

# Down Syndrome - Information sheet for Adults

(Optional sheets to be included where recipient of PHR has Down syndrome)

The material in this sheet has been adapted from the Therapeutic Guidelines book 'Management Guidelines for People with Developmental and Intellectual Disabilities' and updated from the 2005 version 'Management Guidelines – Developmental Disability' which can be consulted for more detailed information.

Down syndrome (Trisomy 21) is the commonest identifiable cause of intellectual disability with an incidence of approximately 1 in 660 live births. It is due to the presence of an extra Chromosome 21. It is more common in older mothers but, as the vast majority of babies are born to younger women, most mothers of babies with Down syndrome are aged in their twenties. Despite a growing trend towards women having children later in life, the annual rate of Down syndrome births in Australia is remaining fairly constant due to advances in prenatal screening.

Ninety two percent of cases of Down syndrome have trisomy 21 in all cell lines, with the remainder being split evenly between mosaic forms and translocated forms. The commonest translocation leading to Down syndrome is from the 21st to the 14th chromosome.

In at least 90% of cases of trisomy 21 the extra chromosome is maternally-derived, although determining the parent of origin usually serves no useful purpose. The chance of a couple who have conceived one trisomy having a further pregnancy with Down syndrome is considered to be 1%, except in the case of balanced parental translocation in which case the chance of recurrence is much higher.

## Principles of Medical Care

Down syndrome is one of the few causes of intellectual disability in which there are many specific medical issues related to the syndrome. These include higher prevalences of:

- hypothyroidism
- visual and auditory deficits
- congenital heart defects
- gastrointestinal malformations
- epilepsy
- atlantoaxial instability
- obstructive sleep apnoea
- depression
- dementia of Alzheimer type 2
- immunodeficiency
- leukaemia

Nevertheless, most medical interactions between people with Down syndrome and their GP will be for everyday medical problems which are unrelated to the syndrome.

General practitioners can expect to be involved in the medical care of people with Down syndrome from the time of first diagnosis through to old age. The GP who is most successful in providing this care tends to be comfortable in communicating with the patient and willing to value that person equally within the patient population. In particular, adults with Down syndrome express the full range of human personality traits and should not be stereotyped as "happy eternal children".

## Unproven Therapies for Down syndrome

General practitioners can expect to be asked about the many controversial nutritional or physical therapies which have been promoted as being "cures" or at least significantly efficacious in ameliorating the disability associated with Down syndrome. Rather than dismissing the parent's request for advice, however, practitioners can help individuals make informed choices about the use of such therapies. Practitioners may also use the opportunity to talk about practical issues which may impact on the individual's well being and development, such as family functioning, stress of family members, knowledge of resources available (eg respite care), and whether the individual has a loving and stimulating environment.

## **Medical Issues in Adulthood**

Early adult life is the peak time for development of hypothyroidism. Mitral valve prolapse may also occur at this age, and preventive health screens should continue throughout adulthood. The incidence of epilepsy begins to rise steadily through adult life after an initial peak during infancy, due to the deposition of proteinaceous material in the brain.

### **Recommendations**

- Annual thyroid function screening.
- Annual cardiovascular examination and, if clinical uncertainty exists, echocardiogram.
- Biannual screening for visual and auditory impairments.
- Annual otoscopic examinations.
- Usual blood pressure monitoring.
- Usual cholesterol screening.
- Usual breast or testes examination.
- Usual cervical cytology (if ever been sexually active).
- EEG if clinically indicated.

## **Aged Care**

Life expectancy for people with Down syndrome has increased significantly in the last forty years with many now living beyond the sixth decade. Reasons for this include surgical correction of congenital abnormalities of the heart, improved nutrition and healthier living conditions.

### **Dementia and Alzheimer Disease**

There is evidence that Alzheimer disease can occur up to two decades earlier in people with Down syndrome than in the general population. This is thought to be related to the metabolic impact of Trisomy 21 with over-expression of certain genes related to ageing on chromosome 21. The true incidence of the disease in people with Down syndrome is difficult to assess as there is not a close correlation between the neuropathological changes and the behaviour changes.

### **Assessment and Management**

The GP's primary role when consulted by a person with Down syndrome who seems to have deteriorated in function is to exclude causes such as:

- depression
- situational crisis
- hypothyroidism
- menopause
- diabetes
- visual and hearing impairment
- cerebrovascular accident
- adverse effects of medication
- occult infection
- carcinoma

Work towards a formal diagnosis of dementia should only proceed after exclusion of these conditions and treatment of any detected pathology.

