion		
Response	N =366	93%
Yes	198	54
No	168	46

Only 42% of parents stated that they were satisfied with service that they had received relating to this communication whilst 58% of respondents were not satisfied.

3.1.3.3 Information provided at birth relating to Down syndrome

The majority of parents (69% of the 95% of parents who responded) reported that they were given information about DS whilst 31% stated that they were given no information at the time of their child’s diagnosis (Table 3.19). When asked if they found this service satisfactory, 57% responded, 40% were satisfied and 60% were not satisfied.

Table 3.19 Information provided on Down syndrome at diagnosis		
Received	N = 373	95%
Yes	256	69
No	117	31

When asked who gave them information about Down syndrome after their baby was born 87% of the parent(s) responded; 47% had been given the information by their consultant and 29% by either their paediatrician or general practitioner. The midwife, ward sister, nurse or the social worker gave the information to the remaining 24% of parents. One mother knew the diagnosis before the birth of her baby and had been given the information by her consultant at the time of prenatal diagnosis (Table 3.20).

Table 3.20 Person who gave information about Down syndrome		
	N =343	87%
Consultant	160	46.6
Paediatrician	7	2.0
GP	94	27.4
Midwife	58	16.9
Ward Sister	5	1.5
Nurse	6	1.7
Social Worker	12	3.5
Knew before birth	1	0.3

3.1.3.4 Chromosomes and genetic counselling

Parent(s) were asked whether chromosome testing had been performed on their child. All parent(s) responded to this question and confirmed that 79% of children had a blood test to detect chromosomal abnormalities, 2% said that their child’s blood was not checked for chromosomal abnormalities and 19% reported that they did not know if their child’s blood had been taken for chromosomal analysis and they held no records (Table 3.21).

Table 3.21 Down syndrome chromosome screening		
	N = 394	100%
Yes	312	79
No	7	2
No record held	75	19

In order to identify the specific karyotypes of Down syndrome information was crosschecked with the EUROCAT register. General practitioners, consultant paediatricians, medical development services, maternity hospitals, the departments of genetics at both Our Lady’s Hospital for Sick Children, Crumlin, and Galway University Hospital Genetic Centre, Great Ormond Street Hospital, Guys & St Thomas NHS Foundation Trust (S E Thames Regional Genetic Centre) London were contacted in an effort to trace the results of the unknown cases (Table 3.22). Cases were identified as follows: EUROCAT 39; Maternity Hospitals 7; Private Hospital 1; GPs 6; Genetic Centre OLHC 12; Guys & St Thomas Cytogenetic Department 4.

Table 3.22 Down’s syndrome karyotype		
	N = 394	100%
Trisomy 21	368	93
Translocation	6	2
Mosaic	7	2
Karyotype unavailable/ clinical diagnosis	13	3

Parent(s) where asked if they were offered genetic counselling and 94% responded; 23% were offered this service and 77% were not offered the service. Only a minority of parents (16% of 78% who responded) reported that they had availed of the service.

3.1.3.5 Parent support by social worker

Parent(s) reported that the majority of the 95% who responded were visited by a social worker whilst 30% were not (Table 3.23).

Table 3.23 Parent support by social worker		
Social Worker	N = 374	95%
Yes	261	70
No	113	30

Almost two thirds of parent(s) were visited by a social worker within the first six week period, a further 4% were visited between two and nine months after the birth of their baby, however 35% of parent(s) perceived it was not necessary to be visited by the social worker (Table 3.24).

Table 3.24 When visited by social worker		
Time after birth	N =317	81%
1-4 Days	93	29
1-6 Weeks	100	32
2-9 Months	12	4
Not Necessary	112	35

3.1.3.6 Information on services available to parents

Parents were asked what information was given to them relating to the services available to them and their child and 80% responded (Table 3.25).

Table 3.25 Information on services available for children with DS		
Responses	N=314	80%
DSAI booklet	32	10.2
Early Service	105	33.4
DSAI booklet and Early Services	20	6.4
Early Service and tax grant	3	1.0
Special schools	1	0.3
Physiotherapy	4	1.3
Breastfeeding	1	0.3
Very little	7	2.2
Mum given a picture of what to expect for child	1	0.3
DSAI mother visited	4	1.3
Physiotherapy and speech therapy	1	0.3
Everything there was to know	1	0.3
Perceived not necessary	134	42.7

The major delay in provision of services related to speech therapy alone or in combination with other services. One parent said she had to wait for all services whilst another mother said she did not have to wait for any of the services she required for her child (Table 3.26).

Table 3.26 Delay in services		
Service	N =135	34%
Speech Therapy	63	46.7
Speech Therapy and Psychology	9	6.7
Speech Therapy and Home Teacher	4	3.0
Speech Therapy and Early Services	2	1.5
Speech Therapy and Physiotherapy	3	2.2
Speech Therapy/Social Worker/Physiotherapy	24	17.8
Home Teacher	8	6.0
School	8	6.0
Psychology	5	3.7
Social worker	2	1.5
Physiotherapy	2	1.5
Psychology, Occupational therapy	1	0.7
Occupational therapy	1	0.7
School bus	1	0.7
Everything	1	0.7
Nothing	1	0.7

3.1.3.7 Early intervention

Birth to two years

Parent(s) were asked if they used early services from birth to two years of age for their child and 98% responded. Seventy-six percent (76%) stated that they had used these services and 24% had not (Table 3.27).

Table 3.27 Early services (birth to 2 years)		
Received service	N =386	98%
Yes	294	76
No	92	24

When asked if they were satisfied with the services received 57% responded, 87% were satisfied and 13% were not satisfied with these services (Table 3.28).

Table 3.28 Early services satisfaction (birth to 2 years)		
Satisfaction	N =244	57%
Satisfied	194	87
Unsatisfied	30	13

Three to five years

Parent(s) were asked if they had used early services from three to five years of age for their child. The response rate was 91%, 60% had used early services whilst 40% had not used the early services (Table 3.29).

Table 3.29 Early services (Three to five years)		
Received service	N =359	91%
Yes	217	60
No	142	40

The response rate regarding satisfaction with early services was 42% indicating that 85% were satisfied and 14% were not satisfied with this service (Table 3.30).

Table 3.30 Early services satisfaction (Three to five years)		
Satisfaction	N =165	42%
Satisfied	141	85
Dissatisfied	24	14

3.1.3.8 Pre-school participation

Parent(s) were asked if their child participated in pre-school education and 94% responded. Forty-six percent (46%) of children attended special school and 25% attended private Montessori preschool (Tables 3.31 and 3.32)

Table 3.31 Special school attendance		
Attended	N =362	92%
Yes	166	46
No	196	54

Table 3.32 Montessori school attendance		
Attended	N =349	86%
Yes	88	25
No	261	75

Parent(s) indicated that 97% were satisfied whilst 3% were not satisfied with the services provided by the Montessori school (Table 3.33).

Table 3.33 Montessori school satisfaction		
Satisfied	N =68	17%
Yes	66	97
No	2	3

3.1.3.9 Primary school education

Parent(s) were asked if their child had attended primary school with additional educational support and 84% responded, 13% had attended and 87% had not (Table 3.34).

Table 3.34 Primary school + support attendance		
Attendance	N =329	84%
Yes	43	13
No	286	87

Satisfaction with this service was reported by 8% of parent(s), 67% were satisfied and 33% were not satisfied with the service (Table 3.35).

Table 3.35 Primary school and support service satisfaction		
Satisfied	N =33	8%
Yes	22	67
No	11	33

Only 17% of children had attended primary schools with no additional educational support whilst 84% had not (Table 3.36)

Table 3.36 Primary school without support		
Attendance	N =327	83%
Yes	54	17
No	273	84

Parent(s) were asked if they were satisfied with this service and 12% responded, which represents 83% of parent(s) whose children attended primary schools without additional educational support. Satisfaction was reported by 82% of parent(s) and 18% were not satisfied with the service provided (Table 3.37).

Table 3.37 Primary school service without support satisfaction		
Satisfied	N =45	11%
Yes	37	82
No	8	18

3.1.3.10 Special school education

Parent(s) were asked if their child attended special school at any time and 92% responded, 46% attended special schools and 54% did not. The average age of starting at special school was 5.1 years (median age 5 years, range 2 to 15 years) (Table 3.38).

Table 3.38 Child attend special school		
Attended	N =362	92%
Yes	166	46
No	196	54

Only 3.2% of parents responded to the question enquiring if they were satisfied with the service of the special schools and they reported that they were not satisfied with the service received (Table 3.39).

Table 3.39 Special school service satisfied		
Satisfied	N =13	%
No	13	100.0

3.1.3.11 Secondary school education

Parent(s) were asked if their child attended secondary school and 62% responded. Seven percent (7%) attended secondary schools and 93% did not (Table 3.40)

Table 3.40 Secondary schools participation		
Attended	N =237	%
Yes	16	7
No	221	93

Only 3% of parents responded to the enquiry as to whether they were satisfied with this service and the majority (82%) were satisfied with the service at the secondary school whilst 18% were not satisfied (Table 3.41).

Table 3.41 Satisfaction with service of senior school		
Satisfied	N =11	3%
Yes	9	82
No	2	18

3.1.4 Uptake of childhood immunisations

The uptake of childhood vaccines is motivated by the Public Health Nurses and administered by the General Practitioner.

3.1.4.1 Bacillus Calmette-Guerin (BCG) vaccine

The BCG vaccine is an active immunizing agent prepared from Bacillus Calmette-Guerin and is administered for immunization against tuberculosis.

Parent(s) were asked if their child had received the BCG vaccine in the newborn period and 95% responded reporting an 84% uptake of the vaccine (Table 3.42).

Table 3.42 BCG Vaccine in newborn period				
	Yes (%)	No (%)	Not yet (%)	Total
Vaccine	329 (84)	47 (12)	18 (4)	394

Parent(s) reported that 10% of children had received the booster BCG vaccine (part of the Republic of Ireland Immunisation programme is offered to those who are known to be tuberculin negative and have not previous BCG.) between the ages of 10 to 14 years (Table 3.43).

Table 3.43 BCG Vaccine at 10 to 14 years				
	Yes (%)	No (%)	Not yet (%)	Total
Vaccine	41 (10)	80 (20)	273 (69)	394

3.1.4.2 Diphtheria, Tetanus and Pertussis (Three in One) vaccine

The ‘three in one’ vaccine is given to immunize against Diphtheria, Tetanus and pertussis (whooping cough). It is generally administered with the oral polio vaccine. Children also receive Haemophilus influenza type b (Hib) vaccine which provides protection against bacterial meningitis, pneumonia and also joint, bone and throat infections. Infection occurs mainly in the first 5 years of life. The ‘three in one’ vaccine is generally given at 2 months, 4 months and 6 months of age. Uptake of 68% was reported for all three doses (Table 3.44).

Table 3.44 Diphtheria, Tetanus, Pertussis + Hib Vaccines					
3 in 1	Yes (%)	2 in 1	Yes (%)	No	Total
1 st	260 (68)	1 st	112 (29)	20 (5)	394
2 nd	259 (68)	2 nd	111 (29)	24(6)	394
3 rd	258 (68)	3 rd	106 (28)	30(8)	394

3.1.4.3 Diphtheria, Tetanus (Two in one) vaccine

The ‘two in one’ vaccine is an immunization against Diphtheria and Tetanus provided for children whose families don’t want the child to have pertussis vaccine or where there was thought to be a contra indication. Parent(s) were asked if their child had received the ‘two in one’ vaccine and 97.2% responded. Uptake of 29% was reported for the first vaccine, 29% for the second vaccine and 28% for the third vaccine (Table 3.44). The results of the 3 in 1 and the 2 in 1 vaccines were then cross-referenced and

we found that the overall uptake was 94% for first vaccine, 94% for second vaccine and 92% for third vaccine.

3.1.4.4 Measles, Mumps and Rubella (MMR) Vaccine

Children are generally offered their first MMR (Measles, Mumps and Rubella) vaccine at approximately 15 months of age. Uptake of 83% was reported for this vaccine.

Children generally receive the second MMR vaccine at either 4-5 years of age or 11-12 years of age. When asked if their child had received the second MMR 63% of parents responded, 16.7% had received the vaccine whilst 20.3% had not and 63% were not yet old enough to receive it (Table 3.45).

Table 3.45 Measles, Mumps and Rubella (MMR) Vaccine					
Vaccine	Yes (%)	No (%)	Not yet (%)	Eligible	Total
MMR at 15 months	297 (83)	43 (12)	20 (5)	3	331
MMR at 4-5 Years	153 (46)	78 (24)	100 (30)	87	331
MMR at 11-12 years	56 (18)	68 (20)	211 (62)	94	335

3.5 Feeding and Gastrointestinal system

3.1.5.1 Methods of feeding

Parent(s) were asked if they could recall how their baby was fed. There was a 96% response rate, which indicated that 64% were bottle fed, 21% breast fed, 8% fed by a combination of bottle and breast, 7.4% were tube fed over a period of time ranging from six to eighteen months and one child was fed intravenously in the first three months of life (Table 3.46).

Table 3.46 Method of feeding baby		
Response	N =380	100%
Bottle fed	243	64
Breast fed	78	21
Bottle and breast	30	8
Bottle and tube	4	1
Tube fed	24	6
Intravenous	1	0.3

3.1.5.2 Ability to suck well

Responding to a question whether their baby could suck well when feeding, 93% of parents responded, 51% of children could suck well whilst 45% could not (Table 3.47). This may have an effect on the baby’s growth and speech development.

Growth monitoring is therefore important and nutritional support may be required.

Early intervention to develop better sucking reflexes may also be necessary.

Table 3.47 Baby’s ability to suck well		
Response	N =369	93%
Yes	187	51
No	167	45
Not sure	15	4

3.1.5.3 Ability to chew well

The majority of the children (60%) were chewing well by one year of age, 28% by two years of age whilst a small proportion did not chew well until aged four year. Parent(s) of four children reported that their child had never chewed well (age range 3 to 12.6 years).

Table 3.48 Child chew well		
Age (months)	N =230	60%
3-6	54	24
6-12	84	37
12-24	65	28
24-36	18	8
36-48	5	2
Never	4	2

3.1.5.4 Age of taking semisolid feeds

Parent(s) were asked if they could recall when their baby started to take semi solids when feeding and 67% responded, reporting that 75% took semisolids by 12 months of age, however, some children were between 24 and 36 months of age before taking semisolid foods (Table 3.49).

Table 3.49 Age of starting semi-solid feeds		
Age (months)	N =266	67%
1-3	16	6.
3-6	49	18
6-9	80	30
9-12	54	20
12-15	8	3
15-18	21	8
18-24	20	8
24-36	18	7

3.1.5.5 Age of starting solid foods

The majority of children (75%) took solids by age of 12 months, however, some children were not taking solids until almost five years of age. Six children were not

yet taking solids (age range 6 months to 12.6 years) of whom four were under 3 years of age and two were aged over 11 years of age (Table 3.50).

Table 3.50 Age of taking solid foods		
Age (months)	N =260	75%
6	80	30.8
6-12	115	44.2
12-18	26	10.0
18-24	21	8.1
24-36	7	2.7
36-48	4	1.5

3.1.5.6 Recurrent Diarrhoea

A history of recurrent diarrhoea, 97% was reported in 17% of children. (Table 3.51).

Table 3.51 Recurrent diarrhoea		
Response	N =382	97%
Yes	64	16.8
No	318	83.2

The causes of recurrent diarrhoea varied and included; dairy products allergy, bowel

disorders, coeliac disease, infection, and cystic fibrosis in one case (Table 3.52).

Table 3.52 Cause of recurrent diarrhoea		
Cause	N =48	12%
Infection	15	31
Allergy to Dairy Products	5	10
Bowel disorder	5	10
Coeliac disease	3	6
Cystic Fibrosis	1	2
Unknown	19	40

Treatment included oral medication in 50% of cases, diet in 27% and surgery in 10% for duodenal atresia, necrotising enterocolitis and Hirschsprung disease.

Table 3.53 Treatment of recurrent diarrhoea		
Response	N =30	8%
Medical treatment	15	50
Diet	8	27
Surgery	3	10
None	4	13

3.1.5.7 Recurrent Constipation

Parent(s) were asked if their child had a history of recurrent constipation and 95% responded, 26% had a history of chronic constipation (Table 3.54).

Table 3.54 Chronic constipation		
Response	N =374	95%
Yes	96	26
No	278	74

Parent(s) reported wide ranging causes including; poor muscle tone, diet, bowel disorders, coeliac disease, hiatus hernia and hypothyroidism (Table 3.55).

Table 3.55 Cause of recurrent constipation		
Response	N =52	13%
Poor muscle tone	8	15
Diet	4	8
Bowel disorder	6	12
Hypothyroidism	2	4
Medical treatment	2	8
Hiatus Hernia	1	2
Coeliac disease	1	2
Unknown	27	52

Treatment included medication, improved diet and in 2 cases massage (Table 3.56).

Table 3.56 Treatment of chronic constipation		
Response	N =50	13%
Medical treatment	31	62
Diet	15	30
Massage	2	4
None	2	4

3.1.5.8 Recurrent Vomiting

Parent(s) were asked if their child had a history of recurrent vomiting and 95% responded, 12% has a positive history (Table 3.57).

Table 3.57 Recurrent vomiting		
Response	N =372	95%
Yes	46	12.
No	326	88

The main causes as reported by the parents were bowel disorders. Other reported causes included poor muscle tone, infections, dairy products intolerance, recurrent chest infections and in one child it was attributed to a nervous disposition.

Table 3.58 Reported cause of recurrent vomiting		
Response	N =28	7%
Bowel disorder	11	39
Poor muscle tone	3	11
Infection	2	7
Diary products intolerance	1	4
Recurrent chest infections	1	4
Nerves	1	4
Unknown	10	36

Treatment included dietary alteration for 4 children; oral medication for 3 children and one child with hiatus hernia was nursed in a sitting position after feeds. Three children had surgery for diaphragmatic hernia, hiatus hernia and bowel obstruction.

Table 3.59 Treatment of recurrent vomiting		
Response	N =17	4%
Surgical treatment	5	29
Diet	4	24
Medical treatment	3	18
Sitting position	1	6
None	4	24

3.1.6 History of surgical procedures

Parent(s) were asked if their child had ever had an operation and 97% responded reporting that 68% children had surgery performed. The results have been sub-grouped as follows; cardiac, ears nose and throat, gastrointestinal, genitourinary, eyes, orthopaedic, neurological and dental operations.

3.1.6.1 Cardiac

Corrective heart surgery had been performed on 15% of children whilst 85% had not required heart surgery.

3.1.6.2 Ear nose and throat

ENT surgery was wide ranging in the areas of ears, nose and throat and accounted for 43% of those who had surgery performed. The most common operations included insertion of grommets and removal of tonsils and adenoids (Table 3.60).

Table 3.60 Ear, nose and throat surgery		
Response	N =170	43%
Grommets	100	59
Tonsils adenoids grommets	34	20
Tonsils	16	9
Tonsils adenoids	8	5
Adenoids	4	2
Grommets and adenoids	2	1
Coanal atresia	2	1
Removal of wax	1	1
Cleft palate and grommets	1	1
Cleft lip and palate	1	1

3.1.6.3 Gastrointestinal Tract (GIT) and Urinary systems

Fifty-one children (13%) had corrective surgery performed for gastrointestinal tract and urinary system problems. The most common operations included correction of undecended testes, imperforate anus and bowel obstruction (Table 3.61).

Table 3.61 Operations of Gastric/Urinary systems		
Response	N =51	13%
Undecended testes	9	18
Imperforate anus	5	10
Diaphragmatic hernia	5	10
Circumcision	4	8
Blocked bowel	4	8
Hirschsprung disease	3	6
Duodenal atresia	3	6
Reflux	3	6
Umbilical hernia	2	4
Hiatus hernia	2	4
Inguinal hernia and undescended testes	2	4
Trachea oesophageal fistula	2	4
Necrotising enterocolitis	1	2
Duodenal atresia and undescended testes	1	2
Duodenal web	1	2
Hiatus hernia and duodenal web	1	2
Hiatus hernia and appendix	1	2
Hirschsprung disease and gall stones	1	2
Vesico-ureteric reflux (Sting)	1	2

3.1.6.4 Eyes

Parent(s) reported that 12% of children had corrective eye surgery which included cataract, strabismus and blocked tear ducts (Table 3.62).

Table 3.62 Eye operations		
Response	N =46	12%
Cataract	16	35
Blocked tear ducts	11	24
Strabismus	17	37
Blocked tear ducts and strabismus	2	4

3.1.6.5 Orthopaedic

Orthopaedic corrective surgery was performed on 3% of children and included fusion of the atlanto axial joint in 3 children for cervical spine instability. There were also operations to correct patella instability, scoliosis, toe joint correction and one child required separation of fused fingers.

Table 3.63 Orthopaedic operations		
Response	N =11	3%
Cervical spine instability	3	27.3
Patella instability	2	18.2
Tendon repair ankle	2	18.2
Scoliosis	1	9.1
Patella instability and toe joint repair	1	9.1
Toe correction	1	9.1
Separation of joined fingers	1	9.1

3.1.6.6 Neurological

One child had corrective surgery undertaken (cranioplasty) to correct early fusion of the skull bones for trigonocephaly (a form of craniosynostosis where there is premature fusion of the metopic suture resulting in a prominent, keel shaped forehead with hypertelorism (e.g. eyes widely apart).

3.1.6.7 Dental health

Parents reported that one child had extraction of teeth performed under general anaesthesia. On clinical examination the majority (89%) of the children were found to have good oral hygiene whilst 7% had poor oral hygiene, of whom 3 had gingivitis. In addition 41% were found to have overcrowding of teeth and 26% had received orthodontic treatment. It was also found that 2 children had a cleft lip and palate and that both had been surgically repaired successfully.

3.1.7 Development

In many children there is a great deal of individual variation in the age at which the different skills develop and psychomotor skills are achieved. Some children will have ongoing health problems which may slow their developmental milestones and thus they may require additional help in the identified developmental area in early childhood. Because of the increased health problems in children with Down syndrome this is an area that can be of great concern to the parents.

3.1.7.1 Developmental Milestones

Parents were asked if they could remember when their child was able to sit unsupported and 82% responded. Over 60% were able to sit alone by 12 months of age, with a further 13% sitting alone before 2 years of age. A small number of children sat alone by five years. The 15 children who were not yet sitting alone included 9 children under one year of age, 3 children under 3 years and 1 child under 4 years of age. The other two children were physically handicapped and were 15 and 17 years of age respectively (Table 3.64).

Table 3.64 Age when child sat unsupported		
Age (months)	N =323	82%
4-6	30	9.
6-12	220	68
12-18	38	12
18-24	14	4
2- 3 years	5	2
3- 5 years	1	1
Not Yet	15	4

The majority of children (70%) were able to stand-alone by 3 years of age, however only 15% could stand at the age of 12 months. The children not yet able to stand unsupported were all less than 3 years old (Table 3.66).

Table 3.65 Age when child able to stand unsupported		
Age (months)	N =321	82%
8-12	48	15.
12-18	110	34.
18-24	78	24.
24-36	39	12.
36-48	8	3
48-60	1	1
60-72	1	1
Not Yet	36	11

Only 47% of children were walking alone at the age of 2 years. The majority of children were walking alone by 3 years of age. There were 36 children not walking yet (age range from 3 months to 4 years) (Table 3.66).

Table 3.66 Age of walking unsupported		
Age (months)	N =363	92%
12-18	60	17
18-24	107	30
2-3 years	114	31
3-4 years	24	7
4-5 years	7	2
5-6 years	1	1
Not Yet	40	114

3.1.7.2 Speech development

Parent(s) were asked at what age their child could say single words and 72% responded. Only 25% of children were able to say single words by 12 months of age. The majority were able to say single words by the age of 3 years, whilst a smaller number of children had only achieved this by 7 years of age. There were 24 children not yet saying single words, 22 of whom were under 4 years with the remaining 2 children aged 11 and 12 years (Table 3.67).

Table 3.67 Age when able to say single words			
Age (months)	Yes (N=261)	Not yet (N=24)	72%
6-12	98	11	38
12-24	101	5	37
2-3 (years)	42	4	16
3-4	13	2	5
4-5	2		1
5-6	2		1
6-7	2		1
11		1	0.5
12		1	0.5

Most children were stringing two words together by four years of age, whilst some were not able to achieve this until 10 years of age. Sixty-five children had not yet started to put two words together, although the majority were under 5 years of age,. 8 children were aged between 5 and 16 years (Table 3.68).

Table 3.68 Age when child able to say two words together			
Age (months)	Yes (N =192)	Not yet (N=65)	65%
6-12	10	12	9
12-24	57	18	29
2-3 (years)	61	15	30
3-4	36	5	16.
4-5	13	7	8
5-6	5	3	3
6-7	4	1	2
7-8	4		2
8-9	1		1
9-10	1	1	1
Over 10		3	2

Only 21% of children were able to verbalize recognisable sentences by 3 years of age. A further 21% had achieved this by 5 years of age. However 40 children were able to speak recognisable sentences between 6 and 12 years and 101 children were unable to speak recognisable sentences (75 under the age of 5 years and 25 were between 6 and 18 years) (Table 3.69).

Table 3.69 Age when child speak recognisable sentences			
Age (years)	Yes (N =146)	Not yet (N=101)	63%
Under 3	53	52	42
3-4	31	11	18
4-5	22	13	14
5-6	15	5	8
6-7	4	2	2
7-8	6		2
8-9	4	2	2
9-10	5	2	3
10-11	3	1	2
11-12	3	2	2
12 +		4	2

3.1.7.3 Independence with feeding, dressing and tricycle riding

Parent(s) were asked at what age their child was feeding independently and 77% responded. The majority of children were able to feed themselves by the age of 3 years. There were 34 children not yet able to feed independently, 32 of whom were under the age of 3 years (Table 3.70).

Table 3.70 Age when child feeding independently			
Age (months)	Yes (N =234)	Not yet (N =34)	77%
8-12	31	13	16
12-18	45	16	23
18-24	63	3	25
2-3 (years)	87		33
3-4	31		12
4-5	6		2
5-6	3	1	1
6-7	2		1
11 +		1	1

The majority of children could dress themselves by 5 years of age. There were 89 children who were not yet dressing themselves, 76 of whom were under the age of 5 years (Table 3.71).

Table 3.71 Age when child able to dress independently			
Age (years)	Yes (N =201)	Not Yet (N =89)	74%
2	10	52	21
3	34	12	16
4	55	12	23
5	38	5	15
6	23	1	8
7	17		6
8	12		4
9	3		1
10-12	7	2	3
12-14	2	3	2
14 +		2	1

The majority of children could ride a tricycle by 5 years of age. There were 149 children not yet able to ride a tricycle, 34 under the age of 2 years (Table 3.72).

Table 3.72 Age when able to ride a tricycle			
Age (years)	Yes (N =147)	Not yet (N=149)	75%
2-3	42	58	34
3-4	41	14	19
4-5	35	18	18
5-6	17	10	9
6-7	7	7	5
7-8	2	1	1
8-9	2	3	2
9-10	1	6	2
10-12	1	10	4
12 +		22	7

3.1.7.4 Behavioural Development

Parent(s) were asked whether they had concerns about their child’s behaviour with regard to sleeping, eating, playing with other children, child tantrums and autism. The most common concerns related to sleeping patterns (23%), eating ability (18%), ability to play with other children (15%), temper tantrums (11%), whilst 1% of parents had concerns about autism (Table 3.73).

Table 3.73 Parent concerns			
Response	N =	Yes	%
Sleep	382	87	23
Eating	388	69	18
Playing with others	388	58	15
Temper tantrums	387	41	11
Autism	386	4	1

Professional help had been sought for these problems by 37% of parents who responded (Table 3.74).

Table 3.74 Seek professional help for behavioural problems		
	N =217	55%
Yes	81	37
No	136	63

The wide range of healthcare professionals who provided assessment and/or advice relating to behavioural problems are shown in Table 3.75. The most common health

professionals providing this service were the psychologist and paediatrician located either in the Special Services or at a hospital.

Table 3.75 Who helped with behavioural problem		
Response	N =77	20%
Psychologist	19	25
Paediatrician at Special Services	15	20
Paediatrician at hospital	13	17
Dietician	9	12
General Practitioner	8	10
Public Health Nurse	3	4
Nurse at hospital	3	4
Family members	2	3
General Practitioner and Nurse	1	1
Osteopath	1	1
Nurse at Special Service	1	1
Parents and school	1	1
Special teacher	1	1

The age at which the child presented with behavioural problems ranged from infancy to 15 years of age (Table 3.76).

Table 3.76 Age of presentation with behavioural problem		
Age of child (years)	N =107	27%
Birth to 1	33	30.8
1-2	15	14.0
2-4	21	19.6
4-6	25	23.4
6-8	8	7.5
8-10	3	2.8
10-15	2	1.9

3.1.7.5 Parent comments

Parent(s) were asked for additional comments relating to concerns. Responses provide a very vivid picture of concerns relating to their child’s behavioural problem, the service received, the healthcare professionals and also the perception of their child’s feelings in relation to his/her health and condition. Parents’ comments have been categorised as follows: -

Speech problems

“Poor speech results in child being left out, as others don't understand him”

“Difficulties arise from lack of verbal communication”

“Not sure how other children relate to him or understand him”

“Lack of speech is a major communication drawback”

Sleeping problems

“Child has trouble sleeping all night

“There is no medical person who understands her sleeping pattern other parents did help”

“Advice from home teacher re sleeping and eating. No problems now”

“He slept all day due to epilepsy. Once the epilepsy was treated problem solved”

“Physicians don't have same understanding of this (sleep) problem as social workers in the Special Service and Paediatricians”

Feeding problems

“Child used to choke on food for first one and a half years”

“Child fed very well but failure to thrive was attributed to Down syndrome (Child was later diagnosed with Cystic Fibrosis)”

“Choking while eating - supervision necessary”

Behaviour problems

“It was more or less a discipline problem getting child to do what he was told”

“Reason for behavioural problem was break up of marriage, starting new school and moving house”

“Child plays well at school but is still jealous of any child his own age coming to play at home, but this is improving”

“Tantrums - banged head off walls. Only lasted a couple of weeks”

“Child was a bit rough with other children for a period after starting school”

“Child watches other children. He is nervous of them”

“Child frustrated & angry when other children will not play with her”

“Hitting other children – trying to communicate”

3.1.7.6 Convulsions

Parent(s) reported that 12 (3%) of children had suffered from convulsions. All children had neurological tests and parents of 9 children recalled that an EEG had been performed and 1 child also had a computerised tomography (CAT) scan performed. Three children had been diagnosed with epilepsy requiring medication and continuous medical management whilst one child was still undergoing investigations for epilepsy. The remaining 8 children had febrile convulsions.

3.1.7.7 Family Support

Only 45% of parent(s) reported that they were members of a family support group. These included Down Syndrome Association of Ireland (34%), Parent Groups (6%), Bernardos (1%) and Special Services Support Groups (5%).

3.2 GROWTH

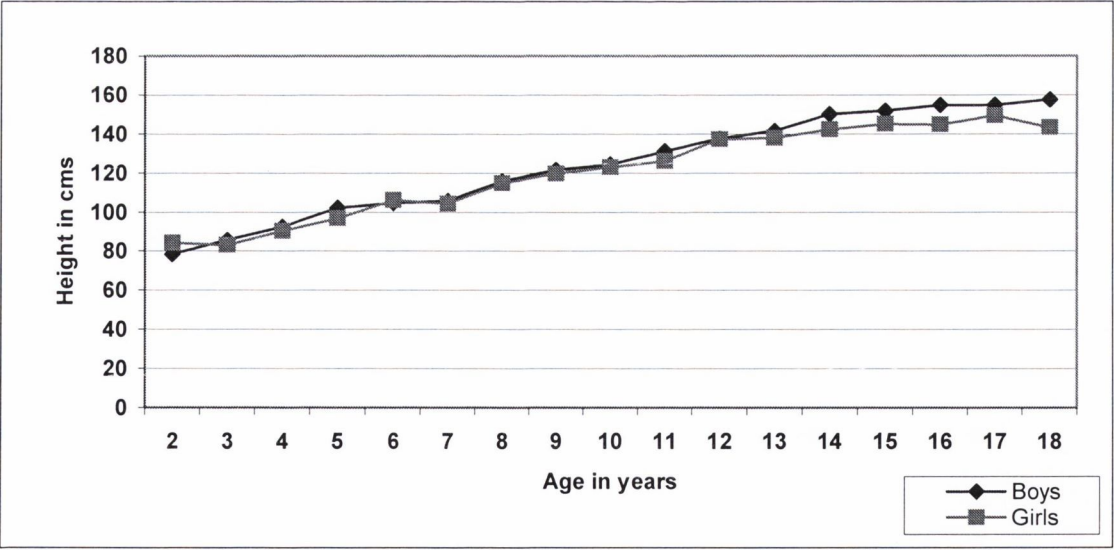
3.2.1 Growth Measurements

Growth measurements were attempted on all 394 children participating in the study, and a small number of children declined one or more measurements as they were unable to co-operate on the day of the study. The total number of children measured was 384 (97.5%) for height, 388 (98.5%) for weight and 379 (96.2%) for head circumference.

3.2.2. Irish Down Syndrome Population

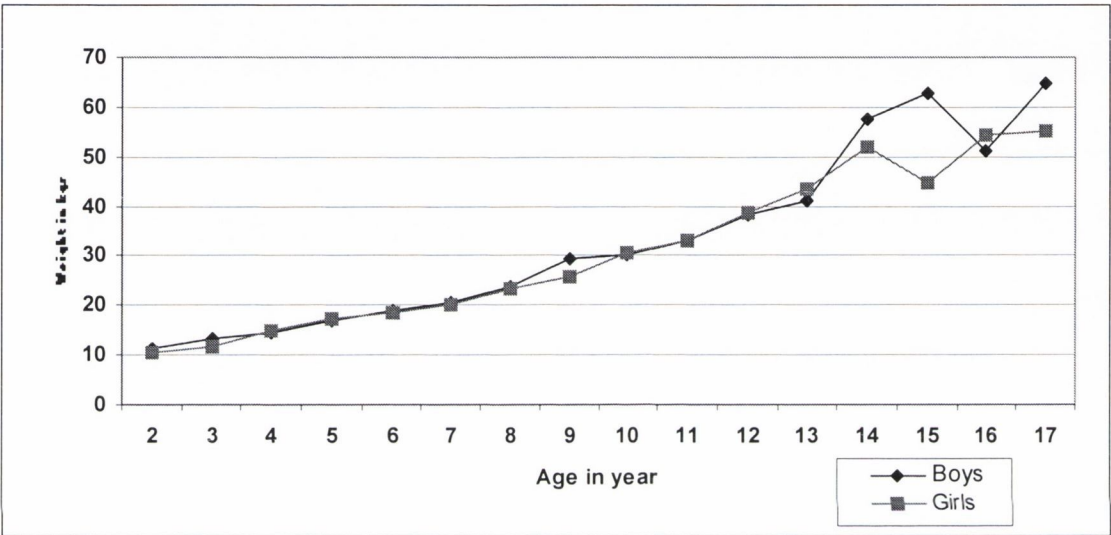
Height: - Irish boys with Down syndrome were consistently taller at all ages than girls with Down syndrome (Figure 3.5, Table 3.77). The mean final height of boys was 157.5 cms and 143.4 cms for girls.

Figure 3.5 Comparison of mean height of boys and girls with Down syndrome



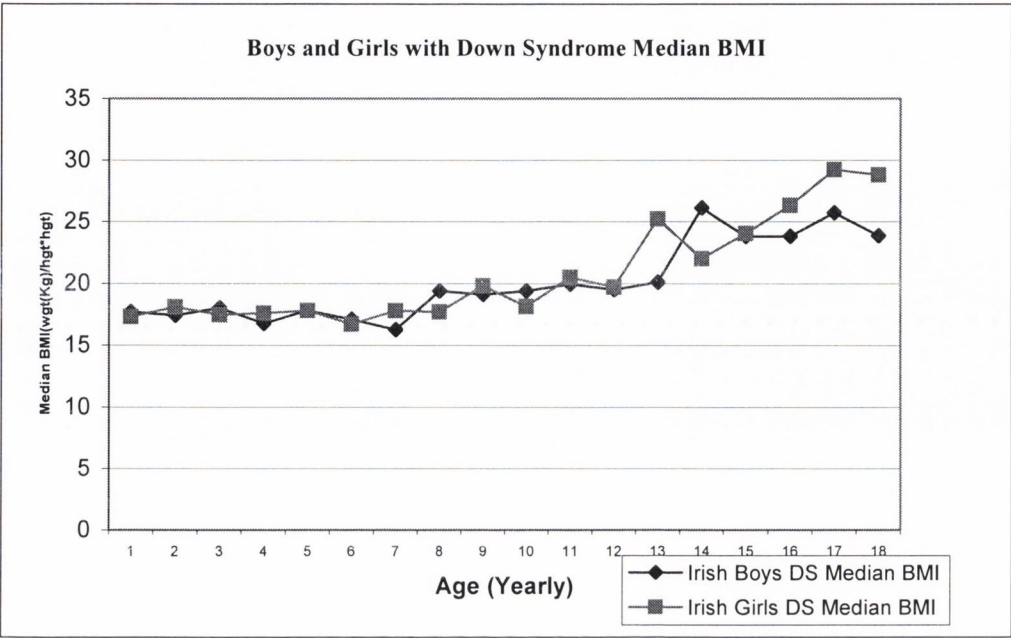
Weight: - Irish boys with Down syndrome were marginally heavier than girls under the age of 7.5 years and weight increased with greater divergence in boys thereafter (Figure 3.6, Table 3.78). Boys showed an increment in weight gain from 13.5 years compared to girls showing a greater weight gain at 12.5 to 13.5 years of age.

Figure 3.6 Comparison of median weight of boys and girls with Down syndrome



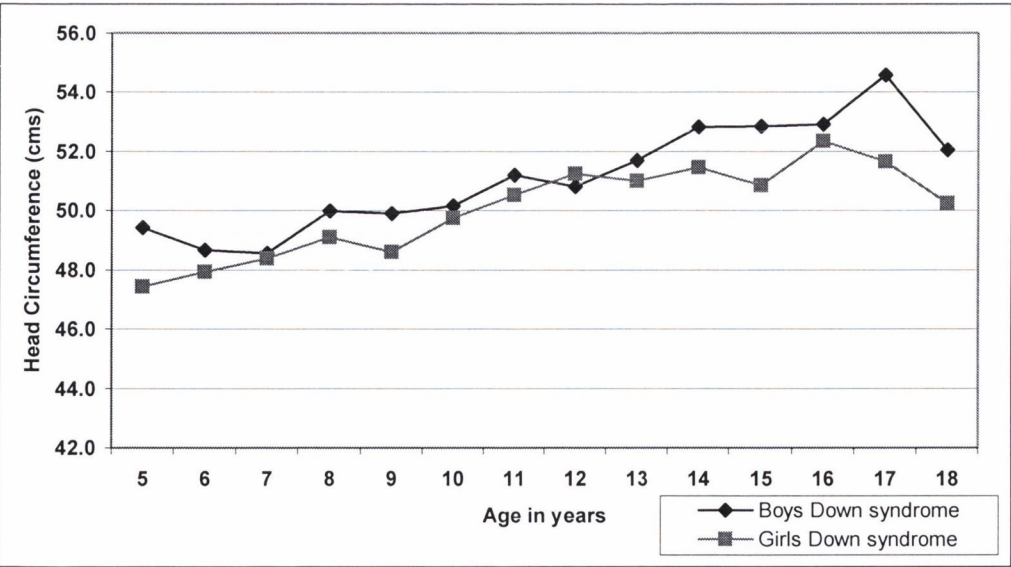
Body Mass Index (BMI); - Median BMI was similar in boys and girls with Down syndrome up to the age of 10 years with greater divergence in girls thereafter (Figure 3.7, Table 3.79).

Figure 3.7 Median BMI of Irish boys and girls with Down syndrome



Head circumference: - The mean head circumference of boys with Down syndrome was marginally larger than girls and increased with greater divergence from 13.5 years (Figure 3.8, Table 3.80)

Figure 3.8 Comparison of mean head circumference of boys and girls with Down Syndrome

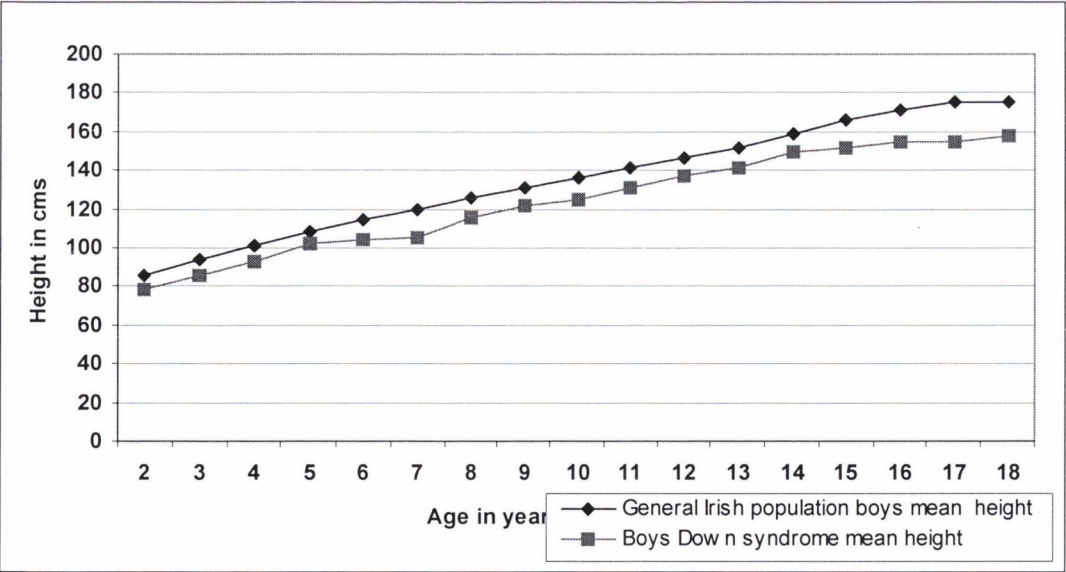


Growth cannot be assessed in isolation and comparisons with present Irish national standards were undertaken as follows: - height and weight (Hoey 1987); body mass index (Cole 1990); head circumference (birth to 5 years) (Nelhaus 1968) and head circumference (5-18 years) (Hoey 1990). In addition growth comparisons were undertaken with Down syndrome specific growth standards as follows:- American height and weight (Cronk 1988), American head circumference (Palmer 1992) and Dutch height and weight (Cremers 1996).

