Fifteen-minute consultation: The review of a child with trisomy 21 (Down’s syndrome)

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ABSTRACT

Down’s syndrome (DS) is the most common chromosomal abnormality seen in live born children and it is the most common genetic cause of intellectual disability. It is associated with abnormalities in many body systems, some of which can cause life threatening complications. This article aims to cover the important aspects to cover when seeing children with DS for their routine follow-up in the neurodevelopmental or general paediatric clinic.

INTRODUCTION

Down’s syndrome (DS) occurs in 1 in every 1000 live births and is the most frequent chromosomal abnormality seen in liveborn infants. It is caused by having an extra full or partial copy of chromosome 21 in some or all of the body’s cells. DS is associated with a wide range of health conditions affecting multiple body systems. It is important that the care of children with DS is standardised so that complications are not missed. The Down Syndrome Medical Interest Group (DSMIG) has very useful guidelines for and examination at each appointment.

GENERAL PRINCIPLES

- Children with DS should ideally be cared for by a paediatrician with expertise in DS (this could include a community paediatrician, paediatrician with a neurodisability interest or general paediatrician).
- Children should be reviewed regularly in the first year of life, then yearly after this.
- Children with DS should have access to a multidisciplinary neurodevelopmental team including speech and language therapy, physiotherapy, paediatric cardiologist (in the neonatal period), paediatric ophthalmologist and audiologist. A range of other medical specialist and allied healthcare professionals should be available if required.

- Children with DS should be screened for common associated conditions including cardiac, hearing, vision, thyroid dysfunction, growth, development and sleep disordered breathing. See figure 1 for the DSMIG recommended screening, taken from the personalised child health record DS insert. Box 1 contains a link to the DSMIG website and other useful sources of information.

- At transition, care coordination should be taken on by a clearly defined person, usually a General Practitioner.

There can be a lot to cover when seeing children with DS during their regular review. We have summarised some important points that should be covered at different age points in table 1. During the majority of regular appointments, a ‘full review’ is needed; please see box 2 for details of what to cover in the history and examination at each appointment.

COMPLICATIONS OF DS

Table 2 is a summary of some of the key complications seen in DS. In the following section, we will discuss these in more detail. It is important for anyone seeing children with DS to be aware of these.

Development

Children with DS usually have delayed early development. The Personal Child Health Record Book DS insert contains useful tables to give indications of the typical ages for developmental milestones.

Intellectual disability in children with DS ranges from mild to profound. Speech and language difficulties are usually more pronounced than other areas, with greater difficulties in expressive language compared with receptive language. Deficits in working memory are also more
Studies have shown that children with DS who are offered a ‘rich’ developmental environment can have significantly improved IQ scores. This should be offered from birth.

**Neurological**
Seizures occur in around 5%–6% of patients with DS, 40% of these patients present in infancy. The most common types of seizures are infantile spasms and then tonic clonic seizures with myoclonus. Children with DS have a relatively better prognosis and response to treatment than other causes of infantile spasms.

**Mental health and neurodevelopmental disorders**
In the past, psychiatric and neurodevelopmental disorders in children with intellectual disability were minimised or felt to be a consequence of the underlying developmental disorder. We have since come to acknowledge the high prevalence of these issues in people with DS with at least half of all children and adults experiencing a major mental health issue during their lives. The most common issues include general anxiety, obsessive-compulsive behaviours, neurodevelopmental disorders, depression and sleep related disorders. Children with DS are particularly sensitive to changes in their environment, including chronic illness or loss of a family member, death of a household pet or absence of a teacher. New behavioural symptoms can also be a manifestation of an underlying medical disorder such as hypothyroidism, sleep problems or bowel related issues.

Autism spectrum disorders (ASD) are present in around 16% of children with DS, much higher than in the general population. Compared with other children with ASD, children with DS and ASD tend to have similar difficulties with communication and repetitive and stereotyped behaviours, but better reciprocal social interaction skills and less peer related and emotional problems.

