Health assessment of children and young people with Down's Syndrome
Guidance for healthcare professionals

SCS Disability Pathway

[Final Version November 2020]

About this guidance

This guidance document has been developed for healthcare professionals in Specialist Community Paediatrics within NHS Greater Glasgow and Clyde.
It sets out the specific recommendations for health screening in children and young people 0-18 years with Down's Syndrome based on current Down Syndrome Medical Interest Group (www.dsmig.org.uk) guidelines.
The guidance includes information which will help clinicians identify health conditions known to be of a higher prevalence in individuals with Down's Syndrome supporting early recognition and intervention to ensure the delivery of safe and effective high quality healthcare.

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The guidance has been reviewed by the Specialist Community Paediatric Professional Advisory Group and the Clinical Governance Executive Group.

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Initial consultation advice from Down's Syndrome Scotland has been included.

Update	Change
06/01/2021	Alteration to Appendix 2 to comply with DSMIG copyright
14/01/2022	Page 3 Link to Information for parents on Health and Wellbeing updated
14/01/2022	Page 9, 21 and References Link to British Gymnastics Information Pack updated
14/01/2022	Page 25 References and Resources Link to Down Syndrome Scotland Family Support service updated
14/01/2022	Page 25 References and resources Link to T Layton updated

Specialist Children's Services Disability Pathway

Health assessment of children and young people with Down's Syndrome
Guidance for Professionals

Health assessment of children and young people with Down's Syndrome

Introduction

Down's Syndrome (Trisomy 21) is one of the most common genetic disorders; occurring in about one per 1,000 babies born each year. Down's Syndrome (DS) is caused by an extra copy of chromosome 21 inside some or all of the body's cells.
There are three types of DS: Trisomy 21-where an extra chromosome 21 appearing in each of the body's cells (94%),Translocation in which extra chromosome 21 material is attached to another chromosome (4%), and Mosaic in which only some of the cells have an extra chromosome 21 (2%).
Although there are an associated range of physical characteristics with DS each child will have their own individual physical attributes and personality. All children born with DS have some degree of learning disability and delayed development but this varies widely between individual children. A wide range of health problems are associated with DS; with a higher frequency of congenital and acquired medical conditions including congenital heart defects, audiological, visual, gastro- intestinal, haematological and thyroid issues, healthcare professionals need to have heightened awareness of the associated conditions.

This guidance supports good practice in the recommended schedule for comprehensive health assessment of children and young people who have DS. The aim is for all children and young people with DS to have equitable access to services which ensure early recognition and appropriate management of their health and developmental needs.

Information for Parents

Some families are aware that their baby is going to be born with DS due to antenatal screening, while others will only discover that their baby has DS when they receive a diagnosis after birth. Parents of children diagnosed with DS should be offered accurate and unbiased verbal and written information about the condition, the birth of their baby should be celebrated and negative perceptions and attitudes avoided-. Health and Wellbeing - Downs Syndrome Association (downs-syndrome.org.uk)
All parents of babies diagnosed with DS should be given a New Parent Information Pack; available from Down's Syndrome Scotland (DSS) as soon as possible after diagnosis and information on how to access further help and support.
https://www.downs-syndrome.org.uk/for-new-parents/new-parent-pack/
A DS insert containing additional information for parents and professionals is available for the Personal Child Health Records (**PCHR**) which includes:

- General information
- Expected developmental progress
- Possible health problems
- Suggested schedule of health checks
- Advice about immunisation, feeding and growth
- DS specific growth charts
- Sources of additional help and advice

Referral

Babies with DS should have neonatal screening undertaken prior to discharge from maternity services, as per MCN for Neonatology West of Scotland Neonatal Guideline Trisomy 21 Care Pathway.
http://www.knowledge.scot.nhs.uk/media/CLT/ResourceUploads/4094079/2c123989-050e-4995-a84f-c7993ba5e88d.pdf

Following referral (**appendix 1**) to the local Specialist Community Paediatric Team (SCPT) the nurse will be responsible for contacting the family to arrange an initial assessment. Prior to initial contact with the family, the nurse should check child's electronic records (Clinical Portal, Trakcare and EMIS) to:

- Gather relevant clinical information
- Check blood for genetic testing ,FBC , blood film, TSH obtained
- Check ECHO requested/carried out/cardiology appointment arranged
- Check Neonatal Hearing Screening completed, audiology follow up
- Check new parent information pack given
- Check Personal Child Held Record (PCHR) insert given, growth plotted
- Check follow up arrangements , professionals involved (include RSV vaccine if meet criteria)

Schedule of health and development assessments

As the child with DS grows and develops, additional needs will become evident that may require other specialists and strategies. Each child should receive a range of screening tests and health checks to ensure issues are identified and addressed as quickly as possible.

Age	Health and developmental assessment
First appointment (within first month)	**Initial Contact** SCP nurse should liaise with family health visitor to discuss involvement of SCPT and to arrange an initial joint appointment, with parent/carer to introduce the service either via a home visit or clinic appointment. The first appointment is an opportunity to: • Discuss the role of SCPT in the provision of support and ongoing health /developmental monitoring and review • Respond to any questions/concerns the family may have and /or signpost to further information • Offer information on family support services, local groups • Ensure family have DS insert for personal child health record • Complete a comprehensive assessment, utilising knowledge of health conditions that may be associated with Down's syndrome , offer advice and support as indicated and respond to any 'red flag' symptoms • Down Syndrome Scotland AHP postcards can be introduced to help support this assessment • Record Read Code on EMIS (if not already applied) PJ0/PJ0z • Complete SCP Nursing Assessment Report , copy to Parent, EMIS, GP , Clinical Portal • Arrange further review (this will be dependent on assessed needs)

