COCHLEAR NERVE APLASIA: THE AUDIOLOGIC PERSPECTIVE

A CASE REPORT

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Congenital absence or underdevelopment of the cochlear nerve has been described in association with congenital hearing loss.

Description of cochlear nerve integrity may be important for diagnosis and management of sensorineural hearing loss.
1 - INTRODUCTION

- absence (aplasia) of the cochlear nerve constitutes a contraindication to amplification and/or cochlear implantation

- it is still not very clear what circumstances may justify or not justify the audiological /otological intervention

- what are the elements of an audiological/otological assessment in case of aplasia/hypoplasia of the cochlear nerve?
electrophysiology indicating profound hearing loss

imaging suggestive of auditory nerve absence

Behavioural audiometry revealing useful residual hearing
A 9 months old girl, with reported profound hearing loss and facial malformation, was referred for cochlear implant selection.

CLINICAL FEATURES:

marked facial asymmetry with right facial underdevelopment
(including right anophtalmia with hypoplastic orbit; abnormally shaped right pinna; right external auditory canal stenosis; right coanal atresia; right mandibular hypoplasia)

delayed linguistic and motor milestones
auditory brainstem response (ABR) with click stimulus:
bilaterally absent response at max intensity level (110 dB nHL)

otoacoustic emissions (OAEs):
TOAEs and DPOAEs bilaterally absent

Acoustic immittance assessment
left tympanometry type “A” with
absent acoustic reflexes

transtympanic electrocochleography (ECochG):
Compound action potential (AP)
bilaterally not identified (at 125 dB SPL)

parental report and observational audiometry:
no reaction to high intensity sounds
Internal auditory canal not present on right (underdeveloped) side

narrow internal auditory canal (yellow arrow) on left side

left side HRTC, axial, thk 0.6 mm
No fluid in the internal auditory canal on the right side (but a present cochlea); narrow internal auditory canal on the left side, with a single nerve (facial nerve)
• hemifacial microsomia (Goldenhar syndrome)

• profound hearing loss

• Bilateral absence of the cochlear nerve

• proposal for sign language habilitation

This non random association of anomalies (also known as oculo-auriculo-vertebral spectrum) represents abnormal morphogenesis of the 1. and 2. branchial arches. While there is no agreement on the minimal diagnostic criteria, the facial phenotype is characteristic when enough manifestation are present.
The little girl was seen for a further audiological control at 12 months of age. The parents reported some changes in her auditory behaviour:

- stops moving because of a sudden noise (a slamming door, someone Screaming, a barking dog)
- awakes due to a sudden loud noise.
- is attracted by loud sounds
- stops crying when mom calls
- loud familial sounds evoke a predictable reaction (sound of a spoon on a plate when it is time to be fed, clap hands etc)
free field V.R.A.
(visual reinforcement audiometry)
at 12 months
The little girl was fitted with BTE hearing aids

speech detection threshold: 45 dB SPL

Free field aided threshold
Good awareness of the sound

Consistency of turn taking involving voice

Good shared attention

Good use of eye contact with the speaker

Vowel discrimination, initial word discrimination and identification

Speech production: vowels, some consonants
1) lack of correlation between radiology, objective audiometry and hearing function in this case

2) cochlear aplasia is a radiological entity

3) congenital narrow internal auditory canal and eighth nerve hypoplasia/aplasia can be expression of different diseases
1) Hemifacial microsomia is generally sporadic, clinically heterogeneous disorder, with probably heterogeneous causes.

2) There is a wide variety of external and middle ear malformations that have been described in association with the syndrome (result from abnormal development of structures derived from the 1. or the 1. and 2. branchial arches)

3) Inner ear malformations are rare, and have not been generally considered to be a feature of the syndrome.

4) Pathogenetically, abnormally shaped, malpositioned internal auditory canals may be considered to be a manifestation of abnormal cranial base development rather than the consequence of a primitive nerve absence.
1) clinical features of cochleovestibular nerve aplasia/hypoplasia/dysplasia need to be investigated

2) there is a lack of clear correlation between imaging, clinical findings and hearing function evaluations in some cases.

3) always seek for protocols that allow the correlation between clinical, pathogenetic and radiological findings for a complete diagnosis, correct and safe treatment planning.
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