

Guidelines for aetiological investigation into mild to moderate permanent childhood hearing impairment

April 2015

SUMMARY DOCUMENT

This document is an executive summary of the guidelines for quick reference. It is meant for all children with mild to moderate permanent sensorineural and conductive hearing loss of pre-lingual nature or late onset. 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] **CMV testing** (for sensorineural hearing loss) [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.

[5] **Blood test for GJB2 [Connexin 26] mutations**(sensorineural hearing loss) [Recommendation C] Informed consent necessary prior to genetic testing

[6] **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.

[7] **Ophthalmic assessment** [Recommendation B/C]: Guided by the Vision care document by NDCS/SENSE.

- Following the diagnosis of PCHI
- At any time if parents or the education service have concerns
- At 1 to 3 years of age
- At 4 to 5 years of age [orthoptist]
- At 7-9 years of age
- Transition to secondary school

Consider electro-retinography if there is evidence of vestibular hypofunction [e.g. delayed motor milestones] without an identifying cause or symptoms of retinitis pigmentosa e.g. night blindness, visual field loss.

[8] Urine examination [Recommendation D]: for microscopic haematuria and proteinuria

Level 2 investigations

[1] Serology to exclude congenital infection (for sensorineural hearing loss) [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] Further genetic testing: Depending on clinical features 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

Chromosomal studies/CGH microarray is indicated if the child has history of developmental delay, or dysmorphic features

[3] Renal ultrasound [Recommendation D]: Consider if

- Preauricular pits or sinuses, deformity of ear [microtia, anotia, cup/lop ear], branchial cleft or cysts
- Mondini defect or EVA on imaging.
- Permanent conductive or mixed hearing loss
- Features of syndrome with kidney involvement e.g CHARGE

[4] Haematology and Biochemistry [Recommendation D]: Not recommended as routine. Thyroid function tests are indicated if there is family history of thyroid disease, goitre, EVA or Mondini deformity.

[5] Investigation into autoimmune diseases [Recommendation D]

[6] Metabolic screen on blood and urine: Consider if epilepsy, neuroregression

[7] Referral to a Geneticist: Consider if

- Family history of hearing loss, parental consanguinity
- A syndrome is suspected,
- Child has multiple abnormalities,
- Parental request
- Opinion on interpretation of genetic mutation testing

[8] Vestibular investigations [Recommendation D]: Consider if

- Motor milestones are delayed
- Progressive deafness,
- Conditions known to be associated with vestibular dysfunction e.g. meningitis
- Vertigo/dizziness
- Vestibular malformations