	• Progress referral via Care Co-ordination Meeting –this may result in joint appointment via Joint Assessment Clinic (JAC) ❖ There may occasionally be a failure to diagnose cardiac conditions prior to discharge from maternity services. Babies who present with symptoms of poor feeding, breathlessness, poor colour and are failing to thrive should have immediate referral for clinical assessment. ❖ Some babies may have a prolonged hospital stay due to their health condition it is of value to for the family to be introduced to an SCP nurse and have information on the service prior to their discharge. ❖ CCN-home team may have greater input initially if baby has complex health needs, liaise and agree roles and responsibilities to ensure assessed needs are being met. Nurse should schedule a routine 3 month review into their EMIS diary or add a Disability nursing CRT
Three Months	Check previous assessment report and electronic records for new information prior to appointment date. **SCP Nurse Review** ▪ Explore if parent/carer have any concerns, AHP pathway postcards may be helpful ▪ Check cardiac status; any symptoms e.g. poor feeding, breathlessness, poor colour, failure to thrive, if present refer immediately for clinical assessment. Check cardiac assessment has been carried out by Paediatric Cardiologist and ECHO report is available, contact RHC to check progress/refer to Paediatric Cardiology if still awaited ▪ Check feeding pattern, particularly ask about Gastro Oesophageal Reflux symptoms and contact GP to initiate treatment where indicated ▪ Check bowel pattern and whether there are any concerns ▪ Check if any hearing concerns ▪ Check if any vision concerns e.g. poor fixing and following ,squint, nystagmus ▪ Any recurrent infections (respiratory, ear infections) or unusual infections (any hospital admissions) ▪ Explore sleep pattern, check for signs of sleep related breathing disorder (**appendix 4**) ▪ Check immunisation status – support uptake of routine primary immunisation ▪ Developmental progress ▪ Discuss with family what is important to them to find out if referral to other support services would be beneficial **Observation** ▪ General appearance, colour, tone, response, check/ record up to date weight, length and head circumference and check /record on DS growth chart in PCHR ▪ Eye examination to look for squint, nystagmus and visual behaviour (e.g. fixing and following) **Follow-up** ▪ Any immediate concerns should be discussed with Paediatrician ▪ Discuss Disability Living Allowance (DLA) if family agree start process as application accepted from 6 months of age ▪ Discuss available support - Down's Syndrome Scotland , carers' centres ▪ Check that new born hearing screen results are reported and follow-up organised

	▪ Discuss at CCM /refer to SLT if there are concerns with feeding ▪ Discuss at CCM /refer to paediatrician if any health concerns **JAC** ▪ Discuss at CCM/ refer to physiotherapy if any concerns with motor development ▪ Complete SCP Nursing Assessment Report copy to Parent, EMIS, GP , Clinical Portal, plot growth on I-Grow DS chart ▪ Finger prick TSH will initially be carried out by SCP nursing team between 4-6 months ▪ Ensure added to EMIS schedule for ongoing annual TSH finger prick measurement (Guthrie-consent required for thyroid screen database). ❖ Frequency of nursing review will depend on child and family's assessed health and support needs, involving liaison with family health visitor, SCPT (General/Specialist Paediatricians) and other nursing support (CCN-Home).
Six Months	**Paediatrician Assessment /JAC** ▪ Check full history ▪ Explore if parent/carer have any concerns ▪ Check cardiac status. Cardiac assessment should have been carried out by Paediatric Cardiologist and ECHO report available ,contact RHC to check progress/refer to Paediatric Cardiology if still awaited ▪ Check feeding pattern ▪ Check bowel pattern and whether there are any concerns ▪ Check if any hearing concerns ▪ Check if any vision concerns e.g. poor fixing and following ,squint, nystagmus, ▪ Any recurrent infections (respiratory ,ear infections) or unusual infections (any hospital admissions) ▪ Explore sleep pattern , check for signs of sleep related breathing disorders ▪ Discuss with family what is important to them ▪ Discuss chromosome result and chance of recurrence ▪ Check immunisation status – support uptake of routine primary immunisation and advise on annual influenza vaccine **Examination** ▪ Full physical examination ▪ Eye examination to look for cataracts, squint, nystagmus and visual behaviour ▪ Plot growth on I-Grow DS chart in records and PCHR ▪ Developmental assessment **Follow up** ▪ Genetics referral if indicate ▪ If not present request consultancy appt from SLT* *The national AHP pathway proposed a multi-disciplinary appointment at around 1 year where the child and family meets with relevant AHPs and the Paediatrician. This approach has had positive results in Lothian.
One Year	**Paediatrician Review** ▪ Discuss any parent/carer concerns ▪ Assess developmental progress ▪ General health, including full history of any respiratory, cardiac, or bowel symptoms (have a low threshold for investigation of coeliac disease **(appendix 6)**

	▪ Any unusual or recurrent infections ▪ Signs of sleep related breathing disorders- If clinical symptoms are present refer to ENT ▪ Behaviour ▪ Link with involved clinicians ▪ DLA and other benefits ▪ Highlight DS are at increased risk of developing C1-C2 atlantoaxial cervical spine instability , provide reassurance that it is uncommon, but of importance to be aware to seek further advise if concerned , offer information leaflet / signpost to further information ▪ Check immunisation status – support uptake of routine primary immunisation and advise on annual influenza vaccine **Examination:** Full clinical examination with particular attention to the following: ▪ Growth - Plot on I-Grow DS chart and PCHR ▪ Cardiovascular ▪ Neurological - Look for any signs of cervical spine instability or cord compression ▪ ENT - Signs of middle ear disease or upper airway obstruction ▪ Eyes - Squint, cataract, nystagmus or blepharitis. Nasolacrimal duct obstruction sometimes needs referral **Investigations:** ▪ Audiology –RHC has a specialist joint ENT/audiology surveillance clinic which is held monthly reviewing children from across Glasgow and Clyde for their annual screening audiogram until they are school age (there are some locality variations e.g.IRH) then the follow-up is transferred to community audiology N.B Some children are not within NHS GGC, need follow up in Lanarkshire /Ayrshire ▪ Check Immune function only if concerns on reported increased history of infections (**appendix 5**) ▪ Thyroid function tests (T4, TSH and thyroid autoantibodies). Finger- prick TSH will be carried out annually by SCP nursing team (**appendix 8**) **Follow up** ▪ Referral to Early Years Assessment Service (EYAS Glasgow only) or local early years education service, if not already notified. EYAS will contact referrer when the child is about 18 months for update before contacting the parent/carer to introduce education services
Two Years	**Paediatrician Review** ▪ Discuss any parent/carer concern ▪ Assess developmental progress ▪ General health, including full history of any respiratory, cardiac, or bowel symptoms (have a low threshold for investigation of coeliac disease) ▪ Any unusual or recurrent infections ▪ Signs of sleep related breathing disorders- (**appendix 4**) ▪ Behaviour ▪ Therapy input ▪ DLA and other benefits ▪ Cervical spine instability ▪ Discuss immunisation – advise re: annual influenza vaccine

