AUDIOLOGICAL ISSUES IN A CASE OF CHARGE SYNDROME

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Abstract

Background: CHARGE syndrome is a genetic disorder resulting in the association of multiple congenital anomalies. The diagnosis of CHARGE syndrome is based on a combination of major and minor characteristics. Approximately half of individuals with CHARGE have severe to profound hearing loss.

Materials and Methods: The authors present a case of a 4 year old child diagnosed with CHARGE syndrome that was referred to our department due to complete absence of expressive language and reaction to sounds. The protocol included: acoustic immittance, observation audiometry and click ABR on air and bone conduction under natural sleep and short duration general anesthesia.

Results: The investigations revealed mixed profound hearing loss with middle ear effusion on the left side and severe sensorineural hearing loss on the right ear. The first option of treatment was classic bilateral hearing aid, after grommet insertion in the left tympanic membrane and effusion drainage.

Conclusions: Any infant suspected of having CHARGE syndrome should undergo complete audiological examination since the frequency of sensory-neural hearing loss or vestibular problems are as high as 75-80%. Evaluation of hearing sensitivity during infancy and provision of amplification are important components in the process of auditory habilitation. Negligence in monitoring for auditory disabilities may result in increased developmental delay.

Key words: CHARGE • hearing loss • auditory habilitation • multiple malformations

Background

CHARGE syndrome is a non-random association of anomalies and malformations usually caharacterised by the association of Coloboma, Heart, Atresia choanae, Retarded growth and development, Genital hypoplasia and ear anomalies/deafness [1]. It has estimated incidence of 0.1–1.2 of 100 000 live newborns and it is supposed to have its origin in a mutation on the chromosome 8 consisting in microdeletions and mutations of the CHD 7 gene [2,3]. For educational purposes the CHARGE is considered a deaf-blind syndrome although complete deafness and blindness are very rare. These children have unusually shaped external ears, conductive and/or sensori-neural hearing loss ranging from mild to severe and also malformations of the structures of middle and internal ear [4]. There is no characteristic hearing loss in CHARGE syndrome. The most common features are asymmetrical mixed losses that are severe-to-profound in degree. Conductive components, which are due to a combination of ossicular anomalies and middle ear effusion, are often asymmetrical and fluctuating in nature; cochlear hearing loss, often due to cochlear malformations, is typically greater in the high frequencies [4]. This added to mental retardation and vision impairment generate a lack of sensorial input to which the child responds with behavioural changes that can become extremely severe, such as destructive or auto mutilation [5]. All this make audiological rehabilitation extremely difficult but sometimes extremely beneficial for the child, since the self hurting behaviours tend to disappear after audiological intervention [6].

Materials and Methods

We present the case of a 4 year old child diagnosed with CHARGE syndrome at the age of 2, after identification of multiple malformations: unilateral choanal atresia, multiple outlet right ventricle, heart septum double defect, bilateral external ear malformations, micropenis, cryporchidism, motor and mental retardation, behavioral changes and delayed somatic growth.

He underwent multiple surgeries for the life threatening malformation and the prolonged hospitalisation delayed the audiological diagnosis and rehabilitation.

The child had bilateral external ear malformations, complete lack of speech development and medium mental retardation (Figure 1).

The audiological evaluation protocol began with acoustic immittance and air and bone conduction click ABRs (auditory brainstem responses) in natural sleep. We identified a fluid collection in the left middle ear consistent with a permanent status of serous otitis media. Therefore a new air conduction click examination was performed after drainage of the effusion from the left ear. Observation audiometry in free field and bone conduction completed the audiological protocol.

