Shropshire Community Health NHS
NHS Trust

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Local Ref (optional)	
Main points the document covers	Clinical guidance on how to medically manage children and young person with Down Syndrome
Who is the document aimed at?	Community Paediatricians, Community Children's Nurses, School Nurses, Health Visitors
Author	Dr Preetha Raveendran Community Paediatrician Narinder Kular, Nurse Consultant Paediatrics Complex Care
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Sub Category	
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Required by CQC	No
Required by NHSLA	
Other	

Amendments History		
1	V1 April 2012	
2	V2 February 2014	Discussed at Community Pediatric Medical Team Meeting
3	V3 November 2017	Discussion Community Medical Team Meeting Edited
4	V4 Sept 2021	Discussion Community Medical team Meeting Edited with formulation of age appropriate screening and recently updated coeliac guidance.

Table of Contents

1. Introduction

Down syndrome (DS), or trisomy 21, is the most common genetic form of intellectual disability, occurring in 1 in 691 births. In addition to having characteristic physical features, short stature, and hypotonia, individuals with DS have a distinctive cognitive and behavioral profile. They also have a higher frequency of associated medical conditions, including congenital heart defects, and gastrointestinal, thyroid, haematological, audiological, and visual abnormalities.

The average IQ of young adults with Down is around 50 compared to normal children with an IQ of 100. A large proportion of individuals with Down syndrome have a severe degree of intellectual disability. Many children with Down syndrome who have received family support, enrichment therapies and tutoring manage to perform well at school and are able to do paid work and some participate in post- secondary education as well. Early childhood intervention, screening for common problems, medical treatment where indicated, a conducive family environment and vocational training can improve overall development of children with Down syndrome. Education and proper care will improve quality of life significantly despite the genetic limitations (Roizen *et al 2003*).

2. Purpose

The purpose of these guidelines is to provide a standardized and consistent approach to the medical management of children with Down syndrome for Shropshire Community Health Trust [SCHT] in order that children with Down syndrome can reach their full potential and are screened accordingly for any potential health care problems. This clinical guideline applies to all medical/clinical healthcare professionals who are routinely involved in detection / screening and the diagnosis and medical management of children with Down syndrome.

3. Definitions / Glossary

Ab	Anti-bodies
Atlanto-axial	The Atlanto-axial joint in common terminology used is actually a composition of the 3, 2 lateral and 1 median atlantoaxial joints
Atresia	Atresia describes a congenital absence or closure of a normal body opening or tubular structure
BMI	Body Mass Index
BMJ	British Medical Journal
Brushfield spots	Brushfield spots are small greyish/brown spots on the peripheries of the iris in the human eye due to aggregation of connective tissue, a normal iris element in the eye
CDC	Child Development Centre
Chromosomes	Chromosomes are the blueprint of the bodies; they are found in every cell in our body and determine our physical and mental characteristics. The usual number of chromosomes is 46 (arranged in 26 matched pairs) People with Down syndrome have an extra chromosome 21 in their genetic makeup

Dermatoglyphs	Dermatoglyphs is the scientific study of finger prints, all primates have ridged fingers, people with DS mainly have ulnar loops.
Diplegia	Diplegia refers to paralysis affecting symmetrical parts of the body
DSMIG	Down Syndrome Medical Interest Group
Epicanthic folds	 Is a skin fold of the upper eye lid, covering the inner corner (medial canthus of the eye).
ECG	Electrocardiography
ENT	Ear, Nose and Throat
ESPGHAN	European Society for Paediatric Gastroenterology, Hepatology and Nutrition
FASP	Fetal Anomaly Screening Programme
Flexion	In anatomy, flexion is the position that is made by the joint angle decreasing (bending)
GP	General Practitioner
HCG	Human Chorionic Gonadotropin
IQ	Intelligence Quotient is a score devised from one of several standardised tests designed to assess intelligence.
Iris	The iris is a thin circular structure in the eye, responsible for controlling the diameter and the size of pupils this the amount of light reaching the retina
Macroglossia	Macroglossia is the medical for unusual enlargement of the tongue.
Microgenia	Microgenia is the medical term for unusually for small unusually formed chin
Muscle Hypotonia	Hypotonia is a state of low muscle tone
MDT	Multi-Disciplinary Team
PAPP-A	Placenta Associated Plasma Protein A
SATH	Shrewsbury and Telford Hospitals
Short Stature	Short stature refers to human height which is below expected
SaLT	Speech and Language Therapy
Trisomy 21	Trisomy is a type of polysomy in which there are 3 copies, instead of the normal 2, of a particular chromosome. Down syndrome is common trisomy's 21 that survive to birth in humans
TFT	Thyroid Function Tests
TSH	Thyroid Stimulating Hormone
HLA	Human Leucocyte Antigen
IUGR	Intra uterine growth retardation

4. Duties

Chief Executive

The Chief Executive has ultimate accountability for the strategic and operational management of the Trust, including ensuring there are effective and appropriate processes in place for the medical management and health surveillance of children with Down syndrome.