	Examination: Full clinical examination with particular attention to the following: ▪ Growth - Plot on the I-grow DS chart and PCHR ▪ Cardiovascular ▪ Neurological - Look for any signs of cervical spine instability or cord compression, full active flexion and extension of the neck should be possible ▪ ENT - Signs of middle ear disease or upper airway obstruction ▪ Eyes - Squint, cataract, nystagmus or blepharitis. Nasolacrimal duct obstruction sometimes needs referral to opthalmology ▪ Developmental progress **Investigations:** ▪ Audiological / ENT assessment ▪ Thyroid function tests (T4, TSH and thyroid autoantibodies). Finger- prick TSH will be carried out annually by SCP nursing team (**appendix 8**) ▪ Check Immune function only if concerns on reported increased history of infections (**appendix 5**) ▪ Ensure child known to early years education service ▪ Hip screening – for any child not yet weight bearing request Hip x-ray and liaise with physiotherapy (increased risk of hip subluxation and dislocation).
Three - Five Years	**Paediatrician Review** These are as per 2 year review ❖ Some children will be seen more frequently than these recommendations according to their individual health needs. ❖ Some children may be attending reviews by General/Specialist Paediatricians depending on their health needs, consideration should then be given to who carries out scheduled assessment and review to avoid duplication. (parental choice, negotiated between paediatricians).
Transfer to school Five Years	Children with DS will be referred to EYAS / early years education with the majority of children able to attend a mainstream educational placement with a supportive learning plan Some following educational assessment will have placement within an additional support for learning school. **Review** ▪ At this stage, there should be discussion between the paediatrician and nurse and a decision taken on whether ongoing review will be by the paediatrician, the nurse or will require a joint review ❖ This will depend on the complexity of the child's health needs, where uncomplicated or where known health issues clearly being addressed elsewhere, nursing review will be more likely.

<table>
<tr>
<td><u>Six Years Onwards</u></td>
<td>
<u>Paediatrician/ Nursing/Joint Annual Reviews</u>

• Explore if parent/carer/ CYP have any concerns
• Assess Developmental progress
• General health, focussing on symptoms of diseases which are more common in CYP with DS (low threshold for investigation as atypical presentation of coeliac disease)
• Any recurrent infections (respiratory ,ear infections) or unusual infections (any hospital admissions)
• Any reported hearing/visual problems
• Explore sleep pattern , check for signs of sleep related breathing disorders
• Behaviour
• Dental Health
• Check immunisation status – support uptake of annual flu vaccine and routine schedule of vaccinations
• Menarche /Menstruation/ Puberty
• Therapy and educational input
• Check benefits e.g. mobility, bus pass
• Check access to other services e.g. sport, clubs

Observations:
▪ Growth - Plot on I-Grow DS chart (there is a BMI look up included on the chart giving the BMI centile)
▪ Neurological - Look for any signs of cervical spine instability or cord compression. Full active flexion and extension of the neck should be possible , any mobility difficulties

Follow Up:
▪ Audiology to be carried out yearly, this should happen routinely through audiology department. Check family have attended and results are available
▪ 2 yearly ophthalmological assessment
Some children with recognised visual problems will have schedule of review arranged via ophthalmology if no visual problems normal vision screening with optician (In ASL schools should be seen annually within school)
▪ Annual TSH screening
▪ Referral or discussion within/ out with SCPT if any concerns

❖ Scottish Transitions Forum https://scottishtransitions.org.uk/ recommends transition planning from age 14 and that health, education and other support services work together.
❖ Children with DS should not be automatically excluded from sports such as gymnastics but the requirements of national governing bodies which include a clinical screening protocol should be followed).file (british-gymnastics.org)
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<tr>
<td><u>School Leaver (Age 16-18)</u></td>
<td>
<u>As per Annual Review</u>

▪ Review and summary of health to date
▪ Requirements for ongoing care
▪ Further education and adult placement
▪ Preparation of medical report for GP, carers and the young person
▪ Testicular examination – discuss if self-examination is appropriate or alternative arrangement as testicular tumours are more common in Down Syndrome
▪ Fertility and need for contraception
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	Examination: ▪ Careful auscultation of heart (increased incidence of mitral valve prolapse and of aortic regurgitation) if has had AVSD repair cardiology follow up may be ongoing **Referrals:** ▪ Transition to Adult Learning Disability Team, where indicated ▪ GP for ongoing health surveillance: https://www.downs-syndrome.org.uk/for-professionals/health-medical/annual-health-check-information-for-gps/ www.**rcgp**.org.uk has an adult DS specific annual check list **Investigations:** ▪ Audiology ▪ Vision ▪ Thyroid function ▪ Echocardiogram (recommended) ❖ Consider most efficient method to capture relevant information which will support effective transition; joint appointment (paediatrician and nurse), joint assessment clinic appointment, multi-agency assessment.
	❖ Regular measurements of height and weight plotted on the DS specific charts are likely to be sensitive early indicators of the medical problems that can occur in this population.

