LATE DIAGNOSIS OF ENLARGED ENDOLYMPHATIC DUCT AND SAC SYNDROME IN A CASE OF UNILATERAL SUDDEN HEARING LOSS WITH RETROCOCHLEAR PATTERN. CASE REPORT

SIMONA ŞERBAN[1], MARIAN RĂDULESCU[2]

[1,2]Institute of Phonoaudiology and ENT Functional Surgery Prof. Dr. "Dorin Hociotă" Bucureşti, "Carol Davila" University of Medicine and Pharmacy Bucureşti

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Abstract: *Enlarged Vestibular Aqueduct (EVA) is one inner ear malformation found in 35% of children with unilateral sensorineural hearing loss.(1) Case reports of late diagnosis of enlarged vestibular aqueduct in young adulthood are extremely rare. The authors present a case of unilateral sensorineural hearing loss with sudden onset and retrocochlear pattern in an 18-year-old patient. MRI examination showed an enlargement of the left endolymphatic sac, without any other anomalies of the inner ear. The controlateral ear had normal hearing and no imaging abnormalities were found. This is a very rare case of unilateral enlarged endolymphatic duct and sac syndrome (EEDSS) discovered on MRI (Magnetic Resonance Imaging) assessment in a case of sudden hearing loss in early adulthood.*

Cuvinte cheie: *sindrom de duct şi sac endolimfatic lărgit, surditate brusc instalată, leziune retrocohleară*

Rezumat: *Apeductul vestibular lărgit (AVL) reprezintă malformaţia urechii interne găsită la 35% dintre copiii cu hipoacuzie neurosenzorială unilaterală.(1) Situaţiile în care hipoacuzia debutează acut la vârsta adultă sunt extrem de rare. Autorii descriu cazul unei paciente de 18 ani, cu surditate brusc instalată în urmă cu două luni la urechea stângă. Examinarea audiologică evidenţiază hipoacuzie neurosenzorială cu caracteristici de afectare retrocohleară. Examinarea imagistică prin rezonanţă magnetică nucleară (RMN) arată lărgirea ductului şi a sacului endolimfatic de partea stângă, fără alte anomalii ale urechii interne. Urechea contralaterală este normală, atât din punct de vedere audiologic cât şi imagistic. Este un caz foarte rar de duct şi sac endolimfatic lărgit (DSEL) unilateral, care debutează clinic la vârsta adultului tânăr prin surditate brusc instalată, cu caracteristici de leziune retrocohleară.*

INTRODUCTION

Large vestibular aqueduct (LVA) or enlarged vestibular aqueduct (EVA) is a congenital anatomical defect, in which the mid vestibular aqueduct is larger than 1 mm in diameter.(2) Hearing loss or balance symptoms associated with an EVA can occur when the endolymphatic duct and sac expand to fill the larger space. When EVA is associated with such symptoms, it is referred to as enlarged vestibular aqueduct syndrome (EVAS). The anomaly can be diagnosed by imaging, such as high resolution CT which accurately measures the diameter of the aqueduct and / or by MRI which highlights the dilated endolymphatic duct and sac.(3) It is often associated with other congenital inner ear anomalies, the most common being an abnormally large vestibule, an enlarged semicircular canals, or a hypoplastic cochlea. EVAS is the only radiographically detectable inner ear anomaly associated with sensorineural hearing loss.(4)

Enlarged vestibular aqueduct is usually diagnosed in early childhood, 12% of these patients being under the age of 4.(5) The exact mechanism which causes hearing loss in EVAS is not well defined, especially the cases of sudden hearing loss where a possible haemorrhage is the cause.(6) Patients with EVAS have residual hearing and most of them experience sudden hearing deterioration, particularly following minor trauma or infection. The rupture of Reissner's membrane(7), the vulnerability of dysmorphic cochlea to pressure changes and the reflux of hyperosmolar content into the endolymphatic sac (ES)(8) have also been proposed as causes for progressive hearing loss.

The authors present the case of an 18-year-old female patient who presented one month after experiencing sudden hearing loss. The audiological assessment was highly suggestive for a retrocochlear lesion, however, the MRI found an enlarged endolymphatic sac.