Director of Nursing & Medical Director

The Director of Nursing & Medical Director have responsibility for ensuring that children with Down syndrome are offered appropriate medical management and health surveillance checks in place and support patient safety at all times.

Service Managers

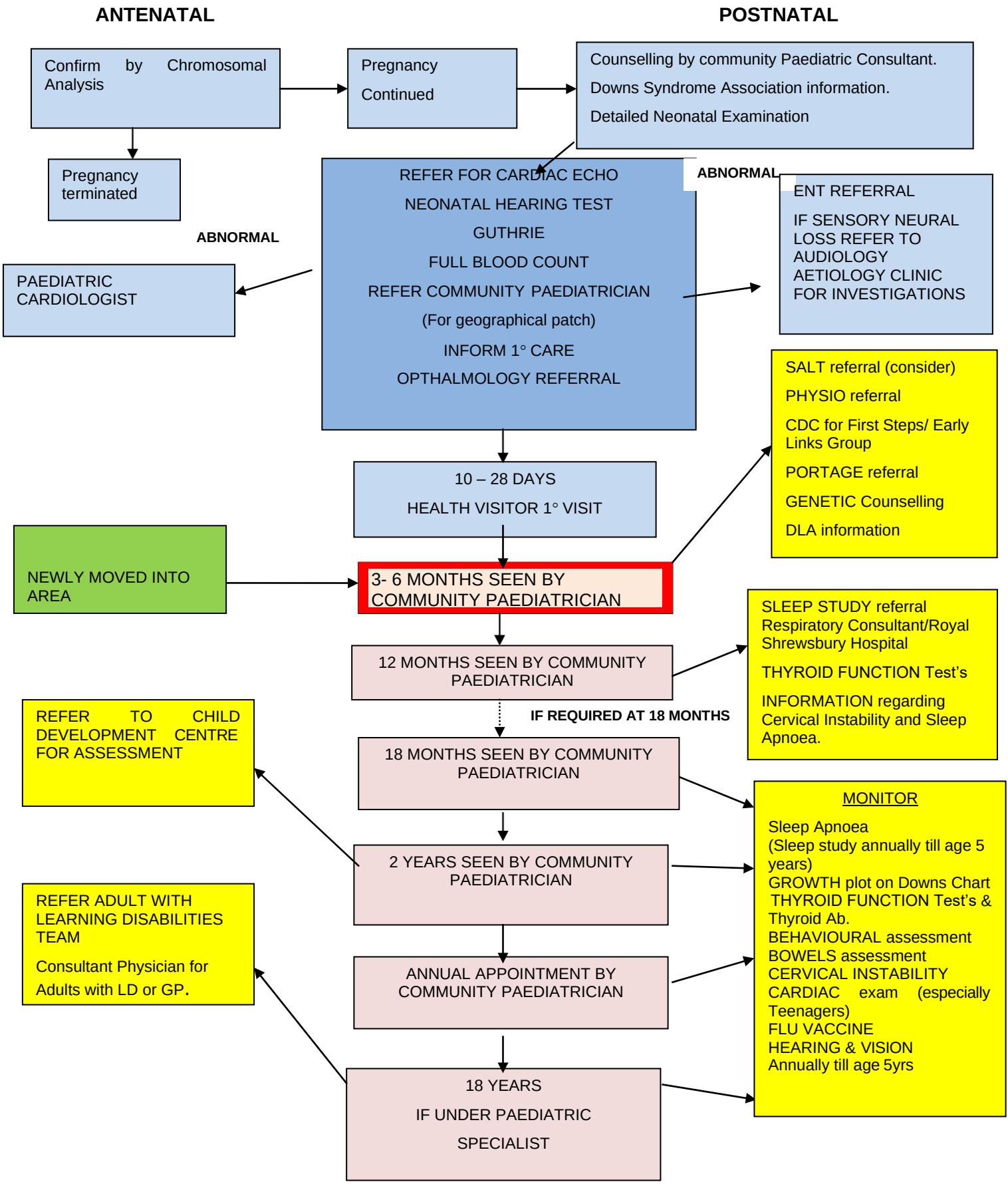
Service Managers are responsible for the day to day operational management and coordination of the medical management and health surveillance of children with Down syndrome in line with the clinical guideline and ensuring their teams are aware of their Multi –Disciplinary Team MDT responsibilities in the care of children with Down syndrome.

