

## Nottingham Guidelines for the Care of Children with Down Syndrome

# Down Syndrome

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Evidence base: (1-5)		4
1a	meta analysis of randomised controlled trials	
1b	at least one randomised controlled trial	
2a	at least one well-designed controlled study without randomisation	
2b	at least one other type of well-designed quasi-experimental study	
3	well –designed non-experimental descriptive studies (ie comparative / correlation and case studies)	
4	expert committee reports or opinions and / or clinical experiences of respected authorities	
5	recommended best practise based on the clinical experience of the guideline developer	
Consultation Process		Paediatric Clinical Guidelines Group
Target audience		Clinicians and healthcare professionals caring for children and young people with Down Syndrome
This guideline has been registered with the trust. However, clinical guidelines are guidelines only. The interpretation and application of clinical guidelines will remain the responsibility of the individual clinician. If in doubt contact a senior colleague or expert. Caution is advised when using guidelines after the review date.		

## Nottingham Guidelines for the Care of Children with Down Syndrome

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### Document Control

#### Document Amendment Record

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V1	October 2013	Dr. Liz Marder Consultant Paediatrician Dr Claire McCall Associate Specialist
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V4	July 2022	Dr Liz Marder Minor amendment – removal contact details only
V5	August 2022	Dr Liz Marder Dr Anastasia Michael, Registrar in Neurodisability Dr Hannah Winterton, Paediatric Registrar Dr Christopher Kingsnorth, Paediatric Registrar

### Summary of changes for new version:

Changes to this iteration of the guideline (minor amendments only):

- New thyroid screening guidance as per DSMIG guideline
- Minor updates in line with new DSMIG neonatal guidance
- Streamlining of advice and presentation throughout the document
- Updated references and weblinks

### Statement of Compliance with Child Health Guidelines SOP

This guideline has followed Child Health Guideline SOP. It has been circulated to all Paediatric Senior staff and comments incorporated before uploading to the Trust Guideline site.

Maria Moran

Clinical Guideline Lead

27 Sept 2022

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## Nottingham Guidelines for the Care of Children with Down Syndrome

### Introduction

These guidelines have been written to describe what is considered to be the appropriate medical input and support for children with Down Syndrome (DS). In Nottingham the service is focused around the Down Syndrome Children's Clinic at the Childrens Development Centre, and this is the programme we aim to offer there.

The information within them should mean that children and young people can be followed by health professionals working within different settings. We hope that this will ensure that all children with Down Syndrome in Nottingham receive a comprehensive service which can be provided in the most appropriate place according to the needs and wishes of the child and family.

These guidelines are largely based on work done by the Down Syndrome Medical Interest Group (DSMIG, UK and Ireland) who have produced guidelines for basic medical surveillance in children with Down Syndrome. These are, as far as possible, evidence-based and, where evidence is not available, draw on the expertise and consensus of clinicians and the relevant professional bodies. These guidelines, and a wide range of other health information, can be found at [www.dsmig.org.uk](http://www.dsmig.org.uk).

For further information about these guidelines, or advice on any aspect of medical management for children with Down Syndrome, please contact the Down Syndrome team based at the Childrens centre, City Hospital.

### Overview of Nottingham Down Syndrome Children's Service

#### **Antenatal**

- If a diagnosis is made at this stage, and parents are clear they are continuing with the pregnancy, a referral to the Down Syndrome Team may be made for further information about Down Syndrome and the services available. .

#### **Neonatal**

- Parents to be informed of the likely diagnosis as soon as possible, usually by a Senior Paediatrician and by the responsible Consultant as soon as feasible.
- A comprehensive medical assessment according to the guidelines must be carried out prior to discharge and information documented on the supplied form (**Appendix 1**) to accompany the discharge letter.
- Referral to the Down Syndrome Team who will make an initial visit as soon as appropriate.

#### **First year**

- Children are invited to come at age 3 months, 6 months and one year for a review.
- Referrals and introductions will be made to the other members of the Multidisciplinary team.
- Ongoing follow up by specialty teams and referrals as appropriate

## Nottingham Guidelines for the Care of Children with Down Syndrome

### Pre- school

- Annual review at the Down Syndrome clinic.
- Transfer to local Community Paediatric team usually following the 4<sup>th</sup> year review.

### School age

- Annual review by local Community Paediatric team.

