Why do we need to investigate a hearing loss?

Dr Shankar Rangan and **Dr Veronica Kennedy**, both Consultant Audiovestibular Physician (Paediatrics),

explain the processes involved in aetiology investigations

Introduction

Diagnosing the cause of the hearing loss in a deaf child is as important as management and rehabilitation of the deafness itself. Across the country, there is a wide variability of professionals undertaking aetiological investigations for hearing loss in children. There is also a marked difference in protocols followed in different regions for carrying out investigations to diagnose the cause of the deafness. The aim of this article is to try and clarify the importance of the various investigations for hearing loss in children and also to highlight the role of a medic in the multidisciplinary team looking after a deaf child.

At the outset we need to consider the reasons for investigating childhood hearing loss. These are:

- Parents often may want to know why their child is deaf.
- Predict if hearing loss is likely to get worse.
- Enable treatment that could prevent the progression of hearing loss as in the case of Congenital Cytomegalovirus (cCMV) infection.
- Diagnose any associated conditions.
- Give advice on lifestyle.
- Inform genetic counselling.

There are national guidelines for aetiological investigations for permanent hearing impairment in childhood. These have been produced by the British Association of Audiovestibular Physicians (BAAP). The guidelines can be accessed at: www.baap.org.uk/documents-guidelines-pathways-and-clinical-standards.html

Every child with any degree or laterality of permanent hearing impairment must be offered aetiological investigations. The investigations can be carried out by a medical practitioner with appropriate knowledge and skills. Some of the investigations are labelled as level 1 investigations and these should be offered in all cases of bilateral severe to profound sensorineural hearing loss (SNHL). The level 2 investigations can be offered under specific conditions.

In the case of a child with bilateral severe to profound SNHL the level 1 investigations would be:

- history:
  - general history, including that of pregnancy and birth
  - family history particularly of hearing loss going back up to three generations
- examination including clinical, developmental and vestibular
- Cytomegalovirus (CMV) testing
- ophthalmic assessment
- Connexin/genetic testing
- imaging: MRI/CT of inner ears
- family audiograms of first degree relatives
- urine test for haematuria, proteinuria
- ECG.

The level 2 investigations, depending on the individual case, could be:

- serology (HIV, rubella, toxoplasma, syphilis)
- thyroid function tests
- renal ultrasound
- autoimmune screen
- metabolic screen
- chromosomal studies
- further genetic testing and/or referral to clinical geneticist.

It is common for a child with a severe to profound hearing loss to also have issues with their balance. The incidence of vestibular problems can be significantly higher in children with sensorineural deafness and this can vary with the aetiology of the deafness. Vestibular loss may manifest in children as motor developmental delay. This requires specific attention as delayed motor development may be mistakenly attributed to developmental delay. It is recommended that all children with a permanent childhood hearing impairment (PCHI) should have a clinical vestibular assessment.

Review of the investigations

History and clinical examination

The importance of a thorough history and clinical examination cannot be overemphasised. This includes obtaining detailed prenatal, perinatal and postnatal histories. Perinatal factors have been reported to account for around a quarter of the cases of permanent hearing impairment in children. Various factors in the perinatal period, including length of stay in the neonatal intensive care unit, prematurity, low birth weight, asphyxia, respiratory distress syndrome, hyperbilirubinaemia, sepsis, ototoxic medications and intracranial haemorrhage, have all been reported to be risk factors for PCHI. It is believed a combination of risk factors is more likely to cause sensorineural hearing loss.

While the newborn hearing screen is effective at picking up hearing loss, it can only pick up any loss that is present at birth. The incidence of a PCHI is approximately 1:1000 at birth: this doubles in the first three years of life. There may be a family history of a late onset hearing loss in childhood. There would be an increased awareness with subsequent siblings if one is diagnosed with a late onset hearing loss.

In the postnatal period, meningitis is the most frequent cause of acquired sensorineural hearing loss. The hearing

loss after meningitis can be progressive and has been reported to deteriorate after many years.

