

DOWN'S SYNDROME – INITIAL MANAGEMENT • 1/4

INTRODUCTION

- Congenital disorder arising from a chromosome defect
- Majority due to trisomy of chromosome 21
- 4% translocations
- 1% mosaics
- Antenatal screening and subsequent termination of pregnancies results in incidence at birth of 0.8/1000
- Incidence increases with increasing age of mother from 1:1500 at aged 20 yr to 1:100 aged 40 yr

DIAGNOSIS

Antenatal

- Confirm cases identified through antenatal screening/high-risk women by amniocentesis/chorionic villi sampling
- Arrange for parents to be seen by **neonatal/paediatric consultant**
- **Complete local paediatric alert register for postnatal care**
- Give parents opportunity to visit **NNU**
- **if visits not possible direct parents to virtual tour (if available)**

Postnatal

- Approximately 30% of cases are not identified before birth – mainly due to screening declined/not undertaken
- If suspected on newborn and infant physical examination (NIPE), request immediate detailed clinical examination by paediatrician/advanced neonatal nurse practitioner
- Identify any urgent medical needs (e.g. feeding, cardiac or respiratory problems)
- **Consultant paediatrician** to discuss testing with parents
- Send EDTA blood sample to **regional genetic laboratory** for confirmation by testing QF-PCR
- referral laboratory will request lithium heparin sample for karyotyping if necessary

Parent consultation

- Parents may have conflicting emotions
- Parents to be seen by consultant:
 - antenatally diagnosed: ≤ 24 hr of birth
 - postnatally diagnosed: ≤ 24 hr of suspicion
 - use interpreter for non-English speaking parents
 - if possible/appropriate both parents to be present during consultation
 - deliver explanation of baby's features and diagnosis sensitively
 - give parents time to absorb information
- Repeat visits may be necessary to deal with questions and distress
- If possible, same consultant to continue to see baby and parents until discharge
- if not possible, named/follow-up consultant must have clear handover

DOWN'S SYNDROME – INITIAL MANAGEMENT • 2/4

INITIAL MANAGEMENT

Age	Professional	Tasks
Birth	Consultant paediatrician/neonatologist	<ul style="list-style-type: none"> • Neonatal examination • QF-PCR genetic testing +/- karyotyping to confirm Down's syndrome • Blood for chromosomes, FBC and manual blood film for peripheral blasts <ul style="list-style-type: none"> • if blasts present discuss with paediatric haematologist • Counselling of parents by consultant (see Parent consultation) • Give written information to parents (e.g. Down's Syndrome Association pack with new parent leaflet – available from http://www.downs-syndrome.org.uk/for-new-parents/new-parent-pack/) • Notify midwife, obstetrician, GP, and health visitor • Cardiac assessment including: <ul style="list-style-type: none"> • pre and postductal pulse oximetry • ECG (if available locally) – discuss with consultant • if cardiac symptoms/signs or abnormal ECG, detailed clinical cardiac examination including echocardiogram within 2 weeks • if no cardiac symptoms and normal ECG, cardiac review and echocardiogram within 4–6 weeks • Gastrointestinal atresia – observe for vomiting (bile stained) • Hirschsprung's disease – ensure meconium passed ≤24 hr of birth • Visual assessment: <ul style="list-style-type: none"> • check visual behaviour and red reflexes for congenital cataract and nystagmus • if concerns refer to ophthalmologist • Follow-up with a paediatrician/neonatologist • Refer to community paediatric team with detailed summary and copies of all other referrals (e.g. ophthalmology, cardiology) • Discuss referral to early support services • Nurse specialist/dietician to provide feeding advice • Speech and language assessment/therapy referral where necessary • Provide parents with information and/or additional sources of help and advice • Replace growth charts in Personal Child Health Record (PCHR) and notes with specific Down's syndrome insert/chart and plot growth parameters • Check automatic referral to audiology has been made
≤5 days	Midwife	<ul style="list-style-type: none"> • Risk of congenital hypothyroidism – ensure heel prick test performed
2–4 weeks	Consultant paediatrician	<p>Follow-up appointment</p> <ul style="list-style-type: none"> • Review parental concerns and medical history, particularly cardiac symptoms, feeding and bowel habit • Ensure Down's syndrome insert in PCHR and growth parameters plotted on Down's syndrome growth chart • Cardiac examination <ul style="list-style-type: none"> • check seen in cardiology clinic • Examine eyes for cataract and nystagmus • Verify results of TSH screen • Check referral to child development centre • If concerns refer to dietician and community speech and language therapy (SALT)
6 weeks	Health visitor/GP	<p>Routine Child Health Service – primary birth visit</p> <ul style="list-style-type: none"> • Plot growth on Down's syndrome chart • Issue Down's syndrome specific pages and growth chart for PCHR if not already issued

DOWN'S SYNDROME – INITIAL MANAGEMENT • 3/4

8 weeks	Health visitor/GP	<ul style="list-style-type: none"> • Primary immunisations
3–4 months	Paediatrician/ community paediatrician /child development centre	Initial assessment <ul style="list-style-type: none"> • Developmental assessment • Refer to physiotherapy as appropriate • Ensure referred to ophthalmologist and SALT • Review newborn hearing screening programme results (in PCHR) • Hearing screening <ul style="list-style-type: none"> • if no clear bilateral or unilateral response: refer for audiological assessment • if bilateral clear response: ensure referral for targeted follow-up aged 7–9 months • Refer to early support services • TSH, FT4 and thyroid antibodies

- Complete neonatal checklist for management of babies with Down's syndrome (if available locally)

LATER REVIEWS

- At all stages review/discuss:
 - parental concern
 - developmental progress
 - growth using **specific Down's syndrome chart**
 - hearing and visual problems
- Formal ophthalmological and audiology assessment every 2–3 yr
 - more often if abnormal
- Copy clinic letters to parents and all professionals involved

Age	Review/action
9 months	<ul style="list-style-type: none"> • Follow surveillance check list, if available locally • Exclude squint • Audiology assessment • Developmental progress
12 months	<ul style="list-style-type: none"> • TSH, FT4 and thyroid antibodies, then annually • if TSH levels elevated/positive antibodies present, discuss with endocrine team
18 months	<ul style="list-style-type: none"> • Developmental progress review <ul style="list-style-type: none"> • discuss schools/nurseries as appropriate • Monitor growth and plot on Down's syndrome chart every visit • Check dental health and refer to specialist community paediatric dentist • Check gastrointestinal symptoms – constipation/diarrhoea, increased risk of coeliac disease • If symptoms of obstructive sleep apnoea present refer to ENT team • Assess gait, bowel and bladder function <ul style="list-style-type: none"> • risk of atlanto-axial subluxation – suspect if new symptoms of gait disturbance, abnormal neck posture and/or deterioration in bladder/bowel function • Increased incidences of: <ul style="list-style-type: none"> • type 1 diabetes (10 x normal) • autism • leukaemia • Advise parents about relevant benefits e.g. disability living allowance (DLA) <ul style="list-style-type: none"> • DLA has 2 parts – care and mobility <ul style="list-style-type: none"> – all children with Down's syndrome will eventually receive DLA – advise parents to consider application when child requires more help than children of a similar age (local authorities have denied applications on the basis that a baby with Down's syndrome has the same needs as any other baby) – application for mobility element of DLA can only be made when child aged ≥3 yr

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| | <ul style="list-style-type: none">• Give information about local and national Down's syndrome support groups |
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FURTHER USEFUL INFORMATION

- Down's Syndrome Association: www.downs-syndrome.org.uk
- Down Syndrome Medical Interest Group: www.dsmig.org.uk