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MANATŪ HAUORA

The Clinical Assessment and Management of Children, Young People and Adults with Down Syndrome

Recommended Clinical Practice

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Preface

A person with Down syndrome should be able to share the same experiences in life as any typically developing individual. The child, and later the adult, with Down syndrome benefits from a well informed, supportive and supported family working in partnership with professionals from health, developmental therapy, education and disability support services.

This document began from a suggestion to provide more consistency in the medical management of young children with Down syndrome. However, the needs of individuals with Down syndrome change as they grow and develop – at different times the focus will involve medical care, therapy, education, vocational and social support. Ongoing regular multidisciplinary review by professionals well known to the family allows assessment and screening, the provision of advice, and advocacy for health, education and disability resources. As a result, this Recommended Clinical Practice document has evolved to provide guidance for individuals throughout the lifespan.

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Introduction

This document aims to ensure that the health, developmental, educational and psychosocial needs of individuals with Down syndrome and their families are identified and addressed, and that individuals with Down syndrome receive care that is appropriate, timely and well co-ordinated between primary and secondary care. The recommended clinical practice (RCP) provided here should be used to supplement routine well child and adult care.

This RCP document has been developed mainly for medical health professionals to use with parents and other caregivers to ensure comprehensive and collaborative health care. Inevitably they address the 'typical' needs of the individual with Down Syndrome, and do not take into account the differences in personality and achievement that are as varied as they are for all individuals. In some cases a greater level of care will be necessary. For the individual with more complex needs, this RCP helps to ensure that standard medical screening is not overlooked, especially when responsibilities are shared between primary and secondary care. The recommendations for medical visits are therefore considered a minimum, and some individuals will have more frequent contact. The recommended GP visits have been planned to coincide with the current immunisation schedule (2001).

The impact of frequent visits to health providers and associated professionals should not be overlooked. They can be disruptive and time demanding. Also, the passing of milestone ages without attainment and a repetitive focus on what the child cannot do are often an extra source of grief to family members. Health professionals, including the GP and paediatrician, can exert an important influence by having a positive approach. This means looking at how well the child is doing within their own frame of reference, and phrasing statements positively.

It is important to remember that children, young people and adults with Down syndrome may have better understanding than verbal expression. This should be taken into account if discussing issues when the child is present. When children are present it is important, as with all children, that the child is not discussed in the 'third person' but is included in a way that is appropriate to their level of understanding. As with all children, information should be presented in a developmentally appropriate way that takes into account their cognitive level and level of maturity.

It is vital to emphasise the strengths of the child and family, not the problems and procedures. Assessment and treatment should address the holistic needs of the child and family. Children, young people and adults with Down syndrome may have similar medical problems, but they are all individuals who have unique personalities and characteristics. They also have a right to participate as fully as possible in society, and this includes both them and their families being active, informed participants in the care they receive from health professionals. When any medical procedures are undertaken, appropriate support and explanation for the child is important which should be appropriate to the child's development. As the child's capacity to comment increases, consideration should be given to them being an increasingly active and informed participant in the care they receive (see *Consent in Child and Youth: Information for Practitioners*, Ministry of Health, 1998, refer http://www.moh.govt.nz). Informed consent is an important issue to be aware of in this area for adults with Down syndrome also. The Health and Disability Codes should be used as the guide in this area.

The development of this document involved a review of the literature with the assistance of the New Zealand Health Technology Assessment (NZHTA) unit of the Christchurch School of Medicine. One member of the group assessed these reviews according to the quality of their search processes, the rigour of their analysis and the specific topics covered. The areas of interest covered included screening, medical assessments and treatments, anticipatory guidance regarding health checks, and the identification and appropriate referral for developmental, psychosocial, mental health, educational, vocational and support needs.

In general, while certain recommendations are based on observational studies, many represent a consensus of expert opinion and a review of the literature (AAPCG 1994; AAP 2001; Down Syndrome Medical Interest Group 1999; Pueschel et al 1995; Piachaud et al 1998; Trumble 1998; Cooley and Graham 1991). The 'Health Guidelines for Children with Down Syndrome' (AAPCG 1994) and the 'Down Syndrome Preventive Medical Check List' (Down Syndrome Medical Interest Group 1999) were especially useful.

The New Zealand Disability Strategy, released on 30 April 2001, aims to eliminate barriers that prevent or reduce the participation of people with disabilities in their communities and New Zealand society. These barriers range from the physical, such as access to facilities, to the attitudinal, due to poor awareness of disability issues. By poor awareness, the medical impact is not the issue but rather how people with disabilities are valued and supported in society. When delivering medical services, it is important to remain aware that the medical impact does not exclude the need to value people with disabilities, and to present an attitude that values the lives of people with disabilities and aims to continually enhance the full participation of disabled people in all aspects of life.

The philosophy of disability support services is to ensure that people with disabilities have control over and make real decisions about their own life. Services funded by DSS aim to promote a person's quality of life and create an environment that enables community participation and maximum independence. A 'needs assessment' involves the needs assessment and service co-ordination process, which is a disability support service. Attendant support can be provided by a range of support organisations.

In the anticipatory guidance section for different developmental periods there are a number of recommendations made in relation to psychosocial aspects of care. Health professionals involved with the individual need to determine whether they have the time, skills and resources available to address these or whether they are better addressed by other services. In doing so it is important to balance not having so many professionals involved that it is overly intrusive versus addressing the needs of the individual by providing comprehensive high-quality care.

Part I: A General Overview of Down Syndrome

1 Overview of Major Clinical Features

Neonatal features

The diagnosis of Down syndrome is generally made at birth on the basis of the physical examination. A confident clinical diagnosis can usually be made and patients informed on this basis with confirmation in due course from chromosome analysis.

Based on 48 neonates with Down syndrome, Hall (1966) set forth the following features.

Neonatal features of Down syndrome	(%)
Hypotonia	80
Poor Moro reflex	85
Hyperflexibility of joints	80
Excess skin on back of neck	80
Flat facial profile	90
Slanted palpebral fissures	80
Unusual shaped ears	60
Dysplasia of pelvis	70
Dysplasia of midphalanx of fifth finger	60
Single transverse palmar crease	45

Additional common physical features are a small brachycephalic head, epicanthic folds, flat nasal bridge, speckling of iris (Brushfield spots), small mouth, and a wide space (often with a deep fissure) between the first and second toes (Hall 1966).

Growth

Growth is relatively slow, with average height progressing along the third percentile of typical individuals and final height attained around 15 years of age (Cronk et al 1988). Excessive weight gain can be a problem (Luke et al 1994; Rubin et al 1998). Head size is usually in the microcephalic range (Palmer et al 1992). Growth charts are available (see Appendix 8).

Neurodevelopment and cognitive ability

While the neurodevelopmental progress and outcome varies significantly between individuals, common patterns exist (Cicchetti and Beeghley 1990). These are described in more depth below.

Motor development

Hypotonia, which is nearly universal at birth, tends to improve with age. Hypotonia and joint laxity significantly delay the development of gross motor skills and increase the risk for joint dislocations. Co-ordination is often poor. The impact of these on the functional development of the infant is major.

Cognitive development

The rate of developmental progress slows with age, and this is underscored by a decline in intelligence quotient (IQ) (Carr 1985). This does not necessarily apply to a decline in functional skills however. Individuals continue to learn new skills, so the effect of this on the adult's actual ability to cope with the world and have an independent or supported lifestyle varies (Wishart 1993). Children and adults with Down syndrome show a wide range of intellectual impairment, from mild to severe, with the majority falling in the mild to moderate range. In adults the corresponding mental ages range from one to nine years, with a mean of about 5.5 years (Carr 1988). While attributing a mental age to a child may be useful in an academic context, the individual's skills and experience of the world will influence their ability to function with success and personal satisfaction in the world.

Specific difficulties with speech and language occur in addition to their intellectual impairment (Chapman et al 1991; Chapman et al 1998; Mundy et al 1995). Auditory skills are weaker than in any population in the hearing community, and early use of visual supports is important. Academic skills such as reading/symbol recognition and concepts of numeracy are important skills for individuals with Down syndrome to acquire. Although the level of social and cognitive functioning is variable, children with Down syndrome often function better in social situations than might be expected from their cognitive ability.

Immunity

Recurrent low-grade infections (in particular chronic rhinitis, chronic serous otitis media, conjunctivitis and periodontal disease) are common. There are subtle non-specific immune deficiencies, with altered antibody levels that change with age and impaired cell-mediated immunity. This predisposes to not only infections, but also auto-immune diseases and malignancy (Nespoli et al 1993; Ugazio et al 1990).

Specific conditions

Over the individual's lifespan, there is an increased risk of congenital abnormalities and acquired conditions that may affect survival and/or quality of life (see Appendix 6 for a list of readings).

Important conditions are:

Condition	Frequency (%)
Hearing impairment	38–75
Serous otitis media	50–70
Eye disease	
Congenital cataracts	4
Acquired cataracts	30–60
Severe refractive errors	50
Congenital heart disease	44
Sleep-related upper airway obstruction	31
Atlanto-axial instability * (see Appendix 7)	15
Thyroid dysfunction	15
Gastrointestinal tract anomalies	12
Hip abnormalities	8
Seizures	5–10
Leukaemia	<1
Psychiatric disorder	22–38
Alzheimer's disease	Increasing after 35 years

2 General Information about Down Syndrome

Incidence

While the generally reported incidence of Down syndrome averages approximately 1:660 live births, the true birth incidence in New Zealand as reported by the New Zealand Birth Defects Monitoring Programme from 1995 to 1998 is just over 1:1000. This difference is due to both a natural and artificial rate of termination. The incidence is similar across ethnic groups (Hook et al 1983), but increases with increasing maternal age: 1:1500 (15–29 years), 1:800 (30–34 years), 1:270 (35–39 years), 1:100 (40–44 years), 1:50 (over 45 years) (Ferguson-Smith and Yates 1984). However, most children with Down syndrome are born to mothers who are less than 30 years, as there are a greater number of pregnancies in this age group.

Genetics and recurrence risk

The Down syndrome phenotype results from the presence of a triple amount of the chromosome region 21q22.2-q22.3 (Mattei et al 1981). This is usually the result of the existence of an extra chromosome 21 in all cells, which is why it is commonly called Trisomy 21. Two other less-common forms occur: namely Translocation Down syndrome and Mosaic Down syndrome (Richards et al 1965). A chromosome analysis should be

performed in all Down syndrome neonates to identify a translocation or mosaic genotype, as these can modify the recurrence risk in subsequent pregnancies and an individual's prognosis respectively (Gardner and Sutherland 1989).

Trisomy 21

Approximately 95% of Down syndrome is due to standard Trisomy 21. This occurs as the result of non-disjunction (failure of separation of chromosome homologues) during either meiosis I or meiosis II. The actual cause of non-disjunction and the association with increasing maternal age is currently unknown. As standard Trisomy 21 is almost never inherited through families, chromosome analysis is not necessary in either parent. For women below 30 years of age, the recurrence risk for live-born Down syndrome is 0.5%. However, for women over 30 years the recurrence risk appears the same as, or only minimally greater than, the age-specific occurrence figure.

Translocation Down syndrome

Between 1% and 5% of karyotypes reveal an unbalanced translocation, usually between chromosomes 14 and 21. A translocation arises when there is a two-way switch of chromosomal material between two non-homologous chromosomes. Therefore both parents of a child with Translocation Down syndrome should have chromosomal analysis to clarify the recurrence risk. Individuals with Translocation Down syndrome do not differ developmentally or medically from those with non-disjunction Trisomy 21. For the mother with a balanced translocation, the risk of having a liveborn child with translocation Down syndrome is approximately 12% and for the father with a balanced translocation the risk of this is 1.2%.

Mosaic Down syndrome

The incidence of Mosaic Down syndrome is traditionally said to be 1–2%, but probably many are undiagnosed. Within the Mosaic Down individual there are two cell lines: one Trisomy 21 and the other normal. The proportions of the cell lines vary between individuals. Mosaicism results from non-disjunction, or from an anaphase lag (transient delay in cell cycle), occurring after conception. Some individuals with Mosaic Down syndrome arise from initially trisomic-21 zygotes, and others from normal conceptuses. Genetic counselling should be the same as for the child with standard Trisomy Down syndrome, so parental chromosome analysis is not necessary. Individuals with Mosaic Down syndrome may score higher on IQ tests and may have fewer medical complications than other children with Down syndrome (Fishler and Koch 1991).

3 A Māori Perspective

Māori families more familiar with oral tradition may view the child born with Down syndrome as the result of a previous breach of tapu within the extended family. Failure to recognise the spiritual component in the meaning of the child's life to such a family could lead to tensions for the health professionals involved. It will also be difficult to develop satisfactory working relationships without involving all the key people in the extended family in suggestions and decisions about the special needs of child with Down syndrome.

Acting as if the child merely has a myriad of special physical problems that can be seen in isolation and require things to be done to the child at specific times will be seen as crass and offensive.

Even the concept of an *individual* development plan may be seen by some as a rather curious Eurocentric notion. The child cannot be seen as separate from the whānau (extended family). Each child represents hope for the future and carries its own particular whakapapa (hereditary pattern). Each newborn will always be someone's special mokopuna (grandchild). Where the child is identified as having special needs, then the awhitanga (special help) and arohatanga (special warmth) requirements will be recognised as even greater from the whole family.

At a practical level, Māori children continue to be over-represented in smaller or more isolated communities where ready access to the multiple special workers mentioned in these guidelines will be difficult. Health professionals will also be aware of the growing concept of health services for Māori being delivered by Māori. Such service possibilities are increasing in both urban and rural communities.

Children who have Down syndrome and are of Māori ethnicity deserve the best standards of care we can offer – as is the case with all children. It could be that a paediatrician sensitive to the special nuances of being Māori, and who can work alongside the whānau and with a key Māori health worker, will offer the best prospects for such a service. In this way Māori families will not be as likely to feel overwhelmed by multiple service providers, who may be sought only to provide care for specific aspects relating to the child.

Part II: Recommended Management of Infants, Children and Adults with Down Syndrome

4 Prenatal Counselling and Future Care Planning

Prenatal counselling

The counselling of a family in which a foetus has Down syndrome should cover the following topics (AAPCG 1994):

- Review and demonstrate the laboratory or imaging studies leading to the diagnosis, and explain the mechanism for occurrence of the condition in the foetus and the potential recurrence rate for the family.
- Review the prognosis and manifestations, reminding parents that some variability is expected.
- When applicable, recommend further investigations that may refine the prognosis (for example, foetal echocardiogram, ultrasound examination for gastrointestinal malformations).
- Review the available treatments and management options. This discussion needs to include the efficacy, potential complications and/or side effects, costs, or other consequences of these treatments.
- Explore the options available to the family for management and rearing of the child using a non-directive approach. In cases of early prenatal diagnosis, this may include discussing rearing the child at home, alternate within-family placement or adoption, continuation or termination of the pregnancy. A plan for delivery and neonatal care must be developed with the Lead Maternity Carer and the family. As the pregnancy progresses, further investigations may be useful in modifying this management plan (for example, detection of a complex heart defect on echocardiography). When appropriate, referral to a clinical geneticist should be considered for a more extended discussion of recurrence rates, future reproductive options, and evaluation of the risks of other family members.
- Discuss the personal, family, financial and medical support programmes available to the family.
- Offer prenatal diagnosis for any future pregnancies of the parents of an individual with Down syndrome, regardless of genotype. A chromosome result by either chorionic villous sampling at 10–12 weeks or amniocentesis at 16 weeks gestation provides a definitive diagnostic result. However, it is important for couples to be informed that maternal serum screening and ultrasound examination of foetal

morphology are not diagnostic tests, and therefore only modify the age-specific risk status (Hecher et al 1993).

- A multidisciplinary team (with a range of skills and experience) may be able to provide counselling if necessary. Alternatively, a referral for formal counselling may be indicated. Such practitioners should have experience in grief and crisis work.

