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Familial Large Vestibular Aqueduct Syndrome: A Report of Two Siblings

Ailesel Geniş Vestibüler Kanal Sendromu: İki Kardeşe Ait Olgu Sunumu

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ABSTRACT

The large vestibular aqueduct syndrome is a disorder in the spectrum of congenital inner ear malformations with distinct features. The syndrome is associated with bilateral progressive sensorineural hearing loss marked with decreases in the hearing level following minor head trauma or an event causing increased intracranial pressure. We present two siblings with inherited large vestibular aqueduct syndrome and discuss clinical, audiologic, radiographic and surgical findings. Cochlear implantation was performed to the first patient, while her sister had perilymphatic fistula during the follow-up. This family with unaffected parents provides a better understanding of the pathophysiology of large vestibular aqueduct syndrome.

Keywords
Vestibular aqueduct; inner ear; abnormalities; sensorineural hearing loss

ÖZET

Geniş vestibüler kanal sendromu belirgin özellikleri ile konjenital iç kulak malformasyonları yelpazesinde değerlendirilen bir hastalıktır. Sendrom minör kafa travmaları veya kafa içi basıncını arttıran bir olay sonrası azalan işitme seviyesi ile belirginleşen bilateral ilerleyici sensörinöral işitme kaybı ile ilişkilidir. Geniş vestibüler kanal sendromu tespit edilen iki kardeşi sunmakta ve klinik, odyolojik, radyolojik ve cerrahi bulguları tartışmaktayız. İlk hastaya koklear implant uygulanırken, kardeşinde takipler esnasında perilenfatik fistül tespit edildi. Hastalıktan etkilenmeyen ebeveynler ile bu aile geniş vestibüler kanal sendromu patofizyolojisinin daha iyi anlaşılmasını sağlamaktadır.

Anahtar Sözcükler
Akueduktus vestibuli; iç kulak, anormallikler; sensörinöral işitme kaybı

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INTRODUCTION

A large vestibular aqueduct (LVA) is one of the most common congenital inner ear malformations, associated with sensorineural hearing loss (SNHL).[1-4] Although LVA is reported in incomplete partition type II malformation (Mondini deformity),[5] large vestibular aqueduct syndrome (LVAS) is considered to exist only when enlargement of the vestibular aqueduct (VA) is the sole anomaly of the inner ear evident on radiographic studies.[6-8] The patients typically experience bilateral and progressive SNHL, with acute decreases in the hearing level following head trauma or activites involving a Valsalva maneuver.[1,2,7,9] Currently, there is no treatment to prevent the progression of SNHL in patients with LVAS, but avoidance of head trauma is essential for patient care.[2,6,9]

We present two siblings with LVAS with clinical, audiologic, radiographic, and surgical analysis and emphasize the importance of considering isolated LVAS as a separate entity in the spectrum of inner ear diseases.

CASE REPORT

CASE 1

The first patient (Pt.1) was first suspected to have hearing loss at 3 years of age, had the audiological evaluation elsewhere, and was fitted with a hearing aid on the left ear. Her first audiological evaluation at the age of 9 years demonstrated profound SNHL on the right ear, and moderate to severe mixed type hearing loss on the left ear (Figure 1). Acoustic immitancemetry indicated type As tympanograms bilaterally, with acoustic reflexes present only on the left ear contralaterally at the maximum intensity levels at 500 Hz, 1 kHz and 2 kHz. The patient was managed with conventional hearing amplification.

The birth history was normal with the patient delivered at full term by normal labor. The mother was not exposed to any known potential teratogens. The neonatal course included physiologic jaundice, requiring no treatment. Pt.1 had no history of ototoxic medication exposure, head trauma or meningitis. Developmentally, she had met appropriate neurologic milestones. There was no history of consanguinity or genetic disorders in the family.

The otorhinolaryngologic and neurologic examinations revealed no abnormalities. The tympanic membranes were intact and mobile with no evidence of ossicular malformations. The cranial nerves were intact bilaterally. She had no dysmorphic features or goitre. Routine blood chemistry tests were normal. Complete metabolic workup including thyroid function tests and thyroid antibody screening were within normal limits. A temporal bone computed tomography (CT) scanning demonstrated bilateral LVA (Figure 2). The right and left VAs measured 4.2 mm and 4 mm, respectively, according to the technique of Valvassori and Clemis.[8] The radiologic anomaly was consistent with isolated LVAS.