Appendix 1

Down's Syndrome Neonatal Discharge Form

Patient Name	**DOB**	**CHI**	**Consultant**

Clinical Findings	**Date**	**Organised/ Performed by**	**Results/Details**	**Follow-up**
Cardiac Examination				
Age at which meconium passed				
Other major abnormalities				
Blood tests				
QF-PCR & Karyotype				
FBC and Blood film				
Thyroid Function (newborn screening)				
Investigations				
Hearing Screen				
ECHO				
RSV vaccination(if applicable)				
Referral				
GP by phone				
HV by phone				
Specialist Children's services Nurse by phone				
Paediatric Physiotherapist				
Consultant community Paediatrician letter at discharge				
Cardiology				
Other				

Appendix 2

Down Syndrome suggested schedule of health checks

Additional information for both parents and professionals on the suggested schedule of health checks by age for children and young people with Down Syndrome is included with the Personal Child Health Records (**PCHR**) inserts produced by the Down Syndrome Medical Interest Group (DSMIG 2020)

The PCHR insert can be accessed at: https://www.dsmig.org.uk/information-resources/personal-child-health-record-pchr/

Appendix 3

Complications associated with Down's Syndrome

Complication	Likelihood
Congenital heart disease About 50% of babies have congenital heart disease Atrioventricular septal defect is the most common form of CHD in DS. Evaluation by a paediatric cardiologist, including an echocardiogram, is recommended in all newborns with DS (even in the absence of a	High
Duodenal atresia Infants with DS may be born with a GI defect, including duodenal or anal stenosis and duodenal or anal atresia.	High
Hirschsprung's disease May be indicated by treatment of chronic constipation not responding to diet change or stool softeners.	Low
Short stature Children with DS may have shorter arms and legs than age-matched peers without DS. Their growth rate and height may be decreased. Likelihood for obesity may be greater, possibly caused by inactivity and low muscle tone.	High
Acute myeloid leukaemia Children with DS have a higher risk of acute myeloid leukaemia (AML) than the general population, with a median onset of 2 years compared with 8 years in the general population. Most cases of AML in children with DS occur between the ages of 1 and 5 years, with an average age of 2 years. After age 3, acute lymphocytic leukaemia (ALL) occurs more frequently, with most cases occurring by age 6. Children with DS are more responsive to chemotherapy, and disease resistance and recurrence are uncommon.	Medium
Myeloproliferative disorder Develops in about 10% of infants with DS. Typically resolves spontaneously within 3 months. Despite most infants with transient leukaemia recovering on their own, 20% to 30% of these cases will later be diagnosed with a more serious disease, acute megakaryocytic leukaemia (AMLK), which is a subtype of acute myeloid leukaemia (AML).	Low
Epilepsy Rate is about 8%. Types of seizure disorder vary: 47% partial seizures, 32% infantile spasm, 21% to 69% tonic-clonic seizures.	Low
Intellectual disability Many children with DS have global developmental delay. Cognitive abilities vary greatly, with challenges in the expressive language domain compared with receptive language. IQ can range from mild to moderate intellectual disability, between 40 and 72. Chronic ear disease and hearing loss can further affect language development Ongoing developmental assessment and psycho-educational evaluation is recommended.	High

Obstructive sleep apnoea An estimated 30% to 60% of children with DS have obstructive sleep apnoea. Predisposing factors include small upper airway, mid-face and mandibular hypoplasia, large adenoids, protruding tongue, hypotonia, and obesity, they are more susceptible to respiratory tract infections, reactive airway disease, and acute and chronic airway obstructions. Referral for a sleep study or polysomnogram for all children with DS is recommended between 3 and 4 years of age.	High
Hearing loss The higher frequency of (mostly conductive) hearing loss in children with DS is secondary to their anatomical anomalies, including mid-face hypoplasia, easily collapsible eustachian tube, stenotic ear canals, and small external canal. Hearing screen is required in all newborns in general. Hearing evaluations should be repeated at 6 months, 12 months, and once a year thereafter.	High
Respiratory infections The immune system in children with DS develops more slowly, predisposing the children to a higher incidence of upper respiratory tract infections, chronic middle ear effusions, and chronic otitis media.	High
Thyroid disorders Hypothyroidism is particularly common in DS (16% to 20%) and may be detected by the newborn screening or during yearly routine screening. Hyperthyroidism occurs at a lower frequency than hypothyroidism All new-borns are required to have a new-born screening, including thyroid testing. If the screen is normal, a follow-up for thyroid function (T4 and TSH) at 6 months and then yearly is recommended.	High
Obesity As a result of low resting metabolic rates, about half of all girls with DS are overweight by the third year of life and half of all boys with DS are overweight by early childhood (3-8 years of age). Refer to a physician with expertise in paediatric sleep for any child with signs or symptoms of obstructive sleep apnoea or abnormal sleep study result. Discuss obesity as a risk factor for sleep apnoea. Consistent exercise and a balanced diet are recommended. Routinely screening for obesity and sleep apnoea is important.	Medium
Visual abnormalities Congenital cataracts are seen in 4% of children with DS, nasolacrimal duct may be obstructed as a complication of mid-face hypoplasia but it improves with age. Other abnormalities include strabismus (23% to 44%), accommodative esotropia, myopia, hyperopia, and blepharitis	Medium
Dental anomalies Include delayed primary and secondary dentition, missing teeth, small or misshapen teeth, or severe crowding as a result of small oral cavity. Average age of eruption of the first tooth is between 12 and 20 months, as compared with 6 months in typically developing children.	Medium
Coeliac disease Occurs in 7% to 16% and may present with diarrhoea, bloating, or growth failure. About one third of individuals with DS who have coeliac disease may not manifest GI signs and symptoms	Low
Skin disorders Hair may be fine and hypo pigmented, alopecia areata is seen in 6-9% of	Medium

