Sensorineural Hearing Loss – Investigation & Referral

Epidemiology

1-3/1000 live births - 1/1000 at birth and 2/1000 at 9-16 years (due to delayed diagnosis, acquired hearing loss, late onset or progressive hearing loss).
In developed countries with immunisations completed there is decreased incidence of sensorineural hearing loss (SNHL) caused by congenital rubella, meningitis (especially H influenza B), perinatal events, ototoxic drugs, consanguinity.
Relatively increased incidence of congenital CMV.
GJB2 gene mutation variability related to ethnicity - commonest in each group are:
- 35delG in Caucasians
- 167delT in Ashkenazi Jews
- 235delC in East Asians.

NZ Hearing Screening

- UNBHS-automated oto-acoustic emissions (OAE and AABR if no emissions at first screen). Likely to change to AABR only screening
- 4 year screening pure tone audiometry – Well Child Check

Referral Pathway

Screened children needing a diagnostic assessment are referred to Audiology services for sleep ABR. Diagnostic testing should be complete by 3 months of age. Appropriate amplification should be complete by 6 months of age.
In Auckland: Audiology at Manukau Super Clinic and at Starship (for ADHB and WDHB).

Important Services

- Audiology
- ORL- at MMH: Andrew Gordon, Lesley Salkeld, David Flint
- ORL- at SCH: Colin Brown, Michel Neeff, Lesley Salkeld
- Paediatrics
- Auckland Genetics: Nerine Gregersen, Ian Hayes, Juliet Taylor
- Ministry of Education Special Education – Advisor on Deaf Children and SLT
- Kelston School for Deaf Education
- Hearing House and North Island Cochlear Implant Team
When to Investigate

1. **Bilateral permanent mild to moderate Hearing Loss**
   Sensorineural, conductive or mixed hearing loss with average hearing level of 20-69 dB HL measured in the better hearing ear averaged across 0.5, 1, 2, 4 kHz. If asymmetric hearing loss, investigate according to the worst hearing ear.

2. **Bilateral severe to profound Hearing Loss**
   Over 70dB hearing loss measured in the better hearing ear averaged across 0.5, 1, 2 and 4 kHz.

3. **Unilateral SNHL**
   Investigate aggressively, as good hearing ear may decline later.

4. **Auditory Neuropathy Spectrum Disorder (Auditory Dys-synchrony)**
   Bilateral and unilateral. (Normal OAE but absent ABR, abnormal cochlear microphonics). Approximately 10% of all children diagnosed with sensorineural hearing loss at birth will have this condition. Other neurologic conditions not common, but should be considered.

5. **Permanent conductive hearing loss** (Usually atresia of the external auditory canal)

Initial Assessment

**Family history, history, and physical examination (all):**
- Consanguinity
- Ethnicity
- Mitochondrial history: family history of diabetes and deafness, aminoglycoside sensitivity
- Cardiac history: sudden deaths, SIDS, known long QT syndrome

**Patient History**
- Preterm, IUGR, perinatal asphyxia, infections during pregnancy, jaundice requiring exchange transfusion
- Meningitis especially S pneumonia
- Noise exposure
- Ototoxic drugs
- Head injury
- Otitis media

**Developmental History**
Of patients with SNHL 50% have intellectual disability, 12% have cerebral palsy and 10% have autism spectrum disorder.
SENSORINEURAL HEARING LOSS – INVESTIGATION & REFERRAL

Physical Examination - Look for syndromes:

Autosomal Dominant: Waardenburg, Branchio-Oto-Renal (BOR), Neurofibromatosis 2, Stickler, Treacher Collins, Osteogenesis Imperfecta

Autosomal Recessive: Usher type I, II, III (associated with retinitis pigmentosa and poor balance), Alport, Jervell and Lange-Nielson, Pendred

X-linked: Norrie disease, Otopalatal digital, Alport, congenital stapes fixation with perilymph Gusher

Also: CHARGE, OAV spectrum (hemifacial microsomia)

Referrals

The principle of initial diagnosis is that the audiologist, advisor on hearing impaired and deaf children, and the otorhinolaryngologist (or paediatrician) work as a team.

Otorhinolaryngology – refer all patients:
Initial work-up. Contact with Otolaryngologist within 48 hours of diagnosis, preferably immediate.

Ophthalmology – refer all patients:
- 50% have eye abnormalities, especially refractive errors
- Retinopathy from intra-uterine infection
- Retinitis pigmentosa
- Cataracts
- Severe myopia

Audiograms: 1st degree relatives (all siblings, consider parents and other family members)

Genetics Service – refer selected patients:
- Where family request further information about the cause of deafness, especially if they are concerned about recurrence risks, are planning pregnancies, or want to discuss prenatal testing or pre-implantation gestational diagnosis
- Where there is a family history of deafness
- Patients with syndromes
- Patients with deafness and other features raising concerns of a possible genetic condition

Paediatric Clinic – refer selected patients:
- History of developmental delay
- Medical or syndrome diagnosis and management
- Auditory neuropathy spectrum disorder

Ministry of Education – refer all patients:
Refer to Special Education for Speech Language Therapy and Advisor on Deaf Children

WINZ
Consider Child Disability Allowance & Taikura Trust assessment
Investigations

1. Imaging of temporal bones, inner and middle ear and brain stem – request on all:

MRI or High Resolution CT (HRCT) or both.
MRI under “feed n wrap” protocol is preferred if possible. Request 3D MRI of inner ears, with brain views and direct sagittal images of the internal auditory canals to demonstrate the 4 nerves in the internal auditory canals.
Approximately 20% have an abnormal CT or MRI.
Radiation dose of HRCT = 0.8 milli-Sievert (mS). HRCT also irradiates the orbits, hence mildly increasing risk of cataracts. NB: Compare to radiation exposure dose of 2 mS for head CT, 0.04mS for CXR, and 0.04 mS equates to one long-haul airplane flight.

MRI essential for Auditory Neuropathy Spectrum Disorder, developmental delay or neurologic abnormalities.

MRI recommended as first line investigation in adults to detect acoustic neuroma.

If LVA (Large Vestibular Aqueduct syndrome) diagnosed advise of increased risk of sudden deterioration of hearing following a head injury. Advise against playing contact sports with risk of head trauma.

In young children diagnosed and assessed later than 3 months, who will need GA for imaging, only request imaging if:
   i) hearing is deteriorating
   ii) child likely to need cochlear implant (profound deafness)
   iii) parents are very keen for genetic reasons.

If hearing is stable and i) – iii) don’t apply suggest wait until 3 years as can cooperate with HRCT without GA.

2. TORCHS (request CMV on all +/- other tests as indicated)

   CMV – Rapid urine test if able to be done within 3 weeks of birth – request on ALL
   Alternatively, if MRI and genetic testing non contributory request retrospective PCR on neonatal heel prick test. Need parental permission, request sent to National (newborn) Testing Centre (ntc@adhb.govt.nz) – email must contain date and place of birth and NHI, and mother’s name at time of birth, and a statement in the email that the requestor has permission.
   Abnormal in up to 20% of all SNHL. Beware this test is specific but not sensitive (i.e. false negative rate is high).

   Congenital rubella
   If under 15 months, i.e., no MMR, positive rubella antibodies are very suspicious, as wild rubella in an infant is rare. Recheck eyes and heart. Consider congenital rubella in overseas born children or NZ born children from women from overseas.
   Congenital toxoplasmosis and herpes as a cause of deafness are exceptionally rare.
3. Molecular genetics (request on all).
A 66 gene analysis covering all known hearing loss mutations is now available. The cost is approximately NZ $2000. Best requested in discussion with genetic services.

Alternative options are 11 gene chip test at ASPER Opthalmics, Estonia - done as a sendaway via DHB laboratories (cost approx. 240 Euros)— includes Pendrin analysis which may affect up to 9% of NZ Population - takes 8 weeks for results, or CDHB laboratory currently testing connexin 26 and 30 as initial screen test (current cost approx. NZ $170). CDHB can do Pendrin sequencing subsequently at increased cost.

4. Molecular karyotype /microarray
Only with associated developmental delay or dysmorphic features.

5. Urinalysis
Infrequently abnormal. Consider if no diagnosis from other testing. Alport syndrome-almost always have haematuria. Alport syndrome (XD, AR, usually type IV collagen synthesis genes for basement membrane of kidney, inner ear and eye).

6. ECG (request if severe congenital bilateral SNHL)
Severe congenital bilateral SNHL only. Test all if a family history of sudden death. Otherwise consider testing if no diagnosis from other testing. False negatives occur. Long QT syndrome in Jervell and Lange-Nielsen syndrome (AR, KCNQ1 and KCNE1 genes involved in formation of a potassium channel protein).

7. Neurometabolic tests.
Consider if globally developmentally delayed or neurologically abnormal
Urine MPS, amino acids, organic acids, pyruvate/lactate, ammonia, CSF lactate

8. Thyroid Function Tests.
Probably not useful. Pendred syndrome children can have normal or borderline thyroid function, so normal thyroid function studies does not exclude it. Goitre (usual onset after 10 years) and abnormal TFT in only 10%. Do thyroid testing if Pendred confirmed on genetic testing. Will need follow up testing - best interval not yet established.

9. Autoimmune disease screen.
Rare cause of SNHL. No specific diagnostic test. Consider ANA when progression of hearing loss, vestibular symptoms, and/or other systemic symptoms.

10. Renal Ultrasound.
If syndromal features: preauricular pits or sinuses, deformity of ear, branchial cleft or cysts; Mondini defect on imaging; multiple congenital abnormalities; permanent conductive or mixed hearing loss.
Indications for Cochlear Implantation

Unilateral implants are funded in NZ.

Severe-profound SNHL not responding sufficiently to hearing aids to establish normal speech and language.

Immediate referral to Cochlear Implant Programmes upon diagnosis of severe hearing loss or worse, including all cases of bilateral Auditory Neuropathy Spectrum Disorder.

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References