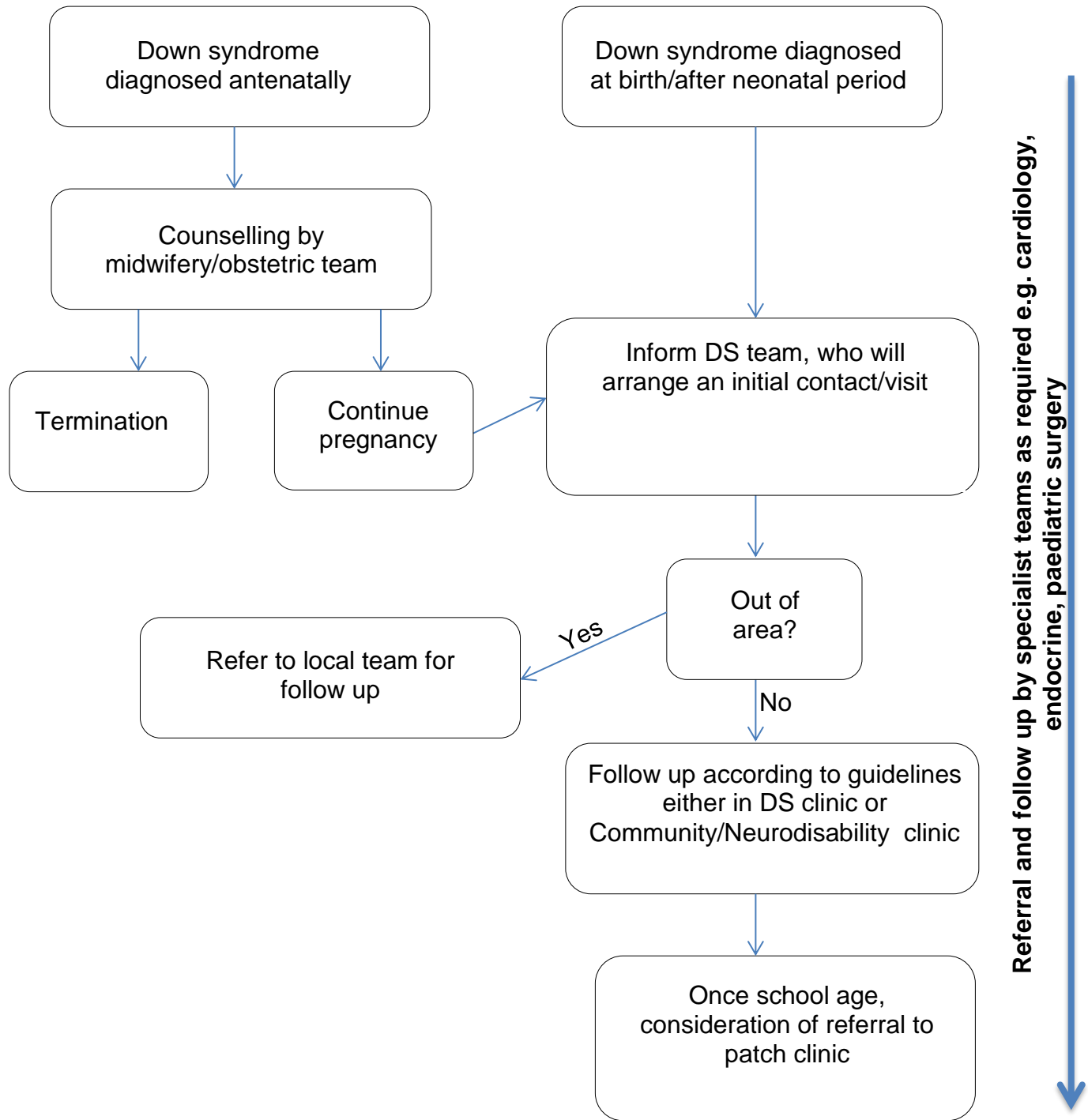
### School leavers

- Review by Community Paediatrician and preparation of a comprehensive report.
- Ensure all young people age 14 years and over have an annual health check with their GP (offered to all young people with Intellectual disability, ensure this is on diagnosis list), the GP should be the main point of contact and take over medical input from community/neurodisability team.
- Signpost both families and primary care teams to advice on annual health checks on the Down Syndrome Association website (advice and Wellbeing for Adults with Down Syndrome) <https://www.downs-syndrome.org.uk/about-downs-syndrome/health-and-wellbeing/#annualhealthchecks>
- Ongoing follow up by specialist hospital teams as required including adult intellectual disability Team (Under mental health service) if indicated

Children should be routinely offered appointments at Down Syndrome clinic or community patch clinic as per the schedule above but more often if indicated. At each review, a full history and examination should be conducted, carefully eliciting any parental concerns and screening for conditions seen more frequently in children and young people with Down Syndrome. While not exhaustive, a list of points to cover in the suggested review schedule and a table of medical conditions that occur more commonly in children and young people with Down Syndrome have been summarised below. Please also refer to the relevant sections for further information. See also **Appendix 3** for proforma that can be used for clinic visits.

## Nottingham Guidelines for the Care of Children with Down Syndrome

### Overview of Nottingham Down Syndrome Children's Service



## Nottingham Guidelines for the Care of Children with Down Syndrome

### Suggested Review Schedule

	Newborn	3 month	6 month	1 year	2-4 year	5y +	Transition
<b>History</b> -always take a full systems History		Special focus on: Reflux and feeding - especially in early reviews Bowels- low threshold for coeliac screen Infections (unusual/recurrent), breathing/sleep Developmental progress and behaviour Support- therapy/education/benefits				As previous, also dental health, puberty	As previous Fertility/contraception Further education/placement
<b>Examination</b> -always do a full systems examination	Newborn exam Check for conditions seen more commonly	Careful check of cardiac status Development		Cardiac status Development Cervical spine instability/cord compression, ENT/signs of upper airway obstruction		As previous, also Orthopaedic problems/Scoliosis	As previous, also Testicular exam/advise self examination
<b>Growth monitoring</b>	Plot on appropriate DS growth chart and red book Check BMI if overweight						
<b>Thyroid blood checks</b> (minimum)	Routine Blood spot		Check Bloods at 6 months and then yearly (either blood TFTs, or Blood spot TSH)			2 yearly checks if no concerns	
<b>Eye checks</b>	Check carefully for nystagmus, cataracts, squint, visual behaviour, blepharitis etc. Nasolacrimal duct obstruction sometimes needs referral Refer if concerns				As previous routinely to ophthalmology at 2 years for yearly review	Can be seen by optician, yearly	
<b>Hearing checks</b> (minimum)	Newborn hearing screen		All children should be sent follow up for 8 months, check at 1 year review this is done. Regular hearing checks as per audiology schedule below (see Hearing section)				
<b>Heart Checks</b>	Echo routinely either on NNU or within 4 weeks	Check had echo & result Referral to cardiologist if required	Always review heart status and low threshold for referral/investigation if concerns				Repeat echo routinely even if asymptomatic/no concerns Then by examination/history