CASE REPORT

18-year-old female patient was attended the audiology department for investigations regarding acute onset sensorineural hearing loss in the left ear with no history of trauma. Pure tone audiometry showed left sensorineural hearing loss with ski slope profile (figure no. 1). There were normal hearing thresholds in the right ear. Speech audiometry with bisyllabic words revealed speech detection threshold at 10 dB HL for the right ear and at 45 dB HL for the left ear. Maximum speech recognition score for the right ear was 100% and 70% for the left ear, with a rollover curve and a rollover index of 0.57 suggestive for retrocochlear lesion (figure no. 2).

Tympanometry indicated normal pressures and compliances of the middle ear bilaterally. Acoustic reflex was absent at 500 and 1000 Hz through ipsilateral stimulation but present when contralateral stimulation was used. Left ear Auditory Brainstem Response (ABR) revealed abnormal traces with prolonged I-V interval (4.57 msec) at maximum sound intensity (100 dB nHL) and an interaural difference of more

[2]Corresponding author: Marian Rădulescu, Str. Mihail Cioranu, Nr. 21, Sector 5, Bucureşti, România, E-mail: focaargintie@yahoo.com, Tel: +40788 414497

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than 0.3 msec (figure no. 3). There were no receptor microphonic potential recordings.

Figure no. 1. Pure tone audiogram shows sky slope sensorineural hearing loss on the left ear

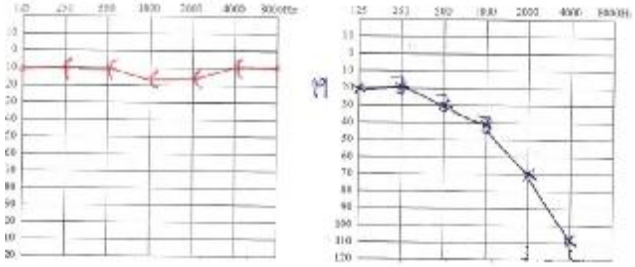


Figure no. 2. Speech audiometry with bisyllabic words with rollover curve on the left ear

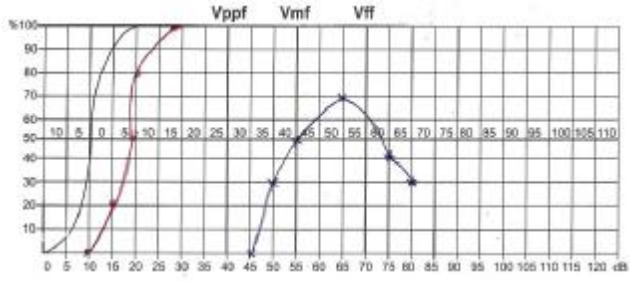
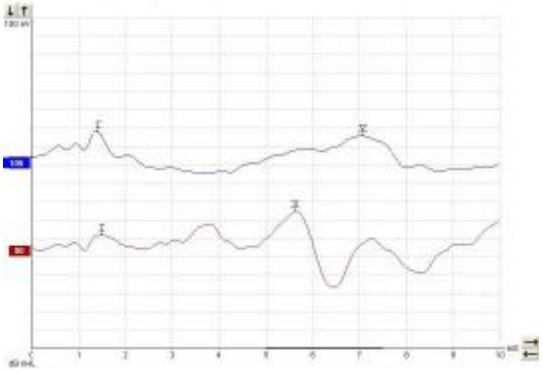
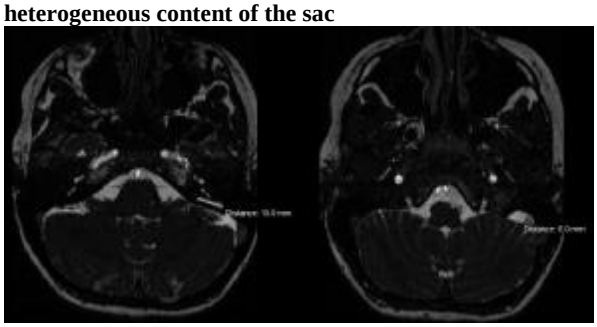


Figure no. 3. Abnormal click Auditory Brainstem Response (ABR) recordings with prolonged I-V interval on the left side



Transient otoacoustic emissions were present in the right ear but absent in the left ear. An MRI examination was requested due to high suspicions of a pathological process involving the internal acoustic meatus, pontocerebellar angle or posterior fossa.

