

# Down Syndrome

<b>Title of Guideline (must include the word "Guideline" (not protocol, policy, procedure etc)</b>		<b>Guideline for the Management of Children with Down Syndrome</b>
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<b>Directorate &amp; Speciality</b>		Family Health, Paediatrics Speciality: General
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<b>Explicit definition of patient group to which it applies (e.g. inclusion and exclusion criteria, diagnosis)</b>		Children and young people aged 0-18 years with Down Syndrome
<b>Abstract</b>		
<b>Key Words</b>		Children; Young People; Paediatrics; Down Syndrome; trisomy 21
<b>Statement of the evidence base of the guideline – has the guideline been peer reviewed by colleagues?</b>		Peer reviewed by Paediatric clinical guidelines development group, Neonatology guidelines group and relevant specialists Also based on Evidence based guidelines developed by Down Syndrome Medical Interest Group (UK and Ireland)
<b>Evidence base: (1-5)</b>		4
<b>1a</b>	<b>meta analysis of randomised controlled trials</b>	
<b>1b</b>	at least one randomised controlled trial	
<b>2a</b>	at least one well-designed controlled study without randomisation	
<b>2b</b>	at least one other type of well-designed quasi-experimental study	
<b>3</b>	well –designed non-experimental descriptive studies (ie comparative / correlation and case studies)	
<b>4</b>	expert committee reports or opinions and / or clinical experiences of respected authorities	
<b>5</b>	recommended best practise based on the clinical experience of the guideline developer	
<b>Consultation Process</b>		Paediatric Clinical Guidelines Group
<b>Target audience</b>		Clinicians and healthcare professionals caring for children and young people with Down Syndrome
<p><b>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.</b></p>		

# Nottingham Guidelines for the Management of Children with Down Syndrome

## Document Control

### Document Amendment Record

Version	Issue Date	Author
V1	October 2013	Dr. Liz Marder Consultant Paediatrician Dr Claire McCall Associate Specialist
V2	December 2016	Dr Liz Marder

### General Notes:

#### Summary of changes for new version:

Changes to this iteration of the guideline( in the end it was really relatively minor

- Updated names and contact details of personnel and services
- Updated references and weblinks
- Additional information to the guidelines for the neonatal period in line with New (2017) DSMIG neonatal guidance on feeding , weight , TAM and genetic testing
- New thyroid screening flow chart

#### No major changes to this version

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### Statement of Compliance with Child Health Guidelines SOP

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This guideline has had only minor changes made and therefore this version has not been circulated to all for review. A previous version had been approved by circulation to senior team members.

Martin Hewitt  
Clinical Guideline Lead  
10.03.2017

# Nottingham Guidelines for the Management of Children with Down Syndrome

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# Nottingham Guidelines for the Management of Children with Down Syndrome

## Introduction

These guidelines have been written to describe what we consider to be the appropriate medical management for children with Down Syndrome (DS). In Nottingham the service is focused around the Down Syndrome Children's Clinic, and this is the programme we aim to offer there. Some families, however, prefer not to come to the clinic and are seen elsewhere, usually by a hospital-based Paediatrician. Also, children of school age are often more conveniently and appropriately seen by their local Community Paediatrician who is linked to their school and other local resources.

These guidelines suggest the appropriate management, referrals, tests, etc. at each stage, from prenatal diagnosis, neonatal care and throughout childhood and adolescence. The information within them should mean that they 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 Children's Service Team** :-

