

Incidence, Prevalence, and Aetiology of Childhood Hearing Loss

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Key Facts

- ❖ Incidence of permanent sensorineural hearing loss at birth: 1-2 per 1000 births
- ❖ Prevalence among children 9-16 years in Australia: 3.2 per 1000
- ❖ Aetiology of childhood hearing loss:
 - 50-60% genetic:
 - 30% syndromic
 - 70% non-syndromic
 - 40-50% non-genetic:
 - congenital infections (especially cytomegalovirus)
 - perinatal, e.g. prematurity, severe jaundice

Incidence and prevalence

The incidence of permanent sensorineural hearing loss at birth in Australia and other developed countries is estimated to be between 1 and 2 babies per 1000 live births.^{1 2} The prevalence increases during childhood, and the figure for Australian 9 to 16 year-olds with all degrees of hearing loss has been reported as 3.17 per 1000.³ There are several reasons for the increase over the childhood years. Some genetic and congenital non-genetic hearing losses have a post-natal onset or are progressive. Some children will acquire a hearing loss through childhood infections such as meningitis. As well, congenital hearing loss that is mild or unilateral may not be detected at neonatal hearing screening and may not be diagnosed until after infancy.³

Australian Hearing figures for 2012 indicate that the total number of Australian children and young people up to the age of 21 years who had been fitted with hearing aids or cochlear implants was 17,394. Almost 10% of these identified as Aboriginal or Torres Strait Islander.⁴

The majority of children with permanent hearing loss have a mild or unilateral loss. The distribution of aided hearing loss (3 Frequency Average Hearing Loss in the better ear) in Australian children and young people up to the age of 21 in 2010 was: 55% 0-40dB; 22% 41-60dB; 12% 61-90 dB; 10% 91+dB (1% unknown).⁵

Aetiology

The aetiology, or cause, of permanent childhood hearing loss is either genetic or non-genetic.





Genetic causes

The American College of Medical Genetics and Genomics estimates that up to 60% of educationally significant congenital and early-onset hearing loss is caused by genetic factors, even though around 95% of deaf and hard of hearing children are born to hearing parents. Most genetic hearing loss is inherited in an autorecessive pattern, where both parents must carry the gene for hearing loss to be present in their children, and there is usually no family history of hearing loss.⁶ Recent advances in genetics have enabled the identification of many genes associated with hearing loss. Genetic deafness can be either syndromic or nonsyndromic.

Syndromic genetic causes

Approximately 30% of genetic hearing losses are part of a syndrome, where there is the presence of distinctive associated clinical conditions.

Pendred syndrome is the commonest form of syndromic deafness. It typically involves enlargement of the vestibular aqueduct as well as thyroid problems. The hearing loss is usually profound and bilateral, but is not always present at birth. Onset can occur during infancy and early childhood, and the hearing loss can rapidly progress.

Waardenburg syndrome can cause bilateral or unilateral hearing loss of variable severity. The syndrome typically involves pigmentation abnormalities and gastrointestinal symptoms. Only 20-50% of individuals with Waardenburg syndrome will have hearing loss.

Usher syndrome involves deafness and retinitis pigmentosa, a progressive degeneration of the retina that leads, eventually, to blindness. In Usher syndrome Type 1 there is a profound congenital hearing loss and a relatively early onset of retinitis pigmentosa, often during the first decade of life. Type 1 affects around 40% of all individuals with Usher syndrome. Usher syndrome Type 2, affecting about 57%, involves a less severe hearing loss and a later onset of retinitis pigmentosa. Type 3, affecting around 3%, has a variable severity of hearing loss which can be progressive.

CHARGE syndrome involves cochlear and semicircular canal anomalies. Other characteristics can include congenital heart defects, kidney problems, cleft lip or palate, and growth retardation. The hearing loss can be sensorineural, conductive, or mixed. CHARGE syndrome is rare, estimated to affect about 1 in 10,000 live births.

There are many other syndromes that can involve permanent hearing loss, including Branchio-oto-renal (BOR) syndrome, Jervell, Lang-Neilsen syndrome (JLNS), Down syndrome, and Alport syndrome.^{6,7}

Non-syndromic genetic causes

About 70% of cases of genetic hearing losses are not associated with a syndrome. The most common cause of genetic non-syndromic hearing loss is a mutation on the autorecessive gene GJB2 that produces the protein connexin 26. These hearing losses are usually present at birth, bilateral, not progressive, and range from mild to profound. However, there are cases of connexin 26 hearing loss that have a later onset and are progressive.⁷



Non-genetic causes

Non-genetic, or environmental, causes of childhood hearing loss include maternal infections during pregnancy, prematurity, and early childhood diseases such as meningitis. In recent decades the proportion of environmental causes has changed considerably. Congenital rubella, once a common cause of infant deafness, is now rare in many countries due to successful immunization programs.

Maternal infections

Currently, the most common cause of congenital environmental hearing loss is cytomegalovirus (CMV) infection in the mother during pregnancy. CMV is a common, flu-like virus. It affects 1-2% of newborns, and an estimated 10-15% of these will develop sensorineural hearing loss. The majority of babies with congenital CMV show no symptoms at birth, and the onset of hearing loss may not occur for months, or even years, after birth. The hearing loss can be bilateral or unilateral, fluctuating, or progressive.^{1 8}

Perinatal causes

Risk factors in newborns include events occurring during or shortly after birth, such as prematurity and low birth weight, asphyxia (a lack of oxygen to the brain and other organs), severe jaundice, neonatal convulsions, and ototoxic drugs given for infections. Survivors of neonatal intensive care units have a much higher incidence of permanent hearing loss than the whole population of newborns, with estimates ranging from 4% to 18%.⁹

Childhood infections

Meningitis (meningococcal disease affecting the brain) contracted in childhood has a high risk of causing permanent hearing loss. Cases of bacterial meningitis have declined in Australia due to the implementation of meningococcal C vaccination programs from 2003. However, there are other strains of meningococcal disease that are not currently routinely covered by immunization programs.

In a large number of children, often around 40%, the cause of hearing loss is unknown. Many of these undetermined cases are likely to be due to genetic non-syndromic causes or congenital CMV infection.^{10 11}

Auditory Neuropathy Spectrum Disorder

A subgroup of children with permanent hearing loss has auditory neuropathy or auditory dyssynchrony, commonly known as Auditory Neuropathy Spectrum Disorder, (ANSD). Typically in ANSD there are normally functioning cochlear outer hair cells but abnormal auditory brainstem responses. It may be present in 10 to 15% of all children with hearing loss and in 15 to 20% of children with severe or profound hearing loss. The degree of hearing loss can range from mild to profound, but often involves difficulties with speech discrimination. Hearing aids are helpful for many children with ANSD, but are ineffective for some who find speech sounds very distorted. Depending on the development of the auditory nerve, cochlear implants may work well. ANSD can be genetic in cause, and can be part of syndromes including Charcot-Marie-Tooth syndrome and Friedreich's ataxia; it is also associated with perinatal causes including asphyxia and severe jaundice.¹²



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