■ BRIEF REPORT

Enlarged Vestibular Aqueduct Syndrome. Report on Four Cases

Muhamad Taha,[a] Guillermo Plaza,[a] José Montojo,[a] María Urbasos,[b] and Ana Hernando[b]
[a]Servicio de Otorrinolaringología, Hospital de Fuenlabrada, Fuenlabrada, Madrid, Spain
[b]Servicio de Radiodiagnóstico, Hospital de Fuenlabrada, Fuenlabrada, Madrid, Spain

Although enlarged vestibular aqueduct syndrome (EVAS) has been reported to be one of the most common anomalies associated with sensorineural hearing loss, the mechanism of such loss is unclear, and there are no established criteria for diagnosis.
We report 4 patients with sensorineural hearing loss that were diagnosed during the last year in our department of EVAS. We review the diagnostic criteria of this syndrome.

Key words: Enlarged vestibular aqueduct syndrome. Sensorineural hearing loss. Computed tomography. Magnetic resonance imaging.

Síndrome del acueducto vestibular dilatado. A propósito de cuatro casos

Aunque el síndrome del acueducto vestibular dilatado (SAVD) ha sido considerado una de las anomalías más frecuentemente asociadas a la pérdida auditiva neurosensorial en la infancia, el mecanismo fisiopatológico que conduce a dicha pérdida aún no está claro, y no hay criterios establecidos para su diagnóstico.
Presentamos a 4 pacientes con hipoacusia neurosensorial que fueron diagnosticados de SAVD en nuestro servicio el último año. Revisamos los criterios diagnósticos de esta enfermedad.

Palabras clave: Síndrome del acueducto vestibular dilatado. Hipoacusia neurosensorial. Tomografía computarizada. Resonancia magnética.

INTRODUCTION

Enlarged vestibular aqueduct syndrome (EVAS) was first described by Valvassori et al[1] in 1978. It is a congenital malformation of the temporal bone involving both the auditory system and the vestibular systems, causing sensorineural hearing losses and vestibular alterations at a very early age in affected patients.[2-4]

The basic process causing deformity of the vestibular aqueduct is the abnormal growth of its contents: the endolymphatic sac and canal.[1,2,5] However, the physiopathology of the hearing loss is not yet clear.[6-8] Some studies have found a relationship between EVAS and other anomalies of the inner ear such as enlarged horizontal semicircular canal[9] or Mondini's dysplasia,[10,11] as well as between EVAS and syndromic hearing losses as in CHARGE syndrome,[12] Alagille syndrome,[13] von Hippel-Lindau disease,[14,15] or Pendred's syndrome.[16,17]

Correspondence: Dr. M. Taha.
Servicio de Otorrinolaringología. Hospital de Fuenlabrada.
Camino del Molino, 2. 28942 Fuenlabrada. Madrid. España.
E-mail: mtaha.hflr@salud.madrid.org

Received July 1, 2007.
Accepted for publication September 1, 2008.

With the continuous advances in imaging techniques, this entity is gaining ever greater interest in recent years.[18,19] We report on 4 patients with sensorineural hearing loss due to EVAS, diagnosed at our department over the last year.

CASE STUDIES

Case 1

A 25-year-old male, without any medical or surgical history of note, came to the clinic complaining of hearing loss in the right ear from 3 years previously. He did not present any other audiological signs or symptoms. In the tonal audiometry (TA), he had normal hearing in the left ear and mixed hearing loss in the RE, with a pure tone audiometry (PTA) threshold of 60 dB and a gap threshold of 20 dB. A speech audiometry confirmed the findings, with a speech reception threshold (SRT) of 53 dB. Hearing measurement and tympanogram were normal. Magnetic resonance imaging (MRI) showed an enlarged vestibular aqueduct in the right ear (Figure 1).

Case 2

Male, 54 years of age, complaining of bilateral hearing loss lasting for several years after wearing bilateral hearing

aids for approximately 15 years. He occasionally suffers from a clinical condition of spinning vertigo. TA revealed moderate asymmetric bilateral sensorineural hearing loss, more marked in the RE with airway PTA of 60 dB and a gap threshold of 10 dB. Speech audiometry presented an SRT of 55 dB. The tympanogram was normal. The image of the enlarged right vestibular aqueduct in an MRI scan confirmed the diagnosis (Figure 2).