3.2.3 General Irish Population

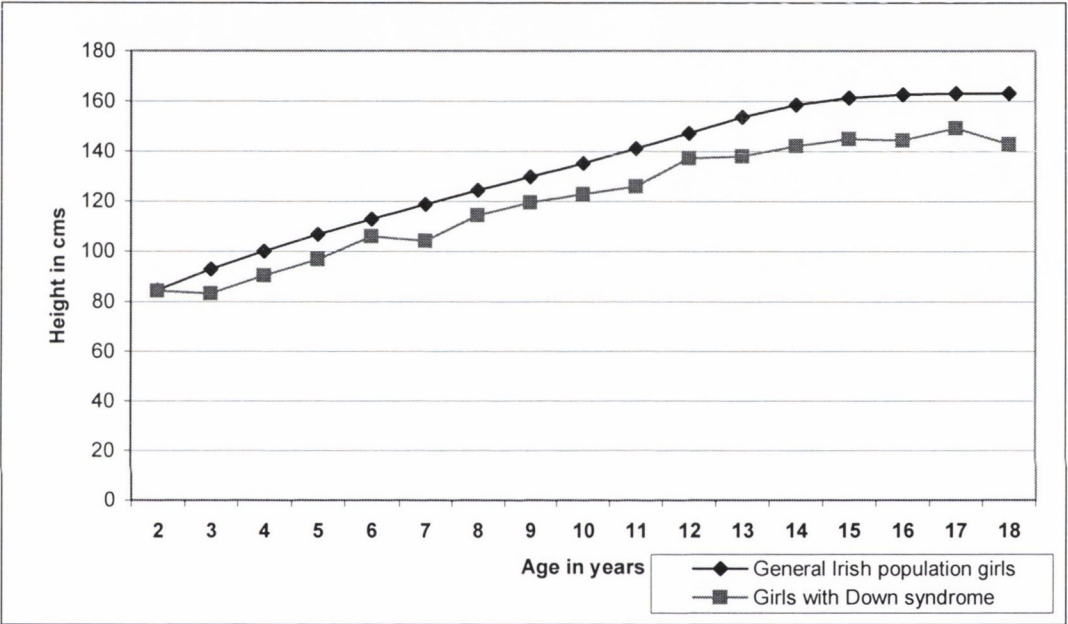
Height: - Compared with the general Irish population (Hoey 1987) the overall height of boys with Down syndrome was significantly shorter (-2.05 SD scores Table 3.81) with greater divergence from the age of 15 to 18 years (Figure 3.9).

Figure 3.9 Comparison of mean height of Irish boys with Down syndrome and general Irish population



Likewise, the overall height of girls was significantly shorter (Table 3.82), which was more marked from 12 to 18 years (Figure 3.10).

Figure 3.10 Comparison of mean height of Irish girls with Down syndrome and general Irish population (cms)



Weight: - When compared with the general Irish population the weight of those with Down syndrome was lighter in boys from birth to 5 years and in girls from birth to 6 years (Figures 3.11 and 3.12,Table 3.83). Boys showed increasing weight gain from 8 years whilst girls showed an increased weight from 11.0 years.

Figure 3.11 Comparison of median weight of boys’ DS with general Irish population

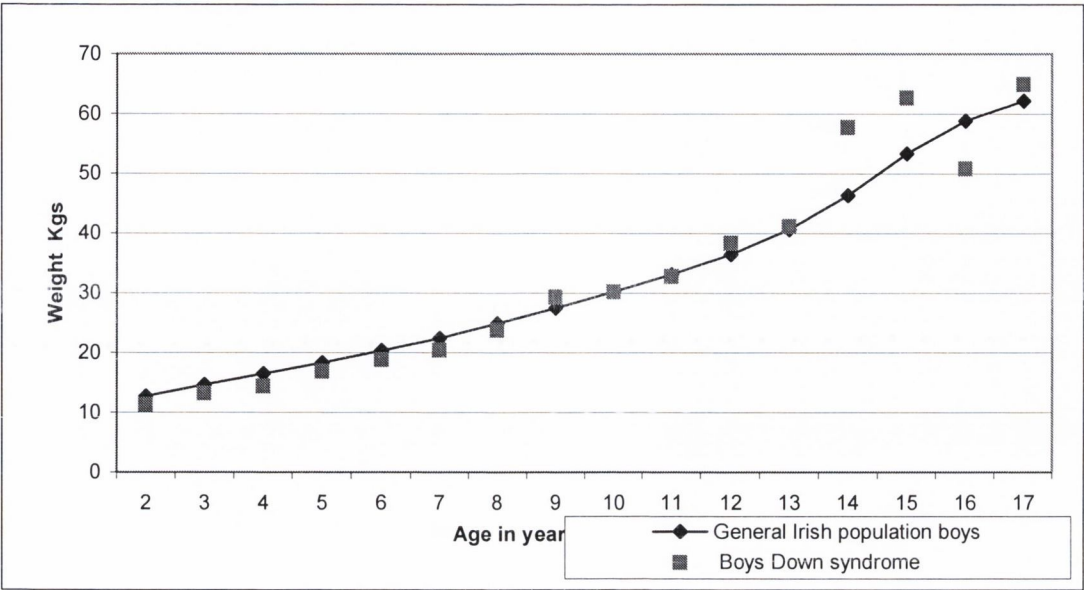
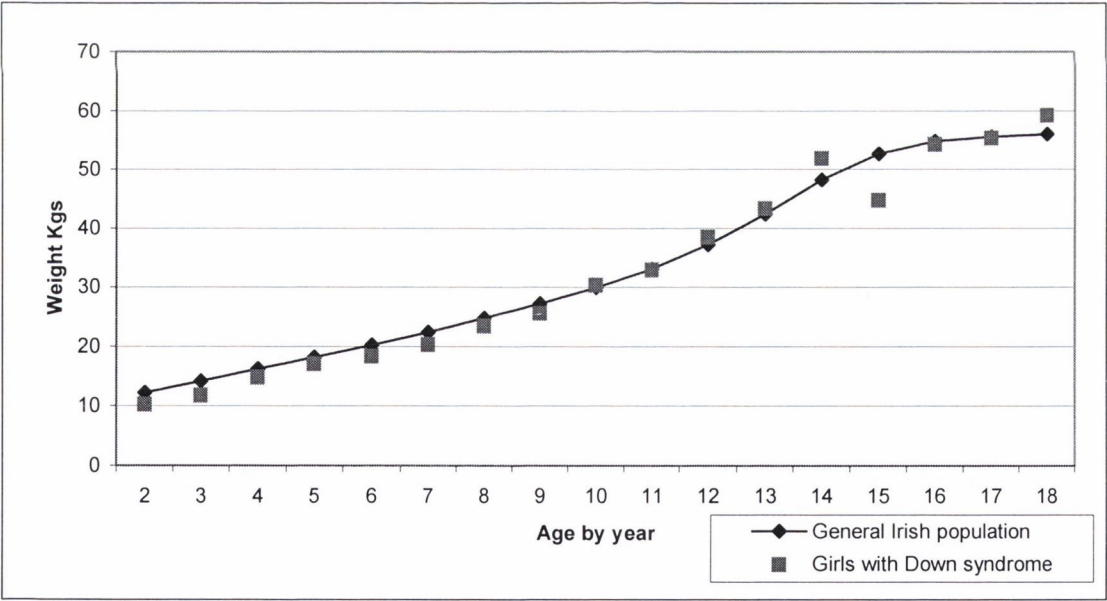
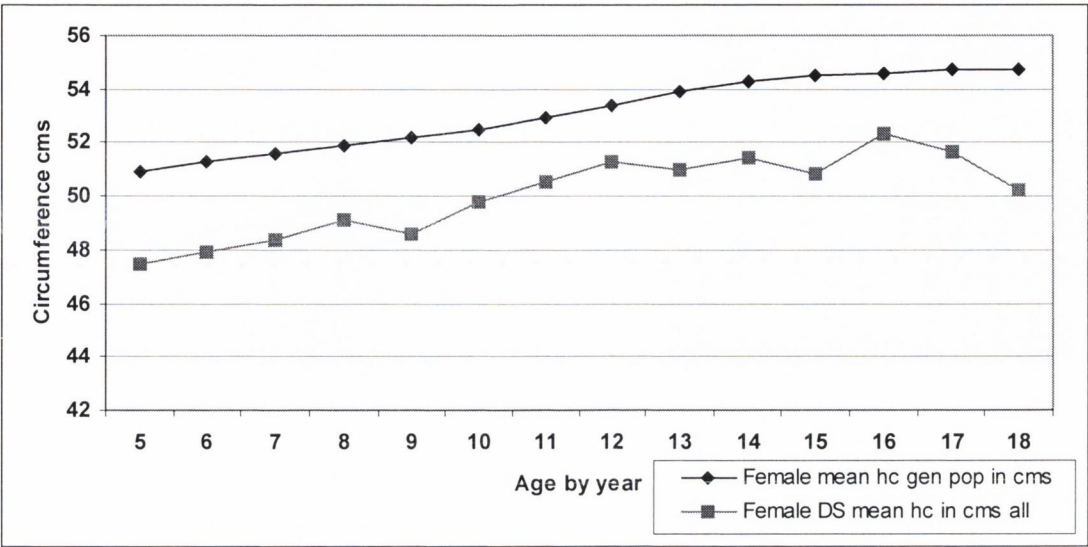


Figure 3.12 Comparison of median weight of girls’ DS with general Irish population



Body Mass Index (BMI) was therefore greater for children with Down syndrome as they had shorter stature in comparison to the general Irish population. Overweight and obesity are defined as those above the 91st percentile and 98th percentile respectively on the UK BMI reference charts (Cole 1996) presently in use in Ireland as the national standards. The data revealed that 17.3% of the children had a BMI greater than the 91st

Figure 3.14 Comparison of mean head circumference of girls with DS and general Irish population (cms)



3.2.4 American and Dutch DS populations

American Down syndrome specific growth charts (Cronk 1988, Palmer 1992) showed that Irish boys with DS were slightly taller (mean SD score 0.23) and heavier (mean SD score 0.3) from 3 months to 16 years whilst the girls were similar for height (mean SD score -0.3) and weight (mean SD score 0.3) (Tables 3.90 to 3.93) (Pages 297 to 300). In comparison to the American Down syndrome population, the Irish boys and girls with Down syndrome had a similar head circumference in the birth to 1 year of age groups. However Irish boys and girls had marginally larger head circumference at 3 years of age than the American Down Syndrome population (Figures 3.17 and 3.18 - pages 306 and 307).

Compared with the Dutch population with Down syndrome the data showed the Irish boys and girls were shorter at all ages (mean SD score -0.85SD and -1.21SD respectively). Irish boys and girls with Down Syndrome were lighter from birth to 13

and 12 years respectively following which the boys were heavier with increased weight gain with greater divergence from 14 – 18 years while the girls were heavier at 13 years and again at 17 years of age indicating their increased weight for height. Dutch head circumference data were not available for comparison (Tables 3.90 to 3.93).

3.2.3 Growth Subpopulations Comparisons

3.2.3.1 Karyotype

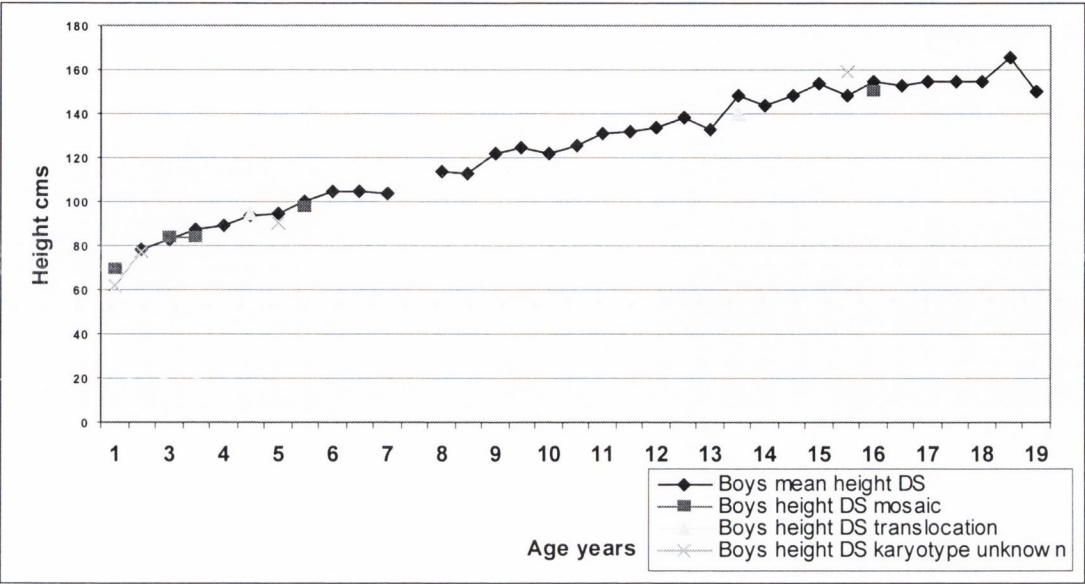
The majority of children had been diagnosed with Trisomy 21 (n= 368), translocation (n=6) and mosaic (n=7). However, 13 parents did not know their child’s karyotype and despite extensive research in Ireland and in London, karyotype data was not available for these children (Table 3.91).

Table 3.94 Down syndrome Karyotypes of children in Study

	Number	Percent
Trisomy 21	368	93.4
Translocation	6	1.5
Mosaic	7	1.8
Unknown	13	3.3
Total	394	100.0

The seven children with a mosaic karyotype (5 male, 2 female) had a height range similar to their peers with a trisomy 21 karyotype (Figure 3.19; 3.20). They were all below the 3rd centile for height on growth standards for the general Irish population.

**Figure 3.19 Comparison of boys’ height with Down syndrome karyotype
Trisomy 21 with mosaic,translocation and unknown karyotype**

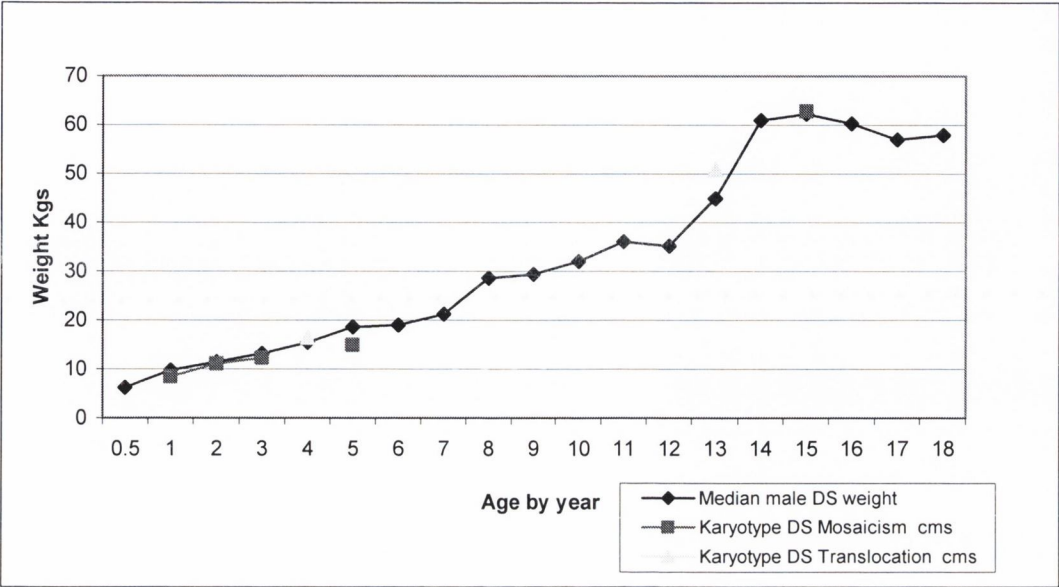


**Figure 3.20 Comparison of girls’ height with Down syndrome karyotype
Trisomy 21 with mosaic and translocation and unknown karyotype**

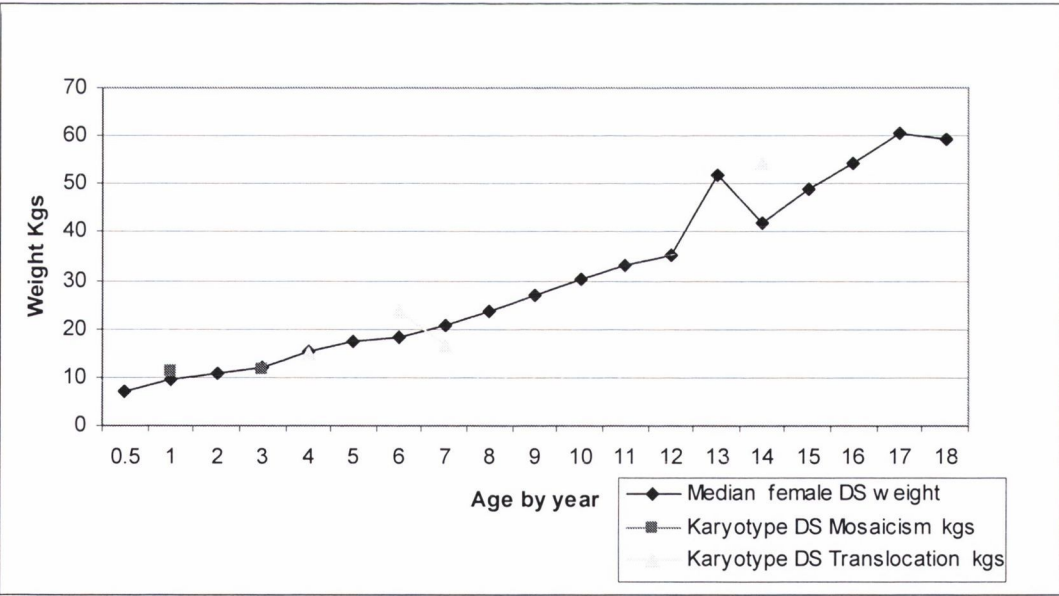


However, their weight was similar to their peers and comparable to the general Irish population (Figure 3.21 -3.22).

**Figure 3.21 Comparison of boys’ weight with Down syndrome karyotype
Trisomy 21 with mosaic and translocation**



**Figure 3.22 Comparison of girls’ Weight with DS Karyotype
Trisomy 21 with Mosaic and Translocation**



The 6 children with a translocation karyotype (3 male, 3 female) were within the height range of their peers and only 1.25 SD below the general Irish population mean height. The heights of the 13 children with no known karyotype compared well with their peers

who had karyotype trisomy 21, for both height and weight and for the purpose of analysis in this study were included in the group with karyotype Trisomy 21 (Figures 3.19 -3.20)

3.2.3.2 Cardiac anomalies

No cardiac abnormality was found in 171 (43.4%) of the 394 children examined. Cardiac anomalies had been diagnosed in 179 (45.4%) children of whom atrial ventricular septal defect (AVSD) was recorded in 52 (29%). Corrective cardiac surgical intervention was performed on 29 children (58%). 7 children (13.5%) were treated medically. One child had an inoperable heart condition, one child was awaiting surgery. Only 14 (27%) children with congenital heart disease had not required medical or surgical intervention.

Comparison of mean height of those with and without cardiac anomalies showed only marginal differences at different age groups and there were no consistent gender differences (mean SD scores boys -0.6, girls -0.3) (Figures 3.23 - 3.24 and Tables 3.95 and 3.96).

Figure 3.23 Comparison of mean height of boys with Down syndrome with and without cardiac anomalies

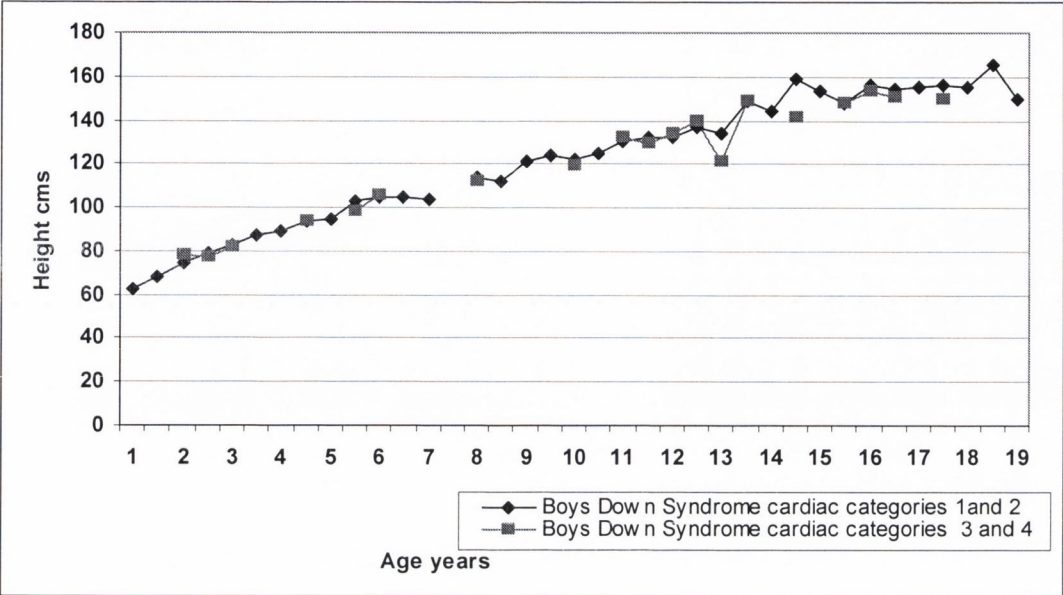
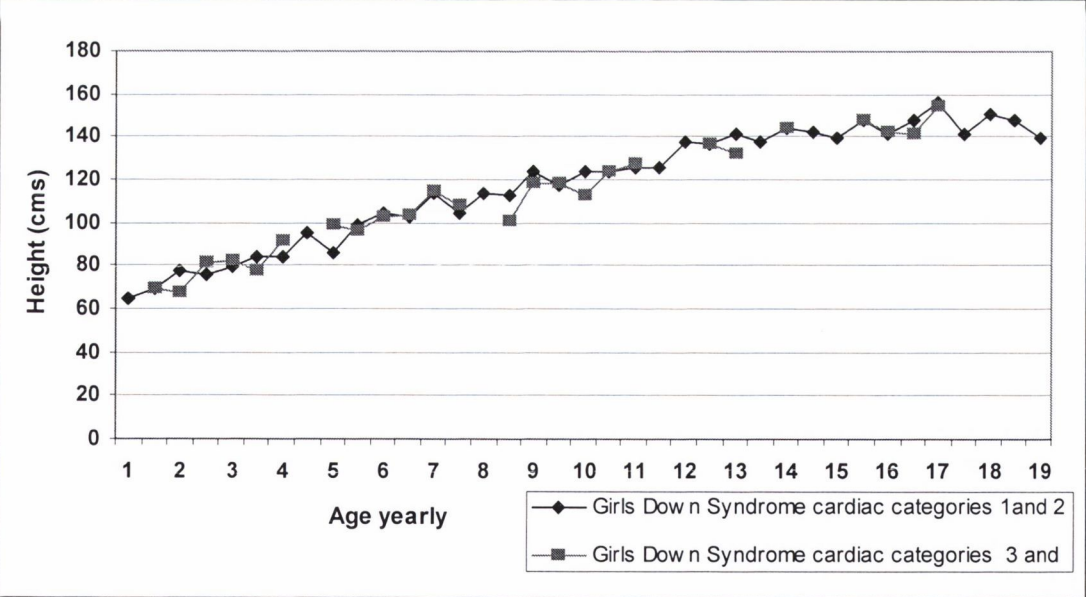


Figure 3.24 Comparison of mean height of girls with Down syndrome with and without cardiac anomalies



3.2.3.3 Thyroid dysfunction

Fifteen children with hypothyroidism were identified in this study (age range 9 months to 16 years). They were predominantly shorter for their age and sex but were within their peer height range. However, 2 children (girls) were significantly shorter than their

peers and had heights of -3.3 SD and -5.9 SD, respectively, below the general Irish population mean height (Figure 3.25-3.26).

Figure 3.25 Comparison of boys height with Down syndrome with and without thyroid dysfunction

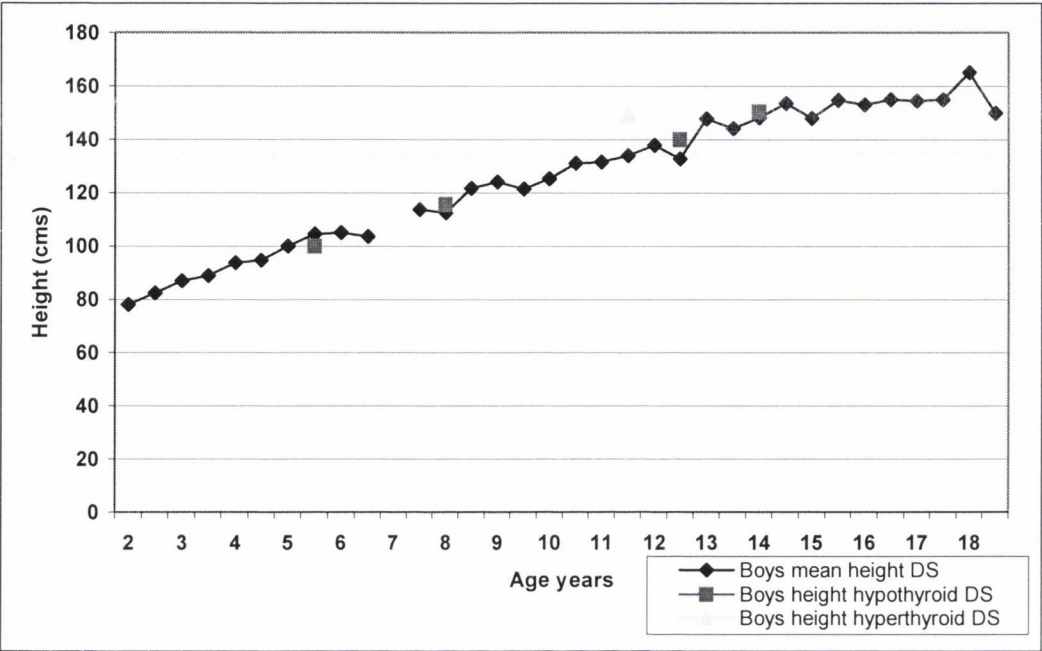
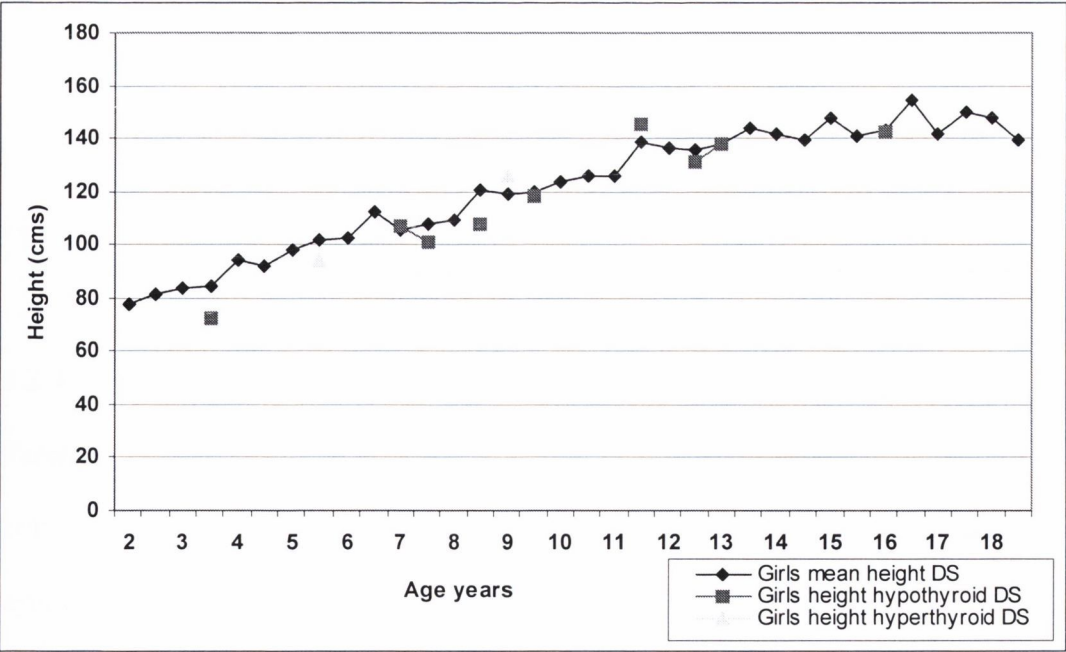


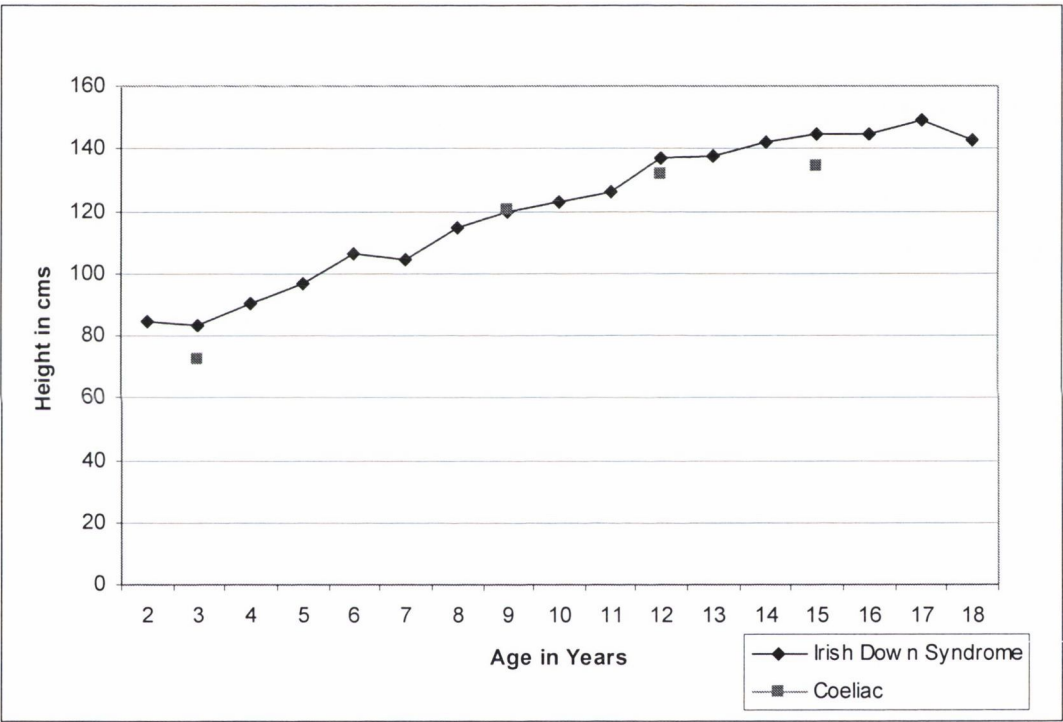
Figure 3.26 Comparison of girls height with Down syndrome with and without thyroid dysfunction



3.2.3.4 Coeliac Disease

Coeliac disease was identified in 4 children (age range 3 – 15 years). They were predominantly shorter than their peers and had a mean height -3.7 SD scores (range -1.6 to -5.9) below the general Irish population mean. One child with severely reduced height (-5.9 SD scores) also had hypothyroidism and Type 1 Diabetes (Figure 3.27).

Figure 3.27 Comparison of girls’ height with Down syndrome with and without Coeliac Disease



Cases at age 3 and 12.5 years also had hypothyroidism

3.2.4 Menarche

Parent(s)/guardian(s) reported that girls commenced menarche (first menstrual period) between 9 and 15 years of age. The mean age of menarche of girls with Down syndrome was determined based on the proportion of those menstruating or not, using probit analysis. The mean age of menarche was 12.6 years. The χ^2 test of goodness to

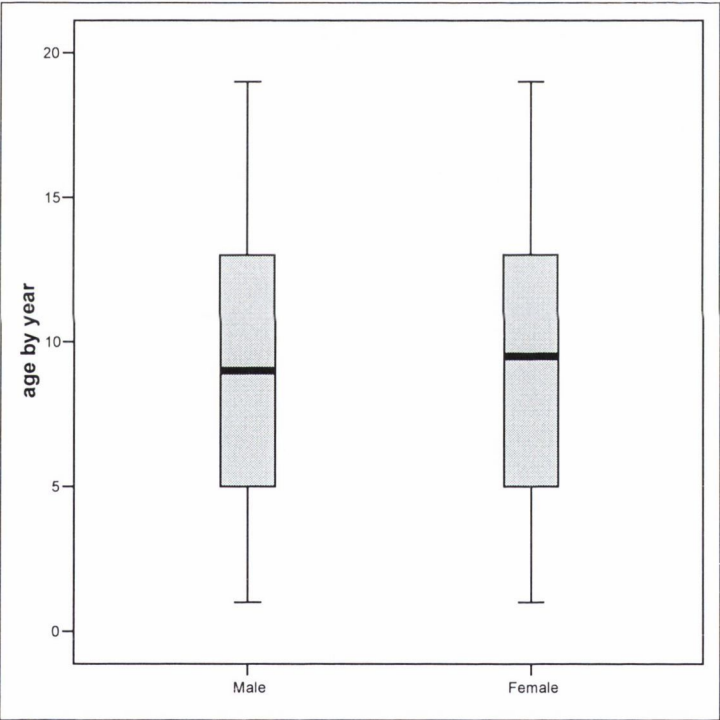
fit of data to the underlying model of the probit curve gave the degrees of freedom (df) of 168 so that the fit of the model was good with a p value = 0.000. The SD was 0.3 giving a 95% C.I. of 12.02 to 13.18 years, which is slightly earlier than the average age of menarche (13.52 years; 95% C.I. 11.0 to 15.0 years) in the general Irish population (Hoey 1986).

3.3 CONGENITAL HEART DISEASE

3.3.1 Cardiac diseases

Congenital heart disease had been diagnosed in 45% of children and was similar in boys (48%) and girls (52%). (Figure 3.24)

Figure 3.28 Cardiac disease in boys and girls by age



Atrio ventricular septal defect (AVSD) either alone or in combination with other heart lesions was found in 29% of those with cardiac anomalies. Atrial septal defect (ASD), ventricular septal defect (VSD) and patent ductus arteriosus (PDA) were the

next most common defects, many of which were self-correcting and did not require medical or surgical intervention. (Table 3.96(a)).

Table 3.96 (a) Congenital heart disease			
Diagnosis	Male N=86	Female N=93	Total N=179 (45%)
AVSD	20	25	45 (25.1)
ASD	19	18	37 (20.7)
VSD	16	16	32 (17.9)
PDA	11	12	23 (12.8)
ASD VSD	6	8	14 (7.8)
AVSD PDA	0	5	5 (2.8)
VSD PDA	5	0	5 (2.8)
ASD PDA	2	2	4 (2.2)
ASD VSD PDA	1	2	3 (1.7)
VSD Eisenmenger syndrome	2	0	2 (1.1)
AVSD Eisenmenger syndrome	1	1	2 (1.1)
ASD VSD Eisenmenger syndrome	0	1	1 (0.6)
ASD VSD Tetralogy of Fallots	1	0	1 (0.6)
ASD PDA Eisenmenger Syndrome	1	0	1 (0.6)
Bradycardia	0	1	1 (0.6)
Cardiomegaly	0	1	1 (0.6)
Epstein’s anomaly	1	0	1 (0.6)
Reversed Heart Chambers	1	0	1 (0.6)

No cardiac anomaly was found in 215 (55%) children, 43 (11%) of whom had an innocent heart murmur with normal cardiac investigations.

3.3.2 Diagnostic tests

Two hundred and eight parents responded to the question relating to cardiac screening tests performed on their child in order to diagnose heart disease. (Table 3.95) Electrocardiograph (ECG) together with echocardiography (ECHO) were the most common tests performed (37.5%), followed by the combination of ECG, ECHO and cardiac catheterisation in 24%. All reports of cardiac disease were crosschecked with a review of hospital records and also with Eurocat findings. (Table 3.97)

Table 3.97 Screening Tests		
Tests performed	N =208	53%
ECG and Echo	79	38.0
ECG Echo and Cardiac Catheterisation	50	24.0
ECG	38	18.3
ECHO	20	9.6
ECG, Echo and Chest X ray	4	1.9
ECG and Cardiac Catheterisation	4	1.9
Unable to recall tests	13	6.3

3.3.3 Age at Diagnosis

The majority (77.7%) of children had their heart disorder diagnosed at birth with a further 7% diagnosed within the first six months of life whilst in 18 children (9%) were diagnosed after the first six months of life (age range 6 months to 14.5 years). (Table 3.98)

Table 3.98 Age at diagnosis of heart disease				
Age diagnosed	Boys N=98	Girls N=101	Totals N = 199	51%
Birth	74	79	153	76.9
1-6 weeks	8	6	14	7.0
6 weeks - 6 months	9	4	13	6.5
6 months - 1 year	3	4	7	3.5
1-2 years	1	3	4	2.0
2-3 years	1	2	3	1.5
3-4 years	0	1	1	0.5
5-6 years	1	2	2	1.0
14.5 years	1	0	1	0.5

3.3.4 Treatment

Corrective cardiac surgery was performed on 59 children (33.3%), the most common operation was AVSD repair (48%), PDA surgery (17%) and ASD together with VSD repair (10%). Two parents reported that their children had inoperable heart conditions. One parent reported that her child was scheduled for cardiac surgery later that year. (Table 3.99)

Table 3.99 Cardiac operations		
Cardiac disease	N =59	15%
AVSD	28	48
PDA	10	17
ASD VSD	6	10
ASD VSD PDA	3	5
AVSD PDA	3	5
ASD	2	3
VSD	2	3
VSD PDA	2	3
ASD PDA	1	2
ASD VSD Tetrolgy of Fallotts	1	2
AVSD Eisenmenger syndrome	1	2

One child with a patent ductus arteriosus and 5 children with heart murmurs required cardiac medication in early infancy only. The remaining 27 children have required medical treatment throughout their lives. (Table 3.100)

Table 3.100 Cardiac Medical Treatment		
Diagnosis	N =33	8%
AVSD	6	18
ASD	6	18
VSD	7	21
PDA	1	3
ASD VSD	3	9
AVSD PDA	1	3
VSD PDA	1	3
VSD Eisenmenger syndrome	1	3
AVSD Eisenmenger syndrome	1	3
ASD VSD pulmonary hypertension	1	3
Heart murmur	5	15

3.4 THYROID DYSFUNCTION

Three hundred and five (77.4%) children (165 boys, 138 girls) were eligible to be screened by finger prick TSH blood sampling. The mean age for boys was 8.7 years (range 7 months - 18.9years) and the mean age for girls was 9.4 years (range 4 months - 18.6 years). One (0.2%) child had a venous thyroid function test, 4 (1%) declined. Eighty-five (21.4%) children (47 boys, 38 girls) were not eligible for finger prick screening as they had already had thyroid function tests performed during the one year prior to commencement of the study by either their family doctor or paediatrician and a further blood test was therefore unwarranted. Of this group the mean age for boys was 7.3 years (range 3 months - 16.4 years) and the mean age for girls was 8.1 years (range 1 - 16 years). Parents reported that 197 (50%) children (range 1 to 17 years) had not been screened since the newborn period. The data of thyroid dysfunction is presented in Tables 3.101 and 3.102 (pages 124 and 125).

3.4.1 Newly diagnosed Hypothyroidism

Hypothyroidism was newly diagnosed in 5 of 305 children (all female) mean age 9.3 years (range 7.7 to 16.1 years) using capillary whole blood TSH. During the study period hypothyroidism (compensated) was diagnosed in one other child (case 1) age 7.7 years, by venous sample when seen in the Accident and Emergency Department with abdominal pain and a history of constipation, making a total of 6 new cases with hypothyroidism. Whole blood TSH values from finger prick blood samples ranged from 13 to 297 mU/l (Table 3.101). At diagnosis 1 child had compensated and 4 had uncompensated hypothyroidism. The capillary whole blood TSH levels were

subsequently confirmed by venipuncture sample, except in one child on whom the report is not available, and correlated well with finger prick samples. Five children proved positive for thyroid microsomal or TPO antibodies and one child was not tested at her mother's request (Table 3.101).

Three children (cases 2, 3 and 5) had no clinical signs or symptoms of hypothyroidism, however on examining previous records case 5 had a TSH level of a 6.2mU/l eight months earlier. Three children had clinical signs and symptoms but they had previously been attributed to DS itself; cases 1 presented at the casualty department with abdominal pain and constipation; case 4 had increased weight gain and was lethargic for a period of 18 months; case 6 had a history of fatigue. The children had an overall mean height -2.4 SD scores below the general Irish population (Hoey 1987) but were within 2SD of the mean height on the DS specific growth charts (Cronk 1988).

All six children (cases 1 to 6) were commenced on Thyroxine and followed up at regular outpatient clinics with substantial improvement in growth and development. In particular, the pre-pubertal child (case 3) with symptoms of weight gain and fatigue showed improvement in; word recognition, speech development, walking and swimming. In addition her weight reduced to within normal range and her height increased by 9.6 cms within one year

3.4.2 Previously diagnosed Hypothyroidism

Ten children (4 males, 6 females) (mean age at diagnosis 7.1 years, range 8 months to 15.5 years) had previously been diagnosed with hypothyroidism by their paediatrician or family doctor.

Age groups	Hypothyroid	Hyperthyroid
< 5 years	4/119 (3.4%)	1 (0.6%)
5-10 years	3/106 (2.8%)	0
10-15 years	2/120 (1.6%)	1 (0.8%)
> 15 years	1/53 (1.9%)	0

There were 69 children under the age of 3 years of whom 4 (7.9%) had been diagnosed with hypothyroidism. Of the 9 cases with available blood results at the time of their diagnosis, 2 had compensated and 7 had uncompensated hypothyroidism. Only 3 children presented with clinical features of hypothyroidism at the time of diagnosis. One child presented with mild fatigue; two children presented with constipation, one of whom also had poor growth for more than 6 months. Of the remaining 7 children, 3 children had also been diagnosed with Type I diabetes

Six children had positive antibodies, five of whom had markedly raised plasma TSH values (23.7 - 87.7 mU/l) at diagnosis. Case 7 (male) had a marginally raised plasma TSH 8.1 mU/l, a plasma T4 within the reference range and negative thyroid antibodies and was not treated but has been observed closely for two years. Case 1 (male) with a recorded raised plasma TSH (8.2 mU/l) and a normal plasma T4 level and negative thyroid antibodies at 8 months of age developed uncompensated hypothyroidism within 7 months and was subsequently commenced on Thyroxine. All other patients were receiving Thyroxine with good clinical effect (Table 3.102).

3.4.4 Previously diagnosed Hyperthyroidism

Two children had previously been diagnosed with hyperthyroidism by their family doctor and paediatrician respectively and were on treatment with good clinical effect. Case 11 (female) presented with exophthalmos and proptosis prior to cardiac surgery and had a plasma total T4 value of 285 nmol/L and TSH <0.2 mU/l on venous sampling. Case 12 (male) with no clinical signs had been diagnosed following routine thyroid function test by his paediatrician.

