

## Audiological findings in large vestibular aqueduct syndrome

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### Abstract

An enlarged vestibular aqueduct is a congenital disorder causing early onset and progressive hearing loss in children. This paper presents the audiological findings at first presentation and the audiological evolution in 10 consecutive cases presenting with hearing loss and showing a large vestibular aqueduct on imaging. The reported onset of the hearing loss is within the first few years of life. Most of the cases (80%) showed bilateral involvement. The sex ratio was 1. Patients presented on average at age 5 with a median hearing loss of 62 dB at the speech frequencies. The hearing loss was essentially asymmetrical with an interaural difference of 33 dB and it was a mixed type of hearing loss in 90% of the cases. The authors claim that the conductive component of this hearing loss is a pure cochlear conductive loss which may be pathognomonic for the disease. The presence of a conductive component in a child is easily misinterpreted as a middle ear ventilation problem or in case of good ventilation as an ossicular problem (type otosclerosis). In addition and in contrast to most literature data, the authors did not find evidence for stabilization of the hearing loss but they found a steady decrease of the hearing at an average rate of 4 dB/year. © 1999 Elsevier Science Ireland Ltd. All rights reserved.

**Keywords:** Large vestibular aqueduct; Congenital malformation; Sensorineural hearing loss; Cochlear conductive hearing loss; Progressive

### 1. Introduction

Large Vestibular Aqueduct Syndrome was first described as a morphological and clinical entity in 1978 by Valvassori and Clemis [1].

The vestibular aqueduct is the bony canal origi-

nating on the medial wall of the vestibule and extending towards the cerebellar face of the petrous pyramid [2]. It contains a vein, an artery and the endolymphatic duct. Enlargement of the vestibular aqueduct is considered to be a minor dysmorphology belonging to the family of Mondini dysplasias. Since in the series of Valvassori this enlargement was often accompanied by other dysmorphologies, such as an enlarged vestibule, an enlarged semicircular canal or a hypoplastic

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cochlea, Valvassori introduced the term 'Large Vestibular Aqueduct Syndrome'. An enlarged vestibular aqueduct is found in almost all Pendred syndromes, but it is more frequently found as an isolated entity.

The diagnosis is made by imaging and different criteria may be used. On CT a vestibular aqueduct with a diameter larger than 1.5 mm is considered abnormal [3]. On MRI, in general and as a rule of thumb, a vestibular duct and/or sac which exceeds in diameter that of the posterior semicircular duct may be considered to be an enlarged vestibular duct and/or sac [4].

The clinical picture is most often that of a child or a youngster presenting with a sensorineural hearing loss. Some variability exists in the audiological features between the different reports [1,3,5–7]. The hearing loss is generally reported as sensorineural, although some authors mention a mixed hearing loss in a minority of their patients (17% [3], 27% [5], 38% [6]). Bilateral involvement is reported in 59–94% of cases and in these cases symmetrical hearing is suggested [2]. The hearing is often reported to be stable (81% [3], 64% [7]). Yet many papers also report a deterioration which may be stepwise and associated with minor head trauma [8]. Jackler and De La Cruz reported an average deterioration of 25 dB in 6 years [5].

The present paper consists of a report on 10 consecutive cases with emphasis on the audiological findings.

## 2. Material and methods

Ten consecutive cases in which a large vestibular aqueduct was diagnosed were studied retrospectively. Several parameters were registered, namely sex, age at first presentation, reported age of onset, family history, history of head trauma.

Audiometry and speech audiometry were performed in a soundproof room with an audiometer calibrated according to ISO (International Standards Organization) standards. Transient click-evoked oto-acoustic emissions were recorded with the Otodynamics ILO88 equipment. Auditory Brainstem Responses (ABR) and electrocochleography were recorded with the Madsen ERA 2250 system.

All patients were studied using enhanced and Gadolinium enhanced 3-mm contiguous T1-weighted images, 500/15/4 (repetition time/echo time/excitations). A three-dimensional Fourier transformation-constructive interference in steady state (3DFT-CISS) gradient-echo sequence was also used and enables detailed evaluation of the membranous labyrinth. The parameters of this sequence are: 0.7-mm-thick slices, in plane resolution of  $0.66 \times 0.66$  mm (field of view of 170 mm and a  $256 \times 256$  matrix), repetition time of 15 ms, echo time of 21 ms, two acquisitions, flip angle of  $65^\circ$  and a total acquisition time of 8 min 6 s.

