CASE REPORTS

LARGE VESTIBULAR AQUEDUCT SYNDROME CAUSING PROGRESSIVE HEARING LOSS IN CHILDHOOD

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ABSTRACT

Background: Sensorineural hearing loss in adults is commonly associated with aging, trauma or autoimmune disease. In children, however, sensorineural hearing loss can also be due to genetic causes, leading to malformation of internal ear structures, like the vestibular aqueduct. In this article we present a rare anatomic malformation which causes progressive hearing loss in children.

Methods: Our patient is a 12 year old boy who presented with deafness and tinnitus after mild trauma to the side of the head. Steroids and carbogen were administered but showed no improvement.

Results: Audiological evaluation demonstrated profound bilateral sensorineural hearing loss. Radiological investigations including CT scan and MRI of the temporal bone showed bilateral enlargement of the vestibular aqueduct.

Conclusion: The patient was diagnosed with Large Vestibular Aqueduct Syndrome and recommended cochlear implantation which has a high success rate and improves the quality of life for the patient.

KEY WORDS: Sensorineural hearing loss, Enlarged vestibular aqueduct, enlargedvestibular aqueduct syndrome.

INTRODUCTION

Hearing loss in early childhood is a matter of great concern because at this age, children use their auditory capabilities to develop their speech and language skills. Children with hearing disabilities are often unable to adjust in various social situations leading to behavioral problems and disturbances in learning. The first line of differential diagnoses for sensorineural hearing loss in the pediatric population is post-infectious causes, including both congenital and acquired infections. Anatomic malformations should also be considered, especially if abnormalities are evident in other organ systems indicating the possibility of a genetic syndrome. Enlarged Vestibular Aqueduct Syndrome should be considered in children with progressive hearing loss as the defect is related to the endolymphatic duct which can cause fluctuating hydrodynamic changes in the membranous labyrinth.

METHODS

Our patient is a 12 year old boy, who had been born and brought up in Ireland. He presented to the Emergency Department with complaints of complete bilateral deafness and tinnitus, 15 minutes after receiving a punch to the left side of the face while playing in school.There was no evidence of bleeding or loss of consciousness. Patient described mild to moderate pain in the left side of the face, including left ear. He complained of a constant whistling sound in both ears, but did not feel dizzy or vertigo.Detailed history revealed non-significant birth history with no gestational complications during pregnancy. Parents reported some delay in speech and language development, but not severe enough to disturb the patient's educational activities.

There were no other family members with hearing loss or genetic abnormalities.

On examination, the boy was a healthy looking 12 years old who was co-operative but anxious due to the sudden loss of hearing. There was no bruise around the ear, no hematoma, bleeding or discharge was noted.Otoscopy and rhinoscopy was within normal limits, with no evidence of nystagmus or battle sign.No gross thyroid enlargement was noted on physical examination.

RESULTS

Initial investigations included routine blood tests to rule out infection, auto immune disease and thyroid imbalance, all of which were normal.

Audiometry revealed profound bilateral sensorineural hearing loss.

CT scans (Figure1)were requested to rule out post traumatic injuries and showed bilateral enlarged vestibular aque-

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ducts inears. Subsequent MRI investigation (Figure2)confirmed the diagnosis.

Genetic testing was not performed as the patient showed no other signs or symptoms of multi system involvement.

CONCLUSION

Steroid treatment and Carbogen inhalation had been initiated as emergency management but was stopped when diagnosis was confirmed to be Large Vestibular Aqueduct Syndrome.