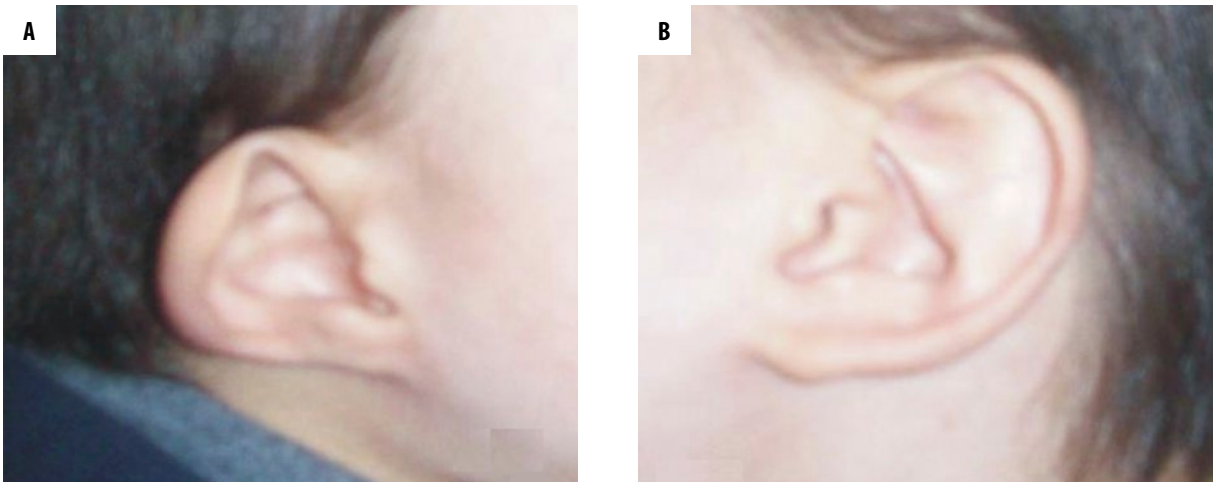

Figure 1. Bilateral external ear malformation. (**A**) Right ear: Outstanding ear, soft cartilage, attached lobule, mild stenosis of the external ear canal. (**B**) Left ear: Soft cartilage, triangular concha, attached lobule, normal external ear canal.

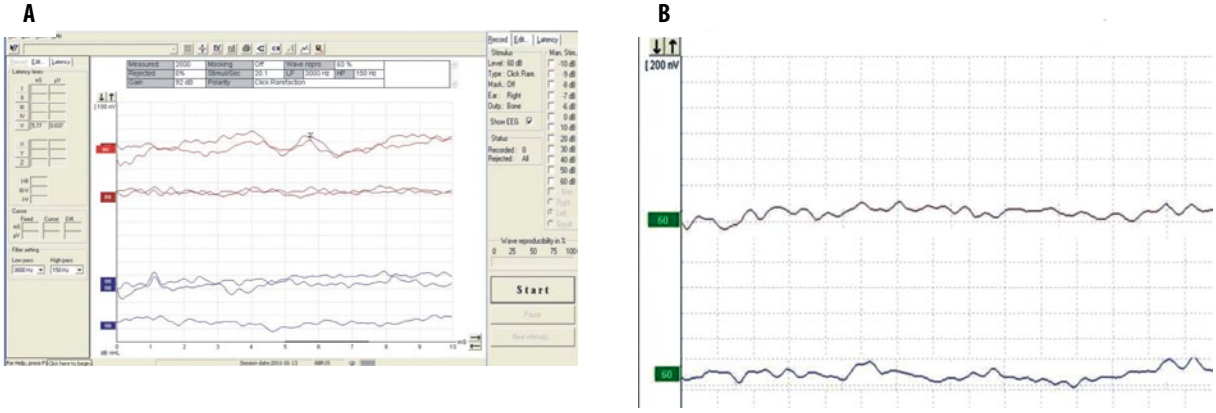

Figure 2. (**A**) AC click ABR wave V threshold present at 90 dB nHL at the right ear. (**B**) No ABR responses using bone conduction stimulation on both sides.

Results

Acoustic immittance revealed for the right ear: type A tympanogram and absent stapedial reflex and for the left ear a type B tympanogram suggesting fluid collection in the middle ear. Objective evaluation was made in two stages. First was performed air conduction (AC) and bone conduction (BC) click ABRs evaluation in natural sleep. We found wave V threshold present at 90 dB nHL at the right ear; the latency of wave V was not shifted out in time. Complete lack of air conduction click ABR evoked response at 90 dB nHL for the left ear and no bone conduction click ABR responses on both sides were observed (Figure 2). We attempted to test the left ear at 100 dB nHL but the child woke up and continuation of examination became impossible.

The second stage of audiometric examination was performed after two weeks under general anesthesia. The left middle ear effusion was drained and a grommet was inserted to the left tympanic membrane. Air conduction click ABR was again performed and wave V threshold at 100 dB nHL was found at the left ear.

Observational audiometry tested in free field revealed reactions of high intensity for stimuli of lower and medium frequency but no reactions were found when testing bone conduction at maximum intensity levels (Figure 3A).

As a management of the child's hearing loss, classic bilateral hearing aids were fitted. At the left ear the hearing aid was added 4 weeks after grommet insertion, because he developed an infectious complication soon after surgery.