Counselling at birth

Providing parents with information about their child's condition – whether a previously undetected Down syndrome, confirming the diagnosis for parents who have already received prenatal counselling, or imparting the finding of a serious complication – should be a medical priority. The informing doctor must be prepared emotionally, undistracted by other issues, well informed with current information, and willing to answer questions. It is important to be patient and understanding, direct and honest, and use plain language. Parents may take their lead from how a professional views and responds to their infant. Touching and handling the infant in an accepting and validating manner is important. Have written material to leave with the parents (refer Appendix 2, Resources for Parents and Caregivers).

Parental satisfaction with this process will be improved if they are told as soon as possible by someone with sufficient knowledge to inspire credibility. It is best to see both parents together, preferably with the baby present, and the baby referred to by name. The location should be a private, comfortable place, away from disturbances. The sensitivity and style with which such information is conveyed will be very important. Consider the impact of the information given. It is important that the medical practitioner conveys hope, possibilities and the uniqueness of the developmental pathway of each child.

Emphasise that they have a new baby who has basic needs that are the same for all newborns. Allow time for the new mother to talk about her experience of giving birth, and debrief if necessary. This may assist with the development of a positive relationship with a new infant. Staying in the present may assist with the establishment of bonding between parent and child. Parents may also need follow-up interviews after sufficient time has elapsed to process the emotional content of the information conveyed.

The information should take the form of a balanced point of view, instead of a listing of problems. A follow-up discussion could be planned and a telephone number to call at any time for information given. Additional information sources, including contact with other parents and early intervention services, should be provided and facilitated and the personal, family, financial and medical support programmes available to the family given. The family meeting should be followed by uninterrupted time for parents and child to be alone together.

It may be useful to include an allied health professional, for example a social worker, to assist with issues that may arise following the interview.

5 Newborns (Birth to 1 Month)

Clinical assessment

A paediatrician should always be involved.

Review parental concerns

Confirm the diagnosis of Down syndrome and review the genetics to establish recurrence risk. Discuss the specific findings with both parents whenever possible, and talk about the following potential clinical features associated with the syndrome. As it may not be possible to discuss everything in one interview, and new issues regularly arise for parents, this aspect of management should be undertaken at every visit, if necessary.

Feeding, nutrition and growth

Hypotonia of the oral musculature can interfere with breastfeeding. Feeding tends to take longer and there may be latching problems due to a protuberant tongue (Frazier and Friedman 1996; Spender et al 1996). Some babies will need supplemental feeding even at the time of discharge home; this is the exception rather than the rule. Consider referral to a speech–language therapist and/or lactation consultant experienced in infant oro-motor function. Support for the mother/child relationship is very important with such problems. Some babies experience difficulties maintaining temperature and will need extra swaddling during feeding. Constipation is more common due to hypotonic gut musculature. With skilled support, breast feeding can often be achieved. It is also an opportunity for bonding between mother and child. An environment free from distraction may assist.

Vomiting or absence of stools are significant early warning signs and should lead to investigation of the gastrointestinal tract. The following conditions may be present (at the indicated incidences):

- duodenal web or atresia (3.0%)
- pyloric stenosis (0.3%)
- annular pancreas
- Hirschprung disease (0.5%)
- imperforate anus (0.9%)
- tracheoesophageal fistula (0.4%)
- tracheal stenosis (1.2%)
- Meckel's diverticulum.

Cardiorespiratory

There is a 44% incidence of heart defects in infants with Down syndrome. Endocardial cushion defect, ventricular septal defect, patent ductus arteriosis, atrial septal defect, and aberrant subclavian artery occur in decreasing order of frequency (Freeman et al 1998). Pulmonary vascular hypertension occurs more rapidly than in children with the same defect who do not have Down syndrome, so a serious cardiac defect may be present in the absence of a cardiac murmur (Clapp et al 1990). Perform a high-quality echocardiogram in all Down syndrome neonates (Tubman et al 1991). The degree of urgency is influenced by a chest x-ray and electrocardiogram (ECG), which should be performed soon after birth (Rosenberg et al 1994). Refer all those with major lesions to a paediatric cardiologist. Medical and surgical management must always seek the best interests of the child. Treatments will usually be basically the same as for others but with special allowance for the needs of those with Down syndrome (Baciewicz et al 1989; Reller and Morris 1998). Reinforce the need for bacterial endocarditis prophylaxis in susceptible children with cardiac disease (see Appendix 3). Discuss increased susceptibility to respiratory tract infections.

Immuno-haematology

A neonatal leukaemoid reaction occurs in up to 4% and polycythaemia in up to 64% (Kivivuori et al 1996; Roizen and Amarose 1993). Check a full blood count (FBC). While most leukaemoid reactions resolve, some develop into acute megakaryoblastic leukaemia. Leukaemia occurs more frequently throughout childhood, but is still rare (less than 1%). Regular screening is not currently recommended.

Neurodevelopmental and orthopaedic assessment

Hypotonia is a constant feature, and underpins the importance of checking the hips for dislocation using the Ortolani-Barlow method.

Endocrine

To check for congenital hypothyroidism (0.7% risk) the neonatal heel prick screen (TSH assay only) is satisfactory. Check TSH and free thyroxine if jaundice is prolonged to two weeks.

Anticipatory guidance

Vision and hearing screening

Refer to an audiologist for initial assessment of hearing at two months of age by auditory brainstem response (ABR). This should detect any congenital unilateral or bilateral sensorineural hearing loss. While oto-acoustic emission (OAE) testing may be easier to perform, it only detects cochlear dysfunction, and so is not a diagnostic test. Check for a red reflex. If cataracts (4%), strabismus, or nystagmus are present at birth or manifest during infancy, refer immediately to an ophthalmologist. Otherwise an initial ophthalmology assessment should be performed by nine months. Glaucoma is also more common.

Support and education services

A discharge planning meeting with parents provides a forum to review care and discuss any issues that have arisen. Early contact with the GP for a well child check, if a GP is not already involved, allows an assessment of the child's baseline medical condition. Establish who the key workers will be for medical problems and therapy/management.

Referral to an early intervention service can ensure a smooth transition to community care by linking the family with multidisciplinary services at an early stage.

Discuss the range of future outlooks, particularly in relation to current community care philosophy, improved access to quality health care, individualised yet integrated developmental and educational programmes, and a growing awareness and acceptance by communities of people with disabilities. Employment in the competitive work force at productive jobs and residential options in more typical settings have brought people with Down syndrome a more healthy involvement in the normal networks of activity and support in their local communities (Carr 1994).

Review the risk of recurrence of Down syndrome and prenatal diagnosis in subsequent pregnancies.

Discuss unproven therapies; for example, dietary modifications, vitamins, piracetam and skull manipulation (Nickel 1996). Cosmetic facial and tongue surgery has not been shown to have any therapeutic benefit and should be discouraged (Jones 2000).

Access to disability services and social supports

Refer to the local child development team – or a multidisciplinary early intervention service. Some parents may wish to utilise additional early intervention services from birth, such as from specialist education services (SES) or NZCCS (see Appendix 2 for a list of support agencies). There are other options in some localities. In Christchurch, for example, the Champion Centre is a major provider of services for children with Down syndrome from birth to six years. Typically a child development service neurodevelopmental therapist will visit at home, working alongside parents to stimulate physical, cognitive, social and language skills. Early intervention services may be able to offer comprehensive multidisciplinary services to assist parents and caregivers with developmental and psychosocial aspects. A speech–language therapist can assist with early feeding difficulties, and later in infancy may teach language enhancement skills such as signing to encourage communication and develop language skills.

Make sure the family is supported to complete the Child Disability Allowance form. This is a non-means-tested allowance provided from birth until 16 years of age. Different allowances are available after this age. A social worker or the needs assessment process can assist the family with this and recommend other means-tested financial assistance as appropriate (for example, the Disability Allowance).

Refer for a needs assessment and service co-ordination

The purpose of a needs assessment is to identify the abilities, resources, goals and needs of the individual with a disability, and their caregiver, and to establish their priorities for these goals and needs. Working in partnership with the family, the assessment facilitator is required to consider, if relevant, all life areas (personal care, domestic and household, training and education/vocation, employment, mobility, recreation and personal development, re/habilitation, communication/speech, accommodation, income). The support needs of the caregiver are also addressed.

The assessment should include relevant specialist assessments (for example, paediatrician, physiotherapy, occupational therapy, speech and language therapy) and copies of these will be requested, or assessments referred for if not already completed. The final needs assessment document belongs to the family. The *Standards for Needs Assessments for People with Disabilities* (1994), available from the Ministry of Health, describes the standards that can be expected when receiving a needs assessment.

The purpose of service co-ordination is to develop a written service plan, arrange and connect to culturally appropriate support services to meet prioritised needs, and act as an ongoing contact and information provider for the disability supports required throughout life. The aim is to help the individual to achieve maximum independence and participate in society according to their abilities and goals.

The Ministry of Health funds many disability support services, and access to almost all of these is via Needs Assessment and Service Co-ordination (NASC) agencies. These services work on the basis of assessed need and equitable sharing of the available resources. For example, some of the possible supports only available via NASC may include the following but are not limited to these examples:

- in-home support
  - household support
  - Carer Support Subsidy
- personal care
  - specialist facility respite
  - special equipment/housing alterations.

Be cautious not to raise family expectations as many of these supports may be unavailable, or available only in a limited amount, until a baby with Down syndrome is older and their support needs higher. It is especially important to note that the previous ‘entitlement’ to 28 days’ Carer Support Subsidy no longer applies, and allocation of this subsidy for a newborn may not occur, or is usually limited to a lesser amount, dependent on relative and assessed need.

Reviews of disability support services are usually carried out annually, or more often at family request. If there is a significant change of needs, which often occurs around the times of transitions (for example, school entry), a new needs assessment is carried out.

It is very important that the key professional working with the family be integral in this process. This is to avoid unnecessary additional assessments and too many people being involved with the family.

Inform the family of the availability of support and advice from the parents of other children with Down syndrome. Such groups include the New Zealand Down Syndrome Association, Parent to Parent and the IHC. Supply current books, pamphlets, contact details and websites as listed in Appendix 2.

Discuss the strengths of the child and positive family experiences

Review the personal, family, financial and medical support programmes available to the family. Talk about how and what to tell other family members and friends. Review ways of living with and integrating the child with a disability into the family to the benefit of all. Consider referring parents, siblings and grandparents for counselling, as necessary. Grief may be a significant issue for family members.

6 Infancy (1 Month to 1 Year)

Clinical assessment

Regular GP visits; paediatrician follow-up at 2 months, 6 months and 12 months.

Review of the information available from the allied services involved, for example from early intervention or child development services, may assist this process.

Feeding, nutrition and growth

Consider a screen for iron deficiency (using ferritin) at 12 months. A referral to a dietitian for assessment of nutritional intake at this age is often useful. Down Syndrome Growth Charts should be used to plot growth (see Appendix 8 but as these charts have included individuals with, for example, congenital heart disease and obesity, they may not be truly representative of the Down syndrome population).

Since coeliac disease is relatively common in Down syndrome, with reported rates of 7–16.7% (Carlsson et al 1998; George et al 1996; Jansson and Johansson 1995; Csizmadia et al 2000), it is advisable to screen for this condition by measuring endomysial antibodies at 12 months. Most cases present between one and five years. If the screen is positive, a duodenal biopsy is needed. Obtain endomysial or antireticular antibodies and IGA levels depending on local availability at other times if there is a clinical suspicion of coeliac disease. If clinical suspicion remains, despite normal antibody levels, perform a small bowel biopsy.

For constipation, use aggressive dietary management and consider hypothyroidism and Hirschprung disease if there is no improvement with dietary changes and stool softeners.

Cardiorespiratory

Severe congenital heart malformations that cannot be definitively repaired remain a major cause of morbidity and mortality throughout childhood. Maintain close liaison with a paediatric cardiologist. Bacterial endocarditis prophylaxis will be needed in susceptible

infants with cardiac disease, including those with residual lesions after cardiac surgery (see Appendix 3 for details).

Upper airway obstruction is more common in individuals with Down syndrome. The midfacial hypoplasia typical of the condition predisposes to narrow airways, complicated by hypotonia, narrow nostrils, recurrent respiratory infections and enlarged tonsils and adenoids. Narrow openings to the paranasal sinuses predispose to frequent sinusitis and naso-pharyngitis. There is an increased risk of tracheomalacia and tracheal stenosis. There is also an increased susceptibility to respiratory tract infections, especially chronic rhinitis, chronic serous otitis media and sinusitis.

Immuno-haematology

Individuals with Down syndrome who have serious recurrent respiratory and systemic infections should be evaluated for immune deficiency. Immunoglobulin levels differ from the normal pattern (refer to page 4, Immunity). Specific antibody production is an important marker of immune competence. If the course of respiratory infections deteriorates, or has unusual features, consultation with a respiratory paediatrician or immunologist is recommended. Over six months of age, children with chronic severe cardiac and respiratory disease are appropriate candidates for the influenza vaccine.

Hearing and vision screening

Assessment during childhood reveals that 28% of individuals with Down syndrome have unilateral hearing loss and 38% have bilateral loss. Although hearing loss may be either conductive, mixed or sensorineural in type, it is usually a mild bilateral conductive loss. Ensure that the family understands the logarithmic nature of hearing loss.

At two months, perform an auditory brainstem response (ABR).

Behavioural audiology requires a developmental age of seven to eight months. Between nine and 12 months an audiologist should perform a hearing test by behavioural distraction testing. Repeated tympanometry may be helpful, although results are often unreliable due to excessive patient movement, a narrow ear canal, blockage with cerumen, and increased compliance of the ear canal and the tympanic membrane.

Because of recurrent otitis media and the 50% to 70% risk of chronic serous otitis media (CSOM), refer to an ear nose and throat (ENT) specialist by nine to 12 months for consideration of tympanostomy tubes (Pappas et al 1994). Fluid accumulation can occur as early as the neonatal period. It should be noted that the cure rate for CSOM may be lower and the sequelae higher (atelectatic eardrum, permanent perforation of the eardrum and middle ear cholesteatoma) than in children who do not have Down syndrome. Otorrhoea from the tympanostomy tubes and antibiotic-resistant bacteria can occur. It may not always be possible to improve hearing significantly after tympanostomy tube placement. Parents must remain vigilant to conductive loss (Iino 1999).
Review concerns about vision and strabismus at each visit. Refer for ophthalmologic assessment by nine months, even if this has already been performed at birth. Eye problems during childhood are congenital cataracts (4%), strabismus (27%), nystagmus (20%), and refractive error – both myopia (22%) and hyperopia (13%). Stenotic nasolacrimal ducts (6%) may lead to tearing. Blepharitis (9%) and conjunctivitis occur frequently.

Neurodevelopmental assessment

A general neurological, neuromotor and musculoskeletal examination, particularly addressing hip dislocation, is essential. Overall 5–10% of individuals with Down syndrome develop seizures, generally starting either under three years or over 13 years of age.

Review development using the appended 'Developmental Milestones' (Appendix 1). Encourage general developmental stimulation using fun/interesting activities as well as specific stimulation, as many infants with Down syndrome are perceived as passive and undemanding. Muscle tone tends to improve with age. The rate of developmental progress slows with age, but the learning continuum continues throughout the individual's life.

Endocrine

Thyroid screening tests (TSH and free thyroxine) should be performed at six and 12 months.

Anticipatory guidance

It is appropriate to advocate for a range of professional services to make sure the needs of the individual and family are being met. At the same time, a single health professional (usually the GP) will be gaining familiarity with what is normal for that child, which helps the early recognition of any health problems.

Review the early intervention services relative to the strengths and needs of the infant and family. Check the status of these services every six months. Prior to six to nine months, evaluation by a speech–language therapist is strongly recommended to maximise communication readiness and opportunities for developing understanding (Kumin et al 1994; Laws et al 1995). Individuals with Down syndrome frequently understand spoken language better than they can express themselves verbally, though they have weaker auditory understanding than other members of the population. Consequently, infants and children may be taught language using a multimodel approach, which may include signing and communication symbols, as well as spoken language. This approach permits children with Down syndrome to communicate and process more effectively at a time when their expressive language abilities and understanding are vulnerable to physical problems (for example, conductive hearing loss, dysarthia and weak auditory skills).

