Why is my child deaf?

Each month we present authoritative advice on the investigation of a common clinical problem, specially written for family doctors by the Board of Continuing Medical Education of the Royal Australasian College of Physicians.

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Permanent sensorineural hearing loss in children is common – at least one in every thousand children has severe or profound deafness, and mild degrees are more common.[1] Unless identified early in childhood, deafness will greatly limit a child's acquisition of speech and language, with severe implications for the child's future life.

Language delay can be minimised in children with sensorineural deafness, but the deafness itself will not get better. Disabling language difficulty also certainly occurs in some children with conductive hearing loss (especially 'glue ear') but can be minimised by prompt detection and management.

Deaf babies can be identified as early as 12 to 24 hours after birth using electrophysiological screening tests, but unfortunately are usually identified much later. Data gathered by Australian Hearing indicate that in the immediate past as few as 11% of all deaf infants were identified by 12 months of age and it has been common to identify them first at 15 to 18 months of age – this has major implications for language development. (Australian Hearing is a government funded organisation that provides hearing assessments, hearing aid fitting and continuing review free of charge to all children and young adults up to the age of 21 years, and adults who are eligible.)

When the diagnosis of permanent sensorineural deafness is made, parents usually go through disbelief, anger, grieving and, sometimes, prolonged depression. These phases are usual with the diagnosis of any chronic disability. The anger is often directed at health professionals or family members. One of the first questions asked is 'Why is my child deaf?'. Increasingly, it is becoming possible to give an answer.

Major advances have been made in the genetics of hearing loss in the past three to four years, and improvements in scanning techniques have made it possible to find a definitive cause of the deafness in many children. In other cases, investigating the deafness may give the parents a lot more information about their child's condition, which will be helpful in management. The approach to childhood deafness outlined below can also be used for investigating an adult who has been deaf since childhood, although it is important to exclude the possibility that the deafness could have been acquired during childhood, for example, due to measles.

Site of deafness

At least 90% of sensorineural deafness in children is thought to be due to cochlear dysfunction. However, problems can sometimes exist in higher parts of the auditory pathway – that is, in the auditory

IN SUMMARY

- **It is now possible to find the cause of sensorineural deafness in many children.**
- **History, examination, family hearing studies and interview enable the exclusion, or establishment, of autosomal dominant and syndromic deafness.**
- **High resolution CT scanning, MRI, ECG and laboratory tests (blood and genetic screening) may determine environmental, nonsyndromic autosomal recessive and mitochondrial causes of deafness.**
- **A very small group of deaf children will have a nonobvious environmental determinant.**

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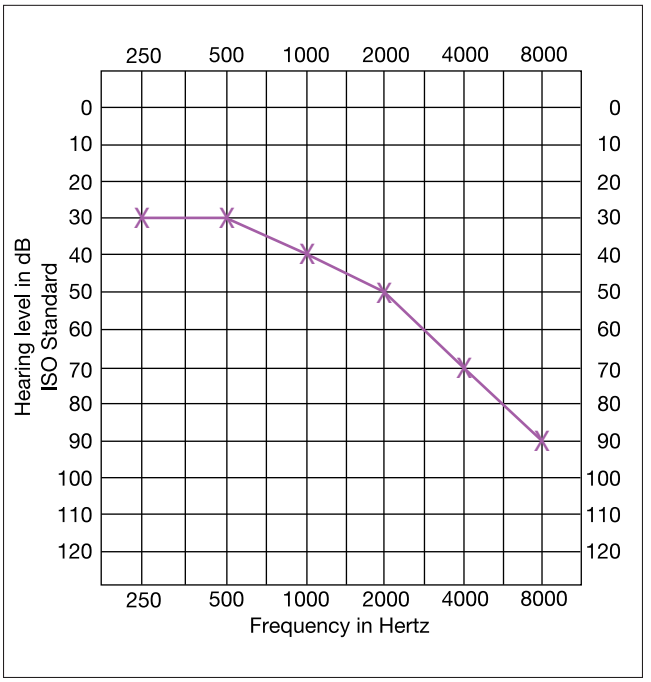
nerve before it joins the brainstem, in the brainstem and in the cerebral cortex. This retrocochlear hearing loss is usually also accompanied by cochlear hearing loss. Research on retrocochlear hearing loss is in its infancy as investigation is somewhat invasive. It appears to be most common in children with extreme prematurity, a history of hyperbilirubinaemia or craniofacial malformations that may involve hypoplasia of the auditory nerve. A search for retrocochlear components of deafness will involve a combination of otoacoustic emission testing, electrophysiological assessment and detailed scanning.

