Audiological features and anatomic abnormalities in rare pediatric syndromes

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During the last 3 years in our department were studied 7 patients with rare syndromes and hearing loss. We describe the anatomic abnormalities and audiological features of 3 cases observed.

Goldenhar syndrome is a multiple congenital syndrome characterized by a variable combination of multiple anomalies: coloboma; choanal atresia/stenosis; cranial nerve dysfunction; characteristic ear anomalies. A case of 1-year-old (F) patient affected by Goldenhar syndrome referred to our clinic. ABR test showed profound bilateral sensorineural hearing loss. No responses to the ECoChG bilaterally were found. The CT scan showed a sovramastoid course of the right facial canal (fig 1a). MRI scans showed that bilaterally, no VIII nerves were present in the cerebellopontine angle (fig 1b). Cochlea and SSC have normal morphology. The patient was candidate for monolateral ABI implantation.

A 4-year-old female affected by VATER syndrome and CHIARI I malformation referred to our clinic. The E-ABR showed profound bilateral sensorineural hearing loss. The CT scan and MRI (fig.2) showed a cystic cochlea to the right side (common cavity) and the lateral semicircular canal is absent from both sides. The internal auditory canals were narrow bilaterally. Despite the normal morphology of the left cochlea, the ECoChG showed no responses bilaterally. The patient was selected for ABI implantation on the right side.

CHARGE syndrome is a rare condition characterized by Coloboma, Heart anomaly, choanal Atresia, Retardation of growth and GENital abnormalities. We studied one patient (1Y, M) with CHARGE syndrome. At the CT scan and MRI he presented a fusion of the middle and apical turns of the cochlea (Mondini abnormality – fig.3), dysplastic vestibular system, and probable aplasia of the cochlear nerves bilaterally. ECoChG and E-ABR showed no response to the maximum output (fig.4). The patient underwent surgical intervention with monolateral ABI.

Pediatric syndromic HL is always associated with other medical conditions, and the degree of severity depends on several factors. In these children, hearing results are very much lower than in non-syndromic patients with only hearing loss, due to the serious associated comorbidities; a systematic and multidisciplinary approach is mandatory for proper management of such cases, and hearing rehabilitation represents one of the major goals for these patients.