There are no primary contraindications to immunisations.

Review the risk of recurrence of Down syndrome and prenatal diagnosis in subsequent pregnancies.

Support and education services

Organisations such as the New Zealand Down Syndrome Association and the needs assessors/service co-ordinators, for example NASCENT, can provide useful information about support groups and resources.

Observe the emotional status of parents and family relationships. Educate and support siblings and discuss sibling adjustments. If necessary refer on, depending on the nature and

severity of problems experienced by siblings. At six to 12 months review the psychological support and family relationships, including long-term planning, financial planning, and guardianship.

Confirm that the family are receiving services from a needs assessment and service co-ordination agency on an ongoing basis and that the support being given is comprehensive.

7 Early Childhood (1 to 5 Years)

Clinical assessment

Regular GP visits; paediatrician follow-up at two, three and four years six months of age.

Review of information available from the allied services involved, for example early intervention and child development services, may assist this process.

Feeding, nutrition and growth

The growth spurts and plateau typical in all children tend to be more prolonged in children with Down syndrome. These growth patterns are not reflected in the smoothed curves of a standardised chart.

As there is a tendency towards obesity, total caloric intake should be below the recommended daily allowance (RDA) for children of similar height and age. Monitor for a well-balanced, moderate-fibre diet. Consider referral to a dietitian for assessment of nutritional intake. Regular exercise and recreational programmes should be established early. Television viewing should be restricted as much as practically possible.

Repeat endomysial antibody and IGA tests (refer page 14) at two years and four years six months of age, or at any other time if there is clinical suspicion of coeliac disease. Consider a screen for iron deficiency.

Cardiorespiratory

Severe congenital heart malformations that cannot be definitively repaired remain a major cause of morbidity and mortality throughout childhood. Maintain close liaison with a paediatric cardiologist.

Sleep-related upper airway obstruction occurs in 31% of individuals and typically starts by the second to third year of life. Predisposing factors include a narrow hypopharynx, large tonsils and adenoids, respiratory secretions, increased fat tissue in the pharynx in obese individuals, glossoptosis and hypotonia. Sleep-related upper airway obstruction may present with the following varied symptoms:

- snoring
- chest wall recessions

- sleep apnoea
- unusual sleeping positions (sitting up or bending forward at the waist with head on knees)
- nocturnal soaking sweats
- fatigability during the day
- poor growth and development
- reappearance of napping in older children
- behaviour change.

Chronic hypoxia due to airway obstruction may cause or exacerbate pulmonary hypertension, leading to cor pulmonale, in addition to causing daytime fatigue and neurobehavioural problems. Refer individuals with significant symptoms to an ENT specialist for consideration of tonsillectomy and adenoidectomy (Bower and Richmond 1995; Jacobs et al 1996). A lateral neck x-ray, ECG and concurrent sleep oximetry and video recording can be diagnostically helpful.

Tongue protuberance and drooling are additional potential problems. Consider referral to a saliva clinic where there is one. One option is to transpose the salivary ducts posteriorly.

Immunology

There is increased susceptibility to respiratory tract infections, especially chronic rhinitis, chronic serous otitis media (CSOM) and sinusitis. Consider an evaluation for immune deficiency. If the course of respiratory infections deteriorates, or has unusual features, consultation with a respiratory paediatrician or immunologist is recommended. Children with chronic severe cardiac and respiratory disease are candidates for use of influenza vaccine over six months and pneumococcal vaccine over two years.

Neurodevelopmental and musculoskeletal assessment

Once walking, orthopaedic disorders including hip dysplasia, patella instability, talipes planus and hallux varus may appear. Check routinely for scoliosis.

Routine cervical spine screening x-rays for atlanto-axial instability are controversial but should be considered before involvement in sports (refer to Appendix 7) (Taylor and Walter 1996; AAPCSMF 1995; Davidson 1988; Ferguson et al 1997; Harley and Collings 1994; Morton et al 1995; Pueschel et al 1990; Pueschel et al 1992).

Over time, it becomes evident that development is globally delayed. It is important to review the different developmental pathway of the child, but to not make too many predictions as to how far and how fast the child will progress.

Endocrine

Perform thyroid screening tests every two years over this period; 30–50% of children and adults with Down syndrome develop subclinical hypothyroidism, with borderline elevated TSH and normal thyroxine. This may reflect a neuroregulatory defect of TSH, which,

when studied by 24-hour sampling, varies between normal and very high levels. This may not need to be treated with thyroid hormone replacement. The presence of thyroid autoantibodies, however, correlates with an increased chance of ultimately developing overt hypothyroidism (7%) (Rubello et al 1995).

Anticipatory guidance

Hearing and vision screening

Due to the high risk of acquired (either persistent or fluctuating) hearing loss, and often inconclusive results, repeated hearing tests are necessary. If no CSOM develops, the next routine hearing test should be at 4.5 years. Regular tympanometry may be helpful, particularly if the child is at risk of CSOM (absence of tympanostomy tubes, 50% to 70% risk of CSOM between three and five years). There is also increased risk of cholesteatoma. Re-refer a child with CSOM to an ENT specialist. Re-refer to an ophthalmologist by 18 months to two years and again at 4.5 years (strabismus and approximately 50% risk of refractive errors).

Personal hygiene

Initial dental evaluation should be carried out at two years of age, with follow-ups every six months (Barnett et al 1986; Giannoni et al 1989; Pilcher 1998; Hennequin et al 1999). Promote twice-daily teeth brushing. Delays and alterations in the sequence of tooth eruption occur, along with malocclusions and missing teeth. Gingivitis can occur early and progresses rapidly, leading to loss of alveolar bone. Cellular immune deficits described in individuals with Down syndrome have the greatest clinical impact on gingivitis and periodontal disease (refer to page 4, Immunity).

Discuss skin care. Atopic dermatitis, cheilitis, ichthyosis, onychomycosis, seborrhoeic dermatitis, vitiligo, syringomas and alopecia areata occur more commonly. With age, generalised or localised dry skin occurs in up to 90% of Down syndrome children, sometimes associated with secondary itching, eczema, or infection. Routine measures to reduce moisture loss are needed on a daily basis, including less-frequent bathing, use of tepid water, use of a mild, moisturising soap, and the addition of unscented oil to the bath water. A hypoallergenic barrier cream should be applied daily, especially after bathing. Thickening and cracking of skin occurs, particularly of the hands, feet, wrists and elbows. The skin ages more rapidly and may be subject to the effects of ultraviolet light. Protective clothing and wide-brimmed hats, avoidance of over-exposure to sunlight, and a sunscreen of level 30 are all recommended.

Enrol with the dental clinic by two years of age but remember that bacterial endocarditis prophylaxis will be needed in susceptible children with cardiac disease, including those with residual lesions after cardiac surgery (refer Appendix 3).

Support and education services

Review the child's Specialist Education Services/NZCCS individual plan and preschool programme, and after four years of age discuss the options for future school placement. While both the child development service and the Specialist Education Services/NZCCS

may be involved from birth, expect an increasing provision of service from the Specialist Education Services/NZCCS when the child walks and starts to attend an early childhood education facility. A small playgroup to provide parent support and early social contact could be appropriate from age 12–18 months. At approximately two years, when the child starts walking, the neurodevelopmental therapist transfers therapy to a child development service physiotherapist and occupational therapist and the child is enrolled in a mainstream early childhood education facility (daycare or kindergarten). The key worker then becomes a Specialist Education Services/NZCCS early intervention teacher.

An individual plan is developed for each child by parents, educators, therapists and other professionals (as appropriate) to guide and set goals for enhancing developmental skills. The plan is typically updated every six months. A speech–language therapist will continue regular communication, language and speech therapy. A psychologist may work with the family and child or young person where such assistance is required. An early intervention teacher can remain involved with the child for up to six months into school (or to six years of age, maximum). While there is little evidence-based research to support the benefits of traditional (either centre- or home-based) neurodevelopmental therapy, it is generally considered beneficial by therapists and parents. There are few randomised controlled studies of early intervention treatment for children with Down syndrome. The evidence that does exist suggests that early intervention can ameliorate cognitive decline in the short to medium term (Guralnick 1998). Long-term effects, however, are less clear. A general goal of developmental therapy is to support the development of the child's functioning to compensate for the limiting effects of the impairments (Guralnick 1998; Harris 1980; Connolly et al 1993; Hines and Bennett 1996; Nilholm 1996; Rogers 1990).

By five years of age review whether the family is receiving comprehensive needs assessment and service co-ordination services.

Assess the need for child management techniques and how child management affects the child's behaviour, talk about sibling adjustment, functional skill development and valued roles, socialisation, and recreational skills. Monitor the family's need for respite care and supportive counselling. If behaviour or cognitive functioning changes, check the following:

- hearing
- thyroid function
- occurrence of sleep apnoea
- spinal cord compression
- depression
- abuse
- provision of family relief.

Referral to appropriate services may be required.

Discuss injury prevention, giving special consideration to developmental skills.

Remember to discuss future pregnancy planning, risk of recurrence of Down syndrome, and prenatal diagnosis.

8 Late Childhood (5 to 13 Years)

Clinical assessment

Yearly GP visits; paediatrician follow-up at 10 years.

Review parental concerns

Review parental concerns and successes, including ear problems/upper respiratory tract infections, sleep problems (snoring or restless sleep may indicate obstructive sleep apnoea), constipation, and current level of academic functioning and functional skills.

Feeding, nutrition and growth

Monitor for obesity. Total caloric intake should be below the RDA for children of similar height and age. Monitor for a well-balanced, moderate-fibre diet, and consider referral to a dietitian for assessment of nutritional intake. Maintaining regular exercise and recreational programmes is an important aspect of weight management.

Cardiorespiratory

Bacterial endocarditis prophylaxis will be needed in susceptible children with cardiac disease, including those with residual lesions after cardiac surgery (refer to Appendix 3).

Neurodevelopmental assessment

Perform a general and neurological examination (with particular reference to symptoms and signs of atlanto-axial instability). Review academic progress and the appropriateness of school placement, with an emphasis on prevocational skills. Children with Down syndrome typically start at a mainstream school with a range of supports already in place. By year seven (Form 1 of intermediate school) it may be more appropriate for some Down syndrome children to be enrolled at a special school, possibly while attending a satellite class at a mainstream school.

Continued monitoring by or therapy from a speech–language therapist would be expected. An individual with significant communication needs may be a candidate for augmentative communication options, which may range from a low technology communication book to a high technology (computer-based) communication device.

If behaviour or cognitive functioning changes, check:

- hearing
- thyroid function
- occurrence of sleep apnoea
- spinal cord compression
- depression

- abuse
- provision of family relief
- medication side-effects
- uncontrolled epilepsy.

There is a slightly increased prevalence of psychiatric disorder relative to the general population. However, psychiatric disorder may be less common than for individuals who do not have Down syndrome but who have a similar level of intellectual functioning (Cooper and Collacott 1994). Consider a mental health referral if symptoms of a possible psychiatric disorder appear to be developing.

Endocrine

Perform thyroid screening tests for hypothyroidism every two years until adolescence, and thereafter yearly. Approximately 10% of school-aged children with Down syndrome have uncompensated hypothyroidism (Noble et al 2000). Immune-mediated hyperthyroidism also occurs more frequently (Pueschel et al 1998). There is an increased risk of other autoimmune disorders such as insulin-dependent diabetes mellitus (1.4–10.6%) (Anwar et al 1998) and alopecia areata.

Anticipatory guidance

Hearing and vision screening

Review concerns about the child's hearing at each visit. Perform an audiologic evaluation at 10 years, and subsequently every two years. Continue to monitor for CSOM. Review concerns about vision and strabismus at each visit. Check the child's vision every two years. At 10 years refer to an optometrist to screen for refractive errors, keratoconus and cataracts.

Personal hygiene and social skills

Reinforce the importance of good self-care skills (grooming, dressing, and money handling skills). Promote twice-daily teeth brushing and dental follow-up every six months.

Review skin care. Adolescents and adults who are overweight seem to be susceptible to recurrent follicular skin infections of their buttocks and thighs with a prevalence of 50%–60%.

Discuss the development of age-appropriate social skills and the development of a sense of responsibility. Also discuss socialisation and family status and relationships, including financial arrangements and guardianship.

Discuss injury prevention, giving special consideration to developmental skills.

Support and education services

Review the need for an updated needs assessment by 10 years (refer to page 16).

9 Adolescence to Early Adulthood (13 to 21 Years)

Currently the New Zealand Youth Health Strategy is being developed by the Ministry of Health. It will include issues in relation to young people with disabilities.

Clinical assessment

Yearly GP visits; paediatrician follow-up at 13 years.

Review the past history of health problems, questioning specifically about the possibility of obstructive airway disease and sleep apnoea. Good general health is reported in 80% or more of young adults with Down syndrome (Carr 1994). Monitor independent functioning.

Nutrition and growth

Continue to monitor for obesity, and provide advice about diet and exercise.

Cardiorespiratory

Mitral valve prolapse with or without tricuspid valve prolapse and aortic regurgitation may occur after 18 years of age (Geggel et al 1993; Pueschel and Werner 1994). Valve regurgitation increases the risk of bacterial endocarditis. Refer to a cardiologist if this is clinically apparent. Remember the ongoing requirement for endocarditis prophylaxis with dental care and certain other invasive procedures (Appendix 3).

Continue to monitor for signs of sleep apnoea, especially if obesity is becoming a problem.

Neurodevelopmental assessment

A general physical and neurological examination should be performed, giving special consideration to the early diagnosis of atlanto-axial instability.

Continue speech–language therapy as needed. An individual with significant communication deficits may be a candidate for an augmentative communication device.

If behaviour or cognitive functioning changes, check:

- hearing
- thyroid function
- occurrence of sleep apnoea
- spinal cord compression

- depression
- abuse
- provision of family relief
- medication side-effects
- uncontrolled epilepsy.

Endocrine

Perform yearly thyroid screening tests from now on, as there is a 3–5% risk of autoimmune hypothyroidism.

A gynaecological examination should only be performed if the young woman is sexually active. Consider referral to a gynaecologist in this situation.

Anticipatory guidance

Vision and hearing

Review concerns about vision and strabismus at each visit. Vision should be tested by an optometrist every two years.

Review concerns about the adolescent's hearing at each visit. Perform an audiologic evaluation every two years. Presbyacusis, manifested by high-frequency hearing loss, may be evident by the second decade.

Personal hygiene and social skills

Reinforce the importance of good self-care skills (grooming and dressing). Promote twice-daily teeth brushing and dental follow-up every six months.

Review skin care. Adolescents and adults seem to be susceptible to recurrent follicular skin infections of their buttocks and thighs, with a prevalence of 50%–60%.

Discuss the development of age-appropriate social skills and the development of a sense of responsibility, for example money-handling skills. Also discuss socialisation and family status and relationships, including financial arrangements and guardianship.

Support and education services

Discuss issues related to the young person's transition into adulthood, including employment, independence and psychological health.

Discuss the appropriateness of school placement, with emphasis on adequate vocational training within the school curriculum.

The family with an adolescent with Down syndrome may experience the usual conflicts as the adolescent tries to establish their own identity, find some private space and pursue their

own interests. Teenagers with Down syndrome are subject to the same tempers, desires and emotions as anyone else, although they are often more frustrated in their experience. They are less able to resolve or manage the conceptual reasoning required at this age. Discuss how the parents can access support for their young person. Discuss psychosexual development, physical sexual development, menstrual hygiene and management, and fertility in both girls and boys (Goldstein 1988; Pueschel et al 1985). Written information and videos are available from the local Down Syndrome Association (see Appendix 2 for resources.) Emphasise socialisation skills. Discuss the need for and degree of supervision and/or the need for contraception. Discuss abuse prevention and talk about the recurrence risk to the patient and her family if she were to become pregnant.

Review an updated needs assessment by 13 years (refer page 16). Currently, adult needs assessment services for individuals as adults begin at 16 years and the disability support services allowances and subsidies change.

Sexual maturation and development in young people with Down syndrome

Primary gonadal deficiency is common, progressive from birth to adolescence, and definitely present in adults.