Parametrical data are expressed as mean and range. Audiometrical data are expressed in terms of five-parameter statistics [9]. A linear regression analysis was performed for the evaluation of the deterioration of hearing over time.

In case of bilateral involvement only one ear was included in the statistical analysis in order to respect the requirement of independency of data. In such a case, a random choice was made as to which ear to include.

## 3. Results

Ten consecutive cases were included, five males and five females. The age at first ENT-visit averaged 5 years (range 0–8 years). All cases presented with hearing loss, two unilateral and eight bilateral. In all cases the diagnosis of Large Vestibular Aqueduct was made on MRI (Fig. 1).

The mean follow-up was 6 years (range 3 months–13 years).

One child with bilateral involvement had a father and a mother who were both bilaterally deaf due to a Large Vestibular Aqueduct, as confirmed on MRI (Fig. 2). The pedigrees of two other children were reported to each contain another case of congenital hearing loss, but further information was lacking.

As to associated congenital anomalies, one case showed mild anomalies of the vestibulum (slightly dilated), one case showed a torsion-anomaly of the kidney and one case showed a hypofunction of the thyroid, of which no further data were available.

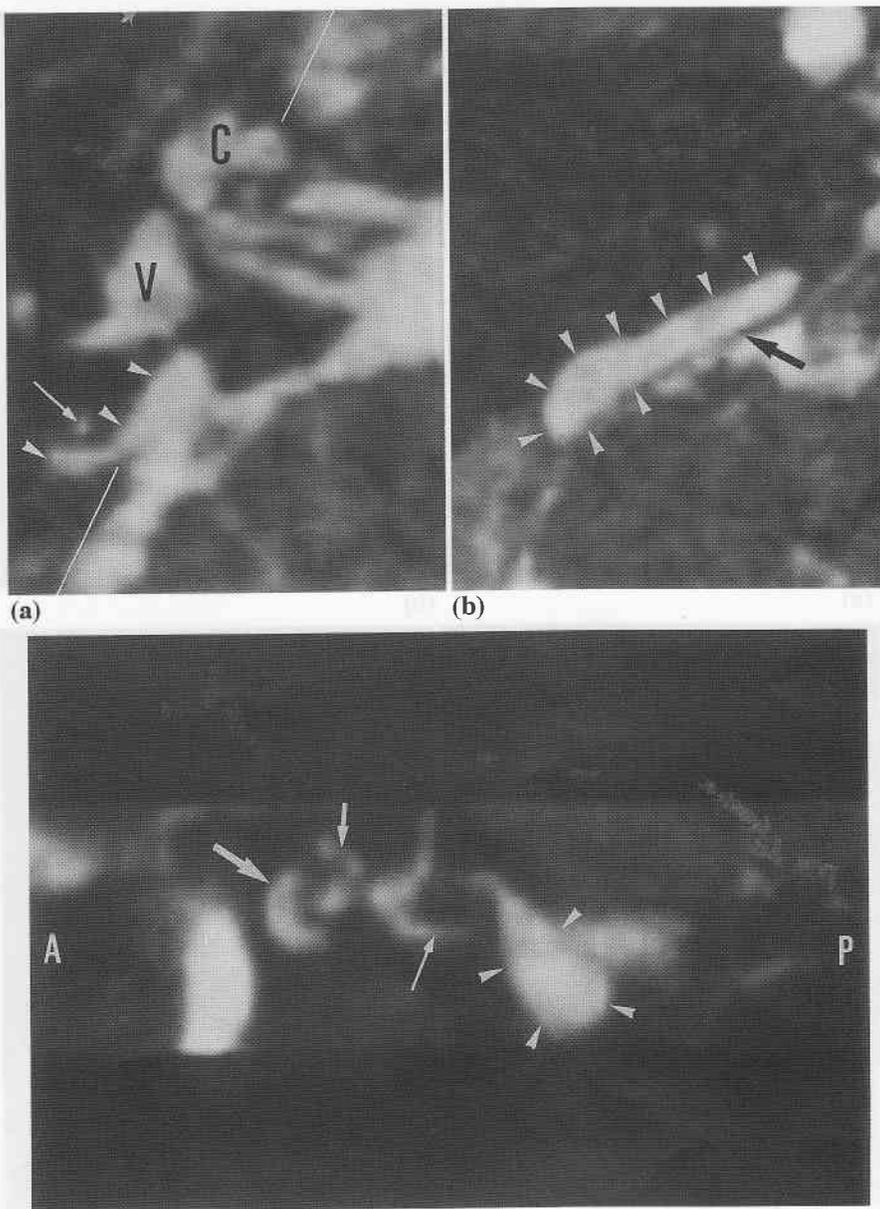


Fig. 1. Axial 0.7-mm thin 3DFT-CISS images (a,b) and a para-sagittal multiplanar reconstruction (c) through the right membranous labyrinth in a 33-year-old man, with an enlarged vestibular duct and sac bilaterally. (a) The endolymphatic duct and sac (white arrowheads) are dilated and their diameter is larger than the diameter of the posterior semicircular canal (long white arrow). Fluid filled cochlea (C) and vestibule (V). (b) Axial image at the level below the membranous labyrinth. The extension of the endolymphatic sac (white arrowheads) in the posterior fossa can be seen on this image. The sac is separated from the cerebrospinal fluid surrounding the cerebellum by the dura mater (large black arrow). This dura mater and the fluid in the enlarged endolymphatic duct and sac can only be seen in a reliable way on thin T2-weighted images (e.g. gradient-echo images, fast spin-echo images, etc.). (c) The extension of the endolymphatic sac in the posterior fossa (white arrowheads) can be recognized on this para-sagittal reconstruction made along the white line in (a). Cochlea (large white arrow), internal auditory canal (small white arrow), posterior semicircular canal (long white arrow), A, anterior; P, posterior.