This study further demonstrated that whole blood TSH screening for hypothyroidism by finger prick sampling is feasible in school age children and particularly the preschool child as parents reported that it was a more acceptable, rapid and non invasive test than venous sampling. In addition only 3/ 305 children screened became upset during the procedure. Parents also found it a more acceptable, rapid and non-invasive test than venous sampling (Table 3.102- page 126)

Table 3.101 Newly Diagnosed Thyroid Dysfunction and Clinical Features

Case Number	Age (Yrs)	Sex	Capillary Whole blood TSH mU/l	Plasma TSH mU/l	Plasma T4 nmol/l	Thyroid Anti bodies	Clinical Features	Height	
								Centiles DS **	SD Scores General Irish Population***
1*	7.7	F	-	18.5	104 (69-141) [†]	Not tested	Constipation	9 th – 25 th	-2.1
2	8.3	F	13	16.3	67 (70-140) [†]	+	None	<9 th	-3.3
3	8.9	F	14 mU/l	-	-	+	None	75 th –91 st	-0.7
4	9.5	F	297	586.7	<5.1 (8-23) ^{††}	+	Weight gain Fatigue	9 th – 25 th	-2.3
5	12.4	F	30	30.0	73 (70-140) [†]	+	None	9 th – 25 th	-2.7
6	16.1	F	81	80.3	<13(70-140) [†]	+	Fatigue	50 th – 75 th	-2.5

* Screened by venous sample

** Styles 2001

***Hoey 1987

† Total T4 nmol/l

†† Free T4 pmol/l

Table 3.102 Previously Diagnosed Thyroid Dysfunction and Presenting Clinical Features and Age of Diagnosis

HYPOTHYROIDISM						
Case Number	Age (yrs)	Sex	Plasma TSH mU/l	Plasma Total T4 nmol/l	Thyroid Antibodies	Clinical Features
1	0.8	M	8.2**	133 (70-140)**	-	None
2	0.9	M	9.5	79 (70-150)	-	None
3	1.4	F*	63.5	57 (70-140)	+	None
4	2.1	M	4.4***	61 (70-180)	-	Constipation poor growth
5	5.5	F	7.1	85 (101-170)	+	Constipation
6	7.1	F	23.9	63 (70-140)	+	None
7	8.1	M	8.1	89 (65-160) †	-	None
8	10.2	F*	62.7	22 (70-140)	+	None
9	11.3	F*	62.7	22 (70-180)	+	None
10	15.5	F	87.7	47 (70-140)	+	Fatigue

*Children with Type 1 Diabetes
 ** Blood vaues 7 months prior to diagnosis of uncompensated hypothyroidism actual values on diagnosis not available
 ***TRH tes positive hypothyroidism TSH post TRH 26.3 mU/l

HYPERTHYROIDISM						
Case Number	Age (yrs)	Sex	TSH mU/L	Plasma T4	Thyroid Antibodies	Clinical Features
11	1.9	F	<0.2	285.0(70-180) †	+	Exophthalmos & Proptosis
12	9.6	M	<0.05	40.3 (13-23) ††	+	None

† Total T4 imc/l
 †† Free T4pmol/l

3.5 VISION DISORDERS

Ophthalmic problems were found in 42% of children with Down’s syndrome. The most common problems being refractive errors in 25%, strabismus in 18%, cataract in 4%, nystagmus in 16% and glaucoma in 0.5% of the children.

3.5.1 Parents concerns

The parents were asked “had they concerns about their child’s vision?” and was there “any family history of visual problems before the age of 5 years?” Parents indicated that 52% had concerns relating to their child’s vision (Table 3.103).

Table 3.103 Parent concerns relating to their child’s vision		
Concerned	N =394	100%
Yes	206	52
No	168	43
Missing	20	5

Parents reported that 17% of children had a family history of visual problems before the age of 5 years (Table 3.104).

Table 3.104 Family history of vision problems < 5 years of age		
Family history	N = 394	100%
Yes	68	17.3
No	294	74.6
Not answered	32	8.1

3.5.2 Vision screening

Vision screening had been performed on 77% of children however 21% reported that

their child had not yet been screened (Table 3.105).

Table 3.105 Child’s vision tested		
Tested	N = 394	100%
Yes	302	77
No	81	21
Don't Remember	6	2
Not answered	5	1

The majority of children had vision screening performed by an eye specialist (65%), 18% by a paediatrician at the special developmental services, 15% in the health clinic and by the doctor in school 1% (Table 3.106).

Table 3.106 Who tested child’s vision		
Tested by whom	N =287	73%
Eye Specialist	189	65
Paediatrician Special Developmental Service	52	18
Health Clinic EHB	43	15
Doctor in School	3	1

Parents reported that the first vision screening assessment was performed in the first year of life in only 18%, from 1 to 2 years in 29%, from 2 to 4 years in 24%, from 4

to 6 years in 11% and 18% were screened for the first time after the age of six years (Table 3.107).

Table 3.107 Age of First Test of Vision		
Age tested (year)	N =257	65%
Birth to 1	46	18
1-2	74	29
2-4	61	24
4-6	28	11
6-8	21	8
8-12	19	7
12-16	8	3

Parents were asked how often their child’s vision was screened, if they wore corrective glasses and whether they wore their glasses all day. Parents reported that 30% were screened yearly, 20% six monthly, 3% 3 monthly and 3% every 2 years. Screening on one occasion only was reported in 16%, 2 - 4 times in 9% and more than 4 times in 1% (Table 3.108). In addition they also confirmed that 39.3% wore corrective glasses with 19.5% wearing them all day.

Table 3.108 How often vision screened		
How often	N =312	79%
Yearly	94	30
6 Monthly	63	20
3 Monthly	10	3
2 Yearly	10	3
Once only	52	16
2-4 times	28	9
> 4 times	4	1
Could not remember	51	16

3.5.3 Clinical history

Parents were asked if they could remember the results of the vision screening tests for both right and left eyes. The most common disorders reported by parents included long and short sightedness, strabismus, nystagmus, cataract and the absence of vision in one eye was reported in two children (Tables 3.109)

Table 3.109 Eye problems history from parents				
Response	Right Eye		Left Eye	
	N =	%	N=	%
Normal vision	41	10.4	39	9.9
Strabismus	28	7.1	28	7.1
Short sighted	32	8.1	40	10.2
Long sighted	27	6.9	31	7.9
Cataract	6	1.5	7	1.8
Short sighted + astigmatism	5	1.3	5	1.3
Short sighted + cataract	0		3	0.8
Astigmatism	3	0.8	3	0.8
Nystagmus	3	0.8	2	0.5
Strabismus + cataract	2	0.5	0	
Fairly good vision	2	0.5	0	
Long sighted + strabismus	2	0.5	2	0.5
Short sighted + strabismus	2	0.5	3	0.8
Strabismus + Nystagmus	2	0.5	1	0.3
No vision	1	0.3	2	0.5
Strabismus + astigmatism	1	0.3	1	0.3
Cataract + glaucoma	1	0.3	1	0.3
Short sighted + cataract	1	0.3	3	0.8
Short sighted + strabismus + cataract	1	0.3	0	
Short sighted + astigmatism + cataract	0		1	0.3
Fairly good vision	0		1	0.3
Poor vision	1	0.3	0	
Poor vision + cataract	0		1	0.3
Long sighted + cataract	1	0.3	0	
No records	155	39.3	146	37.1
Not screened	77	19.5	77	19.5

Comparison of vision results bilaterally provided a clearer picture of refractive errors indicating that generally children had bilateral eye problems and that the occurrence

of several anomalies together was common (Table 3.110).

Table 3.110 Refractive errors					
Refractive errors	Right eye N=75	Left eye N=90	Bilateral N=68	Total 97	%
Short sight	32	40	28	44	45
Short sight + strabismus	2	3	2	3	3
Long sight	27	31	27	31	31
Long sight + strabismus	2	2	1	3	3
Astigmatism	3	3	2	4	4
Short sight + astigmatism	5	5	4	6	6
Strabismus + astigmatism	1	1	1	1	1
Short sight + cataract	1	3	1	3	3
Short sight + strabismus + cataract	1	1	1	1	1
Long sight + cataract	1	1	1	1	1

3.5.4 Treatment

Parents reported that 155 (39%) children wore corrective glasses and that 77 (20%) children wore spectacles all day.

The most common eye operations performed was for strabismus (4%). removal of cataract (4%), and to clear blocked tear ducts (3%) (Table 3.111).

Table 3.111 Eye surgery (n = 46)		
Eyes operations	No	%
Strabismus	17	4
Cataract	16	4
Blocked tear ducts	11	3
Blocked tear ducts and strabismus	2	1

3.5.5 Clinical examination

On clinical examination it was recorded that strabismus was present in 70 children (57 bilateral), cataract was noted in 9 children (6 bilateral), nystagmus was present in 66 children bilaterally, two children had blocked tear ducts and two children had eye infections at time of examination (Table 3.112).

Table 3.112 Paediatric eye examination results					
Eye Problems	Bilateral N=132	Right eye N=137	Left eye N=144	Total N=150	%
Strabismus	57	61	66	70	46.7
Cataract	6	7	8	9	6.0
Nystagmus	66	66	66	66	44.0
Blocked tear ducts	1	2	2	3	2.0
Eye infections	2	1	2	2	1.3

Statistical analysis was performed to assess the association of heart defects with the occurrence of nystagmus and showed significant correlation ($p=0.027$, X^2 test). (40 boys and 26 girls) mean age 11 years (range (2 – 19 years)).

3.6 Hearing Screening

Hearing screening was performed in all 394 children participating in the study using three screening tools (otoscopy, tympanometry and distortion product oto acoustic emissions (DPOAE)) as detailed in methodology (first study). In addition 20 children with Down syndrome and an age matched control group of 20 without Down syndrome were assessed using four screening tools (otoscopy, tympanometry, DPOAE and Pure Tone Audiometry) as detailed in methodology (second study).

3.6.1 First Study of Hearing Screening

The full hearing screening protocol took on average 5 to 7 minutes per child. Some difficulties did exist, however, particularly with regard to explaining and demonstrating the procedures, as some children were quite frightened of the new technology and it was worthwhile spending time to gain their cooperation. Even so there were a number of children who could not tolerate their ears being touched on the day of the study and these have been identified at each screening procedure. Thus the number of ears screened was as follows: - Otoscopy 97% (762 ears); Tympanometry 93% (735 ears) DPOAE 69% (544 ears)

3.6.1.1 Otoscopy

Bilateral otoscopy was attempted on all patients except for 13 children who did not cooperate. The age range of those who did not cooperate was 3.2 –15.8 years. One of

these children had severe intellectual disability and one other child had severe physical disability (Figure 29).

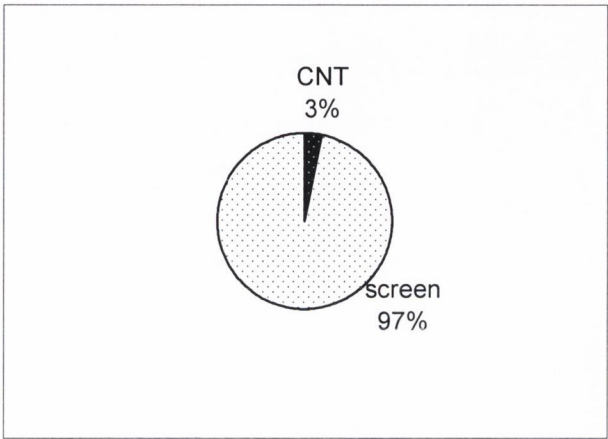


Figure 3.29 Otoscopic Screening

Otoscopy was performed on 381 patients (97% of cohort, 762 ears) by one Paediatrician (MP). Abnormalities were detected in 64% (491 ears) and included mild to severe cerumen (wax); perforated tympanic membrane and very small ear canals. Tympanometry tubes (grommets) were seen in situ in 32 ears and lying in the ear canals in 2 ears.

3.6.1.2 Tympanometry

Tympanometry was performed on 363 children (93% of cohort, 726 ears) while 31 children were not screened, 26 of whom were unable to cooperate with the procedure,

and 5 children had a perforated tympanic membrane (tm). (Figure 30)

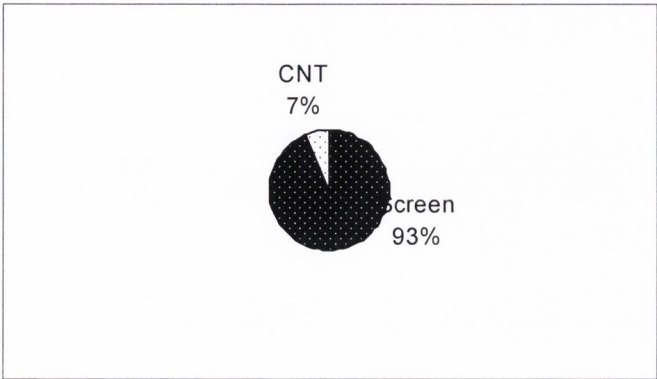


Figure 3.30 Tympanometry Screening

Normal middle ear function was found in 42% of children (309 ears) whilst 58% (426 ears) had middle ear abnormality (Table 106). Abnormalities detected included glue ear, eustacian tube dysfunction, ossicular discontinuity (in one ear) and possible increased stiffness of middle ear structures. The classification of tympanometry findings are detailed using Jerger Classification (1970) (Table 2.2) (Page 245). This classification outlines 6 classes (A (normal); As and Ad (mild abnormalities); and B, C and Cs (moderate to severe middle ear abnormalities)).

Table 3.113 Results of classified tympanograms in children with DS

Tympanograms Type	Number of ears	%
A	309	42.0
Ad	1	0.1
As	60	8.2
B	218	29.7
C	115	15.6
Cs	32	4.4

Of the 309 (42%) ears that passed on tympanometry, 169 (54.7%) had failed on otoscopy and required referral for removal of wax. Also, of the 426 ears that had failed on tympanometry 301 (70.7%) had also failed on otoscopy (Figures 31 and 32)

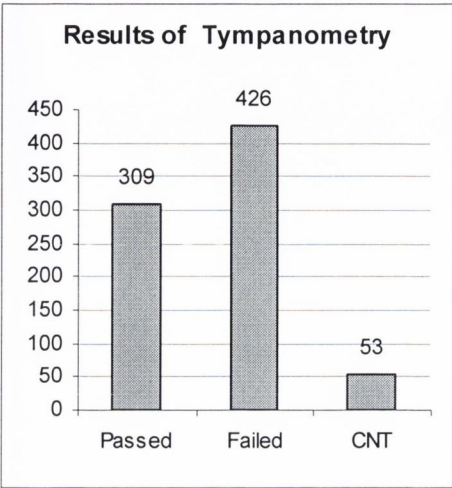


Figure 3.31 Tympanometry results

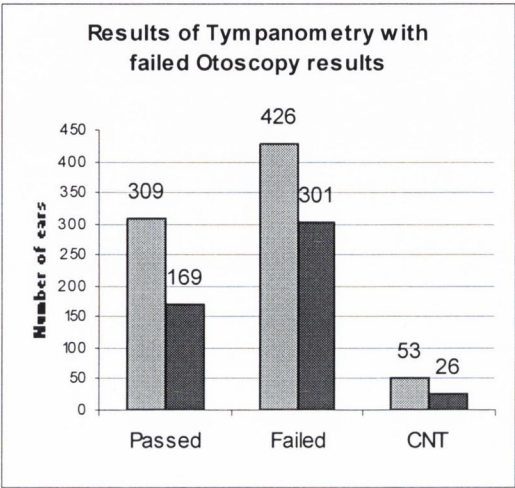


Figure 3.32 Tympanometry/Otoscopy results

Parents reported that the children were prescribed hearing aids from the age of 2 years of age. Reports by parents also indicated that while 15.9% of the children had been prescribed hearing aids, compliance with wearing them was poor as only 6 children (8 hearing aids 1%) were wearing them at the time of the study.

3.5.1.3 Distortion Product Otoacoustic Emissions

In order to assess the percentage of children with DS who could tolerate screening by DPOAE all cooperative children were screened regardless of whether they had failed otoscopy examination and/or tympanometry. DPOAE Test 1 (65/55 dB SPL) and

Test 2 (65/50 db SPL) were attempted on all patients and performed on 544 ears (278 children {266 bilaterally and 12 unilaterally}) (69% children in the study) successfully (figure 33).

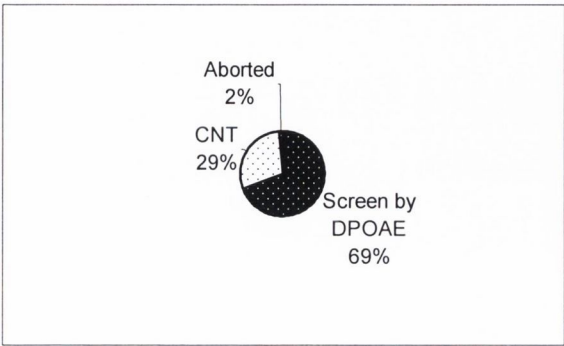


Figure 3.33 DPOAE Screening

232 ears (29% children in the study) were not screened by DPOAE, as 116 children were unable to cooperate for screening by this procedure on the day of the study. This was due to the fact that the children could not tolerate the probe being put in their ears and sit quietly at that time. A further 12 ears (2%) had the screening aborted as the children were unable to sit quietly, or could not stop chatting even for the short period required for this test. (12 (2%) ears aborted {12 children unilaterally} + 232 (29%) ears could not be tested (cnt) {116 children bilaterally} + 544 (69%) ears screened (287 children} = 788 ears).

Technically acceptable emissions were produced in 77% of ears tested (421 ears {258 children {156 bilateral and 95 unilateral}}). Normal inner ear function was found in only 10.7% (58 ears {50 children; 8 bilateral and 41 unilateral}) (passed 3 octaves). And 89% (486 ears) were rated ‘fail’. Of these 29% (158 ears {140 children; 18 bilateral and 122 unilateral}) passed 2 octaves; 37.6% (205 ears (187 children; 16

bilateral and 173 unilateral}) passed 1 octave and 22.6% (123 ears) failed to pass any octave. Review of the failed DPOAE results (123 ears) showed that only 10.5% (13 ears) had normal external and middle ear function. Thus suggesting a possible mixed and or SNHL requiring referral for further investigations. Results illustrated in (Table 3.114).

Table 3.114 Results of Otoscopy Tympanometry and DPOAE in children with DS

	Normal (N=Children)		Abnormal (N=Children)
Otoscopy	35.6% (154 {117*/37 **})		64.4% (254 {227 */37**})
Tympanometry	42% (204 {105*/99**})		58% (258{159* 99**})
DPOAE	10.7%	3 octaves (Pass)	(50 children)
	29.0 %	2 octaves (Review)	(140 children)
	37.6%	1 octave (Review)	(189 children)
	22.6%†	Fail	(123 children)

*Bilateral/ ** Unilateral
†Only 10.5% (13 children) had normal external and middle ear function thus suggesting a possible mixed and/or SNHL thus requiring referral for further investigations

3.5.1.4 DPOAE test 1 versus test 2

Comparison of DPOAE Test 1 (65/55 dB SPL) and Test 2 (65/50 dB SPL) responses showed that Test 1 rated higher overall in acceptable responses and level of amplitude.

3.5.1.5 Clinical History reported by parents

Parents completed a detailed questionnaire that included past clinical history of hearing impairment. They reported that 94.4% of children had been tested for hearing function, however the timing of their first hearing test ranged from 3 months to 14

years, with a median age of 1.5 years. Where children were first screened varied considerably as can be seen from (Table 3.115). 11.2% of parents could not remember and held no record of when their child had been first tested.

Table No 3.115 Where hearing screening first performed

Where tested	Numbers	%
Eye and Ear Hospital	4	1.02
Med. Educational Centre	17	4.31
ENT Consultant	23	5.8
General Practitioner	2	0.51
Local Hospital	18	4.6
National Rehabilitation Board	36	9.1
Paediatrician	2	0.51
Local Health Centre	39	9.9
School	4	1.02
School for deaf	3	0.76
Sr. Lydia	199	50.49
Information not available	44	11.2
Copenhagen	1	0.25
London	1	0.25
Sweden	1	0.25
Total	394	100

Parents reported that 94% children had been tested for hearing function. Their first hearing-screening test was performed at the median age of 1.5 years (range three months to 14.5 years). Parents also reported that 15% of children had been diagnosed with normal hearing and 26% reported that their child had impaired hearing. However, 58% of parents could not remember nor did they hold any record of their child's hearing results. The distribution of reported hearing impairment in each age

group illustrated a high percentage of children with mild hearing loss between 2 to 6 years of age and between 10 to 12 years of age.

Parents also reported that 156 (39.6%) children had grommets surgically inserted bilaterally in 141 children. The number of times children had this operation undertaken varied from a single operation to 9 sets of grommets being inserted in one child. Age at surgery ranged from 6 months to 9 years with a median age of 2.8 years. Hearing aids had been fitted in 49 children unilaterally and 14 bilaterally. Parents reported very poor compliance with wearing hearing aids.

3.5.2. Second Study of Hearing Screening

In order to compare DPOAE and PTA screening results in children with DS and those in the general paediatric population this aspect of the study looked at a group of 20 children with Down syndrome (Group A) and an age matched control group without Down syndrome (Group B) using four screening tools targeting different areas of the auditory axis as follows:- Otoscopy; Tympanometry; Distortion Product Oto Acoustic Emissions (DPOAE) and Pure Tone Audiometry (as detailed in methods)

3.5.2.1 Population

Group A:- 20 children with Down syndrome participated (mean age 12.3 years; range 8 to 16 years male/female ratio 1:1). Five children in each age (8 – 16 years) group were randomly selected. This was undertaken using the random selection facility on the SPSS computer software package from the original large sample of 202 children with Down syndrome in the study. Nine children could not participate because of

illness and 1 child was on holiday. All children were screened in the same sequence using the four screening tools as follows: - otoscopy, tympanometry, DPOAE and then Pure Tone Audiometry. Screening was undertaken at the time and place that best suited the patient.

Group B:- 20 age matched healthy children without Down syndrome were recruited from children of the hospital staff and staff neighbours (mean age 12.10 years; range 8 to 16 years; male/female ratio 2:3) (Table 3.116). Due to the time constraints of this section of the study no attempt was made to match by cognitive function/ability levels. This is a region that would benefit from further study in the future.

Table No 3.116 Gender and age distribution both Groups				
	Group A With Down Syndrome		Group B Without Down Syndrome	
Age (yr)	N	Male/Female	N	Male/Female
8	2	2/0	2	1/1
9	2	1/1	2	1/1
10	2	1/1	2	1/1
11	2	1/1	2	0/2
12	2	2/0	3	1/2
13	3	0/3	2	1/1
14	2	1/1	3	3/0
15	2	0/2	2	0/2
16	3	2/1	2	0/2
Total	20	10/10	20	8/12

3.5.2.2 Otoscopy

Otoscopy was performed on all 40 children. The results were analysed by children and number of ears as abnormalities can affect one or both ears in the same child (Tables 3.117 and 3.118) (pages 148 and 149).

In those with Down syndrome (Group A) 9 children passed bilaterally and 2 unilaterally while 9 children failed bilaterally and 2 unilaterally. This differed greatly with the results of the controls (group B) where only 1 child failed unilaterally. It was interesting to note that the one child in Group B who failed on otoscopy had sustained a perforated ear drum when diving at swimming two days earlier.

There was a statistical difference between the two groups which demonstrates that children with Down syndrome are more likely to have an abnormality on otoscopic examination than the general population children many of which are easily treated.
($\chi^2 = 19.94$; $df=1$; $P < 0.001$)

3.5.2.3 Tympanometry

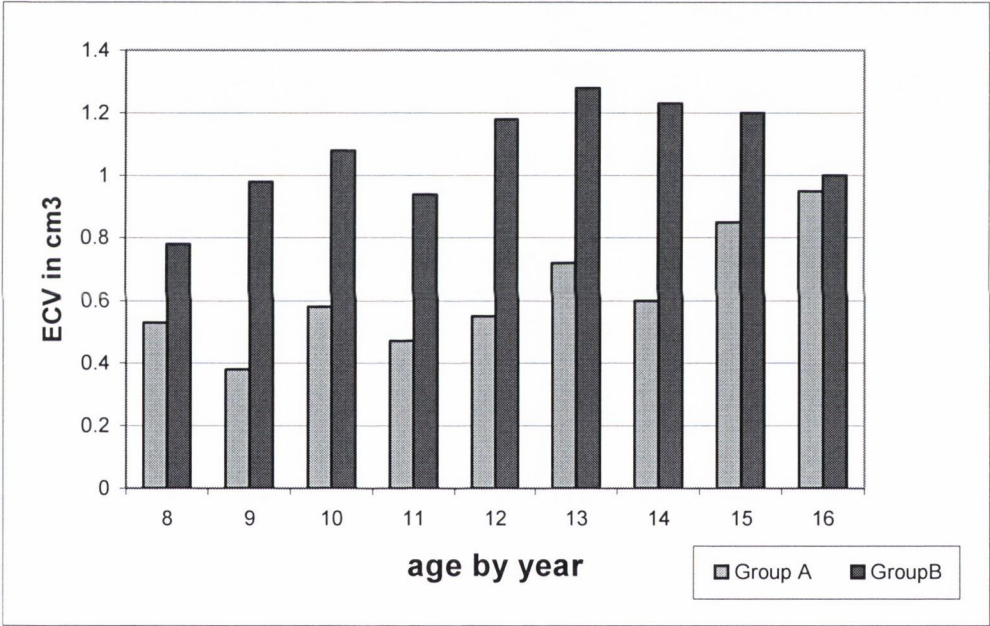
Tympanometry was performed on all of children in groups A and B and the results were categorised as per Jerger classification. These results were classed as either a pass (type A) or fail (types As, B, C and Cs) for the purpose of analysis. (Tables No 3.117 and 3.118).

In those with Down syndrome (group A) 9 children passed bilaterally and 5 unilaterally, while 6 failed bilaterally and 5 unilaterally (Table No 3.117) (Page 148). This differed greatly with the results of the controls (group B) where only 2 children failed bilaterally and 1 unilaterally (Table 3.118) (Page 149). There was a significant difference between the two groups indicating that children with Down syndrome had a much higher incidence of middle ear dysfunction than the general population.
($\chi^2 = 8.58$; $df = 1$; $P < 0.003$)

Ear Canal Volume

Children with Down syndrome may have narrow ear canals. This study sought to compare ear canal volumes of children with Down syndrome with the general population. Ear canal volume can be measured by tympanometry. Normal ear canal volume ranges from 0.2 cm³ to 2.0 cm³ from childhood to adulthood. Ear canal volume was measured by tympanometry and was compared in both groups at each year of age (Figure 3.4 and Table 3.119).

Figure 3. 34 Comparison of ear canal volume by age in those with and without DS



In those with Down syndrome (Group A) ear canal measurement was performed on 19 children (37 ears), 3 ears (2 children) were excluded because of a grommet in situ in one child and the second child had perforated eardrums. Both conditions affect the ear canal volume.

In Group B ear canal measurement was performed on 20 children (39 ears), 1 child was excluded because of a perforated eardrum. Comparison of ear canal volume showed that the general population had larger ear canal volumes than the children with Down syndrome. However, this difference was less marked in the older child (Table 3.119).

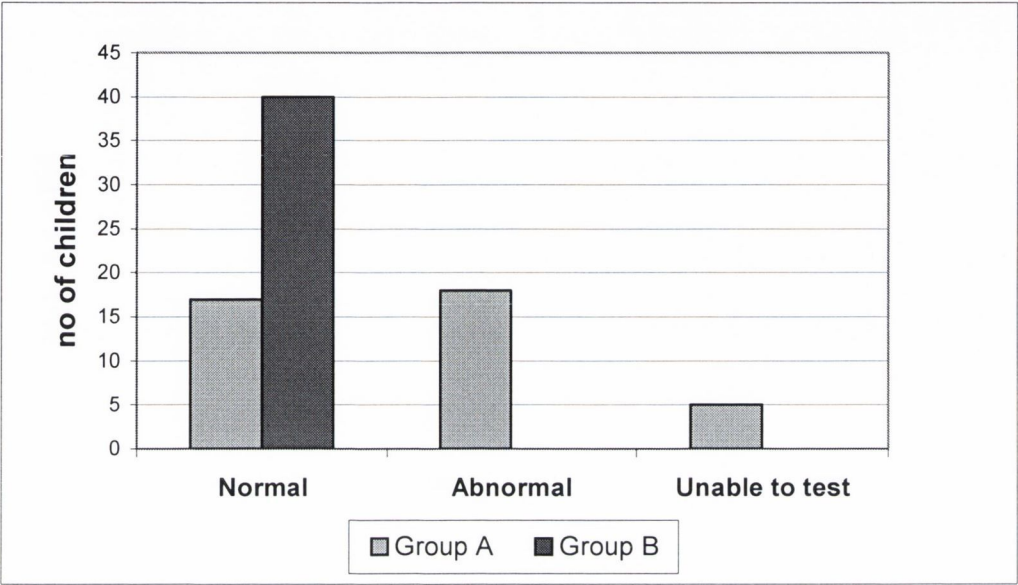
Table 3.119 Comparison of mean ear canal volume by age Down syndrome versus general population				
Age in yrs	Group A (With Down Syndrome)		Group B (Without Down Syndrome)	
	Children	cm ³	Children	cm ³
8	2	0.53	2	0.78
9	2	0.38	2	0.98
10	2	0.58	2	1.08
11	2	0.47	2	0.94
12	2	0.55	3	1.18
13	3	0.72	2	1.28
14	2	0.60	3	1.23
15	1	0.85	2	1.20
16	3	0.95	2	1.00

3.5.3.5 Pure Tone Audiometry (PTA)

Group A (those with Down syndrome) 17 children were tested in both ears and one child only one ear by PTA. Six children had normal hearing bilaterally and 5 children had impaired hearing bilaterally. Five children had normal hearing unilaterally and 6 children had hearing impairment unilaterally. Hearing impairment is graded as per

Lee Classification (Table 2.5) (Page 283). In comparison all those in the Group B had normal hearing on PTA screening (Table 3.118 and Figure 3.35)

Figure 3 35 Comparison of Pure Tone Audiometry in Down Syndrome versus control group

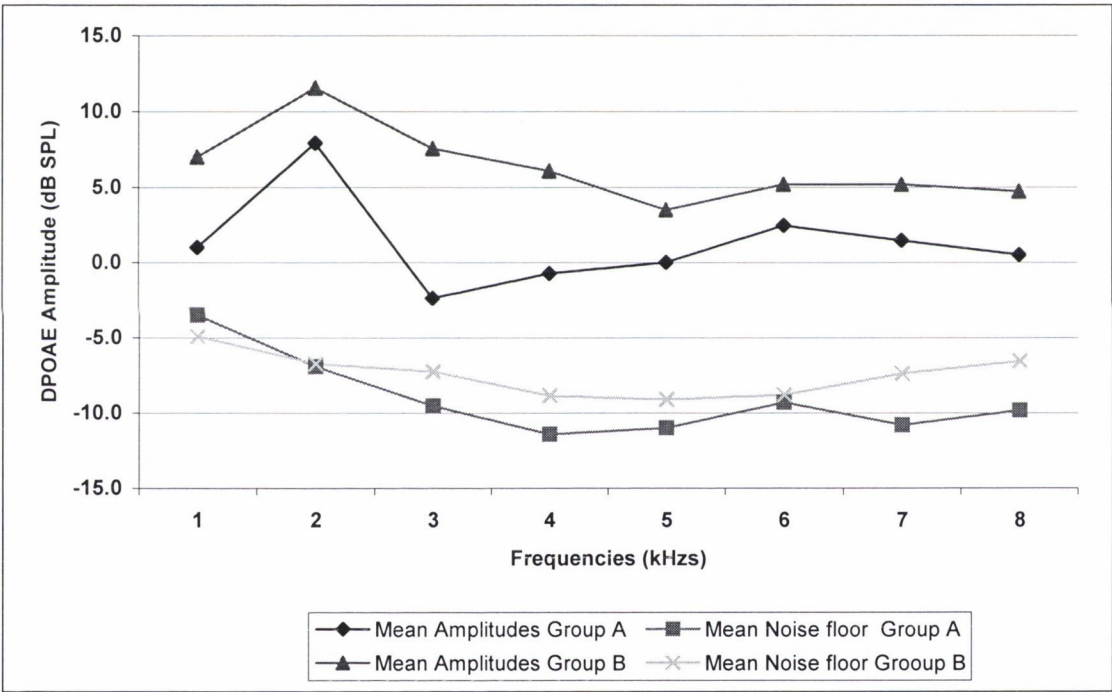


3.5.3.6 Distortion Product Oto Acoustic Emissions (DPOAE)

In Group A, 18 children were screened bilaterally by DPOAE, 2 children (4 ears) in could not be tested due to inability to cooperate. Normal inner ear function was assigned to those who passed 2 or 3 octaves while those passing less than 2 octaves were considered failed. Inner ear function was found to be normal in 5 children bilaterally and 8 children failed bilaterally. Inner ear function was found to be normal in 5 children unilaterally and 5 children failed unilaterally (Table 3.117 - page 148). In Group B, 20 children were screened bilaterally and all children had normal inner ear function bilaterally (Table 3.118 – page 149).

Comparison was made of the mean DPOAE amplitude responses from all children in both groups with normal outer and middle ear results. The mean amplitudes (± 1 SE) are plotted against frequencies (kHz 1-8) on an x y graph showing the mean amplitudes of both groups with the corresponding mean noise floor levels (natural body sound) which is measured automatically as part of the screening operation. (Figure 3.36)

Figure 3.36 Comparison of DPOAE Mean Amplitudes in Groups A and B



(1 kHz $p < .01$; 2 kHz $p < .05$; 3 kHz $p < .001$; 4 kHz p value $< .01$; 8 kHz $p < .05$)

As shown in Figure 35 the mean amplitudes were lower across all frequencies in children with Down syndrome and significantly so (when analysed by independent samples T test) at 1 to 4 kHz and also at 8 kHz in comparison to those in group B.

3.5.3.7 COMPARISON OF PURE TONE AUDIOMETRY (PTA) AND DPOAE

Statistical analysis measuring agreement between both PTA and DPOAE results when the children had normal outer and middle ear results showed perfect agreement (Kappa =1, n=27 children (50 ears)). However when all children were included, with normal and abnormal outer and middle ear results, correlation was weaker but still significant (Kappa: value .77, n=37 children (73 ears)). The DPOAE test outcomes when evaluated against the PTA hearing threshold in this group showed good correlation however further research would be required in larger numbers over time to find the true sensitivity and specificity of these tests.

Table 3.117 Hearing Screening Results (Ears) of Children with Down syndrome (Group A)

Case No	Age (yr)	Otoscopy		Tympanometry		Pure Tone Audiometry		DPOAE	
		Right	Left	Right	Left	Right	Left	Right	Left
1	8	-	-	B	B	could not test	could not test	could not test	could not test
2	8	-	-	A	A	could not test	could not test	2	1
3	9	-	-	B	A	mild	could not test	3	1
4	9	-	-	C	B	mild	normal	-	1
5	10	-	-	B	B	mild/mod	normal	1	1
6	10	+	+	B	C	mild	mild	1	1
7	11	-	-	B	A	mod	mild	2	-
8	11	+	+	A	A	normal	mild/mod	1	1
9	12	+	+	A	B	normal	mild	2	1
10	12	-	+	A	A	normal	normal	1	2
11	13	-	-	B	B	mod[†]	mod/severe[†]	could not test	could not test
12	13	+	+	A	A	normal	normal	2	3
13	13	+	+	A	A	normal	normal	3	3
14	14	+	+	A	A	normal	normal	3	2
15	14	+	+	A	A	normal	normal	3	2
16	15	-	-	B	B	mod	mod	-	-
17	15	+	+	A	A	normal	normal	3	3

Otoscopy: +/- Pass/Fail

Tympanometry: Jerger Classification (1970). [A (normal); As and Ad (mild abnormalities) and B, C and Cs (moderate to severe middle ear abnormalities)]

Pure Tone Audiometry (PTA): (Hearing Threshold Classifications Lee 1999)

DPOAE:- Pass /Refer (pass 2 and 3 octaves / Refer pass only one octave or less)

[†] Sensory Neural hearing loss (SNHL)

Table 3.118 Hearing Screening Results (Ears) of Children without Down Syndrome (Group B)

		Otoscopy		Tympanometry		Pure Tone Audiometry		DPOAE	
Case No	Age (yr)	Right	Left	Right	Left	Right	Left	Right	Left
1	8	+	+	A	A	normal	normal	3	3
2	8	+	+	C	C	normal	normal	3	3
3	9	+	+	A	A	normal	normal	3	3
4	9	+	+	A	A	normal	normal	3	3
5	10	+	+	A	A	normal	normal	3	3
6	10	+	+	A	A	normal	normal	3	3
7	11	+	+	A	A	normal	normal	3	3
8	11	+	+	A	A	normal	normal	3	3
9	12	+	+	C	C	normal	normal	3	2
10	12	+	+	A	A	normal	normal	3	3
11	12	+	+	A	A	normal	normal	3	3
12	13	+	+	A	A	normal	normal	3	3
13	13	+	+	A	A	normal	normal	3	3
14	14	+	+	A	A	normal	normal	3	3
15	14	+	+	A	A	normal	normal	3	3
16	14	+	-	A	B	normal	normal	2	2
17	15	+	+	A	A	normal	normal	3	3
18	15	+	+	A	A	normal	normal	3	3
19	16	+	+	A	A	normal	normal	3	3
20	16	+	+	A	A	normal	normal	3	3

Otoscopy: +/- Pass/Fail

Tympanometry: Jerger Classification (1970). [A (normal); As and Ad (mild abnormalities) and B, C and Cs (moderate to severe middle ear abnormalities)]

Pure Tone Audiometry (PTA): (Hearing Threshold Classifications Lee 1999)

DPOAE:- Pass /Refer (**Pass** 2 and 3 octaves / **Refer** pass only one octave or less)

3.7 ORTHOPAEDICS

This study showed that 71% of children had orthopaedic problems, the most common were pes planus (flat feet) (63%), hip joint instability (2.%), knee joint laxity (2%), ankle joint instability (1%) and atlanto axial joint instability (1%).

3.7.1 Orthopaedic problems

Physical examinations was performed successfully by one paediatrician (MP) on 366 children and showed that 249 (63%) children had flat feet. However, when asked only 9 (2.3%) reported any pain or discomfort. One child had sore pressure areas on both feet.

Parents reported that the most common musculoskeletal disorder was poor muscle tone in 17 children. This was followed by pes planus (11), hip joint instability (10), knee joint instability (11), scoliosis (4), atlanto axial joint instability (4), cerebral palsy (2), ankle and elbow joint instability (2), joint pain (4) and Perthes disease in one child. (Table 3.120).

Table 3.120 History - Musculoskeletal disorders and Orthopaedic problems		
Orthopaedic problems	N =70	%
Poor muscle tone	17	24
Flat Feet	11	16
Hip joint instability	10	14
Knee joint instability	11	16
Scoliosis	4	6
Atlanto axial joint instability	4	6
Cerebral palsy	2	3
Ankle/elbow joints instability	4	6
Joint pain	4	6
Perthes disease	1	1

3.7.2 Treatment

Parents reported that that 63 (16%) of children had received treatment for orthopaedic problems. (Table 8.4) The most common treatment was physiotherapy in 39 (62%) children. Some children required a combination of treatments and a small number received footwear intervention (Table 3.121).

Table 3.121 Treatment for orthopaedic problem		
Treatment	N =63	%
Physiotherapy	39	62
Surgical treatment	11	17
Physiotherapy and surgical treatment	3	5
Special boots	3	5
Medical treatment	2	3
Physiotherapy and medical treatment	2	3
Physiotherapy and occupational therapy	1	2
Shoe lift	1	2
Insoles	1	2
Osteopathy	1	2

Surgical intervention was required in 11 (3%) children and included fusion of atlanto axial joint in 3 (0.8%) for cervical spine instability; patella surgery in 3 (0.8%) and surgical correction of scoliosis in one child. The mean age at diagnosis of atlanto axial joint subluxation was 3.8 years (range 3.5 – 7 years) (Table 3.122).

Table 3.122 Operations for Orthopaedic problems		
Operations	N =11	%
Fusion atlanto axial joints	3	0.8
Patella instability repair	2	0.5
Tendon repair ankle	2	0.5
Scoliosis	1	0.3
Patella instability and toe joint repair	1	0.3
Toe correction	1	0.3
Separation of joined fingers	1	0.3

3.8 SPEECH AND LANGUAGE

A very high proportion comprising of 371 (94.5%) children had speech impairment whilst normal speech for age was found in only 6 children (1.5%). Parents reported difficulties in obtaining speech therapy throughout childhood.

3.8.1 Parent concerns

Parents were asked if they had concerns relating to their child’s ability to speak and 82% indicated that they were very concerned (Table 3.123).

Table 3.123 Speech concerns by parents		
	N =372	%
Yes	304	82
No	68	18

3.8.2 Screening

Eighty percent (80%) of children had been screened for speech whilst 19% had not yet been screened (Table 3.124).

Table 3.124 Child speech assessed		
	N =381	%
Yes	302	80
No	74	19
Don't Remember	5	1

Reports by many parents indicated there was often a major delay in speech assessment as only 19% of children had been screened for speech for the first time before the age of 2 years, 26% between two and four years and 13% between four and

six years, with a further 8% between six and 14 years of age (Table 3.125). Many infants and toddlers with Down syndrome may be sensitive to touch and can be tactilely defensive (Kumin 1994). Early assessment and intervention in developmental needs, such as muscle stimulation and feeding therapy is therefore important in these children as the first steps in developing communication skills.

Table 3.125 When speech assessed		
Age (years)	N =368	%
Under 1	21	6
1 to 2	46	13
2 to 4	94	26
4 to 6	48	13
6 to 8	20	5
8 to 10	3	1
10 to 14	7	2
Did not remember age assessed	60	16
Not assessed	69	19

The location of speech assessment varied greatly. The majority of children (49%) were assessed for speech development within the special development services, followed by 23% at the public health clinics by area medical officers. A further 25% were assessed by speech therapist but parents did not indicate if this occurred within a service or private sector. Four children were assessed at school, two by a home

teacher, three children privately and one child had been assessed outside Ireland (Table 3.126).

Table 3.126 Location of speech assessment		
	N =265	%
Special Developmental Services	130	49
Eastern Health Board Clinic	59	23
Speech therapist	55	21
Speech therapist and TCD Speech Dept	6	2
Speech Therapist and Psychologist	5	2
School	4	2
Privately	3	1
Home Teacher	2	1
Outside Ireland	1	1

3.8.3 Clinical Assessment

On clinical examination by the Lecturer in Paediatrics (MP) speech ability was recorded indicating that only 23 (6%) children could speak clearly and communicate with same aged children in mainstream school. The majority of children 206 (58%) were communicating with between 1-2 and 4 -5 word sentences (age range 2 to 18 years). Single words only were used by 53 children (age range 2 to 18 years), 3 children spoke with a stammer, 9 children had no speech (age range 2 to 12 years), whilst only one of

these children had profound intellectual disability. Two children communicated by sign language alone (Table 3.127).

Table 3.127 Speech and Language milestones										
Age in years										
Speech	0-2	2-4	4-6	6-8	8-10	10 -12	12-14	14-16	16-18	Totals
No speech yet	5	3	2	1		2	1			13
Single words only	12	27	12	5	3	1	1	2	2	65
Sign language only		1				1				2
Babbling	10	1								11
1-2 words	4	4	9	2	1	1	2		1	24
2-3 words		2								2
3-4 words					1		1	1		3
4-5 words		1								1
< 10 words	1	1								2
1-2 word sentences		5	6	2	1	3	2	2	1	22
2-3 word sentences		5	16	9	17	16	18	14	7	102
3-4 word sentences			4	2	11	7	9	2	3	38
4-5 word sentences			1	4	6	7	9	9	8	44
Communicate with peers			1		1	2	1	2	1	8
Clear Speech				1		5	2	4	3	15
Stammer						1	1	1		3
Totals	32	50	51	26	41	46	47	37	26	356

3.8.4 Treatment

Parents recorded that 73% of children had received speech therapy, whilst 27% had not yet received any speech therapy. Parents reported that speech therapy was not always consistent as children had long periods of no therapy when a replacement speech therapist was needed. They also reported that there were very few private speech therapists available who were willing to teach their children (Table 3.218).