Females

The average age of menarche of 13 years six months does not differ from a control group of girls without Down syndrome (Arnell et al 1996). Menses are usually normal and regular, and gonadotropin levels are similar to control groups. Most girls with Down syndrome can manage their own menstruation, either independently or with varying degrees of assistance (Epps et al 1990). Hormonal and surgical treatments should be required no more frequently than in the rest of the population.

Preparation for menstruation should begin before menarche, with careful instruction and demonstrations using sanitary products and the girl's own body. Vaginal smears demonstrate definite ovulation in 38.5% of woman with Down syndrome, probable ovulation in 15.4%, and possible ovulation in another 15.4%. However, abnormal follicular development has been shown to be common among women with Down syndrome. Fertility is severely reduced in females. About half of their children have Down syndrome.

Males

In males primary and secondary sexual characteristics, genital size and hormonal levels are no different from a normative control group. Some men have difficulty attaining a full erection and ejaculation is not always possible. Although there have been several case reports in which a male has reproduced (a genetically normal child), males with Down syndrome are usually infertile (Zuhlke et al 1994).

Contraception

If contraception is required, the usual range of options is available to people with Down syndrome. Barrier methods, however, require a particularly careful process of education, and hormonal methods may need to be supervised. Reliable contraception may be

important, but no more so than education on protective behaviours and appropriate sexual expression.

Promoting healthy behaviour

Promote education on smoking, and the use of drugs and alcohol.

Transfer to adult care

Facilitate transfer to adult specialist medical care, if this is appropriate or desired. At what age the transfer occurs should be based on the best interests of the young person. A referral letter from the paediatrician to appropriate adult specialists would help outline ongoing medical needs. Combined clinics are often helpful during the transition stage to help build trust and confidence in the new clinician. There may be concern that at one of the most vulnerable times in the life of the young person, he or she loses a known long-term support person. This issue needs managing carefully and with the interests and needs of the young the primary consideration. Discuss group homes, workshop settings, and other community-supported employment. These discussions should also cover familial relationships, financial planning, and guardianship.

10 Adulthood

Clinical assessment

Yearly GP visits to systematically screen for treatable diseases: see Chicoine et al 1994; Van Allen et al 1999.

General

Most adults with Down syndrome enjoy good health, but their quality of life can be dramatically affected by chronic treatable conditions. Life expectancy has increased with improvements in health care, community living and antibiotics (Baird and Sadovnick 1987; Chicoine and McGuire 1997). Although life expectancy for any individual is difficult to assess, figures published in 1987 suggest that 44% of people with Down syndrome now survive beyond age 60 and 14% beyond age 68.

Adults with Down syndrome may experience high levels of pain with comparatively little complaint and may localise pain poorly. It is recommended that all reports of pain be thoroughly investigated. In some cases, a change in behaviour may indicate an underlying medical condition causing chronic discomfort or pain. Behavioural disturbance may also indicate anxiety, sexual abuse, reaction to change, or the side effects of medication. People with Down syndrome will often comprehend more than is evident by their speech. Generally, adults with Down syndrome enjoy being addressed directly in an age-appropriate manner. Active involvement in their own health care should be encouraged.

Informed consent

It is very important that practitioners are thoroughly familiar with the Health and Disability Code with regard to informed consent. Informed consent and the right to information are covered by this code. As with all adults the right of someone else to give informed consent on their behalf requires that the necessary legal processes have been undertaken (ie, that the person giving consent on another's behalf has the legal authority to do so). The only legal authorities are the enduring power of attorney or the welfare guardian. The code sets out the options of what to do when neither of these are available (Bray 1999).

Nutrition

Monitor for obesity. Evaluate for late-onset diabetes mellitus. Continue dietary and exercise recommendations.

Cardiorespiratory

Untreated congenital cardiac anomalies will need ongoing monitoring. Mitral valve prolapse with or without tricuspid valve prolapse and aortic regurgitation may occur after 18 years of age, and predispose to infective endocarditis and cerebral emboli. Refer to a cardiologist if this is clinically apparent. An echocardiogram may be indicated. Low blood pressure may lead to light-headedness and fainting, especially on rising in the morning. Bacterial endocarditis prophylaxis will be needed for individuals with cardiac disease, including those with residual lesions after cardiac surgery. Those with untreated atrioventricular canal defects and some ventricular septal defects progress to Eisenmenger syndrome and die in respiratory and cardiac failure any time from their late teens to late twenties.

Ask about sleep apnoea symptoms. Recurrent chest infections, complicated by aspiration, may lead to chronic interstitial lung disease.

Neuropsychiatric

Review the interval medical history and perform a general physical and neurological examination. Continue speech–language therapy as needed. An individual with significant communication needs may be a candidate for an augmentative (computer-based) communication device.

Relatively few studies have been carried out on the prevalence of psychiatric disorders in individuals with Down syndrome, and the majority of these have not been epidemiological studies, so it is difficult to draw conclusions from them. There are conflicting reports regarding the prevalence of psychiatric disorder and most studies have methodological flaws.

Epidemiological studies indicate that psychiatric disorder generally becomes more common with decreasing levels of intellectual functioning. For individuals with Down syndrome, however, this trend appears to be less marked. It would appear that psychiatric disorders occur more commonly in individuals with Down syndrome than in the general population, but less frequently than in individuals with similar degrees of intellectual functioning. Increasing age, degree of intellectual impairment and inability to communicate are associated with a greater risk of psychiatric disorder. In the younger age group, disruptive

behaviour disorders – for example oppositional and aggressive behaviours – are more common, while in the older age group depression occurs more frequently. It is important that depression not be misdiagnosed as dementia (Carr 1994; Gath and Gumley 1986; Meyers and Pueschel 1991).

Currently it is unclear whether there is an increased risk of co-existing autistic spectrum disorders in individuals with Down syndrome. Most of the research has been in the form of case studies, and the findings have been contradictory (Ghaziuddin et al 1992; Howlin et al 1995; Kent et al 1999; Walcabayashi 1979).

Where an individual has an autistic spectrum disorder or significant behavioural problems, considerable support, specialist advice and practical assistance such as regular respite for the person and their family will be required.

Monitor for loss of independence in living skills, behavioural changes and/or mental health problems. Symptoms of dementia include a decline in function, memory loss, ataxia, seizures and incontinence of urine and/or stool.

Behavioural and emotional problems may develop, and symptoms may include one or more of the following:

- decreased self-care
- loss of skills in activities of daily living
- loss of verbal skills
- loss of social skills
- loss of job skills
- withdrawal
- slow-down in activity level
- paranoid features
- increase in talking to self
- aggressive behaviour
- self-injury
- change in sleep patterns
- weight change
- persistent forgetfulness.

These behaviours frequently reflect either intrinsic changes (for example, depression, or hearing loss), changes in the person's life situation (residence, employment), or interpersonal conflict/abuse. Additional causes include hypothyroidism, folate/B12 deficiency, menopause, diabetes mellitus, visual impairment, cerebrovascular disease, adverse effects of medication, uncontrolled epilepsy, occult infection or carcinoma.

It is important to be aware that accelerated ageing may affect the functional abilities of adults with Down syndrome, more so than Alzheimer dementia (Devenny et al 1996). Alzheimer's disease, which invariably occurs to varying degrees, may have an onset at 35 years (Holland and Oliver 1995; Janicki et al 1996). Prevalence rates increase from 3.4% in the 30–39 age group, 10.3% in the 40–49 age group, to 40% in the 50–59 age groups respectively (Holland et al 1998). Late-onset epilepsy is often associated. All efforts should be made to maintain the person in their current accommodation and occupational settings (with appropriate modifications), before moving to more suitable

accommodation. Involvement of geriatric services and contact with specialised support services should occur.

Endocrine

Perform yearly thyroid screening tests (T4 and TSH) to identify hypothyroidism.

Musculoskeletal

Musculoskeletal disorders are common (hip joint instability, patellar instability, pes planus and metatarsus primus varus). Osteoarthritis of the spine occurs due to poor posture and low muscle tone throughout life. Osteoporosis, resulting in fractures of the long bones or vertebral bodies, is more common in all people with Down syndrome, and made worse after menopause. Bone mineral density studies are worthwhile in those at risk of further fractures.

Atlanto-axial instability may present with symptoms of incontinence, spasticity of lower limbs, stiff neck, headache or difficulty walking (refer to Appendix 7).

Anticipatory guidance and screening

Perform normal well adult screening.

Hearing and vision

Review concerns about hearing at each visit. Hearing loss is common, with early onset of presbycusis, and poor hearing often exacerbated by compacted wax and skin. Perform an audiologic evaluation every two years. Reinforce the impact of upper respiratory tract infection and intermittent conductive hearing loss.

Review concerns about loss of vision at each visit. Perform vision testing every two years, looking especially for adult cataracts, which have a prevalence of 30%–60%, and for keratoconus (6%).

Personal hygiene and social skills

Reinforce the importance of good self-care skills, for example, grooming and dressing. Promote twice-daily teeth brushing and dental follow-up every six months. Periodontal disease occurs as a result of lower immunity.

Review skin care. Adolescents and adults who are overweight seem to be susceptible to recurrent follicular skin infections of their buttocks and thighs with a prevalence of 50%–60%. Dry skin, eczema, skin infections, acne and alopecia areata occur commonly.

Discuss the development of age-appropriate social skills and the development of a sense of responsibility, for example, money-handling skills. Also discuss socialisation and family status and relationships, including financial arrangements and guardianship.

Support, education and vocational services

Discuss familial relationships, financial planning, and guardianship. If the person is not competent to give informed consent, a guardian will need to be appointed. Discuss group homes, workshop settings, and other community-supported employment. To ensure success in supported employment, the person needs to have a healthy sense of self-esteem nurtured from early childhood, an ability to complete tasks without assistance, a willingness to separate emotionally from parents and family members, and personal recreational activities. Encourage social and recreational programmes with friends.

Bereavement or grief counselling may be necessary for individuals who have experienced the loss of an important person in their life, either through death or by other circumstances (for example, a sibling moves away after marriage, or goes off to university).

Review an updated needs assessment (refer page 16).

Review financial assistance available through Income Support.

Self-help and independence

The degree to which self-help skills develop varies widely. Two-thirds of young people with Down syndrome are rated as independent in their feeding and toileting, one-third to two-thirds in dressing, about one half in washing and bathing, and about a quarter in hair washing. However, the majority require some degree of supervision with independent living skills, with some tasks completed independently and others requiring more assistance by caregivers. This will partially depend on prior opportunities for learning (Carr 1994).

Living situation

In New Zealand there has been a change in the number of people with Down syndrome living in the community, due to changes in thinking about appropriate care and management. Overseas studies have shown that the majority are either living at home or in some form of supported accommodation (Carr 1994). This appears to be influenced by the philosophy of care in different countries for people with Down syndrome. Traditionally, exclusion from community life has tended to be a societal response rather than a response to the needs and goals of the individual.

Leisure interests

The benefits of exercise that is meaningful or enjoyable for people with Down syndrome for personal health and social development should be encouraged. Sedentary activities – for example, high rates of television viewing (which is reported in some studies) – may lead to increased weight gain and poor health (Carr 1994).

Daytime employment and occupation

Studies suggest that the majority of adults attend adult centres in the daytime. This indicates that more could be done to increase the training, vocational and employment opportunities for young people with Down syndrome (Carr 1994).

Social activities and relationships

The majority of people have social contacts such as friends, and also go on outings. Most of this contact appears to be with other individuals with Down syndrome (Carr 1994).

Like other young people, those with Down syndrome develop an interest in the opposite sex early and wish to be in such relationships. Some studies have found that serious relationships are discouraged by parents and by staff at centres. In addition, less than a third of young people had sex education and their caregivers did not feel it was appropriate. Factors to consider with regard to sexuality include: a higher incidence of sexual abuse in people with intellectual impairment, the rights of individuals with disabilities, and the usual risks associated with unsafe or unprotected sex. While there are only two reported incidents of men with Down syndrome fathering children, many pregnancies have occurred in women with Down syndrome (Carr 1994).

It would appear that the area of sexuality and sex education is an important and underdeveloped area for people with Down syndrome. It will be important to work with caregivers to see how such issues can best be approached. Good communication between parents/caregivers and their children is likely to foster an environment where sexuality can be discussed in the context of interpersonal relationships. The issues of safe sex practices and contraception have been even more controversial, with some in the area of disability arguing that hysterectomies have been undertaken unnecessarily on those with intellectual impairment, and this raises concerns regarding informed consent.

Family factors

Marital discord has been found to be more common in parents of children with Down syndrome, but less common than in the families with other intellectually impaired individuals (Gath and Gumley 1986). This is likely to be contributed to by the additional stressors that occur for the couple. Behaviour disorders in Down syndrome children are associated with psychiatric disorder in the siblings. Psychiatric morbidity may be higher in mothers than in the general population, and similar for other mothers rearing children with an intellectual impairment (Gath and Gumley 1986). Similarly this may be contributed to by the additional stressors which such mothers experience.

Appendix 1: Developmental Milestones

	Children with Down syndrome		Children without Down syndrome	
	Average (mo)	**Range (mo)**	**Average (mo)**	**Range (mo)**
Milestone				
Smiling	2	1.5–4	1	0.5–3
Rolling over	8	4–22	5	2–10
Sitting alone	10	6–28	7	5–9
Crawling	12	7–21	8	6–11
Cruising	15	9–27	10	7–13
Standing	20	11–42	11	8–16
Walking	24	12–65	13	8–18
Talking words	16	9–31	10	6–14
Talking sentences	28	18–96	21	14–32
Self-help skills				
Eating:				
• Finger feeding	12	8–28	8	6–16
• Using spoon and fork	20	12–40	13	8–20
Toilet training:				
• Bladder	48	20–95	32	18–60
• Bowel	42	28–90	29	16–48
Dressing:				
• Undressing	40	29–72	32	22–42
• Putting clothes on	58	38–98	47	34–58

Adapted from Pueschel 1983: 359.

Appendix 2: Resources for Parents and Caregivers

New Zealand Down Syndrome Association

The New Zealand Down Syndrome Association (Inc) (NZDSA) can provide information, resources, support, local group contacts and a quarterly newsletter for parents and caregivers.

Contact the National office or your local group by phoning: 0800 NZDSAI (0800 693724), email: national.coordinator@nzdsa.org.nz

Booklets

Resources include NZDSA booklets entitled:

- *A Life with Down Syndrome*
- *Medical Care of the Child with Down Syndrome* (for parents)
- *Breastfeeding a Baby with Down Syndrome*
- *What is Down Syndrome?*
- *Heart Disease and Down Syndrome*
- *Transitions* (booklets on transitions into and out of school).

Websites

The NZDSA website carries up-to-date information for families in New Zealand and links to reputable international websites: www.nzdsa.org.nz

See also www.denison.edu/dsq/health99.shtml and www.ds-health.com/.