## Nottingham Guidelines for the Care of Children with Down Syndrome

Breathing and Sleep Checks	Careful history of breathing/sleep and enquiry about sleep and quality of sleep, consider sleep study at any stage if concerns			Consider referral for routine sleep study yearly Routine study at 1 year and 4 years minimum		Careful history of breathing/sleep and enquiry about sleep and quality of sleep, consider sleep study at any stage if concerns	
Infections and Immunisations	Take every opportunity to council parents about importance of early identification and management of infection Ask about infections, frequency, antibiotic courses, consider prophylactic antibiotics if indicated, consider testing immune function and referral to immunology if concerns.						
	RSV Prophylaxis if meet respiratory or cardiac criteria in 1 <sup>st</sup> year of life	Discuss importance of immunisation	Discuss importance of immunisation, advise annual influenza	Check immune function 4 weeks after immunisations	Consider pneumovax II after 2 <sup>nd</sup> birthday		
Other Bloods	Done as per clinical concern/indication from history, always have a low threshold for FBC, coeliac screen, checking for autoimmune conditions such as diabetes						
Other		Family adjustment to diagnosis, check chromosome result, refer to genetics if parents wish	Referral to EYSEND Ensure Physiotherapy follow up	Discuss cervical spine instability Application for DLA/other benefits	Hip screening if not weight bearing Discuss/refer for follow up in community patch clinic if appropriate	Should have annual health check at GP from 14y	Refer to adult Intellectual disability teams if required and ensure other adult hospital follow up if indicated

## Nottingham Guidelines for the Care of Children with Down Syndrome

### Medical Conditions that occur more commonly in people with Down Syndrome

The below table, while not exhaustive, is a summary of the medical conditions that occur more commonly in people with Down Syndrome. Awareness of these conditions allows for appropriate and thorough history and examination at each contact. For some of the conditions, further information about routine screening and management is described in this guideline where it may differ for people with Down Syndrome.

Cardiac	ENT & Respiratory	Gastrointestinal	Neurological & Developmental	Endocrine and Autoimmune
Congenital heart disease (AVSD) Pulmonary hypertension (primary or secondary)	Pneumonia OME Laryngo/tracheomalacia Conductive & sensorineural hearing loss Sleep related breathing disorders	Atresia (oesophagus, duodenum, jejunum, anus) Annular pancreas Feeding difficulty Reflux Hirschprung disease Coeliac disease	Hypotonia Alternative development schedule Infantile spasms Epilepsy Autism Mental Health Cerebrovascular Disease Dementia (adults)	Hypothyroidism Hyperthyroidism Diabetes Alopecia areata Coeliac disease Vitiligo
Orthopaedic & Rheumatology	Infection & Immunology	Haematological & Oncology	Eye	Skin
Cervical spine disorders Hip subluxation/dislocation Patellar instability Scoliosis Metatarsus varus Pes planus Inflammatory arthritis	Severe Infection Immune function abnormalities Inadequate response to vaccinations	TL-DS ML-DS Neonatal FBC abnormalities Germ Cell Tumours Retinoblastoma	Refractive errors Blepharitis Nasolacrimal obstruction Cataracts Glaucoma Nystagmus Squint Keratoconus	Dry skin Folliculitis Seborrheic dermatitis Leukaemia cutis Carotenaemia Elastosis perforans serpiginosa Vitiligo Alopecia

## Nottingham Guidelines for the Care of Children with Down Syndrome

### Review in the Neonatal Period (implemented by neonatal team)

#### Diagnosis and Disclosure

Parents will often remember the initial conversations they have with professionals, this can affect how they view the diagnosis and their baby. Confirmation of the diagnosis with genetic testing (Rapid FISH) is usually possible within 48-72 hours; confirmation of the result should not delay informing parents of the suspected diagnosis.

The language used is crucial, begin by congratulating the parents and try to refer to the baby by name. Parents appreciate honesty, and information about Down syndrome must be shared non-judgementally. It is important that this is done by a senior clinician who is able to provide information that is up-to-date and balanced, to include a positive view of what the child's future may hold as well as being realistic about some of the challenges that may be faced. The amount of information given should be based on how much the parents want to know at that stage and your own level of experience, but could include:

- Confirmation of the diagnosis and aetiology (why did it happen?)
- Likely developmental progress (emphasising the wide range)
- Immediate and, if appropriate, future health concerns
- Long term prognosis (What the future may hold)
- Local resources and plan for follow up (including contact details for local team)

The Down Syndrome Team can support these conversations and provide more details as well as written information, including information regarding support organisations and a copy of the Down syndrome insert for the Personal Child Health Record. Many families appreciate being put in touch with another family who have a child with Down syndrome. Families should leave hospital knowing when their next contact will be, and who to approach for concerns and support which can be arranged by the Down Syndrome Team.

#### Medical History and Examination

- Routine neonatal examination should pay particular attention to common complications of Down Syndrome such as bowel atresias, Hirschprung's, cardiac defects and cataracts.
- The findings should all be noted on the Down Syndrome Neonatal Discharge Form (**Appendix 1**) to be sent through to the Down Syndrome Team with the discharge letter.

#### Investigations

- **Chromosomes:** Ask for rapid FISH test first and full test as soon as possible. Two millilitres (less may be possible) in a Lithium Heparin bottle and send straight away or store in a specimens fridge if out of hours.
- **FBC:** Minor abnormalities in blood count are commonly seen and should be managed as for any other baby. Transient leukaemia of Down Syndrome (TL-DS) is reported to occur in 10% of babies with DS and may need advice from the Paediatric Haematologist (see TL-DS section below).

