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# Management of Congenital Deafness of the Syndrome Branchio-Oto-Renal (BOR): A Case Report

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**Abstract** **Case Report**

***Objective:*** Describe the different therapeutic modalities of Branchio-oto-renal (BOR) Syndrome. ***Material and method:*** We report the case of a 7-year-old boy who presented with a Branchio-oto-renal syndrome (BOR). ***Clinical Case:*** T.S, a 7-year-old child presented from the age of 3 years, a progressive, bilateral deafness, with a preserved language. The otological examination found implanted low ears, bilateral prehelic fistulas, and left seromucosal otitis CT scan showed bilateral vestibulo-labyrinthine malformation confirmed by the MRI. Cochlear implant surgery on the right ear was performed. ***Conclusion:*** The therapeutic principle is based on the precociousness of the rehabilitation of a quality auditory canal, which remains a factor of good prognosis of bilateral deafness of the child reached of the syndrome of BOR. A quality ear canal is defined by an ability to perceive and understand speech by the ear canal alone. Severe or deep bilateral deafness is the subject of hearing rehabilitation by hearing aid or cochlear implant, speech therapy and educational support.

**Keywords:** Branchio-oto-renal (BOR) Syndrome, congenital deafness, hearing defects, cochlear implant.



## INTRODUCTON

Branchio-oto-renal syndrome (BOR) combines deafness, multiple gill fistulas and renal malformation. Its prevalence is estimated at 1 / 40,000. Renal malformations can be major (agenesis or major hypoplasia) and sometimes lead to termination of pregnancy. Lesser deformities will be diagnosed by a renal ultrasound which must be requested in the face of deafness suggestive of BOR: deafness is accompanied by malformations of the outer ear (poorly hemmed ears, ear aplasia, enchondromas, stenosis of the ear canals), the middle ear (there is a transmissive component to the audiogram) and the inner ear (various cochleovestibular malformations). We generally find bilateral preheliceal fistulas and fistulas of the second branchial cleft with associated cartilage residues suggestive. In practice, in the event of perceptual or mixed deafness associated with a gill fistula or malformations of the outer ear, it is desirable to do a renal ultrasound [1-19].

Three genes have been located and two identified, EYA1 and SIX1. The EYA1 gene can also be responsible for a branchio-otological syndrome, very close to BOR but without kidney damage [20-25].

## CASE REPORT

This is T.S, a 7-year-old child (Figure-1), 4th in a family of four. Without notion of neonatal suffering, hospitalization in intensive care, or taking ototoxic drugs in his history. No inbreeding among parents. The mother, a brother, an older sister, 3 maternal uncles, two maternal cousins, present more or less the same symptomatology as the child.

