Cochlear nerve aplasia in a child with normal neonatal hearing screening test

Clinical Case

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Abstract

Otoacoustic emissions (OAE) are sounds produced by a healthy cochlea and are used as a neonatal hearing screening test with good sensitivity and specificity, differentiating the ears with normal hearing from those with hearing loss. However, this test does not exclude a neural hearing loss.
The authors present a case of a 3-year-old male referred to an otorhinolaryngology consultation for recurrent otitis media. In the study carried out with complementary diagnostic tests, unilateral hearing loss of the left ear was detected, which motivated the study with magnetic resonance imaging, which in turn revealed aplasia of the left cochlear nerve.
The aplasia or hypoplasia of the cochlear nerve is an extremely rare congenital malformation, and may not be detected in neonatal hearing screening.

Keywords: cochlear nerve, hearing loss, otoacoustic emissions, cochlear implants, auditory brain stem implants

Introduction

Otoacoustic emissions (OAEs) are sounds emitted by the external ciliated cells of healthy cochlea and are used as a test in universal newborn hearing screening (UNHS).[1] OAEs have good sensitivity and specificity for this screening1 in children, without risk factors for hypoacusis, thereby allowing distinction between normal-hearing ears and ears with hypoacusis.

However, the presence of OAEs in UNHS does not necessarily mean normal hearing. Cochlear nerve aplasia or hypoplasia (CNAH) is an extremely rare congenital malformation. Considering the total population of newborns, the proportion of children with significant permanent hypoacusis is 1.2–1.7 for every 1,000 live births, with the main cause being loss of cochlear function.2 Between 20% and 30% of these children have profound deafness.[2] Among children with profound deafness, CNAH accounts for only 0.8%–1.8% of cases.[3] CNAH

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Article received on February 28, 2022.
Accepted for publication on April 3, 2022.

appears to affect both sexes equally[4,5] and is often associated with other malformations of the internal ear[4-6] ENREF_1, with the literature indicating that the proportion of latter cases lies between 40% and 85%.[4] Association with other syndromic or non-syndromic comorbidities is common.[4-6] The best imaging exam for the definitive diagnosis of CNAH is magnetic resonance imaging (MRI), which is superior to computed tomography in the evaluation of nerve structures.[4]

The authors present the clinical case of a child with apparent normal hearing according to newborn hearing screening who subsequently revealed severe/profound deafness due to malformation of the auditory pathways.

Case Description

The authors present the clinical case of a 3-year-old boy who was referred to otorhinolaryngology with a history of recurrent otitis media.

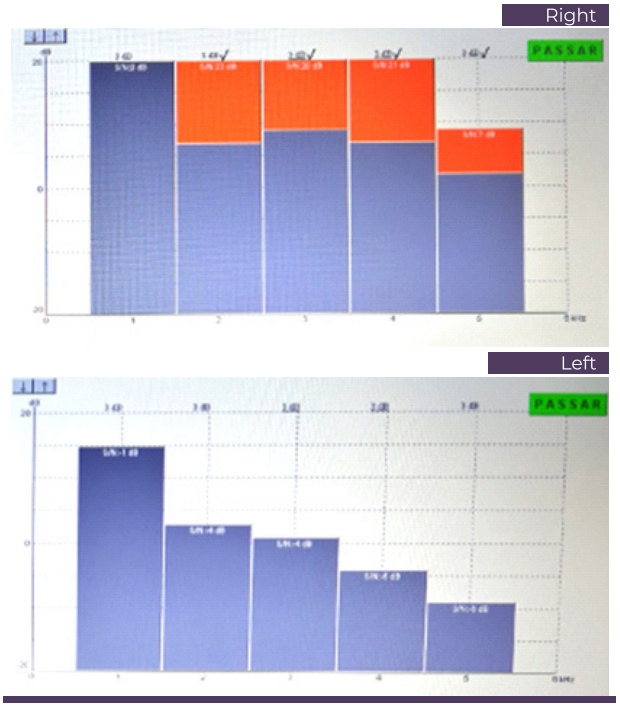
The patient had experienced three episodes of acute otitis media in the previous year, two in the right ear and one in the left ear. He had no clinical history of other episodes of recurrent infections. Further, there was no suspicion of hypoacusis, and the child exhibited age-appropriate speech development, as well as age-appropriate height and weight and psychomotor development. The prenatal period had been monitored, including screening for maternal infections, as prescribed in Portugal for low-risk pregnancies,[7] the result of which was negative. The newborn period was without complications. Newborn hearing screening was performed at 3 days of life via OAE testing, and a negative result ("pass") was obtained in both ears (Figure 1).

The child did not have a family history of hypoacusis or of other medical or surgical conditions.

The otorhinolaryngological objective examination was normal.