Table 3.128 Speech therapy		
	N =373	95%
Yes	274	73
No	99	27

Sign language had or was being used at the time of the study by 43% of children whilst 57% had never used any sign language (Table 3.219).

Table 3.129 Child use sign language		
	N=394	100%
Yes	165	43
No	220	57

The majority of children (86%) used the Lámh sign language (specific for children with intellectual disability) and a small number of children used Makaton (specifically for children and adults with a variety of communication and learning disabilities) and the Irish sign language (a complete language with a unique vocabulary). Some parents reported that the sign language helped their child with communication and in the development of speech. Other parents expressed concern that their child would

never develop speech and would continue to use only sign language (Table 3.130).

Table 3.130 Type of sign language used		
	N =164	42%
Lámh	141	86
Lámh + Makaton	2	1
Lámh & Irish sign language (ISL)	1	1
Makaton	2	1
Irish sign language (ISL)	1	1
Did not remember sign language used	17	10

3.8.5 Social Skills

Children were assessed clinically by the lecturer in paediatrics (MP) in relation to their social skills. It was recorded that 90% were delayed in this area whilst 10% had normal age appropriate social skills (Table 3.131).

Table 3.131 Social skills		
	N =373	95%
Delayed	335	90
Normal	38	10

3.9 QUALITY OF LIFE RESULTS

The response rate for this section of the questionnaire was very high thus giving an indication that it had good face validity and feasibility for ease of use and completion. Disruption to parents 90%, disruption to family routines 90%; reduced time for other children 85%; child's physical problems requiring extra parental care 88%; child's general health 95% and child's quality of life 93%, indicating good face validity and feasibility. The parent(s) willingness and ability to complete /or (uniformity in completing) all sections of the multiple item questionnaire indicated good internal consistency and this was confirmed by Cronbach's α coefficient (Cronbach's α coefficient 0.87 (n=310).

Questions were scored from 1-4 (range of scores 4-20; large = 4; moderate = 3; small = 2 and none = 1), a lower total score indicated less burden. A total score was calculated from simple summation of the items and then averaged thus creating a continuous scale. Data were normalised so that the greatest burden score was 100 and the least burden score was 0. The categories were collapsed from 4 to 3 categories for purposes of analysis as follows large, moderate and small/none. To illustrate the distribution of scores the mean and standard deviation were calculated and family burden subscale scores as perceived by parents are shown in Table 3.132 (page 160). In addition parent's perception of their child's quality of life and general health were scored 1-4 (excellent = 1; fair/good = 2; poor = 3; very poor = 4), a lower score indicated a better quality of life and general health, as shown in Table 3.1323. (page 162).

The major burden related to long-term concerns (with 69% of parents seeing this as a large to small burden) and the smallest burden related to physical problems requiring extra parental care (with only 10% seeing this as a major burden).

3.9.1 Disruption to Parents

Disruption to parents is illustrated in Table 3.132. The majority of parent reported disruption to themselves due to caring for a child with special needs, 64% of parents considered this a small to large burden and 37% reported “no burden”.

Table 3.132 Parent perceived family burden subscales and range of scores

QOL-related items	Family Burden			Mean (SD)	Range
Parent	Large (%)	Moderate (%)	Small /none (%)		
Disruption to Parent time	14	23	63 (26/37)	44.5 (38.2)	0-100
Disruption in family routines	12	22	66(29/37)	38. (35.5)	0-100
Disruption in the time for your other children	14	20	66 (32/34)	36 (34.6)	0-100
Physical problems requiring extra parental care?	10	19	72 (23/49)	38 (34.9)	0-100
Concerns about long-term Health	23	18	59 (28/31)	29.6 (34.1)	0-100

Table 3.133 Parents perception of QOL and general health of child subscales and ranges

	Poor/Very poor (%)	Fair/ good (%)	Excellent (%)	Mean (SD)	Range
Child’s general health	1.8	36	63	12.9 (17.6)	0-100
Child’s QOL	4.1	40	56	16.1 (020.1)	0-100

Disruption to parents was greatest in the early age groups, which was not statistically significant (p value 0.49) less in the 4 to 7 age group with a further upward shift up to 14 years when there was another gradual reduction. This was similar for parents of girls and boys (Figure 3.37).

Figure 3.37 Disruption to parents by age and gender of child



3.9.2 Disruption to family routines

Parents reported disruption to family routines as a small (29%), moderate 21% to large burden (12%) when rearing a child with special needs whilst 37% reported no burden.

3.9.3 Disruption in the time for other children

Parents reported that they had less time for their other children because of caring for a child with Down syndrome. Disruption in parents time for other children in the family showed that whilst 34% reported no burden, the majority (67%) reported a burden (32% small, 20% moderate and 14% large burden).

3.9.4 Child physical problems requiring extra parental care

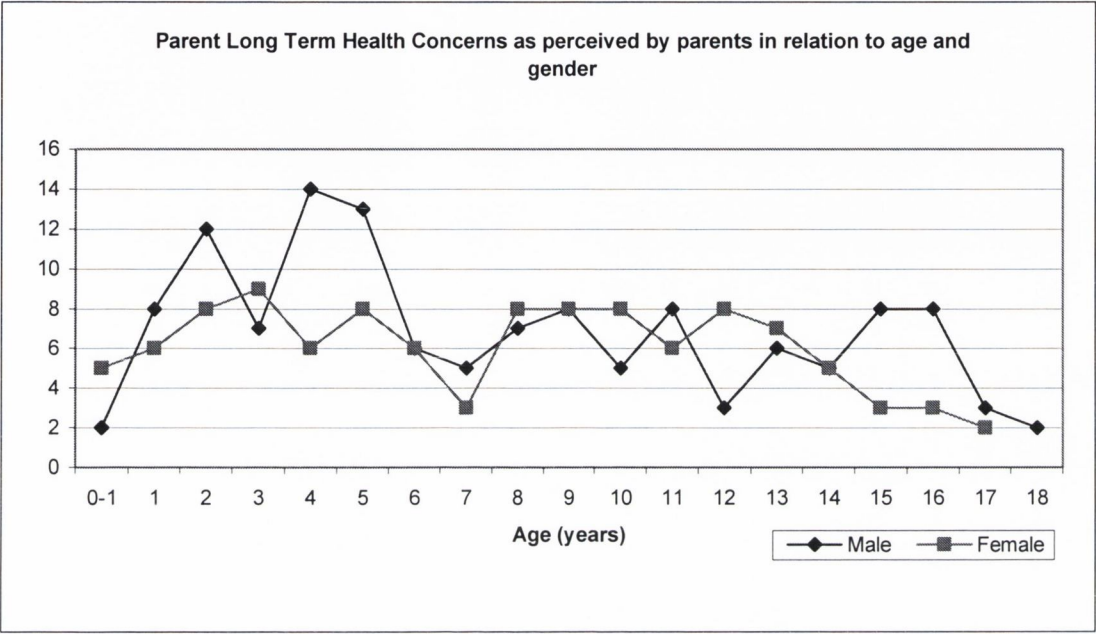
No added burden was reported by 49% of parents because of their child's' physical problems. Parent(s) perceived the burden as small (23%), moderate (19%) and large in (10%) of cases.

3.9.5 Parent long-term health concerns relating to child

A high percentage of parents (69%) reported concern about their child's long term health (23% large; 18% moderate, 28% small burden), whilst 33% reported no burden. Long term health concerns relating to child as perceived by parents is illustrated by age and gender, but not statistically significant (p value 0.21) (Figure 3.38). Parent perceived burden was greater for boys than girls in the first 6 years of life and again in the older

age groups but was similar for both sexes between 7 and 14 year age groups (Figure 3.38).

Figure 3.38 Parent Long Term Health Concerns



3.9.6 Parent perception of child’s general health

The majority of parents (59%) indicated that their child was in excellent general health whilst 36% considered that their health was fair/good. Very few parents rated health as poor (1.3%) or very poor (0.5%).

3.9.7 Parent perception of child’s quality of life

A high percentage of parents (56%) considered that their child’s quality of life was excellent, 38% fair, 1% good, whilst 3% indicated that their child’s quality of life was poor and 0.8% very poor. The five children in the very poor bracket did not have life threatening medical problems, however, they all had hearing and vision problems.

In contrast results showed that for two children with very poor health, one under four years and the other 16 years, parents perceived that their child had an excellent and fair quality of life respectively. The data showed that excellent quality of life dominated from early childhood right through until approximately 7 years of age, following which Quality of Life (QOL) was considerably less with very low levels in teenage years (Figure 3.39 and Table 3.134 page 165).

Figure 3.39 Parent Perception of Child’s quality of life by age

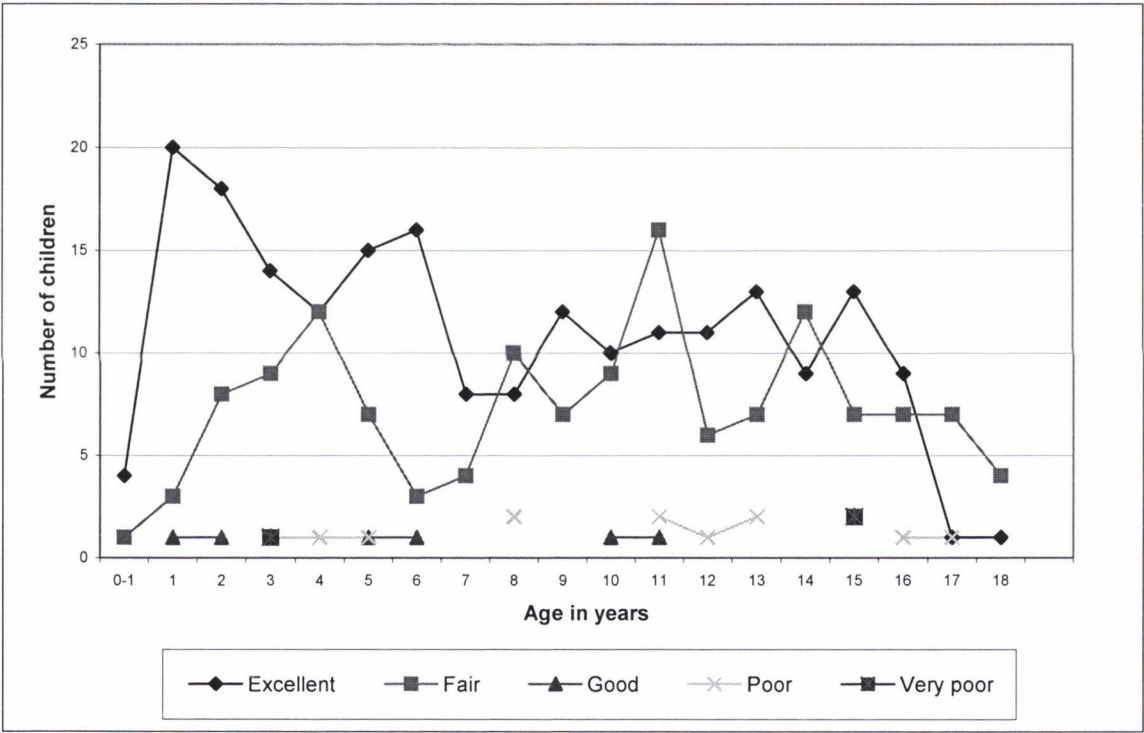


Table No 3.134 Parent Perception of Child’s quality of life by age in percentages

~Age	Excellent %	Fair %	Good %	Poor %	Very poor %
<1`	1.1	0.3			
1	5.5	0.8	0.3		
2	4.9	2.2	0.3		
3	3.8	2.5		0.3	0.3
4	3.3	3.3		0.3	
5	4.1	1.9	0.3	0.3	
6	4.4	0.8	0.3		
7	2.2	1.1			
8	2.2	2.7		0.5	
9	3.3	1.9			
10	2.7	2.5	0.3		
11	3.0	4.4	0.3	0.5	
12	3.0	1.6		0.3	
13	3.6	1.9		0.5	
14	2.5	3.3			
15	3.6	1.9			0.5
16	2.5	1.9		0.3	
17	0.3	1.9		0.3	
18	0.3	1.1			

When the total scores of the QOL questionnaire scale were calculated and the distribution checked it was found to be positively skewed. To apply parametric statistics a transformation of the scores was undertaken using the logarithm function (Armitage

1998). This resulted in a distribution that more closely approached the normal distribution and, consequently, a decision was made to proceed with parametric statistical analysis.

Pearson product-moment correlation analysis indicated that the relationship between parents' feelings of burden, as measured by the summed scales, and the child's general health was statistically significant ($r = .31$, $n = 366$, $p < .01$). However, as the value of r indicates, the relationship was weak suggesting a tendency (weak) for high levels of feelings of burden to be associated with poor levels of health. In addition correlation analysis indicated that the relationship between parent perceived burden (as measured by the averaged summed scales) and child's QOL was also statistically significant ($r = .38$, $n = 359$, $p < .01$). Nevertheless, as the value of r indicates, the relationship was weak positive suggesting a tendency (weak) for high levels of perceived burden to be associated with poor quality of life of the child.

In order to take into account possible confounding factors that might have influenced the parents perceptions correlation analysis were undertaken between the parents' perceived burden scale and the variables - age, gender, family size, parents' working/not working, marital status and indicated that there were no statistically significant relationships. However, three other variables were statistically significantly correlated with the perceived burden scale. These were, concerns about hearing, ($r = .19$ $n = 351$ $p < .01$), speech and language ($r = .21$, $n = 349$ $p < .01$) and vision ($r = .12$ $n = 354$ $p < .01$). However, it is clear that all r values indicate very weak relationships.

3.9.8 Changes Needed to Improve Child’s Quality of Life

Parent(s) were asked to give their opinion relating to changes which would explore the phenomenon of effective ways to improve the quality of life of children with Down syndrome by asking “What do you think could improve the quality of life of your child?”. This section of the questionnaire was completed by 316 parents (80.2%) who provided descriptions of effective changes which they perceived could improve their child’s quality of life. The detailed comments by parents were reviewed and the core perceived needs were divided into themes: - health, education, information, long-term management and community acceptance (Table 3.135).

Table 3.135 Parent Perceived Changes needed to improve child’s QOL

Health	%
Genetic counselling + diagnostic test results	1.5
Breaking the news of diagnosis in a better way	6.1
Early intervention	33.2
Speech & Language	50.5
Improved health service/therapy	34.3
Medical cards for all children with DS	6.0
Education	
Mainstream education	43.1
Remedial support in classroom	26.6
Teacher education re Children with DS	2.5
Long term education	6.6
Employment training	7.7
Additional funding for special schools	1.5
Information	
More specific information on DS	17.3
More awareness of DS in community	12.9
Coordinating body	16.8
Long term management	
Development of Independent living schemes	11.9
Development of long-term care support	6.3
Residential Care	12.9
Community Acceptance	
Integration and acceptance within local community programmes	26.1

3.9.9 Health

Only 1.5% expressed a need for more genetic counselling and diagnostic results.

Parents expressed concern relating to the manner in which they were informed of the diagnosis as in the following statements;

“Doctor’s negative expectations were undermining and disempowering”

“I was too emotionally confused to even think of looking for help”

“I have never recovered from what I was told about my child when he was born. I had been through a tough pregnancy and delivery, my baby had to be resuscitated twice and eventually the consultant brought my baby to my room and handed him to me saying “Here’s your Christmas pudding, it has been nothing but a problem all your pregnancy, it will be nothing but a problem all your life” and left me on my own with my baby”

“I believed the doctor when told that my child would never be able to achieve anything. I lost so much time in her development as it was two years later that she showed me she could do so much”

“We had to wait 1 month for test results to know if our child had Down syndrome. We were very dissatisfied with the way we were told about Down syndrome”

“Staff in maternity hospital don’t know how to deal with situation”

“Parents of babies with Down syndrome to be treated better in maternity hospital”

“Parents should never be made feel that it is their fault”

“The expectations set by the doctors at birth were very negative and something we were not prepared to accept. Our son constantly surpassed these expectations, when he walked, talked, used the toilet etc. If we had accepted these limitations who knows how damaging they would have been to his development and would have certainly reduced his quality of life and ours too”

Parents stressed a great need that the news that a child has Down syndrome be “Said in a more positive way”

Early Intervention

A High proportion of parents expressed their concerns regarding the need for early intervention as follows: -

“Birth to 2 years is very important in development of child with Down syndrome - requires programme for early intervention and speech therapy”

“More intensive early services are required – physiotherapy, occupational therapy and speech and language therapy”

“Parent need more education in early intervention”

“Home teaching at earlier stages”

“Early services could be more imaginative, innovative and dynamic”

“Physiotherapy and occupational therapy are very important for young babies and children with Down syndrome. I would love to see better facilities once or twice a week when baby is very young”

Speech and Language

The need for speech and language assessment and therapy was a major concern of parents as related in the following statements: -

“I am a prisoner in my own home because my son cannot speak and therefore cannot play with other children on the road”

“The speech therapist left our service two years ago and we are still waiting for a new speech therapist – meanwhile our child has to go without and we cannot afford private lessons”

“Proper and continuous speech therapy”

“Speech therapy once a week”

“Children should have legislative right to speech and language service”

“Full assessment of speech development from early childhood”

“Regular assessment of children’s progress”

“Regular speech training on weekly basis”

“Improve parent education in this area”

“Sign language for children and parents”

“Updates of Hammond course for parents”

“Home physiotherapy and occupational therapy to help with speech development”

“Availability of a structured regular speech therapy service especially when very young”

More speech therapy in his own area

Improved Health Service/Therapy

The need for improved health service/therapy was also a major concern of parents as expressed in the following statements:-

“Parents get tired and become embittered trying to access services”

“My son is severely mentally handicapped and needs care 24 hours a day 365 days a year which we have always supplied. I just get more weary. Our quality of life has suffered”

“The hardest part of having a child with a handicap is that you have to fight for their medical needs – one would think you had enough to cope with and there are other members of the family to be considered”

“Often it is too late when needs are attended to and valuable time has been lost in their development. We become tired of struggling with a system which continues to fail our child and with the frustration and the stress associated with it that we often feel like giving up the struggle. But if we give up what hope does our son have for the future”

“I was very confused and upset to find that our daughter had DS as was her mother. But with the inadequate counselling her mother lost interest in her rearing. This has affected our daughter and her development and caused difficulties between us.”

“Regular co-ordinated health checks”

“Regular physiotherapy and occupational therapy”

“Services are disjointed – different people asking the same questions”

“Parents feel they have to look for everything”

“More physiotherapy so child will walk soon”

“Better co-ordination of services”

“More structured services for physiotherapy and occupational therapy”

“General health monitoring”

“The current health board services are superficial – staff appear to be going through the motions and there is an absence of parent and professional working in harmony”

“Child centred services instead of the child having to fit in with the service”

Medical Cards

Parents expressed a need for the introduction of medical cards for all children with Down syndrome.

“Medical card should be issued to every child with Down syndrome as we find it a large financial burden”

“Proper state support from birth as we were made feel like beggars”

3.9.10 Education

A high proportion of parents (43%) wished to have their child in mainstream school

“My child is more stimulated when with normal children”

“My child is afraid of dealing with children with mental handicap”

“I prefer my child to mix with normal population”

“Better integration in the community e.g. schools, sports activities”

“Access to educational psychologist reports”

“Better access to educational assessment with up to date reports available to parents”

“We are in constant fear for the future. No alternative centre with “Normal” children – child picks up mannerisms of other handicapped children”

“Local integration with automatic resource teacher’s for child”

“Integration and inclusion to give her independence”

“After school care and activities – at present child with grandparents after school as no one will take on a child with Down syndrome”

Supply of remedial support in the classroom was a major concern of parents

“Better communication and more educational support- classroom assistant”

“If child goes to normal primary school he will only have a qualified outreach teacher for 1 hour per week. Mum would like service at least 3 hours per day”

“More choice with schools and more classroom assistants”

“More support teachers in mainstream schools”

“Having more expert services available on a regular basis. Resource teachers for longer periods during school hours”

A small number of parents expressed a need for implementation of education programmes relating to Down syndrome for Teachers.

“Education of mainstream teachers to handle children with Down syndrome – Teachers lack confidence”

A small proportion (1.5%) of parents expressed a need for additional funding for special schools

“Increase in financial support to centres in order to enable improved services where necessary”

“More resources in mainstream schools of a specialist nature – speech and language and psychology services”

3.9.11 Information

Parents were very concerned about the lack of information available about Down syndrome itself and also the services available in their areas as can be seen from their comments below,

“I had no clue about Down syndrome babies. I had to ring up the Down Syndrome Association and find out something as I got no information whatsoever in the hospital, I felt very alone those few months after my son was born”

“More information on Down syndrome and heart conditions”

“A guide to services that are available – parents feel they have to look for everything. Contact should be maintained between families and the services as the child grows.

“Easy access to results of recent research on Down syndrome”

“Have an information service on Down syndrome – some one to contact for advice”

“Down Syndrome Help line”

“National register of Down syndrome children with automatic information sent out”

“Parents should have a right to information and so make appropriate choices”

3.9.12 Long term needs

Long term needs both in education, independent living and long-term care were deep concerns in a substantial number of parents as expressed in the following statements:-

“Residential summer camp would be a great benefit – no such service available yet”

“Residential care if parents die - No way will my daughter go into a home”

“I don’t have concerns about our daughter’s long-term health but do have concerns about her welfare and care when her parents are dead”

“Independent living in a paid job and support services to allow him live an ordinary life”

“Increase training after completion of formal school to prepare for work

“Employment opportunities need to be developed”

“More financial resources by government put into area of children with special needs so that each child is encouraged to reach his/her potential in an appropriate setting”

“Independent accommodation and living schemes need to be developed”

“Home alteration to accommodate child and young adult needs”

“Provision for the running of small group homes in the community for the future for adult people with Down syndrome”

“Funding for respite care and long term care”

3.9.13 Community acceptance

Parents expressed concerns regarding community acceptance as made in the following statements:-

“Education of “normal” population to learn about those with special needs”

“Treat as normal by peers and public”

“Children with Down syndrome should be treated equally with other children”

“All need to look beyond the condition Down syndrome and see the child”

“Change peoples attitudes to acceptance of child with Down syndrome in mainstream schools”

“Society could be more tolerant and understanding of people with special needs”

“In our opinion it would be highly beneficial if EHB services were directed where possible to the integration of children with Down syndrome (levels permitting) into local schools. This would enormously increase the child’s acceptability locally and reduce the dependency on special schools”

These results identify the areas in most need of improvements as potential strategies for practice development and management in this population. Findings from this study support the need for more extensive research in this area as well as providing heightened awareness among health care professionals of the perceived stressors experienced by parents caring for a child with Down syndrome in the community.

CHAPTER FOUR

DISCUSSION

CHAPTER 4 DISCUSSION

4.1 INTRODUCTION

Down syndrome is the most frequent chromosomal anomaly which occurs in 1/546 live births in Ireland. This study examined a large representative sample of children and adolescents with Down syndrome in the Eastern Regional Health Authority (ERHA). The high response rate indicated a very high level of genuine parent concern for their children's health and future life. The results of this study provide evidence based data that children and adolescents with Down syndrome have a high incidence of treatable medical disorders and associated conditions, which require early detection and intervention in order to attain a better outcome and quality of life. Therefore, health care providers require accurate information regarding the occurrence of this condition in their population so that they can plan and provide appropriate services to people with Down syndrome. To date no live register has been established for this population and the EUROCAT register is the most accurate and only source that identifies all those born and diagnosed with Down syndrome in the area studied. However, this register is not in operation nationally.

This study sought to capture all those with Down syndrome in the area studied, and identified 571 with the aid of EUROCAT, Down syndrome Ireland register and all the special developmental services. However, whilst 394 participated in the study 352/394 of the children seen in this study were registered with EUROCAT and there was a remaining 224 identified but not seen. In order to check for possible bias all three populations were examined in relation to the incidence of congenital heart disease (CHD). The 352 children seen during the study and on the EUROCAT register were found to have 174 (49%) children with CHD. The 224 children on

register and not seen were found to have 67 (30%) children with CHD. The remaining 42 children seen but not on register were found to have 17 (41%) children with CHD. This may indicate a possible but small bias toward the children with heart anomalies, and could indicate that the parents of children with heart disease might be more motivated to participate in a study on health issues. In addition 33% of those who participated in the study were members of DSI out of a possible 40% identified.

However, this study identified 42 children diagnosed with Down syndrome and not entered on the EUROCAT register for the very good reason that they had either been born outside the catchment area or in the catchment area but to parents resident outside the catchment area at that time. This would suggest that EUROCAT Register should be rolled out nationally, and that additional time points be added for capture other than birth.

4.2 Clinical Questionnaire

The ERHA encompasses almost one third of the country's population and the study group of 394 (217 boys and 177 girls) represented 67% of all children and adolescence with Down syndrome born between 1981 and 1997 in this area. The group comprised of 19% from Wicklow County, 19% from Kildare County and 72% from Dublin County with a relatively even distribution of boys and girls throughout. The majority of the children were born within the ERHA with 3% born elsewhere in Ireland and 3% born outside Ireland. Most children were living at home with their families thus providing the opportunity to assess these children in a natural setting.

The larger family size of over four children represented 44% of the study group. This was greater than the general population of 11% in 2002, which decreased from 27% in 1981. In a previous study by Mercedez Egan (1993) of children with Down syndrome in 1993 the larger family represented 51% in contrast to our findings indicating a similar downward shift in family size with the general population

The proportion of mothers who gave birth prematurely was 15% whilst 85% delivered their babies between 37 and 43 weeks gestation. Twenty two percent (22%) of babies were born by caesarean section which is greater than the national average figures for caesarean section births in the year 1998 (17.8%) In this study 18% of children with Down syndrome were the first child born in the family which contrasts to the general notion of children with Down syndrome being the last child to be born within the family. The maternal age at the birth of the baby ranged from 16 to 49 years of age with 50% of mothers being under 35 years of age. The paternal age at birth ranged from 17 to 61 years of age with 42% under 35 years of age.

The majority of parents were married (83%) whilst 8% were separated, 6% were single parents and 4% were widowed. One family had adopted their child with Down syndrome.

The majority of fathers (93%) were working whilst only 37% of mothers were working full time outside the home, 2% working part-time and 61% of mothers were not working outside the home. Mothers were therefore, the principal person caring for the child with special needs at home

Learning that one's child has Down syndrome is a traumatic experience. However, the location and the amount of time spent giving this information to parent(s) is vital as it will have a long lasting affect and may determine how parents are able to accept the diagnosis and bond with their baby. This study found that 29% of parents were given their child's diagnosis in a non-private location, 37% of mothers were either alone or with their baby when given the diagnosis. Parents indicated that 45% felt that insufficient time was given when informed of the diagnosis, whilst 31% were given insufficient or no information relating to Down syndrome at the time of diagnosis and 46% reported that they were not offered the opportunity of a follow up discussion.

Many parents expressed a need for; more care in the way new parents are told that their baby has Down syndrome; that follow up support with up to date information relating to the condition and the need for support services to be more available and easily assessable.

The uptake percentage of immunisations were higher than the national average from the Regional Interactive Child Health System's database (Fitzgerald et al.2001), indicating the dedication of parents, public health nurses and healthcare professionals for these children.

Developmental milestones in this study correlated well with generally reported development of children with Down syndrome as described by Cunningham 1988.

Many parent(s) expressed the need for improved services particularly in the areas of Speech and Language assessment, delivery of Speech Therapy, educational

assessment and support in mainstream and special schools, financial support (Medical Card available for all children with Down syndrome), long-term needs including training in young adults and the development of independent living.

4.3 Growth

This study of a large representative sample of Irish children with DS reared at home in natural settings, from 3 months to 18 years of age studied height, weight, head circumference and puberty and is the first growth study on this group in Ireland. Prior to this the American Down syndrome specific growth charts for height, weight (Cronk 1981) and head circumference (Palmer 1992) were used. While short stature is well recognised in Down syndrome (McCoy 1992, Cronk 1981) the pattern of growth and puberty is poorly understood. In some children with Down syndrome, associated conditions, such as hypothyroidism (Karlsson 1998, Sharav 1988) sleep related upper airway obstruction (Stebbens 1991) or coeliac disease (George 1996, Jansson 1995) may contribute to short stature and must be promptly excluded to optimise their already compromised growth potential. This study found that boys and girls with Down syndrome have a shorter height with a lesser growth spurt at puberty. In general BMI was greater and a significantly smaller head circumference than the general Irish population was demonstrated at all ages.

Comparison with US Down syndrome specific growth charts showed Irish boys to be marginally taller than those of the US (SD scores + 0.23) while girls were similar for height (mean SD scores – 0.3) (Cronk 1988). The Cronk study (1988) comprised of 4,650 observations on 730 children however there was insufficient information relating to health problems provided and those with poor growth associated with cardiac

problems (140/ 19%) were included. Comparison of height in Irish children with Down syndrome with and without cardiac defects showed no significant differences in height in either boy or girls. Earlier access to improved cardiac care over the previous 15 years may be an explanation for this. Cremers et al (1996) in Holland studied 295 health children with Down syndrome providing 2,045 observations, the lesser height of Irish boys and girls in comparison to the Dutch was expected, as the Dutch population are known to be taller than the general Irish population.

Those with Down syndrome have an increased incidence of overweight and obesity (Chumlea 1981, Prasher 1995) and this weight gain may reflect undetected thyroid disease in some. However, the prevalence of child obesity is increasing rapidly worldwide (World Health Organisation 1997) and is associated with significant adverse health (risk factors) in later life including diabetes and other chronic diseases such as hyperlipidaemia, hyperinsulinaemia, hypertension and early atherosclerosis (Berenson 1993, 1998, Mahoney 1998). Body mass index changes greatly with age in childhood (Rolland-Cachera 1982, Cole 1996). The median body mass index (BMI) in the general population is at birth, is 13 kg/m², to 17 kg/m² at age 12 months, 15.5 kg/m² at 6 years and 21 kg/m² at age of 20 years (Cole 2000). Overweight and obesity have been defined as those above the 91st percentile and 98th percentile respectively on the UK 1990 BMI reference charts (Cole 1996). Irish children compared poorly with children with Down syndrome in the UK study where only 20% were obese (Styles, Cole and Dennis 2001). This study on Irish children showed that 18.4% of girls and 17.4% of boys were overweight with a BMI greater than the 91st percentile while 24.1% of girls and 28% of boys were obese with a BMI greater than the 98th percentile for the general UK population. However, in the older age group the prevalence of obesity, in those over

10 years of age, was 33% of girls and 29% of boys with a BMI greater than the 98th percentile. This may indicate a sharp rise in obesity in those with Down syndrome in Ireland particularly in the older age groups compared with the general UK population.

Comparison with the US Down syndrome specific weight charts, showed an overall greater weight in Irish children (mean SD score boys 0.3kg; girls 0.03kg) with a particular increase at 14 years in boys and 11 and 16 years in girls. Irish adolescents with Down syndrome were also heavier than Dutch children when compared with the Dutch Down syndrome specific weight charts (Cremers 1996). This is particularly concerning and unexpected considering the taller stature in the Dutch Down syndrome population. It suggests excessive weight gain in Down syndrome is not inevitable. Early education and dietetic referral with active intervention in diet and exercise is desirable. Promoting increased activity with enjoyable exercise can prevent excess weight gain in children and adolescents with Down syndrome (Medlen 2002, Cremers 1996). Individuals with Down syndrome have reduced resting metabolic rates (Luke 1994, Allison 1995). A lifelong regimen to monitor growth and prevent obesity should begin at 24 months of age, including food selections, behavioural intervention, physical activities, and social activities. In adults with Down syndrome, a reduced body-mass index correlates with lifestyle variables, especially satisfaction in friendships and access to recreational and social opportunities (Fujiura 1997). Diet should be planned to favour nutrient-rich foods that are high in fibre, and low in calories and fat (Roizen 2003)

Pubertal development in females with Down syndrome has generally thought to be delayed (Benda 1960). However this study found the average age of menarche was 12.6 years, which is slightly earlier than the general Irish population in the 1970-

1980's (Hoey 1987). A study in Japan of 95 females with Down syndrome (age 9 to 18 years of age) indicated the age of menarche as 12.1 years (Takano 1999). However, the Japanese are known to have earlier menarche than women in European countries age in years (Tanner 1973) and 12.55 years (Danubio 2004). A small study of 22 females with Down syndrome in Sweden (Arnell 1996) reported a mean age at menarche of 13.2 years which was related to maternal age of menarche thus similar to that of the general population. Goldstein (1988) studied a group of women in Denmark with a mean age of 18 years. This study included 15 girls with Down syndrome and 33 control girls. There was no difference in the average at menarche (13.6 years), cycle length (28.3 days); and duration of bleeding (5.5 days). In addition, Scola and Pueschel (1992) who studied the menstrual cycles and basal body temperature curves of 51 females with Down syndrome calculated an average age of onset of menstruation at 12.5 years. They found that 76% had average menstrual cycle length of 25 -30 days with 88.5% having an ovulatory pattern.

Therefore many girls with Down syndrome are menstruating regularly, and for many years. This raises the question of how we address their management of menses, in the social situation, from a hygiene and social point of view, but also considering their self awareness and dignity. Premenstrual pain and/or pain during a period can be problematic, particularly as those with Down syndrome may often not understand the source of their pain. This can sometimes manifest as behavioural change with emotional lability and acting out behaviour. It is also recognised that many adolescents with Down syndrome are happy to have menstruation, because it implies being more similar to their mothers and sisters and peers.

However, the other issues that become apparent when we look at menarche in this group are the issues of sexual activity and education in relation to sexuality and contraception. Goldstein (1988) found that there was a difference in relation to sexuality and intercourse in his study where 79% of the controls had been sexually active whereas none of the girls with DS were known to have been sexually active. However, three of those with Down syndrome were using oral contraception and one was using a long acting injectable contraceptive (Depo-Provera), possibly because of period pains/irregularity or because of parental instigation, compared to the reports of 36 (78%) of the controls using contraception (oral contraceptive pill 19; diaphragm 3; condom 16).

Information regarding fertility in women with Down syndrome is limited. The National Down Syndrome Society (NDSS) in America states that 50% of women with Down syndrome are fertile while Scola and Pueschel (1993) found that it could be as high as 88.5%. Bovicelli (1982) reviewed the outcome of 30 pregnancies in 26 women with Down syndrome which resulted in the birth of 10 children with Down syndrome, 18 children (1 set of twins) without Down syndrome and 3 miscarriages. Scientific information about the fertility of men with Down syndrome is very limited. There have, however, been at least two documented cases where the father had Down syndrome. Sheridan (1989) published the first fully documented report case of a non-mosaic male with Down syndrome fathering a child. Zuhlke (1994) in Germany also reported a male with Trisomy 21 fathering a child. It is likely that additional cases may be recognized, especially as more men have the opportunity to live in the community and develop intimate relationships. It is not known yet if the offspring of

men with Down syndrome are more likely to have Down syndrome or other anomalies.

People with Down syndrome are living longer, are increasingly being educated in mainstream schooling and there is no doubt that they are becoming more exposed to what goes on in society. It is very important that adolescents develop an awareness of their own sexuality. The NDSS expresses it well “the emotional changes characteristic of adolescence are present in children with Down syndrome, and may be intensified by social factors. Any adolescent who lives in the community, attends school and is exposed to media inevitably develops an awareness of sexuality. Teenagers and young adults with Down syndrome often express interest in dating, marriage and parenthood. They can be expected to experience typical adolescent changes in mood and outlook”

It is therefore extremely important to anticipate menarche, as it may be an especially traumatic event in those with intellectual impairment. Early education for parents in the area of menstruation and management could be very helpful in preventing anxiety. Supporting the family during this time of development combined with education in relation to sexuality and contraception for both boys and girls is required. A prospective longitudinal study on this subject would be beneficial in identifying appropriate education requirements and management needs, and would support the development of medical and psychosocial management programmes for young adolescents with Down syndrome and their families..

4.4 Congenital Heart Disease

There was a high incidence (45%) of congenital heart disease found in this study with no gender difference. The most common defect found (29%) was complete atrioventricular septal defects (AVSD) which is similar to a prospective study by Tubman et al (1991) in Northern Ireland on 81 babies who found 38% with AVSD. However other studies show varying incidences including a study by Cullen et al (1990) in the Republic of Ireland of 114 children with Down syndrome reported 14% with AVSD. A study by Frid et al (1999) in Sweden of 219 children with Down syndrome reported 42% had AVSD. The next most common cardiac anomalies we found in this study were ASD (21%), VSD (18%), PDA (13%) and ASD VSD (8%). In addition 43 (11%) children had innocent heart murmurs associated with normal cardiac investigations.

Diagnosis and treatment of children with congenital heart disease has dramatically changed over the past few decades. Amark et al (1999) in a retrospective study of cardiac anomalies over a period of 16 years in 115 children with Down syndrome reported that changing attitudes in society and widespread use of echocardiography as a screening method made it possible to plan earlier correction and significantly improve management of heart disease in this group. Fortunately, due to major improvements in health care the majority of congenital heart lesions in children with Down syndrome can now be successfully treated, either medically or surgically. As a result life expectancy in the first year of life has greatly improved in this population and survival to age 10 years rose from 37% in 1958 to 90% in 1997 (Hayes 1997).

Some babies with significant cardiac defects may have no clinical signs or symptoms in the first few weeks of life. Cullen (1990) in a study of 114 children with DS reported that, “despite frequent attendance at clinics, cardiac defects had been overlooked in 14 cases”. Therefore a high index of suspicion regarding cardiac disease should be maintained throughout life even where initial screening has been negative and particularly in those where cardiac echo has not been performed. Tubman et al (1991) found that individual examination methods were insensitive (the sensitivity of: - clinical examination was 0.53, radiography 0.44, and electrocardiography 0.41) but sensitivity improved when the three techniques were combined. Frid et al (2004) in a retrospective study of 801 children with AVSD in Sweden reported a significant reduction over time in age at operation, and also that surgical correction is now equally successful in patients with DS and the general population for both genders. This study found that the majority of children (85%) were diagnosed at birth or within 6 weeks of birth. However, a further 7% were diagnosed between 6 weeks to 6 months of age, with smaller numbers being diagnosed between 1 and 6 years and one child at 14.5 years.

The presence of sleep apnoea is common in children with Down syndrome, particularly in boys. In a prospective study of 108 children with Down syndrome de Miguel-Diez (2004) reported a prevalence of 54.6% with sleep apnoea and a significantly higher prevalence in boys (64.7%). In an earlier study of 5 children with Down syndrome and associated congenital heart disease Loughlin (1981) reported that in three children nocturnal hypoxemia, secondary to sleep-apnoea, significantly contributed to their pulmonary hypertension. Early detection of sleep apnoea in those with cardiac disease is essential and may help prevent complications such as

pulmonary vascular disease developing that could adversely affect the outcome of cardiac surgery (Tubman 1991).

Down syndrome is associated with an increased incidence of periodontal disease and as with all children with cardiac anomalies, and particularly patients with pulmonary hypertension, there is a risk of infective endocarditis. In this study oral hygiene was found to be normal in 89% of the children however 6.6% had poorly managed oral hygiene. In addition clinical signs of gingivitis were found in 3 (0.8%) children. A special consideration in the delivery of their dental care together with antibiotic prophylaxis to prevent further medical complications is therefore important.

4.5 Thyroid dysfunction

This study demonstrated a prevalence of thyroid dysfunction in 46 per 1000 individuals with Down's syndrome. There were 6 new cases of acquired hypothyroidism detected during the study giving an incidence of 1.5% and a prevalence of 4.1%. All new cases of hypothyroidism were found in children from 7.7 to 16.1 years of age. The lack of new cases of hypothyroidism in the younger age groups might have been explained by smaller numbers presenting for fingerprick screening in this group. However, that was not the case in this cohort as 118/163 children were screened under the age of 7 years. However, we did find that 6 children had been previously diagnosed with hypothyroidism from age 0.8 months to 7.1 years. Hypothyroidism distribution was greater in females than in males (ratio of 3:1), occurring from infancy to adolescence. Thirteen patients were thyroid antibody positive and two of these patients were aged less than 2 years. The significance of the role of thyroid antibodies in hypothyroidism is as yet unclear and they were generally

thought to be present only in children over the age of 2 years (Karlsson 1998, Lobo 1980, Loudon 1985).

This study further supports the feasibility of TSH screening by finger prick in those with Down syndrome as found by Noble et al (2000). Parents reported that the procedure was a far more acceptable test for their children because it was extremely quick, non-invasive and caused very little distress to the child. The high proportion 3/38(7.9%) of those with acquired hypothyroidism under the age of 2 years found in this study has not been noted before and supports the need for regular screening from early childhood. This may be particularly important in the preschool age group (under 5 years of age) when growth and development are critical as 119 (30%) children in our study were under 5 years of age. Consequently this method could overcome the reluctance by parents and health professionals, as seen in this study, to undertake routine venepuncture because of the difficulty in performing the test and the trauma caused to the child and would promote increased parental acceptance and participation in ongoing biochemical screening.

Only 6 of the 16 children diagnosed with hypothyroidism had presenting associated clinical signs; increased weight, fatigue, constipation and reduced growth. This supported the findings that the diagnosis of hypothyroidism on clinical grounds in Down syndrome is difficult and unreliable (Noble 2000, Prasher 1995, Mani 1988, Lobo 1980, Steward 1994) particularly in the earlier age groups as the 3 children (2 males, 1 females) under 2 years of age who had been diagnosed with hypothyroidism had no presenting clinical features.

However, clinical suspicion remains important for hypothyroidism as demonstrated by case 4 Table 2, diagnosed with hypothyroidism at 2.1 years of age who had presented with clinical signs of constipation and poor growth together with a plasma TSH level at the upper normal level and reduced plasma total T4. In this instance hypothyroidism was confirmed on TRH testing.

There were two cases of hyperthyroidism previously diagnosed, giving a prevalence of 0.5% in this cohort, similar to findings reported by Pueschel (1991) 0.6% but less than by Karlsson (1998) 2.4% and by Prasher (1995) 3.1%. This further confirms the increased risk of developing hyperthyroidism in Down syndrome and the importance of clinical vigilance, as this condition cannot be diagnosed by capillary TSH sampling. Particularly as only one child diagnosed with hyperthyroidism had presenting clinical features of exophthalmos.

Conversion from compensated hypothyroidism to uncompensated hypothyroidism can be rapid in children and adolescents with Down syndrome (Stewart 1994, Noble 2000,). Stewart reported on a longitudinal study in which two children with DS, aged between 5 and 16 years, had converted to uncompensated hypothyroidism within one year of screening. Noble et al (2000) reported 4 children with Down syndrome to have converted from compensated hypothyroidism to uncompensated hypothyroidism within a period of 3 to 12 months of initial screening. Two children in this study converted to uncompensated hypothyroidism within 7 and 8 month periods respectively of previous blood tests. Case 5, Table 3.101 (page 125) newly diagnosed with hypothyroidism had a previous slightly elevated plasma TSH (6.2 mU/l) with lower border T4 and positive antibodies at 11.8 years of age and subsequently

developed uncompensated hypothyroidism 7 months later. Case 1, Table 3.102 (page 126) previously diagnosed with hypothyroidism had a slightly elevated TSH and normal T4 results 8 months earlier. These rapid conversion reports further support the need for ongoing biochemical screening.