All Clinical Staff

Clinical staff are key members of the MDT, in ensuring that children with Down syndrome are managed appropriately as per national / local guidelines and are offered regular health surveillance checks. All clinical staff is required to comply with this guideline and to report any adverse care related issues to their line manager and to complete a Datix incident report in line with the Trust's Incident reporting policy.

5. Down Syndrome Medical Pathway

The clinical pathway below summarises the medical management of children with Down syndrome in SCHT:



Down Child Health Surveillance

3 Months

- Full History & Examination
- Feeding & Gastroesophageal Reflux
- Bowel Activity
- Cardiac assessment, check ECHO, refer to Cardiology
- Hearing
- Vision
- Immunity
- Infections

6 Months

- Same as 3 month clinical examination
- Immunisation
- Encourage Influenza
- Primary immunization
- Audiology
- SALT
- Sleep study ref
- SEND notification

1 Year

- Consider referral to CTLD if behaviour
- In addition to checklist
- Low threshold for Coeliac disease test
- Examination specific
- CVS, CNS, EN, Ophthalmology
- Plot on Down Syndrome SP chart
- Physio, SALT included in CDC
- Check re
- Respiratory, Cardiac, Bowel symptoms
- Recurrent upper airway
- Portage
- Cervical instability - give leaflet
- DSMIG Schedule
- Exam –Scoliosis?

2 Years

- Any parental concern? Same clinical assessment and examination as previous reviews, with particular attention to growth (plot on DS chart), ENT cardiovascular, neurological/signs of cervical spine instability or cord compression.
- Foot posture
- Behaviour, consider co-morbidities / dual diagnosis, or emerging concerns
- Dental care- referral to specialist dental team indicated?
- Early education, plans under way? Contact SEND team
- If there is indication of dual diagnosis of Autistic Spectrum disorder
- See DSMIG schedule
- Thyroid, Coeliac screen, Audiology. Annual Oximetry test
- Refer to Paediatric Ophthalmology Service

3 Years

- Any parental concern? Repeat full clinical assessment and examination as previous reviews, with particular attention to growth (plot on DS chart), ENT, cardiovascular, neurological/signs of cervical spine instability or cord compression

- Developmental progress and general health. Bowels, thyroid symptoms and coeliac symptoms

Screening and Diagnosis

Down syndrome symptoms vary from person to person and can range from mild to severe. However, children with Down syndrome have a widely recognized appearance.

[Refer to Appendix 1]. Individuals with Down syndrome may have some or all of the following physical characteristics.

Common Physical Characteristics

The following 10 features of DS are typically present in the neonatal period:

- Hypotonia (80% of patients)
- Poor Moro reflex (85% of patients)
- Hyper-flexibility of joints (80% of patients)
- Extra skin on back of neck (80% of patients)
- Flat facial profile (90% of patients)
- Slanted palpebral fissures (80% of patients)
- Anomalous auricles (60% of patients)
- Hypoplasia of iliac wings (70% of patients)
- Short middle phalanx of the fifth finger (60% of patients)
- Single Palmar Transverse crease (45% of patients

General examination in an infant or older child may reveal other findings, although not all features need to be present to suspect the diagnosis:

- Skull: brachycephaly with a flat occiput
- Eyes: epicanthal folds in addition to up slanting palpebral fissures, Brushfield
- spots of the iris
- Nose: short, a low nasal bridge, small nares
- Ears: small, may be low-set
- Tongue: apparent protrusion
- Mouth: down-turned, small oral cavity
- Extremities: short hands, in addition to possible single palmar transverse creases, digital dermatoglyphics, and fifth finger clinodactyly. Wide space between first and second toes, with vertical plantar creases.