Impedance audiometry was performed. The tympanogram showed a type-A curve in the Jerger classification, bilaterally. Moreover, the tone audiogram showed normal hearing in the right ear and cophosis in the left ear, as well as absence of stapedial reflexes only on the left. In view of this clinical presentation, MRI of the ears was performed (Figure 2), which showed absence of the cochlear nerve on the left, without other internal ear anomalies.

Figure 1
Recovery of the hearing screening exam performed in the newborn period using otoacoustic emissions. Right ear (above); left ear (below). The result was a bilateral "pass."

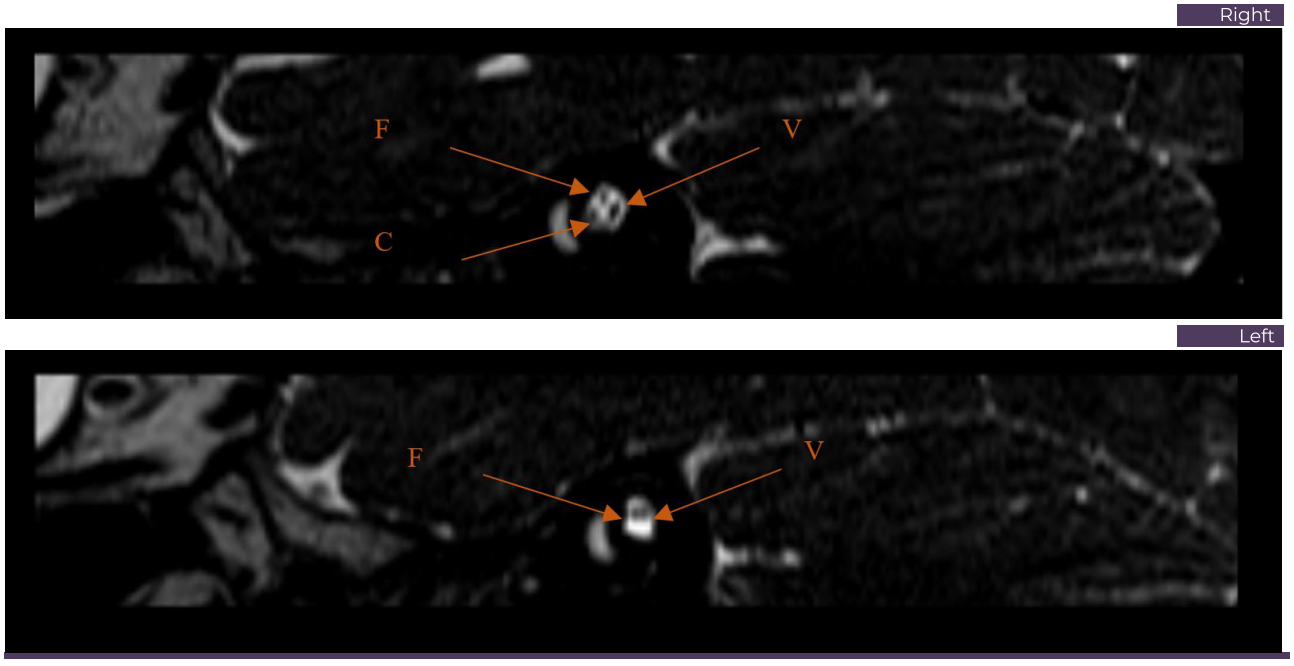


Discussion

CNAH is an extremely rare congenital malformation. The best audiology exam for detection of these cases is the auditory evoked potentials (AEP) test, as opposed to OAE testing, which can be normal in these cases.5 However, AEP testing is only used at birth when there are risk factors for hypoacusis, which was not the case in the exemplar instance. Nevertheless, CNAH is such a rare condition that it does not diminish the sensitivity of OAE as a universal screening exam.

Cases of aplasia or hypoplasia of the cochlear nerve are characterized by the presence of OAEs and the absence of response or anomalies in the AEP test[4-6,8] ENREF_3, as in auditory neuropathy spectrum disorder,[4,5]

Figure 2
Magnetic resonance imaging of the ears, oblique sagittal plane for the visualization of the internal acoustic meatuses, showing absence of the cochlear nerve on the left. F – facial nerve, V – vestibular nerve, C – cochlear nerve



in which the cochlea's external ciliated cells function normally, but there is a dysfunction of the internal ciliated cells or of the nerve fibers of the cochlear nerve[6,9] ENREF_1. Given that CNAH and auditory neuropathy spectrum disorders have similar features in audiology tests, an imaging exam is essential to distinguish these diseases, and MRI is the exam of choice.[5]