Type 1 diabetes mellitus is often associated with additional autoimmune phenomena and previous studies have shown that when it coexists with Down syndrome there is a higher risk of developing hypothyroidism (Griffin 1997). This study found 3 of the 15 (20%) children with hypothyroidism also had Type 1 diabetes mellitus. This further supports that additional thyroid screening vigilance is required for children who have both DS and diabetes mellitus because of their increased risk of developing hypothyroidism (Daniels 1968, Griffin 1997).

This study shows that there was a poor level of awareness among health professionals and parents of the increased risk of hypothyroidism in children with Down syndrome at all ages but particularly in the early years of growth and development. The median age of those screened after the neonatal screening programme was 7.5 years (range 1 to 16.4 years). Parents also reported that 197/305 (64.6%) children had not been screened since neonatal screening (age range 1 to 17 years), this represents (50%) of the cohort. In the year prior to the study only 85 (21.6%) children had been screened for thyroid dysfunction by their family doctor or paediatrician. In addition of those previously diagnosed with thyroid dysfunction 3 were screened as part of a routine follow up management of Type 1 diabetes mellitus, 4 based on presenting clinical features and only 5 on routine screening.

While the sensitivity of whole blood TSH screening by finger prick sampling cannot be judged on this and recent studies further longitudinal surveillance is required in order to identify the true prevalence of thyroid dysfunction and to provide more detailed information on the conversion time from compensated to uncompensated hypothyroidism in this population. However, the high identification of new cases gives further evidence that the method is likely to be very sensitive. Resources for the provision of the service for annual screening are urgently needed for people with Down syndrome.

The United Kingdom guidelines recommend screening at birth, then at least every 2 years from one year of age or annually using dried blood spot TSH measurement (Marder 2001). The Australian and United States of America guidelines recommend biochemical screening at birth, six months, one year and then annually throughout life (Leshin 1998). In view of the possibility of rapid progression of hypothyroidism as shown in some patients in this study we recommend thyroid screening at birth, at one year and then every 2 years by venous blood sampling or annually by finger prick when available. Children with marginally elevated plasma or whole blood TSH levels or positive thyroid antibodies should be tested more frequently than those with normal results.

4.6 Vision

This study recorded an increased incidence of vision disorders (50%) the most common condition being refractive errors in 25% of the children. This was not unexpected as previous studies have reported similar findings. Caputo (1989) in a

retrospective study of 187 patients with Down syndrome over a 10 year period reported that 22.5% had short-sightedness and 20.9% long-sightedness. In a study of 77 children, referred for evaluation by a paediatric ophthalmologist, Roizen (1994) reported that 61% had vision disorders requiring treatment and monitoring. Poor ability to adjust their eye to variation in distance (accommodation) is a common feature in Down syndrome, regardless of the target used and the etiology of the deficit is not yet known. In a study by Woodhouse et al (1993) it was reported that defective accommodation is common in children with Down syndrome even at 3 months of age. In a more recent comparative study by Woodhouse (2000) it was found that children with Down syndrome showed considerably poorer accommodative responses than normally developing children and concluded that it is important that clinicians are aware that near vision is out of focus for children with Down syndrome.

In this study we found 18% of children had strabismus and 4% underwent corrective surgery. A study of 55 children with Down syndrome (mean age 7.4 years) by Berk (1996) in Turkey reported 21.8% with strabismus, while Shapiro & France (1985) found 13% in a study of 53 children (mean age 17.4 years) and Suyugul (1992) reported 31% in a study of 44 children (mean age 4 years).

Cataract were found in 4.6% of children in this study of whom 4% had undergone corrective surgery. A higher incidence of 20% was reported by Berk (1996) and 15% by Suyugul (1992). In addition in this study we found that nystagmus was present in 66 children (16.8%) which compared well with a study by Wagner (1990) in America who reported nystagmus in 19.8% of 188 patients with Down syndrome, and with 11% of 140 with Down syndrome in China (Wong1997). A recent retrospective study

of 58 children by Bromham (2002) in the UK reported an incidence of 19% with nystagmus and also explored the possibility of a connection between nystagmus and congenital heart defects in those with Down syndrome. He found that 9/11 children with nystagmus also had heart defects and that those with heart defects were more likely than those without defects to develop myopia and nystagmus in the first 9 years of life. Myopia and nystagmus can occur following damage to the visual pathways. The clinical association between endocardial cushion defects and Trisomy 21 suggests that genes located on chromosome 21 contribute to the development of the heart (Klewer 1998). An association between the COL6A1 gene region and congenital heart defects in Down syndrome has been reported by Davies et al (1995). COL6A1 codes for part of collagen VI, a component of many ocular tissues (Marshall 1993). In this study we found a significant correlation between 63% of those with nystagmus and heart defects (22 boys, 20 girls; age range 2 to 19 years) (p value = 0.027). This supports the finding by Bromham that those with Down syndrome and heart defects are more likely to develop nystagmus possibly due to the additional genetic material during development. Bilateral congenital cataract were found in only one child in our study and corrective surgery at 3 weeks of age had been performed. In addition this child has glaucoma in both eyes requiring ongoing management. A study of 5 children with Down syndrome with bilateral infantile glaucoma by Traboulsi (1988) reported that delay in management predisposes to poor vision outcome.

Keratoconus is also common in those with Down syndrome where the cornea becomes conical in shape. An incidence of 5.5% was reported in a study of 140 patients by Cullen (1963); and 15% were found by Shapiro and France (1985). No child was found to have keratoconus in our study. However, keratoconus is a major

cause of blindness in patients with Down syndrome and more likely to develop in late adolescence. Early detection and corneal transplantation may be successful which would undoubtedly improve the quality of life of the patient (Volker-Dieben 1993). A study by Vincent (2005) in Canada has reported that computerized videokeratography (CVK) is a useful tool in the ocular assessment of patients with Down syndrome for keratoconus.

Nasolacrimal duct obstruction is a common problem affecting up to 20% of newborns (MacEwen 1991) and is overrepresented in those with Down syndrome (Roizen N 1994). Midfacial hypoplasia leads to increase risk of nasolacrimal duct obstruction with subsequent tearing (DSMIG 1996) The majority (>90%) of congenital nasolacrimal duct obstruction will resolve spontaneously in the first year of life (MacEwen, 2001, Bashour M 2005,) and 60% of the remainder will resolve during the following year (MacEwen 2001). Thirteen children (4%) in this study had their blocked tear ducts cleared under anaesthetic with good effect.

Parents also reported that 39.3% of the children were wearing glasses and that 19.5% were wearing the glasses all day. Previous studies have shown that as many as 50% may require glasses (Turner 1990). It was also found that the majority of children (65%) were screened by an eye specialist.

This study showed that 52% of the parents had concerns relating to their child's vision and that while 77% had received vision screening 21% reported that their child had not been screened. Visual disorders can occur at any age in this group and in particular as the patient gets older high myopia (near sightedness) can occur (Catalano

1990). A study of 77 children by Roizen (1994) reported that the percentage of children with identified vision disorders increased with age from 38% in the 2 to 12 month old group to 80% in the 5 to 12 year old group. Children with Down syndrome, as with all children, will require a variety of visual testing, which depends very much on their developmental age. Some will require more specialized visual tests. A distraction free environment will optimise performance.

4.7 Hearing

Hearing impairment screening was performed on all children in order to assess the hearing function of children and adolescents with DS from a large representative sample, using otoscopy, tympanometry and DPOAE. Pure Tone Audiometry (PTA) was not performed on all patients in the study because of time constraints and the fact that this would have required a full time Audiologist. The high percentage of children (64%) with abnormal otoscopy findings was not unexpected and is consistent with previous studies. Brooks 1972 found 30% with wax in of his cohort. Similar outer ear abnormalities were found by Maroudias 1994 in 40% of the patients in a study of 120 children in Greece. Whilst Hassmann 1998, in a study in Poland, found that all 45 children studied had impacted wax. This study found that patients with significant cerumen were more likely to fail DPOAE screening and that the majority of children who had clear or mild wax were more likely to pass on DPOAE screening, thus illustrating the need to carry out otoscopy prior to screening by DPOAE. This identifies an important treatable health care problem that warrants close monitoring, intervention and parental education.

Impaired middle ear function, demonstrated by tympanograms, was detected in 58% of ears and is comparable with other studies including; Brooks 45%, Fulton and Lloyd 54.8% and Hassmann 89%. With otological assessment and treatment this impairment could be greatly reduced. The proportion of tympanograms of type As (mild middle ear impairment) in this study was 8.2% which was similar to 9% found by Schwartz 1978 in a study of young children, but less than 14.8% found by Keiser 1981, 33.3% found by Hassmann et al. and 30.8% by Dahle 1986 who suggested that this middle ear dysfunction may be due to ossicular chain stiffness.

Only 11% (58 ears) of children fulfilled all DPOAE 'pass' criteria across all frequencies (1 to 8 kHz) thus indicating full normal cochlea function. This was less than the 45% found by Hassmann when he screened 36 ears of 44 children, however, screening was only attempted in his study following removal of impacted cerumen, which could explain the higher number of passes. The DPOAE procedure requires transmission of stimulus and evoked emission through the ear canal and is very susceptible to changing conditions in these areas. Therefore, removal of any wax improves screening result levels.

A further 29% (158 ears) 'passed' 2 octaves (missing 1 octave). These children however, may have normal hearing function as previous studies by Gorga (1997) and Stover (1996) have shown that DPOAE can sometimes perform more poorly at 1-2 kHz, as background noise is sometimes greater in the lower frequencies, which can surround the DP emissions. However, high frequency loss (4-8 kHz) indicating sensorineural hearing loss has been seen to occur in those with DS in studies by

Buchanan and Widen. Therefore, failed DPOAEs at this frequency require further investigations.

DPOAE versus tympanometry: Patients with normal middle ear tympanograms were more likely to pass on DPOAE screening. However in this study we also found that seventeen ears with failed tympanograms (Type - As, B, C and Cs) passed all DPOAE criteria indicating normal inner ear function on screening. This was not expected as the DPOAE procedure requires transmission of stimulus and evoked emission through the middle ear and is very susceptible to changing conditions in these areas. However, on further examination the mean amplitude of these DPOAE emissions were reduced when compared to the mean amplitudes of children with normal middle ear function (A-Type tympanograms). This could indicate that these patients may have had fluid rather than glue ear (where the fluid has become more viscous/thick mucous) which allowed the passage of sound and resulting responses at the lower amplitude. Thus indicating that DPOAE as a screening tool should always be preceded by tympanometry to assess middle ear function and thereby reduce the level of false positive results.

When comparing DPOAE Test 1 (65/55 dB SPL) with DPOAE Test 2 (65/50 dB SPL) responses rated higher overall for Test 1 in differentiating normal versus abnormal inner ear function. This was not unexpected as previous studies, by Stover (1996) on 210 patients and Gorga (1997) on 805 patients, reported that Test Level 1 elicited responses that separated normal from impaired hearing better than either lower level or higher-level stimuli.

A comparative study on group of 20 children with Down syndrome and 20 age matched children without Down syndrome was performed in order to assess the reliability of screening inner ear function by DPOAE when compared to PTA. The high incidence of hearing impairment found in 15 children (75%) with Down syndrome was not reflected in the control group who all had normal hearing.

Outer ear pathology was high (55%) in those with DS in comparison to only 5% in the control group. This was not unexpected and is consistent with previous findings. In a study of 55 people with DS Brooks (1972) found 30% to have severe wax, while Maroudias (1994) in a study of 120 children with Down syndrome found 40% had abnormalities on otoscopic examination. Also Dahle and McCollister (1986) found that 31% of the 30 children studied had impacted cerumen, which resulted in an average 24 dB hearing loss. Build up of wax can reduce hearing and warrants close monitoring as this condition is treatable and preventable.

Abnormal middle ear function was found in 60% of the children with Down syndrome. This was similar to a study in America of 22 children with Down syndrome by Roizen (1994) who reported that 64% had untreated middle ear pathology, while Brooks (1972) found 45%, Fulton and Lloyd (1968) in a study of 79 people with Down Syndrome reported 54.8% had abnormal tympanograms. In addition the study by Hassmann (1998) of 27 children with Down syndrome found 56% had middle ear pathology. In view of this high incidence of middle ear pathology it has also been suggested that otoscopy and tympanometry might even be performed twice a year in those with Down syndrome because of the high incidence of acquired abnormalities throughout their lifespan (Evenhuis H M. 1996). In a recent

5 year longitudinal study in America of 45 children with Down syndrome Shott (2001) reported that after treatment of easily reversible hearing loss from chronic otitis media (either medically or with surgical treatment with grommets) 98% of the children had normal hearing levels at the end of the first year of the study.

Only 15% of the control group failed tympanometry and the abnormal findings in 2 children related to a perforated tympanic membrane in one boy, which had occurred during a swimming lesson and possible poorly functioning Eustachian tubes in one girl bilaterally.

In addition ear canal volume was assessed in both groups and this study found that the mean ear canal volume of those with Down syndrome was significantly smaller than the control group B who had larger ear canal volumes. However, this difference, which may have been affected by the presence of wax in the ear canal, was less marked in the older children. It is not known whether smaller ear canal volume affects the child's ability to hear or their quality of hearing, or whether this may have any possible impact on speech development. This is another area that would benefit from further research to assess any difference in ear canal volume after the removal of ear wax.

Screening by Pure Tone Audiometry showed that 6 children with Down syndrome had normal hearing bilaterally and 5 unilaterally. However, hearing impairment was found in 6 children bilaterally and 6 unilaterally and ranged from mild to severe. Two children had sensorineural hearing loss (SNHL), one child bilaterally and the other child unilaterally. One child was newly diagnosed with severe SNHL in both ears

which was identified as hereditary deafness. The second child had contracted chickenpox at a very young age, which resulted in nerve loss in one ear. In comparison normal hearing levels were found in all controls. It was not possible to assess the children with Down syndrome over all frequencies by PTA due to the children's inability to cooperate. Only 10 children were screened from 1- 8 kHz, while 7 children were screened from 1-4 kHz and 1 child at only 2 kHz. The two children who could not be screened by PTA were both 8 years of age. A previous study by Maurizi (1995) demonstrated that children with Down syndrome over 8 years of age could produce reliable PTA results whilst children in the general population can co-operate from approximately 3.5 years of age. All those in the control group could co-operate over all frequencies 1-8 kHz.

Screening by DPOAE found that 6 children with Down syndrome had normal inner ear function across all frequencies (2 bilaterally, 4 unilaterally); with 7 children passing 2 frequencies (all unilaterally) and 11 children passing one frequency (3 bilaterally, 8 unilaterally) whilst 5 children failed to pass any frequency (1 bilaterally and 4 unilaterally). DPOAE assessment was not possible due to limited co-operation in 2 children on the day of screening (one child 8 years and the second child 11 years of age). In comparison 19 children in the control group were found to have normal inner ear function when screened by DPOAE across all frequencies (18 bilaterally, 1 unilaterally) and 2 children passing 2 frequencies (1 bilaterally, 1 unilaterally). These findings further suggest that those with significant outer and middle ear abnormalities are more likely to fail a DPOAE examination and the majority of children who had clear or mild wax on otoscopy were more likely to pass on DPOAE screening.

In this study statistical analysis measuring agreement between PTA and DPOAE screening evaluations of those with normal outer and middle ear findings showed significant agreement (Kappa: value =1, n=27 children (50 ears)). However when all children with abnormal outer and middle ear pathology were included measurement agreement results were significant but less so (Kappa: value .77, n=37 children (73 ears)). Thus indicating that, with normal outer and middle ear pathology, DPOAE screening results reflect 100% PTA results, however, when abnormal outer and middle ear findings are present DPOAE screening results reflect 80% of PTA results. A study of 806 subjects without Down syndrome by Gorga (1997) suggested that a relation exists between DPAOE measurements and audiometric threshold, such that it would appear that DPAOEs could be used to predict the magnitude of hearing loss.

In addition, statistical analysis measuring the mean DPOAE amplitude results of both groups with normal outer and middle ear function showed they were significantly lower at frequencies from 1 to 4 and 8 kHz in group A in comparison to those in group B. However, the numbers were small and further research in this area would be desirable. What affect this has on hearing quality in those with Down syndrome is not known and will hopefully become part of future research, however, Kimberley (1999) in a study of the multiple clinical applications of DPOAE to an otological practice, has suggested a relationship between pure-tone thresholds and DPOAE responses and that there may be promise of application to novel approaches to the design of hearing aids.

DPOAE screening is limited if there is external and middle ear abnormalities thus the DPOAE technique may have limitations as a screening tool in this population.

However, children with external and middle ear pathology, once treated, could be candidates for DPOAE screening. Therefore, all children should be screened by otoscopy and tympanometry prior to DPOAE screening. A longitudinal study of 48 children with Down syndrome by Shott (2001) reported that “aggressive, meticulous diagnosis and treatment of chronic ear disease in children with DS”, started soon after birth, found 98% had normal hearing levels when screened at the end of year one which is higher than found in previous studies. Further similar studies with control groups are required and would be very beneficial.

Children unable to cooperate for screening require additional time to allay their fears. Some children may require sedation and as DPOAE requires no patient feedback, it can be performed following onset of sleep as demonstrated by Gorga (1995). In his case study a 3 month old child with special needs was sedated with chloral hydrate following which objective tests were performed during sleep and robust DPOAE emissions were observed in both ears.

Historical reports from parents have further confirmed previous reports that those with DS are at high risk of hearing impairment. The median age of first hearing screening was 1.5 years (range 2 months to 14.5 years) in the Eastern Health Board Area.

Studies by Berman (1981) and Downs (1985) indicated that fluctuating hearing loss, even mild, in early childhood may change the sound of acoustic cues and interfere with the child’s ability to recognize speech sounds. Also Lewis (1976) and Yarter (1980) suggested that listening strategies may be permanently affected by otitis media

and that language deficits cannot be overcome unless special remediation is implemented. In addition Yoshinaga-Itano (1998) has identified that the auditory neural pathway continues to develop by external stimulations after birth and the most sensitive time for this final development is in the first year of life. Therefore, aggressive ongoing management of outer and middle and inner ear pathology is desirable from infancy and right through childhood and adolescence in those with DS. Some children with DS may need to be monitored every 3 months by an otolaryngologist in order to assess and keep external canals free from cerumen. Hearing losses need to be clarified by type (conductive, sensorineural or mixed) and treated accordingly. Another advantage of earlier screening would be the possibility of introducing hearing aids at an earlier age where necessary, which could allow better uptake and cooperation in their use.

4.8 Orthopaedics

Orthopaedic disorders are commonly seen in those with Down syndrome (Jacobsen 2000) and are associated with increased joint laxity, reduced muscle tone and abnormal skeletal development (Diamond 1981). Areas of major concern include cervical spine, hip, patellofemoral instabilities, and foot deformities (Winell 2003). In this study we found that 71% of children had orthopaedic problems including atlanto axial joint instability 1%, hip joint instability 2%; knee joint laxity 2%; ankle joint instability 2% and the most common (63%) finding was pes planus (flat feet).

Previous studies have reported atlanto-axial instability as determined by radiographic evidence of an atlanto-dens interval of 5 mm or more occurs in 10% to 30% of patients with Down syndrome by adolescence and that the youngest patient was 3

years old. In addition an occurrence of atlantooccipital instability has been reported in 6 to 63% however the majority were asymptomatic with only 11 symptomatic and all were over 1 year of age Trumble (1994). A recent report of atlantooccipital subluxation was the first case in a neonate with Down syndrome following surgery for repair of a large PDA and suggested the need for increased awareness Winell (2003).

A longitudinal study by Burke (1985) of 32 patients with Down syndrome over 13 years found that one patient had instability at the start of the study and 8/32 were unstable at the final follow-up. Pueschel (1987) also reported 7/95 patients developing atlanto-axial instability over a period of 3 to 6 years indicating that atlanto axial instability is difficult to diagnose and may be acquired.

The value of lateral cervical spine flexion and extension radiographs has been conflicting. Selby (1991) reported in a study of 135 children with Down syndrome (6-14 years of age) that repeat radiographs of the cervical spine (atlanto-axial distance threshold value >4.5mm abnormal) were unreliable in identifying atlanto-axial subluxation in this group. While Cremers (1993) in a study of 400 children with Down syndrome (6 to 17 years of age) reported that repeated radiographs (atlanto-axial distance >4mm as abnormal) showed that there is agreement in repeated radiographs when taken in the flexion position but not in the neutral position. A study by Pueschel (1992) of 141 patients found that 11 patients had abnormal findings on radiograph, however 9/11 had subsequent normal radiographs on follow up screening. In this study we found 4 (1%) children diagnosed with atlanto-axial instability and 3 (0.8%) children had undergone surgical intervention and 1 child did not require surgery. The mean age of diagnosis was 4.8 years (range 3.5 to 7 years).

While the risk is small, to avoid injury of subluxation in 1984 the American Academy of Pediatrics adopted a recommendation of routine radiographic examination before participation in the Special Olympics and that children with Down syndrome and radiological instability of more than 4.5 mm should be barred from participation in sport activities that put them at risk for subluxation. However, this recommendation was reversed in 1995, due to the findings of research studies. The issue remains controversial. Although atlantoaxial instability may lead to diplegia, hemiplegia, or torticollis (Davidson RG 1988, Powel JF 1990) hardly any of these disorders are associated with sport (Grosbovshchek M 1985). In fact infections are implicated more often (Cremers M 1993). Cremers (1993) looked at 400 children with Down syndrome aged 6 to 17 years and compared those participating in sports and those stopped from participating in certain sports. The atlantoaxial distance was more than 4 mm in 91 children (32%) of the subjects; with 75% between 4 and 5 mm; 22% between 5 and 6 mm; and 3% more than 6mm, with a maximum of 6.5mm. They were divided into two groups and reviewed over one year. They found that limiting the sports activities of children with Down syndrome and atlantoaxial instability for one year did not result in a lower rate of neurological signs related to spinal compression at the cervical level. They also found that the functional performance of the children in the control group who continued their usual sport activities was, if anything, better than that of children in whom sports activities were restricted. They reported that, based on their findings, that there is no reason to stop children with Down syndrome from taking part in certain sports activities (Cremers 1993). The four basic objectives of a sports programme for people with special needs are: health and fitness, progressive levels of achievement, social success and rewards (Seekins

NL 1984). Cremers concluded that playing sport does not have a negative effect on the health of children with Down syndrome and does not worsen atlantoaxial instability. Beneficial effects of sports include improvements in stamina, muscle strength and endurance, speed, agility, flexibility, and weight control (Bar-Or O. 1983).

Hip disorders have been shown to occur in 8% to 28%% of those with Down syndrome. (Kioschos M 1999). There is an increased incidence of dislocation, dysplasia, slipped epiphysis, Perthes' disease and avascular necrosis in individuals with Down syndrome. In a study of 114 patients with Down syndrome in America (age range 10 to 43 years) Shaw (1989) reported that 9 (7.9%) had some hip abnormality, including dysplasia, dislocation, avascular necrosis, or slipped capital femoral epiphysis and Perthes' disease. However, hip instability at birth is rare and should be treated along normal lines (Jacobsen FS 2000). In this study we found that only one child was diagnosed with hip instability at birth and was managed conservatively with a Pavlik harness up to 5 months of age. This study also found 8 (2%) children were diagnosed with hip instability yet none required surgical intervention. This is less than seen in other studies, as Lawhon (1990) reported 9/162 (5.6%) (aged from 1 month to 63 years) in non institutionalized patients. Diamond (1981) reported 12/265 (4.5%) and Shaw(1992) reported 8/114 (15%) (10 -19 years of age) patients with hip instability. However their studies included case records, clinical examination and radiographic evaluations. Shaw (1992) reported that hip instability typically presents between the age when the child starts to walk and 10 years of age. The increased mobility of the hip in those with Down syndrome may contribute to the increased incidence of hip abnormality. It has been postulated that prolonged sitting

in the external rotation posture, which is common in this group, may place increased mechanical stress on the growth plate and lead to epiphyseal slipping (Shaw 1992) In this study we found one child diagnosed with Perthes' disease at 2.8 years. Previous studies have shown a slightly higher incidence of Perthes' disease, Lawhon (1990) 4/162 (2.5%), Makley 3/110 (2.7%) and Shaw (1979) 1/114 (0.8%). Treatment does not differ from the general population with the same problem.

Most patients with Down syndrome show some abnormal motion at the knee joint (Jacobsen 2000) where the patella dislocates due to increased laxity of the ligaments. The instability can vary from occasional to permanent dislocation. In this study we found 11 (3%) children had knee joint instability and 3 required surgical intervention. This compares well with a study by Diamond (1981) who reported 4% of cases while Mendez (1988) in a longitudinal study of 252 people with Down syndrome reported 8% of cases with patella instability.

Foot deformities are related to general laxity of the ligaments in those with Down syndrome and are generally well tolerated (Winell 2003). Lawhorn (1990) in a study of 163 patients with Down syndrome reported that 80% had pronated flat feet, half of whom required some form of treatment. Decreased ambulation and/or walking with a limp were common symptoms related to foot problems. While his study population ranged from 1 month to 63 years of age the proportion of children under 18 years was not described therefore it is not possible to know what percentage of children or adolescents had problematic flat feet. This study found that while only 11 parents reported that their child had flat feet. Clinical assessment found that 63% had flat feet when observed walking barefoot and standing up on toes. Unexpectedly only 9

(2.3%) children reported any discomfort, of whom 8 required physiotherapy and 1 required special insoles.

Scoliosis is another problem that occurs in approximately 50% of individuals with Down syndrome (Diamond 1981) and was found in only 4 (1%) children in this study with one child requiring corrective surgery. Most curves patterns are thoraco-lumbar and can cause some spinal imbalance (Lawhorn 1990). However, it has been reported that the curves rarely reach magnitudes which warrant treatment (Jacobson 2000).

Orthopaedic problems in those with Down syndrome may be overlooked due to our failure to recognize their frequent occurrence and the patient's deficient communication skills and inability to identify pain (Jacobson 2000). With their increasing lifespan, those with orthopaedic problems will require early identification and orthopaedic intervention to maintain mobility and improve and maintain long-term quality of life.

4.9 Speech and Language

A very high proportion of children (94.5%) were found to have speech impairment in this study whilst only 5.5% had clear speech appropriate for their age. Language is the most sophisticated of human cognitive skills. Speech and language delay in children with Down syndrome has been suggested to be due to deficits in both speech and language skill development by Fowler (1990) and Miller (1988), however, no single cause has been identified to explain their poor speech and no speech and language problem is unique to children with DS. Spoken language utilizes well formed syllabic units, termed canonical syllables by Oller (1986) who proposed that

the production of significant proportions of canonical babbling marks the entrance of an infant into a developmental stage of meaningful speech. A study by Lynch (1995) reported that infants with Down syndrome are delayed in this process in comparison to the general population. Language learning is a lifelong process but the majority of children lay the foundations for language in the first three years of life (Ni Cholmáin 2002).

Once the child with Down syndrome develops a vocabulary of several words, the vocabulary tends to increase at a slow pace and does not usually show the exponential increase observed in a typical child. Miller (1995) documented individual differences in vocabulary size and vocabulary acquisition in children with Down syndrome and reported that 35% exhibited rates of vocabulary growth consistent with their mental age while 65% exhibited vocabulary growth below 10% for mental age. He found that some children with Down syndrome with a mental age of 24 months had a vocabulary of 250 words while others had a vocabulary of only 10 words. In this study we found that the majority of the children were communicating with between 1-2 and 4-5 word sentences (age range 2 to 18 years) indicating very limited speech ability over the age of 2 years (Cunningham 1988) (Down's syndrome – Child Development Chart DSMIG 2000) (Appendix ???_

Reports of language development have indicated that 60% - 70% of the children with Down syndrome used signing to support communication. In this study we found that 43% of children were signing, the majority using Lámh sign language and a smaller proportion used Makaton with Lámh, while only one child used Lámh and the I SL. Also a small number of parents expressed concern that their child would never

develop speech and would continue to use only sign language. Miller et al (1998) recommends that, during the early periods of development, intervention methods focus on oral motor development and that all communication attempts be encouraged regardless of the mode of communication used. Signing, printed material for reading, and writing should be incorporated into the program. Chapman et al (1998) recommended that to maximize an individual's communicative effectiveness, speech therapy must address comprehension, expressive speech and intelligibility. Buckley (1994) found that children as young as 3 years are able to "read" aloud whole words indicating that the medium of reading can support the development of speech and language from an early age and he suggested that development of expressive language, whether by signing or speech should be the most important focus of therapy.

Children need to be able to hear in order to develop verbal language. Children with Down syndrome are at a greater risk of hearing impairment as have been found in this study. This, if undiagnosed will restrict language learning opportunities (Cholmáin 2002) Yoshinaga-Itano (1998) compared 72 deaf or hard-of hearing children whose hearing losses were identified by 6 months with 78 children whose hearing losses were identified after the age of 6 months. She reported that significantly better language development was associated with early identification of hearing loss and early intervention. Recent literature (Gravel and Wallace, 1995) found a strong relationships between otitis media with effusion (OME), language development, and academic achievement in typically developing children. A recent 5 – year longitudinal study of 48 children with DS by Shott (2001) has reported that "aggressive, meticulous and compulsive diagnosis and treatment of chronic ear

disease in children with DS, started soon after birth, provides significantly improved hearing levels, one year later, than previously reported". This is an area where prospective randomised control trials would be most desirable and very welcome. Treatment of intermittent hearing loss due to impacted wax, middle ear pathology and treatment of permanent hearing loss with hearing aids continue to be important not just in childhood but also in adolescence and adulthood. A study of 672 people in the Netherlands (Evenhuis HM, 2001) compared those with intellectual disability with and without Down syndrome under and over 50 years of age indicated unnoticed increasing hearing impairment problems with ageing requiring regular screening and treatment. Annual audiological monitoring for new or additional hearing loss should continue for the whole life of the individual with Down syndrome (Roizen 1997; American Academy of Pediatrics 1999).

Learning to communicate starts at birth and language acquisition of all children occurs gradually through interaction with people and the environment (Bowen C 1998). A longitudinal study by Mundy (1995) of 37 children with Down syndrome and 25 children with normal development found that language delay in those with Down syndrome appeared to be associated with problems in an earlier nonverbal phase of communication. Their findings indicate that the acquisition of nonverbal communication skills provides an important foundation for the emergence of language in children with and without Down syndrome. The basis for communication is social interaction, and certain conversational skills such as turn taking can be developed at a very young age through play (MacDonald 1989). There are many pre-language skills that can be addressed in treatment before the child is able to talk, therefore therapy should begin before the child speaks the first word (Kumin et al 1991). Speech is an

overlaid function in the human body. Feeding and respiration involve many of the structures and muscles used in speech. Many infants and toddlers with Down syndrome are very sensitive to touch (Kumin 1991). They do not want to be touched, don't want their teeth brushed, or do not like certain textures of foods. Therefore, early intervention to overcome this problem is desirable. (Kumin 1991).

Berglund (2001) studied 330 children with Down syndrome and a control group of 336 typically developing children undergoing early communicative and development therapy. He reported that those with Down syndrome by ages 3 and 4 years had spoken language comparable to the normative group at ages 1.4 and 1.8 years, thus indicating the great potential of early intervention. In this study parents reported difficulties in obtaining speech therapy throughout childhood. The location of speech assessments varied greatly with the majority (49%) assessed within the special developmental services; 23% at their local health centre by the area medical officers and the remaining by speech therapists outside these services. Parents also reported that 73% received speech therapy while 23% had not yet received any speech therapy. Thus, indicating different levels of assessment and management which could be very confusing for any parent. A very worrying finding was that speech therapy was not consistent as parents that many children had long periods of no speech therapy at all while waiting for replacement staff.

The ability to communicate effectively and social skills in children with Down syndrome which are very important for education, integration, work acquisition, reduction in depression, and improved quality of life. As children with Down syndrome have difficulty in developing expressive language and speech development

starts in infancy speech therapy from the earliest stages of development is important and promising (Berglund 2001) Kumin (1991) suggests that parents be provided with home activities so that speech and language experiences will continue in the home environment from birth. Vilaseca (2004) found in a study of 3 children with Down syndrome that language gains were best maintained after intervention if the mothers received at the same time specific advice about verbal interaction at home. In addition, in a cross sectional study by Chapman (2000) of 48 children and adolescents with Down syndrome in comparison to 48 control children reported that expressive language should be a continuing target of language intervention in the adolescent years. At present with the shortage of speech therapists it may be more appropriate to develop a way that parents are educated to support the delivery of speech therapy throughout the deferent developmental stages, with ongoing professional education, supervision and evaluation without overburdening them. This is an area of deep concern of parents and requires further research which would look at both service delivery and need of assessments and speech and language therapy and also how speech and language therapy can delivered at local levels throughout Ireland

4.10 Quality of Life

Over the past two decades, the concept of quality of life (QOL) has increasingly become a focus for research and application in the fields of education/special education, health care (physical and behavioural), social services (disabilities and ageing), and families.(Schalock 2004) Historically, the interest in QOL has come from many sources; the belief that scientific, medical and technological advances alone would result in improved QOL. This has now shifted toward an understanding that better QOL for the person, family, community and social well-being emerge not

only from advances in technology and medial care but also from change from the institutional-based services to community and individual's based services, together with a rise in consumer empowerment and patients' rights with emphasis on the person (Schalock et al 2002). Researchers have made significant progress in conceptualizing and measuring the QOL of individuals with development disabilities (Cummins 1997, Felce, 1997, Hughes & Hwang 1995, Schalock 1996, 1997, 2000). However, QOL of families who have a child with disabilities is at the early stages of research of the conceptualization process.

Within the field of special needs much focus has been placed on the characteristics of the person with special needs as the major stressors on families. However, it has also been suggested that the characteristics that are most likely to put families at risk might include relationships between care and supervisory demands, physical care, marital status and social support, which may relate to stress and are likely to be interconnected (Barnett 1995, Sloper 1991). A longitudinal study of family QOL would be ideal, however, in this cross sectional study family QOL was assessed by a short screening questionnaire. This included burden experienced by parents while rearing a child with special needs and also parent perceived services required to improve their child's quality of life.

The majority of parents (64%) in this study reported small to large burden due to the decreased time for themselves as a couple due to the additional time required to care for a child with Down syndrome. However, when comparisons were made, no significant influencing factors were found in relation to age, gender, family structure and work or health problems were found. A large proportion of mothers (61%) were

not working outside the home suggesting that the demands of caring for a child with Down syndrome required their fulltime input at home. The majority (93%) of fathers were working outside the home, thus indicating single incomes in most families with the possible reduction in socio-economic capacity. Barnett (1995) suggested that for mothers, the substantial shift from time devoted to paid work and social activities, to time spent in child care might reasonably be thought to alter the overall character of daily life as well as family finances. Sloper et al (1991) reported that lack of employment of mothers may have different implications socially, psychologically and financially with reduced opportunity for broader social relationships, interests outside the family and greater independence, which in turn reduces their ability to develop necessary coping skills. Trute (1988) reported that coping strategies are related to outcome measures of family functioning and Sloper et al (1991) suggested that coping strategies have an effect on mothers' reactions to potential stressors. Social factors such as income and severe difficulties with the domestic situation have been found to correlate with maternal depression (Mohan D, Fitzgerald M 1998). The high proportion of mothers working at home in this study may suggest the need for support services in order to allow wider flexibility in this area to give mothers the opportunity to work outside the home, gain confidence and allow them to widen their coping skills.

Parents in the majority of families reported small to large feelings of burden due to disruption to their family routines because of caring for a child with Down syndrome, but no association was found with age, gender, family size, parent/s working or not. However, the parents of 4 children with 3 additional disorders (Type I Diabetes, thyroid dysfunction and coeliac disease) all reported "large" burden due to disruption

to family routines. This might suggest that the added work and concern involved in caring for a child with Down syndrome with multiple health problems required much more time and effort from the parents and left less time for family routines. This may also suggest these parents required more healthcare and or community support than was available to them.

In addition, 67% of parents reported a small to large burden in disruption in the time for other children in the family. This study found that many of the health services were not co-ordinated and the child's needs were divided among various agencies and programmes. This necessitated parents travelling long distances, with medical appointments at many different hospitals, and sometimes with no associated links between the health and medical needs of the child or the overall management required. The need for more coordinated services to overcome these inconsistencies is therefore necessary. It also suggests that better supports for families under additional stress are required and further education may be necessary in order to enable parents mediate better with the services to develop these supports.

It was reassuring to find that 72% of parents reported small to no burden in caring for their child with regard to their child's physical problems requiring extra parental care. However, the parent's perceived major concern (23%) was their child's long term health needs. This may indicate an area parents find difficult possibly because it may be a time when they are no longer around to support, manage and look after their son/daughter. While this was not unexpected, as it was also found that the prevalence of medical problems associated with Down syndrome increased with age, it highlights the importance of identifying and meeting the needs of this vulnerable

group and further demonstrates the need for consensus medical management guidelines for adults with Down syndrome to support healthcare professionals and parents alike throughout the person's lifespan.

A large proportion of parents (63%) perceived that their child's general health at the time of the study was excellent and whilst it was reassuring that very few parents rated health as poor or very poor (1.8%) there were still 36% of parents who rated their child's health as only fair. Similar to general health, many parents (56%) rated their child's QOL as excellent with very few reporting poor or very poor (4.1%), however there was 40% of parents who rated their child's QOL as fair. It was interesting to find that the five children in the 'very poor QOL' bracket did not have life threatening medical problems, but they all had hearing and vision problems. However, while excellent reported QOL dominated in early childhood it was considerably less in the teenage years where parents reported lack of friends and very low community integration.

The transition from childhood to adolescence and adulthood covers less than twenty years but this involves time periods when individuals require different supports and different choices. Adolescence is a period of change that can create stress for both adolescents and parents. Adolescents with Down syndrome go through the same stages as other adolescents, but due to lack of cognitive and behavioural factors they and their parents may find this period particularly challenging (Merrick 2004). While there are special medical problems for this population that require annual medical examinations and surveillance, the focus shifts from health problems to social maturation, developing independence, and transition from school to employment or

work activity (Merrick 2004). Where healthcare is concerned medical transition from a paediatric to family physician provider is necessary. These demonstrate important areas that require further research and development of appropriate management programmes.

Research has shown that to improve the QOL of an individual with disability requires: emotional well-being, interpersonal relations, material well-being, personal development, physical well-being, self determination, social inclusion, and rights (Schalock 2002). However, individual QOL focuses only on what is happening with the individual, whereas the individual lives in the family setting and therefore what is happening with the individual has an impact on other family members and the family as a whole. Therefore improving individual and family QOL is beneficial to both. In a study of 187 families with and without children with disability Poston (2003) reported that improvement in family QOL incorporated two main domains: individual orientation and family orientation. Individual orientation was subdivided into six areas: advocacy, emotional well-being, health, physical environment, productivity, and social well-being. Family orientation incorporated four areas: daily family life, family interaction, financial well-being and parenting. A more recent three country study (Brown et al, in press, Brown, et al in press, and Neikrug et al in press) has reported that, in addition to the Poston study, family QOL outcomes are influenced by: support from other people, community and civic involvement, support from disability-related services and spiritual and cultural beliefs.

Presently many service organisations are designed around programmes, however, some of these programme processes are often too rigid and may not facilitate the

individual needs of children with Down syndrome and their families. Adjusting to life with a child with special needs is often a long process marked with times of joy and achievement but also by times of crises, when extra professional support may be necessary. Virginia Maxwell, a parent and nurse, in her article 'Look through the parents' eyes' suggested that parents and professionals must work together, learning from each other and aiming to increase the quality of life for the person with special needs, the parents and family (Maxwell V 1993).

A high proportion of parents responded to the question regarding changes needed to improve their child's QOL. The parent's responses revealed core themes of concern surrounding health practices, education, information delivery, learning process and learning needs, practice quality, long term management and community acceptance.

It was surprising that whilst 23% of parents had been offered genetic counselling only a small minority availed of the service. However, this may demonstrate reluctance or an inability to continue with the process or that it was not needed as only 1.5% of parents expressed a need for more genetic counselling and diagnostic results. Also, 75 parents held no record of their child's chromosomal diagnosis and it took extensive research to locate 62 of these records with 13 remaining unidentified

Major parental concerns were reported relating to how the diagnosis of Down syndrome was communicated to them as has been portrayed by their comments which express a a very negative picture. Parents very strongly advocated that the news that a child has Down syndrome be "said in a more positive way". An excellent presentation at the 9th Down Syndrome World Congress 2004 "Lets talk about the

Possibilities” by a young adult (Karen Gaffney) with Down syndrome demonstrated very positive achievements by herself including swimming the English Channel. This area of communicating the diagnosis of Down syndrome (telling bad news) requires much improvement and better approaches and attitude.

A high proportion of parents expressed their need for more services with better co-ordination. The particular areas of concern were; early intervention (33%); improved co ordination of health services (34%); most especially speech and language therapy (51%); and continuing parental education in order to empower them with the skills in the area of development, behaviour management and speech and language. Sue Buckley (1993) in her review on ‘Language development in children with Down syndrome’ reports that it is essential for these children to receive early physiotherapy in order to maximise motor progress and prevent delay in cognition and language learning delay. Buckley stressed that any sensory impairments also need to be identified and effectively treated as early as possible. She stressed that children with Down syndrome will benefit from language teaching right from infancy. She also stressed the necessity of drawing the parent’s attention to the importance of early conversations and to encourage them to follow the babies’ cues, and not overwhelm them with too much physical or verbal stimulation without allowing the baby to join in and respond.

When asked about way of improving healthcare 6% of parents expressed a need for the introduction of medical cards for all children with Down syndrome. This was surprisingly low but may be an indication that many of the families who participated

in the study had been successful in already being approved for a medical card for their child.

The need for healthcare guidelines was identified as a major concern by parents whose comments captured the depth of feeling including exhaustion and frustration at having to constantly fight for their child to get appointments to have hearing, vision and developmental screening tests performed on a regular basis through appropriate services. This may also demonstrate that the uncertainty of regular healthcare management deeply affects parent's QOL.

The need for emotional and physical well-being together with personal development has been highlighted by the large proportion (43%) of parents wishing to have their child educated in local mainstream schools. Many parents reported that attending mainstream schools improved their child's stimulation and social skills and supported better community integration. Sue Buckley (1993) reported that integrated education for children with Down syndrome in mainstream schools, in her area in Hampshire, proved to be beneficial for both the children with and without Down syndrome and that they found that most head teachers and staff agreed that all children have a right to grow up, loved and accepted in their local community regardless of disability. Walther-Thomas et al (1996) in a three year study reported improvements in social skills for students with special needs and that all students had developed a new appreciation of their own skills and accomplishments. Gillian Bird (1994) reported that where infant and junior schools are well supported by the child's speech and language therapist, educational psychologist and other appropriate sources of information, mainstreaming is becoming easier. However, mainstreaming in

secondary schools is still in its early days, without sufficient numbers of children on which to base comprehensive advice for good practice. In 1990 Sloper, Cunningham, Turner and Knussen in Manchester undertook a detailed study of 117 children with Down syndrome reported that those attending mainstream schools were likely to have the highest academic attainments in reading, writing and number. Wendy Casey (1988) and colleagues in London followed the progress of 36 children with Down syndrome for two years, assessing their progress at six monthly intervals. Half were in mainstream schools, the other half in schools for children with moderate learning difficulties. They reported that after two years the mainstream children performed better on all outcome measures, which were expressive language, comprehension, numeracy, verbal fluency, drawing ability and reading.

