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CHILDREN'S AUTORY IMPLANTS

A case report of Riboflavin Treatment and Cochlear Implants in a 4-year-old girl with progressive hearing loss and delayed speech development: Brown-Vialetto-Van Laere Syndrome

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Abstract

Brown-Vialetto-Van Laere (BVVL) syndrome is a rare autosomal recessive disorder caused by mutations in intestinal riboflavin transporter genes, resulting in a motor neuron disorder. The motor nerve dysfunction is caused by the riboflavin tran RFVT2 deficiency due to abnormal myelin permeability, which can be associated with sensorineural deafness in childchood. Impaired permeability explains postsynaptic nature of the auditory neuropathy underlying the hearing loss in BVVL syndrome.

The aim of this study was to present a case of Brown-Vialetto-Van Laere syndrome and to describe the method of treating the deep sensorineural hearing loss associated with this syndrome using cochlear implants.

Rapport de Cas

- The case report concerns a girl from Poland who, at the age of 2 years, developed progressive **atypical neurological symptoms of unknown etiology**: ataxia of the upper and lower limbs, gait abnormalities, generalized muscle weakness, visual and hearing problems, and regression of speech development.
- A karyotype study (whole-exome sequencing) revealed alterations within SLC52A2, leading to the **diagnosis of Brown-Vialetto-Van Laere syndrome** and initiation of high-dose riboflavin treatment.
- As a 4-year-old child, she presented to the Institute of Physiology and Pathology of Hearing – World Hearing Center in Poland with **progressive hearing loss and speech regression**.
- Hearing tests revealed **bilateral profound sensorineural hearing loss with auditory neuropathy**.
- Surgical treatment was applied in the form of **bilateral cochlear implantation.** The procedures were performed using **Skarżyński's minimally invasive 6-step procedure**. No complications were reported

Résultats

Audiograms measured 5 months after implantation in the right ear and 4 months in the left ear are shown in Figures 1 and 2. Tests were performed using behavioral observation audiometry (BOA) and visual reinforcement audiometry (VRA). The mean postoperative free-field thresholds determined by BOA with the speech processor on in the right ear (left ear open) were 74 dB and 63 dB for the left ear (right ear open). The parents have noted steady progress in their child's auditory development.

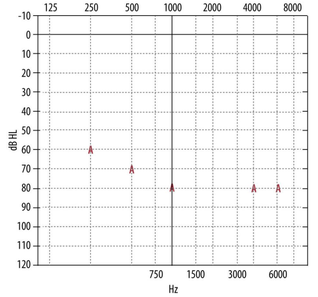

Figure 1. Free-field thresholds determined by BOA 5 months after cochlear implantation in the right ear (left ear open).

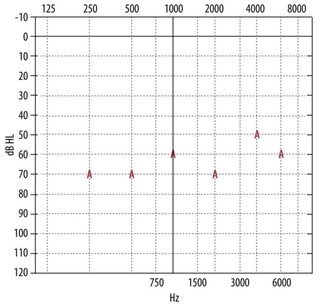

Figure 2. Free-field thresholds determined by BOA 4 months after cochlear implantation in the left ear (right ear open).

Conclusion

This report shows the importance of genetic testing in infants who present with atypical symptoms or signs. In this case, the diagnosis of Brown-Vialetto-Van Laere syndrome resulted in timely correction of the genetic riboflavin (vitamin B2) deficiency and improved hearing following the use of cochlear implants.

Références

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