Symptoms of ADHD are also prevalent in children with DS. If inattention, impulsivity and hyperactivity are disproportionate with the mental age and result in significant impairment to academic or social development, then this diagnosis should be explored.

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**Table 1**
Suggested timetable of follow-up for children and young people with Down’s syndrome

<table>
<thead>
<tr>
<th>Age</th>
<th>MDT members</th>
<th>Key points</th>
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</table>
| 6 months                   | Paediatric and MDT| Full review*  
Discuss genetics, if parents need advice regarding recurrence for future pregnancies, then referral to clinical genetics is advised  
Encourage routine immunisations and annual influenza vaccine  
Examination should include an examination of vision |
| 1 year                     | Paediatric and MDT| Full review*  
Ensure that you enquire about sleep related upper airway obstruction  
Give information about cervical instability, cardiovascular, ENT and eye examinations  
Enquire about behavioural issues and offer advice  
Consider referral to Early Years education |
| 2 years                    | Paediatric and MDT| Full review*  
Ensure you enquire about behaviour, educational input, cervical spine instability. Examination should include signs of cervical spine instability (ensure able to flex and extend neck normally), cardiovascular, ENT and eye examinations |
| 3–5 years annual review    | Paediatric and MDT| Full review*  
Consider complex developmental problems and coexistent neurodevelopmental disorders  
Some patients without complex health issues may be appropriate for nursing review after 5 years of age |
| 6 years and yearly after this | Nurse             | Full review* including dental care, access to services, financial issues, immunisations  
In pubertal girls, enquire about menarche and menstrual management |
| School leaver review       | Nurse and Paediatrician | Full review  
Requirements for ongoing care, further education or adult placements, fertility and need for contraception  
Testicular examination—discuss if self-examination is appropriate or alternative arrangement as testicular tumours are more common in Down’s syndrome  
Prepare full medical report for GP, carers and young person  
Ensure heart auscultated  
In Scotland, discuss guardianship (must be done well before 16 years)  
Refer to adult learning disability team  
GP will be coordinator of care usually. Refer to RCGP adult Down’s syndrome specific annual check list |

*Please refer to box 2 for what to include when doing a ‘full review’.  
MDT, multidisciplinary team; RCGP, Royal College of General Practitioners.
Best practice and Fifteen-minute consultations

DOWN SYNDROME - SUGGESTED SCHEDULE OF HEALTH CHECKS

The following are suggested ages for health checks. Check at any other time if there are parental or other concerns.

<table>
<thead>
<tr>
<th>Birth - 6 weeks</th>
<th>Special checks under 2 years</th>
<th>Preschool checks</th>
<th>School age</th>
</tr>
</thead>
<tbody>
<tr>
<td>Thyroid blood tests</td>
<td>Newborn routine heel prick - blood spot test</td>
<td>Age 4-6 months and then every year from the age of 1 year or more often if clinically indicated. Venous thyroid blood test including thyroid antibodies or finger prick TSH test</td>
<td></td>
</tr>
<tr>
<td>Eye checks</td>
<td>Newborn routine eye check for cataract</td>
<td>Age 18-24 months Normal eye and vision examination including check for squint and refraction for long or short sight</td>
<td>Age 4 years: Normal eye and vision examination including check for squint, refraction and assessment of near and distant vision and visual acuity</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Visual behaviour to be monitored at every review particularly in first year</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hearing checks</td>
<td>Universal newborn hearing screen</td>
<td>Full auditory in review by 10 months including hearing test and impedance check</td>
<td>Annual auditory in review or more frequently and if indicated 2 yearly auditory in review or more frequently if recommended</td>
</tr>
<tr>
<td>Growth monitoring</td>
<td>Length, weight and head circumference should be checked frequently and plotted on Down syndrome growth charts</td>
<td>Height and weight should be checked and plotted on Down syndrome growth charts at least annually (BMi checked if concern regarding overweight)</td>
<td></td>
</tr>
<tr>
<td>Heart checks</td>
<td>By age 6 weeks, formal heart assessment including Echocardiogram</td>
<td>At all ages low threshold for reviewing heart status if signs or symptoms develop</td>
<td>From adolescence onwards as part of routine health checks then to heart for signs of acquired heart disease</td>
</tr>
<tr>
<td>Breathing checks</td>
<td>Enquire at every review for uneven breathing during sleep and poor quality sleep. Screen with an overnight pulse oximetry once in infancy and yearly until the age of 5, or at any age if there are symptoms or concerns. Low threshold for detailed sleep studies if symptoms persist, despite normal tests</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Blood checks</td>
<td>If blood film is abnormal treatment or monitoring may be required</td>
<td>Within the first 3 days of life, full blood count and blood film to check for a serious blood disorder</td>
<td></td>
</tr>
</tbody>
</table>