Her hearing level deteriorated and eventually progressed to profound SNHL over the next six years. The patient was evaluated for the cochlear implantation at the age of 17 years (Figure 3) and found to be a good candidate. Cochlear implantation to the right ear was performed in the year 2000 without perilymphatic gusher during cochleostomy. Full insertion of the electrode ar-

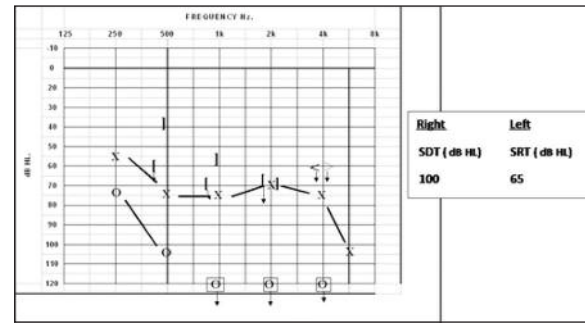

Figure 1. The first audiogram of Pt.1 obtained at the age of 9 years (SDT:speech detection test, SRT: speech reception threshold).

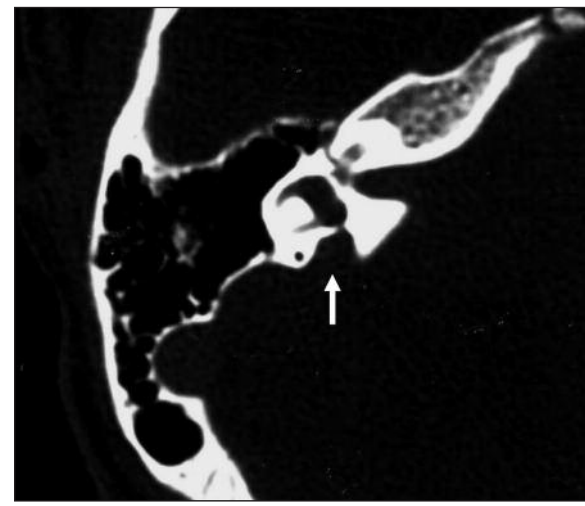

Figure 2. Axial computed tomography scan of Pt.1 demonstrating large vestibular aqueduct (arrow) and vestibule (right ear).

ray was achieved. Electrically evoked stapedius reflexes were recorded and electrically evoked compound action potentials were reliably measured intraoperatively. She has open speech understanding with the cochlear implant.

CASE 2

The first patient's sister (Pt.2) was referred to our clinic for hearing evaluation at the age of 11 years. The audiologic evaluation demonstrated bilateral moderate SNHL with a flat audiometric configuration (Figure 4). Acoustic immitancemetry revealed type As tympanograms bilaterally with present contralateral acoustic reflexes at the maximum intensity levels at 500 Hz, 1 kHz and 2 kHz.

She was the product of a normal pregnancy, had a negative medical history and had proper findings at her physical examination. She had no dysmorphic features or goitre. Routine blood chemistry tests were normal. Complete metabolic workup including thyroid function tests and thyroid antibody screening were within normal limits.

The patient was fitted with moderate gain hearing aids and was followed through routine examinations.

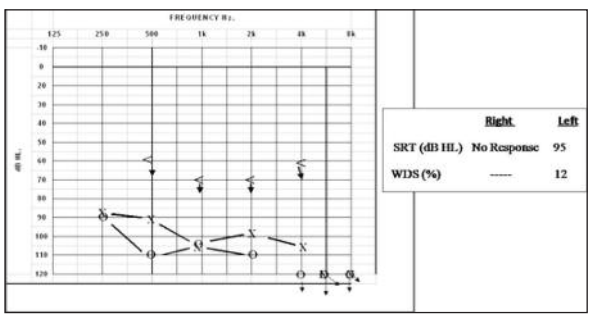

Figure 3. The audiogram of Pt.1 before the cochlear implantation at the age of 17 years (SRT: speech reception threshold, WDS: word discrimination score).

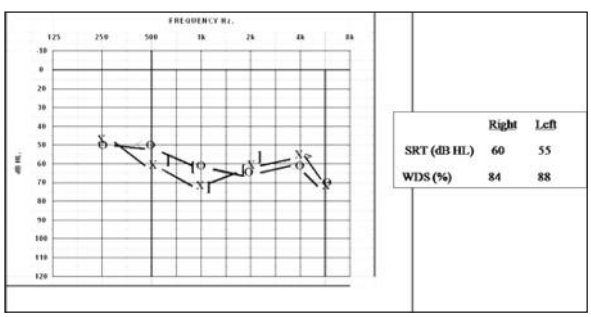

Figure 4. The first audiogram of Pt.2 obtained at the age of eleven years old (SRT: speech reception threshold, WDS: word discrimination score).

The hearing loss progressed gradually from moderate to severe SNHL in the following seven years and the hearing aids' fitting was modified according to the new hearing levels.

In July 2000, the patient was referred with an attack of sudden hearing loss (SHL) and a complaint of aural fullness on the left ear with flat configuration on audiogram. Audiological evaluation indicated profound SNHL on the left ear and severe SNHL on the right ear. Bilateral type As tympanograms were obtained. The pure tone thresholds and the speech tests were found to be deteriorated. Transient evoked otoacoustic emissions were bilaterally absent. Medical treatment with steroids was administered and improvement was observed in the middle and high frequencies. Improvement was also evident on the speech reception threshold and speech discrimination score on the left ear.

One month later, SHL recurred on the right ear. Medical treatment was repeated with an outcome of improvement in both middle and high frequencies as well as speech discrimination scores. The patient experienced unsteadiness with a left beating, first degree, direction fixed horizontal nystagmus. The remaining vestibular examination battery consisted of a negative fistula test and a normal Romberg's test.

A temporal bone CT was obtained, demonstrating a LVA with a measurement of 3.5 mm for both VAs (Figure 5). Magnetic resonance imaging (MRI) scans revealed massive enlargement of the endolymphatic sac, with an enhancement in the mastoid cells and mesotympanum of the right ear isointense to that of endolymphatic system (Figure 6).

After explaining the procedure and the outcome to the family in detail, right explorative tympanotomy was performed with the thorough understanding that there might be no recovery of hearing. Ossicular chain and stapes footplate were hypermobile with a prominent membraneous round window bulging. The escape of perilymph was observed from the oval window with repeated Valsalva maneuver. The middle ear cavity was obliterated with fat tissue and fibrin glue. Postoperative audiological examinations revealed slight improvement in the high frequencies. During the follow-up period of 10 years, she had eight SHL attacks: five times in the right ear and three times in the left ear. Subsequently, Pt.2 is being followed up with progressive SNHL.

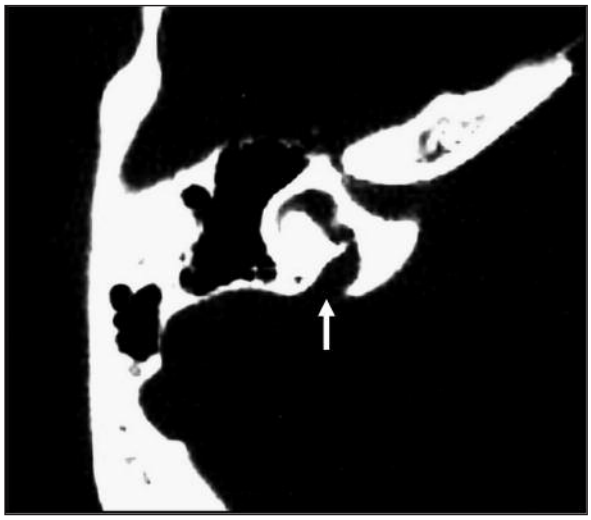

Figure 5. Axial computed tomography scan of Pt.2 demonstrating a large vestibular aqueduct (arrow) (right ear).

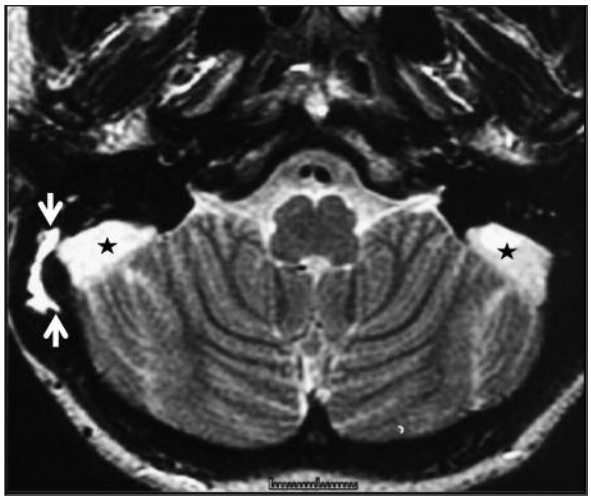

Figure 6. MRI image of Pt.2 showing a large endolymphatic sac (star) with an enhancement in the mastoid cells isointense to that of endolymphatic system (arrow) (right ear).

In order to address the possibility of inheritance of LVA, a thorough evaluation of each parent was performed. The audiologic examinations were normal. HRCT images were obtained from the parents with normal VAs and no detectable inner ear abnormalities, indicating that they are unaffected.

This report was approved by the ethics committee of our department and carried out in accordance with the Declaration of Helsinki. Informed and full consent has been received from the parents for sharing and publishing the data in this study.

DISCUSSION

The VA is the bony canal containing the endolymphatic duct, which is formed by the joining of the utricular and saccular ducts, and the intraosseous portion of the endolymphatic sac.[10,11] It is proposed that an arrest in the development in the 5th week of gestation prior to the lengthening and narrowing of the VA results in a larger-caliber.[1,7,8,12,13] LVA is defined as measured on a radiograph, at half distance between the common crus and its external aperture at the posterior fossa, in the anteroposterior dimension.[3,4,8,12] The radiological diagnosis of LVAS requires that a VA, by a measurement greater than 1.5 mm at the midpoint of the distal limb, be the sole abnormality.[4,6]

With isolated LVA, SNHL which usually begins in early childhood is often experienced with an acute onset, and fluctuating or progressive in course in relation to head trauma and activities involving a Valsalva maneuver.[1,2,7,9,14-16] The SNHL is bilateral in 81% to 94% of cases, ranging from normal to profound deafness with predominantly downsloping audiograms.[1,2,4,7,12] Vestibular symptoms are reported by one third of patients with LVAS.[1,6,17]

Multiple reports have speculated on the pathophysiology of SNHL in LVAS. LVA may not have been able to serve as a resistor to the reflux of the hyperosmotic fluid from the sac into the cochlea after acute pressure fluctuations in cerebrospinal fluid as seen in head trauma, with damage to the cochlear neuroepithelium and vestibular structures.[8,9] Frequent attacks might gradually cause permanent damage to inner ear structures.[1,2] Another possible cause of SHL in LVAS is PLF, which is known to be frequently observed in congenitally malformed ears.[1,18,19] Our patient (Pt.2) had fluctuating and progressive SNHL with a vestibular attack. The suspected PLF was elicited during a right explorative tympanotomy.

The genetic studies mapped the responsibility of LVA and incomplete partition type II malformation to PDS (also known as SLC26A4) gene on chromosome 7q31, which has been known to be associated with Pendred's syndrome (PS).[6,20,21] PS is characterized by a profound SNHL and diffuse goitre.[20] Our patients had thyroid function tests and thyroid antibody screening within normal ranges.

Treatment of LVAS is primarily symptomatic with disappointing results. There exists no beneficial treatment other than amplification with hearing aids.[1,6,9] The

residual hearing of children with LVAS may allow them to acquire excellent speech using these hearing aids before the advent of profound SNHL.[22] If no aidable hearing remains, a cochlear implant may be considered.[22] Pt.1 with profound SNHL had cochlear implantation performed succesfully.

Upon diagnosis of LVAS, the patient must be educated to refrain from even minor head trauma and contact sports.[1,4,6,9] Family members should be screened with audiometry and CT, and discouraged from consanguineous matings. The possibility of LVAS should be considered in children with unexplained progressive SNHL, especially those who develop deafness after head trauma.

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