Epidemiology

The incidence of Down syndrome is estimated to be between 800 to 1000 births (NICE 2006). Down syndrome occurs in all ethnic groups and among all economic classes. Maternal age influences the chances of conceiving a baby with Down syndrome. At maternal age of 20-24, the probability is one in 1562; at the age of 35 to 39 the probability is one in 214, and above the age of 45 the probability is one 19.

Although the probability increases with maternal age, 80% of children with Down syndrome are born to women under the age of 35, reflecting the overall fertility of that age group. Recent data also suggest that paternal age, especially beyond 42, also increases the risk of Down syndrome manifesting.

Pre-natal Screening

The NHS Fetal Anomaly Screening Programme (FASP) uses a combination of tests to test for Down syndrome. These tests cannot provide a definite answer as to whether the child does or does not have the syndrome, but provide a reasonably estimate to the level of risk (such as 1 in 100 chance or 1 24,000). The tests used depend on the stage at which a women requests testing, but they all combine blood test with a special type of ultrasound, known as a translucency scan. This type of scan is similar to a normal ultrasound scan, but measures the space between the spine and the nape of baby's neck. Measuring the thickness of 'nuchal translucency' helps to estimate the risk of the syndrome.

Ideally combined testing is done in the first trimester of pregnancy, blood is taken from the mother between 10-14 weeks to check for levels of certain hormones and proteins, the hormones usually measured are free beta Human Chorionic Gonadotropin HCG and placenta associated plasma protein A (PAPP-A), the levels of these chemicals, combined with nuchal translucency measurement, are used to calculate the risk of the baby having Down syndrome.

Non-invasive Pre-natal Testing: Mum's blood sample at any time from early pregnancy can be analysed for cell free foetal DNA, which is essentially a marker in the mother's blood of the DNA of the baby.

Done at 10 weeks or later it has sensitivity for the detection of Down syndrome greater than 99% and eliminates the risk of miscarriage from invasive testing.

Cognition

Cognitive development of children with Down syndrome is quite variable. It is not currently possible at birth to predict the capabilities of any individual reliably, nor is the number or the appearances of physical characteristics predictive of future ability. Individuals with Down syndrome vary considerably in their language and communication skills.

It is routine to screen for middle ear problems and hearing loss which might affect cognitive development.

Reports have shown that children with Down syndrome display more behavioural problems and show significantly more attention concerns than their siblings

Most individuals with Down syndrome have intellectual disability in mild (Intelligence Quotient [IQ 5-70) to moderate (IQ 35-50) range.

6. Chromosomal Classification

- Down syndrome disorders are based are having too many copies of the genes located on chromosome 21. In general this leads to an over expression of the genes. The three forms of Down syndrome are Trisomy 21, mosaic Down syndrome and translocation Down syndrome. Genetic testing can usually show what type of Down syndrome a baby has.

Standard Trisomy 21

- 95 per cent
- Meiotic nondisjunction during gamete formation
- Karyotype 47XY + 21 (Male) + 47XX + 21 (Female)

Mosaic

- 1% of Down syndrome
- Both typical and trisomy 21 karyotype
- Milder phenotype but behavioural and cognitive profiles vary

Robertsonian Translocation

- 4% of Down Syndrome
- Has 46 chromosomes
- Translocation between 21q and long arm of either 14 or 21
- Higher recurrence if one of the parents is a carrier
- 21q 21q - 100% recurrence risk

7. Health problems associated with Down syndrome

Growth and feeding

- If preterm, standard growth charts for prematurity used until term and then changed to Down syndrome growth charts. Ensure that PCHR growth chart insert is also used
- Early weight loss may be >10% and often takes longer than 2 weeks to regain birth weight as long as baby well no need to refer; if not attained birth weight by 4 weeks, may need evaluation for feeding difficulties or underlying major pathology; Breast feeding should be encouraged and supported.
- Short stature is characteristic
- Growth spurts and plateau occur as in all children but these tend to be more prolonged
- Pubertal growth spurts usually less vigorous and may occur early
- If <2nd centile for weight or falls away on centiles – needs assessment
    - If BMI above the overweight thresholds – offer specialist referral if available for guidance; always check TFTs.

Heart

- 40-60% have congenital heart problems; Of these 30-40% -AVSD – needs early diagnosis and referral for full corrective surgery
- All newborns with DS should have careful clinical examination and ECG
- If abnormal ECG or clinical exam – needs urgent referral for echocardiography (within 2 weeks)
- This also applies to babies diagnosed with Down syndrome later as neonate
- Adolescence with DS should have annual cardiovascular examination and prompt referral for any cardiac symptoms.