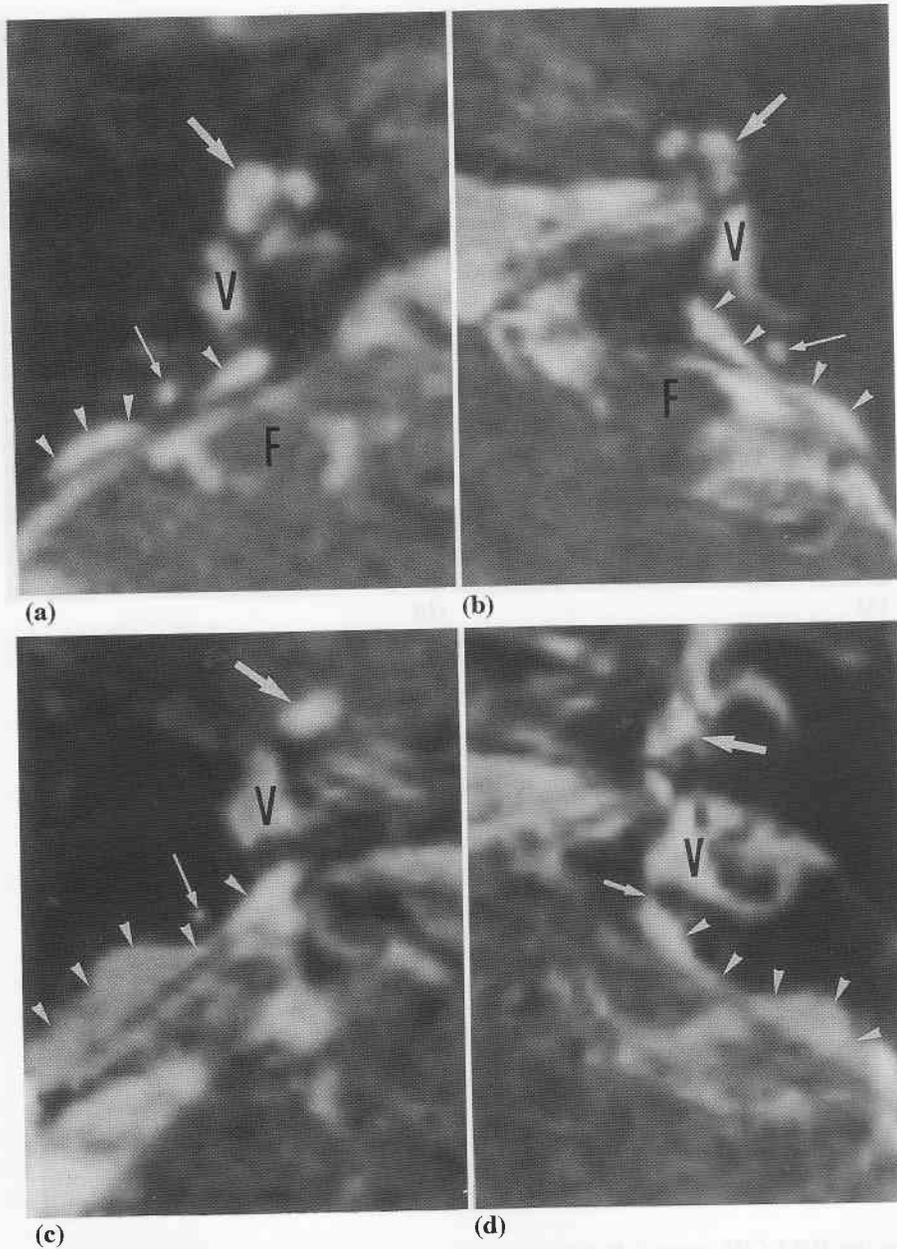


Fig. 2. Axial 0.7-mm thin 3DFT-CISS images through both membranous labyrinths in the wife (a, b) and child (c, d) of the man illustrated in Fig. 1. (a, b) The large fluid filled endolymphatic sacs (white arrowheads) can be depicted posterior to the vestibule (V), then pass the posterior semicircular canal (long white arrow) which has a smaller diameter, and then reach the posterior fossa. Fluid filled cochlea (large white arrow), F, flocculus. (c, d) Again the enlarged endolymphatic sacs (white arrowheads) can be followed behind the posterior semicircular canal (long white arrow) and reach the posterior fossa on both sides. On the left side one can even recognize the endolymphatic duct or connection with the vestibule (small white arrow). Cochlea (large white arrow), vestibule (V).

The audiometrical results at the first visit are shown in Fig. 3 and Table 1. A median hearing loss of 62 dB was recorded at the speech frequencies (Pure Tone Average or PTA, average of 0.5, 1 and 2 kHz). In all cases the audiogram was down-sloping. The better ear showed a median loss of 54 dB and the worse ear of 87 dB at speech frequencies.

In nine cases, the type of hearing loss was mixed with a distinct conductive component. Tympanometry was performed in six cases and a type A tympanogram was found in all of them. Click-evoked oto-acoustic emissions were examined in four cases and were absent in all of them.

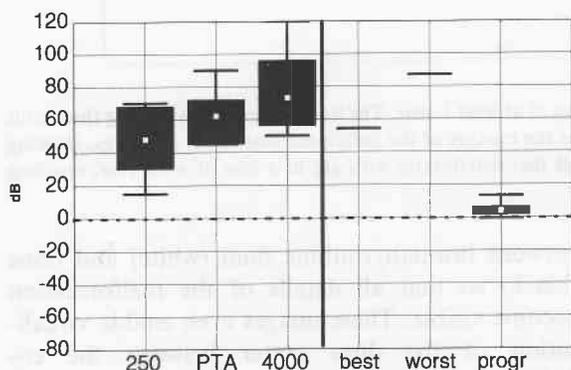


Fig. 3. Box and whisker plots representing some audiometrical variables. 250, PTA, 4000: hearing thresholds (dB HL) at first ENT-visit at 250 Hz, Pure Tone Average and 4000 Hz respectively. Best, worst: PTA at the better respectively the worse hearing ear. Progr: deterioration of the hearing expressed as dB/year at the PTA. Bars, minimum to maximum; large rectangles, 25–75%; small squares, median value.

