MEDICAL CARE OF PEOPLE WITH DOWN SYNDROME

Although Down syndrome is not a medical illness, there are a number of pathological processes, which are more common in people who have the condition. These associations will necessarily bring people with Down syndrome into frequent, and at times intensive, contact with doctors.

Neonatal period:
Immediately after delivery the child should be fully examined to confirm the diagnosis and to identify any immediate medical problems. Paediatric consultation is appropriate in most situations.

Cardiac: Congenital heart disease, usually in the form of endocardial cushion defects, affects 40% of babies and should be screened for by echocardiography soon after birth as it may well be difficult to detect. Septal defects and Fallot's tetralogy also occur. The discovery of severe congenital malformations often raises the issue of how interventive to be. It must be emphasised that exactly the same medical and surgical treatment should be instituted for a child with Down syndrome as for a child without a chromosomal disorder.

Severe congenital heart disease remains a major killer of children with Down syndrome, despite advances in surgical treatments. In the absence of a congenital heart defect, however, the majority of children can expect to live into their sixth decade.

Gastrointestinal: The commonest congenital abnormality of the gastrointestinal tract associated with Down syndrome is duodenal atresia, although pyloric stenosis, Hirschsprung's disease and tracheo-oesophageal fistulae have all been reported. Again, surgical intervention should be evaluated without reference to the chromosomal disorder. The total incidence of GIT malformations is approximately 12%.

Vision: Three percent of newborns with Down syndrome will have dense congenital cataracts which should be removed early. Glaucoma is also more common.

Feeding: Hypotonia is a constant feature of neonates with Down syndrome. This floppiness can interfere with breastfeeding and an experienced lactation consultant may need to be involved to ensure that the process is successful. Feeding tends to take longer and there may be attachment problems due to a protuberant tongue. Some babies experience

difficulty maintaining temperature and may need extra swaddling during feeding. Constipation is more common due to hypotonic gut musculature.

Congenital Hypothyroidism: This condition is slightly more prevalent in babies with Down syndrome. It should be detected by the routine heelprick screen performed on all neonates.

Congenital dislocation of the hips: Joint laxity and hypotonia can combine to increase the incidence of hip dislocation, although true congenital dislocation is quite rare. Extra care should be taken during the usual neonatal examination.

Infancy:
Once any urgent medical conditions have been addressed and feeding has been successfully established, the parents will take their new baby home. If the general practitioner has not been involved during the in-patient stay, early contact is important to allow an assessment of the child's baseline medical condition. This "well child check" means that the doctor need not be confronted for the first time by an unfamiliar and obviously sick child several months later.

Medical care in the first year of life will include the continued management of any problems identified in the neonatal period as well as surveillance for acquired problems such as hearing or visual impairments. Early and regular contact with appropriately-experienced consultants should begin in the first year.

Seizure disorders are more common in children with Down syndrome (approximately 10%) and can occur from an early age. They are usually tonic clonic in nature.

People with Down syndrome have reduced cell-mediated immunity and so babies in particular are likely to suffer more respiratory infections. Upper airway obstruction is also more common due to hypertrophy of tonsils and adenoids. This alteration in immunity has also been implicated in the observed increase in incidence of leukaemia in people with Down syndrome, although the link is not clear.

In practical terms the decrease in immunity has little impact. The normal childhood immunisation program should be commenced at the usual time.

The philosophy of "early intervention" is now well accepted as having benefits for the child

and family. This refers to home-based or centre-based treatment of a disabled child by a variety of health professionals such as occupational therapists, physiotherapists and speech therapists from a very early age. The parents are also involved as therapists. These recognised, government-sponsored programs tend to be preferable to the so-called "fringe" therapists who can be exhausting of parental resources without producing many results.

Childhood:
As the child grows through the preschool years it will become evident that development is globally delayed. Physical milestones will be delayed due to hypotonia and joint laxity, speech is likely to be difficult and socialisation may be delayed. Psychometric evaluation shows that most children with Down syndrome have intellectual functioning in the moderately disabled range but the range is enormous. At this time it is useful to assist the parents in recognising that a different set of milestones is relevant for this child, and that comparing its progress to that of siblings is not much help. Comparing notes with other parents of children with Down syndrome is helpful but it must be remembered that each child will follow their own path. It is important not to make too many predictions as to how far and how fast the child will develop but the mood should be one of optimism and reasonable expectations, as for all children.

The general practitioner's lifelong relationship with the child should be developing well at this point. Familiarity with what is normal for that child will allow the early recognition of any health problems, as will an awareness of which medical conditions are more common. Beyond that, however, the astute GP will remember that the child with Down syndrome is susceptible to the same range of childhood problems as any other and that not all symptoms will be due to the syndrome.

