Down Syndrome - Management of Suspected or Confirmed Down Syndrome on Postnatal Ward / Neonatal Unit

Classification:	Guideline
Authors Name:	Zuzanna Gawlowski
Authors Job Title:	Consultant Paediatrician
Authors Division:	Women's and Children's Health
Departments/Group this Document applies to:	Maternity, Paediatrics, Community Child Health

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Guideline to be followed by (target staff):
Maternity, Paediatrics, Community Child Health

To be read in conjunction with the following documents:

- Congenital Cardiac Defects in Babies - Nursing Management in the Community - PAED/GL/12

Are there any eCARE implications? No

CQC Fundamental standards:
Regulation 9 – person centred care
Regulation 10 – dignity and respect
Regulation 11 – Need for consent
Regulation 12 – Safe care and treatment
Regulation 13 – Safeguarding service users from abuse and improper treatment
Regulation 14 – Meeting nutritional and hydration needs
Regulation 15 – Premises and equipment
Regulation 16 – Receiving and acting on complaints
Regulation 17 – Good governance
Regulation 18 – Staffing
Regulation 19 – Fit and proper

Disclaimer

Since every patient's history is different, and even the most exhaustive sources of information cannot cover every possible eventuality, you should be aware that all information is provided in this document on the basis that the healthcare professionals responsible for patient care will retain full and sole responsibility for decisions relating to patient care; the document is intended to supplement, not substitute

for, the expertise and judgment of physicians, pharmacists or other healthcare professionals and should not be taken as an indication of suitability of a particular treatment for a particular individual.
The ultimate responsibility for the use of the guideline, dosage of drugs and correct following of instructions as well as the interpretation of the published material **lies solely with you** as the medical practitioner.

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Guideline Statement

The purpose of this guideline is to provide a clear pathway for the management of babies suspected of having Down Syndrome in order to ensure the babies receive the required investigations, reviews and support.

Executive Summary

Down Syndrome: Background

Down Syndrome or Trisomy 21 is the commonest autosomal anomaly. In England in 2018, 1,570 babies with Down's syndrome were delivered, representing a prevalence of 25 per 10,000 total births or 1 in every 400 births for Down's syndrome. The live birth prevalence for Down's syndrome was 11.6 per 10,000 live births or 1 in 862 live births." (Public Health England, 2020, p.32)

In the majority of cases (94%) there is a complete/regular trisomy so every cell of the affected individual contains 3 copies of chromosome 21. 4% of cases are translocations, with each cell containing a partial extra copy of chromosome 21. In translocation cases, the prognosis and outcome are the same as with a standard Trisomy 21, though one of the parents maybe a carrier of a balanced translocation and parental karyotypes are therefore recommended. In the remaining 2% of cases there is mosaicism: the affected individual has both 'normal' cells and cells with trisomy 21. This group has the same management, but course may vary with a lesser degree of learning impairment. (Down Syndrome Association: A guide for Healthcare Professionals 2019).

In around a third of cases, a diagnosis of Down Syndrome is made after birth1, often following routine or 'normal' antenatal scanning or 'low risk' (<1:150) Down's testing estimated by blood tests in the pregnancy. (Morris & Springett 2012)

If a baby is suspected to have Down Syndrome, ward staff in Labour and Postnatal Wards or the Neonatal Unit (NNU) should alert the neonatal medical team.

This guideline comprises:

- Management and Investigations required to confirm the diagnosis. This is relevant for babies WITHOUT antenatal diagnosis of Down Syndrome (Section 3.1)
- Relevant for ALL babies confirmed to have Down Syndrome, before or after birth (Section 3.2). Includes checklist to be completed prior to discharge home (Appendix 1)

Definitions

ANNP	Advanced neonatal nurse practitioner
COTW	Consultant of the week
ECHO	Echocardiogram
FBC	Full blood count
FISH	Fluorescent in situ hybridization
NBBS	Newborn blood spot screen
NIPE	Newborn infant physical examination
NNU	Neonatal unit
OFC	Occipitofrontal circumference
Spr	Specialist registrar

1.0 Roles and Responsibilities

Midwife:
The midwife is responsible for performing the initial newborn examination post birth. It is his/her responsibility to inform the Paediatric team regarding any suspected dysmorphic features / other abnormalities identified. It is also his / her responsibility to support the parents in caring for their baby on the postnatal ward whenever possible, including supporting feeding.