Cochlear implants (CIs) are implanted devices that directly stimulate the cochlear nerve and have revolutionized the treatment of profound deafness;[10] they are more effective when placed at an early age.[11] However, in the case of deafness caused by CNAH, the effect of the CI may be minimal or none.[3] In this case, an auditory brainstem implant (ABI) may be an alternative. An ABI is indicated for cases in which complete absence of the cochlear nerve has been shown, that is, in cases of aplasia rather than hypoplasia.[3] However, an ABI requires a higher-risk surgery, and although it yields better results than a CI, the outcome is very variable[3,12] ENREF_7. On the other hand, a response to a CI is possible in cases of hypoplasia, albeit variable.[3] Based on these principles, most authors advocate that if there is any evidence of a potential response to a CI, such as the presence of cochlear nerve hypoplasia rather than aplasia,[3] such patients can receive a CI and eventually (after reevaluation of the response) a subsequent ABI.[3,13]

In this clinical case of unilateral hypoacusis detected at the early age of 3 years in a child with age-appropriate speech development, the risks of an ABI would outweigh its benefits. In sum, the child did not have any hypoacusis risk factor at birth that warranted hearing screening via AEP rather than OAE testing. The strategy in this case consisted of monitoring the child in the clinic given his unilateral hearing loss and recurrent otitis.

Conclusion

The presence of OAEs in newborn hearing screening does not necessarily mean that the child has normal hearing. The aim of this report was to raise awareness of the existence of false negative results of universal newborn hearing screening.

Conflicts of Interest

The authors declare that there is no conflict of interests regarding the publication of this paper.

Data Confidentiality

The authors declare having followed the protocols in use at their working center regarding patients' data publication.

Protection of humans and animals

The authors declare that the procedures were followed according to the regulations established by the Clinical Research and Ethics Committee and to the 2013 Helsinki Declaration of the World Medical Association.

Funding Sources

This work did not receive any contribution, funding or scholarship.

Availability of scientific data

There are no datasets available, publicly related to this work.

Bibliographic references

1.Grupo de Rastreio e Intervenção da Surdez Infantil (GRISI). Recomendações para o rastreio auditivo neonatal niversal (RANU). Acta Pediátrica Portuguesa [Internet]. 2007; 38(5): 209-14. Available from: https://doi.org/10.25754/pjp.2007.4698.

2.Kral A, O'Donoghue GM. Profound deafness in childhood. N Engl J Med. 2010 Oct 7;363(15):1438-50. doi: 10.1056/NEJMra0911225.

3.Freeman SR, Sennaroglu L. Management of cochlear nerve hypoplasia and aplasia. Adv Otorhinolaryngol. 2018;81:81-92. doi: 10.1159/000485542.

4.Levi J, Ames J, Bacik K, Drake C, Morlet T, O'Reilly RC. Clinical characteristics of children with cochlear nerve dysplasias. Laryngoscope. 2013 Mar;123(3):752-6. doi: 10.1002/lary.23636.

5.Cinar BC, Tahir E, Batuk MO, Yarali M, Sennaroglu G, Sennaroglu L. Cochlear nerve hypoplasia: audiological characteristics in children and adults. Audiol Neurootol. 2019;24(3):147-153. doi: 10.1159/000500938.

6.Buchman CA, Roush PA, Teagle HF, Brown CJ, Zdanski CJ, Grose JH. Auditory neuropathy characteristics in children with cochlear nerve deficiency. Ear Hear. 2006 Aug;27(4):399-408. doi: 10.1097/01.aud.0000224100.30525.ab.

7.George Francisco. Exames laboratoriais na Gravidez de Baixo Risco. Norma 37/2011 de 30/09/2011 atualizada a 20/12/2013. Direção Geral da Saúde. Disponível em https://www.dgs.pt/directrizes-da-dgs/normas-e-circulares-normativas/norma-n-0372011-de-30092011-jpg.aspx

8.Liu C, Bu X, Wu F, Xing G. Unilateral auditory neuropathy caused by cochlear nerve deficiency. Int J Otolaryngol. 2012;2012:914986. doi: 10.1155/2012/914986.

9.Carvalho GM, Leão BP, Ramos PZ, Guimarães AC, Castilho AM, Sartorato EL. [Auditory neuropathy: clinical evaluation and diagnostic approach]. Acta Med Port. 2016 Jun;29(6):353-359. doi: 10.20344/amp.6942.

10.Naples JG, Ruckenstein MJ. Cochlear implant. Otolaryngol Clin North Am. 2020 Feb;53(1):87-102. doi: 10.1016/j.otc.2019.09.004.

11.Bouquillon E, Le Gac MS, Godey B. [Cochlear implant in children]. Rev Prat. 2018 Oct;68(8):870-873.

12.Colletti L, Shannon RV, Colletti V. The development of auditory perception in children after auditory brainstem implantation. Audiol Neurootol. 2014;19(6):386-94. doi: 10.1159/000363684.

13.Vesseur A, Free R, Snels C, Dekker F, Mylanus E, Verbist B. et al. Hearing restoration in cochlear nerve deficiency: the choice between cochlear implant or auditory brainstem implant, a Meta-analysis. Otol Neurotol. 2018 Apr;39(4):428-437. doi: 10.1097/MAO.0000000000001727.