Thyroid

- Congenital hypothyroidism – 1-3.6% of children with DS; 10% of school age children have uncompensated hypothyroidism; prevalence increases with age.
- Ensure Guthrie test is negative; If abnormal, repeat TFT, commence thyroxine immediately/ refer to endocrinologist
- Consider hypothyroidism in the differential diagnosis of lethargy, constipation, changes in affect, cognition, growth, depression and dementia.
- All CYP with Down syndrome should be offered regular thyroid function blood tests as part of ongoing surveillance for thyroid dysfunction and treated where indicated in a timely manner..
- Offer a venous blood test for TSH, free T4 and thyroid peroxidase (TPO) antibodies within 5 working days of the initial blood test.
- Consider initiating treatment whilst awaiting blood test results if TSH is very high and there is clinical suspicion of hypothyroidism.
- **Initial venous TSH concentration above 10mU/l, and low free T4:**
- Offer immediate repeat venous blood test to measure TSH, free T4 and thyroid peroxidase (TPO) antibodies.
- Consider initiating treatment whilst awaiting blood test results if TSH is very high and there is clinical suspicion of hypothyroidism.
- Formulate an individualized management plan for the child or young person if blood test results confirm a diagnosis of hypothyroidism and consider discussing the plan with a clinician with expertise in paediatric endocrinology.

Blood test shows a normal TSH and normal free T4 but raised thyroid peroxidase (TPO) antibodies: The guideline group recognized that a TSH level above the local laboratory-defined reference range and below 10mU/l with a normal or low fT4 should raise concern of central hypothyroidism and should be investigated further as soon as possible.

1. The timing of follow up blood tests will vary depending upon individual circumstances including signs and/or symptoms and the CYP's age (Noble et al, 2000; Murphy et al, 2008; Tenenbaum et al, 2012; McGowan et al, 2015). Due to the higher than anticipated prevalence of thyroid disorder in young children (see prevalence table in Appendix K) and the vulnerability of the developing brain the group recommended that in children under the age of 3 years follow up should be done in 1 – 3 months. For CYP aged 3 and over the follow up period is recommended as 3 – 6 months. The guideline group Down' Syndrome special interest group were aware that this age cut-off of 3 years for differentiating management is a year older than the age cut-off of 2 years recommended in the NICE (2019) guideline on thyroid disease, but all members of the Down Syndrome Special Interest group felt strongly that additional caution was appropriate for young children who have Down syndrome.

Hearing

- All newborn babies should have Universal Newborn Hearing Screen
- High incidence of conductive, sensory neural and mixed hearing loss
- Up to a third will not pass their UNHS and will have conductive hearing loss; extremely narrow ear canal interfere with testing and hearing abilities; if failed UNHS, check if referred for further investigation. All children with Down syndrome should have new-born hearing screening followed by targeted hearing assessments, initially at 6 to 10 months and then six monthly till the age of two and to continue at least annually throughout school-age years. A hearing assessment is recommended at least two yearly throughout adult life or more frequently if there are concerns. This should include measurement of auditory thresholds, impedance testing (tympanometry) and otoscopy.

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Vision

- Tenfold increased risk of congenital cataract and infantile glaucoma; Nystagmus present in at least 10%; Careful examination at birth and at 6 weeks for red reflex
- Refractive errors and or squint may present at an early age; Visual behaviour must be

monitored before the formal ophthalmology review and refer earlier if concerned

- Blepharitis occurs in upto 30%; Nasolacrimal duct obstruction is common and needs surgical assessment if present at 9-12 months.
- As with all children newborns with Down syndrome should be examined for congenital cataract and other eye anomalies by a trained person and this should be repeated at 6 weeks. Visual behaviour must be monitored by the child's paediatrician particularly before the first formal ophthalmologic review. Those who start to squint or show other abnormalities of gaze, visual behaviour or attention should be referred for ophthalmological review. Between 18 months and 2 years all children with Down syndrome should have formal ocular/visual assessment by an orthoptist and ophthalmologist/optometrist in accordance with local arrangements. This should include orthoptic assessment, refraction, and fundus examination. At least one third will have ocular/visual defects by this age. Those with deviation from normal should be kept under appropriate specialist review. Refractive errors, most commonly hypermetropia, which often reduce spontaneously in other children, are likely to persist beyond infancy. Correction for hypermetropia may be helpful at a younger age than that for typically developing children especially since the majority will have defective accommodation.