Paediatric ST1 – 3 / Advanced Neonatal Nurse Practitioner:
It is the Paediatric ST1-3 / ANNP's responsibility to identify any abnormalities / dysmorphic features when performing NIPE newborn examinations or when asked to review a baby either on Delivery Suite or in the postnatal ward at any other time and to inform the senior Paediatric team (Registrar and /or Consultant).

Paediatric Middle Grade doctor:
It is the Paediatric Middle Grade doctor's responsibility, if requested by the midwife or junior doctor, to review any baby identified either antenatally as having Down Syndrome, or who, on early postnatal examination, has features suggestive of Down Syndrome or other abnormalities as soon as reasonably possible. If there are any concerns, this should be escalated to the Paediatric Consultant on call or Neonatal Consultant of the Week (COTW). He / she is also responsible for ensuring all necessary referrals are made and the discharge checklist is completed prior to baby being discharged home.

Paediatric Consultant:
The Paediatric Consultant on call, if baby born out of working hours, or the Neonatal COTW if baby born within working hours, should be made aware of any baby with suspected dysmorphic features on the postnatal ward as soon as reasonably possible. If there is doubt as to whether or not to offer genetic testing, then the Consultant on duty should review the baby at the latest the morning following birth and make a final decision whether or not to offer genetic testing. The Consultant on duty should then hand over to the Neonatal COTW to take over care.

Neonatal COTW:
The Neonatal COTW will be responsible for informing the parents of the results of the Fluorescent in situ Hybridisation (FISH) analysis as soon as possible. If possible, he/she should try to perform an echocardiogram before the baby is discharged home. He/she should ensure the parents receive the Down Syndrome Information Pack (folder containing Down Syndrome booklet, Down Syndrome growth charts for the Personal Child Health Handbook – Red Book – and also a Parent Support Group leaflet). He/she is also responsible for ensuring that the Consultant Community Paediatrician with interest in Down Syndrome and also the Community Neonatal Nursing Team, are both informed of the newly diagnosed baby with Down Syndrome.

Consultant Community Paediatrician with Down Syndrome Interest:
The Consultant Community Paediatrician with interest in Down Syndrome will arrange, on receiving confirmation from Neonatal COTW of new diagnosis of child with Down Syndrome, to meet the parents and child in an out patient clinic ideally within 4 weeks and will continue long term follow-up for the child. The Consultant Community Paediatrician will instigate and coordinate onward referrals to therapists / Early Years Support Services as and when appropriate.

Community Neonatal Nursing Team:
The Community Neonatal Nursing Team will meet the family, if possible, together with the Neonatal COTW when the diagnosis is first informed to the parents, or as soon as possible thereafter. They will offer initial support for parents in understanding the diagnosis and initial

support in monitoring of weight and feeding, and will also undertake cardiac monitoring if required as per 'Congenital Cardiac Defects in Babies- Nursing Management in the Community' Guideline.

2.0 Implementation and dissemination of document

This document will be available on the Trust intranet.

3.0 Processes and procedures

3.1 Management and Investigations required to confirm the diagnosis

This is relevant for babies WITHOUT antenatal diagnosis of Down Syndrome. For babies with an antenatal diagnosis, postnatal FISH and karyotype testing should be offered to confirm the diagnosis, following postnatal examination.

3.1.1 On Postnatal Ward

If physical appearance prompts concern:

- Wherever possible, parents should be given explanations that some patterns of physical features require a paediatric assessment. As their baby has one/ more notable features, an assessment has been requested.
- Explanation that a review by a paediatrician will look not only at physical appearance, but will also perform a thorough medical examination, to try to identify any patterns/ diagnosis.

If the baby is otherwise well, ideally a senior paediatrician (SpR or Consultant) should perform this assessment within 12-24 hrs.

If baby is unwell, having difficulty feeding etc then referral for prompt review by the neonatal team is required.

3.1.2 Assessment by Senior Paediatrician

This should be done in an appropriate language, with an interpreter where necessary, and ideally with both parents.