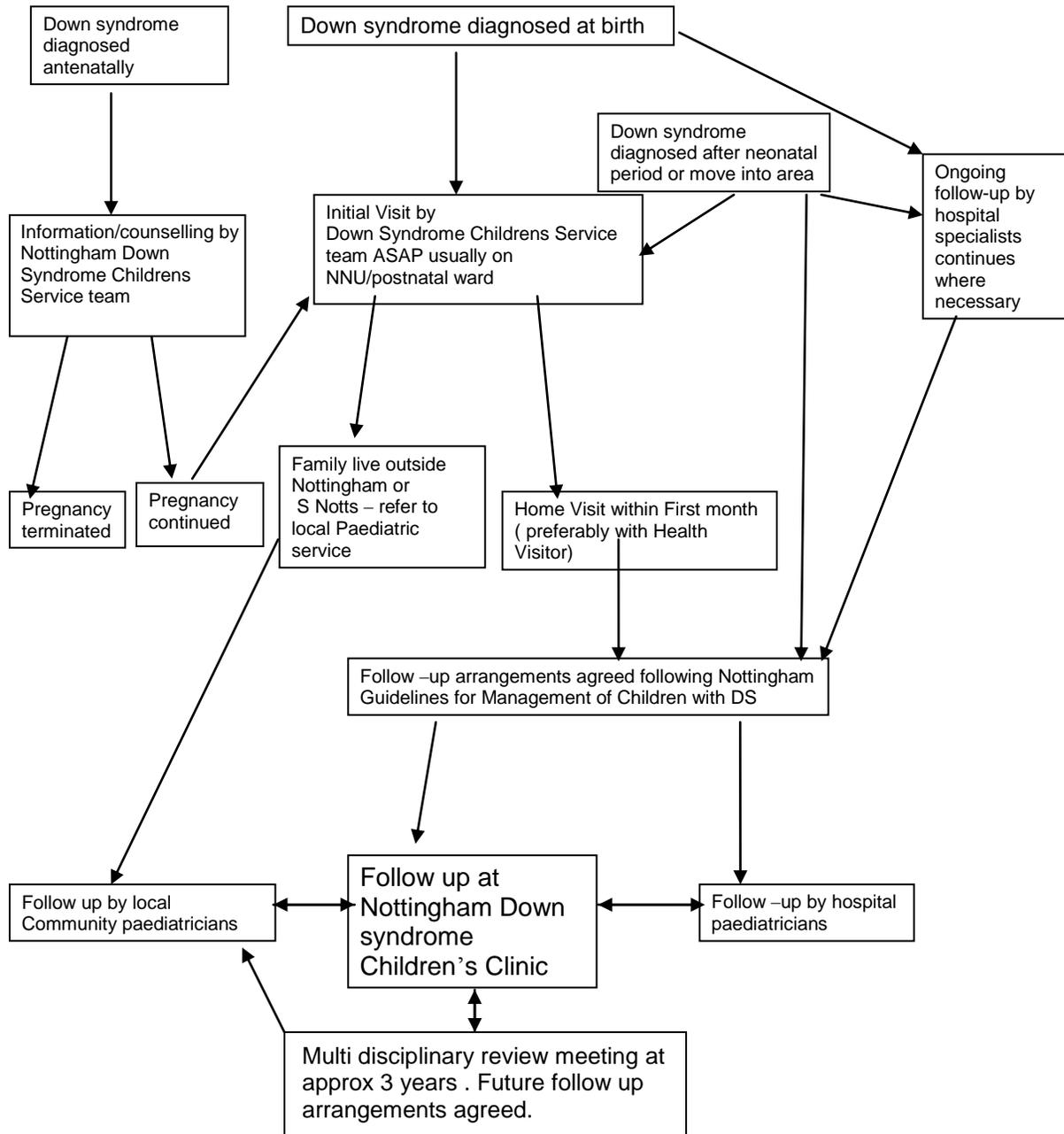
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# Nottingham Guidelines for the Management of Children with Down Syndrome

## Overview of Nottingham Down Syndrome Children's Service



# Nottingham Guidelines for the Management of Children with Down Syndrome

## Overview of Nottingham Down Syndrome Children's Service

### Antenatal

- If a diagnosis is made at this stage, parents may be offered a referral to the Down Syndrome Team 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 A**) to accompany the discharge letter.
- Referral to the Down Syndrome Team who will make an initial visit as soon as appropriate (see **Contact Details** on page 2).

### First year

- The family are invited to attend the specialist clinic for children with Down Syndrome as soon as they feel able to attend. This clinic, which takes place once a month, has open access. Children are asked to come at aged 3 months, 6 months and one year for a specific review. They will only receive a specific invitation by letter if they have not attended around the due time.
- Referrals and introductions will be made to the other members of the Down Syndrome service.
- Continued hospital follow-up continues at the discretion of the hospital consultant, but generally only if medically needed.

### Pre- school

- Annual review at the Down Syndrome clinic.
- Multi-agency review /Team around the Child meeting arranged around age 3 years if not done before.
- Transfer to local Community Paediatric team usually at the 4<sup>th</sup> year review.

### School age

- Annual review by local Community Paediatric team.
- Attendance at transition review in Year 9.

### School leavers

- Review by Community Paediatrician and preparation of a comprehensive report.
- Introduction to Adult Learning Disabilities Team where appropriate

# Nottingham Guidelines for the Management of Children with Down Syndrome

## Guidelines for the Neonatal Period (implemented by neonatal team)

### Diagnosis and Disclosure

- Parents should be told of the diagnosis as soon as possible, preferably by a Senior Paediatrician and by the responsible Consultant as soon as is feasible (please see **Appendix B**).
- Chromosome analysis should be requested urgently, but disclosure of the likely diagnosis should not be delayed to wait for the results.
- The Down Syndrome Team can be contacted at this stage and are happy to join the hospital staff in explaining the diagnosis if practicable. A team member will always try and visit the parents prior to discharge (see **Contact Details** on page 2).

### 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 A**) 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 abnormal myelopoiesis (TAM) is reported to occur in 10% of babies with DS and may need advice from the Paediatric Haematologist.
- **Thyroid screen:** Routine newborn screen is satisfactory, but please chase the result and record it.
- **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 and an **Echocardiogram** if available (if not, must have an **ECG and pre- and post-ductal O<sub>2</sub> saturations**). Assessment should follow the cardiac guidelines (**Appendix C**). **Note:** All babies with a significant left to right shunt should be referred for RSV prophylaxis in season.
- **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 (see **Contact Details** on page 2)
- Social Care in selected cases

# Nottingham Guidelines for the Management of Children with Down Syndrome

## Initial Visits - Down Syndrome Team Guidelines

### First Visit by Community Paediatrician of the Down Syndrome Children's Service Team

- This is usually done in the hospital but if the baby is discharged early it may be arranged at home.

The purpose is to give the parents:

- Written and verbal information about Down Syndrome.
- Information about Nottingham Down Syndrome Children's service.
- An opportunity to talk about the diagnosis and to ask questions.
- An opportunity to agree a plan for follow-up for Nottingham families, or to discuss referral to the appropriate local service for families living outside of Nottingham.

Please see attached guidelines for detailed guidance on the initial visit (**Appendices B and D**).