Recommended books

NZDSA recommends the following books for parents and children and young people:

Mummy, Why Have I Got Down's Syndrome
Caroline Philps, Lion Books
ISBN 0-7459-1921-9

Down Syndrome: The facts
Mark Selikowitz, 2nd ed., Oxford University Press, 1997
ISBN 0-19-262662-0

Down Syndrome: An introduction for Parents

C. Cunningham, 3rd ed., Souvenir Press, 1992
ISBN 0-285-64931-0

Babies with Down Syndrome: A new parents guide
K. Stray Gunderson (ed.), 2nd ed., Woodbine House, 1995
ISBN 0-933149-64-6

Count Us In: Growing up with Down syndrome
Jason Kingsley and Michael Levitz, New York, Harcourt Brace, 1994
ISBN 0-156-22660-X

Down Syndrome: Birth to adulthood: Giving families the edge
I Ryders and J Horribin, Paul Brookes Publishing Co, 1995
ISBN 0-89108-236-0

Differences in Common
Marilyn Trainer, Woodbine House, 1991
ISBN 0-920121-91-8

Uncommon Fathers
Donald Meyer (ed.), Woodbine House, 1995
ISBN 0-933149-68-9

Down Syndrome: Moving through life
Yvonne Burns and Pat Gunn (eds.), Chapman and Hall, 1993
ISBN 412-46180-3

Communication Skills in Children with Down Syndrome: Guide for parents
Libby Kumin, Woodbine House, 1995
ISBN 0-933149-53-0

Medical Care in Down Syndrome: A preventive medicine approach
Paul Rogers and Mary Coleman, Marcel Decker Inc., 1992

Down Syndrome: Advances in medical care
Ira Lott and Ernest McCoy (eds.), Wiley Liss, 1992
ISBN 0-471-56184-3

Medical and Surgical Care in Children with Down Syndrome: A guide for parents
DC Van Dyke, P Mattheis, et al, Woodbine House, 1995
ISBN 0-933149-54-9

Adolescents with Down Syndrome: Toward a More Fulfilling Life
Siegfried Pueschel and Maria Sustova (eds.), Paul Brookes Publishing Company, 1997
ISBN 1-55766-281-9

Gross Motor Skills in Children with Down Syndrome: A guide for parents and professionals
Patricia C Winders, Woodbine House, 1997
ISBN 0-933149-81-6

Improving the Communication of People with Down Syndrome

Jon F Miller, Mark Leddt and Lewis A Leavitt (eds.), Paul Brookes Publishing Company, 1999
ISBN 1-55766-350-5

Down Syndrome: A promising future, together
TJ Hassold and D Patterson (eds.), Wiley-Liss Publishers, 1999
ISBN 0-471-29687-2

Down Syndrome: A review of current knowledge
JA Rondal, Perera, L Nadel (eds.), Whurr Publishers Ltd, 1999
ISBN 1-86156-062-1

New Approaches to Down Syndrome
Brian Stratford and Pat Gunn (eds), Redwood Books, 1996
ISBN 304-33350-6

Meeting the Educational Needs of Children with Down's Syndrome: A handbook for teachers
Gillian Bird and Sue Buckley, University of Portsmouth, 1994
ISBN 1-85302-167-9

The Development of Language and Reading Skills in Children with Down's Syndrome
Sue Buckley, Maggie Ernslie, Gilly Haslegrave, et al, 2nd ed., University of Portsmouth, UK, 1993
ISBN 0-900234-17-2

Books for children

Our Brother Has Down Syndrome
Shelley Cairo, Annick Press Ltd, 1991
ISBN 0-920303-31-5

I Have Down's Syndrome
Brenda Pettenuzzo, Franklin Watts, 1987
ISBN 0-86313-5722

Other resources

IHC Library and Information Service
PO Box 4155, Wellington
ph: 04 472 2247
fax: 04 472 0429
The IHC library holds a specialised collection of books on intellectual disability.

NZCCS, National Office
86–90 Vivian Street
PO Box 6349, Marion Square, Wellington
ph: (04) 384 5677
fax: (04) 482 9353
email: info@no.ccs.org.nz

freephone: 0800 CCSCALL or 0800 227 2255
This is a national organisation providing a range of services for children and adults with physical and other disabilities, and their families, for example, early intervention services, advocacy to families, and vocational and recreational support.

IHC
15th floor, Willbank House, 57 Willis Street
PO Box 4155, Wellington, New Zealand
ph: (04) 472 2247
fax: (04) 472 0429
website: www.ihc.org.nz
Email the webmaster@ihc.org.nz for details on local services.
IHC is New Zealand's largest provider of services to people with intellectual disabilities and their families. It supports people with intellectual disabilities by advocating for their rights, providing a variety of housing and work options, and supporting families. It also provides a number of specialist services such as behaviour support and training.

Parent to Parent
National Office, PO Box 234, Waikato Mail Centre, Hamilton
Freephone: 0508 236 236 or (07) 834 1108
email: p2pnational@compuserve.com
website: http://www.parent2parent.org.nz
Parent to Parent is a support and information network for parents of children with special needs. Support is provided voluntarily by trained support parents who have a child with the same or similar needs. It has a resource library on different conditions.

Disability Issues Directorate
Ministry of Health, 133 Molesworth Street
PO Box 5013, Wellington
ph: (04) 496 2000
fax: (04) 496 2340
website: www.moh.govt.nz
This is the Directorate of the Ministry of Health which deals with disability issues and funds a range of disability support services such as home support and child development services, and is responsible for the development of the New Zealand Disability Strategy (refer www.nzds.govt.nz).

Enable New Zealand
60 Bennett Street
PO Box 4547, Palmerston North
ph: (06) 952 0011 / 0800 171 981
fax: (06) 952 0022
website: www.enable.co.nz
email: enable@enable.co.nz
Enable is a multi-service resource centre for people with disabilities, health professionals, and disability support organisations. It provides access to information, research and funding of equipment, housing alterations, and vehicle modification and purchase. Enable Information and Enable Research are available nationwide. Some services are only available locally and are funded by the Ministry of Health.

Disabled Persons Assembly (New Zealand) Incorporated
Level 4, Wellington Trade Centre, 173–175 Victoria Street

PO Box 27–524, Wellington
ph: (04) 801 9100
fax: (04) 801 9565
website: www.dpa.org.nz
email: gen@dpa.org.nz

The DPA is the umbrella organisation representing people with disabilities, the organisations involved in advocacy on their behalf, and service providers. The disabilities represented include physical, sensory, intellectual, psychiatric, neurological and age related. There are about 400 member organisations and contact details of these organisations can be accessed through the national office.

Appendix 3: Antibiotic Prophylaxis of Bacterial Endocarditis

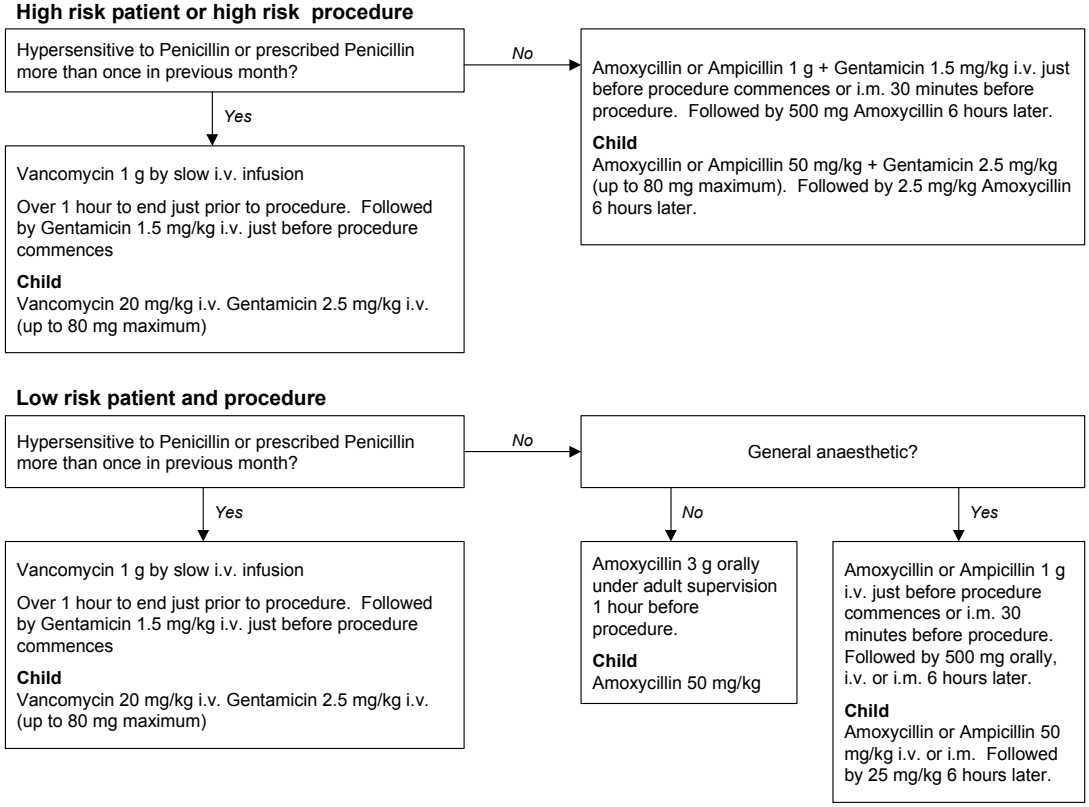
Who should be given prophylaxis?

Prophylaxis is recommended for all patients whose endocardium is damaged or rendered defective by acquired or congenital disease. Some patients are at high risk and some procedures are high risk.

High risk patients include those with a prosthetic valve or a past history of infective endocarditis undergoing dental procedures, oral surgery or upper respiratory tract surgery.

Low risk patients who are undergoing **high risk procedures** – gastrointestinal or genito-urinary procedures – should be treated as high risk.

Children's recommended doses are given in mg/kg. Give the adult dose if the calculated total dose for a child exceeds that recommended for adults.



Paediatric Medicine Referral Guidelines and Prioritisation Criteria, 4 July 2001, Elective Services, Health Funding Authority.

Appendix 4: Disability Support Services

The Ministry of Health funds the Disability Support Services available from the health and disability sector. These agencies provide a range of services and can include home support, personal support, attendant care, respite care and behavioural support.

The Ministry of Health directly funds carer support, eligibility for which is also determined by the needs assessment process. There are two main types of carer support: informal and formal. Informal carer support consists of relief care provided by a friend or family member to a client to enable the full-time carer to have a break. Formal carer support is provided by a professional carer, or within a formal care setting. The amount of time provided is based on the needs assessment. The *Standards for Needs Assessments for People with Disabilities June 1994*, available from the Ministry of Health, describes the standards that can be expected when receiving a needs assessment. Service co-ordination can also involve referring for specialist assessments and any services or support that is required.

Other sectors also have specific disability support services to varying degrees. Education and the Social Services are responsible for a significant range of specific disability support services. The Ministry of Health offers its Disability Support Services through the needs assessment service co-ordination (NASC) agencies. These services are provided after a process of an individual assessment of the needs of the disabled person and their family/ whānau. On the basis of this assessment, a range of services which meet the identified needs will be provided and co-ordinated. This process provides individualised targeted services within the context of prioritisation of resources available.

Appendix 5: Special Education

1 Support for students with high and very high special education needs

Ongoing and Reviewable Resourcing Schemes (ORRS)

Funding for extra teaching, specialist programmes, therapy, consumables and education support. The funds are provided to the 'fundholder' (the school or Specialist Education Services[1]) and then allocated to support each student accepted for the schemes, depending on the level of need.

Speech–language initiative

Therapy usually provided at school for students with high communication needs. Training courses are also run for teachers so they can identify communication difficulties and arrange programmes to meet students' needs.

Severe behaviour initiative

Advice and specialist support for students with severe behaviour difficulties, their schools, families, the community and government agencies. For some students, more intensive support may be provided either on or off the school site.

High health needs

Special education support through Regional Health Schools, for example if students are unable to attend their local school because of health difficulties.

School high health needs fund

Paraprofessional support for students with care and safety issues arising from health needs.

[1] Specialist Education Services (SES) will become a directorate within the Ministry of Education in 2002.

2 Support for students with moderate special education needs

Special Education Grant (SEG)

Funding for all schools as part of their operational funding to be used for special education programmes.

Resource Teachers: Learning and Behaviour (RTLB)

Specially trained teachers who support and work within school settings to meet the needs of students with moderate learning and/or behaviour difficulties.

Resource Teachers: Literacy (RT: Lit)

Specially trained teachers who support and work in schools, assisting staff to meet the needs of students with reading and writing difficulties.

Specialised services

Occupational therapy and/or physiotherapy for students with moderate to high physical impairments, Resource Teachers: Vision Impaired for students with moderate vision impairment, Resource Teachers: Deaf and Hearing Impaired for students with moderate hearing impairment.

Learning support fund

Funding for clusters of schools, used in conjunction with the Resource Teachers: Learning and Behaviour, Resource Teachers: Literacy, Resource Teachers: Hearing Impaired and Deaf.

Other support

Other support available may include:

- transport assistance – a subsidy or allowance for taxi or bus for travel between home and school
- property modifications – capital works such as alterations or additions to school property to enable children and young people with special education needs to enter and carry out regular activities within state schools
- assistive equipment – a wide range of tools for students to help them access the learning curriculum.

Information about each of these initiatives is available from schools, Ministry of Education local offices, or from the Ministry web site www.minedu.govt.nz

Appendix 6: Readings

Buchanan L. 1990. Early onset of presbyacusis in Down syndrome. *Scandinavian Audiology* 19: 103–10.

Cooley WC, Graham JM. 1991. Down syndrome: an update and review for the primary care physician. *Clinical Pediatrics* 30(4): 233–53.

Courage ML, Adams RJ, Reyno S, et al. 1994. Visual acuity in infants and children with Down syndrome. *Developmental Medicine and Child Neurology* 36(7): 586–93.

Cuskelly M, Dadds M. 1992. Behavioural problems in children with Down's syndrome and their siblings. *Journal of Child Psychology and Psychiatry* 33(4): 749–61.

Cutler AT, Benezra-Obeiter R, Brink SJ. 1986. Thyroid function in young children with Down syndrome. *American Journal of Disease in Childhood* 140: 479–83.

Da Cunha RP, Moreira JB. 1996. Ocular findings in Down's syndrome. *American Journal of Ophthalmology* 122(2): 236–44.

Fort P, Lifshitz F, Bellisario R, et al. 1984. Abnormalities of thyroid functions in infants with Down syndrome. *Journal of Pediatrics* 104(4): 545–9.

Freeman SB, Taft LF, Dooley KJ, et al. 1998. Population-based study of congenital heart defects in Down syndrome. *American Journal of Medical Genetics* 80(3): 213–17.

Greene WB. 1998. Closed treatment of hip dislocation in Down syndrome. *Journal of Pediatric Orthopedics* 18(5): 643–7.

Hayes A, Batshaw ML. 1993. Down syndrome. *Pediatric Clinics of North America* 40(3): 523–35.

Holland AJ, Hon J, Huppert FA, et al. 1998. Population-based study of the prevalence and presentation of dementia in adults with Down's syndrome. *British Journal of Psychiatry* 172: 493–8.

Karlsson B, Gustafsson J, Hedov G, et al. 1998. Thyroid dysfunction in Down's syndrome: relation to age and thyroid autoimmunity. *Archives of Diseases in Childhood* 79(3): 242–5.

Kile JE. 1996. Audiologic assessment of children with Down syndrome. *American Journal of Audiology* 5: 44–52.

Knox GE, Bensel RW. 1972. Gastrointestinal malformations in Down's syndrome. *Minnesota Medicine* 55(6): 542–4.

Levanon A, Tarasiuk A, Tal A. 1999. Sleep characteristics in children with Down syndrome. *Journal of Pediatrics* 134(6): 755–60.

Litz CE, Davies S, Brunning RD, et al. 1995. Acute leukemia and the transient myeloproliferative disorder associated with Down syndrome: morphologic, immunophenotypic and cytogenetic manifestations. *Leukemia* 9(9): 1432–9.

Marcus CL, Keens TG, Bautista DB, et al. 1991. Obstructive sleep apnea in children with Down syndrome. *Pediatrics* 88(1): 132–9.

Martin GR, Rosenbaum KN, Sardegna KM. 1989. Prevalence of heart disease in trisomy 21: an unbiased population (abstract). *Pediatric Research* 25: 77A.

Mitchell C, Blachford J, Carlyle MJ, et al. 1994. Hypothyroidism in patients with Down syndrome. *Archives of Pediatrics & Adolescent Medicine* 148(4): 441–2.

Myers BA, Pueschel SM. 1991. Psychiatric disorders in persons with Down syndrome. *Journal of Nervous and Mental Disease* 179(10): 609–13.

Noble SE, Leyland K, Findlayson CA, et al. 2000. School based screening for hypothyroidism in Down's syndrome by dried blood spot TSH measurement. *Archives of Disease in Childhood* 82: 27–31.

Pueschel SM. 1990. Clinical aspects of Down syndrome from infancy to adulthood. *American Journal of Medical Genetics* (Suppl 7): 52–6.

Pueschel SM, Louis S, McKnight P. 1991. Seizure disorders in Down syndrome. *Archives of Neurology* 48(3): 318–20.