## Nottingham Guidelines for the Care of Children with Down Syndrome

- **Thyroid screen:** For the purpose of screening in babies with Down Syndrome, routine newborn blood spot screen is satisfactory.
- **Cardiac assessment:** This should be performed by a senior paediatric trainee or Consultant prior to the baby leaving the hospital and should include a physical examination (including pre/post ductal oxygen saturations) and an ECG. If there are concerns an inpatient Echocardiogram should be performed and the baby discussed with the cardiology team. If no concerns on examination or echocardiogram, the baby needs an echo within 6 weeks (can be as in or outpatient). See cardiology section below also. **Note:** All babies with a significant defect should be considered for RSV prophylaxis in season (discuss with cardiologist).
- **Neonatal hearing screen:** Check this has been done and record the result on the form.

### Referrals/notifications to be arranged by neonatal team

- Primary Care team (GP and Health Visitor)
- Community Midwife
- Obstetrician
- Down Syndrome Team and/or relevant local team
- Social Care in selected cases

### Initial Visit by Down Syndrome Team

#### Referrals

We will usually be contacted by the neonatal team as soon as the diagnosis is suspected and has been discussed with the parents. The baby will usually be on the neonatal unit or postnatal wards. Sometimes we are notified when they are ready to go or have gone home. We would usually try to see as soon as is mutually convenient, if possible before discharge.

#### **Information to gather prior to first visit:**

- Confirm parents ready to meet DS team, for some they may need a time for adjustment.
- Baby's details (DoB, health)
- Family's details (parents names, address)
- Was baby expected to have DS?
- What have they been told and by whom?
- Is chromosome result available and if not when will it be?
- Date, time and venue of first visit (check both parents available, if appropriate)

#### **Information to take to first visit:**

- Nottingham DS welcome pack (from CDC) which includes Personal Child Health Record (PCHR) inserts, information regarding Down Syndrome clinic and Down Syndrome Association (DSA) parents' leaflet
- New baby welcome gift bag supplied by Nottingham Down Syndrome Support group

## Nottingham Guidelines for the Care of Children with Down Syndrome

### Points to Discuss:

This may vary depending on how much parents are ready to hear at that time but discussion should follow a similar format to that described above. Further information can be found in **Appendix 2**.

## Prevention, Investigation and Management of Infections

### Introduction

Infections are more common in children with Down Syndrome. A number of factors contribute to the increased risk of infection:

- Structure of mid face
- Reflux/aspiration
- Other associated medical conditions
- Poor immune function though not necessarily demonstrable by routine immunology tests

Importantly, there is an increased risk of **serious** infections in Down Syndrome.

### General management

- **Consider 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 influenza vaccine for child ( from 6 months 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
  - Pneumovax II should not be repeated within 5 years. If pneumococcal antibodies low despite recent Pneumovax II, please discuss with Immunology
- Current research in children/adults with Down Syndrome and Covid-19 shows that older children/adults are found to be more susceptible to severe disease. Vaccination is recommended for at risk groups as per national guidance and treatments such as monoclonal antibodies may also be available. Please check latest guidance for specific antiviral and antibody treatments relating to covid-19 infection.

## Nottingham Guidelines for the Care of Children with Down Syndrome

### At 12 months

Check bloods at least 1 month after completion of 12 month routine immunisations for:

- immunoglobulins
- functional antibodies
- lymphocyte subsets

(ensure Down Syndrome is written in the clinical details so that the correct subset panel is performed).

If concerns at any review with frequent, atypical, or significant infection consider bloods as above and referral/discussion with immunology team.

### 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.

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

### Alerts

Babies born in Nottingham should have an alert put on their electronic record:

*“Down Syndrome: Risk of serious infection and atypical presentation. Senior Paeds review and low threshold for antibiotics”.*

A similar message should be added to correspondence with GP and into the red book:

*“Children with Down syndrome are more vulnerable to serious infection and may present atypically.*

*If they present with possible infection please have a low threshold for suspecting sepsis or pneumonia, and seek prompt paediatric advice.*

*Also have a low threshold for starting antibiotics and continuing for double the usual length course.*

*Please ensure all recommended immunisations are given including annual influenza and Pneumovax II after the age of 2 years.”*

## Nottingham Guidelines for the Care of Children with Down Syndrome

### Thyroid surveillance ▲

Screening is done in the newborn period as part of the routine newborn screening programme. The results of this will need to be checked and recorded. Full Thyroid Function Tests (TFTs) are only required in the neonatal period if there is a new clinical suspicion of Thyroid Disease.

Thyroid function should then be checked at 4-6m of age, 12 months of age and annually thereafter. A venous/capillary sample should include TSH, free T4 and Thyroid Peroxidase (TPO) antibodies. A dried blood spot sample includes TSH only.

A venous sample should be performed if there are any signs and symptoms of Thyroid disease. If there is a suspicion of hyperthyroidism, Thyroid Receptor Antibodies (TRABs) should also be checked.

Try to coincide surveillance TFTs with other reviews and blood tests, such as the annual review, or opportunistically if the child needs venepuncture for other reasons. Remember that illness can affect the concentrations of TSH, T4 and T3. However, if there are any signs or symptoms of Thyroid Disease, or a child is already on treatment, then earlier testing is indicated.