### Follow- up Visit (within the first month)

- This usually takes place at home, but may need to be in the hospital.
- The Community Paediatrician may be accompanied by colleagues from the Child Development Team and/or the Health Visitor.
- General discussion and provision of further information regarding Down Syndrome and the locally available services (e.g. Children's Centre Baby Group).
- Ensure that cardiac status is checked and that all babies with significant left to right shunt have been referred for RSV prophylaxis in season.
- The specific Down Syndrome literature from the DoH Early Support materials (**Blue Box**) to be given if this was not given at the first visit (**Appendix K**).
- Offer opportunity to meet other families.
- If the child is likely to have a prolonged hospital stay, refer to the Children's Centre Play Team.
- **Note: A letter to the GP and Health Visitor should be written during these two weeks.**

# Nottingham Guidelines for the Management of Children with Down Syndrome

## First Year Reviews - Down Syndrome Team Guidelines

### Three Month Review

This usually takes place at the Down Syndrome Children's Clinic at the CDC.

#### Discussion:

- **Take a full history**
- Feeding, particularly ask about GOR symptoms and treat according to guidelines
- Check whether there are any bowel problems
- Cardiac assessment, including checking ECHO reported by a Paediatric Cardiologist or referral to Paediatric Cardiology if necessary (**Appendix C**)
- Hearing and Vision (**Appendices E and F**)
- Any unusual or recurrent infections (**Appendix I**)
- Discuss immunisation – encourage routine primary immunisation
- Family adjustment to the diagnosis
- Chromosome result and recurrence risk

#### Examination:

- General physical examination, with particular attention to cardiac status.
- Eyes examination to look for cataracts, squint, nystagmus and visual behaviour.
- Plot growth on the Down Syndrome chart in records and in the Red Book.
- Development

#### Referrals:

- Verbal notification to the Physiotherapist who is part of the Down Syndrome service and recommend initial assessment to parents.
- discuss Disability Living Allowance (DLA) (note that this is often more successful when applied for at 6 months but it is good to start the process)
- Referral to Speech Therapy only if there are concerns with feeding
- Genetic counselling if parents wish

# Nottingham Guidelines for the Management of Children with Down Syndrome

## Six Month Review

### Discussion and Examination:

- As for 3 months
- Discuss immunisation – encourage routine primary immunisation plus advise annual influenza vaccine

### Referrals:

- Check that the newborn hearing screen results are known and follow-up organised. Write a letter to Child Hearing Assessment Centre (CHAC) to ensure that an appointment is in the system (**Appendix E**)
- Speech and Language Therapy referral by letter and explain to the parent/carer that they will normally be seen by the time the child is 9 months old (**Appendix G**)
- Pre-school education team/ Inclusive Education team and Educational Psychologist for the child's area
- Ensure physiotherapy input if any particular concerns about motor development including extreme hypotonia.

# Nottingham Guidelines for the Management of Children with Down Syndrome

## Pre-School Reviews - Down Syndrome Team Guidelines

### One Year Review

#### Discussion:

- Parental concerns
- 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 (**Appendix I**)
- Sleep-related upper airway obstruction. If clinical symptoms are present refer to ENT. Arrange for those who are asymptomatic to have a sleep study at 12 months of age according to the guidelines (**Appendix H**).
- Behaviour
- Therapy and educational input
- DLA and other benefits
- Cervical spine and atlanto-axial instability and give information leaflet (please see DSMIG guidelines)
- Discuss immunisation – encourage routine primary immunisation plus annual influenza vaccine

#### Examination:

Full clinical examination with particular attention to the following:

- Growth - Plot on the Down Syndrome charts and also the child's Red book
- Cardiovascular
- Neurological - Look for any signs of cervical spine instability or cord compression
- ENT - Signs of middle ear disease or upper airway obstruction (**Appendices E and H**)
- Eyes - Squint, cataract, nystagmus or blepharitis. Nasolacrimal duct obstruction sometimes needs referral.

#### Investigations:

- Audiological assessment by the CHAC team should be carried out at least once a year but more often as needed
- Thyroid function tests (T4, TSH and thyroid autoantibodies). If a finger- prick/ spot test is done for TSH only this must be repeated yearly.
- Check Immune function ( at least 4 weeks after completion of primary immunisation course) (**Appendix I**)

# Nottingham Guidelines for the Management of Children with Down Syndrome

## Two Year Review

### Discussion:

- Parental concerns
- 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 (**Appendix I**)
- Sleep-related upper airway obstruction. If clinical symptoms are present refer to ENT and/or repeat sleep study
- Behaviour
- Therapy and educational input
- DLA and other benefits
- Cervical spine and atlanto-axial instability
- Discuss immunisation – advise re: annual influenza vaccine and
- Pneumovax II if child in “ at risk group” for pneumococcal infection ( see **Appendix I**)

### Examination:

Full clinical examination with particular attention to the following:

- Growth - Plot on the Down Syndrome charts and also the child’s Red book
- Cardiovascular
- Neurological - Look for any signs of cervical spine instability or cord compression
- ENT - Signs of middle ear disease or upper airway obstruction (**Appendices E and H**)
- Eyes - Squint, cataract, nystagmus or blepharitis. Nasolacrimal duct obstruction sometimes needs referral.

