HYPOTHYROIDISM

SCREENING
- Perform neonatal screening blood test at 5-8 days old for:
  - congenital Hypothyroidism
  - phenylketonuria
  - sickle cell disease
  - medium chain acyl-co A dehydrogenase deficiency
  - cystic fibrosis
- In preterm infants, perform on day 6 and repeat at 36–40 weeks gestational age
- Screening relies on measurement of raised blood spot TSH
- duty clinical biochemist will notify designated consultant or on-call consultant if blood spot TSH >20 mU/L on first testing or ≥10 mU/L on two occasions

IMMEDIATE MANAGEMENT
Informing diagnosis
- If screening test result indicates congenital hypothyroidism, a well-informed healthcare professional (community midwife, neonatal outreach nurse, health visitor or GP) must inform parents face-to-face
- do not communicate an abnormal result on Friday, Saturday or just before a weekend if consultant meeting cannot be arranged within next 24 hr
- provide parents with information leaflet 'congenital hypothyroidism suspected' (available from www.newbornscreening-bloodspot.org.uk)

Consultant meeting
- Consultant to arrange to meet parents on same or next day to:
  - explain abnormal result
  - examine baby using screening laboratory proforma as an aide-mémoire
  - look for other abnormalities (10% in CH versus 3% in normal baby), congenital heart disease (pulmonary stenosis, ASD and VSD) is commonest anomaly
  - commence baby’s treatment
  - stress importance of daily and life-long treatment
  - provide parent information leaflet (available from www.ich.ucl.ac.uk/factsheets/families/F040274/congenital_hypothyroidism.pdf)
- Document discussion and management plan and follow up and send to GP and parents
- Complete and return data form to clinical biochemist at screening laboratory

Obtain further diagnostic tests
- Baby
  - 1 mL venous blood in heparinised container for FT$_4$ and TSH
  - send repeat dried blood spot card to screening laboratory
  - ultrasound or radionuclide scan of thyroid, preferably within 7 days of starting levothyroxine
- Mother
  - take 3 mL venous blood from mother into a heparinised container for FT$_4$, TSH and thyroid antibodies

TREATMENT
- Start treatment with levothyroxine after obtaining confirmatory blood tests. Do not wait for results unless transient hypothyroidism suspected. Treatment must start before 21 days of age, and preferably by 18 days
- after discussion with paediatric endocrinologist, consultant may withhold treatment if transient hypothyroidism suspected
- Starting dose levothyroxine 10 microgram/kg/day with a maintenance regimen to maintain serum FT$_4$ in upper half of normal range, particularly for infants with severe hypothyroidism (e.g. athyrotic, ectopic or those with initial FT$_4$ <5.5 nmol/L)
- Adjustment required depending on thyroid function test results
Hypothyroidism 2009-11

- Tablets are 25 microgram strength
- it is not necessary to divide tablets for intermediate dose as half life >1 week
- administer intermediate dose, such as 37.5 microgram, as 25 and 50 microgram on alternate days
- Crush required levothyroxine dose (e.g. between 2 metal spoons) and mix with a little milk or water, using teaspoon or syringe
- do not add to bottle of formula
- suspensions not advised due to variable bioavailability
- repeat dose if baby vomits or regurgitates immediately
- Record date treatment commenced
- Provide parents with 28 day prescription for levothyroxine
- Arrange continued prescription with GP, emphasising need to avoid suspensions

FOLLOW–UP
- Arrange follow up after commencement of hormone replacement therapy as follows:
  - 2 weeks, 6 weeks, 3 months, 6 months, 9 months, 1 yr, 18 months, 2 yrs, 30 months, 3 yr, yearly thereafter
- At each clinic visit:
  - physical examination, including height, weight and head circumference
  - developmental progress
  - blood sample for thyroid function test (FT$_4$, FT$_3$ and TSH, just before usual daily medication dose)
- request as FT$_4$ priority, then TSH

Interpretation of thyroid function test results

<table>
<thead>
<tr>
<th>Analyte</th>
<th>Age</th>
<th>Concentration</th>
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</thead>
<tbody>
<tr>
<td>FT$_4$ (pmol/L)</td>
<td>0-5 days</td>
<td>17-52</td>
</tr>
<tr>
<td></td>
<td>5-14 days</td>
<td>12-30</td>
</tr>
<tr>
<td></td>
<td>14 days-12 yr</td>
<td>12-25</td>
</tr>
<tr>
<td>TSH (mU/L)</td>
<td>0-14 days</td>
<td>1-10</td>
</tr>
<tr>
<td></td>
<td>15 days-12 yr</td>
<td>3.6-8.5</td>
</tr>
</tbody>
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Check reference ranges with your laboratory’s assay

- Aim for FT$_4$ towards upper limit of normal range
- at higher concentrations of FT$_4$, normal concentrations of T$_3$ (produced by peripheral conversion) are achieved
- if FT$_4$ concentration satisfactory but with significantly raised TSH, consider non-compliance
- TSH concentration does not always normalise under 6 months and may be slightly raised up to 3 yr of age in absence of non-compliance, probably due to reset feedback mechanism
- Overtreatment may induce tachycardia, nervousness and disturbed sleep patterns, and can produce premature fusion of cranial sutures and epiphyses

AFTERCARE
- Reassure parents that baby will grow into healthy adult with normal intelligence
- Stress importance of regular treatment. As half-life is long, it is not necessary to give an extra tablet next day if a day’s treatment missed
- Give details of:
  - British Thyroid Foundation, PO Box 97, Clifford, Wetherby, West Yorkshire, LS23 6XD
  - Child Growth Foundation, 2 Mayfield Avenue, Chiswick, London, W4 1PW
  - www.bsped.org.uk