The assessment of hearing aid benefit revealed free field thresholds with hearing aids at medium intensity levels (Figure 3B). The hearing aid gain of about 60 dB SPL allowed the child to wear the devices with a skull retention system without encountering acoustic feedback. Increase in sound responsiveness and slight improvement of the child behavior were observed.

The family decided that the child should start the rehabilitation therapy in a spoken and sign language rehabilitation center.

Discussion

Behavioral troubles with autistic-like elements delay audiological assessment and hearing loss diagnosis in children suffering from CHARGE syndrome.

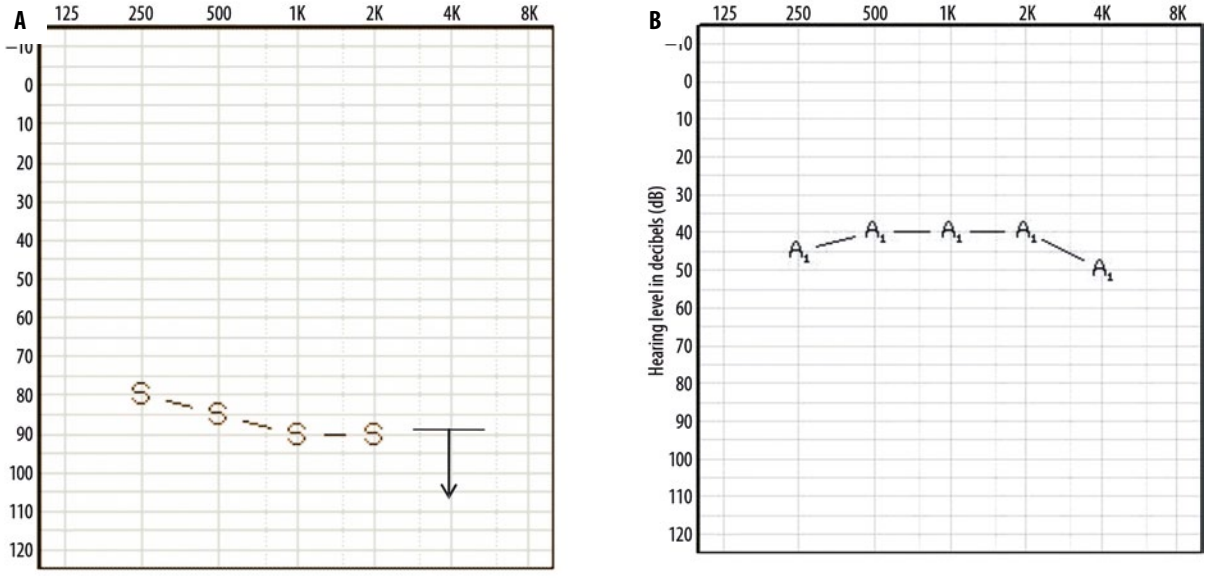

Figure 3. Observational audiometry. (**A**) Primary evaluation. (**B**) With hearing aids on.

The audiological assessment is difficult due to increased anesthetic risks, difficult intubation (previous tracheostomy), pulmonary aspiration risk. The first hearing evaluation of our patient was performed in natural sleep and the reevaluation of the left ear after fluid drainage and grommet placing was done under a short general anesthesia. Unfortunately for the child, due to its other medical conditions, a complete audiological diagnosis of severe sensorineural hearing loss on the right side and profound mixed hearing loss in the left ear was detected and managed late – at age of 4. After the hearing aid fitting the patient started to show reactions to sounds and the self-destructive behavior manifests rarely and mildly. Since he also is moderately mentally retarded, the rehabilitation is slow, but the changes in the patient's behavior are obvious and satisfactory.

Cochlear implantation can be considered if the hearing aids eventually show lack of benefit. Such decision is hard to take, considering the high surgical risks generated by the child's plural-malformative pathology. Even if the parents will accept the risks of a new surgical intervention and of possible complications (aspiration, meningitis, cardiac complication) the child's autistic-like behavior does not allow us to accurately predict the child's probability of learning a certain way of communication.

Conclusions

A child with CHARGE had several major surgeries before 3 years of age. Once the survival of the child is assured, parents shift their focus to development, communication, and learning.

Early evaluation of hearing sensitivity and, when appropriate, provision of conventional or nonconventional hearing aids are important components in the process of auditory habilitation and quality of life improvement for individuals with this complex and multifaceted disorder.

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