Investigation of cochlear hearing loss

Step 1. History and examination

It may be helpful to obtain information about the child's intrauterine development and birth and the postnatal and family history via a questionnaire before the first consultation. This may point to likely environmental causes of deafness. At the first interview, the child's history is reviewed and a careful physical examination is performed, with special emphasis on face, eyes, eardrums, external ears, neck, hair and skin. An examination by an ENT specialist should be organised, if this has not already been done.



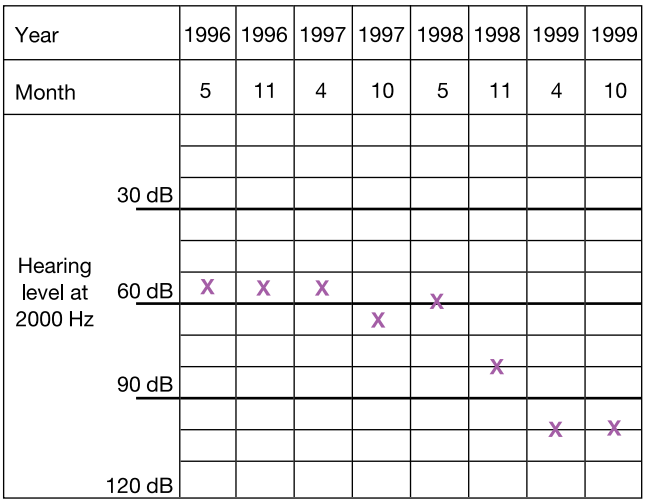


Figure 1 (left). A sloping audiogram of a child with profound hearing loss at high frequencies and a mild loss at low frequencies.
Figure 2 (above). The 2000 Hz segment of a left ear continuous audiogram showing that the threshold at that frequency had deteriorated over three years to a profound loss.

Hearing tests (audiograms) are important and can be obtained from Australian Hearing (Figure 1). It is important to check for fluctuation and deterioration in thresholds over time using the continuous audiogram (Figure 2). Hearing losses are usually rounded to the nearest 5 dB and are classified as follows, although definitions vary slighty according to the reference source:[2]

- a loss of 15 to 40 dB is regarded as 'mild'
- 45 to 65 dB as 'moderate'
- 70 to 90 dB as 'severe'
- greater than 90 dB as 'profound'.

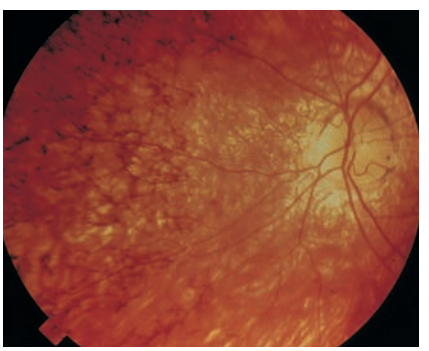


Figure 3. Retinopathy of Usher's syndrome. Note the mottled retina with clumping of pigment.

Most children with an average loss of 90 dB or less can be assisted substantially by hearing aids. However, for losses greater than 90 dB this may not be the case, and cochlear implantation or the addition of sign language may be considered for these children.

Almost all deaf and hearing impaired children in Australia who require hearing aids attend an Australian Hearing centre for their hearing tests and hearing aids.

Step 2. Examination of eyes

A thorough eye examination by an ophthalmologist will not only reveal possible refractive errors, which would be an additional burden to the deaf child, but may also give a clue to the aetiology of the deafness. For example, punctate retinopathy may indicate intrauterine infection by rubella or cytomegalovirus, while retinitis may indicate genetic conditions such as Usher's syndrome (Figure 3).

If a deteriorating visual condition such as Usher's syndrome is suspected, electroretinography is advisable because deteriorating vision will impact heavily on the best communication style for the child. A deaf child with deteriorating vision may be best managed by intensive auditory training and hearing aids or a cochlear implant, rather than sign language.

Step 3. Family hearing studies

The child's parents and siblings should have basic audiometry and, preferably, otoacoustic emission testing (which gives a crude measure of functional outer hair cells in the cochlea). The finding of a sibling with hearing loss, in the face of normal parental audiograms, suggests autosomal recessive or X-linked recessive deafness. The finding of parental hearing loss (that is not adventitious) may suggest that the child has dominantly inherited hearing loss. Rarely, it may suggest mitochondrially inherited hearing loss. Additional cases of hearing loss are commonly found.

Step 4. Family interview

A pedigree of the family should be drawn. It is important to check both the family

history and the child for thyroid disease (?Pendred's syndrome),[3] renal disease (?Alport's syndrome)[4] and skin/hair/eye pigmentation anomalies (?Waardenburg's syndrome).[5] Tiny pits or nodules around the external ear or neck are important genetically.[6] They are a flag to middle or inner ear malformations generally, and are an important feature of some specific syndromes, such as branchio–oto–renal syndrome. Possible parental consanguinity should be checked. Balance problems may point to inner ear malformations such as enlarged vestibular aqueducts. Testing young children for such problems is difficult, but getting them to run, jump and throw balls gives an idea of their ability to balance.