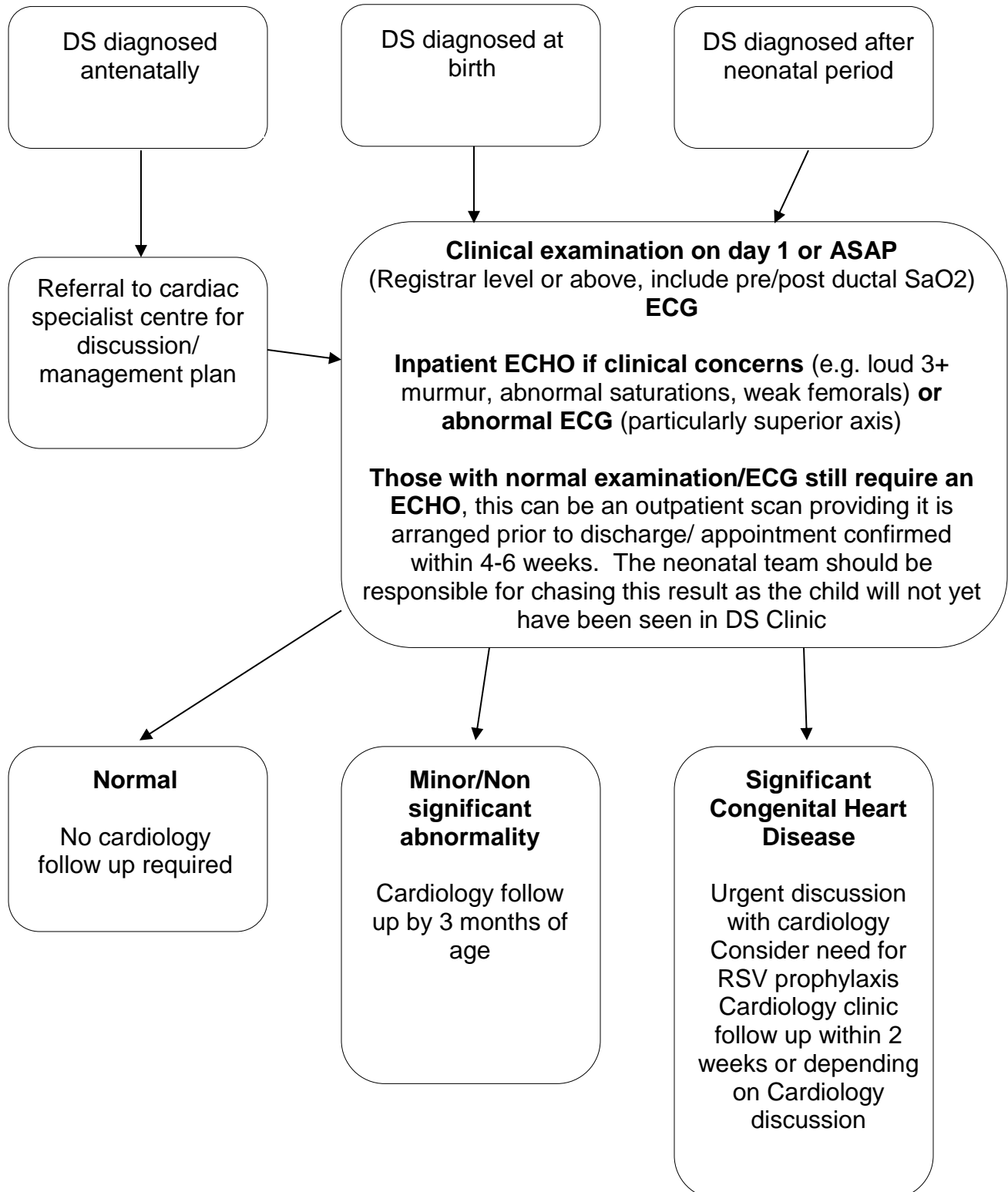
What to do with abnormal results:

Thyroid Function Result If TSH is from dried blood spot, repeat with venous blood sample		Action: At all points consider discussing with paediatrician with expertise in Endocrinology
Hypothyroid	TSH >10mU/l low free T4	Repeat venous TSH, free T4 and thyroid peroxidase (TPO) antibodies to confirm Consider initiating treatment whilst awaiting blood test results if TSH is very high and there is clinical suspicion of hypothyroidism.
Likely Hypothyroid	TSH > 10mU/l normal free T4	Repeat venous TSH, free T4 and TPO antibodies Discuss with a clinician with expertise in paediatric endocrinology
Subclinical Hypothyroid	TSH mildly raised (but < 10 mu/l) normal free T4	Under 3: Repeat TSH and free T4 test in 1-3 months*. Include thyroid peroxidase (TPO) antibodies (as baseline) Over 3: Repeat venous blood test in 6 months* (TSH, free T4 and TPO antibodies) Return to annual surveillance if three consecutive repeat tests show TSH and free T4 remain stable
Possible Central Hypothyroid	TSH normal/low Low free T4	Repeat venous blood TSH, free T4 and TPO antibodies Seek advice promptly from a paediatric endocrinologist if repeat blood test findings show that the abnormality persists- specialised investigations may be required
Raised TPO	normal TSH normal free T4 raised TPO antibodies	Less than 3: repeat in 1-3 months* 3 years and over: repeat in 6 months* Return to annual surveillance if there are no signs and/or symptoms suggestive of thyroid dysfunction
Hyperthyroid	Low TSH high free T4 clinical symptoms	Venous blood test for TSH, free T4, free T3, TPO antibodies and TSH receptor antibody (TRAB) levels Seek advice from a paediatric endocrinologist
*if any clinical concerns regarding signs/symptoms of thyroid disease, repeat bloods sooner		



## Nottingham Guidelines for the Care of Children with Down Syndrome

### Screening For Congenital Heart Disease



For further information please see cardiac guidelines from the DSMIG website.