Detailed recommendations for Medical Surveillance Essentials for children with Down syndrome can be found at www.dsmig.org.uk

Figure 1 The suggested health checks in the PCHR DS insert as recommended by the DSMIG. BMI, body mass index; DS, Down’s syndrome; DSMIG, Down Syndrome Medical Interest Group; PCHR, The Personal Child Health Record Book.

Cardiac

DS is the most common genetic condition associated with congenital heart disease (CHD) with CHD diagnosed in approximately 50% of children.8 Atrioventricular septal defect is the most common defect followed by atrial septal defects, ventricular septal defects and tetralogy of Fallot. All neonates with DS should be seen by a cardiologist and have an echocardiogram within the first 6 weeks of life, preferably within the first 2 weeks in high risk infants.9 Children with DS can develop pulmonary hypertension and develop right sided problems secondary to airway and respiratory problems.10 In later childhood, mitral valve prolapse and aortic regurgitation can develop. The DSMIG recommends yearly cardiac auscultation to identify emerging cardiac problems.9

Box 1 Guidelines, resources and important sources of information

- DSMIG, Down Syndrome Medical Interest Group.

Box 2 Full review, what to include in history and examination

History
- Parental concerns.
- Growth and feeding.
- Gastrointestinal symptoms.
- Cardiac symptoms.
- Hearing and vision.
- Unusual or recurrent infections.
- Immunisations.
- Symptoms of cervical spine instability.
- Upper airway symptoms and sleep disordered breathing.
- Behaviour.
- Developmental milestones.
- Social support, benefits, disability living allowance (DLA), respite.
- Education.
- Dental hygiene and registration with a dentist.

Examination
- Full system examination.
- Development.
- Plotting height, weight (and head circumference) on Down’s syndrome growth chart.
- Examine for cervical spine instability (ensure the patient can fully flex and extend their neck, abnormal head posture).

**Endocrine**

Delayed growth and short stature are common features of DS. A suggested mechanism for this is growth hormone deficiency, secondary to hypothalamic dysfunction.\(^{10}\) DS-specific growth charts should be used to ensure children are following an appropriate growth trajectory. Routine use of growth hormone is not recommended, as it does not improve developmental outcomes.\(^{11}\) Up to ¾ of children with DS are overweight or obese.\(^{12}\) The underlying causes are thought to be linked to increased leptin, more sedentary behaviour, unfavourable diet and lack of exercise.\(^{12}\) Increasing weight gain usually starts after around 2 years of age.\(^{12}\)

Patients with DS are more likely to have low bone mineral density, due to factors including low physical activity, decreased muscle mass, malabsorption syndromes, antiepileptic medication, low calcium and vitamin D3. Enhanced physical activity is recommended to optimise bone health.\(^{10}\)

Vitamin D deficiency is more common due to a variety of mechanisms including poor intake of vitamin D, reduced sunlight exposure, poor absorption from coeliac disease and increased breakdown associated with anticonvulsants. No specific recommendations exist to advise supplementation in children with DS; however, there is an argument that it is beneficial to optimise growth and musculoskeletal development.\(^{10}\)

Hypothyroidism in people with DS can be congenital or acquired. In a study of 85 people aged 0–25 years with DS, 33% developed hypothyroidism in a 15-year period.\(^{13}\) Around 50% also have subclinical hypothyroidism, indicated by high thyroid stimulating hormone (TSH) levels.