Gastrointestinal Tract

- 7% have congenital anomalies of GIT including oesophageal atresia/ TOF, pyloric stenosis, duodenal stenosis/ atresia and anal stenosis/ atresia
- Hirschsprung's disease – incidence 2.6% in DS
  - 8-12% incidence of coeliac disease; mostly asymptomatic; HLA testing for children with Down syndrome has been incorporated into the surveillance for identifying children with coeliac disease, as per the recent recommendation from the Royal College of paediatrics and Child health and Down syndrome special interest group June 2021.
  - Children with coeliac disease will have a predisposing genetic factor DQ 2 or DQ 8 in over 99% of the patients and diagnosis of coeliac disease is unlikely if neither of these is present. If HLA test is positive, then the risk of coeliac disease is present and coeliac antibody test will need to be undertaken. However, if this is negative, then they would not require further screening. If coeliac antibody test is positive or there is a high degree of clinical suspicion, then further referral to the gastroenterologist will be required.
  - Screening for children with coeliac disease will now be HLA testing. Screening will take place within the first six months prior to the introduction of gluten in the diet.
    HLA DQ 2 and HLA DQ 8 will be undertaken if this is positive coeliac antibody screen will also be undertaken.
    The discussion for the above will happen during the clinical consultation.

Haematology

- In neonates: Haemoglobin and haematocrit are higher than normal babies; 20% are polycythaemic; Platelet counts are also lower but important to rule out other causes like sepsis/ IUGR
- Transient myeloproliferative disorder (TMD) – congenital leukaemia unique to neonates with DS; majority resolve spontaneously by 3 months; Severe TMD needs treatment
- 20% of those with resolved TMD develop AML in the first 4 years of life; therefore monitored more closely with frequent blood films and FBC
- All neonates with DS should have FBC and blood film in the first 2-3days of life

Immunisations

- All routine immunisations advised.
- RSV – if relevant cardiac or prematurity factors
- There is very limited evidence of the safety or efficacy of the RSV vaccine in children, and it should not be offered. It may be considered on an individual basis for older children/young people (over 12 years of age) who are extremely vulnerable and at

high risk of exposure.

- COVID-19- The NHS in the UK is now able to offer antibody and antiviral treatments for those with coronavirus (COVID 19) infection who are at highest risk of becoming seriously ill from the infection. Treatments have been available for some people seriously ill in hospital since September 2021. As from 20 December 2021, neutralising monoclonal antibody treatment can be offered to people in the community aged 12 and over who have tested positive via PCR, and are in certain vulnerable groups.
- People with Down syndrome over the age of 12 are included in the list of those with highest risk.
- Those who may be eligible will be identified by the NHS centrally, and local pathways are being set up to administer treatment.
- Further details can be found on the Down's Syndrome Association website or via www.nhs.uk/conditions/coronavirus-covid-19/treatments-for-coronavirus/
- It is important for people who have Down syndrome and their families to understand the implications of the government advice. This is available in an easy read format on the Down's Syndrome Association, Down's Syndrome Scotland and Down Syndrome Ireland websites.

- People with Down syndrome may be at greater risk of other infections aside from COVID-19. It is therefore also important to maximise other measures recommended to minimise the risk of infections including routine and additional immunisations, and treatment of other medical problems that may predispose to infection.

- Therefore it is really important to take strict precautions to prevent the spread of the virus. This includes maintaining strict hand hygiene, social distancing and wearing a mask.

Early Interventional Therapies in Childhood

Early management involves investigations as described in Section 7 along with suggested schedule of health checks for Down syndrome, in addition to which there is a supportive and informative role to be provided to the family. Later management of an individual with Downs syndrome and their family is a complex topic requiring a multidisciplinary approach. In Shropshire Community Health Trust the basis of this approach is formed by local CDC, and early referral to this important. Clinicians are encouraged to refer Children with Down syndrome for an offer of Friday Morning Group / Parent meetings, referral to relevant specialists/ therapist and Portage can be initiated by this team.

