ETIOLOGICAL ASSESSMENT OF CONGENITAL PROFOUND SENSORINEURAL HEARING LOSS

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Introduction:
Congenital profound sensorineural hearing losses SNHL are determined by genetic or nongenetic factors.
Hearing loss is a common symptom in children. It may be congenital or acquired and is classified as conductive hearing loss, sensorineural hearing loss, and mixed hearing loss.

To further evaluate more accurately congenital profound sensorineural hearing losses from etiologic point of view.

Methods:
Congenital profound sensorineural hearing loss etiological assessment is a critical component in infants and children clinical and paraclinical evaluation.
The most frequently identified causes of SNHL are infectious, anatomical, and genetic.
Infectious causes include congenital cytomegalovirus and postnatal bacterial meningitis.
The most common anatomical cause of SNHL is enlarged vestibular aqueduct; and other inner ear anomalies often have a genetic basis.
The most common genetic causes are mutations in the gap junction JJ 2 gene (GJB2) encoding the connexin 26 (Cx26) protein.

In many countries, > 50% ofthe SNHL is genetic, with 30% being syndromic and the remainder nonsyndromic.
Nongenetic factors might be implied in about 25% of congenital hearing losses, while genetic factors (hereditary) are thought to determine more than 50% of all hearing losses.
Evaluation of nongenetic and genetic factors will allow to determine a disease progression pattern and will facilitate associated clinical manifestations and complications monitoring.

Of the nonsyndromic causes, 80% are autosomal recessive, 15 to 17% are autosomal dominant, 2 to 3% are X-linked, and -1% are mitochondrial.

Results:
Auditory dyssynchrony is a subset of SNHL and presents and evolves on a spectrum. Some children will have significant spontaneous improvement, developing useful hearing that supports spoken language development.
Auditory dyssynchrony associated with anatomical anomalies is generally permanent. Children with the bilateral variety who have generally normal temporal bone anatomy, but who do not develop significant usable hearing that supports spoken language may benefit from cochlear implants.

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General neonatal screening for hearing impairment is recommended since diagnosis of hearing impairment is often delayed and early intervention is of great importance. All newborns should be screened during their first days of life. Screening methods should have high sensitivity and specificity, and should be objective as well as time- and cost-efficient. The following methods are available:

- Automated brainstem evoked audiometry (BERA)
- Automated measurements of amplitude modulation following response (AMFR)

If the child fails to pass the screening for hearing impairment, follow-up with eventual rescreening and more extensive auditory testing to confirm or exclude hearing loss is necessary.

Evidence confirms that early identification and treatment, coupled with sustained, appropriate habilitation and educational support can achieve excellent outcomes.

Development of spoken language can proceed at rates similar to that for normal-hearing children even in profoundly deaf children, provided that early identification and cochlear implantation are achieved.

Conclusion:
In congenital profound sensorineural hearing loss etiological assessment is mandatory a multidisciplinary approach: ENT surgeon, neonatologist, pediatrician, geneticist, radiologist and other specialists, depending on each case.

References: