Congenital Hearing Loss

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9.1 Introduction

Hearing loss present at birth (congenital hearing loss) is categorized according to the underlying pathology to hereditary (syndromic or non-syndromic), non-hereditary, and idiopathic. Three types of hearing loss can be induced depending on the site of the lesion: conductive, sensorineural, or a combination of both (mixed) [1].

9.2 Neonatal Hearing Screening

Newborn hearing screening programs show that the incidence of congenital hearing loss is 2–4 children per 1000 births, and it is considered as the most frequently occurring birth defect in the US [2, 3]. Mutations in GJB2 account for 50% of people with severe-to-profound congenital autosomal-recessive non-syndromic hearing loss [4].

Neonatal hearing screening programs are available for the early detection of this condition. More than 50% of cases of permanent hearing impairment in childhood can be detected shortly after birth [5]. Using the 1-3-6 model intends to screen all newborns within the first month of birth for early diagnosis and subsequent early management with a better developmental outcome [6]. View the algorithm below to check the steps of the screening. However, passing the neonatal screening does not rule out hearing impairment in childhood. Progressive or late-onset hearing loss can be undetected by neonatal screening programs [6].

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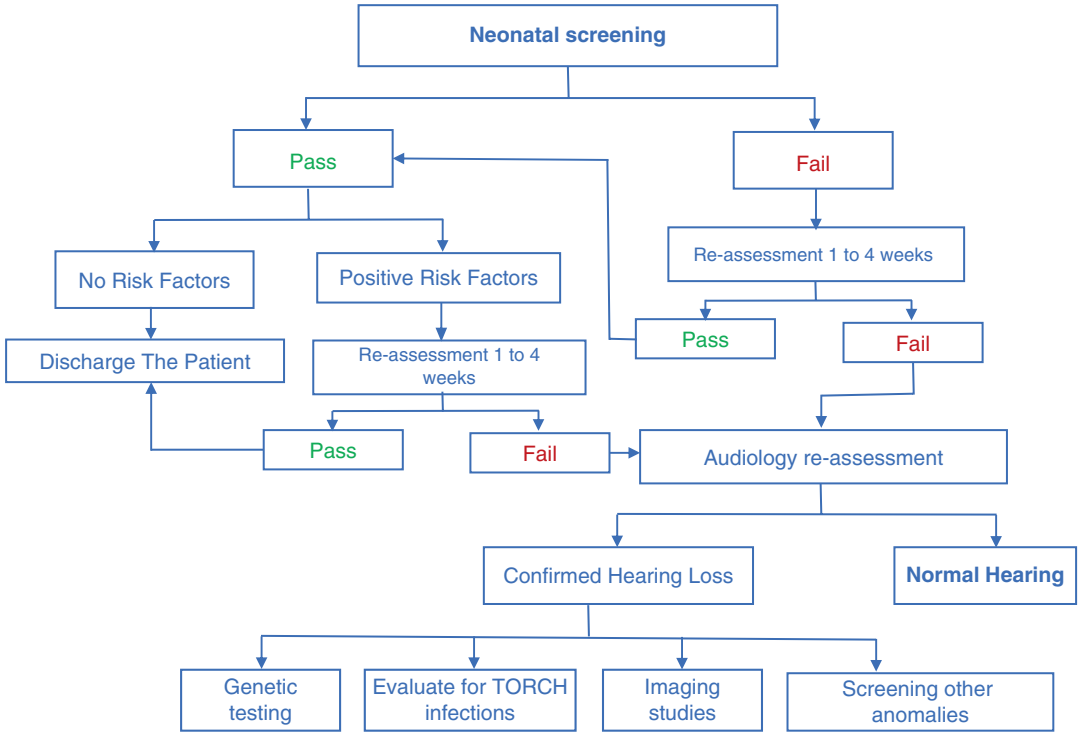

9.3 Evaluation of a Child with Congenital Hearing Loss

The general approach to evaluating the child with suspected congenital hearing loss includes the following:

- **History**: pregnancy history, perinatal and postnatal period, NICU admission, postnatal infection, ototoxic medications, co-existing medical conditions, family history of hearing loss in first- and second-degree relatives, consanguinity, and ethnic origin.
- **Physical examination**: a full head and neck examination should be performed to including any dysmorphic features, the shape, and position of the external ears, neck examination for cysts, sinuses, and scars (branchio-oto-renal syndrome), swelling of the thyroid gland (may indicate Pendred syndrome), and unusual pigmentation of the hair, skin, or eyes (which may indicate an auditory pigmentary disorder).
- **Investigation**: the following should be considered: genetic testing, CT/MRI imaging, thyroid function tests, cardiology evaluation (Echo and ECG, for possible association with Jervell and Lange-Nielsen syndrome or congenital heart conditions), or ophthalmology assessment (electroretinogram if suspected Usher syndrome).

Risk factors for permanent congenital, delayed, or progressive hearing loss in childhood are described in Box 9.1 [5].

Box 9.1 Risk Factors for Permanent Congenital, Delayed or Progressive Hearing Loss in Childhood

- Hearing, speech, language, or developmental delay
- Family history of hearing loss
- Neonatal intensive care unit stay >5 days **or** receiving any of the following treatments: extra corporal membrane oxygenation, assisted ventilation, ototoxic drugs (e.g., gentamycin and tobramycin), loop diuretics, or exchange transfusion for hyperbilirubinemia
- In utero infections (toxoplasmosis, rubella, cytomegalovirus, herpes simplex, or syphilis)
- Craniofacial anomalies, including ear tags, ear pits, and anomalies that involve the outer ear, external auditory canal, and temporal bone
- Physical findings associated with a syndrome known to cause permanent hearing loss (e.g.,, white forelock)
- Syndromes associated with congenital hearing loss or progressive or late-onset hearing loss
- Neurodegenerative disorders or sensorimotor neuropathies
- Confirmed bacterial or viral meningitis
- Head trauma, especially of the basal skull, or temporal bone fractures that require hospitalization
- Chemotherapy

9.4 Categories of Congenital Hearing Loss

1. **Idiopathic (25%).**
2. **Hereditary (50%):**
   - **Non-syndromic (70%)**: more common, the prefix "DFN" to designate non-syndromic DeaFNess, DFN followed by an A implies dominant inheritance, whereas B implies recessive inheritance and X implies X-linked inheritance. Autosomal recessive (AR) in (75–80%), GJ2B mutation (autosomal recessive) coding for the protein gap junction beta 2 (also called connexin 26) results in impaired Potassium (K^+) exchange. Autosomal dominant (AD) in (20–25%), X-linked in (2–4%), and mitochondrial in <1% [5].
   - **Syndromic (30%)**: Divided into AR, AD, and X-linked. The most common syndromic form of hereditary SNHL is Usher syndrome [7], and other AR syndromes include Pendred syndrome and Jervell and Lange–Nielsen Syndrome. The most common AD syndrome is Waardenburg [7], and others include Treacher–Collins Syndrome (Fig. 9.1), Branchio-oto-renal Syndrome (mainly mixed hearing loss), Neurofibromatosis Type 2, and Stickler Syndrome. X-linked includes Alport's syndrome. Box 9.2 describes the features of the most common congenital hereditary hearing loss syndromes.
3. **Non-hereditary (25%)**
   - **Malformations**: arrest in normal development may result in hearing impairment depending on the timing and nature of the developmental insult. About 65% of such abnormalities are bilateral, and 35% are unilateral. Malformations include *Membranous* (Alexander's Aplasia, Scheibe Deformity, and Siebenmann–Bing Dysplasia) and *Osseous and Membranous* (Cochlear Hypoplasia, Mondini, Common Cavity, Cochlear Aplasia, Michel Aplasia, and Small Internal Auditory Canal). Mondini malformation is the most common type of cochlear malformation [1].
     - **Alexander aplasia** is one of the membranous malformations, where cochlear duct differentiation at the level of the basal coil is limited with resultant effects on the organ of Corti and the ganglion cells. Hearing assessment shows high-frequency sensorineural hearing loss with adequate residual hearing in the low frequencies, and amplification devices can be used.
     - **Scheibe aplasia (cochleosaccular dysplasia or Pars Inferior dysplasia)** is one of the membranous malformations, and it is a relatively common cochlear malformation. Scala media is compromised due to the failure of the organ of Corti development affecting the tectorial and the Reissner's membranes. It is

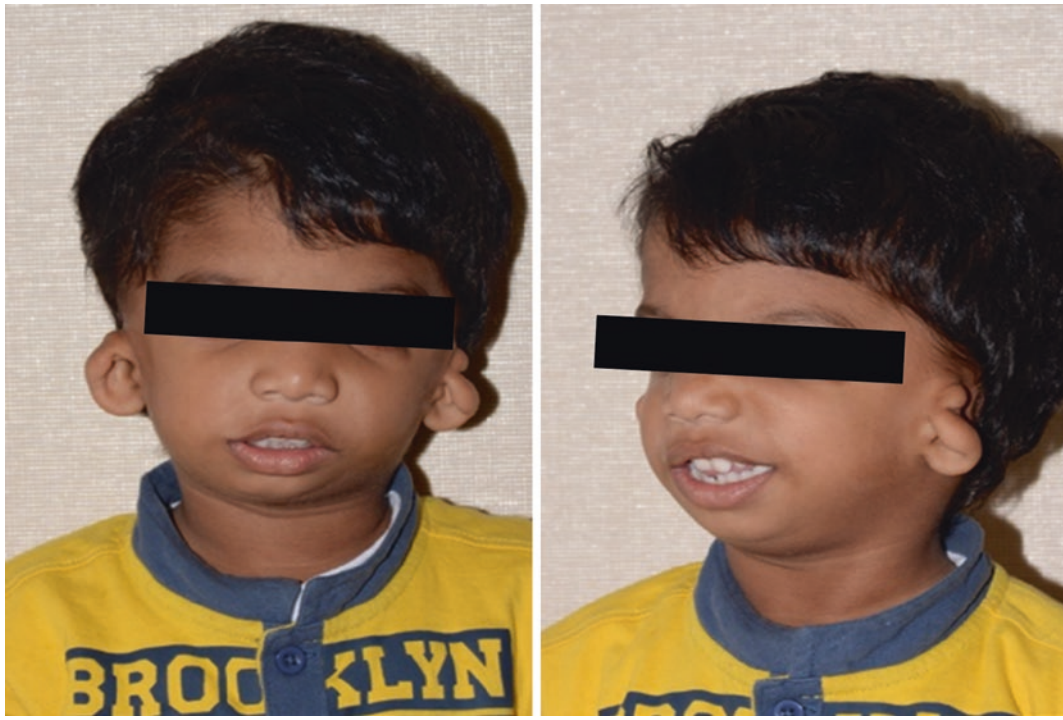

Fig. 9.1 Treacher–Collins syndrome

reported in Jervell and Lange Nielsen, Usher, and Waardenburg syndromes as well as in congenital rubella infants. Amplification devices with rehabilitative intervention are beneficial in many of these children.