School Leaver

- Repeat full clinical examination. Discuss behaviour Self care, independence, personal hygiene, sex and relationship education.
- Refer for support to Learning Disability Team where indicated.
- Dental care- referral to specialist dental team indicated? Discuss immunisation and annual influenza vaccine.
- Testicular Check
- Transition to for adults
- State Learning disability as a diagnosis and please ensure from age 14, annual health checks occur with the GP.Please consider obtaining a cognitive baseline assessment from 18 years in case of early onset dementia.
-

Consider the following if available:

Physiotherapy: to ensure the child avoids developing abnormal compensatory movements for the physical limitations (hypotonia, ligament laxity, short extremities)

Occupational therapy: facilitates mastering self-help skills for independence (feeding, dressing, writing); also focus on oral motor exercise

Speech and language: Expressive language more delayed than receptive; Language intervention involves the family and child's learning style

8. Down Syndrome Medical Interest Group

- Website (http://www.dsmig.org.uk/)
- Offers important information for healthcare professionals on 'best practice' medical care for individuals with Down syndrome
- Created by the UK Down Syndrome Medical Interest Group (DSMIG)
- Includes: medical library; DSMIG information resources, including special insert for the parent held child record (PCHR), guidelines for basic child health surveillance clinical awareness notes and at a 'glance key points'

8. Review and Compliance Monitoring

Element to be monitored	Lead	Tool	Frequency	Reporting arrangements	Acting on recommendations and Lead(s)	Change in practice and lessons to be shared
Adoption of standardised Down chart centile graphs for Weight, Height, Head circumference Growth, Hearing & Vision, Cardiac, TFTs, Monitoring	Service Managers	Patient records audit	Annual	The audit report will be submitted to the Quality and Safety Group. The group is expected to read and interrogate the report to identify deficiencies in the system and act upon them	The audit report will be submitted to the Quality and Safety Group. The group is expected to read and interrogate the report to identify deficiencies in the system and act upon them	The audit report will be submitted to the Quality and Safety Group. The group is expected to read and interrogate the report to identify deficiencies in the system and act upon them
The review of audit data and the instigation of remedial action if deficits are identified.	Service Manager	Patient records audit	Annual	The audit report will be submitted to the Quality and Safety Group. The group is expected to read and interrogate the report to identify deficiencies in the system and act upon them	Any changes to the process will be identified as part of the audit and allocated to a relevant person(s) within a specified timeframe	Relevant clinical staff will be responsible for changes in the risk assessment. Lessons will be shared with all the relevant stakeholders.

9. Consultation

Consulted with Consultant Community Paediatricians Dr Janet Butterworth, Dr Short, Dr Unsworth, Dr Ganesh, Dr Buch, Dr Ogilvie, Dr Postings, Dr Raveendran, Nurse Consultant Narinder Kular, Dr Minnaar in Community Paediatric Doctors Meeting on 20 April 2021

10. Dissemination and Implementation

These guidelines will be disseminated by the following methods:

- Managers Informed via DATIX system who then confirm they have disseminated to staff as appropriate
- Staff via Team Brief
- Published to the staff zone of the trust website

11. Advice and Training

Training will be covered by regular Community Paediatric CPD meetings.

12. References

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3. Dominic A Fitzgerald**,** Annette Paul**,** Clare Richmond. Severity of obstructive apnoea in children with Down syndrome who snore. *Archives of Disease in Childhood* 2007;**92**:423-425
4. Medical Management of Children with Down's syndrome, Current Paediatrics 2001, 11, 57 – 63
5. American Academy of Pediatrics Committee on Genetics (February 2001). "American Academy of Pediatrics: Health supervision for children with Down syndrome: Health supervision for children with Down syndrome". *Pediatrics 107* (2): 442-49
6. Roizen, NJ; Patterson, D (April 2003). "Down's syndrome" (Review). *Lancet* 361 (9365): 1281–89. DOI: 10.1016/S0140-6736(03)12987-X. PMID 12699967.
7. Standards for Services for Children with Disorders of Sleep Physiology Report
    i. *2009 Royal College of Paediatric's & Child Health*
8. BMJ *'Best Practice Guidelines for Down Syndrome'* July 2017
9. ESPGHAN 'updated Coeliac Guidelines' 2012
10. JUNE 2021 DSMIG Presentation BACKGROUND Part 1
11. JUNE 2021 DSMIG Presentation DS SPECIFIC Part 2

13. Associated Documents

- Down Guidelines from Down Syndrome Medical Interest Group (DMIG) http://www.dsmig.org.uk/
- Shrewsbury Friday Morning Parent Meeting Group Criteria 2012
- Shropshire Community Health NHS Trust Consent to Examination and Treatment Policy
- Shropshire Community Health NHS Trust Records Management Policy

15.1 Appendix 1 SATH Neonatal Checklist

Clinical Features of Downs Syndrome		Baseline Investigations - Neonatal period (or time of diagnosis) SATH Neonatal Checklist
Confirm diagnosis of Down syndrome by chromosomal analysis ….		

