Guidelines for aetiological investigation into bilateral severe to profound permanent childhood hearing impairment

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SUMMARY DOCUMENT

This document is an executive summary of the guidelines for quick reference. It is meant for all children with bilateral severe to profound PCHI (thresholds over 70 dBHL in the better ear averaged across 0.5, 1, 2 and 4 KHz). The detailed document should be referred to for further information.

Level 1 investigations should be offered to all children and Level 2 offered to children with specified indications.

Level 1 investigations

[1] Clinical history [Recommendation D]
[2] Clinical examination [Recommendation D]
[3] Family audiograms of parents and siblings [Recommendation D]
[4] ECG for prolongation of the corrected QT interval. [Recommendation D] This is essential in children with evidence of vestibular hypofunction.
[5] Ophthalmological assessment [Recommendation B/C]: Guided by the Vision care document by NDCS/SENSE

- At diagnosis
- Parent or professional concern
- 1-3 years of age
- 4-5 years of age [orthoptist]
- 7-9 years of age.
- At referral for cochlear implantation
- Transition to secondary school

Consider electro-retinography if evidence of vestibular hypofunction [e.g. delayed motor milestones] without an identifying cause.
[6] Urine examination (labstix) for microscopic haematuria and proteinuria [Recommendation D]
[7] CMV testing [Recommendation B/C]
Child less than one year age: Urine or saliva swab x 2 samples for CMV DNA PCR

- If baby is less than 3 weeks age, a positive CMV DNA test in urine or saliva is evidence of congenital CMV.
- If baby more than 3 weeks age, a positive CMV DNA test in urine or saliva could be due to acquired CMV infection and the neonatal blood spot should be tested for CMV DNA. A positive result is evidence of congenital CMV but a negative result cannot reliably exclude congenital CMV

Child more than one year age: CMV IgG +/- Urine CMV DNA PCR

- If either positive, request neonatal dried blood spot

At any age: Mother's CMV IgG. If negative, congenital CMV can be excluded.
[8] Blood test for GJB2 [Connexin 26] mutations [Recommendation C]
Informed consent necessary prior to genetic testing

[9]Imaging: [Recommendation C]
MRI of Internal Auditory Meati and Brain [in Sensorineural hearing loss] or CT Scan of Petrous Temporal bone [in permanent conductive hearing loss]. Both MRI and CT are needed in bacterial meningitis and may be needed prior to cochlear implantation

Level 2 investigations

[1] Serology [Recommendation C]

- **Syphilis:** TPHA and FTA-ABS tests [IgG and IgM]
- **HIV**: In 'at risk' pregnancies
- **Rubella:** Infant less than 6 months of age: Rubella IgM
  Infant more than 6 months of age: Rubella IgG at 1 year age (before MMR)
- **Toxoplasma**:
  Infant less than 1 year of age: Maternal toxoplasma IgG &Toxoplasma IgM/IgG
  Infant more than 1 year_of age: Toxoplasma IgG & Consider maternal Toxoplasma IgG

[2] Haematology/Biochemistry: [Recommendation D] Not recommended as routine. TFT are indicated if: Family history of thyroid disease, Goitre, EVA or Mondini deformity.
[3] Investigation into autoimmune diseases: [Recommendation D]
[4] Metabolic screen on blood and urine: [Recommendation D] Consider if epilepsy, neuroregression
[5] Renal ultrasound [Recommendation D] Consider if

- Preauricular pits/sinuses, deformity of ear, branchial cleft or cysts
- Mondini defect or EVA.
- Permanent conductive or mixed hearing loss
- Features suggesting syndrome with kidney involvement e.g. CHARGE

[6] Chromosomal studies/CGH microarray: If developmental delay/dysmorphism
[7] Further genetic testing E.g. SLC26A4, EYA.
Test for m.1555A>G if:

- Exposure to aminoglycoside antibiotics
- Progressive hearing loss
- Mother/sibling with sensorineural hearing loss
- High frequency sensorineural hearing loss

[8] Referral to a Geneticist: Consider if

- Parental consanguinity
- A syndrome is suspected,
- Child has multiple abnormalities,
- Parental request
- Opinion required on interpretation of genetic mutation testing
- After completion of investigations no cause has been identified.

[9] Vestibular investigations [Recommendation D] Consider if

- Delayed motor milestones
- Progressive deafness
- Conditions associated with vestibular dysfunction e.g. post-meningitis
- Vertigo/dizziness
- Temporal bone malformations