Case 3

Thirty-six-year-old male consulting due to hearing loss in the RE since childhood. No other clinical symptoms were reported. TA revealed unilateral moderate-severe mixed hearing loss in the RE, with PTA of 72 dB and a gap threshold of 17 dB. Speech audiometry revealed an SRT of 65 dB. The MRI scan of an enlarged vestibular aqueduct confirmed EVAS.

Case 4

Female, 32-year-old, complaining of bouts of spinning vertigo for 4 years, with progressive hearing loss in the RE since childhood. Noteworthy personal history included sub-aortic stenosis for which she was operated on in childhood. The otoneurological examination, including videonystagmography, was normal. TA reflected a fall of 40 and 85 dB at 4 and 8 kHz, respectively. Speech audiometry presented discrimination of 100% for words at 60 dB. MRI confirmed the diagnosis of an enlarged vestibular aqueduct (Figure 3).

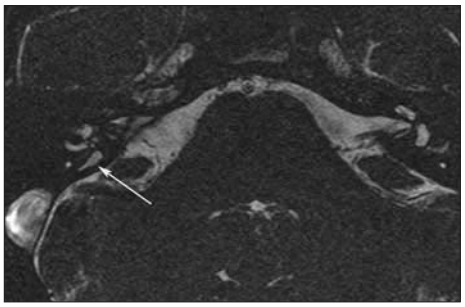

Figure 1. Ultrasound gradient sequence (FIESTA). Enlarged right endolymphatic sac, 1.8 mm, as an isolated finding.

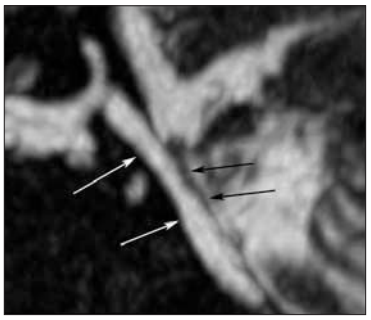

Figure 2. Reconstruction of a maximum intensity projection in axial and coronal views. Enlarged endolymphatic sac, 1.9 mm, within the bony vestibular aqueduct, separated from the cerebrospinal fluid by the dura mater of the posterior fossa, to its posterior extra-osseal extension. Normal vestibule.

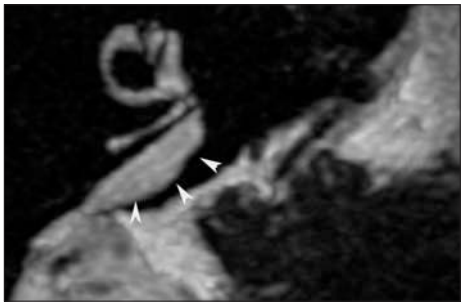

Figure 3. Reconstruction of a maximum intensity projection in oblique axial view. Enlargement of the right vestibular aqueduct to 3.1 mm of maximum diameter.

DISCUSSION

Among the congenital anomalies of the inner ear, EVAS is the one most often detected in imaging studies,[2,16] with an estimated prevalence of 1%-25% in subjects with sensorineural hearing loss, depending on the study.[4,5,18-20] The diagnosis is established by means of the association of an appropriate clinical context, the existence of unilateral or bilateral sensorineural hearing loss and the viewing of the characteristic signs in the corresponding imaging methods, computed tomography (CT) and MRI, among which the latter is the technique that provides the most pertinent data for its diagnosis.[21]

Age at presentation of this clinical picture is variable.[1,18] Albeit more frequent in women, there is not usually any prior family history,[3,6] although some authors have found first or second generation prior cases in 1 out of 3 patients.[22] Nonetheless, recent studies have been unable to prove any clear correlation between the anatomical malformation and genetic mutations or alterations.[23] In fact, some publications have reported on patients with enlarged vestibular aqueduct and normal genetic studies.[24]

In this syndrome, CT provides details of the bony labyrinth, whereas MRI scans present images of its contents.[1-3] Dahlen et al[25] found a good correlation between the diameter of the bony labyrinth as measured by CT and the diameter of its contents using MRI. The vestibular aqueduct is considered to be oversized when it measures more than 1.5 mm in width, measured at the midpoint of its trajectory, from the common crus to its external aperture.[1,3,7,8,18,26] Antonelli et al[22] classified EVAS into 5 degrees (Table 1).