An enquiry about the child's motor developmental milestones is important as there may be gross motor developmental delay in children with bilateral vestibular loss. The delay causes them to stand or walk later than their peers developing normally. It is important to obtain a full family history including details of hearing loss in first and second degree relatives.

Family audiograms

Age-appropriate hearing assessment of first-degree relatives has been recommended, even if there are no concerns, as unsuspected abnormalities may be uncovered and the configuration of the audiogram may also show a similar pattern among family members.

ECG

Sensorineural hearing loss (SNHL) may be associated with cardiac abnormalities as present in the Jervell and Lange-Nielsen Syndrome. In this syndrome, along with severe to profound SNHL, there is abnormal heart rhythm (prolongation of QT interval) which can be triggered by emotion or exercise. This can lead to syncope and/or sudden death.

Ophthalmic assessment

Deaf children will rely, more than a hearing child, on their vision for the development of language, social and communication skills. Ocular abnormalities have been found in more than a third of deaf children with refractive errors being the commonest. Ophthalmic examination will also enable identification of other ocular abnormalities that may be associated with deafness in syndromes like retinitis pigmentosa in Usher syndrome. Eye assessment may have to be repeated when the child is older as some of the abnormalities may not develop till later in life.

Urine analysis

A routine urinalysis or simple dipstick done in the clinic may pick up proteinuria or haematuria that is seen in Alport syndrome, in which SNHL is associated with progressive glomerulonephritis and sometimes ocular abnormalities. However, the haematuria or proteinuria in Alport may not develop until the child is older and hence the urine test may have to be repeated.

Cytomegalovirus (CMV) testing

One of the most significant advances that have been made recently in the field of aetiology of deafness, is in the area of congenital Cytomegalovirus infection (cCMV). Congenital CMV is the most common cause of congenital infections in humans. It is a leading cause of non-hereditary SNHL. Children with both symptomatic and asymptomatic congenital CMV can develop SNHL and the hearing loss can be progressive or of delayed onset.

Early antiviral therapy has been shown to prevent onset or deterioration of hearing loss in both symptomatic and asymptomatic cCMV infections. It is probably the only cause of congenital deafness where prevention of progression of the deafness can be achieved by early treatment. As current evidence stands, to be effective, antiviral therapy has to be started within the first month of life. However, diagnosing cCMV before this period can be tricky. The incubation period for CMV is 21 days, so CMV diagnosed after 21 days of life could be either congenital or acquired. Acquired CMV is not associated with SNHL.

The diagnosis of cCMV is essential to predict progression of deafness and other developmental delays. Vestibular problems, which can cause developmental delay, are also much more likely to be present with cCMV. Not diagnosing cCMV infection can have important implications as parents may not be counselled about the chance of concurrent disabilities. Therefore, routine testing for cCMV is particularly important.

In most areas, by the time babies with SNHL are seen by the medical professionals for investigations, they are older than four weeks. There should be local pathways to overcome this problem. Salivary swabs have been shown to be an easy and effective way of diagnosing cCMV. In some areas the audiologists, who see the babies earlier, take salivary swabs to diagnose cCMV. Another approach is for newborn hearing screeners to take salivary swabs of babies failing the hearing screening. This has been implemented in some areas of the country, but there are still logistical difficulties in making this a uniform practice across the UK

Genetic testing (for Connexin)

There are many genetic mutations associated with deafness. The most common are mutations in the Gap Junction Beta 2 gene (GJB2) located on chromosome 13 q and encoding the protein Connexin 26. The commonest mutation is 35 delG and is reported to account for about 80 % of the GJB2 mutations in the Caucasian population. Genetic testing in the UK is changing with a panel of deafness-related genes soon to be widely available for testing.