Table 1  
Numerical data for the box and whisker plots in Fig. 3, representing some audiometrical variables<sup>a</sup>

	250 Hz	PTA	4000 Hz	Progr.
Maximum	70	90	120	13
Upper quartile	68	72	96	6.5
Median	48	62	73	4
Lower quartile	30	45	56	1.4
Minimum	15	45	50	-0.6

<sup>a</sup> 250 Hz, PTA, 4000 Hz: hearing thresholds (dB HL) at first ENT-visit at 250 Hz, Pure Tone Average and 4000 Hz, respectively. Progr., deterioration of the hearing expressed as dB/year at the PTA. Five parameters as in Ref. [9].

Speech audiometry was performed in four cases and ABR in three cases and neither added any further information. Electrocochleography was performed in one case with unilateral involvement and showed an enlarged negative summing potential with an SP/AP-ratio of 0.70 in the involved ear and a normal summing potential with an SP/AP-ratio of 0.22 in the non-involved ear.

The evolution of the hearing is displayed in Fig. 4. Linear regression analysis shows a linear deterioration of 4 dB/year with an average correlation coefficient of 0.60. As can be seen on the figure, many cases show episodes of more pronounced hearing loss which recuperates totally or partially. This sometimes occurred after minor head trauma, but often without any evidence of head trauma. Most episodes were not associated with vestibular problems. As explained in Section 2, only one ear per patient was included in this statistical analysis. In all cases with bilateral involvement, the evolution of the other ear was similar to the evolution of the ear that has been included.