### Investigations:

- Audiological assessment by the CHAC team should be carried out at least once a year but more often as needed
- Thyroid function tests (T4, TSH and thyroid autoantibodies). If a finger- prick/ spot test is done for TSH only this must be repeated yearly.
- Ensure Child known to Inclusive Education team/Educational psychologist
- Referral to Paediatric ophthalmology. Most children can be seen at the Children’s Centre (**Appendix F**)
- Hip screening – for any child not yet weight bearing request Hip x-ray and ensure physiotherapy input.

# Nottingham Guidelines for the Management of Children with Down Syndrome

## Three Year Review

### Discussion:

- Parental concerns
- 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 (**Appendix I**)
- Sleep-related upper airway obstruction. If clinical symptoms are present repeat sleep study and/or refer to ENT.
- Behaviour
- Therapy and educational input
- DLA and other benefits
- Cervical spine and atlanto-axial instability
- Discuss immunisation – advise re: annual influenza vaccine and Pneumovax II if child in “ at risk group” for pneumococcal infection ( see **Appendix I**)

### Examination:

Full clinical examination with particular attention to the following:

- Growth - Plot on the Down Syndrome charts and also the child's Red book
- Cardiovascular
- Neurological - Look for any signs of cervical spine instability or cord compression
- ENT - Signs of middle ear disease or upper airway obstruction (**Appendices E and H**)
- Eyes - Squint, cataract, nystagmus or blepharitis. Nasolacrimal duct obstruction sometimes needs referral.

### Investigations:

- Audiological assessment by the CHAC team should be carried out at least once a year but more often as needed
- Thyroid function tests (T4, TSH and thyroid autoantibodies). If a finger- prick/ spot test is done for TSH only this must be repeated yearly.
- Multi-agency review meeting ( Team around the child) arranged around the third birthday to include all agencies that are, and will be, involved with the child. Will often be CAF/TAC meeting already ongoing. if not Down Syndrome clinic team to consider need and arrange. Invite local Community Paediatrician.
- Ensure Local Community Paediatrician has been notified about child.

# Nottingham Guidelines for the Management of Children with Down Syndrome

## Four Year Review

- As for Three Year Review
- Sleep-related upper airway obstruction. Asymptomatic children to have a sleep study arranged (**Appendix H**). If clinical symptoms are present also consider referral to ENT.
- Discuss future follow-up with parents and refer to Local Community Paediatrician if appropriate or continue annual reviews at the Down Syndrome Clinic.

## Five and Subsequent Year Reviews

- The 5<sup>th</sup> and subsequent annual reviews are usually best carried out by the local paediatrician for the child's school. This is discussed with the parent/carer.
- Children may continue to be followed up for routine reviews at the Down Syndrome Children's Clinic if felt more appropriate,
- Referral back to the Down Syndrome clinic may be made for specific clinical problems.

# Nottingham Guidelines for the Management of Children with Down Syndrome

## School Age Reviews - Down Syndrome Team Guidelines

### Annual Review

#### Discussion:

- Parental concerns
- Developmental progress
- General health focusing on symptoms of disease known to be more common in Down Syndrome, in particular coeliac disease which may present atypically
- Any unusual or recurrent infections (**Appendix I**)
- Behaviour
- Dental care
- Menarche and menstrual management
- Therapy and educational input
- Advice regarding cervical spine instability (a useful resource for screening prior to participation in competitive sport can be found at <http://www.dsmig.org.uk/library/articles/Atlanto%20Axial%20-%20British%20Gymnastics.pdf> )
- Check benefits e.g. mobility bus pass
- Check access to other services e.g. sport, clubs (refer to Information library at CDC)
- Discuss immunisation – advise re: annual influenza vaccine and Pneumovax II if child in “ at risk group” for pneumococcal infection ( see **Appendix I**)

#### Examination:

- Growth - Plot on Down Syndrome chart and Red Book (according to guidelines plot on BMI charts if child is on or above the 75<sup>th</sup> centile for weight)
- Neurology - look for any signs of cervical spine instability or cord compression
- ENT - Middle ear disease and obstructive sleep apnoea
- Eyes - squint, cataract, blepharitis and keratoconus
- Cardiac status
- Orthopaedic problems e.g. foot posture or scoliosis

#### Investigations:

- Audiology to be carried out yearly by CHAC which should be automatic, unless the family repeatedly DNA. If so, keep referring.
- Yearly Ophthalmological assessment. If the Child has not had any visual problems annual vision check by school nurse or optician is sufficient.
- Thyroid function tests (T4, TSH and thyroid autoantibodies) every two years, more frequently if any suspicion of thyroid disease. If a finger prick/spot test is done for TSH only this must be repeated yearly.