The size of the vestibular aqueduct in our cases varied between 1.8 and 3.1 mm (Table 2). MRI scans allow demonstration of the presence of enlarged endolymphatic sac and canal, as well as its extra-osseal projection onto the posterior fossa. The existence of patients with sensorineural hearing loss, presenting normal CT scans and enlarged endolymphatic sac and canal (false negatives with CT) allows us to state that MRI scans are the most appropriate imaging method for detecting this malformation.[17]

Patients with EVAS usually present a moderate level of hearing deficit in childhood and this gradually increases until it is diagnosed after a course of several years.[4,19] These patients may benefit from a cochlear implant, as shown in recent literature.[18,27]

On occasions, sensorineural hearing loss is fluctuating and associated with vestibular symptoms,[28-30] as in 2 of our patients.

In conclusion, EVAS is a relatively frequent entity and must be suspected in patients with unilateral or bilateral sensorineural hearing loss where the MRI scan shows that

Table 1. Antonelli's Classification of Enlarged Vestibular Aqueduct

Grade I	The contents of the vestibular aqueduct is only visible in the temporal bone
Grade II	The contents of the vestibular aqueduct is visible close to the common crus
Grade III	The contents extend beyond the common crus, but is not visible at the exit to the vestibule
Grade IV	The internal portion of the aqueduct is visible and its diameter at the exit to the vestibule is smaller than or equal to the common crus
Grade V	The internal portion of the aqueduct is visible and its diameter at the exit to the vestibule is larger than the diameter of the common crus

Table 2. Summary of the Patients' Signs and Symptoms

	Gender	*Age*	*Type of Hearing Loss*	*Vertigo*	*Tinnitus*	*VA*
Case 1	Male	25	Unilateral mixed hearing loss	No	No	1.8 mm
Case 2	Male	54	Bilateral SNHL	Yes	No	1.9 mm
Case 3	Male	36	Unilateral SNHL	No	No	2.1 mm
Case 4	Female	32	Unilateral SNHL	Yes	No	3.1 mm

the vestibular aqueduct is large (>1.5 mm in width), especially if the hearing loss is progressive since childhood. An early diagnosis is very important as these patients may benefit from a cochlear implant.