The GP's approach to a child with Down syndrome should be the same as to any other child: friendly, non-threatening and interactive. The parents are usually invaluable sources of information about the child and, after several years of hard work, will have become staunch advocates and "bureaucracy-busters". Their concerns should be treated with due consideration.

Having been involved in early intervention and preschool programs, most children with Down syndrome are well-equipped for entry to mainstream schooling at the usual time. There remains a lot of debate over the relative merits of mainstreaming versus special schooling which is outside the scope of this article. The doctor's role is generally to support

the parents in their decision making and to address any medical issues which may arise when selecting a school.

Congenital Heart Disease: Severe malformations which cannot be definitively treated remain a major cause of morbidity and mortality throughout childhood. Close liaison with a paediatric cardiologist should be maintained.
Sensory deficits: Significant hearing impairments occur in the majority of children with Down syndrome. Annual audiometry and specialist consultation is recommended.

Visual impairment due to refractive errors or strabismus is also common and should be checked annually. Cataracts often develop but are usually outside the visual axis.

Hypothyroidism: With a lifetime incidence of anything up to 30%, hypothyroidism must be screened for on a regular basis. Although most cases develop in teenage years, biannual biochemical screening of younger children is recommended. If any symptoms of thyroid disease are detected, early investigation and treatment are essential.

Atlantoaxial instability: There has been a lot of controversy as to the correct approach to this problem. Up to 15% of children with Down syndrome will have radiological evidence of instability of the atlantoaxial joint but in only a handful of cases will this instability result in an impingement on the spinal cord with resultant neurological signs. The controversy arises over whether to screen radiologically all people with Down syndrome and if so, when. If instability is detected, is it fair to limit a person's sporting and recreational pursuits in an attempt to prevent the rare complication of damage to the spinal cord? Subtle neurological signs are sometimes difficult to detect in people with Down syndrome and the surgery involved in stabilising the joint is major.

The current consensus is marginally in favour of radiological screening before school entry, mainly to reassure the parents of the large majority of children who will be found to have stable AA joints. If instability or anatomical abnormalities are detected, careful counselling should ensure that activities are modified appropriately without restricting the child unnecessarily. Neurological surveillance is essential. Some authorities recommend a second set of screening radiographs before entry to high school for all children with Down syndrome although there is little evidence to support the concept of development of instability.

Physical growth: Physical development is invariably delayed in children with Down syndrome

and modified percentile charts are available for accurate monitoring (). A tendency towards obesity requires special attention to healthy diet and exercise habits in this group.

Dental care: The teeth of children with Down syndrome tend to be small, irregularly spaced and misshapen. Early and frequent dental care is required to ensure adequate dentition for adult life.

Adolescence:
Having Down syndrome does not protect against the hormonal maelstrom, which usually accompanies adolescence. All the usual trials and torments of this potentially difficult phase of development have to be negotiated. This includes the adolescent trying to establish his or her own identity, find some private space and pursue their own interests.

People with disabilities are sexual beings and those with Down syndrome are no exception. It is a grave injustice to hold a stereotype of people with Down syndrome as being "happy eternal children", as any parent will agree. Teenagers with Down syndrome are subject to the same tempers, desires and emotions as anyone else, although they are often more frustrated in their expression.

Some specific medical conditions need attention:
Menstruation & sexuality: Menarche is usually only slightly delayed in girls with Down syndrome. Menstruation usually settles into a regular pattern and, although many cycles will be anovulatory, fertility should be presumed. There are approximately thirty cases in the world literature of women with Down syndrome becoming pregnant.
There is a long history of women with Down syndrome having their menstruation and fertility controlled through the use of medications such as progesterone or surgical intervention. Little attention was paid to the woman's needs and even less to providing appropriate education on menstrual hygiene, relationships and contraception. A hysterectomy does not protect against sexual abuse, particularly when many as compliant and helpless see women with Down's syndrome. More enlightened thinking has allowed these women to successfully manage their own menses and to make decisions about contraception, based on appropriate information from expert counsellors.

It is hard to justify involuntary menstrual suppression or sterilisation unless there are major medical indications. A recent High Court decision has given the Family Court responsibility for consenting to sterilising procedures on children. Most states have legislation giving intellectually disabled adults the same protection through Guardianship Boards.

Adolescent males with Down syndrome usually experience the same sexual drives and frustrations as their peers. The genitals are usually small and underdeveloped although this is by no means invariable. Some men have difficulty attaining a full erection and ejaculation is not always possible. Although the semen of males with Down syndrome shows decreased sperm numbers and motility with increased abnormal shapes, there is at least one recorded case of a child being fathered by a man with Down syndrome.

Education about appropriate sexuality is essential. One of the great obstacles to developing healthy sexual expression for people with Down syndrome is the lack of information which other teenagers have access to through a variety of community sources. Family Planning clinics and women's health resource centres can often be of assistance to families and general practitioners in this area.