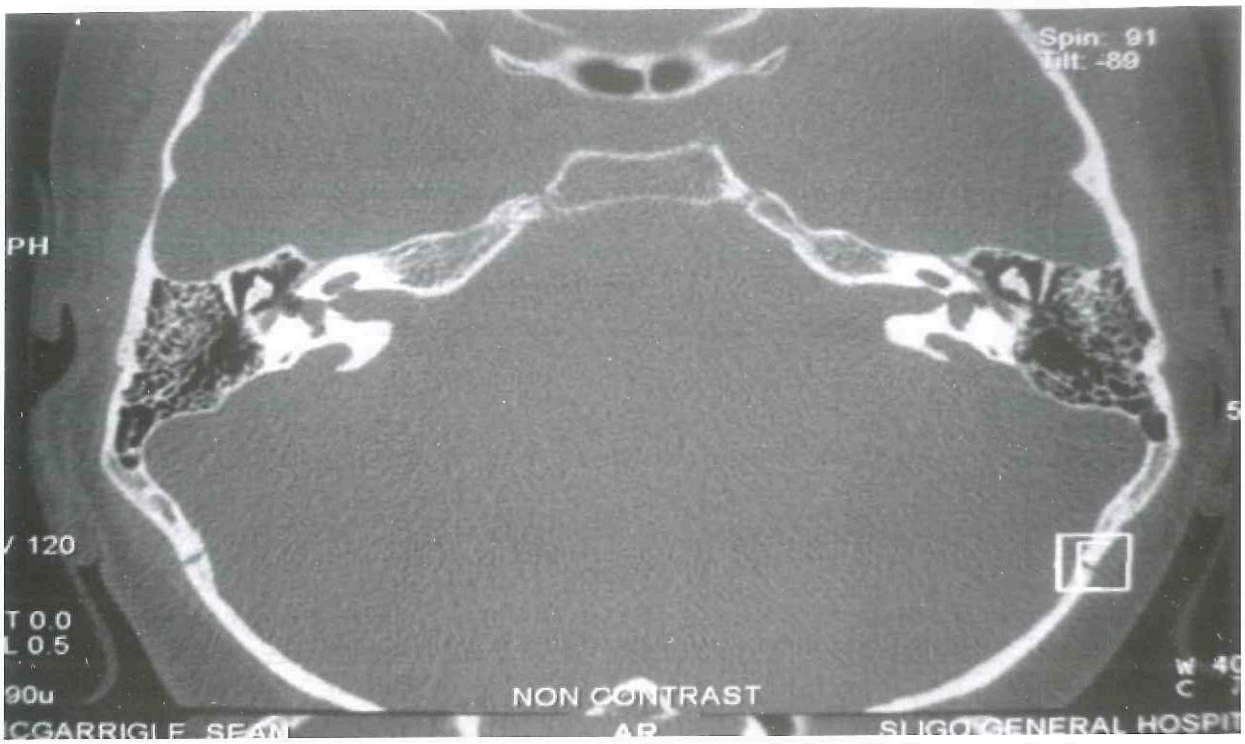

Figure1. CT Scan showed bilateral enlargement of vestibular aqueduct

Table 1: The presence and number of cusps in the IJV valves

ale (n= 40)							Female (n= 22)						
ght side			Left side				Right side				Left side		
one	Uni	Bi	Tri	None	Uni	Bi	Tri	None	Uni	Bi	Tri	None	Uni	Bi
	04	25	00	9	3	28	00	4	0	17	1*	5	2*	15
.50	10.00%	62.50%	00%	22.50%	7.50%	70%	00%	18.18%	00%	77.27%	4.54%	22.72%	9.09%	68.18%

otal Valves on both sides in males and females (n= 124)			
one	Unicuspid	Bicuspid	Tricuspid
)	9	85	1
3.38%	7.25%	68.54%	0.80%

ignificant at p< 0.05 (comparisons were made between the right and left sides of each group)

Figure 2. MRI scan showed enlargement of vestibular aqueduct bilaterally

The family was counseled about the nature of the disease and its prognosis. Management options were discussed and hearing aid was given immediately while cochlear implantation was recommended as early as possible to improve quality of life and patient's well being.

DISCUSSION

The vestibular aqueduct is a bony canal which acts like a passage from the inner ear to the temporal bone. This aqueduct contains the membranous cochlear duct and ends in the endolymphatic sac. The endolymph present in the endolymphatic sac regulates the ion exchange in the cochlear fluids and therefore a change in size of this structure may cause a chemical imbalance leading to changes in auditory sensations.