patients with DS. Seborrhoeic dermatitis may occur in 30%. Skin may be dry and rough, predisposing to itching, eczema, and infection	
Delayed motor developmental skills Children with DS, like other children, develop their skills according to their own timetable, some will be slower than others and some will be faster. In general though, specific physical and medical problems can delay the development of gross motor skills in a child with DS.	High
Behavioural problems Patients with behavioural problems, including inattention, hyperactivity, or withdrawal, should receive a behavioural assessment and tailored behavioural intervention.	Medium
Autism spectrum disorder Present in 1% to 13% of children with DS, although most studies are limited by sample size and ascertainment bias Screening and clinical observation of symptoms suggestive of autism spectrum disorder is recommended.	Low
Atlantoaxial instability About 15% of patients have lax atlanto-axial joint. This may result in spinal cord compression in 1% to 2% of cases. In terms of atlanto-axial subluxation, symptoms of myelopathy should be sought, including neck pain, changes in head positioning or torticollis, spasticity or change in tone, radiculopathy, incontinence, changes to gait, or hyper-reflexia	Low
Joint dislocations Musculoskeletal disorders, such as ligamentous laxity and low muscle tone, may contribute to knee and hip problems, and increase susceptibility to subluxation and dislocations.	Low
***Depression in adulthood** Depression in adults with DS is estimated to be about 11% and may be attributed to several factors: challenges in transitioning from childhood to adulthood, increased awareness that they are different and may change, loss of personal relationship with their peers, obesity leading to a sedentary lifestyle, poor diet, limited exercise and/or social activity, and medical complications such as hypothyroidism and obstructive sleep	
***Dementia/Alzheimer's disease** Neuropathological changes of Alzheimer's disease are seen between 35 and 45 years. Despite early changes, the average age of onset of clinical dementia is between 51 and 54 years, and not all patients with DS will exhibit signs of dementia	
*For information /awareness	

Adapted from BMJ Best Practice Down's Syndrome https://bestpractice.bmj.com/topics/en-gb/700

Appendix 4

Sleep-related upper airway obstruction (SRUAO) or Sleep related breathing disorder (SRBD)

Studies have shown that children with DS are more pre-disposed to sleep related breathing disorder due to having a naturally lower muscle tone and narrower airway. SRBD can affect cognitive abilities, behaviour and growth, however as these are commonly associated with DS the adverse effects of SRBD may not be recognised.
It is extremely important that clinicians check for a history of SRBD at all routine appointments.

The RCPCH Working Party on Sleep Physiology and Respiratory Control Disorders in Childhood reported that:

- Children with DS are at high risk of SRBD and nocturnal hypoxaemia, and the high incidence of congenital heart disease in these children makes the development of pulmonary hypertension a significant risk.
- SRBD may be difficult to identify on symptoms in this group.
- Adenotonsillectomy may have a lower rate of success, but is still indicated.
- Other interventions including CPAP are effective but may be difficult to institute.

Symptoms to consider include:

- **Snoring ***
- Sleep disturbance
- Mouth breathing and halitosis
- **Restless sleep***
- Chronic rhinorrhoea
- Subcostal and sternal recession
- Odd sleep positions, such as hanging over the bed or sleeping upright with head extended to optimise the upper airway
- Swallowing difficulties
- Recurrent upper respiratory tract infections
- Nausea and vomiting
- **Daytime sleepiness ***
- Persistent or secondary enuresis
- Nocturnal sweating
- Cyanosis
- **Apnoea ***

***Interpretation of a history of sleep difficulties can be difficult but restless sleep, snoring and daytime sleepiness may be symptoms that point to SBRD. Observed sleep apnoea is highly suggestive of a significant problem (Charleton et al 2014)

Local Recommendations

Clinicians should use specific questioning at every review to identify any symptoms of SRBD. There should be a low threshold for further investigation.
For children and young people who have any of the presenting symptoms suggestive of SRBD, further discussion and decision on actions should be agreed with relevant members of local SCPT.

RCPCH recommended the following:

- All children with DS should be offered screening for SRBD, using at least oximetry; suggested screening ages are at least once in infancy then annually until age 3-5 years.
- Children with DS with abnormalities on screening for SRBD, or where there is a clinical suspicion of a false negative screening test, should have polysomnography, including oximetry, airflow, effort and CO2 measurement. Video should be included if possible.
- If significant SRBD with hypoxia is present in children with DS, then appropriate treatment should be offered.
- Further research is needed on the benefits and risks of screening for SRBD and DS.
- It is acknowledged in the report that “there is no evidence about how long screening should continue in these children”. We have arbitrarily taken 3-5 years as including the period of highest risk of OSA. If screening tests are negative up to this age it would seem reasonable not to undertake further tests subsequently unless there are suggestive symptoms.

Appendix 5

Infection

Children with DS are more susceptible to infection, particularly respiratory tract infections. They are more likely to require hospital admission due to the severity of their illness and require a longer period of hospital stay.

The reasons for this are multi-factorial but include:

- Structural abnormalities
- Gastro-oesophageal reflux/aspiration
- Other medical conditions
- Immune function

Research is ongoing to examine past and current practice for the management of respiratory tract infections in people with DS to support further development of recommendations on the most effective evidence based treatments.

General advice

Promotion of good general health and nutrition, a healthy environment, good basic hygiene measures and avoiding contact with others when they are unwell are practical measures than can be advised to help reduce the risk of infection.
Inform carers to seek advice early from health care services with any persistent health issues to allow prompt assessment and treatment of infection where indicated.