Hypothyroidism: As most cases of hypothyroidism in people with Down syndrome develop during the teen years, surveillance should be increased to yearly thyroid function tests along with an increasing index of suspicion as to whether this condition could be behind unusual clinical presentations.

Skin: The skin of children with Down syndrome tends to be dry and susceptible to eczema. During adolescence, folliculitis and boils become more common. Alopecia areata is a common manifestation of the disordered autoimmunity, which may accompany Down syndrome.

Adulthood:
Changes in the approach to people with Down syndrome in the latter part of this century has resulted in a threefold increase in their life expectancy. Better and more active medical care coupled with community living has been largely responsible. It is only in the last few decades that people with Down syndrome could reasonably expect to reach adulthood. If the first five years of life (when most cardiac deaths occur) are survived a person with Down syndrome has an 80% chance of reaching the third decade and a 60% chance of living beyond fifty. The average age of death for a person with Down syndrome is in the mid-fifties. All this means that general practitioners need to employ the same preventive health skills as they do for the rest of the community so that people with Down syndrome can be healthy into old age.

People with Down syndrome still complain about the way they are perceived by some doctors. A man of twenty-five should not be called a "boy". He has usually moved out of the

family home and is working in an adult centre. He is probably able to answer questions about his health and should be given the opportunity to do so. He is an adult and should be treated as such. Medical procedures should be explained to him in appropriate language (see figure 3) and consent should be gained in the usual way.

If there is any likely controversy over a medical treatment or procedure for an adult with Down syndrome, a guardian may need to be appointed to make decisions on behalf of that person. This only applies if the person is not competent to give informed consent and it is one of the few areas of the law where common sense has a large role to play. Usually a parent or concerned friend can be seen to be clearly capable of making decisions, which are in the best interests of the disabled person. Uncertainties about the need for a guardian can usually be cleared up by a telephone call to the Guardianship Board or Attorney General's Department in each state.

Preventive care: The usual range of preventive health activities should be offered to adults with Down syndrome. The usual protocols apply for cervical cytology, breast screening and cardiovascular risk factor monitoring. Diet and exercise advice should be realistic, comprehensive and adapted for the person's living and employment situation where necessary.

Hypothyroidism: Throughout the adult years vigilance for thyroid dysfunction should be maintained with annual thyroid function tests. Sensory deficits: On going contact with eye and ear specialists is advisable during adult life due to a high prevalence of acquired problems such as cataracts, keratoconus and sensorineural hearing loss.
Behavioural problems: One of the most difficult challenges confronting a GP in treating a person with an intellectual disability is the assessment and management of difficult behaviours. Our predecessors seem to have relied heavily on the use of major tranquillisers as a way of quelling angry outbursts or destructive behaviour but this approach does nothing to identify the root cause of the problem. People with disabilities rarely behave in a destructive or violent way unless there is a reason. It may be something intrinsic to the person, such as depression or a toothache. It may be due to a change in that person's life situation, such as a change of residence or employment. Or it may be due to interpersonal conflict. The doctor's role is to seek the underlying cause of the problem through a full assessment rather than just to suppress the symptoms.

Psychiatric disorders: Psychiatric illnesses occur in people with Down syndrome with much the same frequency as in the rest of the population. This is a difficult area for psychiatrists,

however, and there are few in Australia who has risen to the challenge of providing comprehensive mental health services to people with intellectual disabilities.
Dementia: Much recent attention has been focussed on the association between Down syndrome and Alzheimer's disease. There appears to be a gene-dose effect where having an extra chromosome 21 gives an individual a higher chance of developing the neuropathological changes of Alzheimer's disease, which are coded for on that chromosome. The association is so strong that some authorities are advocating the inclusion of Alzheimer's disease as one of the invariable features of the syndrome, although it is always a diagnosis of exclusion after other, reversible causes of dementia have been excluded. Late onset epilepsy is often seen in association with Alzheimer,s disease in this group.

From a practical point of view it is helpful to be aware of this association. Due to the increased life expectancy of people with Down syndrome, most of our patients can be expected to show signs of dementia as they age. The recognition of declining intellectual functioning and loss of some social skills may assist the GP in suggesting appropriate strategies to help the patient and family. These may include a move to more appropriate accommodation, involvement in generic geriatric services and contact with specialised support agencies.

Summary:
People with Down syndrome are part of our community and as such are part of the GP's patient population. While there are some medical conditions, which are more common in people with an extra chromosome 21, there is nothing which is unique to this group or which is totally outside the scope of general practitioner involvement. The key to good quality care is to be familiar with the syndrome but, more importantly, to know the person who has the syndrome.

The advocacy efforts of people with Down syndrome and their families have resulted in huge improvements in life quality and expectancy. The medical profession is attempting to match these developments by becoming more sophisticated in its approach to ethical issues, more energetic in its provision of care and more cooperative in its interactions with people who have Down syndrome and their families.

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