Syndrome Specific Medical health check guide – Down’s Syndrome

Introduction

Down’s syndrome results from increased genetic material on all or part of chromosome 21, usually as a consequence of Trisomy 21, and is characterized by intellectual disability and often comorbidities involving multiple organ systems.

The survival of people with Down’s syndrome has improved dramatically in the past few decades, largely as a result of improved surgical repair of congenital heart defects. The median age at death is now the mid-50s, compared with less than 10 years of age in the 1970s. Respiratory infection and dementia are now leading causes of death in adults with Down’s syndrome.

People with Down’s syndrome generally do well with consistent schedules and can blossom in a setting of predictable routine. This also includes dietary habits and physical activity that prevent obesity.

History

As with all people with LD focus on an assessment of:

- eyesight and hearing
- feeding, bowel and bladder function
- behavioural problems and decline in skills.

The differential diagnosis for a **decline in skills** includes: depression, changes to routines, life events, hypothyroidism, sleep apnoea, hearing loss, vision loss, dementia, seizure disorder, developmental regression.

Important causes of unexplained **weight loss** include: coeliac disease and gastroesophageal reflux or dyspepsia, and swallowing problems.

Well over 50% of people with Down’s syndrome have significant **hearing** impairment, which can range from mild to profound. Sensorineural and/ or conductive loss may be present at any age. If undetected it is likely to be a significant cause of preventable secondary handicap. The main cause of conductive loss is persistent Otitis media with effusion (glue ear).
About two thirds have problems affecting their eyesight, such as refractive errors, cataract, glaucoma and keratoconus.

**Obesity** is widespread in people with Down’s syndrome (89-95%), likely due to lower activity levels and a lower metabolic rate, making exercise and energy restriction critical in maintaining a healthy weight.

One third, if not the majority of those with Down’s syndrome, have **obstructive sleep apnoea (OSA)**, which may due to a small jaw and upper airways combined with macroglossia, as well as blocked nose and most of all obesity. OSA can occur at any age and cause daytime sleepiness, behavioural change, loss of skills and other symptoms suggestive of depression or dementia. Complete an Epworth sleepiness score and refer for sleep studies. Weight loss if obese as well as CPAP mask overnight can dramatically improve the symptoms of OSA and the wellbeing of patients.

**Pneumonia**, aspiration pneumonia and flu are a common causes for admission and the second most common cause of death of people with Down’s syndrome. All adults with Down’s syndrome are eligible for Influenza and Pneumococcal immunisation.

**Swallowing difficulties (dysphagia)** can present with coughing, gagging, sighing, burping, or throat clearing during mealtimes, and cause choking with aspiration. Evaluation consists of a modified barium swallow study in conjunction with a SALT assessment.

**Gastro-oesophageal reflux** is also common in people with Down’s syndrome. Like dysphagia, it can present with weight loss, vomiting, decline in skills or behavioural changes.

**Mental health problems** affect 25-30%, mostly depression, anxiety, obsessive-compulsive tendencies, and behavioural issues. Depression is common in older adults, often triggered by bereavement or changes in their living situation. Discriminating depression from dementia can be difficult but is important, since the former is amenable to medical therapy. Symptoms more suggestive of depression include withdrawal and decreased appetite and speech. Autism is ten times more common than in the general population; it can be very difficult to treat, often requiring specialist input.

People with Down’s syndrome have an increased risk of Alzheimer’s **dementia**, with an earlier onset than in the general population. The prevalence is 10-22% in their 40s; 20-25% in their 50s; and 40-77% in those over 60 years, contributing to one third of deaths. Although donepezil and memantine are increasingly used, there is currently no good evidence demonstrating their effectiveness in this population. They appear to be beneficial for some patients, however, hypotension, bradycardia or ataxia may require their discontinuation in some.

Women with Down’s syndrome have an earlier **menopause** around 44 years on average.
Down’s syndrome is an independent risk factor for **osteoporosis**, further increased by early menopause, anti-epileptic medication and other risk factors. There is a high risk of fractures in the over 50s.

**Hypothyroidism** affects 15-37%, increasing with age. Hyperthyroidism is also more common than in the general population.

