AUDIOLOGICAL EVALUATION IN INFANT PATIENTS WITH BIOTINIDASE DEFICIENCY, UNDER EARLY TREATMENT

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INTRODUCTION

Biotinidase deficiency (BD) is an autosomal recessive disease that occurs due to failure in recycle of vitamin B complex[1]. Vitamin B deficiency, if not treated early, can lead to important systemic changes, including hearing loss[2]. Audiological assessments are essential for the child's individual performance to be minimally affected in this way.

OBJECTIVE

Assess the hearing of children up to 24 months of age diagnosed with BD.

METHODS

Cross-sectional observational study with a non-probability sample consisted of 9 children affected by the disease, screened and treated by the Neonatal Screening Program of the state of Minas Gerais. Meatoscopy, Transient Evoked Otoacoustic Emissions (TEOAE), and behavioral audiometry were conducted. The researcht was approved by the Ethics Committee under number 19595319.7.0000.5149.

RESULTS

The sample consisted of 9 children with mean age 13.9 months old. The mean age at diagnosis and beggining of treatment was 1 month, ranging from 10 days to 2 months. Of the 9 participants, only 1 of them had BD in its deep form, the others had BD in the partial form. The average age of treatment onset was 1 month, since when the diagnosis is made, the treatment starts immediately. The average age of audiological evaluations was 16.3 months for male children and 6 months for female children. All were submitted to the audiological evaluation (18 ears), and 100% of them presented TEOAE. When it comes to the assessment of auditory behavior, all 9 children presented cochlear-eyelid reflexes, and a response the bahavioral audiometry was appropriate for each age.

Table 1. Comparison of frequency bands between the participants' right and left ears (N=9).

Ear	1000 Hz	1410 Hz	2000 Hz	2830 Hz	4000 Hz
Right	11,76	13,21	14,59	15,48	14,20
Left	16,01	16,61	13,97	16,39	11,38
p-value	0,929	0,220	0,260	0,164	0,233

DISCUSSION

Neonatal screening is essential for early diagnosis of BD as well as for adequate supplementation, in order to minimize or erradicate symptoms[3]. Despite presenting as a rare metabolic disorder, the incidence of the disease is quite significant and changes caused are very severe if left untreated. The data show that children with BD who are undergoing treatment are less likely to have hearing loss[4].

CONCLUSION

Children diagnosed with biotinidase deficiency who are being treated with biotin did not present hearing loss.

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