Just over 25% of parents reported a need for better integration in the community. One parent reported that she felt like a prisoner in her own home as her child could not go out and play with the local children as they could not communicate with each other. Another parent explained that she was so distressed and felt it so important for her child play with the local children that she arranged with her neighbours to teach their children sign language in order for her child to be accepted into the group and let play on the local street. Poston (2003) in a study of family QOL in a group of 187 individuals with and without disability pointed to the importance of physical environment, including home, school, work and neighbourhood/community environment. Roy Brown (in press 2006) in an international collaborative study of family QOL has reported that they are finding that among the challenges most frequently cited by families are issues concerning respite, support from neighbours and community, issues concerning front line support personnel, and lack of support

for the family in comparison to the child with the disability. For some children, the most nurturing environment available remains a special school, often one for children with severe learning difficulties, where they are wanted and valued whatever their level of learning difficulty or behaviour, and where staff are experienced in behaviour management and general teaching methods. Understanding the children's specific learning difficulties, as a group and individually, and meeting their educational needs, is fundamental for their development whether the children attend a special or mainstream school Bird (1994).

There was a high proportion (47%) of parent concerns relating to limited access to information and current research on Down syndrome. Many parents perceived that this reduced their ability to make informed decisions regarding their child's health and education. Parents identified areas that would require substantial investment and government support in order to develop comprehensive systems for the long-term needs of their children. Of great concern was transition from school to employment and independent living with support by parents. Some parents reported that they were left waiting until the last minute before knowing if their young adolescent/adult had been given a place in a job training school. They reported that there was little or no discussion or collaboration with the family regarding placement. Bellamy and Wilcox (1985) reported that in the past individuals with intellectual disabilities often were placed in workshop situations or in adult day programmes. Such placements usually promote dependence and provide few opportunities for integration with communities. Morton and Wehman (1995) suggested that when a formal interagency transition process facilitates the delivery of appropriate services in the least restrictive environment, one that includes employment, recreation, domestic life, and other

secondary schools does not include training for work placement but this is included in special schools. When a person with Down syndrome leaves their secondary school they have to look for a place to be trained for a work placement and be appointed a work coach. Parents concerns regarding community acceptance may identify that there is still much to be done to bridge the gap in community integration and participation. Wendy Parent (1988) found that successful community involvements, transition to independent or supported independent living can be successful for people with disabilities. They also suggest that the important component that can makes transition a positive experience includes a work educational program that provides for a functional curriculum in a natural and integrated setting, and placement in meaningful employment with an active and supporting work coach. Pueschel (1996) in an overview of transition from childhood to adulthood stated that the ultimate goal for people with Down syndrome who progress through a well planned transition period is to live well-adjusted and happy adult lives. Each individual should be offered a status that observes their rights and privileges as full citizens in a democratic society and that, in a real sense, preserves their human dignity.

This study identified other medical problems including: gastro-intestinal disorders such as constipation, diarrhoea and a higher incidence of coeliac disease. There is also an increased incidence of Type 1 Diabetes and respiratory problems including sinusitis, upper airway obstruction and chest infections, which require identification and management.

This study also identified that services were not coordinated throughout the health board area studied. Parents and healthcare professionals had no clear timetable for

health checks for their child with special needs. This could lead to an associated duplication of services to some families and minimal or no service to others. Progress in service delivery is hampered by the fact that Ireland does not have a national live register of people with Down syndrome. There is presently no central overseeing body to promote and audit the delivery of necessary services for this group and parents and young people with Down syndrome have no forum to appeal this poor performance.

While there are many challenges ahead in the lives of children, adolescents and adults with Down syndrome and their families, there are also challenges for healthcare professionals and particularly those involved in education and research. The way forward will be improved by collaboration across all fields and in believing that an enhanced QOL is a realistic and obtainable goal for those with intellectual disability (Shalock 2004).

CHAPTER FIVE

CONCLUSIONS AND RECOMMENDATIONS

CHAPTER 5 CONCLUSIONS AND RECOMMENDATIONS

Conclusions and Recommendations

This study shows a high incidence of associated medical disorders in children and adolescents with Down syndrome in Ireland. Studies have shown that early intervention carries a better medical outcome. Appropriate medical intervention maximises health outcomes and improves quality of life for the child and whole family.

Growth

This study found that boys and girls with Down syndrome have a shorter height with a lesser growth spurt at puberty. BMI was greater and a significantly smaller head circumference than the general Irish population was demonstrated at all ages. Body Mass Index (BMI) was therefore significantly greater in the older age groups when compared with the general population. Obesity is an escalating health problem associated with coronary disease, hypertension, type II diabetes and orthopaedic complications. There is growing evidence that excess weight gain can be prevented in children and adolescents with DS by early intervention. The onset of puberty in those with Down syndrome has been found to be slightly earlier than the Irish general population, even allowing for secular changes, thus requiring anticipation and education.

It is recommended that (Murphy 2005, Marder 2001) -:

- All children with DS should be given healthy eating advice at all ages and be encouraged to undertake daily physical activity from early childhood.

- Regular growth measurements should be performed frequently (three monthly) in the first two years of life and at least annually thereafter. These measurements should be plotted on DS specific growth charts
- Early failure to thrive may be associated with temporary feeding problems, many of which resolve after the first few weeks. Persistence of poor growth generally requires dietary evaluation and exclusion of pathology such as coeliac disease, gastro-oesophageal reflux, duodenal stenosis, malabsorption
- Comparison of weight alone can be misleading due to the presence of significant short stature and thus monitoring of BMI should also be performed. Those with a BMI above the 91st centile should be carefully monitored and those above the 98th centile require further assessment including thyroid function tests.

Data from this study contributed greatly to the development of the UK/Ireland Down syndrome Specific Growth Charts (Appendix 6)

Congenital Heart Disease

This study found that there is a high incidence of cardiac anomalies in children with DS in Ireland with the most common anomaly that of an AVSD. It was also found that the majority of cases were identified between birth and six weeks of age however, a further 15% were identified between 6 weeks and 14.5 years.

It is recommended that (Murphy 2005, Marder 2001) -:

Clinical examination, electrocardiogram and cardiac ECHO performed by staff with appropriate paediatric experience under the supervision of a paediatric cardiologist.

OR

A clinical examination, electrocardiogram (ECG) and chest X-ray (CXR) at birth and repeated at 6 weeks of age

Clinical examination alone is not sufficient. It is hoped in the future that advances in Telemedicine in Ireland may allow transmission of echo images between paediatricians in peripheral hospitals and the cardiologist at the National Unit.

Parents should receive written instruction regarding prevention of infective endocarditis, where appropriate.

Good dental health should be encouraged and regular dental assessments.

There is an increased incidence of mitral valve prolapse and of aortic regurgitation in adults with DS thus requiring an echo screen in early adult life

Thyroid

This study provides additional evidence that there is an increased prevalence of thyroid dysfunction in those with Down syndrome and that biochemical screening is necessary on a regular basis. Capillary TSH sampling by finger prick was found to be a feasible, reliable and an acceptable screening tool for diagnosing hypothyroidism in this population. This is an important factor in reducing parental reluctance for screening particularly in the preschool population. Hyperthyroidism also requires clinical vigilance, as it cannot yet be diagnosed by capillary sampling by measuring whole blood TSH alone. Further studies are required to identify the sensitivity and specificity of TSH capillary screening and provide a clearer picture on the rate of conversion to hypothyroidism and also the prevalence of the coexistence of thyroid dysfunction and Type 1 diabetes mellitus.

It is recommended that (Murphy 2005, Marder 2001) -:

- Newborn screening of all infants for hypothyroidism as per National Neonatal Screening Programme (Grant 1988)
- Screening of all those with DS for thyroid disease should be performed at 1 year and be continued 2 yearly thereafter or at any time if there is a clinical suspicion. This screening should include TSH, Free T4, and thyroid antibodies.
- Biochemical screening, including T4, TSH and thyroid antibodies, should be carried out at least once every two years from one year of age throughout life.. If finger-prick TSH assessment is available, annual screening of all patients with DS for hypothyroidism with TSH estimation by finger prick could be performed when available. Where TSH is elevated, full evaluation of thyroid function including Free T4 and thyroid antibodies should be undertaken.
- Any abnormality of thyroid function including isolated TSH elevation should be discussed with a Paediatric Endocrinologist.
- As in the general Irish population, clinical pointers of hypothyroidism in those with DS include lethargy, decreased cognition, reduced height, increased weight, depression and suspected dementia.
- The possibility of hyperthyroidism should be borne in mind and clinical vigilance is important. Symptoms may be subtle such as hyperactive behaviour, sleep disturbance without significant physical signs.

Vision

This study shows that vision disorder is common in those with Down syndrome in Ireland. These treatable conditions if undiagnosed and untreated can result in significant impairment of vision. Vision disorders can significantly impair psychomotor development and quality of life. Therefore, early referral and expert assessment is desirable in suspected vision disorders. There is an urgent need for agreed guidelines for assessment and treatment of disorders of vision in this group.

It is recommended that (Murphy 2005, Marder 2001) : -

- All newborns including those with Down syndrome should have an eye examination carried out at birth to exclude congenital cataract and glaucoma and be included in the community-screening programme thereafter.
- Visual behaviour or attention or changes in the appearance of the eye should be monitored regularly, at least, annually by a Paediatrician. The development of a squint or other abnormality should result in ophthalmological review. Distance vision should be checked at every review.
- In the second year of life all children with Down syndrome should have formal ophthalmological assessment including orthoptic, refraction and fundal examination. Subtle abnormalities are often detected on specialist screening and require close monitoring.
- Children and adults with Down syndrome will respond to a variety of visual testing and this depends very much on their developmental age. Some will require more specialized visual tests to be performed. A distraction free environment will optimise performance. Distance vision should be checked at every review.
- In the absence of abnormalities a further examination should be performed at

approximately four years of age by an ophthalmologist.

- After the age of 4 years vision should be checked two yearly by an ophthalmologist or optometrist throughout life.

In addition -:

- Any child identified by the school visual clinic service as less than 6/9 vision should be referred to a community ophthalmologist.
- Any child identified with less than 6/12 on a linear Snellen chart should be referred to a community ophthalmologist for further assessment.
- Local opticians give an excellent service but subjects who are not cooperative in this setting should be referred to a specialist clinic.
- Any child or adult with pain, and/or changing vision, visual disturbance and/or red eye, should be referred for urgent specialist opinion.

Hearing

Hearing impairment is a very an important health issue in children with Down syndrome, and early detection and intervention is essential to minimise secondary handicaps such as delayed speech and language development, communication and literacy skills (Yoshinaga-Itano 2001, Davis 1997, Denyer 1999, Hall 2003). This study has shown that DPOAE screening of inner ear function is feasible, fast, non-invasive and objective and that DPOAE results appeared to compare well with PTA findings in a small sample. Further longitudinal studies on larger numbers need to be undertaken to assess the specificity and sensitivity of this screening regime especially in the younger age groups in this population. DPOAE is a good choice for inner ear screening because of its ease of application and short screening time and the absence

of need for patient feedback. It must be remembered, however, that the DPOAE does not check the entire hearing pathway and it cannot detect retro-cochlea pathologies but allows accurate selection of persons for further diagnostic assessment.

It is recommended that (Murphy 2005, Marder 2001) -:

- Hearing assessment in those with DS must be performed by professionals trained in Audiology
- Newborns should be included in neonatal hearing screening programmes where available, to exclude congenital sensorineural hearing loss by Oto Acoustic Emissions (OAE) and/or brainstem auditory evoked responses (ABR)
- Full Audiological assessment should be performed between 6 and 10 months even where neonatal screening has been normal and thereafter repeated yearly until 5 years of age and then every 2 years for life. Methods used should allow measurement of auditory thresholds, impedance testing and otoscopy (Evenhuis 1996, National Deaf Children Society 1994).
- Those with detected abnormalities require more frequent assessment and ready access to ENT specialist services.
- Guidance for parents should include discussion about hearing problems and their management, supporting by quality written information (Sonsken 1985).

Cervical Spine Instability

In this study we found a high incidence of orthopaedic problems some of which required ongoing management with surgical intervention and physiotherapy and/or occupational therapy. The risk of cervical spine instability, though small, remains the

most worrying for parents and healthcare professionals. Acute dislocation/subluxation at the atlantoaxial joint cannot be predicted by cervical spine X-rays. There is currently no screening procedure, which can predict those at risk.

It is recommended that (Murphy 2005, Marder 2001)-:

- Despite the absence of an effective screening procedure for cervical spine instability in DS, the following signs and symptoms must not be ignored and indicate the need for immediate specialist referral (usually a neurologist or a spinal orthopaedic surgeon) -:
 - Pain behind the ear or elsewhere in the neck
 - Abnormal head posture (for example torticollis)
 - Deterioration of gait and manipulative skills
 - Reduced bowel and/or bladder control
- Cervical spine protection is important in the unconscious patient with DS undergoing anaesthesia. Anaesthetist and recovery room staff should be alerted to the diagnosis of DS in order to avoid cervical spine injury. In addition particular care should be paid to cervical spine management when an individual with DS is involved in a road traffic accident
- Exclusion from sporting activities should not be encouraged as currently there is no evidence that participation in sports increases the risk of cervical spine injury in those with DS.

Speech and Language

A very high proportion of children (94.5%) were found to have speech impairment in this study whilst only 5.5% had clear speech for age. Learning to communicate starts

at birth and language acquisition of all children occurs gradually through interaction with people and the environment. The ability to communicate and social skills in children with Down syndrome are vital for education, integration, work acquisition, reduction in depression, and improved quality of life. As speech development starts in infancy speech therapy is essential from the earliest stages of development. There is growing evidence that those with Down syndrome can develop speech more successfully with the introduction of signing, speaking, pictures and reading. In addition studies have shown that language gains were best maintained after intervention if the mothers received at the same time specific advice about verbal interaction at home. In addition research of language skills supports a need for speech and language therapy to continue into adolescence and adulthood. At present with the shortage of speech therapists in Ireland it may be more appropriate to develop a way that parents be educated to support the delivery of speech therapy with ongoing professional education, supervision and evaluation.

Quality of Life

Rearing a child with Down syndrome is not easy and a major challenge is to maximise Quality of Life (QOL). Assessment of QOL is important in determining whether additional multidisciplinary support is necessary. We found that QOL for parents caring for a child with Down syndrome was likely to be disrupted due to the extra care required, and particularly so when the child had more than one medical problem. Similarly, the majority of parents reported disruption to their family routines. Very poor quality of life was reported when hearing and vision problems were associated in a small number of cases. Early medical intervention, mainstream education, parent information, long term health needs, community acceptance and better integration were parent perceived areas in

most need of urgent improvement. This may be achieved by improved parental education in order to empower them with the skills in the area of development, behaviour management and speech and language. In addition collaboration between families and healthcare professionals in the development of improved co ordination of the health services; the implementation of the new medical management guidelines will support unified health management in children and adolescents; the development of adult medical management guidelines will promote better health throughout the lifespan and improved QOL. Projects in the community will promote awareness as to the abilities of children, adolescents and adults with Down syndrome and show them as valued members of society.

Conclusion

This study showed a high incidence of associated medical disorders in this population. Medical management guidelines for children and adolescents with Down syndrome in Ireland were developed from this study data with the support of the Down Syndrome Medical Interest Group UK. Many health care professionals with expertise in the management of children and adolescents with Down syndrome in Ireland have contributed to the Irish guidelines. These guidelines for Irish children have been approved by the Irish College of General Practitioners, The Faculty of Paediatrics of the Royal College of Physicians of Ireland and the Faculty of Public Health Medicine of the Royal College of Physicians of Ireland. Implementation of the guidelines and is now required and it will be essential to audit present and changing practices to ensure that they are achieving their objectives. This will ideally promote the development of a live register of people with Down syndrome in Ireland which will facilitate an accurate record of health problems and service requirements in this population. In addition the development of a national screening programme for

hypothyroidism for this group by fingerprick blood sample is desirable. Thus promoting a more coordinated, efficient and people friendly health service.

CHAPTER SIX

REFERENCES

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TABLES

Table 2.1 Distribution of Community Care Areas in the EHB				
1	Dun Laoghaire	Boooterstown	Sallynoggen	Ballybrack
		Blackrock	Dalkey	Loughlinstown
		Monkstown	Foxrock	Shankill
		Dun Laoghaire	Cabinteely	Little Bray
		Stillorgan	Killiney	
2	Dublin SE	Ringsend	Rathmines	Ballinteer
		Irishtown	Rathgar	Sandyford
		Sandymount	Donnybrook	Stepaside
		Ballsbridge	Milltown	Kilternan
		Ranelagh	Dundrum	
3	Dublin South Central	James' Street	Churchtown	
		Harold's Cross	Ballyboden	
		Terenure	Whitechurch	
		Rathranhham		
4	Dublin South West	Drimnagh	Kilnamanagh	
		Crumlin	Tallaght	
		Walkinstown	Fourtunestown	
5	Dublin West	Palmerstown	Inchicore	Saggart
		Chapelizoid	Clondalkin	Rathcoole
		Ballyfermot	Milltown	Brittas
		Lucan	Newcastle	
6	Dublin North West	Ward	Blanchartstown	Cabra
		Clonsilla	Tolka Valley	Phibsboro
		Mulhuddart	Glasnevin	
		Finglas	Castleknock	
7	Dublin North Central	Dublin Airport	Drumcondra	
		Santry	Donllymount	
		Ballymun	Fairview	
		Beaumont		
8	Dublin North	Balbriggan	Rush	Balydoyle
		Naul	Donabate	Kilbarrack
		Garristown	Kilsallaghan	Coolock
		Skerries	Swords	Raheny
		Lusk	Malahide	Artane
		Oldtown	Portmarnock	Howth
		Ballyboghill	Darndale	
9	Co. Kildare	Co. Kildare	Co. Kildare	
10	Co Wicklow	Co Wicklow	Co Wicklow	

Table 2.2 Classification of Tympanograms

Predetermined parameters for grading of Tympanometry as described by Jerger 1970

Class	Pressure	Compliance	Volume
A	Normal (-150 - +100 daPa)	Normal (0.3 - 2-5cm ³)	Normal (0.3-2-5cm ³)
As	Normal	Shallow (<0.3cm ³)	Normal
Ad	Normal	Hyper (>2-5cm ³)	Normal
C	Negative (> - 150 daPa)	Normal (0.3 - 2-5cm ³)	Normal
Cs	Negative	Shallow (<0.3cm ³)	Normal
B		Flat	Normal
B		Flat	Large (>1.75cm ³)
Type A Normal function			
Type As The shallow curve suggests increased stiffness of the middle ear structures			
Type Ad: This is the opposite situation in which normal middle ear pressure is present, accompanied by an unusually high compliance. This pattern can often be seen in the presence of a flaccid (floppy) tympanic membrane, and if it exceeded the limits of equipment (>5 cm ³) could be a sign of ossicular discontinuity and may require further investigation			
Type C Possible poorly functioning Eustachian tube			
Type B + normal ear canal volume - possible fluid +large ear canal volume – possible perforation of tympanic membrane			

Note: 1: 0.02 mmH₂O = 1.0 daPa; daPa = decaPascals; (Atmospheric pressure = 0 daPa)

Table 2.3 DPOAE Screening Test Levels

Test No 1 Screening 65/55 dB

DP Stimulus Protocol

L1=65 dB SPL; L2=55 dB SPL

Number of Octaves =3 (1000 - 2000 Hz, 2000 - 4000 Hz, 4000 - 8000 Hz)

Number of points/octave = 3,3,2 total 8

Test No. 2 Screening 65/50 dB

DP Stimulus Protocol

L1=65 dB SPL; L2=50 dB SPL

Number of Octaves =3 (1000 - 2000 Hz, 2000 - 4000 Hz, 4000 - 8000 Hz)

Number of points/octave = 3,3,2 total 8

**DPOAE - Distortion product $\geq$ 3 dB above noise floor amplitude* (mean + 1 sd)
for 2 of 3 frequencies per octave**

* Background noise (noise floor amplitude) is defined as the amplitude (sound level) of background noise that occurs at or near the test frequency at time of recording.

Table 2.4 DPOAE – GSI 60 Pass/Refer Criteria

Table 2.4 GSI 60 Pass/Refer criteria for DPOAE			
Classification	1-2 kHz	2-4 kHz	4-8 kHz
Pass	> 2 Emissions*	> 2 Emissions*	> 2 Emissions*
Refer	< 2 Emissions*	< 2 Emissions*	< 2 Emissions*
<i>Emissions* >= 3 dB above noise floor level</i>			
Screening levels used: 1) 65/55 dB and 2) 65/50 dB SPL at each octave plotted on the Vanderbilt Normal Range			

Table 2.5 Hearing thresholds (Lee, KJ 1999; Essential Otolaryngology)

Degree of Impairment	Threshold (dB HL)
Normal hearing	-10 - 20
Mild hearing loss	21 - 40
Moderate hearing loss	41 - 55
Moderate to severe hearing loss	56 - 70
Severe hearing loss	71 - 90
Profound hearing los	>90

Table 3.77 **Comparison of mean height (cms) of Irish boys and girls with Down syndrome (DS)**

Age (yrs)	Irish Boys DS*	Irish Girls DS
2	78	77.5
2.5	82.4	81.1
3	86.9	83.4
3.5	88.9	84.6
4	93.7	94.3
4.5	94.6	92.1
5	100	98
5.5	104.5	101.7
6	104.9	102.4
6.5	103.6	111.9
7		105.8
7.5	113.8	107.4
8	112.6	108.9
8.5	121.6	120.8
9	124.2	119
9.5	121.4	119.4
10	125.2	123.6
10.5	131.2	126.1
11	131.5	125.7
11.5	134	138.3
12	137.9	136.4
12.5	132.8	135.3
13	147.7	137.5
13.5	144	143.7
14	148.2	141.7
14.5	153.5	139.7
15	148	147.6
15.5	154.6	141.1
16	153	143.5
16.5	154.9	154.7
17	154.4	141.3
17.5	155	150.1
18	165	147.3
18.5	150	139.4

Table 3.78 Comparison of median weight (kgs) of Irish boys and girls with Down syndrome (DS)

Age (yrs)	Irish Boys DS*	Irish Girls DS*
0.5	6.2	6.9
1	9.7	9.6
2	11.5	10.8
3	13.2	12.2
4	15.5	15.4
5	18.6	17.3
6	18.9	18.1
7	21.2	20.9
8	28.6	23.5
9	29.4	26.9
10	32	30.2
11	36	33.2
12	35.2	35.1
13	44.9	51.9
14	60.9	41.8
15	62.2	49
16	60.2	54.4
17	57	60.5
18	57.9	59.3

Table 3.79 **Comparison of median BMI of Irish boys and girls with Down syndrome (DS)**

Age (yrs)	Irish Boys DS Median BMI	Irish Girls DS Median BMI
1	17.7	17..3
2	17.4	18.1
3	18	17.45
4	16.75	17.6
5	17.8	17.8
6	17.1	16.7
7	16.25	17.8
8	19.4	17.7
9	19.1	19.8
10	19.4	18.15
11	19.95	20.5
12	19.5	19.7
13	20.1	25.2
14	26.1	22
15	23.8	24.05
16	23.8	26.3
17	25.7	29.2
18	23.85	28.8

Table 3.80 **Comparison of median head circumference of Irish boys and girls with Down syndrome**

Age (yrs)	Boys Down syndrome	Girls Down syndrome
5	49.4	47.4
6	48.7	47.9
7	48.6	48.4
8	50.0	49.1
9	49.9	48.6
10	50.2	49.8
11	51.2	50.5
12	50.8	51.3
13	51.7	51.0
14	52.8	51.5
15	52.9	50.9
16	52.9	52.4
17	54.6	51.6
18	52.1	50.3

Table 3.81 Comparison of mean height (cms) of Irish boys with Down syndrome (DS) with the general Irish population

Age (yrs)	General Irish Population Boys*	Irish DS** Boys	Z score
2.0	85.8	78.0	-2.4
2.5	90.0	82.4	-2.1
3.0	94.1	86.9	-2.0
3.5	97.9	88.9	-2.2
4.0	101.5	93.7	-1.8
4.5	105.0	94.6	-2.3
5.0	108.1	100.0	-1.5
5.5	110.9	104.5	-1.3
6.0	114.2	104.9	-1.8
6.5	117.2	103.6	-2.6
7.0	120.0		0.0
7.5	122.7	113.8	-1.6
8.0	125.6	112.6	-2.4
8.5	128.3	121.6	-1.2
9.0	131.0	124.2	-1.2
9.5	133.5	121.4	-2.0
10.0	136.2	125.2	-1.8
10.5	138.8	131.2	-1.2
11.0	141.3	131.5	-1.5
11.5	143.9	134.0	-1.5
12.0	146.4	137.9	-1.9
12.5	149.2	132.8	-2.3
13.0	151.9	147.7	-0.6
13.5	154.9	144.0	-1.4
14.0	158.7	148.2	-1.4
14.5	162.6	153.5	-1.1
15.0	166.1	148.0	-2.4
15.5	168.9	154.6	-1.9
16.0	171.5	153.0	-2.6
16.5	173.7	154.9	-2.8
17.0	175.0	154.4	-3.1
17.5	175.3	155.0	-3.1
18.0	175.5	165.0	-1.6
18.5	175.5	150.0	-4.0

*Hoey 1987
 ** DS Down syndrome

Table 3.82 Comparison of mean height (cms) of Irish girls with Down syndrome (DS) with the general Irish population

Age (yrs)	General Irish Population Girls*	Irish DS Girls	Z score
2.0	84.6	77.5	-2.2
2.5	88.8	81.1	-2.2
3.0	92.9	83.4	-2.5
3.5	96.7	84.6	-3.0
4.0	100.2	94.3	-1.4
4.5	103.3	92.1	-2.5
5.0	106.8	98.0	-1.9
5.5	109.9	101.7	-1.7
6.0	113	102.4	-2.1
6.5	115.8	111.9	-0.8
7.0	118.9	105.8	-2.4
7.5	122.2	107.4	-2.7
8.0	124.5	108.9	-2.7
8.5	127.2	120.8	-1.1
9.0	129.9	119.0	-1.8
9.5	132.5	119.4	-2.2
10.0	135.3	123.6	-1.9
10.5	138.2	126.1	-1.8
11.0	141.1	125.7	-2.2
11.5	144.2	138.3	-0.8
12.0	147.3	136.4	-1.5
12.5	150.5	135.3	-2.1
13.0	153.7	137.5	-2.4
13.5	156.2	143.7	-2.0
14.0	158.6	141.7	-2.8
14.5	160.3	139.7	-3.5
15.0	161.3	147.6	-2.3
15.5	162.1	141.1	-3.6
16.0	162.6	143.5	-3.3
16.5	162.8	154.7	-1.4
17.0	163.0	141.3	-3.8
17.5	163.0	150.1	-2.3
18.0	163.0	147.3	-2.8
18.5	163.0	139.4	-4.1
Total Mean SD Score			-2.3

*Hoey 1987
** DS Down syndrome

Table 3.83 Comparison of median weight of boys and girls with Down syndrome (DS) with general Irish population (kgs)

Age (yrs)	General Irish population* Boys	Irish Boys DS**	General Irish population* Girls	Irish Girls DS**
0.5		6.2		6.2
1		9.7		9.7
2	13.1	11.5	13.1	11.5
3	15.0	13.2	15.0	13.2
4	16.4	15.5	16.4	15.5
5	18.8	18.6	18.8	18.6
6	20.8	18.9	20.8	18.9
7	23.0	21.2	23.0	21.2
8	25.4	28.6	25.4	28.6
9	28.0	29.4	28.0	29.4
10	30.8	32.0	30.8	32.0
11	33.9	36.0	33.9	36.0
12	37.4	35.2	37.4	35.2
13	41.9	44.9	41.9	44.9
14	47.9	60.9	47.9	60.9
15	54.8	62.2	54.8	62.2
16	60.0	60.2	60.0	60.2
17	62.6	57.0	62.6	57.0
18	63.7	57.9	63.7	57.9

*Hoey 1987
 ** DS Down syndrome

Table 3.84 Comparison of median BMI of Irish boys and girls with Down syndrome (DS) and 50th Centile UK BMI

Age (yrs)	50th Centile Male BMI UK*	Irish Boys DS Median BMI	50th Centile Female BMI UK*	Irish Girls DS Median BMI
1	17.64	17.70	17.25	17.30
2	16.66	17.40	16.34	18.10
3	16.13	18.00	15.88	17.45
4	15.75	16.75	15.66	17.60
5	15.55	17.80	15.48	17.80
6	15.5	17.10	15.49	16.70
7	15.56	16.25	15.68	17.80
8	15.75	19.40	15.99	17.70
9	16.04	19.10	16.4	19.80
10	16.42	19.40	16.9	18.15
11	16.89	19.95	17.48	20.50
12	17.43	19.50	18.12	19.70
13	18.04	20.10	18.77	25.20
14	18.68	26.10	19.4	22.00
15	19.32	23.80	19.96	24.05
16	19.94	23.80	20.44	26.30
17	20.52	25.70	20.85	29.20
18	21.05	23.85	21.19	28.80

(British Growth Reference for BMI Cole T. 1990)

Table 3.85 Boys with DS identified as overweight and obese (Cole)

Age (yrs)	Total number	Overweight N (%)	Obese N (%)	Total Overweight/ Obese %
1	15	2 (13)	3 (20)	33.3
2	20	4 (20)	4 (20)	40.0
3	12	3 (25)	1 (8)	33.3
4	20	4 (20)	4 (20)	40.0
5	13	3 (23)	3 (23)	46.2
6	13	2 (15)	2 (15)	30.8
7	6	1 (17)	1 (17)	33.3
8	10	1 (10)	4 (40)	50.0
9	9	2 (22)	3 (33)	55.6
10	8	1 (13)	3 (38)	50.0
11	18	1 (6)	6 (33)	38.9
12	13	2 (15)	1 (8)	23.1
13	11	3 (27)	2 (18)	45.5
14	10	2 (20)	6 (60)	80.0
15	12	2 (17)	4 (33)	50.0
16	9	1 (11)	3 (33)	44.4
17	6	2 (3)	2 (33)	66.7
18	2	0 (0)	1 (50)	50.0
Total	207	36	53	

Table 3.86 Girls with DS identified as overweight and obese (Cole)

Age (yrs)	Total number	Overweight N (%)	Obese N (%)	Total Overweight/ Obese %
1	11	0 (0)	3 (27)	27.3
2	10	3 (30)	1 (10)	40.0
3	14	2 (14)	1 (7)	21.4
4	6	1 (17)	2 (33)	50.0
5	13	4 (31)	2 (15)	50.0
6	7	0 (0)	2 (29)	28.6
7	5	2 (40)	0 (0)	40.0
8	11	2 (18)	2 (18)	36.4
9	11	4 (36)	2 (18)	54.5
10	14	3 (21)	2 (14)	35.7
11	11	0 (0)	4 (36)	36.4
12	10	2 (20)	1 (10)	30.0
13	11	3 (27)	5 (45)	72.7
14	12	2 (17)	2 (17)	33.3
15	12	1 (8)	5 (42)	50.0
16	7	2 (29)	3 (43)	71.4
17	7	0 (0)	4 (57)	57.1
18	2	1	1 (50)	100.0
Total	174	32	42	

Table 3.87 Boys and Girls with DS with overweight and obesity in age groups under 10 years and 10 to 18 years of age

Age 1 to 10 years		
	Overweight (%)	Obesity (%)
Girls (n=88)	18 (20)	15 (17)
Boys (n=118)	22 (19)	25 (21)
Ages 10 to 18 years		
	Overweight (%)	Obesity (%)
Girls (n=86)	14 (16)	27 (31)
Boys (n=89)	14 (16)	28 (31)

Table 3.88 Comparison of mean head circumferences (cms) of boys with Down syndrome (DS) with general Irish population*

Age (yrs)	Boys DS** Head Circumference	z Score	General Irish population* Head Circumference	SD
5	49.4	-1.7	51.8	1.45
6	48.5	-2.5	52.1	1.45
7	48.5	-2.6	52.3	1.45
8	50.0	-1.8	52.6	1.45
9	49.8	-2.1	52.9	1.45
10	50.3	-2.0	53.2	1.45
11	51.3	-1.6	53.6	1.46
12	51.2	-1.9	54.0	1.48
13	51.5	-2.0	54.5	1.52
14	53.1	-1.3	55.1	1.51
15	52.7	-2.0	55.7	1.49
16	53.3	-2.0	56.3	1.48
17	53.8	-1.9	56.6	1.47
18	52.1	-3.1	56.7	1.47

*Hoey 1987
 ** DS Down syndrome

Table 3.89 Comparison of mean head circumferences (cms) of girls with Down syndrome (DS) with general Irish population*

Age (yrs)	Girls DS**Mean Head Circumference	z Score	General Irish Population	SD
5	47.4	-2.5	50.9	1.38
6	48.1	-2.3	51.3	1.37
7	48.3	-2.4	51.6	1.37
8	49.2	-2.0	51.9	1.37
9	49.0	-2.3	52.2	1.37
10	49.7	-2.0	52.5	1.37
11	50.5	-1.7	52.9	1.39
12	51.2	-1.5	53.4	1.43
13	51.5	-1.7	53.9	1.43
14	50.6	-2.6	54.3	1.43
15	50.9	-2.6	54.5	1.40
16	52.8	-1.4	54.6	1.33
17	52.2	-1.9	54.7	1.30
18	50.2	-3.5	54.7	1.30

*Hoey 1987
** DS Down syndrome

Table 3.90 Comparison of boys’ mean height of general Irish population and the Irish, American and Dutch Down syndrome (DS) populations (cms)

Age (yrs)	General Irish Population*	Irish DS**	American DS***	Dutch DS****
2	85.8	78.5	81.6	85.18
3	94.1	85.7	86.0	91.57
4	101.5	92.5	90.5	97.7
5	108.1	102.1	95.5	103.61
6	114.2	104.6	101.0	110.6
7	120.0	105.4	107.1	115.44
8	125.6	115.5	113.0	121.27
9	131.0	121.3	119.0	127.2
10	136.2	124.3	125.0	131.4
11	141.3	130.8	131.3	135.71
12	146.4	137.5	136.8	144.03
13	151.9	141.4	141.3	151.13
14	158.7	149.9	145.8	157.21
15	166.1	151.9	148.9	160.26
16	171.5	154.5	150.8	163.03
17	175.0	154.7	151.8	163.03
18	175.5	157.5	152.0	163.03

*Hoey 1987
** DS Down syndrome
***Cronk C 1988 Smoothed chart yearly figures
****Cremers 1996

Table 3.91 Comparison of girls’ mean height of general Irish population and the Irish, American and Dutch Down syndrome (DS) populations (cms)

Age (yrs)	General Irish Population*	Irish DS**	American DS***	Dutch DS****
2	84.6	84.2	81.0	84.60
3	92.9	83.2	86.5	90.74
4	100.2	90.3	92.2	96.31
5	106.8	96.7	98.0	103.67
6	113.0	106.0	103.8	112.45
7	118.9	104.3	109.3	116.44
8	124.5	114.5	115.0	123.76
9	129.9	119.7	120.9	127.52
10	135.3	122.8	126.0	133.67
11	141.1	125.9	131.0	137.59
12	147.3	137.2	135.0	142.15
13	153.7	137.9	139.0	147.26
14	158.6	142.1	142.0	150.12
15	161.3	144.8	144.0	151.42
16	162.6	144.4	145.0	151.99
17	163.0	149.3	145.3	151.99
18	163.0	143.0	145.3	151.99

*Hoey 1987

** DS Down syndrome

*** Cronk C 1988 Smoothed chart yearly figures

****Cremers 1996

Table 3.92 Comparison of boys’ median weight of general Irish population and the Irish, American and Dutch Down syndrome (DS) populations (kgs)

Age (yrs)	General Irish population*	Irish DS**	American DS***	Dutch DS****
0.5		6.2	6.7	
1		9.7	8.2	
2	13.1	11.5	10.5	13.8
3	15.0	13.2	12.1	15.49
4	16.4	15.5	13.2	17.78
5	18.8	18.6	14.4	19.95
6	20.8	18.9	16.6	22.39
7	23.0	21.2	19.7	25.12
8	25.4	28.6	23.3	28.18
9	28.0	29.4	27.0	31.62
10	30.8	32.0	31.2	34.67
11	33.9	36.0	35.7	39.81
12	37.4	35.2	40.1	45.71
13	41.9	44.9	44.6	50.12
14	47.9	60.9	48.8	53.7
15	54.8	62.2	52.1	56.23
16	60.0	60.2	55.7	56.23
17	62.6	57.0	58.4	56.23
18	63.7	57.9	60.0	

*Hoey 1987
 ** DS Down syndrome
 **** Cronk C 1988 Smoothed chart yearly figures
 *****Cremers 1996

Table 3.93 Comparison of girls’ median weight of general Irish population and the Irish, American and Dutch Down syndrome (DS) populations (kgs)

Age (yrs)	Irish general population*	Irish DS**	American DS***	Dutch DS****
0.5		6.9		
1		9.6		
2	12.2	10.8	11.2	13.8
3	14.2	12.2	12.4	15.9
4	16.2	15.4	13.9	18.2
5	18.2	17.3	15.7	20.4
6	20.3	18.1	18.3	23.4
7	21.7	20.9	21.1	26.3
8	24.4	23.5	24.7	29.5
9	27.0	26.9	28.0	33.1
10	29.9	30.2	31.9	36.3
11	33.0	33.2	35.5	39.8
12	37.2	35.1	39.2	44.7
13	42.3	51.9	43.0	49.0
14	48.5	41.8	47.0	52.5
15	52.2	49.0	50.7	53.7
16	53.2	54.4	54.0	53.7
17	55.5	60.5	57.1	53.7
18	56.3	59.3	59.5	

Table 3.95(a) Comparison of mean height SD scores of Down syndrome (DS) boys with cardiac anomalies categories 1-2 and 3-4 and the general Irish Population

Age (yrs)	DS mean height (cms) Categories 1-2*	DS Mean Height SD Scores –v- general Irish Population Categories 1-2*	DS mean height (cms) Categories 3-4*	DS Mean Height SD Scores –v- general Irish Population Categories 3-4**
2	79.1	-2.0	76.9	-2.7
2.5	82.5	-2.1	81.9	-2.3
3	86.9	-1.9		
3.5	88.9	-2.2		
4	93.4	-1.9	93.7	-1.8
4.5	94.6	-1.6		
5	103.3	-0.4	97.9	-1.6
5.5	104.9	-0.7	105.9	-0.5
6	104.9	-1.8		
6.5	103.6	-2.6		
7				
7.5	114.3	-1.5	111.6	-2.0
8	111.6	-2.5		
8.5	121.5	-1.2		
9	124.2	-1.2		
9.5	122.3	-1.9	119.7	-2.3
10	125.2	-1.8		
10.5	130.8	-1.3	132.2	-1.0
11	131.9	-1.4	129.6	-1.8
11.5	132.5	-1.3	134.1	-1.5
12	137.1	-1.3	140.0	-0.9
12.5	134.3	-2.1	121.0	-3.9
13	148.8	-0.1	149.1	0.0
13.5	144.0	-1.4		
14	159.2	0.1	141.6	-2.2
14.5	153.5	-1.2		
15	148.1	-2.4	147.9	-2.4
15.5	156.1	-1.8	153.4	-2.1
16	154.3	-2.5	150.3	-3.0
16.5	154.9	-2.8		
17	155.8	-2.9	150.0	-3.8
Total Mean SD Scores		-1.8		-1.6

Peuschel 1987
*Category 1
* Category 2
** Category 3
** Category 4

No cardiac anomaly.
Single cardiac defects, such as atrial or ventricular septal defects without pulmonary vascular involvement. These children generally did not require cardiac medications nor cardiac surgery and had a normal growth rate.
More complex cardiac problems, such as endocardial cushion defects. These children often required digitalis. Their growth was generally sub optimal.
Serious anatomical heart lesions, such as complex endocardial cushion defects, Fallot’s tetralogy, etc. These children were often in cardiac failure, and most had pulmonary complications and generally reduced growth.

Table 3.96(b) Comparison of mean height SD scores of Down syndrome (DS) girls with cardiac anomalies categories 1-2 and 3-4 and the general Irish Population

Age (yrs)	DS mean height (cms) Categories 1-2*	DS mean height SD scores from general Irish population Categories 1-2*	DS mean height (cms) Categories 3-4*	DS Mean Height SD Scores –v- general Irish Population Categories 3-4**
2	76.1	-2.6	81.6	-0.9
2.5	79.1	-2.7	82.4	-1.8
3	83.8	-2.4	77.6	-4.0
3.5	84.1	-3.1	91.3	-1.3
4	95.0	-1.2		.
4.5	85.8	-3.9	98.4	-1.1
5	98.9	-1.7	95.8	-2.3
5.5	104.2	-1.2	102.9	-1.4
6	102.1	-2.1	103.5	-1.9
6.5	113.3	-0.5	114.4	-0.3
7	104.1	-2.7	108.0	-2.0
7.5	113.8	-1.5		.
8	112.8	-2.1	101.0	-4.1
8.5	123.8	-0.6	118.6	-1.5
9	117.5	-2.1	118.3	-1.9
9.5	123.9	-1.4	113.0	-3.2
10	123.5	-1.9	123.9	-1.8
10.5	125.5	-1.9	127.2	-1.7
11	125.7	-2.2		.
11.5	137.1	-1.0		.
12	136.2	-1.5	136.7	-1.5
12.5	141.6	-1.2	132.3	-2.5
13	137.2	-2.5		.
13.5	143.8	-1.9	143.6	-2.0
14	141.7	-2.8		.
14.5	139.7	-3.5		.
15	147.7	-2.3	147.6	-2.4
15.5	140.9	-3.7	141.7	-3.5
16	148.0	-2.5	141.5	-3.6
16.5	156.0	-1.2	154.2	-1.5
17	141.3	-3.8		.
Total Mean SD scores		-2.4		-2.0

Peuschel 1987

*Category 1 No cardiac anomaly.

* Category 2 Single cardiac defects, such as atrial or ventricular septal defects without pulmonary vascular involvement. These children generally did not require cardiac medications nor cardiac surgery and had a normal growth rate.

** Category 3 More complex cardiac problems, such as endocardial cushion defects. These children often required digitalis. Their growth was generally sub optimal.

** Category 4 Serious anatomical heart lesions, such as complex endocardial cushion defects, Fallot’s tetralogy, etc. These children were often in cardiac failure, and most had pulmonary complications and generally reduced growth.