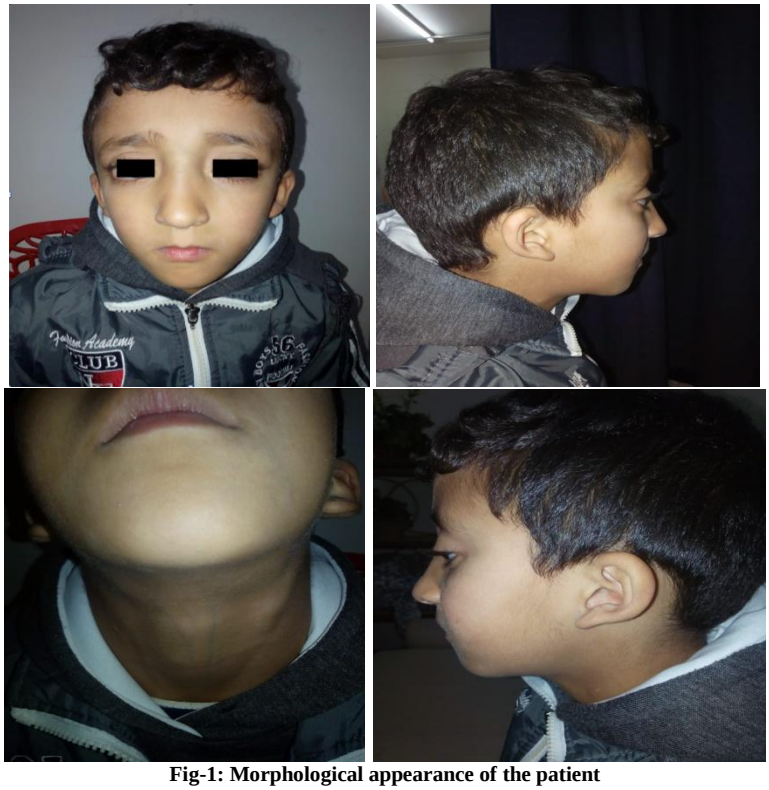

**Fig-1: Morphological appearance of the patient**

The patient presented from the age of 3 years, a progressive, bilateral deafness, with a preserved language. The otological examination found implanted low ears, bilateral prehelic fistulas, and left seromucosal otitis, the child reacts to the otoscopic aspiration. The cervical examination found a second bilateral cleft fistula. Audiological explorations have shown on the auditory evoked potential a bilateral cophosis (Figure-2).

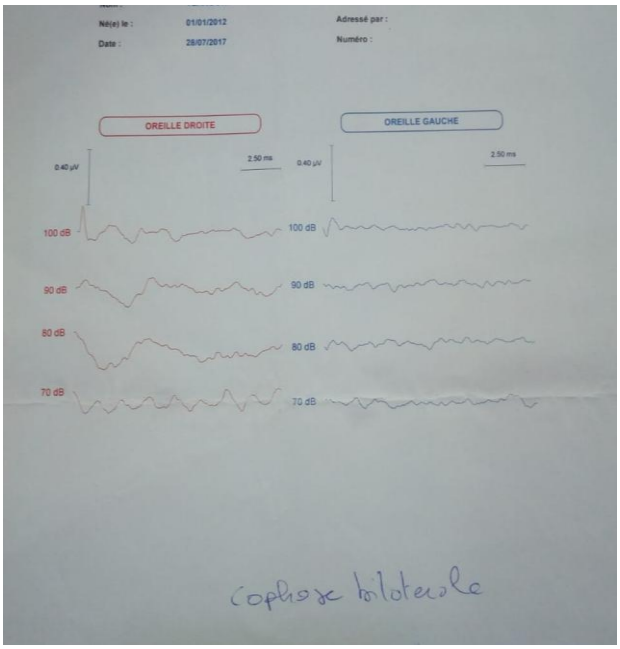


**Fig-2: Evoked auditory potential showing bilateral cophosis in children**

CT scan of of the petrous bones showed bilateral chronic otitis media with mastoiditis, and bilateral vestibulo-labyrinthine malformation in the form of bilateral vestibular ectasia, dilation of the median part of the canals, circular seedlings on both sides, with dilation of the aqueduct of the right vestibule (Figure-3). Kidney ultrasound is normal. No explorations done to family members.

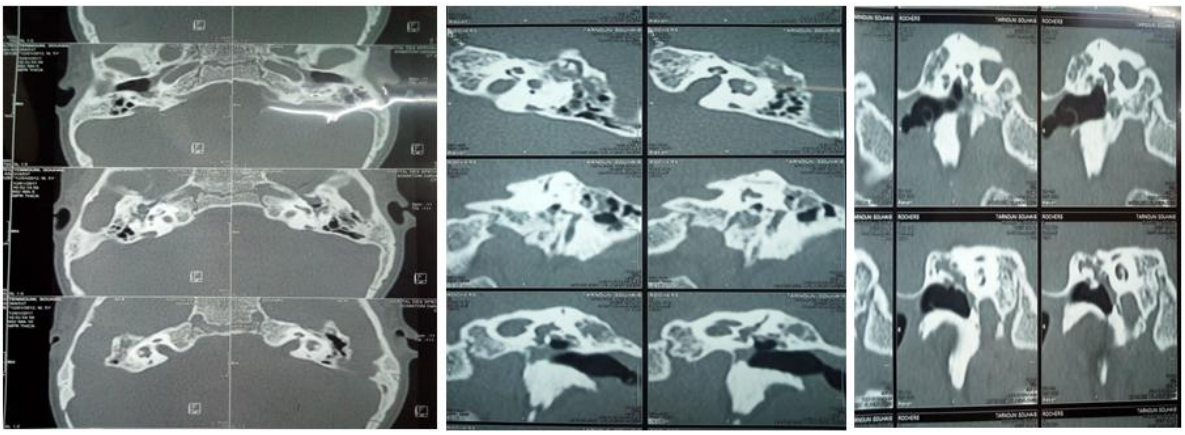

**Fig-3: CT images with gadolinium injection in axial sections showing the different malformations of the inner ear on both sides**

The MRI performed confirmed the malformations visible on the scanner, but these patients did not contraindicate the performance of cochlear implant surgery on the right ear (Figure-4).