Two cases were operated upon for middle ear inspection. In both cases the middle ear showed no anomaly, the ossicular chain was intact and its mobility was absolutely normal. The only noteworthy observation that was found in both operation records was the absence of a 'round window reflex', which means that the round window membrane could not be displaced by moving the stapes in the oval window.

One case was operated upon and received a LAURA<sup>®</sup> Cochlear Implant. The same findings were recorded as in the former two cases. In addition, upon the opening of the basal cochlear winding before the insertion of the cochlear implant electrode, a profuse perilymph leaking was observed, reflecting the higher than normal pressure in the cochlea.

#### 4. Discussion

The large vestibular aqueduct syndrome was first described in 1978 [1]. It was diagnosed on radiological tomography of the inner ears of 50 patients presenting with hearing loss. In subse-

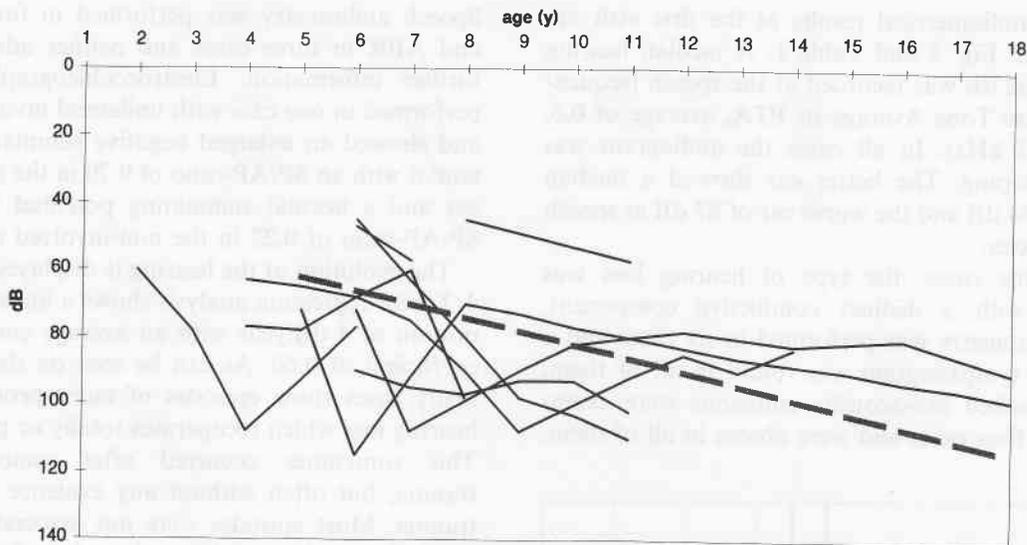


Fig. 4. Evolution of hearing thresholds of nine children with a follow-up of at least 1 year. The lines represent the hearing thresholds (dB HL) at PTA of one child as a function of age. The broken line is the median of the individual best linear fit curves, showing that on average children present at age 5 with a hearing loss of 62 dB that deteriorates with age at a rate of 4 dB/year, resulting in a severe hearing loss or deafness by age 18.

quent years and with the introduction of other medical imaging techniques, the diagnosis has become more common and several papers have reported on this new clinical entity [1–3,5–8,10–12]. It has been called a syndrome because of its frequent association with other inner ear anomalies such as enlarged vestibule, enlarged lateral semicircular canal, hypoplastic cochlea, etc. [1]. The term syndrome may however be illegitimate, since the associated anomalies are localized in the same organ and since 'isolated' large vestibular aqueducts are frequently encountered as well, with the same clinical symptoms. The present authors support the suggestion by Emmett to consider a large vestibular aqueduct as a minor variant of a Mondini deformity [3].

The large vestibular aqueduct can be recognised on CT [3], but only MR images can demonstrate the extension of the large endolymphatic sac in the posterior fossa [4,13]. However this malformation and its extension in the posterior fossa can only be recognised in a reliable way when T2-weighted gradient-echo (e.g. 3DFT-CISS) or fast spin-echo sequences are used. Only these images are thin enough and provide enough contrast

between intralabyrinthine fluid (white) and bone (black) so that all details of the malformation become visible. These images even enable visualisation of the dura mater between the endolymphatic sac and cerebrospinal fluid surrounding the cerebellum. Routine T1-weighted spin-echo images are not sensitive enough. In our experience they can depict a large endolymphatic duct/sac in only 26% of these patients and should therefore not be used as the only sequence to detect congenital inner ear malformations [13].