FIGURES

Figure 3.15

HEAD CIRCUMFERENCE CHART
BOYS - BIRTH TO 18 YEARS

Address	Date of Birth
Consultant:	Hospital

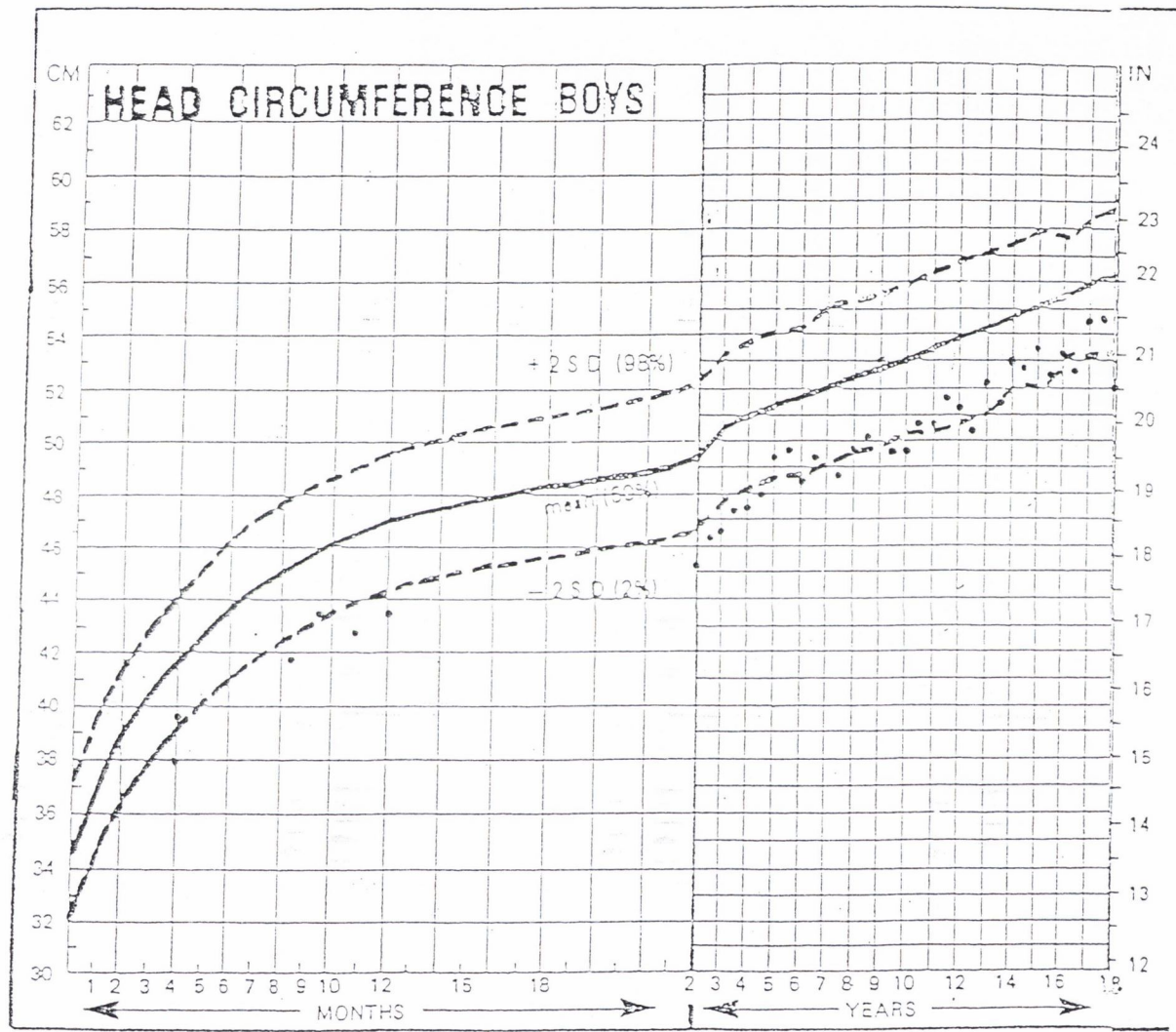


Figure 3.16

FEDERATED DUBLIN VOLUNTARY
HOSPITALS

HEAD CIRCUMFERENCE CHART
GIRLS — BIRTH TO 18 YEARS

Surname	First Name	Hospital No.
Address		Date of Birth
Consultant:		Hospital

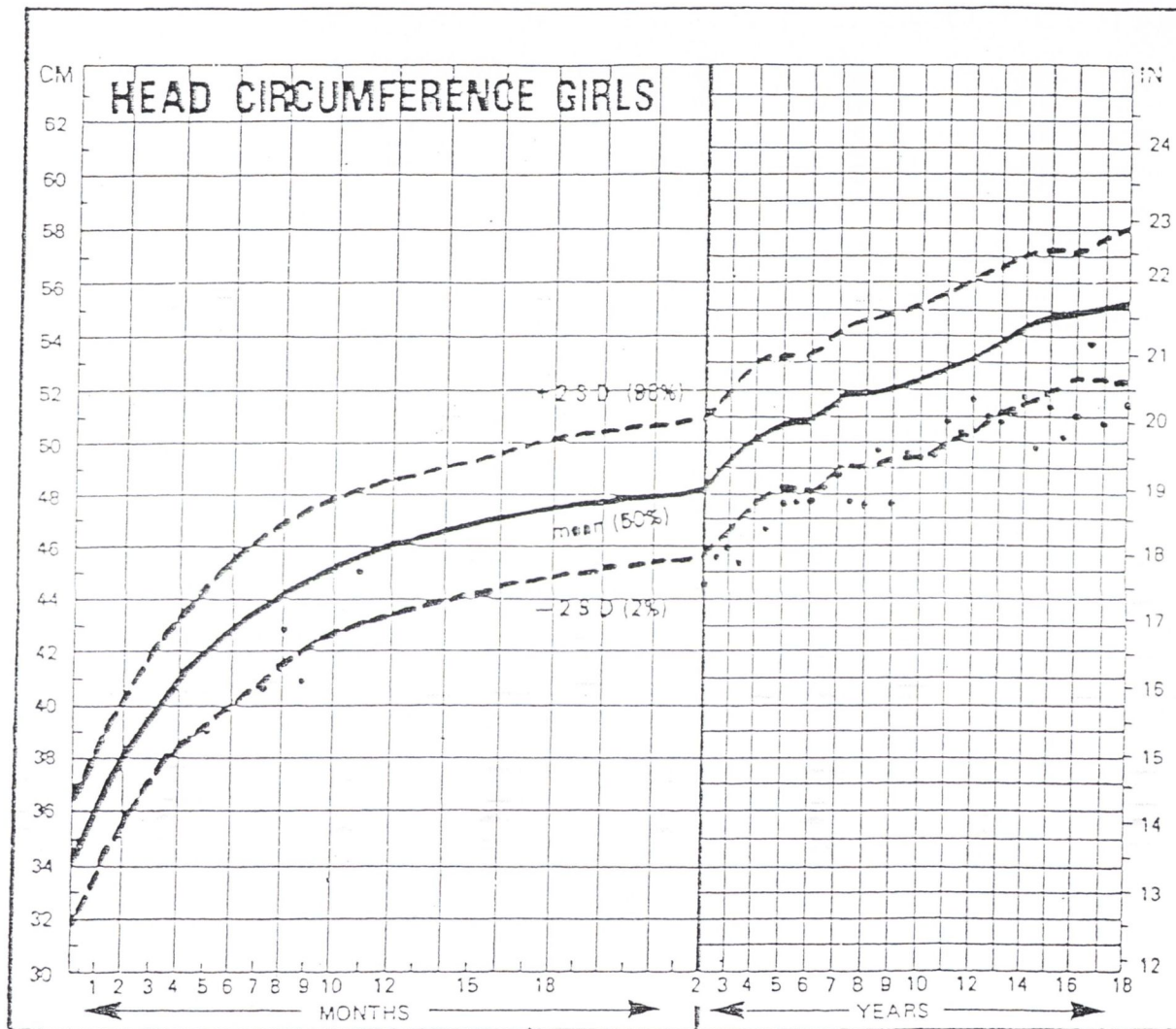


Figure 3.17

Reg. No.	
Surname	
Forename	
Date of Birth	/ /
Hospital/Clinic Name	
Decimal Yr.	

Growth and Development Charts
Boys: Down Syndrome
Birth to 3 years
Head Circumference
NUMBER 2741 - 1992
M.A.S. INTERNATIONAL SERIES

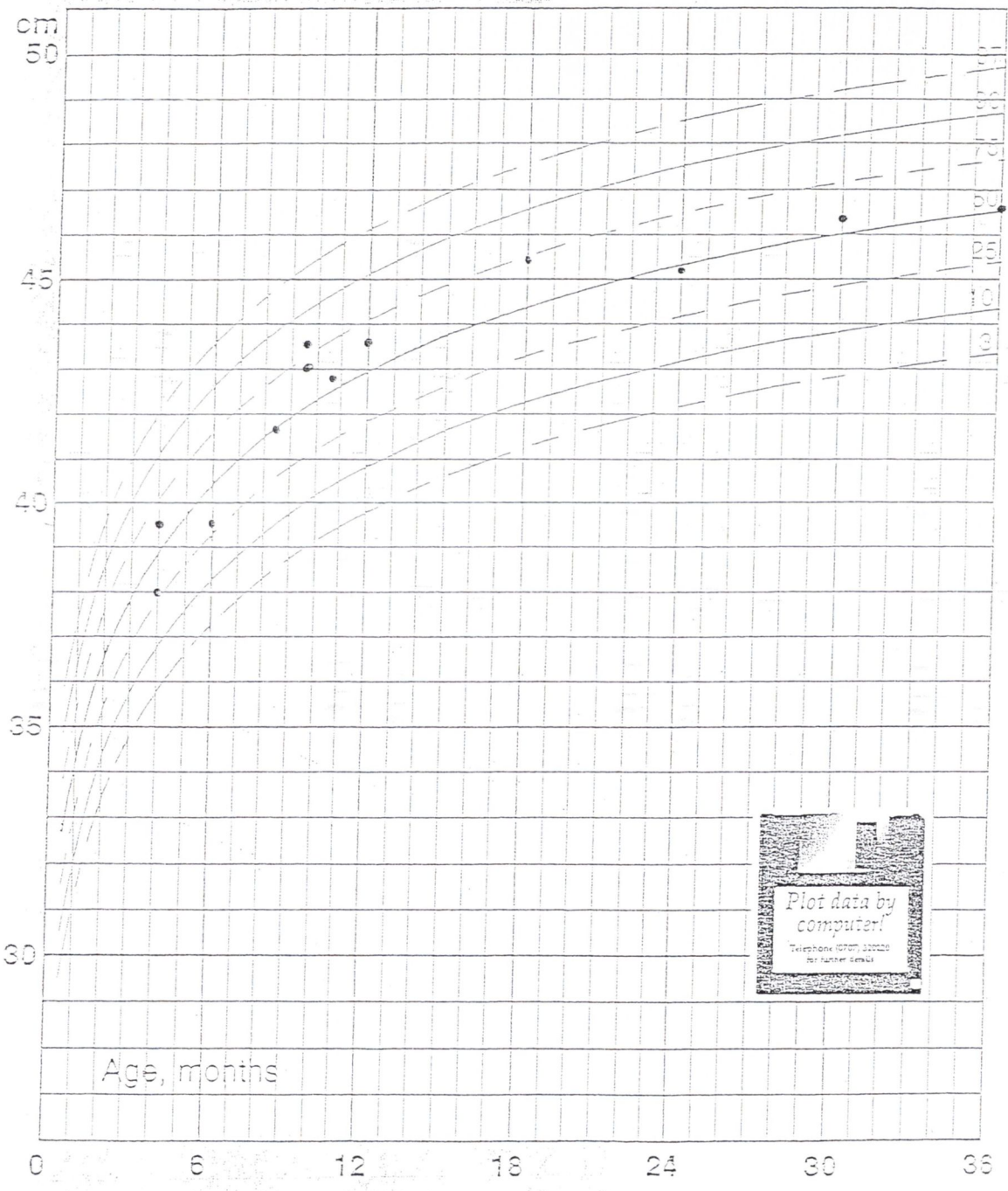
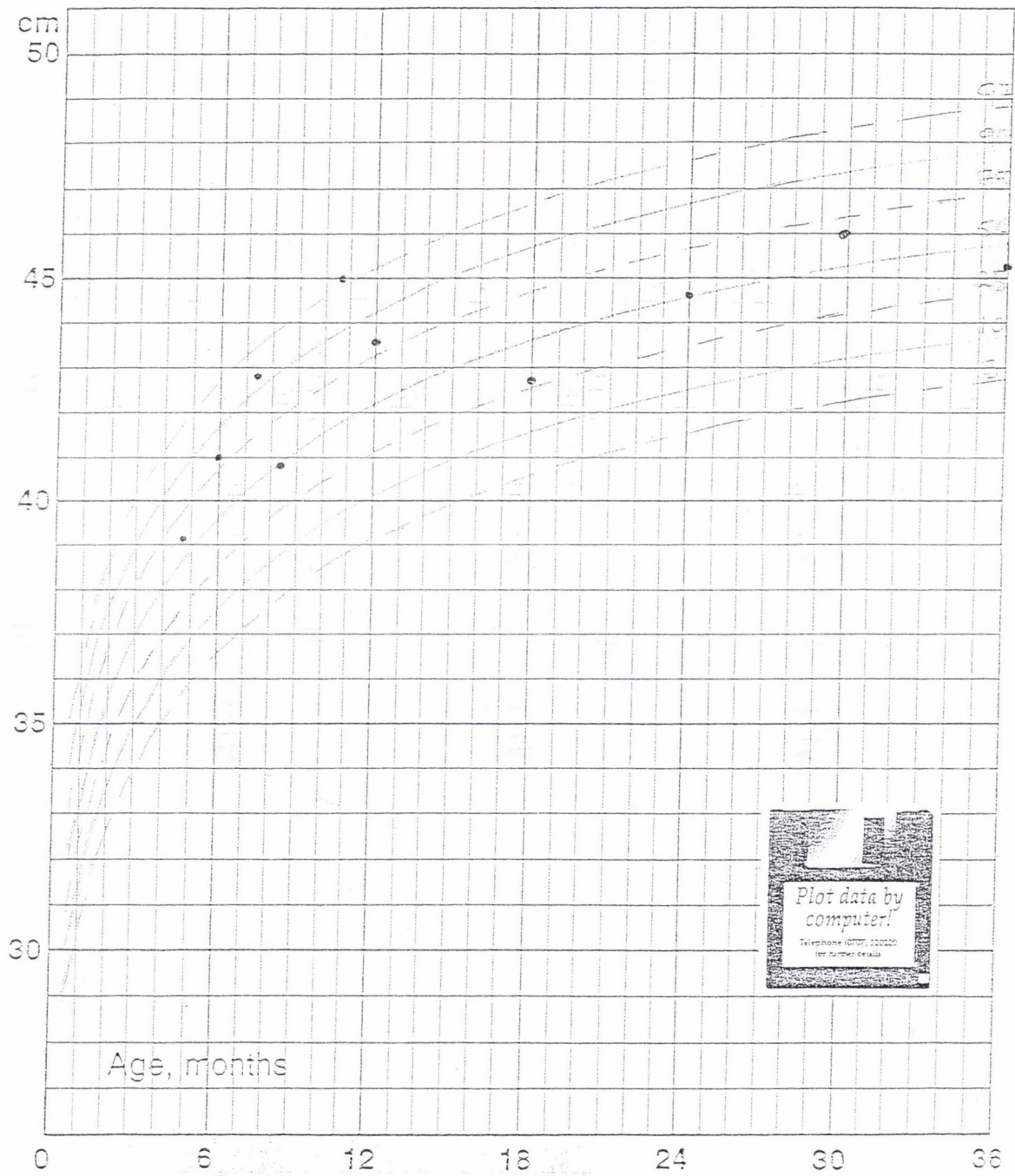


Figure 3.18

Reg. No. _____
Surname _____
Forename _____
Date of Birth / / Decimal Yr. _____

Hospital/Clinic Name _____

Growth and Development Record
Girls: Down Syndrome
Birth to 3 years
Head Circumference
PALMER ET AL. (1992)
Ref 96 INTEGRATED SERIES



APPENDICES

APPENDIX I

APPENDIX 1: PARENT INVITATION AND INFORMATION LETTER

“A Study On Children With Down Syndrome”

Dear Parent,

We are conducting a study on children and adolescents with Down Syndrome from birth to 16 years in the Eastern Regional Health Authority and obtained your name and address from the Down Syndrome Association. The purpose of the study is to assess their medical needs and the specialist services they require at each stage of their lives. We would like to invite you to become part of this study as your opinions and answers are vitally important to identify the additional services your children require.

We will ask you to answer a questionnaire and we will examine your child's height, weight, chest, heart, eyes, ears (including a Hearing Screening Test), and we may need to do a simple blood test (finger prick) to check Thyroid function only if necessary and with your consent. This will be a once off visit and any findings that need medical attention will be referred back to the Paediatrician already in charge.

All your answers will be kept strictly confidential and will be covered by Data Protection Act 1988. When published the information will not be used to identify any individuals or centres. You may withdraw from the study at any time.

Participation in the Study will not disrupt you child's present attendance at his/her Centre, School or Special Service.

I would be grateful if you would complete the attached Consent Form and return it to us. We will then contact you to arrange an appointment to see both yourself and your child at the time and place that suits you best. Thank you for taking the time to read this letter and we look forward to meeting you and your child during the Study.

Should you wish to contact us, our number is 086 833 4916, or 6083785 to leave a message.

Yours sincerely,

Joan Murphy
Research Nurse

Project Director: Professor Hilary Hoey, Head of Paediatrics, University of Dublin, Trinity College, Dublin 2.
Consultant Paediatrician/Endocrinologist, National Children's Hospital, Harcourt Street, Dublin 2.

Project Co-ordinator: Dr. Magued Philip, Lecturer in Paediatrics, University of Dublin, Trinity College, Dublin 2.
Ms Joan Murphy, Research Nurse, Department of Paediatrics, National Children's Hospital, Harcourt Street, Dublin 2.

APPENDIX 2

APPENDIX 2: PARENT CONSENT FORM

“A Study On Children With Down Syndrome
in the Eastern Health Board”
(Birth to Sixteen Years of Age)

I understand the *purpose* of the above study. I also understand that it will be a *once off visit* and any findings that need medical attention will be referred back to the Paediatrician/ Doctor already in charge of my child.

I understand that I have the right to withdraw from the Study at any time and this will not effect the quality of the care my child is currently receiving or future services.

I **give/do not give** my consent for a simple blood sample to be taken *only* if required and after discussing it with you.

I hereby give my consent to have my child _____ participate in this study and agree to the collection of all relevant information for this purpose from the centres my child attends.

Address _____

Telephone No. _____

Date _____ *Signature of* _____

Parent/Guardian

Project Director: Professor Hilary Hoey, Head of Paediatrics, University of Dublin, Trinity College, Dublin 2.
 Consultant Paediatrician/Endocrinologist National Children’s Hospital, Tallaght, Dublin 24.

Project Co-ordinator Dr. Magued Philip, Lecturer in Paediatrics, University of Dublin, Trinity College, Dublin 2.
 Research Nurse Ms Joan Murphy, Department of Paediatrics, National Children’s Hospital, Tallaght,
Dublin 24.

APPENDIX 3

Section 3.

DETAILS OF DIAGNOSIS

This section is concerned with how you were told your child's diagnosis.

How much information you received about Down Syndrome and the Services available to help you.

1. When were you told that your child had Down Syndrome? Straight away Days later Weeks later
☐ 1 ☐ 1 ☐ 1
2. Who was with you when you were told the diagnosis? Alone Your Baby Baby's Father Others
☐ 1 ☐ 1 ☐ 1 ☐ 1
3. Was the location private? Yes No Satisfactory Unsatisfactory
☐ 1 ☐ 2 ☐ 1 ☐ 2
4. Was enough time spent with you? ☐ 1 ☐ 2 ☐ 1 ☐ 2
5. Were you offered a follow up discussion? ☐ 1 ☐ 2 ☐ 1 ☐ 2
6. Were you given information about Down's Syndrome? ☐ 1 ☐ 2 ☐ 1 ☐ 2
7. Who gave you the information? Consultant Junior Doctor Midwife
☐ 1 ☐ 1 ☐ 1
8. Did the Social Worker visit you? Yes No When
☐ 1 ☐ 2 _____
9. Were your child's chromosomes checked? ☐ 1 ☐ 2 Don't Know ☐ 3
10. What were the results if known? Trisomy 21 ☐ 1 Others ☐ 2
11. Did both parents have blood taken for genetic testing? Yes No Do not remember
☐ 1 ☐ 2 ☐ 3
12. Were you offered Genetic Counselling ☐ 1 ☐ 2 ☐ 3
13. Did you avail of this service? ☐ 1 ☐ 2
14. If yes, where did you receive this service? _____
15. Were you given any information about other services available to you and your child? Yes No Do not remember
☐ 1 ☐ 2 ☐ 3
16. If yes, what information did you receive (please specify) _____
17. Who gave you this information? _____
18. Any further comments? _____

Section 4.

CENTRE OF CARE/EDUCATION

This section is concerned with Centre(s) your child is attending and the Services being received there

1. Which Centre(s) has your child attended *since birth*? Please tick the centres involved and years attended.

<u>Centre</u>	<u>Where</u>	<u>When (approx. age of child)</u>
a) Cheeverstown House	☐	
b) County Wicklow Assoc.	☐	
c) St. John of God Brothers	☐	
d) St. Michael's House	☐	
e) St. Vincent's Centre	☐	
f) Stewart's Hospital	☐	
g) Other(s)	☐	
h)	☐	
i)	☐	

2. What *services* have you used since your child's birth?

<u>Services</u>	<u>When (approx. age of child)</u>	Satisfied	Dissatisfied
a) Social Work Services	☐	☐	☐
b) Physiotherapy Services	☐	☐	☐
c) Speech & Language Services	☐	☐	☐
d) Occupational Therapy Services	☐	☐	☐
e) Psychology Services	☐	☐	☐
f) Psychiatric Services	☐	☐	☐
g) Early Services	Birth - 2 yrs ☐	☐	☐
	3yrs - 5 yrs ☐	☐	☐
h) Special School	☐	☐	☐
i) Primary School	National ☐	☐	☐
	National + support ☐	☐	☐
	Montessori ☐	☐	☐
a) Senior School	☐	☐	☐
b) Other(s)	☐	☐	☐
	☐	☐	☐

3. Did you have to wait to avail of any of the above services? Yes ☐ 1 No ☐ 2

4. If Yes, (Please specify)

5. What did you find most beneficial with the services?

6. What did you find least beneficial with the services?

Section 5.

IMMUNISATION DETAILS

1. Did your child receive the following vaccinations? Yes No
- a) BCG Birth - 1 month ☐ 1 ☐ 2
- b) Diphtheria & Tetanus (2 in 1) + Oral Polio + Hib ☐ 1 1st ☐ 1 2nd ☐ 2 3rd ☐ 3
Dose Dose Dose
- c) Diphtheria, Tetanus & Pertussis (3 in 1) + Oral Polio + Hib ☐ 2
- d) Measles, Mumps, Rubella (MMR) 15 months ☐ 1 ☐ 2
- e) Booster vaccination at school entry (D.T. + Oral Polio) 4-5 years ☐ 1 ☐ 2
- f) BCG (check) 10 - 14 years ☐ 1 ☐ 2
- g) MMR 11 - 12 years ☐ 1 ☐ 2

Section 6.

FEEDING

(GASTRO INTESTINAL SYSTEM)

1. How was your child fed at birth? Breast fed ☐ 1 Bottle fed ☐ 2 Combination ☐ 3
2. Did your baby suck well? Yes ☐ 1 No ☐ 2 Not sure ☐ 3
3. At what age did your child start to chew well? _____ (months)
4. At what age did your child start on Semi Solids _____ 1 (months) Solids _____ 2 (months)
5. Did your child ever have any of the following? Cause if known Treatment if known (Please Specify)
- a) Recurrent Diarrhoea? Yes ☐ 1 No ☐ 2 _____
- b) Chronic Constipation Yes ☐ 1 No ☐ 2 _____
- c) Recurrent Vomiting Yes ☐ 1 No ☐ 2 _____
6. Did your child ever have any operations Yes ☐ 1 No ☐ 2

7. If Yes, give details of: Operation Age Hospital
- a) 1 2 3
- b) 1 2 3
- c) 1 2 3

8. Any further comments? _____

Section 7.

DEVELOPMENT

1. At what age (in months) did your child start to:
- Sit alone 1 _____ Stand alone _____ 2 Walk alone 3 _____
2. At what age (in months) did your child start to:
- 1 Say single words _____ 2 Put 2 words together _____ 3 Use recognisable sentences _____
- 4 Feed him/herself _____ 5 Dress him/herself _____ 6 Ride a tricycle _____

Section 8.

BEHAVIOURAL DEVELOPMENT

1. Did you ever have any concerns about your child's behaviour?

Sleeping ☐ 1 Eating ☐ 1 Playing with others ☐ 1 Tantrums ☐ 1 Autism ☐ 1

2. If yes, Did you ever seek professional help? Yes ☐ 1 No ☐ 2

3. If Yes, Who looked after your child? _____

4. What age was your child when this problem presented? _____

5. Any further comment? _____

Section 9.

HEART (CARDIO VASCULAR SYSTEM)

1. Did your child ever have a heart problem? No ☐ 1 Yes ☐ 2

2. If Yes, What was the diagnosis? (Please specify) _____

3. At what age did it present? Birth ☐ 1 Other age (Please specify) ☐ 2 _____

4. What tests did your child have done? ECG ☐ 1 ECHO ☐ 1 Cardiac Catheterisation ☐ 1

Others (Please specify) ☐ 1 _____

5. Was the problem treated? a) Surgically (operation) Yes ☐ 1 No ☐ 2

b) With medicines only ☐ 1 ☐ 2

6. Any further comments? _____

Section 10.

GLANDS (ENDOCRINE SYSTEM)

1. Did your child ever have a Thyroid problem? Yes ☐ 1 No ☐ 2 Don't Know ☐ 3

2. Did your child ever have any tests done for this? Yes ☐ 1 No ☐ 2 Don't Know ☐ 3

3. If Yes, Where _____ When _____ How Often _____

4. Has your child reached puberty yet? Yes ☐ 1 No ☐ 2 Age of onset 3 _____ (in years)

5. Do you have any concerns about his/her puberty? ☐ 1 ☐ 2

6. Is your daughter menstruating? ☐ 1 ☐ 2

7. Does your daughter have any problems in this regard? ☐ 1 ☐ 2

8. Who is the person dealing with this problem? GP ☐ 1 Gynaecologist ☐ 2 Others ☐ 3 _____

9. Any further comments? _____

Section 11.

HEARING

1. Did you ever had any concerns about your child's hearing? Yes ☐ 1 No ☐ 2
2. Was your child's hearing ever tested? Yes ☐ 1 No ☐ 2
3. *If Yes* When _____ By Whom _____ How Often? _____
4. What were the results if known (give details) Right Ear 1 _____ Left Ear 2 _____
5. Did your child ever have grommets? Yes ☐ 1 No ☐ 2
6. *If Yes*, which ear had the grommets? Right Ear ☐ 1 Left Ear ☐ 2 How Often _____ 3
7. Did your child ever wear a Hearing Aid? Yes ☐ 1 No ☐ 2
8. *If Yes*, did your child wear the Aid all day? Yes ☐ 1 No ☐ 2
9. Any further comment? _____

Section 12.

VISION

1. Did you ever have any concerns with your child's vision? Yes ☐ 1 No ☐ 2
2. Is there any family history of eye problems before the age of five years? Yes ☐ 1 No ☐ 2
3. Did your child have his/her vision tested? Yes ☐ 1 No ☐ 2
4. *If yes* When _____ By whom _____ How Often _____
5. What were the results if known (give details) Right Eye _____ Left Eye _____
(For example Long-sighted/Short-sighted)
6. Did your child ever wear glasses Yes ☐ 1 No ☐ 2
7. *If yes*, did your child wear the glasses all day? Yes ☐ 1 No ☐ 2
8. Any further comments? _____

Section 13

SPEECH, LANGUAGE AND COMMUNICATION

1. Did you ever have any concern about your child's Speech? Yes ☐ 1 No ☐ 2
2. Was your child assessed? Yes ☐ 1 No ☐ 2
3. *If yes* When _____ By whom _____
4. Did your child receive any therapy? Yes ☐ 1 No ☐ 2
5. Does your child use sign language? Yes ☐ 1 No ☐ 2 Type _____
6. Any further comments? _____

Section 14.

Lungs and Breathing

(RESPIRATORY SYSTEM)

	Yes	No	Mild	Moderate	Severe	How often yearly
1. Did your child ever have Ear Infections?	☐ 1	☐ 2	☐ 1	☐ 2	☐ 3	4
2. Did your child ever get Chest Infections?	☐ 1	☐ 2	☐ 1	☐ 2	☐ 3	4
3. Did your child ever have Asthma?	☐ 1	☐ 2	☐ 1	☐ 2	☐ 3	
4. Did your child ever snore?	☐ 1	☐ 2	☐ 1	☐ 2	☐ 3	

5. Any further comments? _____

Section 15.

BONES MUSCLES AND JOINTS

(MUSCULOSKELETAL SYSTEM)

1. Did your child ever have any joint problem? Yes ☐ 1 No ☐ 2 (Please specify) _____

2. Did your child require treatment? Yes ☐ 1 No ☐ 2

3. What treatment was required? Physiotherapy ☐ 1 Occupational Therapy ☐ 1
 Medical ☐ 1 Surgical ☐ 1 Other ☐ 1 _____

4. Any further comments? _____

Section 16.

CONVULSIONS

	Yes	No	Don't Know
1. Did your child ever suffer from convulsions?	☐ 1	☐ 2	☐ 3
2. If Yes, What tests did your child have done? EEG ☐ 1 CT Scan ☐ 1 MRI Scan ☐ 1 Others ☐ 1 (Please specify) _____			
3. Where were the tests done? _____			
4. Any further comments? _____ _____ _____ _____			

Section 17.

FAMILY SUPPORT

1. Are you a member of a family support group? Yes ☐ 1 No ☐ 2
2. Name of the support group (s) _____
3. Any other comments which you feel will be helpful _____

Caring for a child with Down Syndrome can be difficult in different ways for parents and siblings. The following questions ask about the difficulties or burden you may experience in certain areas:

- | 4. How much burden (difficulty) is there because of : | Large Burden | Moderate Burden | Small Burden | No Burden |
|--|---|------------------------------------|------------------------------------|---|
| a) Disruption to Parent(s) while caring for this child? | ☐ 1 | ☐ 2 | ☐ 3 | ☐ 4 |
| b) Disruption in family routines because of caring for this child? | ☐ 1 | ☐ 2 | ☐ 3 | ☐ 4 |
| c) Disruption in the time for your other children while caring for this child? | ☐ 1 | ☐ 2 | ☐ 3 | ☐ 4 |
| d) Physical problems in this child requiring extra parental care? | ☐ 1 | ☐ 2 | ☐ 3 | ☐ 4 |
| e) Concerns about this child's long-term Health | ☐ 1
Excellent | ☐ 2
Fair | ☐ 3
Poor | ☐ 4
Very Poor |
| 5. Do you feel that your child's general health at present is: | ☐ 1 | ☐ 2 | ☐ 3 | ☐ 4 |
| 6. Do you feel that your child's quality of life is: | ☐ 1 | ☐ 2 | ☐ 3 | ☐ 4 |

7. What do you think could improve the quality of life of your child? _____

8. Is your child on any Nutritional Supplements: Yes ☐ 1 No ☐ 2 Type 3 _____

9. *If Yes* What changes have you seen in your child since starting? _____

YOUR CHILD'S NEEDS

1

This image shows a single sheet of white paper with horizontal blue ruling lines. The lines are evenly spaced and run across the width of the page. There are no margins, text, or other markings on the paper.



Thank you for participating in this Study
Your answers and comments are vitally important

Section 19.

CLINICAL EXAMINATION

Part Two

1. Child's Height _____ cms Centile _____
2. Child's Weight _____ kgs Centile _____
3. Child's Head Circumference (birth to 1 year) _____ cms
4. Mother's Height _____ cms
5. Mother's Head Circumference _____ cms
6. Father's Height _____ cms
7. Father's Head Circumference _____ cms

8. Pubertal Development

- a) Breast Development Stage : 1 2 3 4 5
- b) Pubic Hair Stage : 1 2 3 4 5
- c) Genitals Stage : 1 2 3 4 5

Section 20

Dental Examination

1. Oral Hygiene Normal ☐ 1 Abnormal ☐ 2
2. Gingivitis Yes ☐ 1 No ☐ 2
3. Overcrowding Yes ☐ 1 No ☐ 2
4. Orthodontic Treatment Yes ☐ 1 No ☐ 2
5. Cleft Lip / Palate Yes ☐ 1 No ☐ 2
6. Others _____
7. _____

Section 21.

Skin Examination

1. Skin : 1 Normal ☐
- 2 Abnormal ☐ -> Dry ☐ 1 Infected ☐ 1 Others : _____

Section 22.

C.V.S & Respiratory Examination

1. Pulse _____ (per minute)
2. Resps _____ (per minute)
3. Blood Pressure _____ (systolic/diastolic)
4. Colour Normal ☐ 1 Abnormal ☐ 2 Specify _____
5. Clubbing No ☐ 2 Yes ☐ 1
6. Chest deformities No ☐ 2 Yes ☐ 1 Specify _____
7. Scars No ☐ 2 Yes ☐ 1
8. a) Breath sounds Normal ☐ 1 Abnormal ☐ 2 Specify _____
- b) Heart sounds Normal ☐ 1 Abnormal ☐ 2
- c) Murmur No ☐ 1 Yes ☐ 2 Specify _____

Section 23.

Abdominal Examination

1. Liver Normal ☐ 1 Abnormal ☐ 2 Specify _____
2. Spleen Normal ☐ 1 Abnormal ☐ 2 Specify _____
3. Kidney Normal ☐ 1 Abnormal ☐ 2 Specify _____
4. Hernial Orifices Normal ☐ 1 Abnormal ☐ 2 Specify _____

Section 24.

Musculoskeletal System Examination

1. Tone Normal ☐ 1 Abnormal ☐ 2 Specify _____
2. Joint Dislocation No ☐ 1 Yes ☐ 2 Specify _____
3. Flat Feet No ☐ 1 Yes ☐ 2
4. Did it cause any problem _____ (discomfort on walking)

Section 25.

Vision

Right Eye

Left Eye

- | | | | | | | |
|-----------------|-------------------------------|--------------------------------|------------|-------------------------------|--------------------------------|------------|
| 1. Inspection | N ☐ 1 | Abn ☐ 2 | Spec _____ | N ☐ 1 | Abn ☐ 2 | Spec _____ |
| a) Strabismus | N ☐ 1 | Abn ☐ 2 | Spec _____ | N ☐ 1 | Abn ☐ 2 | Spec _____ |
| b) Nystagmus | No ☐ 1 | Yes ☐ 2 | Spec _____ | No ☐ 1 | Yes ☐ 2 | Spec _____ |
| c) Cataracts | No ☐ 1 | Yes ☐ 2 | Spec _____ | No ☐ 1 | Yes ☐ 2 | Spec _____ |
| d) Keratoconus | No ☐ 1 | Yes ☐ 2 | | No ☐ 1 | Yes ☐ 2 | |
| 2. Light Reflex | N ☐ 1 | Abn ☐ 2 | | N ☐ 1 | Abn ☐ 2 | |
| 3. Eye Movement | N ☐ 1 | Abn ☐ 2 | | N ☐ 1 | Abn ☐ 2 | |
| 4. Others | N ☐ 1 | Abn ☐ 2 | Spec _____ | N ☐ 1 | Abn ☐ 2 | Spec _____ |
| 5. _____ | N ☐ 1 | Abn ☐ 2 | Spec _____ | N ☐ 1 | Abn ☐ 2 | Spec _____ |

/ Kay Pictures / Snellen Letters

Section 26.

Development

1. Gross Motor Normal ☐ 1 Delayed ☐ 2 Specify _____
2. Fine Motor Normal ☐ 1 Delayed ☐ 2 Specify _____
3. Speech Normal ☐ 1 Delayed ☐ 2 Specify _____
4. Socialisation Normal ☐ 1 Delayed ☐ 2 Specify _____

Section 27.

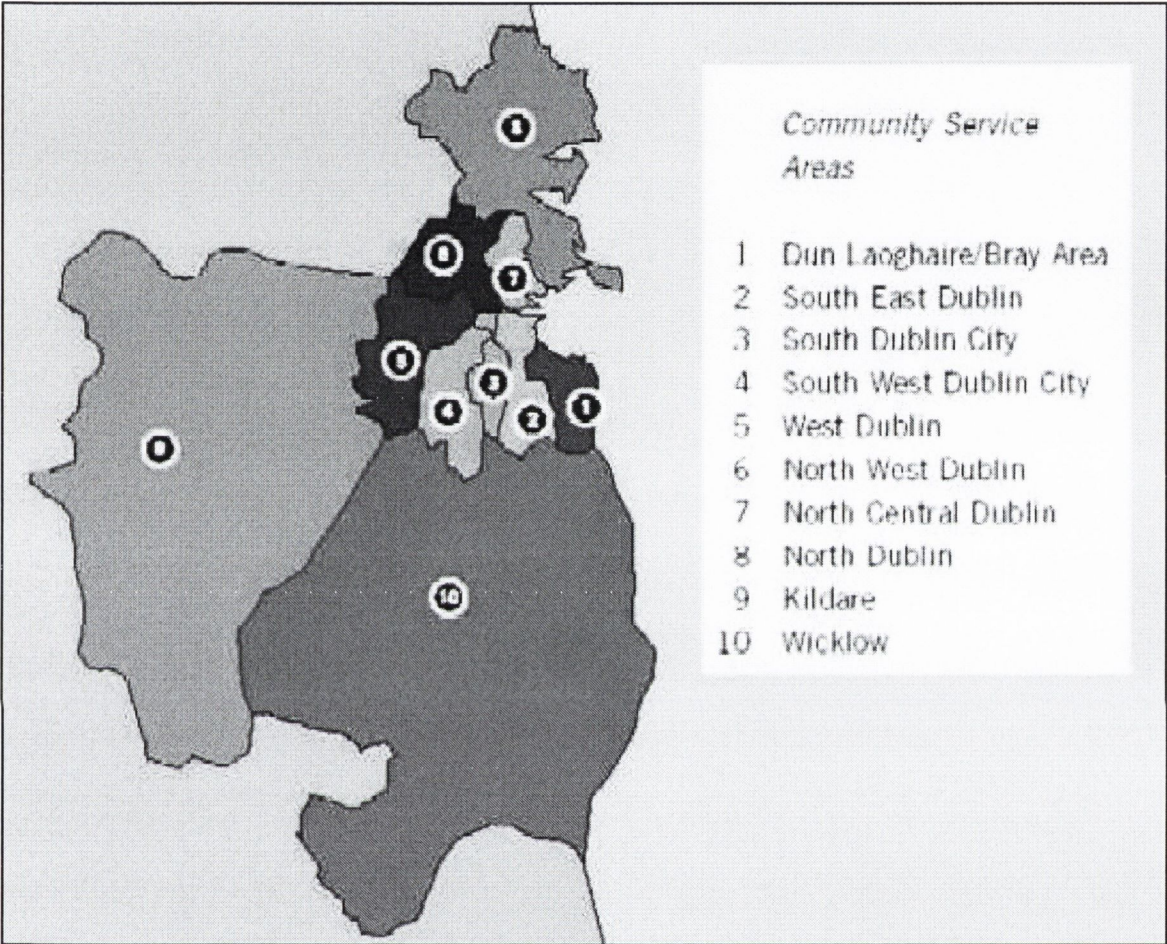
Hearing

- | | | | | |
|--|------|-----------|---|----------|
| 1. Otoscopy | 1 | Right Ear | 2 | Left Ear |
| 2. Distortion Product Oto Acoustic Emissions Results | 1 | Right Ear | 2 | Left Ear |
| 3. Tympanogram Results | 1 | Right Ear | 2 | Left Ear |
| 4. Pure Tone Audiometry | 1 | | 2 | |
| 5. Thyroid TSH Results | mU/l | | | |

APPENDIX 4

Appendix 4: Study Area

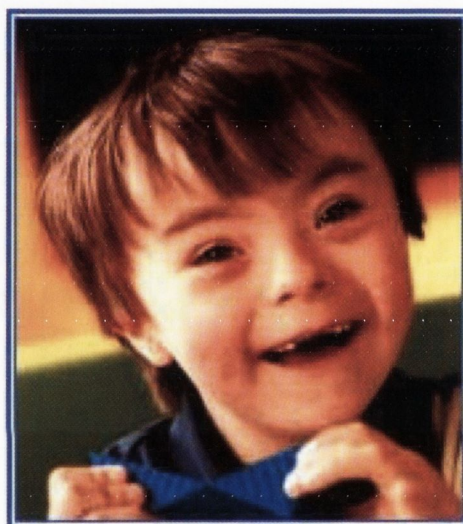
Community Service Area in the Eastern Regional Health Authority



APPENDIX 5



**Medical Management
of
Children & Adolescents
with
Down Syndrome in Ireland**



APPROVED GUIDELINES

**Down's Syndrome Medical Interest Group
(DSMIG) (UK & Ireland)**



**Department of Paediatrics
University of Dublin, Trinity College
The National Children's Hospital, AMNCH, Tallaght**



Medical Management
of
Children & Adolescents
with
Down Syndrome in Ireland

Approved Guidelines

Professor Hilary MCV Hoey

Joan Murphy RSCN MSc Paediatrics

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INTRODUCTION

Down Syndrome is the most common congenital cause of developmental disability in Ireland with a birth prevalence of 1 in 546 live births, which is the highest in Europe. It is well recognised that as a group they have a high incidence of treatable medical disorders. All studies show that early intervention carries a better outcome for their general health, quality of life and life expectancy. With medical progress many now live into their sixties.

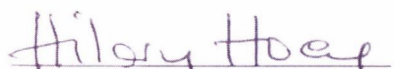
In order to assess the medical and psychosocial needs of children and adolescents with Down syndrome in Ireland we conducted a surveillance study in the Eastern Health Board (now the Eastern Regional Health Authority).

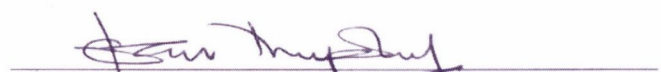
Medical guidelines were originally developed by the Down Syndrome Medical Interest Group for use in the United Kingdom. Our research provided the necessary evidence based data to construct medical management guidelines appropriate for children and adolescents with Down syndrome in Ireland. Many health care professionals with expertise in the management of children and adolescents with Down syndrome in Ireland have contributed to the Irish guidelines. These guidelines have now been approved by The Irish College of General Practitioners, The Faculty of Paediatrics of The Royal College of Physicians of Ireland and the Faculty of Public Health Medicine of The Royal College of Physicians of Ireland. Implementation of these guidelines is now urgently required.

We are very grateful to Dr. Jennifer Dennis, Director of Information and Research, DSMIG, Dr. Liz Marder, Vice Chairman DSMIG and all members of the group for the enormous amount of work that they have undertaken in the development of the guidelines.

We are also very grateful to the many Irish health professionals who supported and assisted us in carrying out the study and in particular we wish to express our sincere thanks to all the parents and children who took part in the study.

We wish to thank Dr. Sheila Macken, Mr. Don McShane, Dr. Philip Mayne, Dr. Desmond Duff, Dr. Myra O'Regan, Ms Aoife Walsh, Mr. Michael O'Keefe, Mr. Esmond Fogarty, the late Dr. Zachary Johnson, Ms. Virginia Delaney, Dr. Siobhan Murnaghan, Dr. Frances Kelly, Professor Denis Gill, Dr. Owen Hensey, Dr. Mary McKay, Dr. Edwina Daly, Dr. Edna Roche, Dr. Judith Meehan, Ms Mary Cronin, Mr. Michael Harney, Dr. Colm Costigan, Dr. Louis Ramsey, Dr. Jervais Corbett, Bro. Finnian Gallagher, Dr. Brendan McCormick, Dr. Noel McDonnell, Dr. Mary Staines, Dr. Mona Byrne, Dr. Martin McLaughlin, Dr. Mona O'Donnell, Professor O. Conor Ward, Dr Austin O'Carroll, the Area Medical Officers in the Eastern Regional Health Authority and all the Paediatricians and Staff in the three Children's Hospitals and the Developmental and Educational Centres, who supported us in many ways throughout the study and accommodated us at all times.