## Nottingham Guidelines for the Care of Children with Down Syndrome

### Sleep Related Breathing Disorders

Sleep disorders affect up to 85% of children with Down syndrome, the most common being obstructive sleep apnoea (OSA) seen in 75%, compared to 1-3% of typical population. Sleep disordered breathing is more common due to craniofacial anatomy and low muscle tone. It is associated with neurocognitive difficulties including developmental delay, behavioural difficulties, tiredness, pulmonary hypertension and faltering growth. OSA can be difficult to diagnose from symptoms alone (such as restlessness, snoring, daytime tiredness) and therefore screening is recommended even in the absence of reported problems.

Symptoms to consider include that may indicate sleep disordered breathing or OSA:

- 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
- Pulmonary hypertension
- Heart failure.

Sleep studies (overnight oximetry) are recommended once in infancy and then should be considered yearly thereafter, but more frequent if concerns. Abnormal results should result in prompt referral to the ENT and/or respiratory team particularly if severe OSA is seen. There should be a low threshold for further investigation with more sophisticated respiratory investigation such as polysomnography where there is ongoing clinical concern despite normal oximetry.

## Nottingham Guidelines for the Care of Children with Down Syndrome

### Transient Leukaemia of Down Syndrome (TL-DS)

#### **Background**

5% to 30% of children with Down syndrome (DS) are born with transient leukaemia of Down syndrome (TL-DS), also known as transient abnormal myelopoiesis (TAM) and transient myeloproliferative disorder (TMD). TL-DS is the preferred terminology as TAM and TMD give a misleading impression of benignity. TL-DS is a congenital leukaemia unique to neonates with DS or mosaic trisomy 21. It is caused by a mutation in the GATA1 gene, which codes for a key haematopoietic transcription factor.

#### **Clinical Features**

No single clinical feature is entirely specific to TL-DS but there are several characteristic features seen in TL-DS which are uncommon in neonates with DS who do not have a GATA1 mutation including organomegaly, hepatopathy (raised transaminases with conjugated hyperbilirubinemia), skin rash, pericardial and pleural effusions, extreme leucocytosis, coagulopathy. Presence of any of the above features without a clear explanation should trigger early consideration of TL-DS. TL-DS should be defined as the presence of a GATA1 mutation together with a peripheral blood blast percentage >10% and/or clinical features suggestive of TL-DS in a child with Down syndrome (DS) or mosaic trisomy 21

#### **Antenatal**

If fetal USS shows features suggestive of TL-DS, fetal FBC, blood film, LFTs and GATA1 mutation analysis should be performed to confirm the diagnosis. This allows close multidisciplinary management of the pregnancy, which is likely to improve outcomes via timing of delivery and judicious use of blood product support.

#### **Postnatal**

All neonates with confirmed or suspected Down Syndrome should be examined for features suggestive of TL-DS. A full blood count and blood film should be performed in the first 3 days of life for all infants with Down Syndrome. If there is suspicion of TL-DS (blast percentage >10% and/or clinical features) should be discussed urgently with the paediatric Haematology team, additional tests should be requested to include; GATA1 mutation analysis, LFTs (including conjugated bilirubin if the baby is jaundiced), CXR, abdominal ultrasound, echocardiogram.

Any child who did not have a peripheral blood blast cell percentage performed in the first 3 days of life or in whom there was significant IUGR (when blast counts may be suppressed) should be considered to be still at risk of TL-DS in the first 4-8 weeks of life. The baby should be monitored appropriately and an FBC and blood film sent. GATA1 mutation analysis should be considered.

Babies with Down syndrome of mosaic trisomy 21 with a peripheral blast percentage  $\leq 10\%$  in the first week of life do not appear to be at risk from TL-DS, thus screening for GATA1 mutations in these babies is not routinely recommended except in those cases where the neonatal blast percentage was not assessed or is deemed unreliable

## Nottingham Guidelines for the Care of Children with Down Syndrome

### Hearing Screening and Follow up

- All children will have the newborn hearing screen and then will automatically be sent an appointment for follow-up at around 8 months. If the diagnosis of Down Syndrome is not known at this point, however, this may not happen.
- At the One Year Review it should be confirmed that the child has had a Children's Audiology appointment.
- All children with normal hearing will be called for follow-ups as below and may need referral back if discharged for any reason (e.g. non-attendance).
- The routine hearing assessment pathway for normal hearing at each stage is: 8 months, 18 months, 2 years, 2.5 years, 3 years, 3.5 years, 4 years, 4.5 years, 5 years then annual review until 16. There will be transition to a specialist Adult Audiology clinic at around 16 years old.
- Children who present with a hearing loss will be managed within audiology with onward referral to ENT as required. Children's audiology will often fit hearing aids to manage any hearing loss. For OME (otitis media with effusion) this is often a softband bone conduction hearing aid but may also be behind the ear hearing aids.

### Vision Screening and follow up

The possibility of visual problems should be considered at all ages.

- Routine neonatal examination: check for cataracts.
- At 3 and 6 month review ask the parents about and examine for visual behaviour, squint, nystagmus etc. If there are any concerns refer for ophthalmology assessment.
- Repeat above at the 1 year review.
- Blepharitis and nasolacrimal duct obstruction is common and may need referral.
- At the 2 year Review examine and refer all children to Paediatric Ophthalmologist unless they are already in the system.
- Typically the ophthalmology team will review children annually. Older children can be seen by the optician.

