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Australians with Down syndrome

Health matters

Background

The health and life expectancy of Australians with Down syndrome has improved dramatically over recent decades, resulting in more people living into adulthood and accessing community and hospital based health and social services.

Objective

This article presents information and resources helpful to general practitioners providing healthcare to patients who have Down syndrome. Healthcare issues through the lifespan are explored, the importance of proactive management is emphasised and strategies are outlined.

Discussion

Australians with Down syndrome are an interesting and rewarding group of people with whom to work. They present us with particular challenges in the way we provide healthcare and, in doing so, offer us an opportunity to improve the way we work with other patients who have cognitive and communication difficulties; chronic, complex health and social needs; family or paid carers involved in health management and those who require health advocacy as a part of healthcare provision.

Keywords: Down syndrome; general practice; preventive health services; disability health, disablilty healthcare

CPD

Knowing a person has Down syndrome (DS) alerts us to their increased risk of a range of medical conditions and provides an excellent example of how understanding the aetiology of disability informs medical care. This article provides specific information on the care of a patient with DS, and general principles relevant to the care of anyone with cognitive impairment.

Over recent decades the lives of Australians with DS have been transformed almost beyond recognition. In the 1950s, attitudes toward people with DS were often negative, expectations low and opportunities limited.[1,2] Now people with DS grow up with their parents, siblings and peers; enjoy better health and longer lives; have opportunities for participation and contribution in education, employment and recreational activities; and contribute in many ways to their families and communities.[3,4]

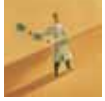
The change in life expectancy over this period has also been remarkable. In the 1950s children born with DS in Australia had a life expectancy of 15 years;[1] now it approaches 60 years.[5] This dramatic change is due both to medical advances and to changing social attitudes toward people with disabilities. More people are therefore living into adulthood and old age and accessing community and hospital based health and social services.

Current health issues

People with DS still experience significant barriers to the receipt of high quality healthcare, and health outcomes compare unfavourably with those of the general population.[6,7] They are at increased risk of range of physical and mental health conditions, often live with multiple unrecognised health issues, and have limited access to disease prevention and health promotion interventions.[6–8]

Prevalence

Down syndrome is the most common known cause of intellectual disability. In Australia DS is identified in 1:1150 live births (around 260 births each year).[9,10] The chance of a woman conceiving a child with DS varies from 1 in 1400 for a woman 20 years of age to 1 in 32 at age 45 years.[11] Although the chance of having a child with DS increases with maternal age, most babies with DS are born to



mothers less than 35 years as this is the most fertile age group. The prevalence of people with DS in our community has increased over recent decades due to the dramatic increase in life expectancy.[5]

Diagnosis

The diagnosis of DS may be made before or soon after birth. Many Australian women are routinely offered prenatal screening; guidelines for pretest counselling are readily available.[12] Testing includes nuchal translucency and biochemistry in the first trimester; and triple/quadruple testing in the second. When screening indicates a high risk of the fetus having DS, a diagnostic test (chorionic villi sampling [CVS] or amniocentesis) is offered.[12]

Antenatal screening raises many ethical issues.[3] If trisomy 21 is identified antenatally, the parents must decide whether to continue with the pregnancy. This decision is influenced by many factors relating to the parents, family circumstances, socio-cultural environment, professional advice – as well as the child's likely pattern of abilities and disabilities. When the diagnosis of DS is made after birth, the parents' experience is also stressful and they may fear what the future will bring. Life, however, often unfolds in ways very different from those imagined: view Novita Services'[4] online video of a mother describing her experiences of diagnosis and beyond.

Genetics of Down syndrome

Down syndrome usually (95%) results from nondisjunction of chromosome 21 leading to the embryo having trisomy 21. Rarer causes are translocation (3–4%) and mosaicism (1–2%).[11,13] Most instances of DS are not hereditary, however occasionally one parent has a balanced genetic translocation resulting in an embryo with DS. If translocation is identified, parental karyotype is important in genetic counselling.