# Nottingham Guidelines for the Management of Children with Down Syndrome

## School Leaver Review - Down Syndrome Team Guidelines

### School Leaving Guidelines

- As for Annual Review

#### **Discussion:**

- Review 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

#### **Referrals:**

- Transition co-ordination Social Care Team if mental health issues present
- Adult Learning Disability Team, where necessary
- GP for ongoing surveillance

#### **Investigations:**

- Audiology
- Vision
- Thyroid function
- Echocardiogram (A repeat is now recommended in early adult life, because of the high incidence of mitral valve prolapse and aortic regurgitation in adults with Down Syndrome who are asymptomatic)

# Nottingham Guidelines for the Management of Children with Down Syndrome

Appendix A

## The Down Syndrome Neonatal Discharge Form

When completed please send with discharge summary to:

Dr Liz Marder  
Consultant Paediatrician  
Nottingham Down Syndrome Children's Service  
The Children's Centre  
City Hospital Campus  
Nottingham  
NG5 1PB

<b>Name:</b>	<b>DoB:</b>	<b>Hospital Number:</b>	<b>Consultant:</b>
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	Date:	Organised/ performed by:	Result/Details:	Follow up arranged:
<b>Clinical findings</b> <ul style="list-style-type: none"> <li>• Cardiac examination</li> <li>• Age at which meconium passed</li> <li>• Other major abnormalities</li> </ul>				
<b>Blood tests</b> <ul style="list-style-type: none"> <li>• Chromosomes</li> <li>• FBC</li> <li>• Thyroid (newborn screen)</li> </ul>				
<b>Referral to DS service</b> <ul style="list-style-type: none"> <li>• Telephone call</li> <li>• Letter</li> </ul>				
<b>Investigations</b> <ul style="list-style-type: none"> <li>• Echocardiogram</li> <li>• RSV vaccine (if needed)</li> <li>• Cardiology follow-up (if needed)</li> </ul>				
<b>Hearing screening</b>				
<b>GP and HV notified</b>				
<b>Other referrals made</b> <ul style="list-style-type: none"> <li>a)</li> <li>b)</li> <li>c)</li> </ul>				

# Nottingham Guidelines for the Management of Children with Down Syndrome

## Appendix B

### Sharing news with Families

Please see document “**Working with families affected by a disability or health condition**” published by Contact A Family

Particularly the information sheets:

- “**Good practice in sharing news**”
- “**Support for fathers, siblings and grandparents**”

These documents are available online [www.cafamily.org.uk/HealthSupportPack.pdf](http://www.cafamily.org.uk/HealthSupportPack.pdf) and also from the **Resource Library** at the Children’s Centre, City Hospital. Please speak to Joyce Judson (Tel: 0115 8831100)

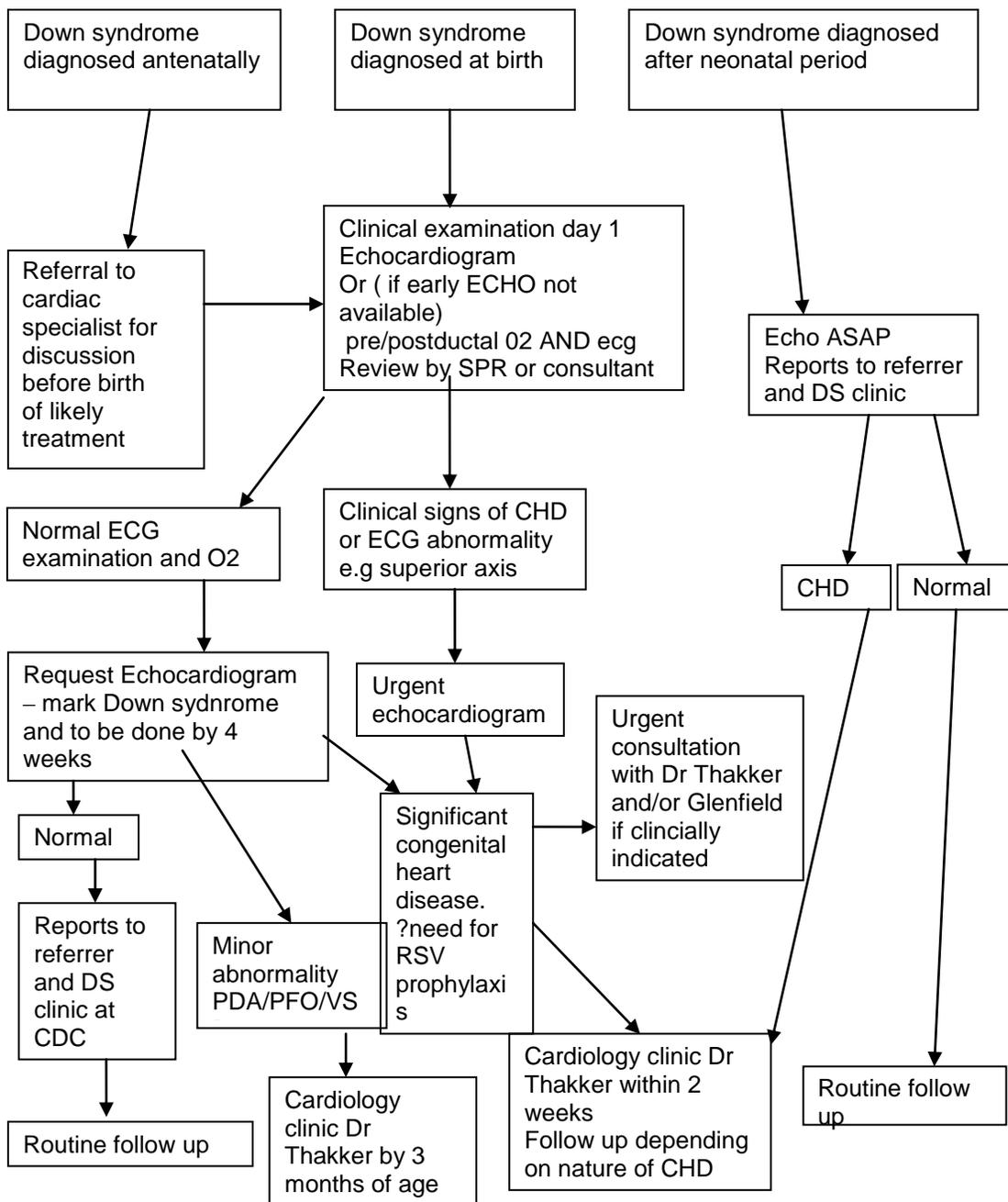
Also helpful:

**Contact A Family information pack “About diagnosis: for families and disabled children”**. This is also available at the Resource Library and online ([www.cafamily.org.uk](http://www.cafamily.org.uk))

# Nottingham Guidelines for the Management of Children with Down Syndrome

## Appendix C

### Screening for Congenital Heart Disease in Children with Down Syndrome



Note: For further information please see Cardiac guidelines from the DSMIG [www.dsmig.org.uk](http://www.dsmig.org.uk)

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

# Nottingham Guidelines for the Management of Children with Down Syndrome

## Appendix D

### Guidelines for Initial Visit

#### 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!
- 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 (get from CDC – Mary Lane) which includes Personal Child Health Record (PCHR) inserts, information regarding Down Syndrome clinic and Down Syndrome Association (DSA) parents' leaflet
- Parents folder if available (we have a few loan copies but may be out)
- DoH Early Start materials - Down Syndrome booklet (CDC Information Office)

Also sometimes, but usually wait until subsequent visits:

- DoH Early Start materials - Developmental manual (CDC Information Office)

#### **Setting up the visit:**

- Ensure both are parents available, if appropriate
- Ask if they wish their named nurse to be present
- See in private: side room on ward or parents room on NNU. Have baby present if possible
- If baby needs to stay on NNU ask if you can meet the baby first

#### **Introduction:**

- Congratulate them on the birth of their baby
- Introduce yourself as a doctor from the team that runs the DS Children's Service and clinic (or our stand in!)
- Explain your role as giving them further information about Down Syndrome, issues they may face and services available to support them and the baby
- Find out:
  - If they had been expecting baby to have DS
  - What they already know
  - Have they yet been given anything to read?
  - Do they have any personal experience of people with DS?
  - It is sometimes useful to ask their own professional backgrounds as this may help you pitch your talk at the right level

# Nottingham Guidelines for the Management of Children with Down Syndrome

- How has receiving the news been for them?
- How much do they want to hear now?

## Points to Discuss:

This may vary depending on how much parents are ready to hear at that time

### • Cause of DS

- Extra chromosome 21
- We don't know why this happens
- It can occur at any age but risk increases with maternal age
- Most cases are standard trisomy 21, but there are a few percent with mosaicism or translocation
- The recurrence risk 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

### • 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 necessarily 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
    - Hearing - mainly otitis media with effusion (glue ear)
    - Vision - squints, myopia, hypermetropia
    - Minor problems - dry skin, thin hair, blepharitis
  - Suggested programme of medical surveillance

# Nottingham Guidelines for the Management of Children with Down Syndrome

- 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 learning 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, but 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
- **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
      - Details in the welcome pack,
      - 1st Wednesday of the month at CDC

# Nottingham Guidelines for the Management of Children with Down Syndrome

- Drop-in clinic, they are welcome any time
- First formal visit at around 3 months
- Nottingham DS guidelines can be followed in ordinary 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 SALT by 9 months unless there are feeding concerns when they may be seen earlier
- **Education**
  - Will have special educational needs
  - Referral to Preschool services at 6 or 12 months (differs in City/County)
  - Most children with DS go to mainstream school with support
  - Some will have statutory assessment of Special Educational Needs or 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
  - HV or DS clinic team will help with application
- **Check neonatal screening is done**

Neonatal team should have arranged the following investigations and completed the form (**Appendix A**). Please check they have been done!

- Echocardiogram done and reported by a Paediatric Cardiologist (follow up arranged if necessary) OR ECG and pre- and post-ductal O<sub>2</sub> saturations checked with appointment for echocardiogram made for 2 weeks (**Appendix C**)
- Hearing screen
- Karyotype and, if parents not aware of result, when and how this will be communicated with them
- GP and Health Visitor informed
- **Follow-up arrangements**
  - Offer to see family again in hospital if there is likely to be a prolonged admission
  - Offer a home visit, preferably with the Health Visitor in approximately 2 weeks (and if so contact the HV to arrange)
  - Check if the neonatal clinic plan to follow up and if there are any planned appointments at other clinics e.g. cardiac, ophthalmology, hip clinic
  - Ask the family if they would like to meet another family of a child with DS
  - Invite to DS clinic as soon as the family wish, but with a formal appointment at 3 months of age

# Nottingham Guidelines for the Management of Children with Down Syndrome

- If family do not wish to attend DS clinic discuss and arrange suitable Hospital or Community General Paediatric clinic, with an appointment for 3 months of age

## Appendix E

### Hearing Guidelines for Children with Down Syndrome

- All children will have the newborn screening and then those with known Down Syndrome will automatically be sent an appointment for follow-up at 8 months. If the diagnosis of Down Syndrome is not known at this point, however, this may not happen.
- At the **Six Month Review** please refer to CHAC to check that the child is in the system and will be sent an appointment by 8 months.
- At the **One Year Review** it should be confirmed that the child has had a CHAC appointment and the report is in the notes.
- All children will be called for yearly follow-ups BUT if the child DNAs they may be discharged according to protocol. The Paediatrician will be informed with a request for further referral.