Professor Hilary MCV Hoey


Joan Murphy RSCN MSc Paediatrics, Dip Stats

We are very grateful to Footsteps, Down Syndrome Ireland, the Minister for Health and Children and the Provost, University of Dublin, Trinity College for sponsoring this Research. We also thank the National Children's Hospital Foundation for sponsoring the scientific meeting to launch the guidelines in May 2001

BASIC MEDICAL SURVEILLANCE ESSENTIALS

FOR PEOPLE WITH DOWN SYNDROME

GROWTH

Short stature is a recognised characteristic of most people with Down syndrome. Average height of children with Down syndrome at most ages is around the 2nd centile for the general population. For the majority the cause of growth retardation is not known.¹ Some conditions leading to poor growth - congenital heart disease,^{2,3} sleep related upper airway obstruction,⁴ coeliac disease,^{5,6} nutritional inadequacy due to feeding problems, and thyroid hormone deficiency^{7,8} occur more frequently among those with the syndrome. Regular surveillance of growth, general health, nutritional and thyroid status should aid in early identification of pathological causes of growth retardation.

UK/ Ireland growth charts for healthy children with Down syndrome from birth to 18 years are now available.⁹ These reference values are essential for assessing linear growth. However, as many older children and adults with the syndrome tend to be overweight,^{10,11} the reference values for weight should not be used as a standard that children should aim to achieve. Instead the body mass index (BMI) data included on the charts should be used to aid the assessment of overweight.

Guidelines:

1. We suggest that it is good practice to record and chart height and weight frequently in the first two years using Down syndrome specific charts. Thereafter measurements should be made at least annually throughout childhood and at regular intervals in adult life. Regular measurements of this sort are likely to be sensitive early indicators of the many medical problems which are over represented in the syndrome
2. Children normally lose weight after birth but regain the weight by approximately day 10. Preliminary data suggest that many babies with Down syndrome do not regain birth weight until around 1 month of age.¹² This is not reflected in the growth charts because of their cross sectional nature. This early failure to thrive is usually due to feeding difficulties many of which resolve after the first few weeks. From 1 month weight should increase parallel to the centiles. Failure to do so should be investigated.
3. Of those with measurements below the 2nd centile some will have major pathology but some may be failing to thrive for other reasons – e.g. feeding difficulties.¹³ Such children should have their dietary intake evaluated and may need to be referred to a paediatrician or paediatric endocrinologist for assessment.
4. The Down syndrome specific growth charts clearly reflect the tendency to excess weight gain among the UK and Irish study sample particularly in later childhood. Hence standard BMI charts have been included on the growth charts. We suggest that all those over age 5 years with weight above the 75th centile should be charted on these BMI charts. Those above the 91st BMI centile should be carefully monitored. Those above the 98th BMI centile should be considered for further assessment and guidance.
5. Although there is a high prevalence of overweight/obesity^{10,11} among people with Down syndrome this is not inevitable. As with the general population weight is influenced by environmental^{11,14} as well as biological factors.

6. Appropriate anticipatory guidance regarding diet and physical activity should be given for all those with the syndrome.
7. Thyroid function should always be checked in those with accelerated weight gain.
8. In childhood growth spurts and plateaux occur as in all children but among the Down syndrome population these tend to be more prolonged. They are not reflected in the smoothed curves of a standardised chart.
9. The Down syndrome specific chart suggests an absence of pubertal growth spurt. However those with the syndrome do have an adolescent growth spurt. It is usually less vigorous than in the general population. Puberty may occur at an earlier age and requires *anticipation together with education and support for parents and child*.¹⁶ Early onset of puberty has a limiting effect on final height.
10. As with all children, head circumference should be measured regularly and charted on Down syndrome specific charts. If there is any cause for concern subsequent measurements should be made.
11. The use of growth hormone in Down syndrome is still being evaluated. There is no evidence that it should be prescribed except in the unusual situation of concurrent primary growth hormone deficiency.¹⁷⁻²⁰
12. The influence of parental height on target height appears to be variable.²¹

UK/Ireland Down's Syndrome Specific Growth Charts are available from Harlow Printing Ltd, South Shields, Tyne and Wear. NE33 4PU. Tel 0044 191 455 4286

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BASIC MEDICAL SURVEILLANCE ESSENTIALS FOR PEOPLE WITH DOWN SYNDROME

CARDIAC DISEASE CONGENITAL AND ACQUIRED

1. Between 40 and 50% of babies with Down syndrome have congenital heart defects. Of these 30-40% have complete atrioventricular septal defects (AVSD).^{1,2} Most AVSD can be successfully treated if the diagnosis is made and the baby referred for full corrective surgery before irreversible pulmonary vascular disease is established.³
2. There must be a high level of clinical suspicion of congenital heart disease for all newborns with the syndrome
3. It is essential to establish the cardiac status of every child by age 6 weeks.⁴⁻¹⁰
4. Clinical examination alone is insufficient to detect even *some of* the most serious abnormalities.^{2,8,11}
5. It is very unlikely that a serious abnormality requiring early intervention (AVSD) will be missed if one of the following courses of action is taken.^{2,10,12}
 - (a) Clinical examination plus electrocardiogram (ECG) and chest X-ray (CXR) for all newborns and again at age 6 weeks, followed by echocardiography only for those with abnormal findings.
 - or**
 - (b) Clinical examination, ECG plus echocardiogram in the newborn period, both carried out by an appropriate person (see below 6).

However, even if early investigations are reported as 'normal', if a child develops signs or symptoms of cardiac disease appropriate investigations must take place, as structural problems may not have been evident at an earlier age.

6. It is not always essential to refer newborn babies with the syndrome to a cardiologist. However, all clinical examinations should be by a doctor experienced in the care of newborns; an experienced paediatrician should review CXR and ECG findings; echocardiograms should be carried out and reviewed by staff with appropriate paediatric experience *under the supervision of a paediatric cardiologist. Telemedicine may provide a useful intermediate step between paediatrician and cardiologist.* Those with suspected problems should be referred for immediate cardiological review so that intervention, if necessary, can take place before pulmonary vascular disease develops.
7. It is recognised that minor heart defects (atrial septal defect and small ventricular septal defects) may be missed in those children who do not have echocardiograms but these should declare themselves clinically, as for any child, in the normal course of child health surveillance.
8. Parents and carers of all children with Down syndrome with heart lesions should be given verbal and written information about infective carditis preventive measures.
9. It *should* be remembered that despite a normal echo at birth children with Down

syndrome, like all other children, can develop *symptoms and signs of* heart disease at a later age *e.g.* secondary to airway problems.¹³

10. There is an increased incidence of mitral valve prolapse and of aortic regurgitation in adults. This has implications for infective carditis prevention particularly because of the high incidence of periodontal disease among this population. We therefore recommend an echo screen for all people with Down's syndrome early in adult life.^{9,14}
11. If a potential risk situation for infective endocarditis arises for an adult with Down syndrome who has not had an adult echo, preventive prophylactic measures should be started.

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We are very grateful to Dr Desmond Duff, Consultant Cardiologist, Our Lady's Hospital for Sick Children, Crumlin for his support with the development of the guidelines for children and adolescents with Down syndrome in Ireland

Professor Hilary MCV Hoey

Joan Murphy RSCN MSc Paediatrics

BASIC MEDICAL SURVEILLANCE ESSENTIALS FOR PEOPLE WITH DOWN SYNDROME

THYROID DISORDER

1. At all ages thyroid disorder (usually hypothyroidism) occurs more frequently in people with Down syndrome than in the general population.¹⁻⁵ Around 10% of the school age population have uncompensated hypothyroidism. The prevalence increases with age.⁶ If undiagnosed, thyroid disorder constitutes a significant cause of preventable secondary handicap. Diagnosis on clinical grounds is unreliable.^{7,8} Biochemical screening is essential. As in the general population those with significant abnormalities of any thyroid function test (TFT) should either be treated (if there is uncompensated hypothyroidism) or kept under close clinical and biochemical surveillance.
2. All babies in the U.K and Ireland have a neonatal screen for hypothyroidism.⁹
3. Biochemical testing, including estimation of T4, TSH, and thyroid antibodies should be carried out at least once every two years from age 1 year and throughout life.^{6,10}
4. Information is currently coming in from several areas where the feasibility of fingerprick TSH Guthrie screening is being investigated. Preliminary evaluation suggests that this may prove an effective screening procedure, *which may be possible annually, once the appropriate structures, personnel and funding are in place.*
5. Transient changes may occur.^{10,11} Mildly raised TSH ($5-10\text{mU/l}$) or the presence of antibodies with normal T4 and no clinical evidence of hypothyroidism may not warrant treatment.^{12,13} It does however indicate increased likelihood of developing uncompensated hypothyroidism. Such people should therefore be tested more frequently than those with normal test results. A specialist opinion may be required.
6. Clinicians should always bear in mind the high prevalence of thyroid disorder in people with Down syndrome and have a low threshold for testing thyroid function if there is any clinical suspicion at times between biochemical testing.
7. As in the general population key clinical pointers are lethargy and/or changes in affect, cognition, growth, or weight.
8. Consideration of hypothyroidism is mandatory in the differential diagnosis of depression and dementia.^{14,15}
9. The possibility of hyperthyroidism should also be born in mind.^{5,16}

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BASIC MEDICAL SURVEILLANCE ESSENTIALS FOR PEOPLE WITH DOWN SYNDROME

OPHTHALMIC PROBLEMS

1. There is a high incidence of ocular disorder among people with Down syndrome.¹⁻⁶ Refractive errors and Strabismus (squint) may occur at an early age. *Over 50% of people with Down's syndrome wear glasses.* Cataract and/or Glaucoma may occur in infancy.⁷ Nystagmus is present in about 10% and *Brushfield Spots are present in the eye in many children at birth.* Keratoconus,^{8,9} is more common in adolescents and young adults. *In particular as the patient gets older, high myopia (near sightedness), can occur.*³ If untreated, many of these disorders are a significant cause of preventable secondary handicap.
2. All newborns with Down syndrome should have an eye examination carried out at birth to exclude congenital cataract *and glaucoma* and should thereafter be included in community screening programmes.
3. Visual behaviour must be monitored by a paediatrician. Those who start to squint or show other abnormalities of gaze, visual behaviour or attention or changes in the appearance of the eye or excessive watering in the first year of life should be referred for ophthalmological review in the normal way.
4. All children with Down syndrome should have a formal ophthalmological examination including orthoptic assessment, refraction and fundal examination during the second year of life. The majority may have some deviation from normal and should be kept under close review
5. A further *formal ophthalmological* examination should be performed at around the age of four years. If hypermetropia is not present at this age it is not likely to occur later on.
6. Children and adults with Down's syndrome *will respond to a variety of visual testing and this depends very much on their developmental age. Some will be able to perform the routine, ordinary visual testing. Others will require more specialized visual tests. A distraction free environment will optimise performance.* Distance vision should be checked at every review.
7. After the age of 4 years vision should be checked every two years *by an ophthalmologist or optometrist throughout life.*
8. As with all children, if at any age visual acuity deteriorates a specialist opinion is required.
9. If at age 4 years or thereafter, a child has visual acuity assessed as at least 6/9 on a linear Snellen chart then subsequent testing by the school visual clinic service is adequate.
10. Any child identified by the school visual clinic service as less than 6/9 vision should be referred to *a Community Ophthalmologist.*

11. Any child identified *with less than 6/12* on a linear Snellen chart should be referred to Community Ophthalmologist for further assessment.
12. *Local opticians* give an excellent service but subjects who are not cooperative in this setting should be referred to a specialist clinic.
13. Any child or adult with pain, and/or changing vision, *visual disturbance and/or* red eye, should be referred for urgent specialist opinion.

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BASIC MEDICAL SURVEILLANCE ESSENTIALS FOR PEOPLE WITH DOWN SYNDROME.

HEARING IMPAIRMENT

1. Well over 50% of people with Down syndrome have significant hearing impairment, which may be mild, moderate, severe or profound (30 - >95 dB HL).¹⁻³ Sensorineural and/or conductive loss may be present at any age.³⁻¹⁰ Hearing impairment can be successfully managed in this population.¹¹⁻¹⁵ If undetected it is likely to be a significant cause of preventable secondary handicap.^{3,6,12,14} Lifelong audiological surveillance is essential for all.¹⁶⁻²¹ The main cause of conductive loss is persistent otitis media with effusion (OME, glue ear). The natural history of OME and response to intervention differ from that in the general population hence local surveillance and management protocols need to be set up specific to people with Down's syndrome.^{4,6,13,15,22}
2. People with Down syndrome of all ages should have rapid access to specialist audiology services.⁴
3. Because of an increased incidence of congenital sensorineural loss newborns with Down syndrome should be included in neonatal screening programmes where available.^{1,14,19} This does not preclude the need for ongoing surveillance.¹⁶
4. Guidance for parents of children with Down syndrome should include discussion about hearing problems and their management, supported by good quality written information.¹⁹
5. Whether or not a baby with Down syndrome has passed a neonatal screen all should have full audiological assessment between age 6 and 10 months. This should include measurement of auditory thresholds, impedance testing and otoscopy.^{10,19,21} To ensure inclusion of the child with Down's syndrome participation in existing child health hearing surveillance programmes should be encouraged.
6. Therefore by 10 months it should have been established whether or not a child with Down syndrome has any degree of permanent hearing loss with or without OME. A clear management plan must have been agreed with the parents and intervention instigated where necessary.
7. In the second year (usually around 18 months) all children with Down syndrome – whatever their previous hearing status - should have further audiological review carried out in a manner appropriate for a child with learning disabilities. This should include assessment of auditory thresholds, impedance testing and otoscopy. This should be repeated at least yearly until age 5 and thereafter 2 yearly for life. More frequent testing will be necessary if problems exist.
8. Transition of care from paediatric to adult services should involve direct transfer of care to a named person.
9. At all ages people with Down's syndrome have narrow ear canals, which predispose to accumulation of wax.⁵ This may affect impedance testing and hearing. Early management to clear wax would be desirable to remove any further impact on hearing loss.

10. Most people with Down syndrome are able to respond to standard tests – e.g. distraction; speech discrimination; pure tone audiometry (*play or standard*); and visual reinforcement audiometry. These tests must be *performed by professionals trained in audiology with experience in working with people with learning disabilities*. Threshold measurement tests appropriate to developmental age must be used.^{7,21}
11. Because of increased incidence of sensorineural as well as conductive loss the frequency range tested should include 8000Hz whenever feasible as failure at this level may be an early warning of impending high frequency sensorineural deafness.^{3,23}
12. Diagnostic Auditory Brain Stem (ABR) responses in people with Down syndrome must be interpreted with caution.^{2,23} *A Child with Down syndrome with a failed ABR may require Oto Acoustic Emissions (OAEs) to distinguish cochlear from neurological pathology.*⁶
13. As in the general population all those who are hearing impaired should have access to specialist hearing support services (Speech and Language Therapy; Teachers of the deaf; etc)
14. At all ages particular attention should be paid to the treatment of suppurative nasal and ear conditions.^{4,20}
15. In adults with the syndrome hearing assessment is essential in the differential diagnosis of depression and dementia.³

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BASIC MEDICAL SURVEILLANCE ESSENTIALS FOR PEOPLE WITH DOWN SYNDROME.

CERVICAL SPINE INSTABILITY

1. People with Down syndrome have a small risk for acute or chronic neurological problems caused by cervical spine instability.^{1,2}
2. Currently there is no screening procedure, which can predict those at risk. In particular cervical spine x-rays in children have no predictive validity for subsequent acute dislocation/ subluxation at the atlantoaxial joint.³⁻⁷
3. Children with Down syndrome should not be barred from sporting activities because there is no evidence that participation in sports increases the risk of cervical spine injury any more than for the general population.^{6,8}
4. Although the risk of injury is small, if any child or adult with Down syndrome needs an anaesthetic, the anaesthetist and recovery room staff must always be reminded of the diagnosis, so that appropriate care can be taken to avoid cervical injury, whilst manipulating the head and neck in the unconscious subject.⁹
5. Although the risk of injury is small, if a person with Down syndrome is involved in a road traffic accident personnel involved in their care should be alerted to the possibility of cervical spine instability and of the need for particular care relative to this.^{1,5}
6. If a person with Down syndrome develops pain behind the ear or elsewhere in the neck, abnormal head posture, torticollis, deterioration of gait, manipulative skills, or bowel and /or bladder control they should be referred immediately to an appropriate specialist (usually a neurologist or a spinal orthopaedic surgeon).

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DOWN SYNDROME MEDICAL MANAGEMENT GUIDELINES

Suggested schedule of health checks taken from Guidelines

	Growth	Heart	Thyroid	Sight	Hearing
Birth–6 wks	Length/weight/head circumference - Plot on Down Syndrome Specific Growth Charts*	Clinical examination ECG + Chest X-ray Birth and 6 wks or Clinical examination + ECG + Echocardiogram Birth and 6 wks	Routine Guthrie test	Eye Examination, exclude cataract and glaucoma	Neonatal screening Where available
6-10 months	Growth assessment as above at each routine visit*			Visual behaviour exclude squint	Full audiological review (Otoscopy, Impedance, Hearing thresholds)
12 months	Growth assessment as above at each routine visit*	Dental Advice	Full Thyroid function tests or TSH (Guthrie) yearly when available	Visual behaviour exclude squint	
18-24 months	Growth (height/weight) as above*	Dental Advice & examination of teeth	Full Thyroid function tests or TSH (Guthrie) yearly when available	Ophthalmological examination including Orthoptic screening, refraction and fundal examination	Full audiological review as above
3 – 3 ½ years	Growth (height/weight) assessment as above*	Dental Advice & examination of teeth	Full Thyroid function tests or TSH (Guthrie) yearly when available		Full audiological review as above
4 – 4 ½ years	Growth (height/weight) assessment as above*	Dental Advice & examination of teeth	Full Thyroid function tests or TSH (Guthrie) yearly when available	Ophthalmological examination as above	Full audiological review as above

* Encourage a healthy lifestyle (healthy eating and regular exercise) at all times.

From Age 5 to 19 years

Paediatric Medical Review Annually

Cardiology	Echo in early adult life to rule out mitral valve prolapse
Hearing	2 yearly audiological review as above
Vision	2 yearly ophthalmological examination including refraction and fundal examination
Thyroid	2 yearly from 5 years (venous) or annual fingerprick TSH, when appropriate structures, personnel and funding are in place

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Down's Syndrome Medical Interest Group (DSMIG UK & Ireland)
Down's Syndrome Medical Information Services (DSMIS)

DSMIG was launched in 1996. It is a network of doctors from the UK and Ireland whose aim is to share and disseminate information about the medical aspects of Down's syndrome and to promote interest in the specialist management of the syndrome. The group meets twice a year at the Royal Society of Medicine in London.

Initiatives to date include:

- The production of guidelines for basic medical surveillance essentials for people with Down's syndrome
- The production and nationwide distribution of a special insert for babies born with Down's syndrome for the UK national parent held personal child health record (PCHR)
- Production of UK/Ireland growth charts for children and adolescents with Down's syndrome.
- Organisation of regional DSMIG road shows for health care professionals
- Setting up of Down's Syndrome Medical Information Services (DSMIS) – the information arm of DSMIG The remit of this organisation is to provide information to the health care professionals about the medical aspects of Down's syndrome. This service is complementary to that provided for parents by the National Down's Syndrome Support Groups
- Launch of a temporary website in 2000 with a projected launch date for a full information site in June 2001

We have no health service funding or corporate sponsorship. We have to date received financial help towards specific projects and administration costs from the DSA: Marks and Spencer plc; Mencap City Foundation; The David Solomon Trust; Harlow Printing; and Children Nationwide. We ourselves accrue some monies from fees charged for medical road shows; from occasional personal donation of lecture fees etc and from Royalty payments on the growth charts and PCHR

Currently we work under the charitable umbrella of the Nottingham Community Health NHS Trust Charitable Funds but are seeking charitable status in our own right.

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BASIC MEDICAL SURVEILLANCE ESSENTIALS FOR PEOPLE WITH DOWN SYNDROME



**Guidelines of the
Down's Syndrome Medical Interest Group
DSMIG (UK & Ireland)
IRISH EDITION**

We are grateful to the Association for the Prevention of Disabilities for financial support for initial meetings of the surveillance essentials development group and to the Learning Disability Forum of the Royal Society of Medicine and Mencap City Foundation for ongoing support for a number of initiatives.



Background notes

(As service delivery varies in different countries some minor adjustments to the guidelines have been made for Ireland and these appear in italics)

People with Down syndrome do not have unique medical problems, which differ from the general population. However some medical conditions are heavily over represented among those with the syndrome. Most of these are treatable disorders, which if undiagnosed, impose an additional but preventable burden of secondary handicap.

These surveillance guidelines have been developed on the basis of available evidence by a group of clinicians with a special interest in Down syndrome. They are updated as new research and audit evidence becomes available. The overall aim is to help ensure equitable provision of basic essential medical surveillance for all children with Down's syndrome in the UK and in the Republic of Ireland. The Royal College of Paediatrics and Child Health has been supportive of the venture and we have had guidance from the Centre for Evidence Based Child Health.

A set of background notes is being developed which cover the evidence on which the guidelines are based. Currently these are completed for the cardiac and hearing sections and are available electronically via jendennis@dsmig.org.uk

The guidelines are not a blueprint for Gold Star services. Their purpose is to set out a minimum safe standard of basic medical surveillance which we consider essential for all those with the syndrome. This we consider to be the identification of **cardiac disease, thyroid disorder, hearing impairment** and **ophthalmic problems** and the appropriate monitoring of **growth**. We have also included information which we hope will increase understanding of the complex issues surrounding **cervical spine instability**. We are currently producing clinical awareness notes covering other conditions which are over represented in the syndrome. In parallel with the guidelines we have produced **UK/Ireland Down's syndrome specific growth charts and a special insert for UK Personal Child Health Record** (The Red Book) for children born with Down's syndrome.

These are available from:

Harlow Printing Ltd, South Shields, Tyne and Wear. NE33 4PU. Tel 0044 191 455 4286

The guidelines are available in electronic format on www.dsmig.org.uk The site also features the PCHR insert and an order form for the growth charts and PCHR

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Dr Liz Marder. Paediatrician. Nottingham.
Medical Adviser. Down's Syndrome Association.

Dr Marion McGowan. Paediatrician.
St Helier NHS Trust. Surrey

Professor Ben Sacks. Psychiatrist in Learning Disability
Charing Cross & Westminster Medical School. London.

Mr Ross Scrivener. Research and Audit Co-ordinator
Royal College of Paediatrics and Child Health

Dr Ajay Sharma. Paediatrician.
Optimum Health Services. Southwark.

Dr Barbara Stewart. Paediatrician. Oxford.

Professor O. Conor Ward. Paediatrician/Cardiologist. Dublin
Past Medical Advisor, Irish Down's Syndrome Association

APPENDIX 6

D.o.B

Weight	Signature
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BOYS DOWN'S SYNDROME GROWTH CHART

UK/Republic of Ireland cross-sectional reference: 2000

STICKER

These charts are based on data from around 6000 measurements of 1100 children living throughout the UK and Republic of Ireland (Styles et al - in preparation). Growth can be charted from term to 18 years. Children with significant cardiac disease or other major pathology are excluded from the study population. In addition, data for those born before 37 completed weeks were excluded up to age two. The charts are therefore representative of healthy children with Down's syndrome growing in the U.K. and Republic of Ireland.

The charts were commissioned by the UK Down's Syndrome Medical Interest Group (DSMIG) and the data collected by Dr Mary Styles on DSMIG's behalf. The centiles were compiled under the guidance of Professor Michael Preece with statistical analysis provided by Professor Tim Cole, both of the Institute of Child Health, London. Data were analysed by Cole's LMS method. Dr Styles' data collection was funded by the Child Growth Foundation and remains the copyright of DSMIG.

PCHR - The charts are also available in A5 format for inclusion with the special PCHR Down's Syndrome insert.

Preterm babies - We do not as yet have sufficient information to compile centiles for preterm babies with Down's syndrome. Measurements for those born before 37 completed weeks should not be plotted on the charts until the expected date of delivery (EDD) is reached. Thereafter they should be charted relative to EDD for at least a year. Those born at 38 weeks or later should be charted in the normal way from the EDD line.

More information about growth monitoring for children with Down's syndrome is included in the Medical Surveillance Guidelines for people with Down's Syndrome produced by the Down's Syndrome Medical Interest Group. These are available from the address given below.

Overweight and underweight - Action guidelines:

Many older children with Down's syndrome are overweight and this is clearly reflected in this study population. Hence this reference data should not be used as a standard that children should aim to achieve. As with all children weight must be related to stature. Any child aged 5-18 years whose weight falls within the shaded area above the 75th centile should be charted on the BMI chart (see right). Those above the 98th centile on the BMI charts are significantly overweight and referral for further assessment and guidance should be considered.

Of those falling below the 2nd centile some will have major pathology, but some may be failing to thrive for other reasons - eg because of feeding difficulties. Again such children may need further assessment and guidance.

How to calculate BMI (Body Mass Index)

Divide weight (kg) by square of length/height (m²)
e.g. when weight = 25kg and length/height = 1.2m (120cm),
BMI = $25 \div (1.2 \times 1.2) = 17.4$

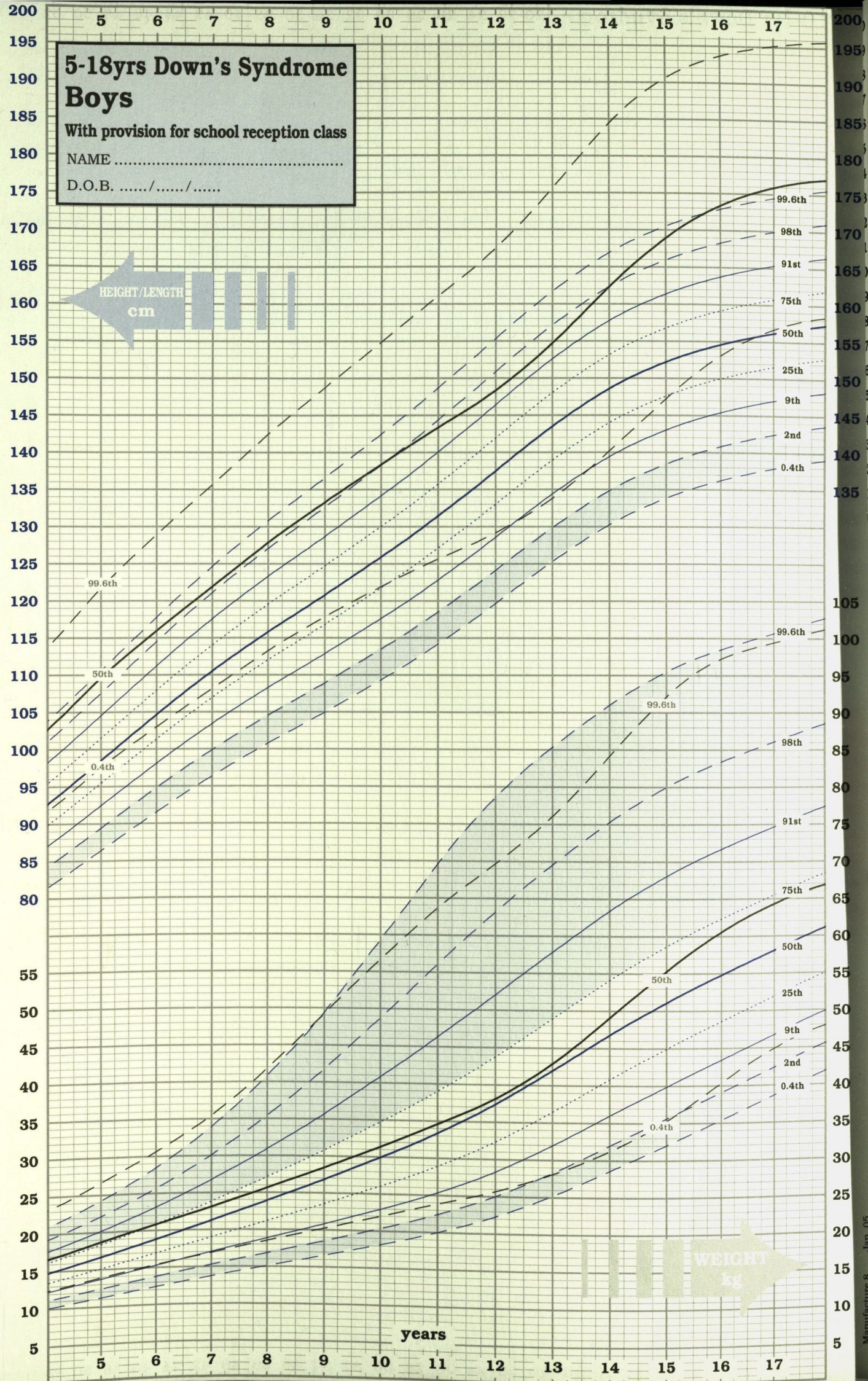
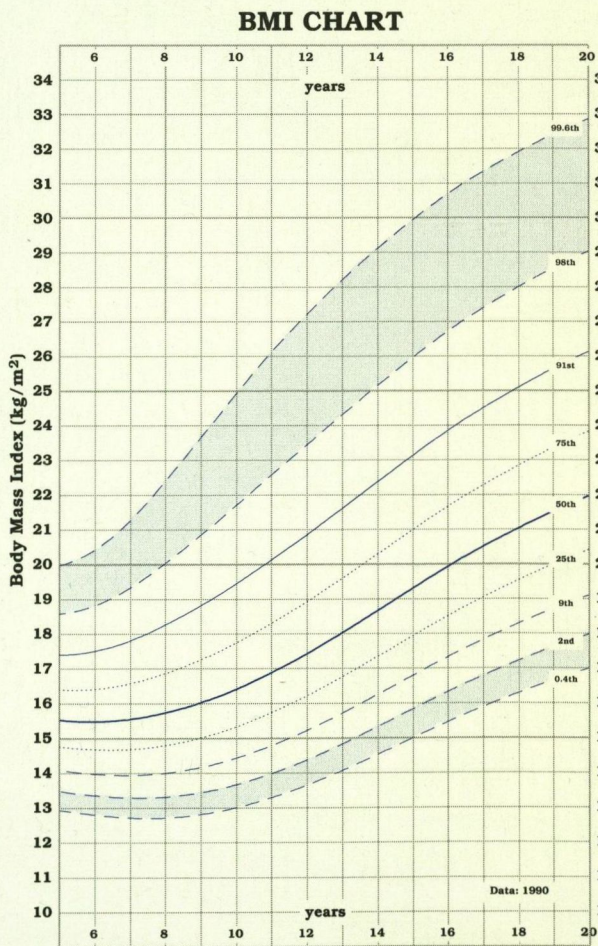
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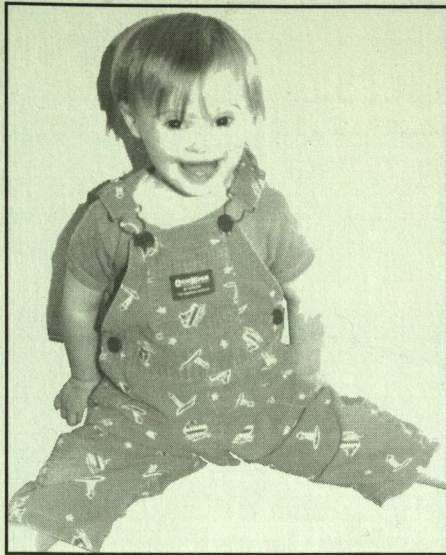
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GIRLS DOWN'S SYNDROME GROWTH CHART

UK/Republic of Ireland cross-sectional reference: 2000

STICKER

These charts are based on data from around 6000 measurements of 1100 children living throughout the UK and Republic of Ireland (Styles et al - in preparation). Growth can be charted from term to 18 years. Children with significant cardiac disease or other major pathology were excluded from the study population. In addition, data for those born before 37 completed weeks were excluded up to age two. The charts are therefore representative of healthy children with Down's syndrome growing in the U.K. and Republic of Ireland.

The charts were commissioned by the UK Down's Syndrome Medical Interest Group (DSMIG) and the data collected by Dr Mary Styles on DSMIG's behalf. The centiles were compiled under the guidance of Professor Michael Preece with statistical analysis provided by Professor Tim Cole, both of the Institute of Child Health, London. Data were analysed by Cole's LMS method. Dr Styles' data collection was funded by the Child Growth Foundation and remains the copyright of DSMIG.

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Overweight and underweight - Action guidelines:

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Of those falling below the 2nd centile on the height and weight charts some will have major pathology, but some may be failing to thrive for other reasons - eg because of feeding difficulties. Again such children may need further assessment and guidance.

How to calculate BMI (Body Mass Index)

Divide weight (kg) by square of length/height (m²)
e.g. when weight = 25kg and length/height = 1.2m (120cm),
BMI = $25 \div (1.2 \times 1.2) = 17.4$

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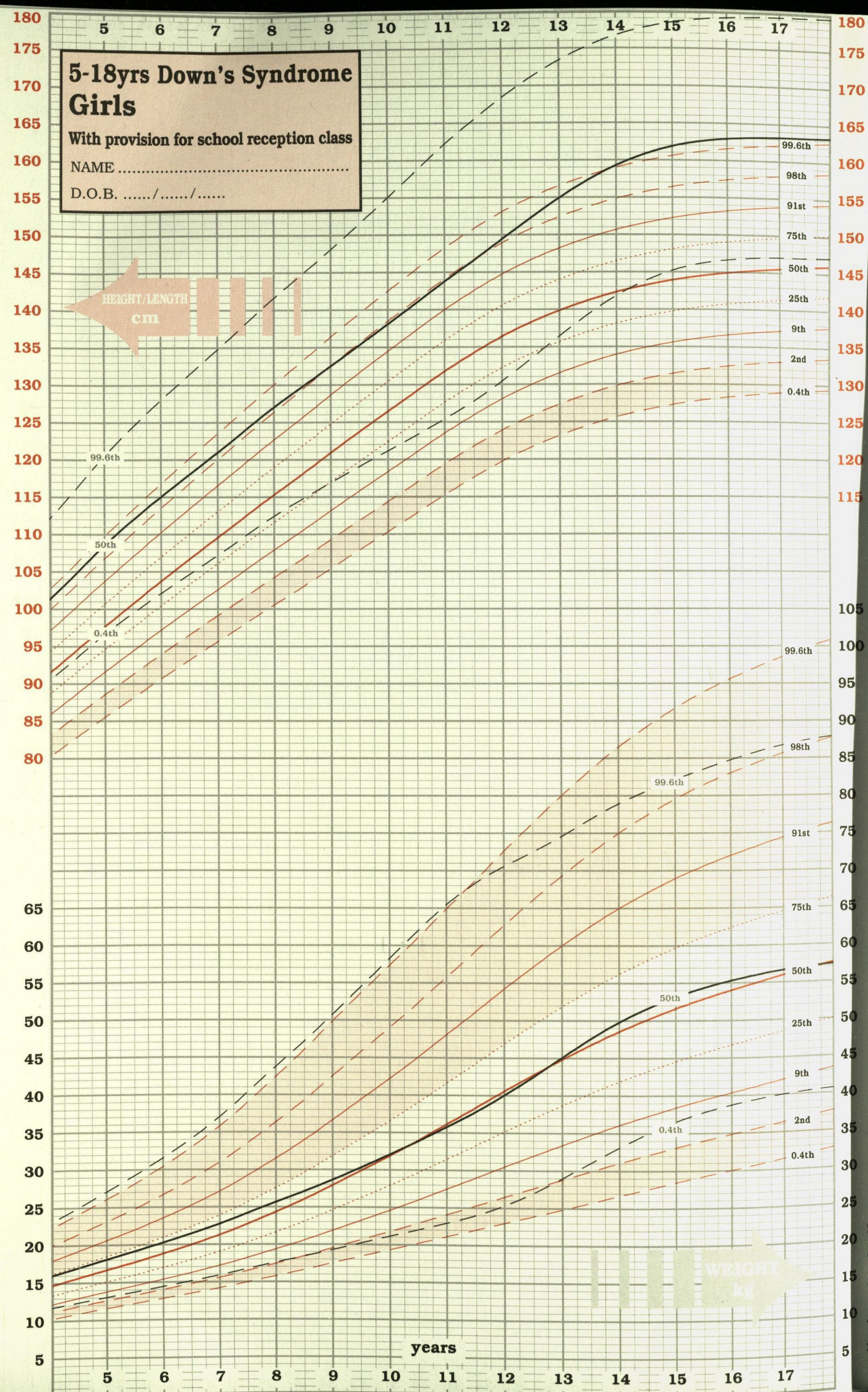
www.dsmig.org.uk



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APPENDIX 7



**The Irish College of
General Practitioners**

Coláiste Dhochtúirí
Teaghlaigh Éireann

25 September 2001

Prof. Hilary Hoey
The Adelaide & Meath
Hospital Dublin
Dept. of Paediatrics
Tallaght
Dublin 24

Dear Hilary,

Further to our phone conversation last week, I am writing to confirm that the DSMIG Guidelines have been discussed during two meetings of the College Executive during the Summer. I enclose some formal comments and observations for your interest from Dr. Richard Brennan and Dr. Paul Armstrong.

The overall response towards the Guidelines is positive, and there is clearly a need to implement the guidelines in general terms. It is probable that much of the work is not getting done in a systematic and uniform basis for these individuals and their families.

There are two critical aspects which emerged in our discussions however. The formal input from General Practice in the design of these Guidelines appears minimal, and indeed, in the list of those responsible for devising the Clinical guideline, there is no primary care physician participating. This is a serious omission.

Secondly, the role of the General Practitioner is not clearly defined, particularly in terms of care co-ordination. This is significant, particularly in cases such as these, where good care co-ordination is quite important.

The consensus among the Executive however is that despite these shortcomings, the Guidelines are important, and the ICGP position would be to formally support and promote the guidelines, subject to the design of a Combined Care Card or Record, preferably as a patient held record, clearly setting out an agreed schedule of health surveillance, with a defined GP role in care Co-ordination.

4-5 Lincoln Place, Dublin 2

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e-mail: info@icgp.ie

www.icgp.ie

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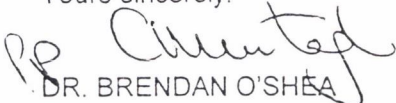
Registration Number 100456

F Ó Cuinneagáin
Chief Executive

If you are happy to proceed along these lines, I will undertake to provide a first draught, through the Communications and Publications Committee, within the next 6 weeks.

A final issue relates to the adequate resourcing of this standard of care. If a uniform standard of care is to be achieved, it will require to be resourced. Most individuals with Down's Syndrome have well above average medical needs, most have a GMS Entitlement, and there is a significant shortfall between GMS Payments and actual costs involved in providing adequate care in general Practice. Introducing a guideline such as this one has a significant cost implication for General Practitioners, and if a final draught of a Combined care card can be agreed during the next 3-4 months, I hope we could count on the support of your group in highlighting this.

Yours sincerely,


DR. BRENDAN O'SHEA
COMMUNICATIONS CHAIRMAN
ICGP

APPENDIX 8



The Royal College of Physicians of Ireland

Faculty of Paediatrics



27th November 2001

Prof. Hilary Hoey,
Consultant Paediatrician,
The National Children's Hospital,
Tallaght,
Dublin 24.

Re. Guidelines on Children's with Down's Syndrome.

Dear Hilary,

Your excellent document of the Guidelines for Management for Children with Down's Syndrome was discussed at the Faculty Board meeting on Friday 9th November 2001. I am delighted to inform you that the Board and the Faculty of Paediatrics have officially endorsed it.

Kindest regards.

Yours sincerely,

Siobhán
PP

Dr Siobhan Gormally,
Honorary Secretary

APPENDIX 9



The Royal College of Physicians of Ireland
Faculty of Public Health Medicine

6 Kildare Street, Dublin 2

September 9th 2002

Telephone: 01 - 661 6677
Fax: 01 - 676 3989

Professor Hilary Hoey,
Dept of Paediatrics,
The National Childrens Hospital,
AMNCH,
Tallaght,
D24

Dear Hilary,

Further to our meeting in relation to the Guidelines on the Medical Management of Children and Adolescents with Down Syndrome in Ireland, the Faculty Board met last Tuesday Sep 2nd 2002 and endorsed the report. The report will be an excellent help to service providers and families of children with Down Syndrome and the Faculty will do what it can to promote the guidelines. We will distribute it to our Directors of Public Health members who can distribute it further amongst Senior Area Medical Officers and Area Medical Officers but we would suggest that it should be sent by your Faculty to the Chief Executive Officers of the Health Boards as well to ensure wide spread dissemination.

As I mentioned at our meeting, it would be useful to laminate page 23 which summarises the schedule of health checks taken from the guidelines so that these can be displayed in doctors' surgeries, health centres and other locations. I imagine that 5,000 of those could be distributed throughout the country. I will also contact Dr Ailish Quinlan of Best Health for Children who might also be able to disseminate the guidelines.

Yours Sincerely,



Dr Joe Barry,
Dean,

Faculty of Public Health Medicine

APPENDIX 10

DOWN'S SYNDROME – CHILD DEVELOPMENT

This table gives an outline of the usual development of children with Down's syndrome. Just as with all children there is a great deal of individual variation in the age at which the different skills develop. In the table we show the usual range for some milestones. A few children will have additional health problems which may slow their development. However all will continue to develop at their own pace. You can use the table to find out if your child has particular difficulty in any area which may require extra help.

Area of development	Milestones	Age range		For you to record the age at which your child acquires these skills
		Down syndrome	Other children	
Gross motor skills (moving around)	Holds head steady in sitting position	3-9m	1-4m	
	Sits alone	6-16m	5-9m	
	Stands alone	12-38m	9-16m	
	Walks alone	13-48m	9-17m	
Fine motor skills and eye hand coordination	Follows object with eyes	1.5-8m	1-3m	
	Reaches out and grasps objects	4-11m	2-6m	
	Passes objects from hand to hand	6-12m	4-8m	
	Builds a tower of two 1" blocks	14-32m	10-19m	
	Copies a circle	36-60m	24-40m	
Communication skills	Babbles "Dada", "Mama"	7-18m	5-14m	
	Responds to familiar words	10-18m	5-14m	
	First words spoken with meaning	13-36m	10-23m	
	Shows needs by gesture	12-30m	11-19m	
	Two word phrases	18-60m	15-32m	
Personal and Social Skills	Smiles when talked to	1.5-4m	1-2m	
	Feeds self with biscuit	6-14m	4-10m	
	Drinks from cup	12-23m	9-17m	
	Dry by day	18-50m	14-36m	
	Bowel control	20-60m	16-48m	

Adapted with permission from Cunningham 1988 Down's Syndrome – An introduction for Parents. Souvenir Press Ltd. Human Horizon Series