Physical examination must be documented in separate baby notes on eCARE and to specifically include:

- Cardiac examination including pulse oximetry (pre- and post-ductal saturations)
- Meconium – time passed
- Eye examination, visual behaviour, red reflexes
- Tone/ feeding assessment

3.1.3 Diagnosis

If the diagnosis is clinically evident, then discussions re: diagnosis should not be delayed until blood tests results available.

Explanation of the baby's features and diagnosis should be delivered sensitively and by the most senior person available – ideally the Neonatal COTW. See Down's Syndrome Association (2019) Our 'Tell it Right' top tips on how this can be done.

If there are only subtle features or any uncertainly, explain to the family that a rapid FISH test will confirm the diagnosis within 48 hours.

3.1.4 Investigations

- Request rapid FISH test for Down Syndrome/ Trisomy 21
- Contact Genetics laboratory in Oxford to specifically request urgent sample

Note in the case of clinical uncertainty, the majority of babies remain in hospital for assessment of tone, feeding and general observation.

However, the baby may be discharged home if the following criteria are met:
- with the agreement of the responsible Neonatal Consultant
- the baby is medically well and fit for discharge, with documented NIPE examination completed
- clear documentation of examination above, with no concerns regarding feeding
- in agreement with the parent(s)
- an agreed date and time MUST be given to the family at discharge to return to NNU to discuss the results (48-72 hours after the sample is sent)

3.1.5 Results

Positive FISH for Down Syndrome
See Section 3.2.

Negative FISH for Down Syndrome
If the FISH results are negative then Section 3.2 recommendations are not applicable.

3.2 Relevant for ALL Babies Confirmed to Have Down Syndrome, Before or After Birth

It includes checklist (Appendix 1) to be completed prior to discharge home.

After confirmed antenatal diagnosis or positive rapid FISH test after birth:

Physical examination must be documented in separate baby notes, and to specifically include:
- Cardiac examination, pulse oximetry (pre- and post-ductal saturations)
- Meconium – time passed
- Eye examination, visual behaviour, red reflexes
- Tone/ feeding assessment
- Weight, Length, OFC

Low tone and feeding difficulties can frequently cause issues in the neonatal period. This should be emphasised to responsible postnatal ward midwives. The baby should have close monitoring to ensure successful establishment of feeding. A referral can be made to the Infant Feeding Team for support with feeding care plans.

3.2.1 Investigations (Whilst Inpatient)

- Blood tests
  - Karyotype – 2mls Lithium Heparin - please request copy of results to relevant Consultant in Community Paediatrics, Milton Keynes
  - FBC and film
  - Newborn Blood Spot Screening (Thyroid function). Ensure Community midwife does NBBS if baby already home by Day 5-7.

- Echocardiogram – ideally as inpatient or, if not available, within 4 weeks of birth.
- Newborn Hearing Screen.
- Further investigations and referrals as an inpatient to be guided by results of anomalies found on general examination.

3.2.2 Discharge Home

- Checklist in Appendix 1 MUST be completed prior to final discharge home. Please include a copy for the baby's notes.
- Ensure parents have received a Down Syndrome information pack containing the following:
  - Down Syndrome Information Booklet – see reference list for example booklet available from the Down Syndrome Organisation
  - Down Syndrome Growth Charts
    - Details of the Down Syndrome growth charts and the Personal Child Health Record insert are available online at the Down Syndrome Medical Interest Group (DSMIG) website:
    - Growth charts: https://www.dsmig.org.uk/information-resources/growth-charts/
    - Personal Child Health Record insert: https://www.dsmig.org.uk/information-resources/personal-child-health-record-pchr/
- Down Syndrome Parent Support Group Leaflet

4.0 Statement of evidence/references

Statement of Evidence

References

Public Health England (2020) National Congenital Anomaly and Rare Disease Registration Service statistics 2018: Summary Report – updated 12 August 2020. Available from https://www.gov.uk/government/publications/ncardrs-congenital-anomaly-annual-data. Last accessed 17-02-2021.