As discussed and agreed with  
Head of Service CHAC, Claire Benton

Note: - For further information please see hearing guidelines from the DSMIG  
[www.dsmig.org.uk](http://www.dsmig.org.uk)

# Nottingham Guidelines for the Management of Children with Down Syndrome

## Appendix F

### Vision Guidelines for Children with Down Syndrome

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 visual behaviour, squint, nystagmus etc. Examine for squint, nystagmus, cataracts and visual behaviour. If there are any concerns refer for ophthalmology assessment.
- Repeat above at the **One Year Review**. Treat and give advice as necessary e.g. blepharitis. Nasolacrimal duct obstruction is common and may need referral.
- At the **Second Year Review** examine and refer all children to Paediatric Ophthalmologist unless they are already in the system. They will usually see children with Down syndrome at the Children's Centre.
- All children should automatically then see ophthalmology team yearly.
- Older children can be seen by the optician.
- Routine follow-up yearly throughout life.
- Corneal disease (i.e. keratoconus) is diagnosed by slit lamp examination. However it may be picked up in the early stages by decreasing visual acuity due to astigmatism.

Note: - For further information please see Vision guidelines from the DSMIG [www.dsmig.org.uk](http://www.dsmig.org.uk)

# Nottingham Guidelines for the Management of Children with Down Syndrome

## Appendix G

### Pre-school Speech and Language Therapy Service in Nottingham for Children with Down Syndrome

#### **What do Speech and Language Therapists do?**

We assess and support the needs of children who have communication or eating and drinking difficulties.

#### **When does my child have access to the Speech and Language Therapist?**

We operate an open referral system which means that you can refer your child to Speech and Language Therapy at any time, or a professional can do it on your behalf. Usually the paediatrician will refer to the SLT service after their 6 month check, or sooner if there are eating and drinking difficulties requiring a SLT involvement

#### **How does Speech and Language Therapy help my child's eating and drinking?**

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( floppy muscles), instability of the jaw or nasal congestion. Some children are sensitive to certain textures of food. We can advise on positioning for mealtimes, utensils, appropriate textures of the food for your child, and strategies to help children with their eating and drinking.

#### **How does Speech and Language Therapy help my child's communication?**

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.

Here are a few examples of how we can help:

- Early Play ideas: By sharing information about strategies which help children learn to interact and actively communicate, for example, following their lead, adding language to their play, using repetitive rhymes and songs.
- By looking at ways of creating opportunities for communication e.g. by offering choices, by giving them enough time, by using pauses during games, by playing interactive games.
- Making communication visual: e.g. signs, photos, symbols as well as speech. This can help them focus their attention, understand better and express themselves. We run training sessions for parents and carers.
- By working on new vocabulary and sentence building.
- By giving advice on developing the children's lip and tongue movements for speech.

#### **How do we decide on what to work on?**

By considering...

- What is the biggest barrier to communication?

# Nottingham Guidelines for the Management of Children with Down Syndrome

- What is most likely to increase the child's success in communicating?
- What is the potential for change?

## What are the principles of the Speech and Language Therapy Service?

- We provide care based on evidence, good practice guidance and professional standards.
- We work in partnership with you and your child, and other professionals to reduce the impact of their communication difficulties on the child's well-being and ability to participate in daily life. Working through others is the key to create change in the child's communication skills.
- We assess children's communication skills through observations, information gathering and assessment. We often link in with other professionals to help with this. E.g. Early Years Teachers.
- Every child can communicate. We help build on their strengths and use those to support areas they find more difficult.
- We tailor strategies to the needs of the individual child and focussing on people they communicate with and places they go, as well as the child's own skills.

## Useful resources/contacts:

These are some examples of resources available in the library at the Children's Centre, City Hospital.

- 'It takes two to talk. A parent's guide to helping children communicate'. Ayala Manolson.
- 'Early Communication Skills for Children with Down Syndrome'. Libby Kumin.
- Books and DVDs e.g. 'Something Special' ideal for practicing signing skills.

## Other resources

- Down Syndrome Educational Trust [www.downsed.org](http://www.downsed.org)
- Down Syndrome online [www.down-syndrome.org](http://www.down-syndrome.org)
- Makaton. Makaton Vocabulary Project, 31 Firwood Drive, Camberley, Surrey, GU15 3QD. [www.makaton.org](http://www.makaton.org)

## To find out more, please contact us...

South Base (Nottingham City, Broxtowe, Gedling and Rushcliffe) Children's Centre City Hospital Hucknall Rd Nottingham NG5 1PB 0115 831101	North Base ( Mansfield, Ashfield and Newark) Children's Therapy Centre Kings Mill Centre Mansfield Road Sutton in Ashfield Notts 01623 785019
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# Nottingham Guidelines for the Management of Children with Down Syndrome

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

The RCPCH Working Party on Sleep Physiology and Respiratory Control Disorders in Childhood report September 2009 [http://www.rcpch.ac.uk/doc.aspx?id\\_Resource=5215](http://www.rcpch.ac.uk/doc.aspx?id_Resource=5215) concluded that:

- Children with Down Syndrome 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.