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**Table 2** Summary of the complications of DS

<table>
<thead>
<tr>
<th>System</th>
<th>Complications</th>
<th>Relevant information</th>
</tr>
</thead>
<tbody>
<tr>
<td>Neurological</td>
<td>Learning disability</td>
<td>Early developmental impairment varies from mild to profound in children with DS</td>
</tr>
<tr>
<td></td>
<td>Epilepsy</td>
<td>Infantile spasms are the most common type of seizure seen</td>
</tr>
<tr>
<td></td>
<td>Neurodevelopmental disorders</td>
<td>Increased incidence of ASD/ADHD</td>
</tr>
<tr>
<td>Psychological</td>
<td>Behavioural difficulties and mental illness</td>
<td>Enquire about mood in older children</td>
</tr>
<tr>
<td>Cardiac</td>
<td>Congenital heart disease, most commonly AVSD</td>
<td>Routine echocardiogram within 6 weeks of birth</td>
</tr>
<tr>
<td></td>
<td>Mitral valve prolapse</td>
<td>Monitoring for development of cardiac failure in those with diagnosed cardiac</td>
</tr>
<tr>
<td></td>
<td>Aortic regurgitation</td>
<td>conditions</td>
</tr>
<tr>
<td>Endocrine</td>
<td>Hypo/hyperthyroidity</td>
<td>Follow recommended screening protocol</td>
</tr>
<tr>
<td></td>
<td>Propensity for obesity</td>
<td>Careful dietary control</td>
</tr>
<tr>
<td></td>
<td>Growth retardation</td>
<td>Refer to appropriate growth chart</td>
</tr>
<tr>
<td></td>
<td>Low bone mineral density</td>
<td>Ensure physical activity and adequate vitamin D intake supplementation if necessary</td>
</tr>
<tr>
<td>Reproductive</td>
<td>Infertility</td>
<td>Discussing fertility is important in patients of childbearing age</td>
</tr>
<tr>
<td>Haematological</td>
<td>Haematological malignancy</td>
<td>Acute myeloid leukaemia and acute lymphoblastic leukaemia, both usually present by</td>
</tr>
<tr>
<td></td>
<td>Thrombocytopenia</td>
<td>5 years of age</td>
</tr>
<tr>
<td>Gut</td>
<td>Feeding issues</td>
<td>May need additional support with breastfeeding</td>
</tr>
<tr>
<td></td>
<td>Congenital GI defects</td>
<td>Particularly duodenal atresia, other GI stenosis or atresia more common</td>
</tr>
<tr>
<td></td>
<td>Constipation</td>
<td>Treat as any other child</td>
</tr>
<tr>
<td></td>
<td>Coeliac disease</td>
<td>Routine screening not recommended, low threshold for testing if symptoms present</td>
</tr>
<tr>
<td></td>
<td>Hirschsprung disease</td>
<td>Consider diagnosis</td>
</tr>
<tr>
<td>Vision</td>
<td>Refractive errors</td>
<td>Follow suggested screening schedule and have a low threshold to refer if concerns</td>
</tr>
<tr>
<td></td>
<td>Nystagmus</td>
<td>occur outside of this</td>
</tr>
<tr>
<td></td>
<td>Congenital cataracts</td>
<td>Enquire about sleep disordered breathing at every review, with an oxygen</td>
</tr>
<tr>
<td></td>
<td>Glaucoma</td>
<td>saturation study if symptoms present</td>
</tr>
<tr>
<td>ENT</td>
<td>Sleep apnoea</td>
<td>Follow suggested screening protocol</td>
</tr>
<tr>
<td>Hearing</td>
<td>Sensorineural hearing loss</td>
<td>Enquire about sleep disordered breathing at every review</td>
</tr>
<tr>
<td></td>
<td>Otitis media with effusion</td>
<td>with an oxygen saturation study if symptoms present</td>
</tr>
<tr>
<td>Immune</td>
<td>Immunodeficiency</td>
<td>At risk of serious infection</td>
</tr>
<tr>
<td></td>
<td>Routine vaccinations, yearly influenza vaccine and</td>
<td>Routine vaccinations, yearly influenza vaccine and one off pneumococcal</td>
</tr>
<tr>
<td></td>
<td>one off pneumococcal polysaccharide vaccine</td>
<td>Some high-risk infants will qualify for RSV vaccine</td>
</tr>
<tr>
<td>Respiratory</td>
<td>Lower respiratory tract infections</td>
<td>Treat underlying conditions</td>
</tr>
<tr>
<td>Musculoskeletal</td>
<td>Craniovertebral instability</td>
<td>Ensure symptoms are checked for during every review</td>
</tr>
<tr>
<td>Maxillofacial</td>
<td>Delayed dentition</td>
<td>Delayed dentition is normal for children with DS</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Ensure child is registered with a dentist and parents are brushing teeth. Teeth</td>
</tr>
<tr>
<td></td>
<td></td>
<td>brushing can be more difficult due to sensory sensitivities</td>
</tr>
</tbody>
</table>