Characteristics of Down syndrome

People with DS usually have readily recognisable characteristic facial features and assumptions about their abilities and potential are often made on that basis alone. In fact, people with DS are as different from each other as any other group in the community, and have physical and personality features that reflect their individuality, life experience and family, as well as their DS.

Everyone with DS has some degree of cognitive impairment and learning difficulty, although the degree varies from mild to profound. Many other features reflect the pervasive effect of trisomy 21 on body organs (*Table 1*). An individual may have few or many associated conditions; no-one has them all![6,8,14,16]

The provision of healthcare

People with DS tend to have lifelong complex health and social issues and require the contribution of health professionals from a range of disciplines to monitor health status, address current issues, and plan for future needs. The general practitioner plays a central role in referral and coordinating the work of this multidisciplinary team and monitoring the health and wellbeing of both the patient and their family.

When providing healthcare to people with DS, consider current and future health issues with these questions in mind:

- What health issues would I consider for any patient of this age and gender?
- What particular health issues should I consider for someone with DS of this age and gender?

Children

Children with DS, like all children, require optimal health to play and learn. In early life, medical care focuses on the identification and management of congenital anomalies and monitoring common conditions (*Table 2*). Medical care is usually provided by the GP in collaboration with a paediatrician. Most children with DS go to mainstream schools, and the family, GP and school need to work together to ensure adequate support for the child. Mixing with peers promotes social and language development and other children benefit from the opportunity to understand and appreciate individual differences and contributions.

Adolescents

Adolescents with DS (*Table 3*) face many of the same pubertal and social issues as other teenagers, although they tend to do so at a later chronological age. Issues of puberty and sexuality need careful attention and management strategies will depend on the abilities and degree of independence of the young person and their social supports and circumstances. Where appropriate, the adolescent should be encouraged to spend part of the consultation alone with the GP to provide an opportunity to discuss personal issues and build a sense of autonomy and responsibility with respect to their healthcare.

Adults

Adults with DS (*Table 4*) rely on their GP to monitor their health and wellbeing. Knowledge of conditions occurring more commonly in people with DS informs assessment, investigation and management. Many adults develop sensory impairments that compound their disabilities and undermine their ability to interact with others, learn, study, work and enjoy their environment and experiences.

Annual health assessments (MBS Items 705, 707) provide the opportunity to identify health issues and address disease prevention and health promotion. Assessment includes medication review, preventive health, detection of comorbidities and risk factors, and management of active health issues. Findings prompt investigation, referral, and potentially, GP Management Plan/Team Care Arrangements.

Communication and behaviour

It is important to always include the person with DS in the consultation conversation, both verbally and nonverbally. Inclusion is courteous, builds rapport and encourages cooperation. The patient and their accompanying family or paid carer will 'pick up' on the respect and care conveyed. Simple strategies employed by the GP can greatly improve communication.[14,15]

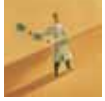
Table 1. Implications of trisomy 21 for body systems

