DOWN SYNDROME – INITIAL MANAGEMENT

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 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/neonatologist to discuss testing with parents
- Send EDTA blood sample to regional genetic laboratory for confirmation by testing QF-PCR and lithium heparin sample for karyotyping

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

INITIAL MANAGEMENT

Age	Professional	Tasks
Birth	Consultant paediatrician/ neonatologist	• Neonatal examination • QF-PCR genetic testing +/- karyotyping to confirm Down syndrome • Blood for chromosomes, FBC and manual blood film for peripheral blasts ◦ if blasts present and/or clinical signs suggestive of (transient leukaemia of Down syndrome) TL-DS, discuss with paediatric haematologist and send peripheral blood sample for GATA1 mutation analysis • 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: ◦ 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: ◦ 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 syndrome insert/chart and plot growth parameters • Check automatic referral to audiology has been made
≤5 days	Midwife	• Risk of congenital hypothyroidism – ensure heel prick test performed
2–4 weeks	Consultant paediatrician	**Follow-up appointment** • Review parental concerns and medical history, particularly cardiac symptoms, feeding and bowel habit • Ensure Down syndrome insert in PCHR and growth parameters plotted on Down syndrome growth chart • Cardiac examination ◦ 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	**Routine Child Health Service – primary birth visit** • Plot growth on Down syndrome chart • Issue Down syndrome specific pages and growth chart for PCHR if not already issued • Recheck for red reflex and other eye anomalies
8 weeks	Health visitor/GP	• Primary immunisations
3–4 months	Paediatrician/ community paediatrician/child development centre	**Initial assessment** • Developmental assessment • Refer to physiotherapy as appropriate • Ensure referred to ophthalmologist and SALT • Review newborn hearing screening programme results (in PCHR) • Hearing screening ◦ if no clear bilateral or unilateral response: refer for audiological assessment ◦ if bilateral clear response: ensure referral for targeted follow-up aged 6–10 months • Refer to early support services • TSH, FT4 and thyroid antibodies at aged 4–6 months, then aged 12 months and annually thereafter, unless: ◦ already receiving treatment for thyroid disorder **or** ◦ develops signs and symptoms of thyroid dysfunction – earlier testing indicated

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

LATER REVIEWS

- At all stages review/discuss:
  - parental concern
  - developmental progress
  - growth using specific Down syndrome chart
  - hearing and visual problems
- Formal ophthalmological assessment aged 18 months–2 yr, then aged 4 yr
  - more often if abnormal
- Formal audiology assessment aged 6–10 months then
  - 6-monthly until aged 2 yr then
  - annually aged ≤18 yr
  - more often if abnormal
- Copy clinic letters to parents and all professionals involved

Age	Review/action
9 months	• Follow surveillance check list, if available locally • Exclude squint • Audiology assessment • Developmental progress
12 months	• TSH, FT4 and thyroid antibodies, then annually • if TSH levels elevated/positive antibodies present, discuss with endocrine team
18 months	• Developmental progress review • discuss schools/nurseries as appropriate • Monitor growth and plot on Down 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 • 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: • type 1 diabetes (10 x normal) • autism • leukaemia • Advise parents about relevant benefits e.g. disability living allowance (DLA) • DLA has 2 parts – care and mobility – all children with Down 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 syndrome has the same needs as any other baby) – application for mobility element of DLA can only be made when child aged ≥3 yr • Give information about local and national Down syndrome support groups

FURTHER USEFUL INFORMATION

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