MCN for Neonatology West of Scotland Neonatal Guideline	

Trisomy 21 - Care Pathway

This Pathway is intended to guide the initial care for a newborn infant with a diagnosis of Trisomy 21. It is intended for use by all health professionals involved in the care of these infants in the West of Scotland

Antenatal Diagnosis of Trisomy 21

- **No major structural abnormalities identified in detailed scan**
  – Counselling will be performed by the local Obstetric and Neonatal team

- **Cardiac Anomaly or Duodenal atresia suspected on detailed scan**
  - Referral to Fetal medicine at the Queen Elizabeth University Hospital (QEUH)
  - Antenatally detected cardiac anomalies will require joint assessment and counselling by Consultants in Fetal Medicine and Fetal Cardiology. Discussions will include a decision on place of delivery (Cardiac Centre or local unit if clinically appropriate).
  - Antenatally detected duodenal atresia will require joint assessment and counselling by Consultants in Fetal Medicine and Paediatric Surgery. Delivery will be planned at QEUH
  - Any specific plans for intrapartum or immediate postnatal management will be summarised and recorded in the maternal notes (according to local arrangements for highlighting such care plans)

- **Cord Blood Sampling**
  - 2ml Lithium Heparin – Karyotype & 2ml EDTA - QF-PCR (unless genetics confirmed antenatally)
  - If cord blood sample is taken, a sample of at least 3mls EDTA is required from the mother to be sent with the cord blood sample, appropriately labelled to exclude maternal contamination
  - 0.5ml EDTA - FBC and Film

Postnatal Diagnosis and Disclosure of Trisomy 21

- **Communication of diagnosis / suspected diagnosis**
    - Parents should be informed of the diagnosis / suspected diagnosis, as soon as possible, by a Senior Paediatrician
      (This would be the attending consultant for the relevant clinical area if they are in the hospital. Where the consultant is not immediately available the 2nd on Paediatrician should inform the parents of their provisional diagnosis and this should be followed by a consultant review at the next available opportunity (The following morning at the latest)

Cord Blood Sampling

- 2ml Lithium Heparin – Karyotype & 2ml EDTA - QF-PCR
- 3ml EDTA maternal sample appropriately labelled to exclude maternal contamination sent with cord blood sample from neonate
- 0.5ml EDTA - FBC and Film.

- **Confirmation of the diagnosis**

- Once the diagnosis is confirmed, the attending consultant for the relevant clinical area should inform the family and provide relevant counselling regarding the diagnosis
    - An Information Pack (Down Syndrome Association Scotland) should be provided for the family.
    - Down syndrome specific growth charts should be used both for the clinical notes (select Down syndrome charts in iGrow), and for the red-book. Please provide inserts for the latter even though they will not receive the red book until their first visit from the Health Visitor

Medical Examination

- A comprehensive medical assessment should be carried out, with particular attention to

    - Gastrointestinal tract - increased risk of duodenal atresia, oesophageal atresia and anorectal malformations (especially, Hirschsprung's disease)
    - Eyes - increased risk of cataracts
    - All neonates with known, or a high suspicion of, DS should be examined for features suggestive of TL-DS (Transient Leukemia of Down syndrome) , exclude organomegaly, cholestasis and other liver disease, skin rash, pericardial and pleural effusions

    - Cardiac assessment – increased risk of cardiac anomalies including septal defects and Fallot's tetralogy

Other Investigations

- ***Thyroid screen****:* Routine newborn screen is satisfactory.

- **Full Blood Count and Blood Film**

  -All neonates with known, or a high suspicion of, DS full blood count and blood film should be requested in the first 3 days of life and a formal assessment of the peripheral blood blast cell percentage performed by a haematologist with experience in reviewing neonatal blood films.

  - Babies in whom clinical examination and blast cell percentage indicate that TL-DS is likely should have additional tests considered: liver function tests including conjugated bilirubin if the baby has significant jaundice, chest X-ray, echocardiogram and abdominal ultrasound (Grade 1B)[1].

  - Any neonate with a blast percentage >10% and/or clinical features suggestive of TL-DS should be discussed urgently with the regional Paediatric Oncology Principal Treatment Centre and a peripheral blood sample sent for GATA1 mutation analysis (Grade 1A) [1].

  - 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 intra-uterine growth retardation (when blast counts may be suppressed) should be considered to be still at risk of clinical problems of TLDS in the first 4-8 weeks of life and should be monitored accordingly. GATA1 mutation analysis should be considered (Grade 1B) [1].

  - Any GATA1 positive babies are referred to haematology for ongoing surveillance of counts until the age of 4 [1].

- **ECHO**

  ECHO, if expertise is available locally, may provide useful information.

  If there is no local Paediatrician available, with expertise in cardiology, the baby should be examined by a senior paediatrician and have pre & post ductal saturations and an ECG performed.
  If the Cardiac Assessment is normal a routine referral can be made to cardiology for assessment within 6 weeks. If the clinical examination or any of the investigations raise the suspicion of a cardiac abnormality then advice should be sought from the on-call consultant for cardiology.

Referrals/notifications to be arranged by neonatal team prior to discharge

- Local Neonatal /Paediatric clinic
- Cardiology. All babies should be referred for a formal cardiology review even if the initial cardiac exam was normal.
- Community Child Health – refer as soon as diagnosis made, but shared care with the local neonatal team may be required for a period until acute medical issues have resolved. These may include feeding difficulties, jaundice, home oxygen and babies with transiently elevated TSH who will require repeat testing until hypothyroidism has been excluded.
- If local community child Centre is not known (especially children living outside Greater Glasgow), contact family Health Visitor to direct you to the local community paediatrician/ Child Centre.
- Primary Care team (GP and HV) by telephone

In Glasgow, please contact "The Disability Service within a Specialist Community Paediatric Team (SCPT)", by email on discharge, see contacts for Glasgow and Clyde below, Please refer to your local arrangement if out with Glasgow and Clyde

- Physiotherapy, Refer to community physiotherapy either directly or via your Local Neonatal Physiotherapist, as per local arrangements.
- Referral for Palivizumab – If the infant has cardiac disease and meets the criteria outlined in the immunisation guideline.

Babies diagnosed after hospital discharge

A small number of babies are diagnosed after hospital discharge where the clinical features were not appreciated in the immediate postpartum period. Such babies should be referred back to a neonatal consultant to ensure that the necessary investigations and referrals are completed. This will include

- Referral to audiology
- Cardiac examination and referral to cardiology
- Genetic sample to screen for GATA1 mutations (If older than 3 days, as the blood film appearances indicative of TL-DS may have resolved)

Reference

1. Guidelines for the investigation and management of Transient Leukaemia of Down Syndrome. Tunstall O, Bhatnagar N, James B, Norton A, O'Marcaigh AS, Watts T, Greenough A, Vyas P, Roberts I, Wright M; British Society for Haematology. Br J Haematol. 2018 Jul;182(2):200-211. doi: 10.1111/bjh.15390. Epub 2018 Jun 19.

2. Dalrymple RA, Somerville LH, Hamza S*, et al*. Fifteen-minute consultation: The review of a child with trisomy 21 (Down's syndrome) *Archives of Disease in Childhood - Education and Practice* 2022;**107:**88-94.

Local Arrangements for community follow up

GG&C

Team Leader Information

"The Disability Service within a Specialist Community Paediatric Team (SCPT)" Team Lead information

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Implementation / Review Dates

Implemented – 01/01/17 Reviewed - 21/06/22 Review – 01/06/25

The Down syndrome Neonatal Discharge Form

Patient Name	DOB	CHI	Consultant

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