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CLINICAL GENETICS

Bronchio-oto-renal syndrome – a case report

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Abstract

The **branchio-oto-renal syndrome** (BOR syndrome) is characterized by congenital defects of the outer, middle and inner ear, fistulas and cysts in the neck, and abnormalities in the structure and function of the kidneys. Patients are often accompanied by hearing loss.

Major Criteria	Minor Criteria
• Second branchial arch anomalies • Deafness • Preauricular pits • Auricular malformation • Renal anomalies	• External auditory canal anomalies • Middle ear anomalies • Inner ear anomalies • Preauricular tags • Other: facial asymmetry, palate abnormalities

Table 1. Diagnosis criteria in BOR syndrome* by Chang et al.

* At least three major criteria must be met to make a diagnosis; two larger and at least two smaller or one larger criterion and having a family member with BOR syndrome in the first degree of kinship

Rapport de Cas

- A 7-year-old female patient diagnosed with BOR syndrome came to the IFPS due to bilateral hearing loss observed from birth.

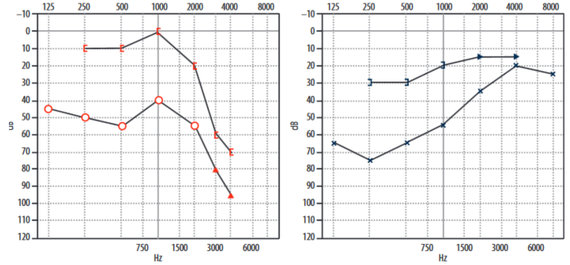

Figure 1. The patient's pure tone audiometry test results. Red: right ear; blue: left ear.

- The CT and physical examination revealed features characteristic of the brachio-otorenal syndrome, as well as effusion in the right ear and perforation of the eardrum of the left ear. Additional examinations showed mixed bilateral hearing loss and a defect of the inner and middle ear.

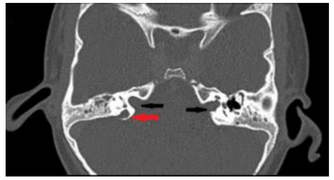

Figure 2. Computed tomography of the temporal bones; the red arrow marks the enlarged of the vestibular aqueduct; the inner ear canals are marked with black arrows.

- The patient was qualified for bone conductive device implantation surgery. Eventually parents decided to fit conventional hearing aids.

Résultats

Patients with BOR syndrome should be under the care of otolaryngologists, audiologists, and nephrologists from early childhood. Management is highly individualized and depends on the manifestation of disorders and the severity of symptoms. Preauricular fistulas require monitoring, and in cases of recurrent inflammation, surgical removal may be necessary. Malformations of the auricle can be corrected surgically, especially in cases of microtia. Patients may also require the creation or widening of the external auditory canal or reconstructive surgery to improve hearing.

Accurate diagnosis of hearing loss and early initiation of treatment are crucial. The type of hearing loss will determine the course of action.

Patients for whom hearing improvement through tympanoplasty is not feasible should consider using hearing aids. If the results with hearing aids are unsatisfactory, cochlear implant implantation should be considered. In cases of severe hearing loss, cochlear implants may be necessary.

Conclusion

BOR syndrome is one of the most common causes of inherited autosomal dominant hearing loss in children. Patients with this disease require multi-specialist care, especially otorhinolaryngological, audiological and nephrological care. Treatment in BOR syndrome depends on the type of developmental disorders

Références

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