Conclusions so far

Having completed investigations to this point, the physician is likely to have excluded (or established) autosomal dominant and syndromic deafness. The remaining alternatives now comprise nonsyndromic autosomal recessive deafness, mitochondrially determined deafness (rarely), and a very small group with a nonobvious environmental determinant.

Step 5. Scanning of the inner and middle ear

High resolution CT scanning of the inner and middle ear is used to determine any abnormalities. Magnetic resonance imaging is useful if there is uncertainty about abnormality, and is strongly recommended if the child has any central nervous system abnormality or if cochlear implantation is being considered.

The most important abnormalities are:

- dilated vestibular aqueducts, which are often associated with fluctuating or deteriorating hearing
- Mondini abnormality of the cochlea (that is, less than 2.5 clear turns), either by itself or with dilated vestibular aqueducts.

Although Mondini syndrome has been reported to occur in deafness due to intrauterine rubella, it commonly appears to be genetically determined. Familial occurrence of these inner ear abnormalities is now established.[7]

Step 6. Electrocardiogram

An ECG should be done to exclude Jervell and Lange Nielsen syndrome (i.e. long QT, deafness, sudden death or collapse).

Step 7. Laboratory tests

Blood tests

Blood sampling in a child should be a 'one-hit' event. If the child has not yet been immunised with measles–mumps–rubella vaccine, positive serology for rubella in a child over 7 months of age suggests possible intrauterine rubella. Some cases of progressive hearing loss in

childhood are known to be due to intrauterine infection with cytomegalovirus.[8] However, it is difficult to establish intrauterine cytomegalovirus as a cause of deafness in an older child presenting with deafness because postnatal cytomegalovirus infection is a common, usually trivial, infection in young children and renders the child seropositive. Testing both the child and the mother for antibodies to cytomegalovirus can frequently lead to the exclusion of intrauterine cytomegalovirus as a cause of the deafness: if the child is negative for cytomegalovirus antibodies or if the child is positive but the mother is negative, the infection can be excluded.

In the same way, congenital toxoplasmosis can usually be excluded as a cause of deafness in the child. However, congenital toxoplasmosis seems to be an exceptionally rare cause of deafness in children seen in our clinics.

Genetic screening tests

Over 60% of nonsyndromic autosomal recessive deafness is caused by defects in the connexin 26 gene, which influences potassium ion transport in the endolymph of the inner ears.[9]

Approximately 70% of Caucasian children with defects in the connexin 26 gene have the common 35delG genetic mutation (deletion of guanine at position 35). Screening for the 35delG mutation is available after telephone discussion with the DNA Diagnostic Group at The Murdoch Children's Research Institute in Melbourne. Searching for other connexin 26 mutations may be indicated in certain patients (particularly some who are of non-Caucasian extraction). At present, this necessitates labour intensive and expensive sequencing of the whole connexin 26 gene.

Testing for abnormalities of the pendrin gene (which appears to determine Pendred's syndrome) is available from a few laboratories overseas and may become available in Australia in the near future.

Step 8. Counselling

After completing these investigations, the physician may have identified, or have a strong suspicion of, the cause of the child's deafness. Probably the single most common environmental agent implicated in deafness in young children is 'complicated prematurity'. The scenario of very low birthweight, need for ventilatory assistance, disturbance of acid-base balance, significant jaundice and possible use of aminoglycoside antibiotics appears to result in hearing impairment in around 3% of premature neonatal intensive care unit babies. The exact factor responsible has not been determined. However, ascribing deafness to prematurity should be done cautiously and only after other investigations have proved inconclusive.

If a clear-cut environmental determinant has not been identified, the family should be told that their child probably has a genetic deafness and warned that there is a recurrence risk for future children. This may best be done with the help of a geneticist.

In many cases, autosomal recessive deafness will have been diagnosed by connexin 26 testing or will be strongly suspected. The concept that both parents are 'carriers' and that the child is affected because he has a double-dose of the defective gene can be difficult for many families to understand. Forgetting genetic advice and misinterpretation is greatly reduced by giving families a personal letter about the recurrence risks for them.

It is not usually appropriate to suggest to parents that they curtail plans of future children because they have one hearing-impaired child.

Conclusion

Although sensorineural deafness is not yet curable, habilitation of the deaf child using hearing aids, education and, if necessary, cochlear implants or signed communication can be highly successful if deafness is identified early.

However, in most cases a deaf child places extra demands on his or her family. Many parents will want to space future pregnancies carefully. Despite anxieties and uncertainties, this will also give them the opportunity to enjoy their deaf child. MT

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