Immunisation

It is important that children with DS receive the full schedule of immunisation.
Seasonal flu vaccination is also recommended.
Children who have congenital heart disease, respiratory problems or are premature may also meet the criteria for RSV prophylaxis.
Pneumococcal Polysaccharide Vaccine (PPV) in addition to the usual Pneumococcal Conjugate Vaccine, may be recommended particularly in those with cardiac and respiratory disorders.

Local recommendations

Where parent/carer reports a history of recurrent infections, a prolonged illness or a serious infection which has resulted in hospital admission at a review appointment, the clinician should check if immunological investigations have been ordered /carried out.
For children and young people who have a history which is suggestive of a poor immune response who have not previously had immunological investigations carried out, further discussion and decision on actions should be agreed with relevant members of local SCPT.

Nottingham Down's Syndrome service routinely carry out immunological investigations following the child's first birthday (see p18).

Guidelines for prevention, investigation and management of infection

At 12 months

Check immunoglobulins, functional antibodies and Prevenar antibodies ensuring timing of blood test is at least 1 month after completion of 12 month routine Hib/MenC/PCV booster immunisations. In addition send EDTA sample for lymphocyte subsets (ensure Down's syndrome is written in the clinical details so that the correct subset panel is performed).

At any review if above assessment not done at 12 months or history of infection as follows:

- 4 or more infections over 6 month period requiring visit to GP or ill health for >5 days
- A hospital admission for sepsis

Check immunoglobulins, functional antibodies and Prevenar antibodies (wait until at least 4 weeks post completion of primary immunisations for the latter 2). In addition send EDTA sample for lymphocyte subsets (ensure Down syndrome is written in the clinical details so that the correct subset panel is performed).

Prophylactic antibiotics

If significant infection history, it may still be appropriate to commence prophylactic antibiotics even if tests normal. These should be considered in children with frequent infections either to be used throughout the year or just September to April.

Options -Azithromycin 10mg/kg od 3 days per week
Amoxicillin, standard dose tds
Co- trimoxazole – single daily dose

The need for prophylactic antibiotics should be reviewed at each visit. If stopping, suggest choose to do this in late spring/early summer.

At any review if history of unusual infection

Check immunoglobulins, functional antibodies, Prevenar antibodies and lymphocyte subsets and consider further discussion.

General management

- Double the length of usual antibiotic course
- Maximise immunity by ensuring appropriate vaccines have been given (as per childhood schedule and any additional vaccines – see below).
- Yearly influenzae vaccine for child and household members
- Pneumococcal vaccines
- If child is in additional 'at risk group' for pneumococcal infection i.e. because of congenital heart disease, risk of aspiration or neuromuscular complications then to also be offered additional pneumococcal immunisation:
    - Age 2-5 yrs: single dose of Pneumovax II(at least 2 months after final dose PCV)
    - Age > 5 years: single dose of Pneumovax II (*)

If pneumococcal antibodies are low or concern regarding immunodeficiency discuss further action with relevant specialist.

* Pneumovax II should not be repeated within 5 years. If pneumococcal antibodies low despite recent Pneumovax II, please discuss with relevant specialist.

Nottingham Down's Syndrome Guideline (2017)

Appendix 6

Coeliac disease

Coeliac disease occurs more commonly in people with DS with a prevalence of 4-17% depending on age and country of origin. Clinical diagnosis is difficult because of overlap with features commonly seen in DS. In the UK routine screening has not been adopted, however it is recommended that clinicians should have a high awareness of coeliac disease as a possible problem in children and adults with DS and a low thresh-hold for testing if there is a clinical suspicion.

Coeliac disease may present with a wide range of symptoms. Serological testing should be considered for any of the following:

- Persistent unexplained abdominal or gastrointestinal symptoms
- Recurrent abdominal pain
- Abdominal distension/ flatulence
- Reflux and vomiting,
- Faltering growth
- Unexpected weight loss
- Irritability
- Prolonged fatigue
- Severe or persistent mouth ulcers
- Unexplained iron, vitamin B12 or folate deficiency
- Type 1 diabetes at diagnosis
- Autoimmune thyroid disease at diagnosis
- Irritable bowel syndrome(adults)
- First degree relatives of people with coeliac disease

Local Recommendations

Clinicians should use specific questioning at every review to identify any symptoms of coeliac disease. Referral for coeliac screening will be based on overall assessment and findings alongside the family profile. There should be a low threshold for further investigation.
For children and young people who have any of the presenting symptoms suggestive of coeliac disease further discussion and decision on appropriate actions should be agreed with relevant members of local SCPT.

Appendix 7

Cervical Spine Instability (Atlanto-Axial Instability)

It is imperative that any person with Down syndrome presenting with new signs or symptoms which may be indicative of cervical spine instability (CSI) or myelopathy be examined and investigated urgently.
There should be a low threshold for suspicion as there is good evidence that early warning signs are often missed and diagnosis of CSI made late with otherwise preventable catastrophic consequences.

It is essential that parents, relatives, carers and all healthcare professionals are made aware of these clinical signs and symptoms.

Warning Signs

- Neck pain
- Abnormal head posture,
- Torticollis, (Wry neck)
- Reduced neck movements
- Deterioration of gait and/or frequent falls
- Increasing fatigability on walking
- Deterioration of manipulative skills