The enlargement of Large Vestibular Aqueduct Syndrome is the most common inner ear malformation, with an incidence rate of 1%. [1] It can be an isolated defect but may also be associated with some genetic diseases including CHARGE syndrome and Waardenburg syndrome. The most common association is Pendred Syndrome which is an autosomal recessive condition in which enlarged vestibular aqueduct is associated with goiter. Studies show the genetic mutation responsible for Pendred Syndrome involves pendrin (SLC26A4), a chloride/iodide transporter. [2]

Symptoms vary widely with most cases being bilateral and not associated with other inner ear deformities. This problem could present at birth but often becomes a problem after the first few years of life. The age of onset and rate of progression of the deafness is variable but as seen in our case, patients usually present with mild, slowly progressive deafness which gets suddenly exacerbated by a sudden onset of complete deafness after the patient experiences some form of trauma, like the physical blow received by this boy. Reports show that even mild changes like air pressure can cause similar situations. Although hearing loss is of varying severity and fluctuates frequently, pure tone audiometry showed that high tone hearings were worse than low tone hearings, with fluctuations in high tone hearing being smaller than low tones. [3]

Less frequently, patients with Large Vestibular Aqueduct Syndrome complain of balance issues. It is more of a problem when the patient is a child and is not able to explain his condition to others.

Investigations usually begin with audiometry to confirm the type of deafness. When physical examination and routine investigations have ruled out more common causes of sensorineural hearing loss, physicians turn to CT scans and MRI tests to detect any anatomic reason behind the loss of hearing.

Vijaysekaran et al reviewed CT scans of temporal bones in 73 individuals with normal hearing and concluded that the upper limit of normal vestibular aqueduct size is 0.9mm for midpoint and 1.9 mm for opercular width. Anything greater than these measurements is considered as enlarged vestibular aquaduct. [4]

In a study focused on the diagnosis of Large Vestibular Aqueduct Syndrome, Campbell et al pointed out that measurement of the endolymphatic width near the vestibule was more accurate when correlated with severity of deafness ascompared to measurements of endolymphatic width at its mid-point.[5]In a comparison of radiological investigations, it was concluded that while CT scans assess the size of the vestibular aqueduct, MR imaging can provide additional information about the abnormalities of the fluid spaces related to the membranous labyrinth[6] and therefore they both carry their own weightage.

Although CT scans and MRI have proven to be the diagnostic modality of choice, other methods have also been investigated. Taylor et al showed augmentation of air-conducted ocular Vestibular Evoked Myogenic Potentials (oVEMP) in a patient with bilaterally enlarged vestibular aqueducts [7] Auditory brain potentials were also measured in another set of Large Vestibular Aqueduct patients and results showed that ASNR in 2 3 cm by the ABR testing also aided the diagnosis.[8]

Steroid therapy is the gold standard for treating these patients but different therapies have been tried including hyperbaric oxygen [9] and carbogen inhalation which was offered to our patient.These treatment options are directed towards increasing cochlear circulation and oxygenation and have shown good results in patients with sudden onset of deafness, but are of limited value in Large Vestibular Aqueduct Syndrome.

Cochlear implantations are the mainstay of treatment and have been used in the treatment of various similar syndromes including Wardenburg syndrome, Jervell-Lange-Neilsen syndrome and Cogans syndrome. It has a high success rate with improvement in quality of life and early complications are seen in only approximately 6.8% of implantations.[10] Rare complications include infections such as bacterial meningitis and there is also one case of pneumolabirynth reported after cochlear implantation [11]

A study conducted on over forty pediatric patients with Large Vestibular Aqueduct Syndrome showed that cochlear implantation performed before the age of five years produced better results in terms of speech perception and intelligibility.[12]A similar study conducted on over 400 patients showed similar results where auditory skills of infants were seen to rapidly improve after cochlear implantation as measured by the Infant-Toddler Meaningful Auditory Integration Scale (IT-MAIS) [13]

In summary, we identified a case of Large Vestibular Aqueduct Syndrome, in a boy with acute-on-chronic hearing loss. This is the most common inner ear malformation with easily accessible diagnostic modalities and therefore should be kept in mind by both the radiologist and the otorhinolaryngologist when dealing with sensorineural deafness in the pediatric population.

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