They recommended the following:

- All children with Down Syndrome 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 Down Syndrome 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 CO<sub>2</sub> measurement. Video should be included if possible.
- If significant SRBD with hypoxia is present in children with Down Syndrome, then appropriate treatment should be offered.
- Further research is needed on the benefits and risks of screening for SRBD and Down Syndrome.

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.

Symptoms to consider include:

- Snoring
- Sleep disturbance
- Mouth breathing and halitosis
- Restless sleep
- Chronic rhinorrhea
- Subcostal and sternal recession
- Odd sleep positions, such as hanging over the bed or sleeping upright with head extended to optimise the upper airway

# Nottingham Guidelines for the Management of Children with Down Syndrome

- Swallowing difficulties
- Recurrent upper respiratory tract infections
- Nausea and vomiting
- Daytime sleepiness
- Persistent or secondary enuresis
- Nocturnal sweating
- Cyanosis
- Apnoea
- Pulmonary hypertension
- Heart failure.

## Local recommendations for screening for SRUAO/SRBD in Down Syndrome

As well as the arbitrary times for recommending screening as mentioned above there is concern regarding a lack of robust evidence to support the use of oximetry as a screening tool, with concerns particularly about false negative results. Further research is needed.

In the meantime we suggest that locally we do introduce screening with overnight oximetry at the age of 1 year and again at 4 years of age, supported by specific questioning for symptoms of SRBD at every review, with a low threshold for investigation for children with possible symptoms. This will be reviewed as further evidence becomes available.

- All children with Down Syndrome to have overnight oximetry at 1 year of age
- Specific questioning for possible symptoms at every paediatric review
- Children with no suggestive symptoms and normal oximetry will have routine follow up , with repeat oximetry at 4 years of age
- Children with abnormal oximetry will be referred for further assessment by respiratory paediatricians, including polysomnography, or to ENT surgeons if clinical symptoms dictate
- Children with significant suggestive symptoms, but “normal” oximetry should also be referred for further assessment as above
- Children with suggestive symptoms between or beyond the age of routine screening should be referred for oximetry

# Nottingham Guidelines for the Management of Children with Down Syndrome

## Appendix I

### **Guidelines for prevention, investigation and management of infections**

#### **Introduction**

Infections are commoner 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. We have had a number of adverse outcomes in children with Down Syndrome where prompt treatment of infection may have made a difference.

#### **General measures**

All children with Down Syndrome should have an alert put on electronic record E.g “this child has Down Syndrome – if they present to the emergency department or are admitted with possible sepsis please start promptly on antibiotics and refer to the Down Syndrome guideline section on prevention, investigation and management of infections”

Similar message should be added to correspondence with GP

#### **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 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).

If abnormal, suggest discuss with Dr Liz Mcdermott/Dr Lucy Cliffe (need to have vaccine history available).

# Nottingham Guidelines for the Management of Children with Down Syndrome

## 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 discussion with Dr Liz Mcdermott/Dr Lucy Cliffe

## 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 Dr Liz Mcdermott/Dr Lucy Cliffe

\* Pneumovax II should not be repeated within 5 years. If pneumococcal antibodies low despite recent Pneumovax II, please discuss with Dr Liz Mcdermott/Dr Lucy Cliffe

# Nottingham Guidelines for the Management of Children with Down Syndrome

## Appendix J

### **Physiotherapy Service in Nottingham for Children with Down Syndrome**

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 DSCC. 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 DSCC.

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.

# Nottingham Guidelines for the Management of Children with Down Syndrome

## References and further information

### **Medical management of children with Down Syndrome,**

Marder E. and Dennis J.,  
Current Paediatrics (2001) 11, 57 – 63.

### **Medical and Surgical Care for Children with Down Syndrome: A Guide for parents**

Van Dyke et al (Editors)  
Woodbine House. 1995. ISBN 0-933149-54-9

### **Early Support Programme Publications (Blue Box)**

Information for Parents – Down Syndrome

<https://www.education.gov.uk/publications/standard/publicationDetail/Page1/ES13>

Developmental Journal For Babies and Children with Down Syndrome

<https://www.education.gov.uk/publications/standard/publicationDetail/Page1/ES49>

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

DSMIG(UK) (2000) Insert for Personal Child Health Record for babies born with Down Syndrome (2nd edition). DSMIG, UK (available from Harlow Printing, NE33 4PU, UK)

### **Down Syndrome Medical Interest Group**

Children's Centre  
City Hospital Campus  
Hucknall Road  
Nottingham NG5 1PB

Website: [www.dsmig.org.uk](http://www.dsmig.org.uk)

Detailed guidelines and background evidence on which these guidelines are based can be found here, as well as access to a wide range of further information and resources on health issues

### **Down Syndrome Association**

Langdon Down Centre  
2a Langdon Park  
Teddington TW11 9PS  
Tel: 0845 230 0372

Website [www.downs-syndrome.org.uk](http://www.downs-syndrome.org.uk)

Parent support, and information on range of issues including health , education and legal issues

### **Down's Heart Group**

PO Box 4260  
Dunstable  
Beds LU6 2ZT  
Tel: 0845 166 0861

Website: [www.dhg.org.uk](http://www.dhg.org.uk)

Parent support group – useful information leaflets on cardiac issues

# Nottingham Guidelines for the Management of Children with Down Syndrome

## List of Contributors

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