Figure no. 4. Nonenhanced axial T_2 weighted images show dilatation of the endolymphatic sac on the left side; heterogeneous content of the sac



The MRI scan, using the Siemens Magnetom Avanto–1.5 T system did not reveal any space-occupying lesion in the left internal acoustic meatus, or in the left pontocerebellar angle, however, it showed a dilated left endo-lymphatic sac (figure no. 4) with sizes of 6/15mm. There were no other morphological anomalies of the cochlea or semicircular canals bilaterally. Axial T2 weighted MRI revealed a heterogeneous aspect with low intensity signal area in the endolymphatic sac. Clinical vestibular examination was normal and there were no laboratory results for thyroid pathology. The patient's family declined a CT–scan.

DISCUSSIONS

The vestibular aqueduct originates from the medial wall of the inner ear vestibule and extends its orifice to the cerebellar part of the petrous pyramid. The first description of EVAS was attributed to Valvassori and Clemis.(3) EVAS has many causes, not all of which are fully understood. The most well-known cause of EVA and hearing loss is the mutations of the PDS gene on chromosome 7.

EVAS is usually diagnosed in young children with known bilateral sensorineural hearing loss and confirmed by imaging. There are cases in which this pathology is associated with other inner ear congenital anomalies or thyroid gland disorders such as Pendred syndrome. Auditory dyssincrony and EVAS is also a possible finding but it is usually discovered in early childhood.(9) The enlarged vestibular aqueduct without cochlear anomalies may cause acquired sensorineural hearing loss and appears to be a high risk for sudden deafness. In some patients with sensorineural hearing loss and enlarged endolymphatic sac (EES), the vestibular aqueduct may not appear dilated on CT–scan. MRI is therefore necessary for the correct diagnosis of this syndrome, which should more correctly be named enlarged endolymphatic duct and sac syndrome (EEDSS). Prominent EES may predict a poor prognosis in this syndrome.

The type of hearing loss in enlarged vestibular aqueduct syndrome is controversial. All three types of hearing loss (sensorineural, mixed and conductive) have been reported in the condition. Although some authors describe sensorineural hearing loss in the majority of patients, others believe that nearly all patients with enlarged vestibular aqueduct have an air bone gap, particularly at the lower frequencies.(10)

Cases with congenital unilateral vestibular aqueduct malformation or enlarged endolymphatic duct and sac and sudden onset of sensorineural hearing loss in young adults are very rare. The article presents a case of unilateral enlarged endolymphatic duct and sac syndrome characterised by unusually late diagnosis in early adulthood in the context of sudden unilateral sensorineural hearing loss with retrocochlear pattern.

It is not well documented how sensorineural hearing loss develops in patients with EVAS. One theory suggests that a chemical mechanism is involved, characterised by an endolymph high Ca2+ intake and secondary acidosis. Another theory states that cochlear membranes fragility to pressure changes and hyperosmolar reflux secondary to head trauma or inner ear haemorrhage is the cause.(6) In this case, the heterogeneous aspect of the sac content visible on T2 weighted images may reflect bleeding into the sac or chronic changes in fluid homeostasis due to increased fluid flow rates with hyperosmolar reflux.

The type of auditory hearing loss is purely sensorineural, without any air bone gap in the low frequencies. Interestingly, the retrocochlear pattern revealed by Auditory Brainstem Response (ABR) testing in which the mechanism

incriminated remains highly controversial. As the wave I is well defined and neither cochlear microphonics nor otoacoustic emissions were found, unilateral auditory neuropathy associated with EEDSS should be excluded. A retrocochlear sudden hearing loss due to other aetiologies than space occupying lesions, like cochlear neuritis, remains questionable. In such hypothetical cases the discovery of enlarged endolympahatic sac on MRI images is purely incidental.

We are unable to predict if the hearing loss will remain unchanged or if it will progress to deep deafness and it is always possible for the patient to develop vestibular symptoms.

Therefore, further management of this case requires audiological and vestibular monitoring of the patient.

The patient should be advised to avoid head trauma and sudden pressure changes and hazardous activities such as scuba diving, ski diving or mountain biking.

CONCLUSIONS

This is a rare and interesting case, due to the unexpected discovery of an enlarged endolymphatic sac in a young patient. We would have expected to find imaging evidence of a space occupying lesion in the internal acoustic meatus or the pontocerebellar angle, in a patient with acute sensorineural hearing loss and audiological retrocochlear findings.

From the studied literature, there is no reported case of enlarged endolymphatic sac with sudden onset sensorineural hearing loss and an underlying retrocochlear lesion.

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