**Diabetes**: Increased prevalence of Type 1 diabetes and Type 2 Diabetes associated with obesity. The onset of type 2 diabetes is often at a younger age than the general population and can present with subtle symptoms.

**Skin conditions**: Dry skin and eczema are particularly common and are managed in the usual way.

**Cervical spine**: Atlanto-axial instability has mostly been described in children. In adults, degenerative changes and cervical spondylosis are more common, with a prevalence of 35-70%. Routine cervical spine X-ray is not recommended, but we need to be alert to signs of spinal stenosis with cord compression and assess these promptly.

Congenital heart disease is common and usually treated surgically in early childhood. In adults, consider the possibility of **acquired valve disease**, specifically mitral valve prolapse (in 45%, often with mitral regurgitation) and aortic regurgitation. It may be asymptomatic and a murmur may not always be audible. The incidence of coronary artery disease in adults with Down’s syndrome is decreased compared with the general population.

With the exception of childhood leukaemia, the incidence of **cancer** - whether hematologic or solid tumours - is also decreased in all age groups with Down’s syndrome. **Full blood counts** frequently show leukopenia, macrocytosis and mild polycythemia, which do not appear to be of clinical relevance, but B12-deficiency and hypothyroidism should be excluded and the rare possibility of adult leukaemia be borne in mind.

**Examination**

1. **Sensory**
   - Full assessment by optician/optometrist at least every 2 years.
   - If examination is difficult, refer to specialist optician or ophthalmologist for assessment.
   - Otoscopy annually - gentle examination as short auditory canals
   - Auditory assessment every 2 years has been recommended (including auditory thresholds, impedance testing).
2. Dental
- Dental Review at least annually, as periodontal disease is common.
- Look for signs of oesophageal reflux.
- Ask about swallowing problems and aspiration.

3. Respiratory
- Examine nose for blockage, the oral cavity, and lungs for lower airway disease
- Ask about daytime sleepiness and sleep apnoea. Consider Epworth sleepiness score and sleep studies.

4. Cardiovascular
- Auscultation of the heart annually.
- A single Echocardiogram should be performed in adult life.
- Echocardiogram for new murmurs and signs of cardiac failure.
- Adults with a pre-existing structural abnormality should be informed of applicable prophylactic antibiotic protocols.

5. Gastrointestinal
- Ask for signs and symptoms of Coeliac Disease annually
- Coeliac antibody test in those with suspicious symptoms or signs: disordered bowel function with loose stools or new onset constipation, abdominal distension, general unhappiness and misery, arthritis, rash suggesting dermatitis herpetiformis.
- Coeliac antibody test in those with existing thyroid disease, diabetes or anaemia.

6. Endocrine
- Thyroid function blood tests (TFTs) including thyroid antibodies every 1 or 2 years.
- Check TFTs if weight gain or loss, generally unwell, possible diagnosis of depression or dementia.
- Consider HgbA1c annually (diabetes defined as greater than 48 mmol/mol) and finger prick blood glucose.
- Ask women over 40 about hot flushes and menopausal symptoms.
- Osteoporosis screening should start begin in their 40s.
- Screen early especially in the presence of risk factors, such as poor mobility or non-weight bearing status, anti-psychotic or anti-epileptic medication, poor nutritional status, or early menopause.
7. Mental Health
   - From the age of 40, ask about symptoms of dementia, which include: loss of skills and independence, no longer remembering or managing routines, need for prompting, appearing confused, change in behaviour, also urinary and/or faecal incontinence, ataxia, seizures, impaired mobility.
   - Ask family members and/or carers about these symptoms.
   - When considering a dementia assessment and diagnosis, consider deafness, hypothyroidism, sleep apnoea and depression.

8. Orthopaedic
   - Ask about signs of spinal stenosis associated with atlanto-axial instability, which may be acute or chronic, such as: hyperreflexia, ataxia, clonus, unsteadiness, deterioration in bladder or bowel control, or quadriparesis, and consider urgent neurosurgical assessment if present.

Resources
Managing the care of adults with Down’s syndrome, Clinical Review, BMJ 2014: http://www.bmj.com/content/349/bmj.g5596

Down’s Syndrome Association: https://www.downs-syndrome.org.uk/for-professionals/health-medical/annual-health-check-information-for-gps/