ASD, autism spectrum disorder; AVSD, Atrioventricular septal defect; DS, Down’s syndrome; GI, Gastrointestinal; RSV, Respiratory syncytial virus.
hormone (TSH) but normal T3/T4. Hyperthyroidism is also seen more frequently than in the normal population. Children need yearly screening from the age of one, more frequently before this.

Delayed puberty is a common feature of DS in both sexes. Girls can get delayed menarche and adrenarche. Boys have been reported to have ambiguous genitalia, cryptorchidism, micropenis and small testes.

Haematological
Children with DS have a 150-fold increased risk of myeloid leukaemia (known as myeloid leukaemia of DS), it is nearly always diagnosed before 5 years of age and it is preceded by transient leukaemia of Down Syndrome (TL-DS), (previously known as transient abnormal myelopoiesis) as an infant. Children with DS also have a 30 fold increased risk of acute lymphoblastic leukaemia.

Gastrointestinal
Hirschsprung disease affects around 2% of children with DS. If children fail to pass meconium after birth and have recurrent issues with constipation, vomiting and abdominal distention then consider a possibility of Hirschsprung disease or an as yet undiagnosed gastrointestinal stenosis.

Gastro-oesophageal reflux occurs frequently due to reduced lower oesophageal tone and being slower to sit up. Consider aspiration pneumonia if children present with chronic cough and recurrent pneumonia.

Coeliac disease is present in around 5% of the DS population; there are some advocates of including regular screening in the surveillance programme, using either regular coeliac serology or human leukocyte antigen (HLA) screening. This is not currently recommended by the DSMIG and testing should be guided by clinical symptoms and risk (eg, positive family history).

Visual
Refractive errors, squints, abnormalities in accommodation, nystagmus, congenital cataracts and infant glaucoma are all more common in children with DS. Examination of the child’s visual system should be taken at each review, including in younger children looking for squint, abnormalities in gaze, visual behaviour and attention. They should have a formal ocular/visual assessment by an orthoptist and ophthalmologist or optometrist at 18–24 months, but if abnormalities are evident before this, earlier referral should be made. One-third of children will have a ocular/visual defect detected at this time. Due to the high prevalence of visual disorders, there is a specific screening programme recommended (see table 2).

ENT
Obstructive sleep apnoea is seen in half to three quarters of children with DS. Predisposing anatomical abnormalities including midface hypoplasia, large tongue and mandibular hypoplasia combined with relatively large tonsils and adenoids, leads to airway obstruction. Sleep-related breathing disorders result in nocturnal hypoxaemia, which can result in pulmonary hypertension. The presence of a CHD compounds this risk. See box 3 for the symptoms of sleep disordered breathing.