Organ/system affected	Resulting conditions
Heart	• Congenital heart disease (50% neonates) • Mitral valve prolapse (50% adults)
Gastrointestinal tract (GI)	• Congenital GI anomalies (12%) including: oesophageal or duodenal web/atresia, tracheoesophageal fistula, pyloric stenosis, Meckel diverticulum, imperforate anus, Hirschsprung disease (1%) • Chronic constipation: relates to hypotonia, diet, exercise • GORD (common): relates to congenital GI dysmotility, hypotonia, weight, acquired dysmotility in Alzheimer disease (AD), *Helicobacter pylori* infection • Coeliac disease (5–15%)
Neurological	• Intellectual disability/learning difficulties (100%), epilepsy (6% children, 80% in those with advanced AD), AD (% increases as age increases – rare under 40 years, ~50% at 60 years) • Cord compression from subluxation/dislocation of atlanto-axial joint (~2%)
Sensory impairment	• Hearing loss (75%): sensorineural and/or conductive (otitis media, glue ear) • Vision: refractive errors (50%), strabismus (44%), nystagmus (20%), conjunctivitis, tearing from small/blocked nasolacrimal ducts, congenital (3%) and acquired (50%) cataracts, keratoconus (adults)
Respiratory	• Vulnerability to respiratory tract infections including URTI, otitis media, pneumonia (12-fold risk related to immunodeficiency, aspiration, hypotonia) • Sleep apnoea (50–75%): relates to hypotonia, anatomy of mid-face, weight
Musculoskeletal	• Characteristic physical features: small stature, single palmer crease (20%), brachycephaly, microcephaly • Hypotonia: contributes to feeding difficulties, sleep apnoea, hyperflexibility, joint subluxation/dislocation, eg. hips (6%), patella • Hypoplasia of midface: contributes to otitis media, mouth breathing • Atlanto-axial instability (14%, symptoms/signs in ~2%)
Immune system	• Recurrent respiratory tract infections including pneumonia • Folliculitis, blepharitis, conjunctivitis, periodontal disease, aphthous ulcers • Autoimmune disease: alopecia, hypothyroidism, diabetes, coeliac disease
Dermatological	• Dry skin, infections, eg. folliculitis, atopic dermatitis, seborrhoea, blepharitis, psoriasis, alopecia (~8%)
Endocrine	• Hypothyroidism (3–54%, increases with age) • Failure to thrive, obesity, diabetes • Women: early menopause (~44 years), decreased fertility • Men: hypogonadism, undescended testes, decreased fertility • Osteoporosis: increased risk related to diet, anticonvulsants, sun exposure, weight bearing exercise, hypogonadism, early menopause
Genitourinary	• Congenital renal tract anomalies, hypospadius, cryptorchidism
Haematological	• Functional defects in white cells related to decreased immunity, macrocytosis, transient leukaemia or thrombocytopenia in neonates. Leukaemia is more common in children/young adults with DS (~1%)
Dental	• Delayed and abnormal dentition (eg. partial adontia 50%, microdontia 40%), periodontal disease, malocclusion, difficulties with oral hygiene
Mental health	• Bio-psycho-social risk factors: increased risk of anxiety, depression, obsessive compulsive disorder, AD (rare under 45 years, ~50% by 60 years)

People with DS have difficulties with receptive communication (understanding) that relate to cognitive ability and are compounded by hearing or visual impairments. Expressive communication (being understood) may also be affected by articulation difficulties relating to anatomical differences of the oropharynx. Early hearing impairment may undermine language development and speech quality.

Changes in behaviour are important communications and may be a sign of distress, physical or mental illness. Consider attempts to communicate wants and needs (eg. for company, an activity); emotion (sadness, fear, loneliness, excitement); difficulty understanding (confusion, misunderstanding); physical pain/illness (including dental, GORD); mental illness (anxiety, depression, mania, psychosis); sensory deterioration (hearing, vision).

Pain may be inferred from a grimace, or cold from a shiver. The behaviour may indicate pain at a specific site (eg. rubbing jaw/dental pain, limping/hip pain); general distress (eg. irritability



relating to earache, oesophagitis or depression); or other feeling of discomfort (eg. food refusal-nausea, withdrawal-depression). The diagnosis is usually in the history and observation. Energetic investigation is also required in people who have difficulty reporting or describing their symptoms.

A change of behaviour may be an indication of physical or mental illness. Effective management depends on accurate diagnosis.