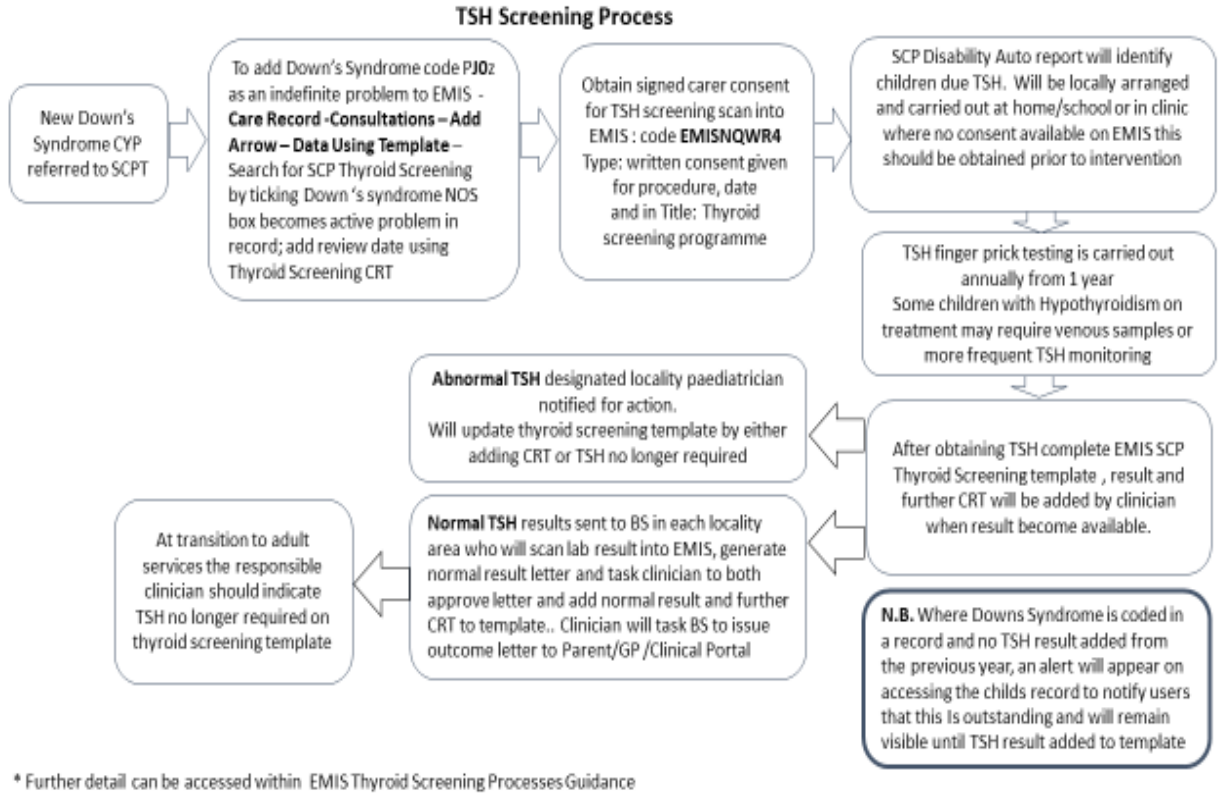
Sport

Asymptomatic individuals with Down syndrome should not be barred from normal sporting activities because there is no evidence that participation in sports increases the risk of cervical spine injury any more than for the general population. For specialised sport, such as gymnastics, children with DS should not be automatically excluded but the requirements of national governing bodies which include a clinical screening protocol should be followed:- file (british-gymnastics.org)

Adapted from DSMIG further information can be found at: **https://www.dsmig.org.uk/wp-content/uploads/2015/09/CSI-revision-final-2012.pdf**