Children with DS should have screening for a sleep-related breathing disorder by enquiring about the above symptoms. The DSMIG recommend yearly screening with overnight oximetry until 5 years of age, but this will be guided by local availability. Those with suggestive symptoms should have overnight oximetry and referral to ENT. Those whose results are abnormal, or there is suspicion of a false negative should have a more detailed sleep study with polysomnography including oximetry, airflow, effort and CO₂ measurement. Adenotonsillectomy may be of benefit and continuous positive airway pressure can be used to treat it.

Hearing
Over 50% of people with DS have a significant hearing impairment, with sensorineural and or conductive hearing loss being present at all ages. Otitis media with effusion is the most common cause of conductive hearing loss. All babies should have a full audiological assessment at 6–10 months of age and a regular programme of screening should be in place after this (see table 2).

Infections
Children with DS are at increased risk of infections, due to abnormalities of their immune systems and anatomical differences. Immune system abnormalities include mild to moderate T and B cell lymphopaenia, with abnormal T cell proliferation, reduced response to vaccination and defects of neutrophil chemotaxis. Infections may be serious and present atypically compared with non-DS children.
Anatomical abnormalities of the airway like laryngomalacia and tracheomalacia, impaired clearance of secretions, increased gastro-oesophageal reflux and aspiration are the mechanisms leading to increased lower respiratory tract infections. The ENT abnormalities in the above section also predispose to respiratory tract and ear infections.

Lower respiratory tract infections are a frequent reason for children with DS to be admitted to hospital, and the most common reason to be admitted to intensive care.

In children with history of recurrent, severe or atypical infections a referral to the local immunology team should be considered. They may advise longer antibiotic courses, additional vaccination and occasionally prophylactic antibiotics.

Vaccination
Normal routine vaccinations are recommended plus consideration of annual influenza vaccine and pneumococcal polysaccharide vaccine.

Musculoskeletal
Inflammatory arthritis is 18–21 times more common in people with DS,9 and the diagnosis is often missed due to having a higher pain threshold, so there should be a low index of suspicion. People with DS have lower muscle tone and hyperflexibility.10

There is an increased susceptibility of people with DS to get craniocervical instability due to hypotonia and ligamentous laxity. This occurs between atlas and axis (atlantoaxial subluxation) and also between occiput and atlas (occipito-atlantal subluxation).

Warning signs of craniocervical instability include9
► Neck pain.
► Abnormal head posture including torticollis.
► Reduced neck movement.
► Deterioration of manipulative skills.
► Deterioration of gait and frequent falls.
► Increased fatigability on walking.

If these signs are present and no alternative is found to explain the symptoms, a cervical spine x-ray in flexion and extension should be performed.9 There is no good evidence that x-rays in asymptomatic children are useful.

General approach
The DS charities advocate a ‘people first’ approach to our language around people with DS. Using certain terminology such as ‘Down’s baby/child’ or describing DS as a ‘disability/handicap or illness’ are incorrect. Instead, saying that a they are a ‘baby with Down’s syndrome’ and describing it as a ‘condition’ are factually correct and more sensitive.

Children with DS may have additional communication needs, so it is important to speak clearly and if possible use visual aids. Remember that a child’s parents know the most about their child and are a valuable source of information, particularly when they are acutely unwell, always listen to the parents!

CLINICAL BOTTOM LINE
► Children with DS can develop a range of medical disorders linked to their condition, and it is important that clinicians caring for children in the community are aware of these complications.
► Screening pathways exist to monitor for abnormalities in the following systems: cardiac, hearing, vision, thyroid dysfunction, growth, development and sleep disordered breathing.

Correction notice This article has been corrected since it first published. The provenance and peer review statement has been included.

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REFERENCES
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