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Down's Syndrome Association (2014) *New Parents Pack* [Online]. Available from: https://www.downs-syndrome.org.uk/download-package/new-parents-pack/ [Accessed 25 November 2020]

Office of Communications (2018) Down Syndrome. *National Institute of Child Health and Human Development* [Online]. https://www.nichd.nih.gov/health/topics/factsheets/downsyndrome [Accessed17-02-2021]

External weblink references:

5.0 Governance

5.1 Document review history

Version number	Review date	Reviewed by	Changes made
1	11/2015		
2	11/2017		
3	10/2020		
4	2/2024	Zuzanna Gawlowski	

5.2 Consultation History

Stakeholders Name/Board	Area of Expertise	Date Sent	Date Received	Comments	Endorsed Yes/No
Women's Health CIG	Governance			Approved on 02/06/2021	
Children's Health CIG	Governance			Approved on 26/04/2021	
Lisa Viola	Neonatal Matron	11/05/2021		Reviewed and happy	
Shveta Chana	Consultant Paediatrician	08/05/2021			
Denise Campbell	Quality Lead Paediatrics	05/2021			

5.3 Audit and monitoring

Audit/Monitoring Criteria	Tool	Audit Lead	Frequency of Audit	Responsible Committee/Board
Compliance with guideline	Notes Audit	Lead Clinician for Down Syndrome	3 yearly	Child Health Clinical Governance Board.

5.4 Equality Impact Assessment

As part of its development, this Guideline and its impact on equality has been reviewed. The purpose of the assessment is to minimise and if possible, remove any disproportionate impact on the grounds of race, gender, disability, age, sexual orientation, religion or belief, pregnancy and maternity, gender reassignment or marriage and civil partnership. No detriment was identified. Equality Impact assessments will show any future actions required to overcome any identified barriers or discriminatory practice.

Equality Impact Assessment			
Division	Women and Children	Department	Paeds
Person completing the EqIA	Zuzanna Gawlowski	Contact No.	
Others involved:	Denise Campbell	Date of assessment:	02/03/2021
Existing policy/service	Yes	New policy/service	no

Will patients, carers, the public or staff be affected by the policy/service?	Yes
If staff, how many/which groups will be affected?	Medical / Nursing / Midwifery

Protected characteristic	Any impact?	Comments
Age	NO	Positive impact as the policy aims to recognise diversity, promote inclusion and fair treatment for patients and staff
Disability	NO	
Gender reassignment	NO	
Marriage and civil partnership	NO	
Pregnancy and maternity	NO	
Race	NO	
Religion or belief	NO	
Sex	NO	
Sexual orientation	NO	

What consultation method(s) have you carried out?
E-mail, Teams, Governance meetings – Paediatrics PIG,CIG , Maternity Guidelines meeting , CIG
How are the changes/amendments to the policies/services communicated?
E-mail, Teams, Governance meetings – Paediatrics PIG,CIG , Maternity Guidelines meeting , CIG
What future actions need to be taken to overcome any barriers or discrimination?

What?	Who will lead this?	Date of completion	Resources needed
Nil	Nil	Nil	Nil
Review date of EqIA	02/03/2021		

Appendix 1: Checklist

☐ ECHO date performed: ……*

If not as inpatient then planned date: ……

**Referral to Paediatric Cardiology at the John Radcliffe Hospital, if indicated.*

☐ Referral letter required

☐ Information to parents with the standard Down Syndrome Information Pack which is in the NNU consultant's room

Telephone numbers for NNU/ medical team if concerned, community nurses/ health visitors

☐ Referral to Community Neonatal Nurses

☐ Risk assessment form for Newborn Hearing Screen completed and sent to audiology

☐ Referral letter to Community Paediatric Consultant.

Please include the following details in the referral letter:

1. On NNU/Postnatal ward: details of chromosome testing/ what tests sent and dates, results of rapid test
2. FBC/ film: date sent/ result
3. Cardiac examination, ECHO findings if performed or, date referral for ECHO made.
4. Eye examination details, plus details of any additional physical features noted
5. Discharge measurements: weight/length/OFC.
6. Length of stay in hospital after birth.
7. Comment on tone, feeding, any special measures in place at time of discharge
8. List of any referrals made e.g., Paediatric Cardiology, Speech & Language Therapy etc.
9. Please photocopy discharge checklist and include with referral letter.

☐ Inform Community Paediatric Consultant's secretary via telephone (if not available, leave answerphone message) of new confirmed diagnosis of Down Syndrome.