– **Michel aplasia** is one of the osseous and membranous malformations, where there is complete agenesis of the petrous portion of the temporal bone resulting in absence of normal inner structures and therefore complete deafness. The developmental arrest occurs early prior to the end of the third gestational week.
– **Mondini deformity** is one of the osseous and membranous malformations, and it is the most common type of cochlear malformation [1]. The developmental arrest occurs at the sixth week of gestation. It presents with an incomplete partition of the cochlea where it contains only about 1.5 turns with only the basal coil and absence of the apical modiolus and interscalar septum. Mondini deformity is associated with enlarged vestibular aqueduct and dilated vestibule. It is seen in Pendred, Waardenburg, Treacher–Collins, and CHARGE syndromes, and it is also seen in congenital cytomegalovirus (CMV) infection.
– **Enlarged Vestibular Aqueduct Syndrome** is defined as a vestibular aqueduct measuring more than 1.5 mm on CT scan. It is thought to be one of the most common congenital causes of sensorineural hearing loss. It presents with early onset bilateral progressive sensorineural hearing loss. It is associated with Pendred syndrome, vestibular anomalies, Mondini malformation, and other cochlear anomalies. Mainstay of treatment is conservative management, including avoidance of head trauma and contact sports.

- **TORCH Infections**: It includes Toxoplasmosis, Other (syphilis, varicella-zoster, parvovirus B19), Rubella, Cytomegalovirus (CMV), and Herpes.
  - Cytomegalovirus: the most common cause of congenital viral deafness [8].
  - Mumps: the most common infectious cause of acquired sensorineural hearing loss.
- **Other perinatal factors**: prematurity, low birth weight, teratogenic drugs, maternal diabetes, hyperbilirubinemia, and neonatal sepsis.

Box 9.2 Overview of Congenital Hearing Loss Syndromes

Syndrome	Features	Comments
Autosomal recessive		
Usher syndrome	Sensorineural hearing loss, retinitis pigmentosa, and vestibular symptoms **Type 1**, profound hearing loss, vestibular symptoms, and retinitis pigmentosa beginning in first decade **Type 2**, stable mild to severe hearing loss and retinitis pigmentosa in first to second decade **Type 3**, progressive hearing loss, variable vestibular symptoms, and variable onset of retinitis pigmentosa	Commonest cause of deafness with blindness, early diagnosis of retinitis pigmentosa possible with electroretinography, and cochlear implants effective
Pendred's syndrome	Sensorineural hearing loss (severe to profound), goiter, and abnormality of the bony labyrinth (Mondini dysplasia or enlarged vestibular aqueduct)	Iodine transport defect diagnosed by perchlorate discharge test in those homozygous for Pendred's syndrome (SLC26A4) and euthyroid goiter
Jervell and Lange-Nielsen syndrome	Profound sensorineural hearing loss with prolongation of QT interval, syncopal episodes, and risk of sudden death	Should have a thorough cardiac evaluation, carriers also at risk for sudden death, and treatment effective
Autosomal dominant		
Waardenburg syndrome	Most common type of AD; hair (white forelock 1), eyes (heterochromia iridis) **Type 1**, white forelock and dystopia canthorum **Type 2**, dystopia absent **Type 3**, upper limb defects **Type 4**, increased incidence of Hirschsprung's disease	Hearing loss caused by defective migration of pigment cells to stria vascularis
Treacher–Collins syndrome	Conductive hearing loss, underdevelopment of facial bones (malar and zygomatic hypoplasia) with malformed ossicles, microtia, cleft palate; micrognathia, downward slanting eyes, coloboma of the eyelid	
Branchio-oto-renal (BOR) syndrome	Hearing loss; preauricular pits; malformed pinnae; Branchial cysts or fistulae; renal anomalies (structural malformations to agenesis)	
X-linked		
Alport's syndrome	Hematuria with progressive renal failure, progressive late-onset high-frequency sensorineural hearing loss; anterior lenticonus and macular flecks	Results from mutations in 1 of 3 collagen genes expressed in glomerular basement membrane

Take-Home Messages

- The most common prenatal cause of hearing loss is intrauterine infection (CMV).
- Mutations in GJB2 account for 50% of severe-to-profound autosomal-recessive non-syndromic deafness.
- The goal of newborn testing is to screen by 1 month of age, diagnose hearing loss by 3 months of age, and initiate intervention by 6 months of age.
- Screening based on risk factors only detects 50% of infants with substantial congenital hearing loss.

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