Social support

People with DS usually live with family or paid staff who assist them in their daily lives. Knowing the home circumstances and support available is relevant to healthcare; a home visit may provide valuable insights. Disability support workers usually have training in disability but no health training. Expectations of their role in monitoring health and illness, medication effects, and implementing management recommendations

Table 2. Healthcare for children with Down syndrome

General: for all children
Primary care: monitoring health and development. Health promotion and disease prevention: diet, exercise, weight management, immunisations
Specific: for children with Down syndrome
Neonatal examination and investigation to identify congenital anomalies, including cardiac and gastrointestinal effects
Feeding difficulties: Down Syndrome Association, Australian Breastfeeding Association. Dieticians can also offer information and support (note: calcium, vitamin D for osteoporosis prevention)
Families: link to services and supports, including early intervention services, Centrelink for entitlements, dental services, allied health services, support services including Down Syndrome Association
Hearing: Auditory Brainstem Evoked Response at 0–6 months; audiology annually from 1–5 years; 2 yearly from age 5–18 years AND at any time concern regarding hearing loss raised by parents, carers or screening
Vision: ophthalmological examination at 0–6 months; annually to age 5 years; 2 yearly to age 18 years AND at any time concern regarding vision is raised by parents, carers or by screening
Thyroid function: check at birth; then annually throughout childhood AND if suggestive symptoms or signs are noted
Gonadal: undescended testes, hypogonadism
Dental/oral health: 3–6 monthly dental review from first teeth for monitoring development PLUS prevention and treatment of oral disease
Consider requirement for subacute bacterial endocarditis prophylaxis for dental treatment (congenital heart disease, mitral valve reflux)
Gastrointestinal: monitor diet and weight. Consider GORD, *H. pylori*, coeliac disease, constipation
Atlanto-axial instability: monitor for symptoms and signs of cord compression
Haematological/immunological: be alert to increased risk of infections, leukaemia

Table 3. Healthcare for adolescents with Down syndrome

Care as for children, PLUS:
Girls: education and support to manage periods (see Resources). Advice regarding sexuality and contraception; require additional education and support for learning
Boys: advice regarding sexuality and contraception; require additional education and support for learning
Sexuality: vulnerable to sexual exploitation/abuse. Require education about appropriate behaviours (themselves and others), how to keep themselves safe, and how to get help if needed. For those unable to advocate for themselves, adequate protection and supervision is required to ensure their safety
Folliculitis/acne: may affect self esteem and the way others interact. Energetic treatment is required
Mental health: biological, psychological and social factors contribute to increased risk of disorders of mental health, especially anxiety and depression
Transition from paediatric to adult services: consider all services used by the child and their family: recreation, respite services, school/post school options (employment, education, other), paediatric to adult medical and mental health services. The GP has an important role in providing support and guidance through this time of change
Note: Encourage the adolescent to take an active role in their health management where appropriate

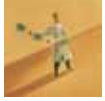


Table 4. Healthcare for adults with Down syndrome
General: for all adults
Health monitoring, risk factor identification (eg. cardiovascular disease)
Health promotion and disease prevention: diet, exercise, weight management, cancer screening (may include Pap test if indicated, mammography, bowel cancer screening), immunisations
Specific: for adults with Down syndrome
Annual health assessments: MBS Item 705 and 707 underpin proactive healthcare: enable early detection and health promotion and disease prevention
Behaviour change: a change in behaviour is a communication – consider disorder of physical, dental or mental health
Dental and oral health: 6 monthly dental review. Consider prophylaxis for subacute bacterial endocarditis for dental treatment (congenital heart disease, mitral valve)
Cardiovascular: mitral valve prolapse may develop during adulthood (50% of adults) – regular cardiac examination and/or echocardiography required
Atlanto-axial instability: cervical cord compression (~2% of people with DS). Symptoms/signs: neck pain, torticollis, limb weakness, increased reflexes, change in gait and/or bladder/bowel function, sensory changes
Hearing: audiology 3 yearly AND if suspicion of hearing loss raised by carers or screening
Vision: ophthalmological review at 30 years; every 5 years thereafter AND if suspicion of visual loss raised by carers or screening
Thyroid function: annually AND whenever suggestive symptoms or signs are noted
Mental health: biological, psychological, social factors contribute to increased risk of disorders. Anxiety and depression more common. Alzheimer disease – average age of diagnosis early 50s; rare under 45 years; exclude physical/mental cause of functional decline.
Gastrointestinal: increased risk of GORD and coeliac disease and chronic constipation
Osteoporosis: bone mineral density testing in early adulthood and repeat at menopause and ~40 years for hypogonadal men. Prevention: diet (calcium, vitamin D) and exercise – consider vitamin D supplement if levels low, anticonvulsant use
Medication: regular (eg. 3 monthly) review. Monitor response and side effects; re-evaluate indications (medication only if indicated), efficacy (cease if ineffective); ensure lowest effective dose; educate patient and carers about expected response and side effects. Consider pre-packaging of medication by pharmacy to ensure accurate dosing and safety. Sensitivity to psychoactive medication – 'Start low, go slow' and be vigilant for side effects (behaviour change may be an early symptom)