In agreement with the literature data, the present series shows mainly bilateral involvement (80%). So far, not much attention has been given in literature to the symmetry of the hearing loss. We found a median interaural difference of 33 dB, with thresholds of 54 dB in the better hearing ear at the time of first presentation.

The average age of presentation was 5 years, which is in line with other reports. Zalzal et al. [7], Levenson et al. [12], Arcand et al. [2] and Belenky et al. [11] mentioned an average age of 4.9, 4.5, 3.1 and 3.5 years, respectively at initial presentation. The mean age at the first ENT-visit was higher in other papers: 11 [5,8], 16 [6], 20 years [3].

The reported age of onset of the symptoms (hearing loss) is even lower and can be assumed to be prelingual.

The type of hearing loss is commonly being reported as sensorineural [7,8,11,12], although some papers mention a mixed type of hearing loss in a minority of cases: 33% [1], 27% [5], 38% [6]. In our series, nine patients out of 10 showed a mixed type of hearing loss. In six cases tympanometry was performed and showed a type A curve, meaning that the conductive component could not be explained by middle ear impedance problems (such as effusion). In addition, middle ear inspection in three cases showed normal mobility of the ossicular chain and absence of a round window reflex. The latter finding is suggestive of a cochlear mechanical problem. This was confirmed in one case where an excessive perilymph leakage was observed upon opening the basal cochlear turn for the insertion of a cochlear implant electrode. A similar finding was also reported by Schessel and Nedzelski [14]. In addition electrocochleography was performed in one patient and showed an enlarged negative summing potential and this was also reported to be the case in four out of 14 patients in another study [3]. All these findings contribute to the idea of an intrinsic cochlear conductive hearing loss. The cause of this functional anomaly is still unknown, but it seems reasonable to speculate that an enlarged vestibular aqueduct with an enlarged endolymphatic duct may cause mechanical endocochlear problems, either by volume or by pressure effects.

The hearing loss is said to be stable (though fluctuating) in most cases and progressive in some. The present data give evidence of a steady decrease of the hearing over time. Sudden attacks of hearing loss (10–20 dB) may occur, sometimes following minor head trauma. In most cases the hearing recovers to its former level. Since many patients may present just after a sudden hearing loss, their thresholds may be assumed to be at their worst and to recover in the next few days or weeks. If afterwards the hearing slowly goes down at a rate of 4 dB/year, as suggested by our data, it may take a couple of years before the thresholds will reach the same level as the level at presenta-

tion. During this period the observer will believe the hearing to remain stable. The present authors therefore believe that the idea of a stable hearing may be untrue and may be due to too short a follow-up period. They feel the data of Jackler and De La Cruz [5] confirm this: they reported an overall deterioration of 25 dB over a median follow-up period of 6 years in 12 patients.

In general, a large vestibular aqueduct is believed to be congenital, yet not inherited. In this regard it is remarkable that in one family, both father and mother and their only child have bilateral large vestibular aqueducts with associated hearing loss. This finding may be a coincidence but it may also be some evidence of an autosomal recessive trait. The family history did not report consanguinity. No audiometric or imaging results are available from the rest of the family, but the family history does not mention other members with hearing loss. On the other hand the relatively closed community of the deaf may increase the likelihood of two affected homozygotes meeting one another. Recently a report has been published on two brothers with large vestibular aqueducts [15] and of several families with large vestibular aqueducts in one generation [16], which may also be suggestive of an autosomal recessive manner of inheritance.

In conclusion, the authors believe that the clinical picture of a large vestibular aqueduct may be refined on the basis of the present data. They advocate the clinical entity should not be named a syndrome. It is rather a congenital anomaly that may be considered a minor variant of the Mondini dysplasia and that is characterized by a prelingual hearing loss that is probably mild in the first few years of life, but that deteriorates at a rate of 4 dB/year, resulting in severe hearing loss or deafness by adulthood. Episodes of sudden hearing loss may occur and they usually recover totally or partially. The hearing loss is basically asymmetrical with an interaural difference of approximately 30 dB and it is mixed with its conductive component being due to cochlear mechanical disturbances. Some evidence exists that it may be inherited in an autosomal recessive way and that it may be associated with thyroid dysfunction.

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