REFERENCES

1. Valvassori GE, Clemis JD. The large vestibular aqueduct syndrome. Laryngoscope. 1978;88:723-8.
2. Jackler RJ, De La Cruz A. The large vestibular aqueduct syndrome. Laryngoscope. 1989;99:1238-43.
3. Levenson MJ, Parisier SC, Jacobs M, et al. The large vestibular aqueduct syndrome in children. Arch Otolaryngol Head Neck Surg. 1989;115:54-8.
4. Arcand P, Desrosiers M, Dube J, et al. The large vestibular aqueduct syndrome and sensorineural hearing loss in the pediatric population. J Otol. 1991;20:247-50.
5. Emmett JR. The large vestibular syndrome. Am J Otol. 1985;6:387-403.
6. Griffith AJ, Arts HA, Downs C, et al. Familial large vestibular aqueduct syndrome. Laryngoscope. 1996;106:960-5.
7. Kodama A, Sando I. Postnatal development of the vestibular aqueduct and endolymphatic sac. Ann Otol Rhinol Laryngol Suppl. 1982;96:3-12.
8. Kodama A, Sando I. Dimensional anatomy of the vestibular aqueduct and the endolymphatic sac in human temporal bones. Ann Otol Rhinol Laryngol Suppl. 1982;96:13-20.
9. Ieda Maria I, Makoto S. Lateral semicircular canal and vertigo in patients with large vestibular aqueduct syndrome. Otol Neurotol. 2006;27:788-92.
10. Gussen R. The endolymphatic sac in the Mondini disorder. Arch Otorhinolaryngol. 1985;242:71-6.
11. Chen-Chi W, Yuh-Shyng C. Common clinical features of children with enlarged vestibular aqueduct and Mondini dysplasia. Laryngosope. 2005;115:132-7.
12. Murofishi T, Ouvrier RA, Parker GD, et al. Vestibular abnormalities in CHARGE association. Ann Otol Rhinol Laryngol. 1997;106:129-34.
13. Okuno T, Takahashi H, Shibahara Y, et al. Temporal bone histopathologic findings in Alagille's syndrome. Arch Otolaryngol Head Neck Surg. 1990;116:217-20.
14. de Minteguiaga C, García Ibáñez L, Tran Ba Huy P. Tumor del saco endolinfático y enfermedad de Von Hippel-Lindau. Revisión de la literatura. Acta Otorrinolaringol Esp. 2002;53:515-20.
15. Williamson RA, Coker NJ. Endolymphatic sac tumor in von Hippel-Lindau disease. Otol Neurotol. 2003;24:832.
16. Phelps PD, Coffey RA, Trembath RC, et al. Radiological malformations of the ear in Pendred syndrome. Clin Radiol. 1998;53:268-73.
17. Moshe G, Glaser M. CT of the ear in Pendred syndrome. Radiology. 2005; 235:537.
18. Silke S, Brockmeier S-J, Kiefer J. The large vestibular aqueduct – case report and review of the literatura. Acta Oto-Laryngologica. 2006;126: 788-95.
19. Boston M, Halsted M, Meinzen-Derr J, Bean J, Vijayasekaran S, Arjmand E, et al. The large vestibular aqueduct: A new definition based on audiologic and computed tomography correlation. Arch Otolaryngol Head Neck Surg. 2007;136:972-7.
20. Zalzal GH, Tomaski SM, Vezina LG, et al. Enlarged vestibular aqueduct and sensorineural hearing loss in childhood. Arch Otolaryngol Head Neck Surg. 1995;121:23-8.
21. Damborenea T, Hernandez D, Fraile R, Llorente A, Garcia A, Vicente G, et al. Síndrome del acueducto vestibular dilatado en la infancia. A proposito de un caso. An Otorrinolaringol Ibero Am. 2001;28:5-11.
22. Antonelli PJ, Nal AV, Lemmerling MM, et al. Hearing Loss with coclear modiolar defects and large vestibular aqueducts. Am J Otol. 1998;19:306-12.
23. Ramirez-Camacho R, Garcia Berrocal JR, Arellano B, Trinidad A. Familial isolated unilateral large vestibular aqueduct syndrome. ORL J Otorhinolaryngol Relat Spec. 2003;65:45-8.
24. Gonzalez-Garcia JA, Ibanez A, Ramirez-Camacho R, Rodríguez A, Garcia-Berrocal JR, Trinidad A. Enlarged vestibular aqueduct: looking for genotypic-phenotypic correlations. Eur Arch Otorhinolaryngol. 2006;263: 971-6.
25. Dahlen RT, Harnsberger HR, Gray SD, Shelton C, Allen R, Parkin JL, et al. Overlapping thin-section fast spin-echo MR of the large vestibular aqueduct syndrome. AJNR. 1997;18:67-75.
26. Vijayasekaran S, Halsted MJ, Boston M, et al. When is the vestibular aqueduct enlarged? A statistical analysis of the normative distribution of vestibular aqueduct size. S Am J Neuroradiol. 2007;28:1133-8.
27. Claros P, Sanz JJ, Claveria MA, Costa C, Claros A. Implante coclear en paciente con dilatación del saco endolinfático y del acueducto del vestíbulo. Acta Otorrinolaringol Esp. 2005;56:132-4.
28. Broohhouser PF, Worthington DW, Nelly WJ. Fluctuating and/or progressive sensorineural hearing loss in children. Laryngoscope. 1994;104:958-64.
29. Schessel DA, Nedzelski JM. Presentation of large vestibular aqueduct syndrome to a dizziness unit. J Otol. 1992;21:265-9.
30. Oh AK, Ishiyama A, Baloh RW. Vertigo and the enlarged vestibular syndrome. J Neurol. 2001;248:971-4.