Pueschel SM, Scola FH. 1987. Atlanto-axial instability in Down syndrome: epidemiologic, radiographic and clinical studies. *Pediatrics* 80(4): 555–60.

Pueschel SM, Sustrova, Kostalova L. 1998. Hyperthyroidism in children with Down syndrome. *Down Syndrome Quarterly* 3(2): 1–4.

Roizen NJ, Mets MB, Blondis TA. 1994. Ophthalmic disorders in children with Down syndrome. *Developmental Medicine and Child Neurology* 36(7): 594–600.

Roizen NJ, Wolters C, Nicol T, et al. 1993. Hearing loss in children with Down syndrome. *Journal of Pediatrics* 123 (1): S9–12.

Stafstrom CE. 1993. Epilepsy in Down syndrome: clinical aspects and possible mechanisms. *American Journal of Mental Retardation* 98 (Suppl): 12–26.

Stafstrom CE, Patxot OF, Gilmore HE, et al. 1991. Seizures in children with Down syndrome: etiology, characteristics, and outcome. *Developmental Medicine and Child Neurology* 33: 191–200.

Stebbens VA, Dennis J, Samuels MP, et al. 1991. Sleep related upper airway obstruction in a cohort with Down's syndrome. *Archives of Disease in Childhood* 66(11): 1333–8.

Watson MS, Carroll A, Shuster JJ, et al. 1993. Trisomy 21 in childhood acute lymphoblastic leukemia: a Pediatric Oncology Group study (8602). *Blood* 82(10): 3098–102.

Zipursky A, Brown E, Christensen H, et al. 1997. Leukemia and/or myeloproliferative syndrome in neonates with Down syndrome. *Seminars in Perinatology* 21(1): 97–101.

Appendix 7: Clinical Aspects of Atlanto-axial Subluxation

Approximately 15% of Down syndrome individuals under 21 years old have atlanto-axial instability (AAI), radiologically defined by an atlanto-odontoid interval of greater than 4.5mm (Pueschel and Scola 1987). Almost all are asymptomatic. Symptomatic AAI results from subluxation that is severe enough to injure the spinal cord, or from dislocation at the atlanto-axial joint. Instability can also involve the atlanto-occipital interface. Bony anomalies of the cervical spine occur with increased frequency and may contribute to the risk of subluxation of an unstable joint.

The natural history of AAI in Down syndrome and its relationship to spinal cord injury are still incompletely understood. No study to date has shown that radiographic findings can predict which children will develop neurologic problems. A recent review has stressed the importance of measurement of the neural canal width on lateral cervical spine x-ray (Brockmeyer 1999). Therefore obtaining x-rays is optional.

Neurologic manifestations of spinal cord compression include:

- neck pain
- head tilt or limited neck mobility
- increased clumsiness
- limping or refusing to walk
- weakness; tingling of an arm
- hyperreflexia or clonus
- upgoing plantars
- changes in bowel or bladder function
- unexplained behaviour change
- easy fatigability.

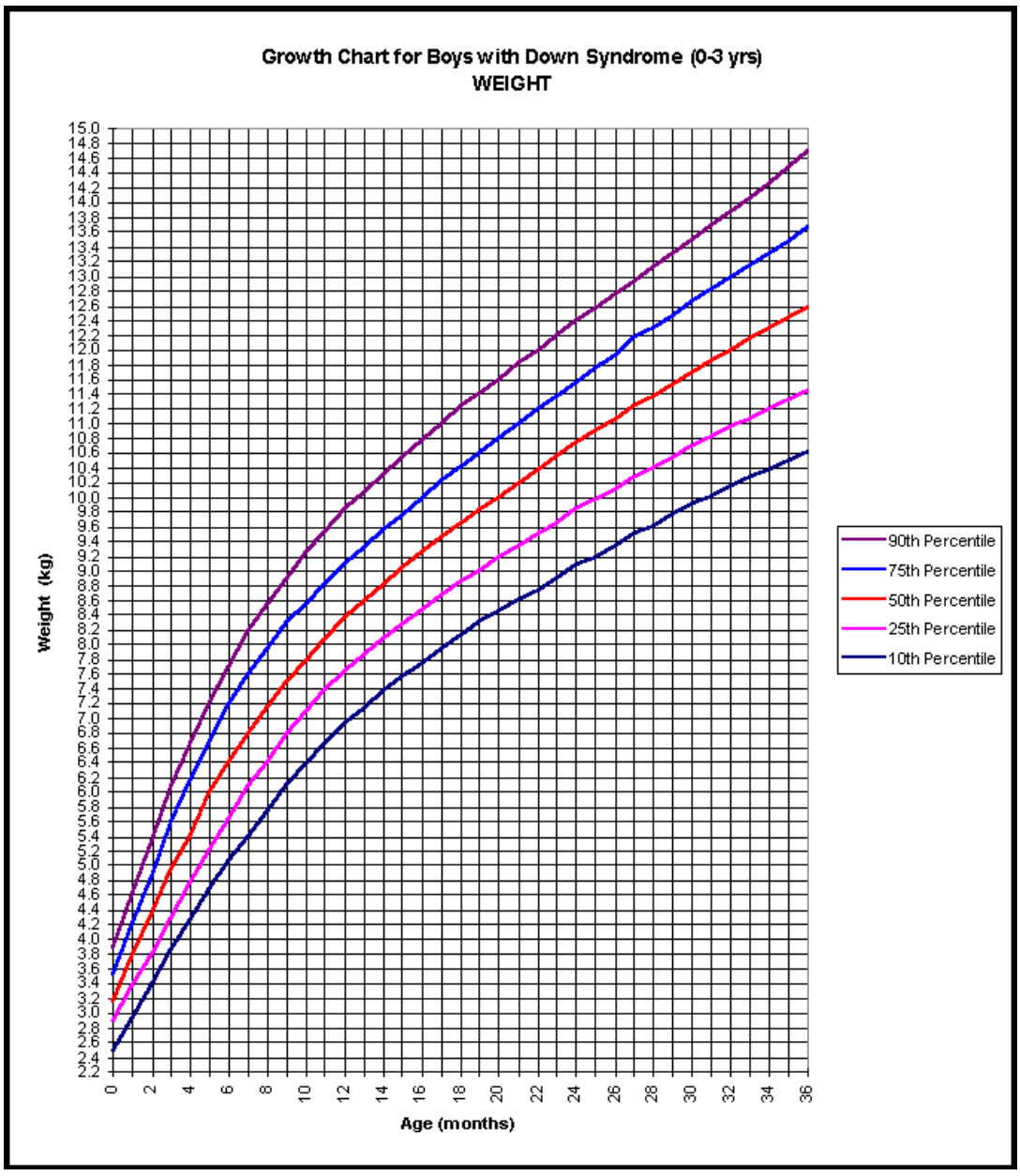
Such symptoms and signs often remain relatively stable for months or years. Occasionally they progress, rarely even to paraplegia, hemiplegia, quadriplegia or death. Trauma rarely causes the initial appearance or the progression of these symptoms. Nearly all the individuals who have experienced catastrophic injury to the spinal cord have manifested weeks to years of preceding, less severe neurologic abnormalities. The majority of patients develop symptoms before 10 years of age, when ligament laxity is most severe. Therefore, probably of greater importance than screening cervical spine x-rays is the need for the individual's caregivers to be aware of the manifestations of symptomatic AAI. They should be instructed to contact their doctor immediately if any symptoms appear. Symptoms of spinal cord compression warrant an urgent neurological review and consideration of magnetic resonance imaging of the cervical spine.

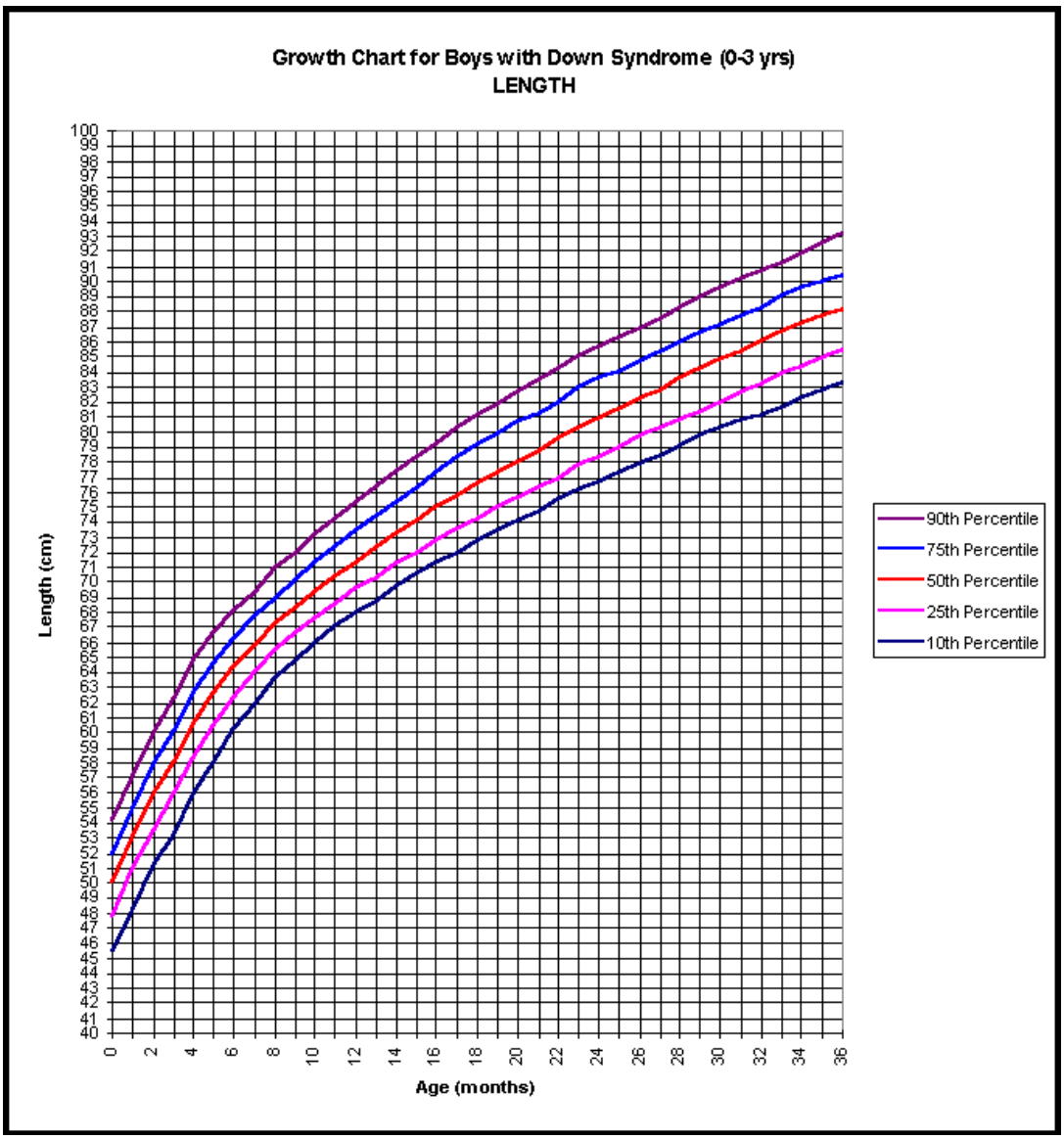
If x-rays are taken, they should be performed at an institution accustomed to taking and reading these x-rays. A series of x-rays in neutral, flexion and extension lateral is necessary. Do not force into passive flexion; encourage the patient to flex their neck actively. Particularly in younger children in whom radiographic interpretation is more difficult, normal x-rays do not exclude atlanto-axial instability. Therefore, with normal x-rays anticipatory guidance needs to be balanced: acknowledging both the sometimes

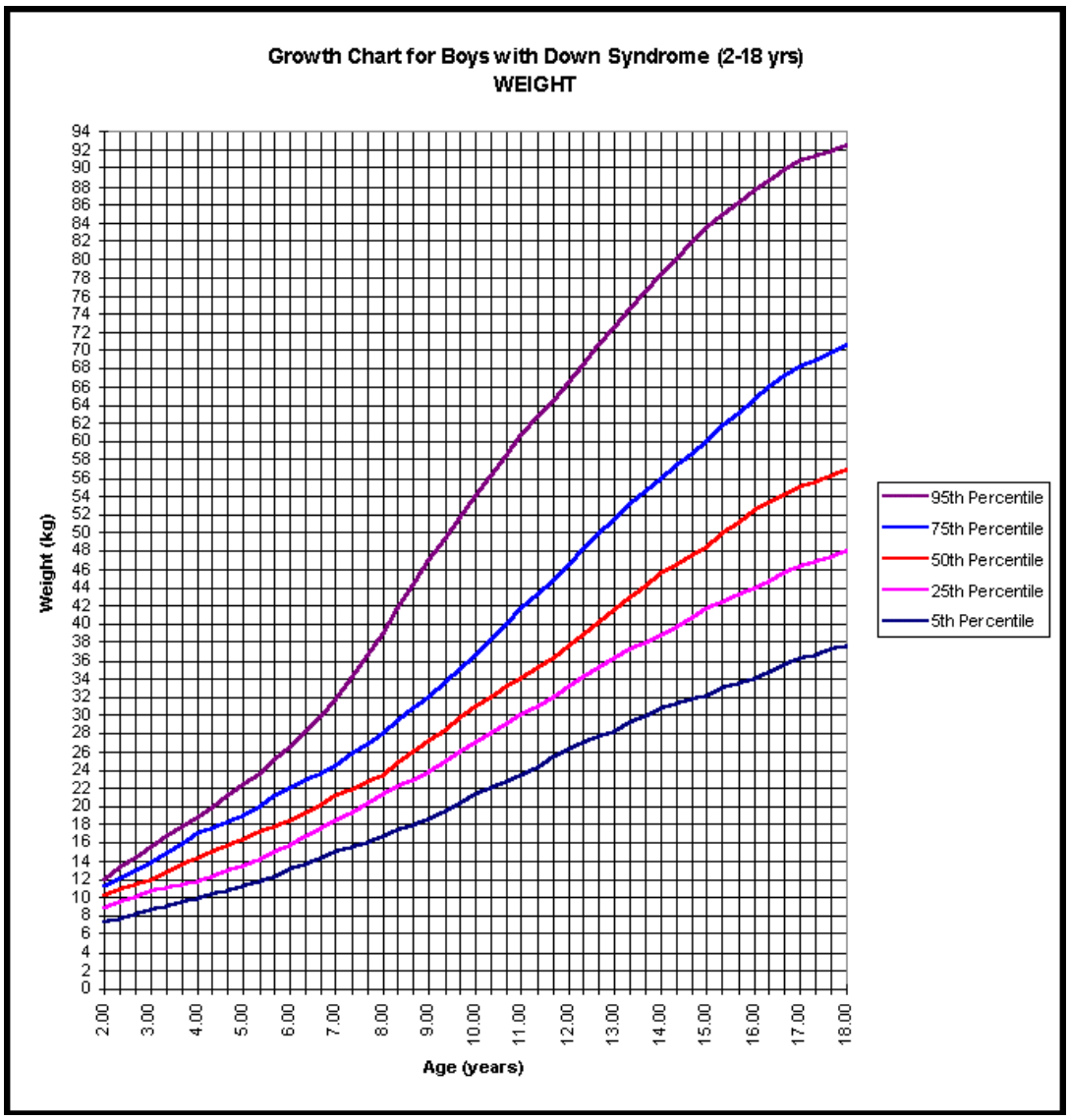
boisterous nature of this age group and the rare occurrence, but serious nature of neurologic impairment.