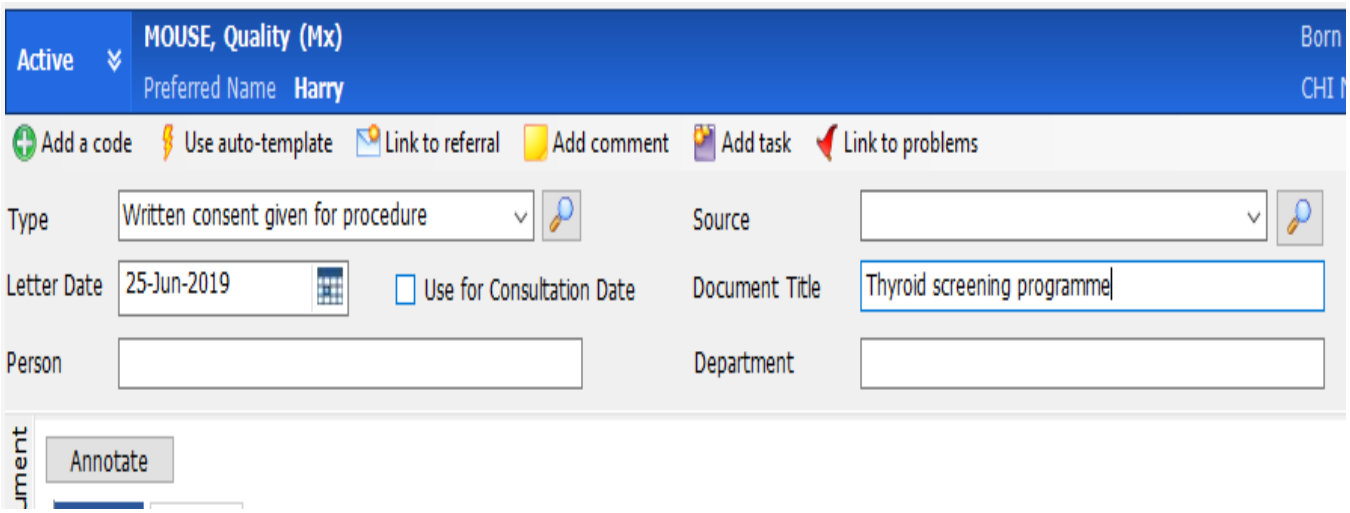
Appendix 8

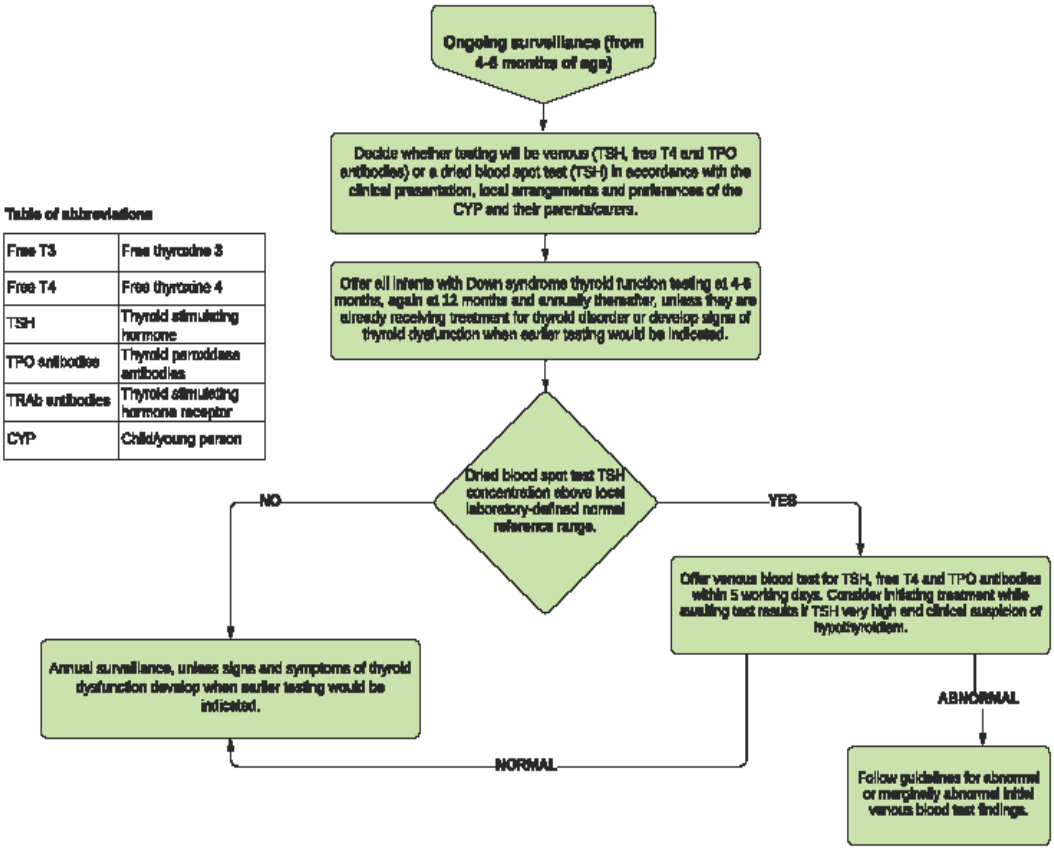
Flow chart TSH Screening



Adding consent form for TSH Screening Programme

Scan code **EMISNQWR4**





Thyroid disorder in children and young people with Down syndrome; surveillance and when to initiate treatment

Flow Chart Guidance Thyroid Disorder in Children and Young People with Down Syndrome (2020) Further Flow Charts can be accessed at:

https://www.dsmig.org.uk/information-resources/by-topic/thyroid-disorder/

Appendix 9

Health and Wellbeing Surveillance Checklist for Children and Young People with Down's Syndrome

Age	Birth	6/52	3/12	6/12	1	2	3	4	5	6	7	8	9	10	11		12	13	14	15	16	17	18
Genetic result FBC ,blood film																							
Guthrie result																							
Neonatal Cardiac assessment ECHO																							
Neonatal Hearing Screening																							
Red book insert																							
Info DS family support /DSA																							
Thyroid screening TSH																							
Growth plotted																							
Hearing review																							
Visual review																							
Health (infections,																							
Sleep disordered breathing																							
Cervical spine instability																							
Gastro-intestinal (reflux ,coeliac)																							
Immunisation (flu vaccine)																							
Early years education																							
Sexual Health & Puberty																							
Transition planning																							

References/Resources

Association of Paediatric Chartered Physiotherapists
https://apcp.csp.org.uk/documents/parent-leaflet-down-syndrome-updated-2016

BMJ Best Practice Down's Syndrome (2018)
https://bestpractice.bmj.com/topics/en-gb/700

British Gymnastics **Atlanto-Axial Instability** Information pack 2018
Persons with Down's Syndrome Atlanto-Axial screening information sheet
file (british-gymnastics.org)

Charleton, M, P, Dennis. J, Marder. E (2014) *Medical Management of children with Down's Syndrome* Paediatrics and Child Health Volume 24, Issue 8, August 2014, Pages 362-369
https://doi.org/10.1016/j.paed.2013.12.004

Coeliac disease: recognition, assessment and management (September 2015)
NICE Guideline (20) https://www.nice.org.uk/guidance/ng20/resources

Corker. McCall. Marder. E. (7th March 2017) *The aetiology of Down's syndrome, the screening process and management of comorbidities in childhood and adulthood.*
Clinical review: Down's syndrome
https://www.gponline.com/clinical-review-downs-syndrome/genetics/article/1103527

Down Syndrome Medical Interest Group (www.dsmig.org.uk) guidelines for basic medical surveillance in children with Down's Syndrome.
https://www.dsmig.org.uk/information-resources/guidance-for-essential-medical-surveillance/

Down's Syndrome Scotland Family Support Service
Family Support Team | Downs Syndrome Scotland (dsscotland.org.uk)

NHS HULL Down's Syndrome Pathway (2018)
http://www.hullccg.nhs.uk/downssyndromehull/

Nottingham Down's Syndrome Guideline (2017)
https://www.dsmig.org.uk/wp-content/uploads/2017/10/Nottingham-Down-syndrome-guidelines-2017.pdf

Scottish Transition Forum
https://scottishtransitions.org.uk/

The DSA Health Series is an information resource for parents and carer's which covers a range of health and medical conditions.
https://www.downs-syndrome.org.uk/for-families-and-carers/health-and-well-being/health-series

Thomas L. Layton, Ph.D. (2004) *Developmental Scale for Children with Down's Syndrome*
(Microsoft Word - Developmental Scale for Children with Down Syndrome comple\205) (dsacc.org)