• Features of Down's Syndrome post delivery	SATH	• Senior Paediatrician and Midwife to discuss with both parents
		• Take blood for:
		  ○ FBC and blood film
		  ○ Confirmatory genetic testing. Initial QR-PCR testing results may be available in 3 days whereas karyotype results take 7-10 days.

• Diagnosis confirmed	SATH	• Give a new parent information pack, including the Down Syndrome 2011 PCHR insert • Inform Obstetrician, Midwife, Health Visitor community paediatrician and GP
• Detailed newborn examination	SATH	• Plot growth using 2011 updated DS growth charts / PHCR inserts • Ensure bowels opened <24 hours and feeding established • Check urine passed and antenatal scan normal • Refer on to Ophthalmology if in doubt about congenital cataract • Cardiac referral: Symptomatic babies require immediate referral. If asymptomatic but there are abnormalities on examination - to be seen within 2 weeks; if cardiac examination and ECG are normal , cardiac referral can be seen within 6 weeks • Ensure neonatal hearing screen completed and audiology follow-up is arranged for 8 months, even if screening is normal. • Treat polycythaemia and thrombocytopenia if indicated. If abnormal blood film, discuss with a paediatric haematologist
	SATH	• Typed letter to GP with copies Health Visitor, Obstetrician, Parents, Child health Longbow House, Congenital Abnormality Survey (Dr Tyler's secretary)

Appendix 2 SCHT Clinical Management

Recommendations for the management of Downs Syndrome In childhood

DOWNS SYNDROME CLINICAL MANAGEMENT GUIDELINES

[1]Recommended Testing / Screening / what to look out for	Clinical Management Recommendation
• **Cardiac** Congenital malformations; Cor pulmonale; Acquired valvular dysfunction	• Joint care between Local Paediatric SATH Cardiologist / Birmingham Children's Hospital Clinical cardiac abnormality, ECG, Echocardiogram, FBC
• **Thyroid disorder** Growth retardation; Hypothyroidism Hyperthyroidism; Diabetes	• Follow Up TFTs at 6 months then annually
• **Hearing impairment** Conductive hearing loss Sensory neural hearing loss	• New-born hearing screening followed by targeted hearing assessments, initially at 6 to 10 months and then six monthly till the age of two and to continue at least annually throughout school-age years.
• **Growth** Short Stature, can be prone to obesity,	• Plot height, weight, and head circumference on updated Downs Syndrome centile charts/ BMI refer dietician / available weight loss programmes
• **Ophthalmology problems** Refractive errors; Blepharitis; Nasolacrimal obstruction; Cataracts; Glaucoma Nystagmus; Squint; Keratoconus	• Formal eye assessment of all children between 18mths-2yrs & then at 4yrs- refer PRH Clinic, nasolacrimal obstruction if present at 9-12mths, needs surgical referral
• **Cervical spine instability** Hip sublaxation/dislocation Patellar instability; Scoliosis Metatarsus varus; Pes planus	• Refer Physiotherapy; Orthotics devices; Symptomatic Individuals neck pain, abnormal posture, Torticollis, reduced neck movements, deterioration in gait, increased fatigue - X-rays, referral to RJAH Othpod opinion
• **ENT & Dental** Keep upper airway obstruction Chronic catarrh; Sleep apnoea/habitual snoring / Prone to Viral URTI; chest infection	• Refer ENT, Sleep Apnoea Test (baseline at a year and then annual referral to respiratory paediatrician till 5 years); Check Childhood Immunisation Status / Flu Vaccination, Dental checks aged 2yrs and 6mthly thereafter
• **Gastrointestinal** High risk of asymptomatic coeliac disease	• Anti TTG & Total IgA, asymptomatic children every 2-3yrs (IgA deficient do IgG Anti TTG) referral to gastro consultant if necessary

Appendix 3 SCHT Clinical Management Guidance 5-18 years