Early diagnosis of Alzheimer's disease is important in people who have Down syndrome as treatment with cholinesterase inhibitors can produce some recovery of skills and slow the progress of the subsequent deterioration. It is important to ensure there is no cardiac contraindication for the use of cholinesterase inhibitors. Special provisions allow the NHS prescription of these drugs for people who do not meet the usual Mini Mental State Examination (MMSE) criteria.

If Alzheimer disease is diagnosed as the cause of a decline in functional ability a person with Down syndrome, the GP should become involved in efforts to allow that person to remain in their current accommodation and occupational settings with appropriate modifications and supports.

# Down Syndrome - Information sheet for Children and Adolescents

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- coeliac disease
- leukaemia

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## Neonatal Care (0-6 Weeks) -recommendations

- An early paediatric review, including ophthalmology and cardiology examination.
- Liaison with a trained lactation consultant if hypotonia causes feeding difficulties.
- Contact with a trained representative of an official Down syndrome organisation.

## Infancy (6 weeks - 1 year) - recommendations

- A "well child check" at about 6 weeks can set a baseline for that child's health and to allow the GP to support the parents in coming to terms with their child's disability.
- Annual biochemical screening for hypothyroidism.
- Auditory brainstem evoked response within first six months.
- Immunisations should be given with the usual doses of vaccines at the usual intervals.
- Use special growth charts (available from "Down Syndrome Society" State offices).
- Referral to useful support services such as early intervention therapeutic services (physiotherapy, occupational and speech therapy), a local support group for parents of disabled children, and Centrelink for information about the Child Disability Support Allowance.

### **Pre-school and Kindergarten Age (1 year- 5 years) - recommendations**

- Annual hearing tests (usually standard techniques are sufficient) and tympanometry.
- Annual ophthalmological examination.
- Regular dental checks.
- Continue annual biochemical screening for hypothyroidism.
- Kindergarten -most children attend mainstream kindergartens and enjoy the socialising experience.
- Assessment for school readiness may require specialist advice.

### **Primary school age ( 5 - 11 years) - recommendations**

- Biannual screening for visual and auditory impairments.
- Annual otoscopic examinations.
- Monitoring for overgrowth of oropharyngeal and nasopharyngeal lymphoid tissue which can lead to sleep apnoea and cor pulmonale..
- Preventative dental care.
- Biannual biochemical screening for hypothyroidism is necessary at this age.
- Atlantoaxial Instability - care should be exercised with procedures that involve hyperextension of the neck. Symptoms which could be due to spinal cord compression such as neck pain, torticollis, altered gait, increased reflexes, clonus, up-going plantar reflexes, sphincter disturbances, or unexplained behaviour change warrant thorough neurological review and consideration of magnetic resonance imaging of the cervical spine.
- Education - most children can do well in mainstream settings with varying degrees of integration support.

### **Adolescence (12-18 years) - recommendations**

- Sexuality and Fertility
  - Preparation for menstruation should begin before menarche, which occurs at the usual time, with careful instruction and demonstrations using sanitary products and the girl's own body.
  - Boys with Down syndrome are usually presumed to be infertile. However education on protective behaviours and appropriate sexual expression is still vital.
  - If contraception is required, the usual range of options is available. However, a careful process of education and supervision may be required and education on protective behaviours and appropriate sexual expression is vital.
- Atlanto-axial Instability - continue surveillance for neurological symptoms of spinal cord compression and to encourage adolescents who are known to have large atlanto-dens intervals (greater than 5mm) to avoid high risk activities such as contact or high speed sports
- Obesity and Obstructive Sleep Apnoea
  - The establishment of good diet and exercise habits is important to avoid obesity.
  - Obstructive sleep apnoea is more common in people with Down syndrome and this is worsened by obesity. It can cause headache, irritability, somnolence and behaviour change.

### **Transition to Adulthood - Employment, Independence, and Psychological Health**

- The transition from adolescence to adulthood can be a difficult time for people with Down syndrome due to changes in family relationships over time and perceived limitations of opportunities. Depression, loss of skills, social withdrawal and aggression can all be caused by problems negotiating this passage.
- Adult life can be relatively independent for many people with Down syndrome while others require varying degrees of support from their family or external agencies.
- Employment is usually available in a range of supported environments and options for accommodation include independent living with outreach support.