## **Nottingham Guidelines for the Care of Children with Down Syndrome**

### **Pre-school Speech and Language Therapy**

Some children with Down Syndrome can have difficulties with their lip and tongue movements for eating and drinking. This may be because of low muscle tone, instability of the jaw or nasal congestion. Some children are sensitive to certain textures of food.

Most children with Down Syndrome have speech difficulties and most have difficulties with their language development to varying degrees. Some children may also need help with their interaction skills.

Early referral should be made to the SLT team if there are concerns with feeding e.g. coughing/choking with food. Later referral should be made to support communication, this is most commonly done at the 1 year review depending on the individual child's level of communication.

### **Physiotherapy**

There is an open referral system to the physiotherapy service at the Child Development Centre. Babies are usually seen after their 3 month review at the Down Syndrome Children's Clinic but earlier assessment can be requested if necessary. Appointments are on a drop-in basis by parents requesting a physiotherapy review when attending the Down Syndrome Children's clinic.

At the initial appointment an assessment is undertaken and information and advice given. The majority of babies are then seen as drop-in appointments during the monthly DS Clinic. If the child has not attended for 12 months an opt in letter is sent to parents requesting them to bring the child for review. The GP and Paediatrician are notified if the child is discharged either if there are no physiotherapy related problems or if there has been no attendance when requested for review.

Some children with increased physiotherapy input requirements will be transferred to an individual caseload and seen for appointments outside of the DS Clinic.

After the age of 5 years if ongoing physiotherapy is required the child's care is transferred to the over 5's team. (Local services if from out of area.)

Hip surveillance is important in children with Down Syndrome due to the increased risk of hip subluxation and dislocation. Any child with DS not weight bearing by 2 years should have paediatric and physiotherapy review and hip x ray undertaken.

Once a child becomes eligible for an early years or preschool placement, support can be provided around transition to identify staff training needs and appropriate equipment to maximise participation and inclusion. Physiotherapy can also contribute to the statutory EHCP process if a child is eligible for assessment

## Nottingham Guidelines for the Care of Children with Down Syndrome

### APPENDIX 1

### Down Syndrome Neonatal Discharge Form

#### PATIENT DEMOGRAPHIC STICKER

Name:

DOB:

NHS/K:

Address:

Neonatal Consultant:

		Details/Result:	Follow up (date/time of appointment):
Clinical Findings	Additional diagnosis/ concerns (include other congenital anomalies)		
	Cardiac examination (including pre/post ductal SaO <sub>2</sub> )		
	Age meconium passed		
Blood Tests	Chromosome result		
	FBC		
	Thyroid (newborn blood spot adequate)		
Investigations	ECG		
	Echocardiogram		
	Hearing screening		
DS team	Seen by DS team (Phone/email to arrange)		
	Referral letter to clinic		
	GP and HV notified		
Referrals	Cardiology follow-up (if needed) RSV vaccine (if needed) Other (please detail)		

## Nottingham Guidelines for the Care of Children with Down Syndrome

### APPENDIX 2

#### Information New Parents may wish to know

Always check what parents have been told, or how much information they would like to know at that stage. For some families it may be appropriate to give limited information at the first visit with signposting for further reading, and to re-visit and offer to explain further at a later date. The information below is designed to be used as a guide and reference if required, but consultation should be guided by families.

#### **Cause of DS**

- Extra chromosome 21
- We don't know why this happens
- Can occur in pregnancies at any age, but the chance increase with maternal age
- Most babies will have standard trisomy 21, but there are a few percent with mosaicism or translocation
- The chance of having a further pregnancy with a fetus with Down syndrome for any woman is 1% but may be higher if there is a translocation or if mum is older than her early 40's
- The possibility of genetic counselling prior to the next pregnancy and early antenatal testing if the parents wish.

#### **Features of DS**

- Physical appearance- this is only really important because it is usually why we first suspect that the baby has DS
  - Usually not necessary to point out the feature of DS, but may be necessary if not previously done or the parents ask
  - Babies with DS do NOT all look exactly the same and DO also resemble their families
  - Appearance is not linked with ability!
  - People with DS are generally smaller than the general population
- Associated medical problems
  - Point out that there is a long list of things that are more common in DS but these are the same sort of things that other people can get AND nobody with DS gets all of them – some people with DS are very healthy
  - Diagnoses that may cause significant concern in the neonatal period or that they will have screening for in next few days if not done already
    - Heart
    - Gastrointestinal atresias/ Hirschprung's
    - Vision - cataracts or nystagmus
    - Hearing
  - Other problems that are more common
    - Increased susceptibility to infection - stress the importance of routine immunisations
    - Thyroid disorder
    - Bowel problems- Hirschprung's, coeliac disease, constipation

## Nottingham Guidelines for the Care of Children with Down Syndrome

- Hearing - mainly otitis media with effusion (glue ear)
  - Vision - squints, myopia, hypermetropia
  - Minor problems - dry skin, thin hair, blepharitis
- Suggested programme of medical surveillance
  - Wherever they are followed up this should follow a schedule with regular check ups to screen for problems that are more common - refer to the suggested schedule of checks in PCHR which is in their welcome pack
- Development and Learning disability
  - All children with DS will have a intellectual disability BUT there is a huge range of abilities- see “ variability” below
  - General development is along the same lines as other children, but at a slower rate - refer to developmental chart in PCHR of typical and ranges of milestones
  - Babies with DS are often floppy and their motor progress slower
  - Typical pattern of strengths and weakness - stronger in visual learning, weaker with auditory processing
  - Language is often the biggest problem - combination of factors including overall learning ability, auditory processing and oromotor skills