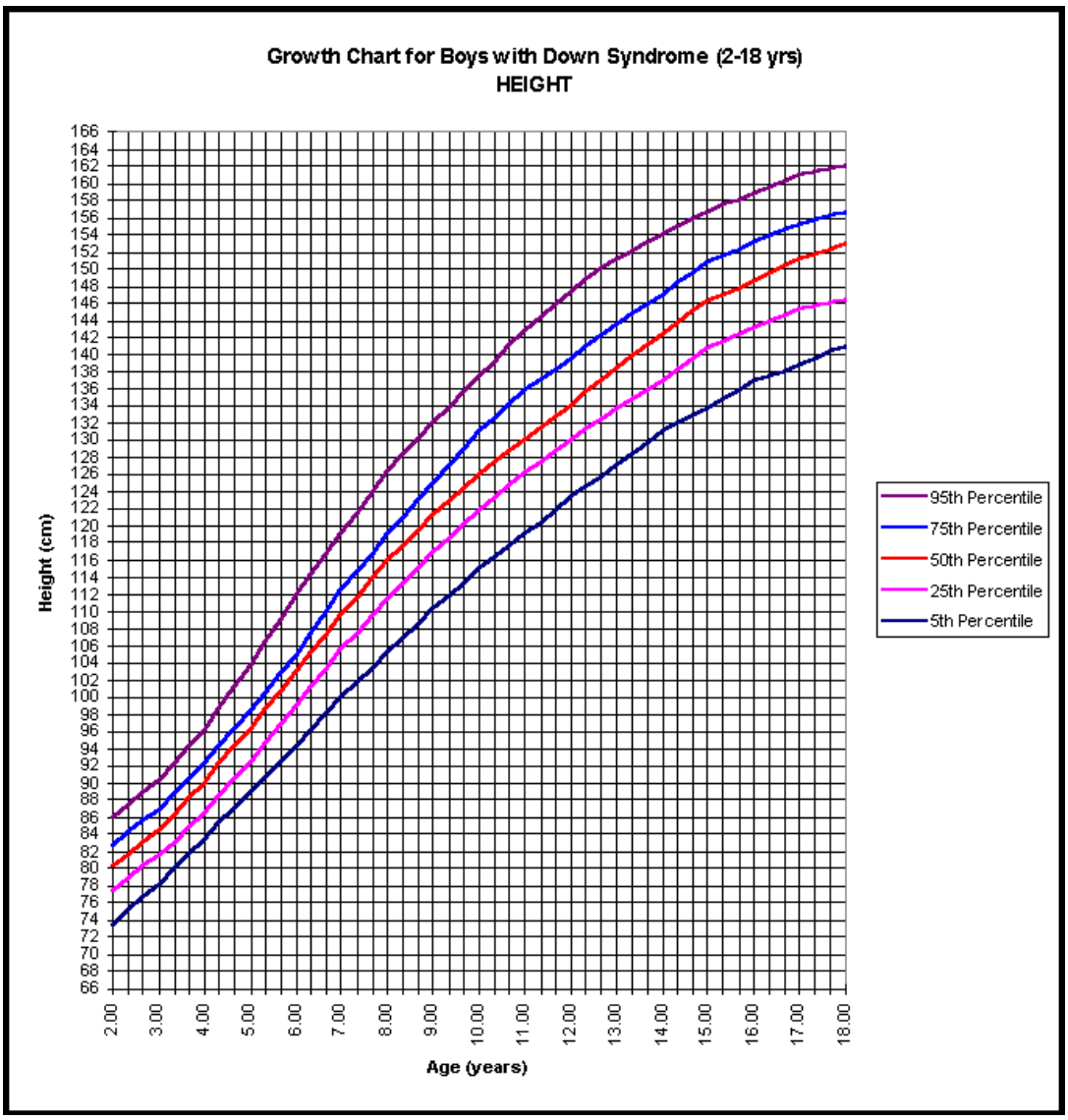
If the x-rays show either an abnormal odontoid, an increased atlanto-odontoid interval on flexion/extension lateral radiographs of more than 4.5mm, or AAI, then the patient should refrain from high-risk sports such as gymnastics, diving, butterfly stroke in swimming, high jump in track and field, soccer, trampolining, and any exercises that place excessive pressure on the head and neck muscles. These patients should also be referred to an orthopaedic surgeon. Care should be taken with procedures that involve hyperextension of the neck, such as instillation of eye drops. Yearly neurological surveillance is essential. Furthermore, anaesthetists and ambulance officers need to be aware of the potential cervical spine instability in asymptomatic individuals and act accordingly. Most anaesthetists do not routinely perform preoperative cervical spine x-rays (Kobel et al 1982; Litman et al 1995; Williams et al 1997).

Appendix 8: Growth Charts




Growth Chart for Boys with Down Syndrome (0-3 yrs)
LENGTH
Length (cm)
Age (months)
90th Percentile
75th Percentile
50th Percentile
25th Percentile
10th Percentile


Growth Chart for Boys with Down Syndrome (2-18 yrs)
WEIGHT
Weight (kg)
Age (years)
95th Percentile
75th Percentile
50th Percentile
25th Percentile
5th Percentile


