Aetiological investigation into bilateral mild to moderate permanent hearing loss in children

Produced by the British Association of Audiovestibular Physicians and British Association of Paediatricians in Audiology
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Aim

The aim of these guidelines is to provide an evidence based approach to medical evaluation of the cause of mild to moderate hearing loss in children.

Guidelines are not intended to restrict clinical freedom, but practitioners are expected to use the recommendations as a basis for their practice. Where possible recommendations are based on, and linked to the evidence that supports them.

Areas lacking in evidence are highlighted and may form a basis for future research.

Categories of evidence:

Ia Evidence from meta-analysis of randomised controlled trials
Ib Evidence from at least one randomised controlled trial
IIa Evidence from at least one controlled study without randomisation
IIb Evidence from at least one other type of quasi-experimental study
III Evidence from non-experimental descriptive studies, such as comparative studies, correlation studies, and case-control studies
IV Evidence from expert committee reports, or opinions or clinical experience of respected authorities, or both

Strength of recommendations is expressed thus:

A. directly based on category I evidence

B. directly based on category II evidence or extrapolated recommendation from category I evidence

C. directly based on category III evidence or extrapolated recommendation from category I or II evidence

D. directly based on category IV evidence or extrapolated recommendation from category I, II, or III evidence

Search methodology : Medline, Embase, Cochrane
By NLH Specialist Library for ENT and Audiology , and hand search of journals.

Why investigate hearing loss

There are several reasons why it is important to perform medical evaluation of children with hearing loss:

1. To try and answer parents who ask "Why is my child deaf?"
2. To identify and treat medical conditions e.g. congenital infection, Alport, syndrome, Neurofibromatosis type 2, and vestibular hypofunction.
3. The results of investigations can assist the professionals in making decisions about the most appropriate management plan e.g further investigations, likelihood of progression, follow ups and Educational placement e.g. Usher type II Syndrome as well as counselling the family appropriately.
4. To inform genetic counselling
5. The information from investigation of childhood deafness informs epidemiological research

The timing of investigations will depend on the family's readiness to proceed with tests and how well the child can cooperate with tests.

Subjects

All children with bilateral permanent sensorineural, conductive or mixed hearing loss with average hearing level of 20-69dB HL measured in the better hearing ear at 0.5, 1, 2, 4kHz.

If there is an asymmetric hearing loss investigate the child according to the worst hearing ear using the appropriate guideline for example if the average hearing level is >69 dB HL use the guideline for severe to profound hearing loss.

Guidelines for Good Practice

Level 1 investigations:

Level 1 investigations should be considered for every child. Timing will depend on several factors, including the family's agreement to proceed with tests, availability of local test facilities and how well the child can cooperate with tests.

In cases where aetiology has not been diagnosed then further aetiological investigations may need to be arranged and some repeated.

Paediatric history: Strength of evidence - D

Detailed history of :

(a) onset of audiovestibular symptoms and progression of symptoms

(b) History of exposure to risk factors e.g.

- noise
- ototoxic medications/ radiation
- head injury
- ear disease
- meningitis
- bacterial and viral illness
- immunisation status

c) pregnancy, delivery and postnatal period

d) developmental milestones including speech, language, motor milestones as well as social development

e) Family history of deafness or risk factors associated with hearing loss in first and second degree relatives.

f) History of consanguinity and ethnic origin

2) **Clinical Examination**: **Strength of evidence - D**

Should include

a) measurement of height, weight and head circumference

b) Inspection of craniofacial region.

c) Examination of the ears, neck, skin and nails, limbs, chest, abdomen and gait.

d) Developmental assessment

3) **Family audiograms**: **Strength of evidence - D**

Parents and first degree relatives as mild hearing losses in family members may be undiagnosed (1).

4) **Ophthalmological assessment**: **Strength of evidence - D**

assessment of visual acuity and fundoscopy which may require repeating in the second decade of life (2) .

Consider electroretinography for children with visual symptoms, those with high frequency hearing loss and those with auditory neuropathy (as other nerves may be affected) (3,4,5).

5) Urine examination (labstix) for microscopic haematuria (6,7,8) **Strength of evidence - D**

6) Cytomegalovirus DNA Testing (9,10,11). **Strength of evidence - D**

< 1 yr : urine and/or saliva x 2
>1yr: if taking blood: IgG
if not taking blood: urine and/or saliva (if they are negative, test IgG)

If either are positive, request Dried Blood Spot for CMV DNA testing:
Requires:

1. mother's address during first weeks of baby's life
2. signed parental consent
3. newborn screening laboratory address:

see http://www.newbornscreening.org/laboratories.asp

N.B. consider maternal IgG: if negative to exclude congenital CMV

In cases with Moderate hearing loss the following are strongly suggested : Strength of evidence – D

a) Blood test for Connexin 26 mutations (12,13) with consent from parents, an explanation that DNA is stored afterwards in lab, that genetic testing can take a long time and for permission to share results with other family members/professionals (see guidelines for consent for genetic testing (14)

b) MRI of Internal Auditory meati should be considered in children with progressive hearing loss, dysmorphic features, history of tinnitus vestibular symptoms and those with other cranial nerve abnormalities especially of 5,6 and 7th nerves and/or developmental delay.

c) CT Scan of Petrous Temporal bone should be considered in children with permanent conductive hearing loss, fluctuating hearing loss, Tullio phenomenon.(15,16, 17,18,19)

For Mild hearing losses the yield from the above investigations will be low but the decision to request these tests will be informed by clinical findings

Other investigations: **Strength of evidence - D**

These will be indicated from history and clinical findings.

(a) Autoimmune disease screen where there is evidence of progression of hearing loss and or other systemic symptoms (20)

(b) Serology for congenital infection: where there is a need to exclude congenital rubella, toxoplasmosis, syphilis. Investigation may involve testing maternal stored (booking) serum if available.

(c) Metabolic Screen and Chromosomal studies:

May be indicated if there is a history of developmental delay or Dysmorphic features (Follow Local Child Development Team protocol)

(d) Renal ultrasound:

- If child has preauricular pits or sinuses, deformity of ear, branchial cleft or cysts
- Mondini defect on imaging.
- permanent conductive or mixed hearing loss
- multiple congenital abnormalities

(e) Vestibular investigations. Consider in cases where there are vestibular symptoms,, motor milestone delay or where there is fluctuating and/or progressive deafness (21).

Consider referral to Clinical Geneticist especially

- if the parents are consanguineous
- a syndrome is suspected
- child has multiple system abnormalities
- parental request
- opinion required on interpretation of genetic mutation testing
- after completion of investigations if a genetic disorder is diagnosed

In some cases following medical evaluation no cause for the deafness is found. These cases require continual review and in the future further investigations may need to be arranged and some previously normal investigations repeated.

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