Radiology (MRI/CT)

Radiological imaging can have a high diagnostic yield in children with SNHL showing anatomical abnormalities causing hearing loss. The guidelines state all children with SNHL should have radiological imaging (MRI/CT) of their inner ears and brain. High resolution MRI has the added advantage of enabling other neuro-developmental abnormalities to be detected. Characteristic abnormalities in the white matter of the brain can be seen in congenital CMV infections and this can help in the diagnosis of cCMV. A dilated vestibular aqueduct (DVA) is the commonest abnormality noted on imaging for congenital SNHL. Identifying these structural abnormalities can be extremely helpful both in diagnosing the cause and in advising parents on the likely prognosis and management for that condition. The hearing loss in DVA can be progressive or there may be sudden drops in hearing triggered by even minor head trauma. The child may also experience episodes of dizziness. Parents of children with DVA should, therefore, be warned about the effects of any type of head trauma including those which can be caused by contact sports.

Nationally there is a wide variation in requesting MRI scans ranging from being requested in 20% to 100% of

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Norrie Disease

Theresa Peacock, a Trustee of The Norrie Disease Foundation, details more about the dual sensory loss

Norrie Disease (ND) is a rare, lifelong genetic condition that mainly affects males. ND is thought to affect approximately 40 people in the UK and 500 people globally, although this may be an underestimate.

What are the common features and symptoms of Norrie Disease?

- Children and young people with ND have vision impairment from birth, and most develop hearing loss that starts during adolescence.
- Some people with ND also experience problems with their development, growth, behaviour, sleep and feeding, as well as seizures and autism.
- The features and symptoms may vary in different children and young people with ND.

Further information about Norrie Disease can be found on the Norrie Disease Foundation website, there is also an educational leaflet available which gives guidance to schools and educational settings in supporting children and young people with Norrie Disease.

Below are the experiences of two families who have a child with Norrie Disease. The first is about my own son Ethan, the second is written by another parent about her son.

Experience 1

We became concerned about Ethan's vision when he was around 6 weeks old. Following a referral to see an ophthalmologist, we were told that he was essentially blind and the cause was Norrie Disease. Ethan was fortunate in that he had a small amount of useful vision, to begin with, but he would still need to be a tactile learner, using Braille for reading and writing and tactile diagrams for other aspects of learning. Ethan attended a mainstream primary school and is currently in year 10 at a grammar school. He is supported at school by a QTVI (Qualified Teacher of the Visually Impaired) team, has one-to-one support in lessons where needed and is provided with Braille and tactile resources.

Over the last couple of years, the level of useful vision Ethan had when he was younger has diminished and he is ▶

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PCHI cases. Scans that are done as soon as PCHI is diagnosed and before the child is three months of age, are advantageous because they can be done under natural sleep when the baby is still very young. As the child gets older, general anaesthetic or sedation would be required for the scan. Having a medic as part of the multi-disciplinary team looking after deaf children aids this process of arranging early scans and further appropriate management of any abnormalities detected in the scan.

Summary

Establishing the cause of the hearing loss is important for several reasons. It not only helps in better management of the hearing loss but can also play a pivotal role in preventing the progression of the deafness. Other conditions that may coexist with deafness can be picked up early and thus can be more effectively managed. The role of the doctor in the multidisciplinary team looking after a deaf child is crucial in establishing the aetiology of deafness. The doctor should take a lead in establishing early aetiological diagnosis and, as some associated conditions present later in childhood, should provide medical input at every stage of the care pathway throughout the child's development. The medical input is especially important in the management of a child with associated imbalance, dizziness or tinnitus and to coordinate the management of visual, developmental or other problems. Early diagnosis and rehabilitation of deafness leads to better outcomes for the child, not only in terms of speech and language development, but also in the child's development as a whole. Likewise, early diagnosis of the aetiology of the hearing loss leads to better management of the deafness.

Key messages

- Aetiological investigations have to be offered to every child with permanent childhood hearing impairment. Establishing the aetiology of the deafness at a very early stage of the care pathway leads to better management of the deafness. The role of the doctor in the team is vital to achieve this.
- There are national guidelines for aetiological investigations for hearing loss in children, they can be accessed online at www.baap.org.uk/documents-guidelines-pathways-and-clinical-standards.html
- Some investigations have to be done early
  - CMV testing within three weeks of age
  - MRI/CT scan within three months of age to enable the scan to be done under natural sleep
- Some of the investigations have to be repeated later in life, eg urine check and ophthalmic assessment.

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