Growth Chart for Boys with Down Syndrome (2-18 yrs)
HEIGHT
Height (cm)
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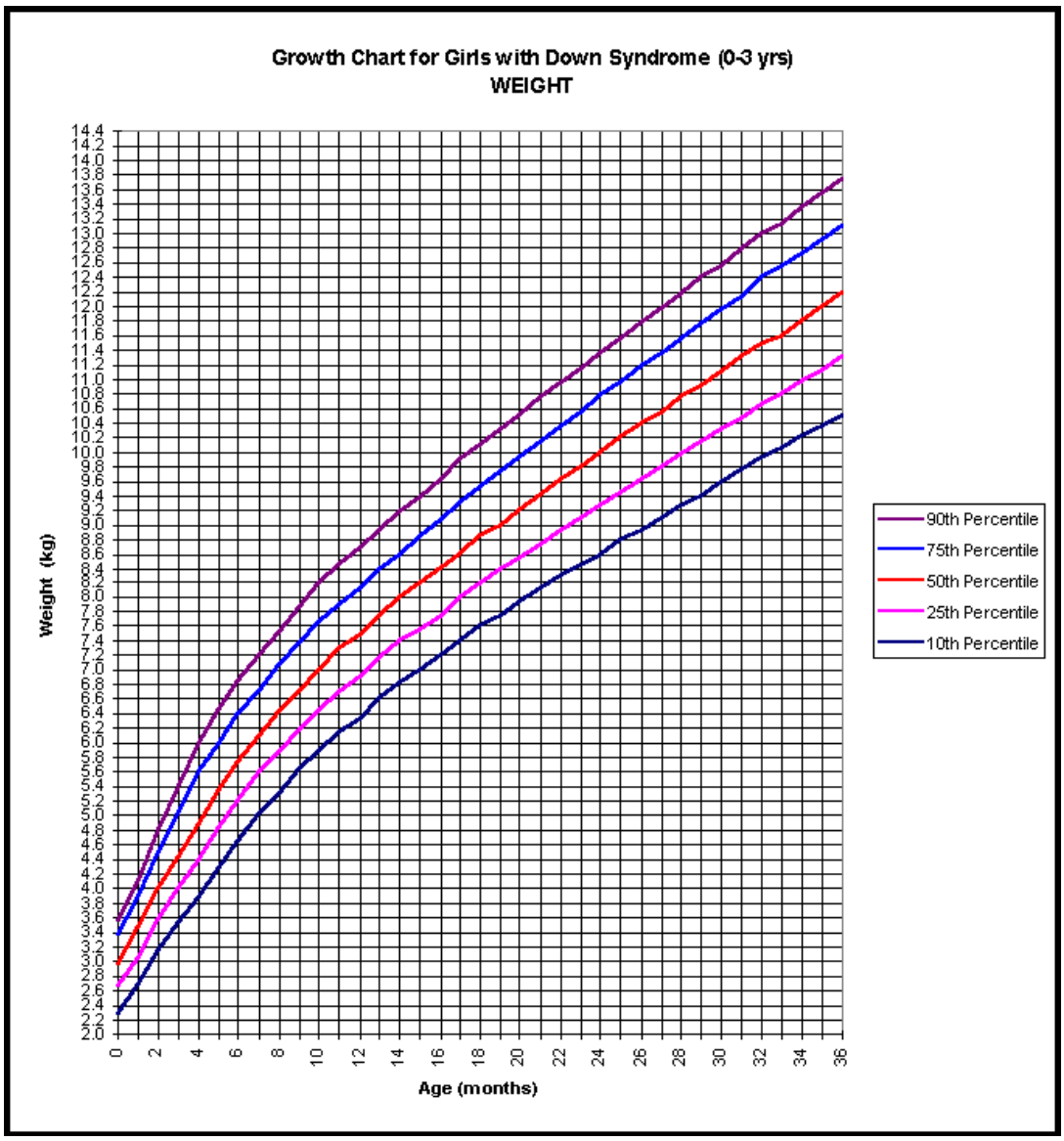
Age (years)
95th Percentile
75th Percentile
50th Percentile
25th Percentile
5th Percentile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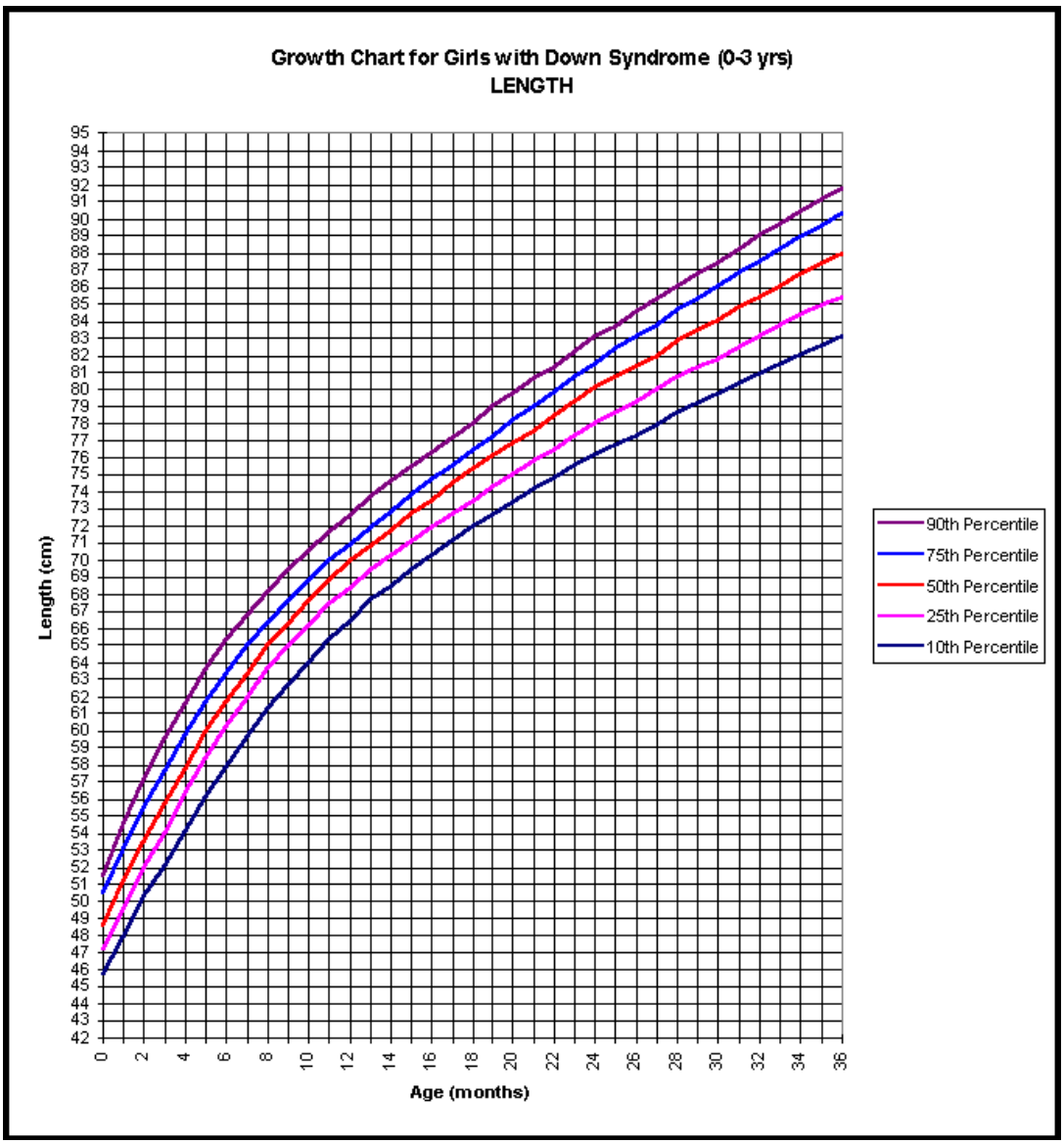
Weight (kg)
Age (months)
90th Percentile
75th Percentile
50th Percentile
25th Percentile
10th Percentile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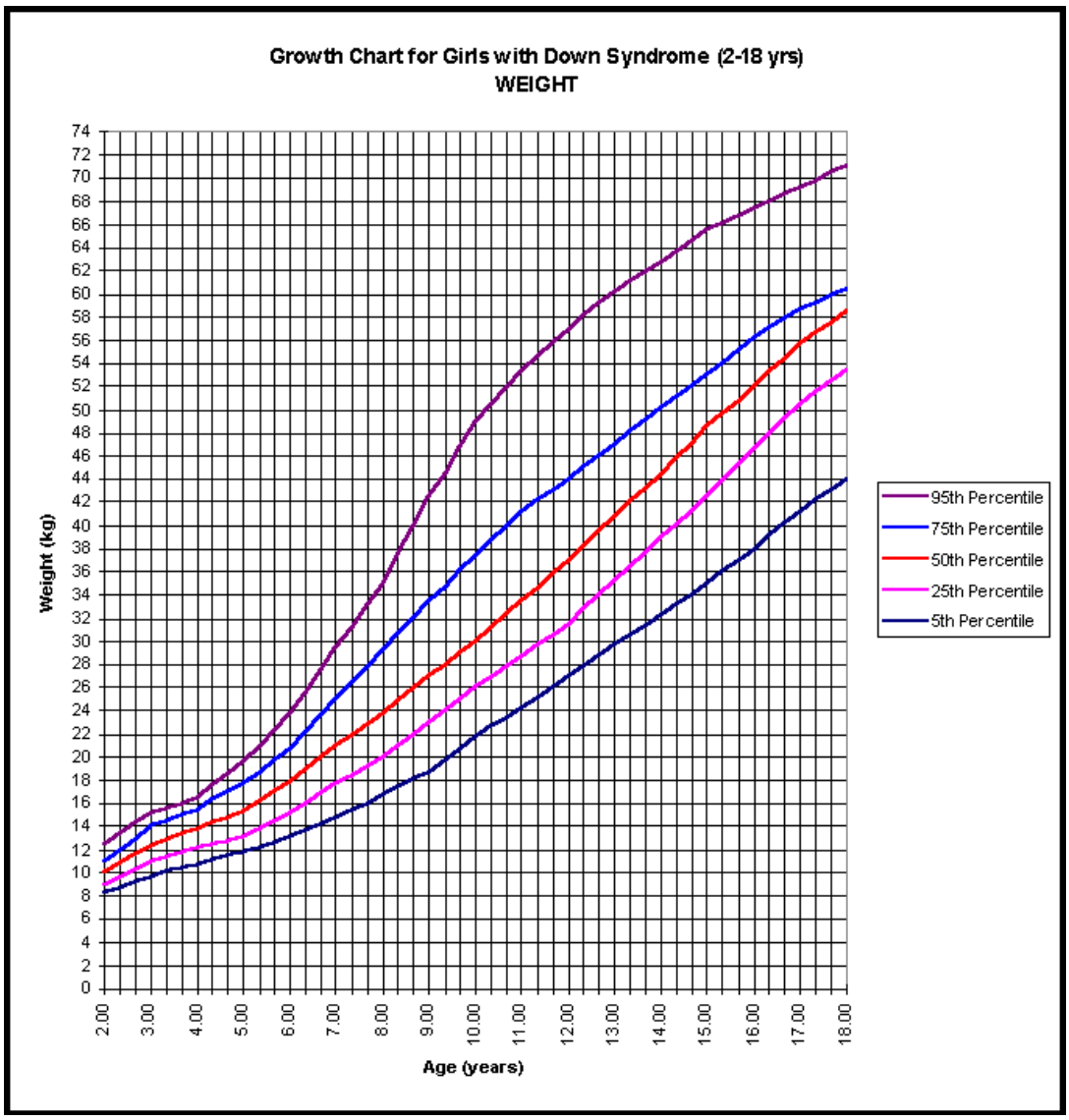
LENGTH
Length (cm)
Age (months)
90th Percentile
75th Percentile
50th Percentile
25th Percentile
10th Percentile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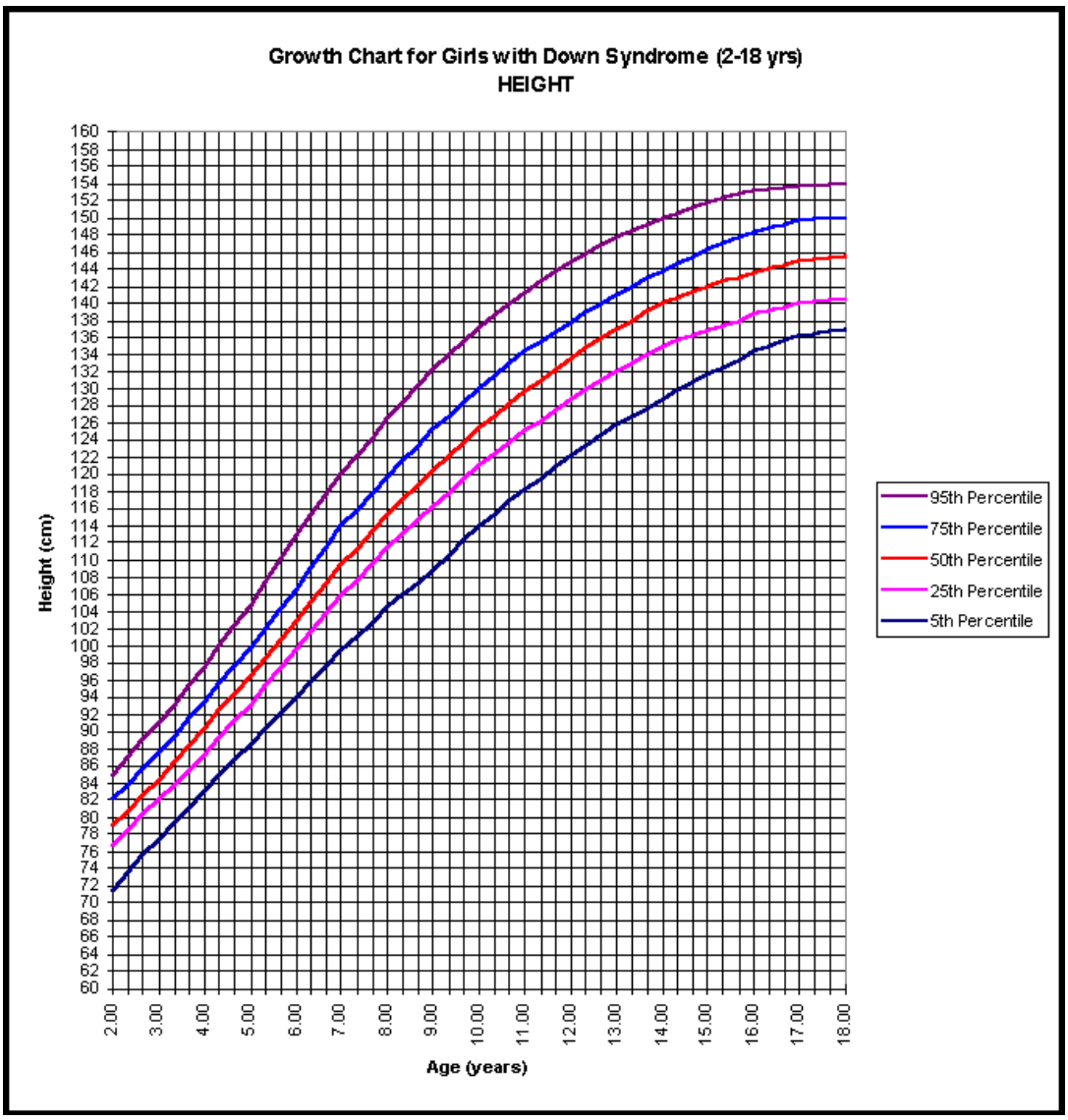
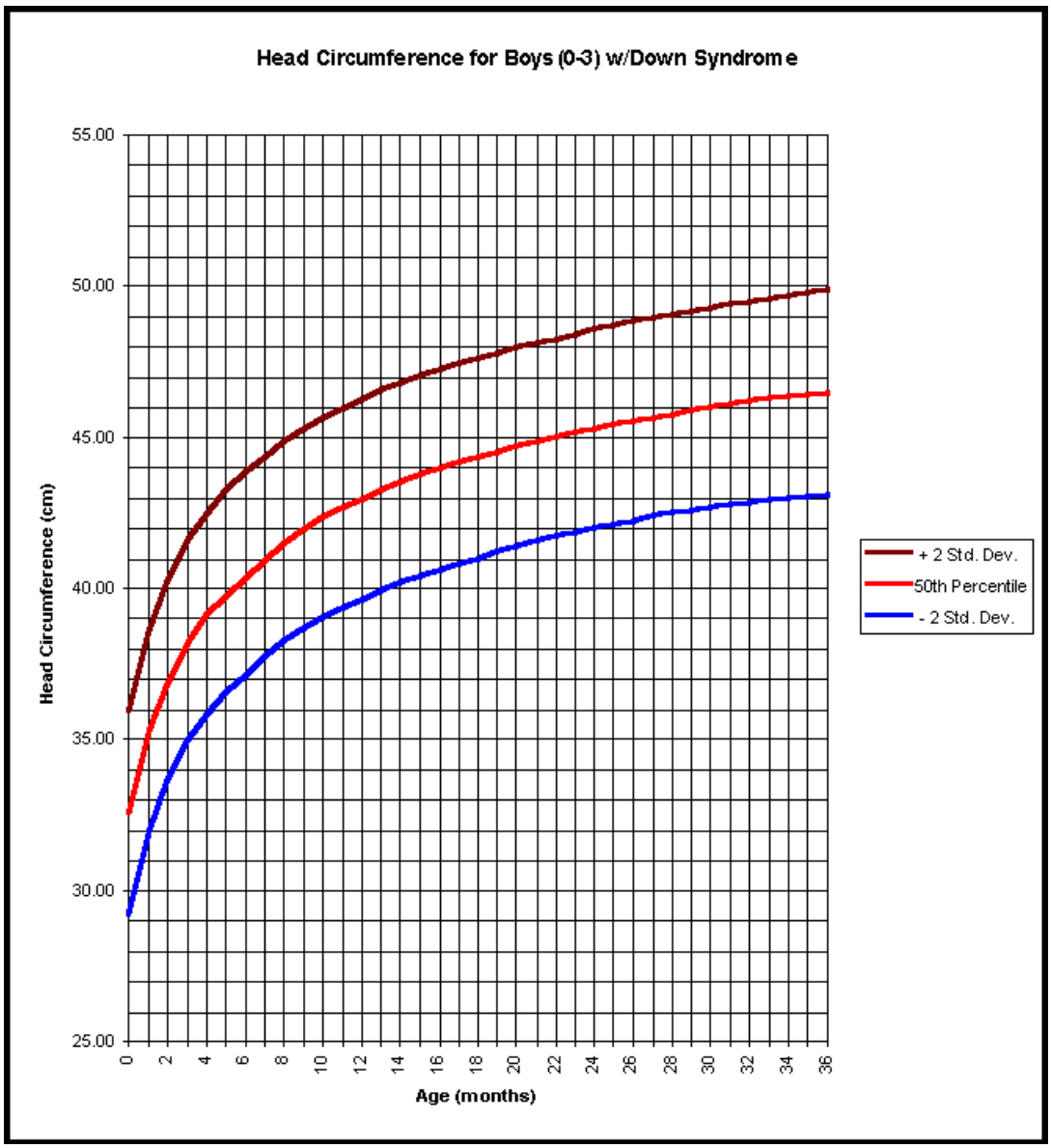
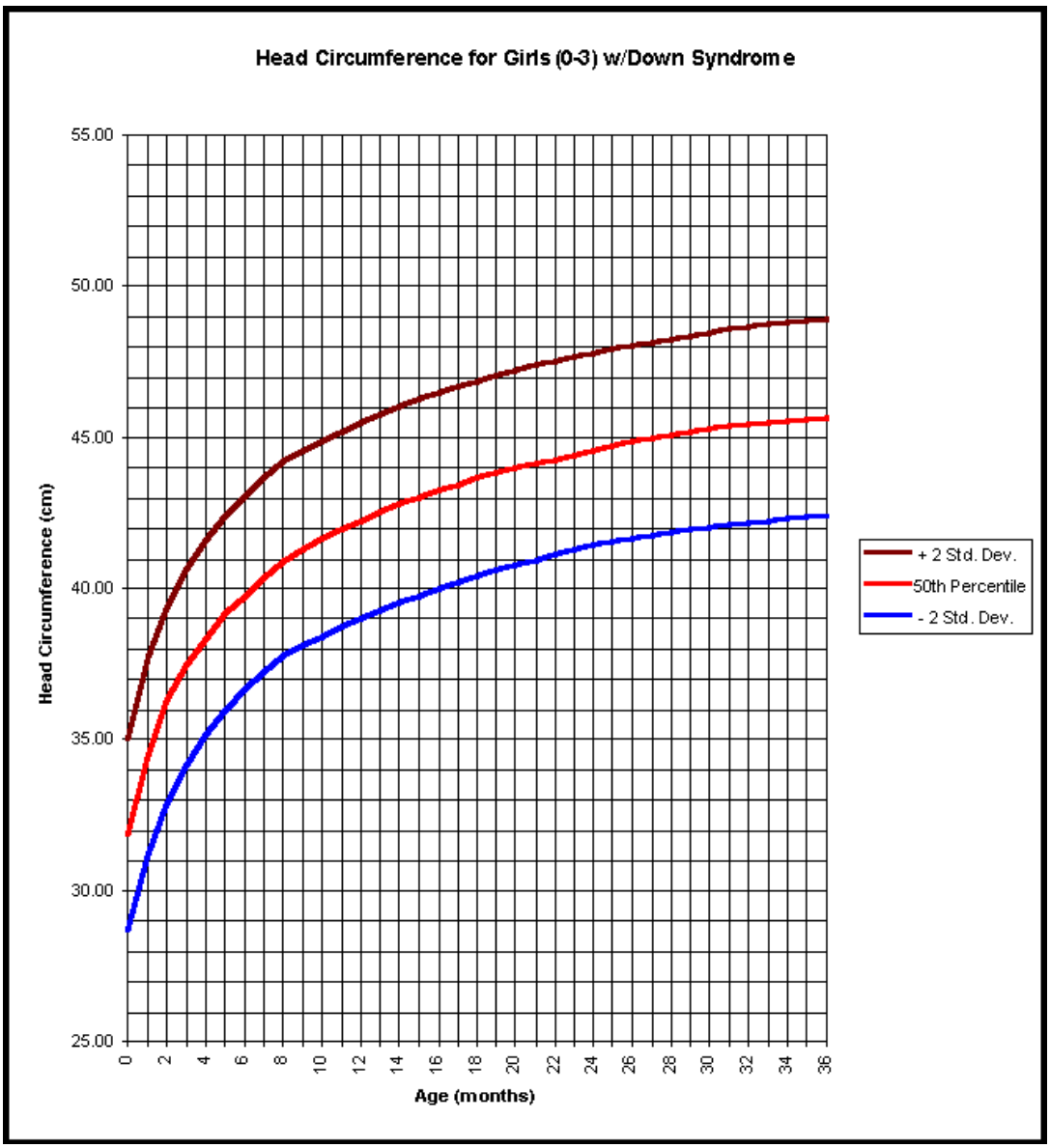
50th Percentile
25th Percentile
5th Percentile


Growth Chart for Girls with Down Syndrome (2-18 yrs)
HEIGHT
Height (cm)
160
158
156
154
152
150
148
146
144
142
140
138
136
134
132
130
128
126
124
122
120
118
116
114
112
110
108
106
104
102
100
98
96
94
92
90
88
86
84
82
80
78
76
74
72
70
68
66
64
62
60
2.00
3.00
4.00
5.00
6.00
7.00
8.00
9.00
10.00
11.00
12.00
13.00
14.00
15.00
16.00
17.00
18.00
Age (years)
95th Percentile
75th Percentile
50th Percentile
25th Percentile
5th Percentile


Head Circumference for Boys (0-3) w/Down Syndrome
Head Circumference (cm)
55.00
50.00
45.00
40.00
35.00
30.00
25.00
0
2
4
6
8
10
12
14
16
18
20
22
24
26
28
30
32
34
36
Age (months)
+ 2 Std. Dev.
50th Percentile
- 2 Std. Dev.


Head Circumference for Girls (0-3) w/Down Syndrome
Head Circumference (cm)
55.00
50.00
45.00
40.00
35.00
30.00
25.00
0
2
4
6
8
10
12
14
16
18
20
22
24
26
28
30
32
34
36
Age (months)
+ 2 Std. Dev.
50th Percentile
- 2 Std. Dev.

Appendix 9: Health and Welfare Chart

	Neonatal	Infancy						Early childhood						Late childhood	Adolescence/adulthood
Paediatrician follow-up			2mo		6mo		12mo		2yr	3yr		4yr 6mo		10yr	13yr
GP visits[1]		6w	3mo	5mo		9mo		15mo	2yr	3yr	4yr		5yr	Yearly visits	Yearly visits
Diagnosis															
Karyotype review[2]	●														
Phenotype review	●														
Genetic counselling[3]	●				●		●		●			●			
Medical evaluation	●[4]	●[4]	●[4]	●[4]	●[4]	●[4]	●[4]	●[4]	●	●	●	●	●	●	●[5]
Feeding/nutrition/growth[6]	S / ■	S / ■	S / ■	S / ■	S / ■	S / ■	S / ■	S / ■	S / ■	S / ■	S / ■	S / ■	S / ■	S / ■	S / ■
Upper airway obstruction	S	S	S	S	S	S	S	S	S	S	S	S	S	S	S
Spinal cord compression								S	S	S	S / (■)	S	S	S	S
Echocardiogram	●														
FBC[7]	●														
Thyroid screening	●				●		●		●			●[8]		●[8]	●
Endomysial antibodies							●		●			●			
Medical referrals															
Hearing/audiologist			S / ■	S	S	S / ■	S	S	S / (■)	S / (■)	S	S / ■	S	S / ■	S / ■[8]
ENT specialist						●			●						
Vision			S	S	S	S	S	S	S	S	S	S	S	S / ■[8]	S / ■[8]
Ophthalmologist	(●)[9]				●			●				●			
Dental[10]									●	●	●	●	●	●	●
Developmental/academic/social adaptation															
Development and behaviour			S / ■	S / ■	S / ■	S / ■	S / ■	S / ■	S / ■	S / ■	S / ■	S / ■	S / ■	S / ■	S / ■
School performance													S / ■	S / ■	S / ■
Socialisation							S	S	S	S	S	S	S	S	S
Disability services and support															
Child Development Service referral	●														
SES referral	(●)								●						
CDA form	●														
Needs assessment[11]	●											●		●	●
Recommend support groups	●								●						
Long-term planning	●						●		●			●		●[12]	●[12]

Notes

S = subjective, by history; ● = to be performed; ■ = objective, by a standard testing method; ( ) = optional

1 General practitioner reviews
2 Genetics referral for translocations, mosaicism
3 Recurrence risks, prepregnancy planning, prenatal diagnosis
4 Gastrointestinal symptoms
5 Gynaecological exam, if sexually active
6 Down syndrome growth curves are available
7 Leukaemoid reaction and polycythaemia
8 Test every two years
9 Must see red reflex to rule out cataracts
10 Check-up twice yearly
11 Required to access certain Disability Support Services
12 Information on vocational programs, sexuality, independent living

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