must take this into account. Issues impacting on staff communication such as staff turnover, shift work and the use of casual staff also impact on their ability to fulfil their important role as part of the healthcare team.

Healthcare for older people with Down syndrome

The longevity of people with DS means more people experience the conditions of aging including menopausal symptoms, arthritis, cardiac disease, osteoporosis, sensory loss, and dementia.[5] The GP plays a vital role in health monitoring. Vigilance is required with regard to a change in behaviour or function that may indicate physical disease, sensory deterioration, medication side effects or mental health disorder.

Alzheimer disease occurs earlier in people with DS, with an average age of diagnosis in the early to mid 50s.[5] Symptoms may be difficult to differentiate from physical disease (eg. hypothyroidism, anaemia); medication effects (eg. nausea, confusion, dizziness); mental illness (depression, psychosis); and sensory deterioration (hearing and vision). Energetic assessment and investigation is required to exclude treatable causes of functional decline.[5–7,16]

While the leading cause of death in those aged less than 40 years is congenital heart disease; pneumonia is the leading cause in older adults with DS.[5]

Loss of skills in an adult with DS may indicate physical or mental illness, medication effect, sensory deterioration, changes in the physical or social environment, or Alzheimer disease.

Conclusion

People with DS are part of our practice populations and there is much the GP can do to achieve and maintain optimal health and wellbeing for this interesting and rewarding group of patients. Regular health assessments and review, a strong collaborative relationship with the patient and family/carers, and a multidisciplinary healthcare team underpin good healthcare, and enable identification and management of health and social issues that arise throughout life.

Resources

- Health guidelines for adults with intellectual disabilities. Consensus statement: www.iassid.org/pdf/healthguidelines-2002.pdf

- Understanding intellectual disability and health: www.intellectualdisability.info
- Therapeutic Guidelines. Management guidelines: developmental disability: www.tg.org.au/index.php?sectionid=93
- Down Syndrome Medical Interest Group. Evidence based recommendations and checklists: www.dsmig.org.uk/publications/pchrhealthchk.html
- Oral health: www.dhsv.org.au/oral-health-resources/guides-and-resources/#Oral_health_information
- Centre for Developmental Disability Health in Australia: www.cddh.monash.org
  - Guide to the primary care of people with intellectual disability: www.cddh.monash.org/disability-health-assessment.html
  - Supporting women: guidelines for GPs and carers supporting women to manage their menstruation: www.cddh.monash.org/products-resources.html#supporting-women
  - About Alzheimer disease and Down syndrome: www.cddh.monash.org/products-resources.html#healthg
- Queensland Centre for Intellectual and Developmental Disability: www2.som.uq.edu.au/som/Research/ResearchCentres/qcidd/Pages/default.aspx
- NSW Centre for Disability Studies: www.cds.med.usyd.edu.au/
- SA Centre for Disability Health: www.sa.gov.au/subject/community+support/Disability/Corporate+and+business+information/Disability+SA/Centre+for+Disability+Health
- Queensland Down Syndrome Association: www.dsaq.org.au (includes links to other state Down syndrome associations).

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