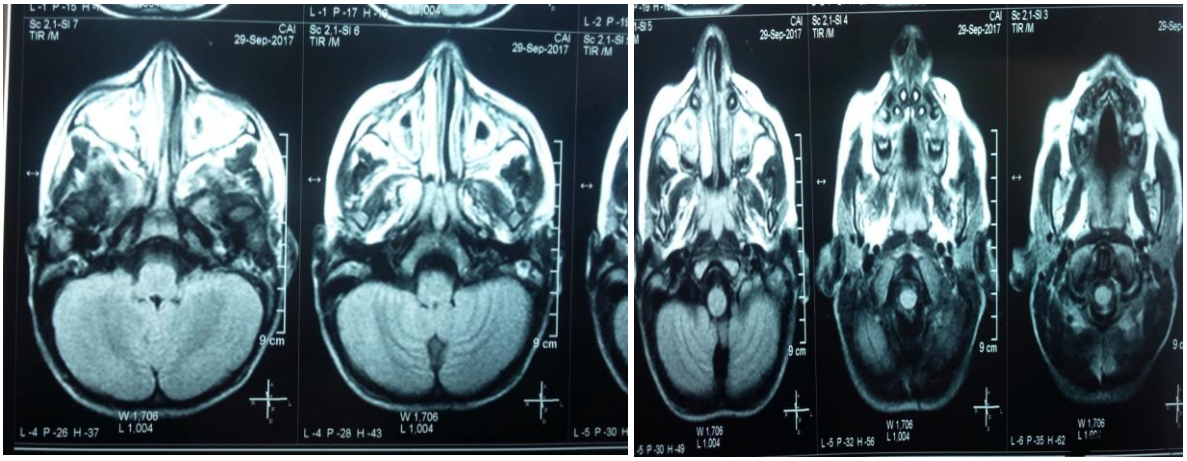

**Fig-4: MRI images in axial sections with gadolonium injection in T2 sequence showing the different site malformations**

## DISCUSSION

The branchio-oto-renal syndrome (BOR syndrome) has a complete penetration (any carrier of an anomaly in one of the genes involved presents clinical signs; no "generation jump"). Hearing loss is the most constant of the anomalies and affects 90% of patients. It can be transmission (30%), neurosensory (20%) or mixed in 50% of cases. Deafness is severe in a third of patients and progressive in a quarter [26-36].

The treatment of BOR syndrome deafness, like all congenital deafness, imperatively concerns two aspects: on the one hand, on hearing loss itself, on the other hand on its consequences on learning. Early rehabilitation is essential, therefore, rehabilitation must be organized as soon as reliable and consistent arguments are obtained for permanent deafness: Early acoustic evoked potentials (PEAP) or Auditory Steady State Response (ASSR), and behavioral audiometry [37-49].

Hearing rehabilitation must start from the first months of life in cases where the deafness is severe or profound bilaterally. Speech therapy is essential. Indeed, in most cases, compensation for hearing loss is not enough to make up for the delay in language development or learning essential for good schooling [50-56].

In infants and children, only hearing aid hearing aids are used, due to the small size and continuous growth of the external ear canal, which in most cases is malformed. In-ear devices are reserved for the adolescent, in case of refusal of another solution [57-62].

When deafness is progressive, having initially allowed access to oral language, the cochlear implant must be proposed as soon as speech perception without lip reading becomes less than 50% at 65 dB with two hearing aids [63-65].

Cochlear implantation remains a real surgical challenge given the different anatomical malformations found in this syndrome. Bilateral implantation is recommended because it allows better localization of sounds and better perception in noisy environments than unilateral implantation [66, 67].

## CONCLUSION

The significant advances in early hearing screening, imaging, and cochlear implantation have dramatically changed the future of children with BOR syndrome, allowing for better consideration of the impact of conductive hearing loss.

Once the diagnosis has been established and rehabilitation has started, the frequency of associated disorders and the potential development of deafness justify careful monitoring throughout schooling and multidisciplinary care in many cases.

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