### Variability

- Point out that DS is a diverse condition and that there is a wide range of ability. Some brighter individuals with DS overlap with the lower end of the typical range with only minor difficulties, whilst some have very severe disability, most fall somewhere in between.
- Information about typical abilities that they may come across will be very dependant on where it comes from:
  - Different countries/societies have different attitudes and approaches which will influence outcomes
  - Things have changed enormously over past few decades, and children brought up in the UK today may well be healthier and do better than those who are currently adults

### Prognosis as an adult

- Repeat the variability!
- Most adults with DS are mobile, active and are able to manage their own basic self-care needs
- Most can communicate sufficiently to make their needs known – many are quite articulate, but for some it is an area of more difficulty
- Some will require support with daily living
- Most will require support throughout their lives with finance, housing, employment etc.
- Some are able to live independently or semi-independently



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**Services available** – Note that this information is relevant for Nottingham residents only – if they are out of the area explain the service we offer and that there will be similar services in their local area that we can refer them on to.

- **Health**
  - Nottingham DS clinic
  - Nottingham DS guidelines can be followed in other hospital or Community Paediatric Clinic if they choose not to come to the DS clinic
- **Therapy** - they will meet the physiotherapy team at the DS clinic who will see when, and if, needed and will be referred to SLT early for feeding concerns, otherwise later on for communication support
- **Education**
  - Will have special educational needs
  - Most children with DS go to mainstream school with support
  - Some will go to a special school
- **Social Services**
  - The family would be entitled to support as a “child in need” if necessary
- **Benefits**
  - Will be entitled to DLA at some point
  - They may get turned down in the first few months if there are no obvious additional needs



## Nottingham Guidelines for the Care of Children with Down Syndrome

### APPENDIX 3

<b>AFFIX ADDRESSOGRAPH LABEL HERE</b>  Name:  DOB:  Hospital/NHS Number:  Address:	<b>Date:</b>  <b>Age:</b>  <b>Height:</b>  <b>Weight:</b>  <b>Head Circumference:</b> (plot on DS growth chart & red book)
<b>Medical Problems:</b>     	<b>Allergies:</b>  <b>Medications:</b>    <b>Routine Immunisations:</b> Up to Date Y / N RSV: Y / N / NA      Flu Vaccine: Y / N / NA Pneumovax: Y / N / NA
<b>Attended with:</b> <b>Parental Concerns:</b>    	
<b>Respiratory Problems/ Infections/Symptoms of sleep related upper airway obstruction:</b> <b>Last sleep study/Result:</b>   	
<b>Cardiac Problems/Result of ECHO:</b>   	
<b>Feeding/Bowels/Reflux/Vomiting Symptoms:</b>   	
<b>Development (review all 4 domains):</b> <b>Hearing/Vision (any parental concerns):</b> Result of Newborn hearing screen/Last hearing appointment   	

## Nottingham Guidelines for the Care of Children with Down Syndrome

<b>Other:</b>																											
<table style="width: 100%; border: none;"> <tr> <td style="width: 60%; vertical-align: top; padding: 5px;"> <b>Examination:</b>  <b>Cardiovascular:</b>   <b>Respiratory:</b>   <b>ENT:</b>   <b>GI/Genitalia:</b> </td> <td style="width: 40%; vertical-align: top; padding: 5px;"> <b>Eyes:</b>   <b>Development/Neurology:</b> </td> </tr> </table>		<b>Examination:</b> <b>Cardiovascular:</b>  <b>Respiratory:</b>  <b>ENT:</b>  <b>GI/Genitalia:</b>	<b>Eyes:</b>  <b>Development/Neurology:</b>																								
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<b>Signed:</b>																											

## Nottingham Guidelines for the Care of Children with Down Syndrome

### References and Further information

#### **Personal Child Health Record for Babies with Down Syndrome**

DSMIG (UK) (2020) Insert for Personal Child Health Record for babies born with Down Syndrome (5<sup>th</sup> edition). DSMIG, UK (available from Harlow Printing, NE33 4PU, UK)

#### **Further reading article overview**

Michael A, Marder E. 2021. Keeping Children With Down Syndrome Healthy. Paediatrics and Child Health. 31(9):340-346

#### **Further reading textbook (Professional reference)**

Newton R, Puri S, Marder L. Down's Syndrome Current Perspectives. Mackeith Press. (2015)

## Nottingham Guidelines for the Care of Children with Down Syndrome

	<p><b>Down Syndrome Medical Interest Group</b> Guidelines for Essential Medical Surveillance, Keypoint Series and Clinical Awareness Notes. Available online at: <a href="https://www.dsmig.org.uk/information-resources/guidance-for-essential-medical-surveillance/">https://www.dsmig.org.uk/information-resources/guidance-for-essential-medical-surveillance/</a></p>
	<p><b>Down Syndrome Association</b> Parent support, and information on range of issues including health , education and legal issues, useful leaflets Website <a href="http://www.downs-syndrome.org.uk">www.downs-syndrome.org.uk</a></p>
	<p><b>Down's Heart Group</b> Parent support group – useful information leaflets on cardiac issues Website: <a href="http://www.dhg.org.uk">www.dhg.org.uk</a></p>
	<p><b>Nottingham Down Syndrome Parent support group</b> Support, advice, events, activities and meet ups Website : <a href="http://www.ndssg.org.uk">www.ndssg.org.uk</a> Facebook or email: <a href="mailto:nottinghamshiredsgroup@outlook.com">nottinghamshiredsgroup@outlook.com</a></p>