

Enlarged Vestibular Aqueduct Syndrome (EVAS)

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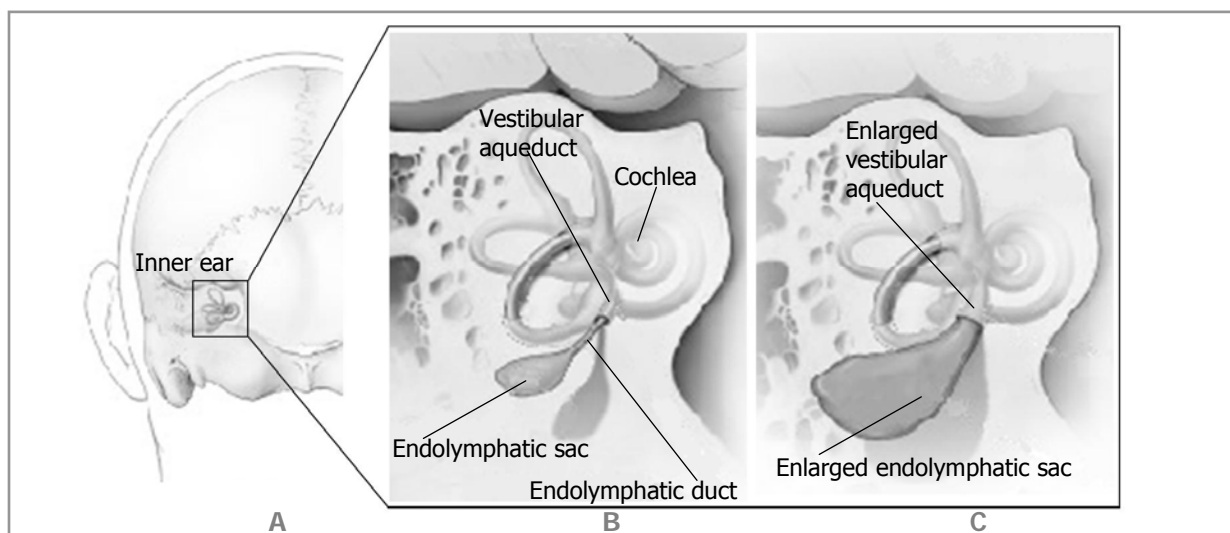
The vestibular aqueduct is a tiny, bony canal that extends from the inner ear endolymphatic space toward the brain. It is shielded by one of the densest bones in the body, the temporal bone, which also houses the sound-sensing cochlea and motion-sensing vestibular organs vital to our ability to hear and maintain balance. Inside the vestibular aqueduct is the endolymphatic duct, a tube that connects the endolymph (a fluid) in the inner ear to the endolymphatic sac. The function of the endolymphatic duct and sac is not totally understood, but it is believed that they help maintain the volume and ionic composition of endolymph necessary for transmitting hearing and balance nerve signals to the brain.

When a vestibular aqueduct is larger than normal, it is known as a large vestibular aqueduct (LVA)

or by the term used here, enlarged vestibular aqueduct (EVA). Hearing loss or balance symptoms associated with an EVA can occur when the endolymphatic duct and sac expand to fill the larger space (see Figure). When EVA is associated with such symptoms, it is referred to as EVA syndrome (EVAS).

Causes

During fetal development, the vestibular aqueduct starts out as a wide tube. By the fifth week it narrows, and by midterm it approaches adult dimension and shape; however, the vestibular aqueduct continues to grow and change until a child is 3 to 4 years old. As yet incompletely understood genetic or environmental conditions cause EVA, which is often congenital (present at birth) or occurs during early childhood.



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Figure: The inner ear as seen from the back of the head, left side (panel A). Close-up view of the inner ear comparing a normal (panel B) and enlarged (panel C) vestibular aqueduct and endolymphatic sac. Adapted by VEDA with permission from the National Institute on Deafness and Other Communication Disorders (NIDCD).

Hearing loss associated with EVAS can be *syndromic deafness*, a loss of hearing accompanied by physical signs and symptoms affecting other parts of the body. More commonly, it is *nonsyndromic deafness*, affecting only ear function (nonsyndromic deafness accounts for approximately 70% to 80% of all genetic hearing loss¹). Just as the cause of EVAS remains unclear, much of what is known about it—why the hearing loss pattern differs among patients, how many people actually have it, how it causes symptoms, how to effectively treat it, and what the prognosis might be—comes from isolated clinical observations and small studies, not from comprehensive scientific research.

Genetic testing often but not always reveals that EVA is associated with mutation of the SLC26A4 gene (also called the PDS gene) which also causes Pendred syndrome, a condition associated with syndromic hearing loss and thyroid disease. Pendred syndrome occurs in an estimated one-third of all cases of EVA² and is an autosomal-recessive genetic disorder, meaning each parent must be a genetic carrier.

EVA can be associated with branchio-otorenal syndrome, which affects the anatomy of the ears, kidney, and neck. Additionally, it can be associated with other anatomical defects such as a Mondini malformation, an incomplete cochlear development that is also linked to a mutation of the PDS gene.

Prevalence

EVAS is considered to be rare, but as with many inner ear disorders, its true prevalence is difficult to assess because it is not always recognized during a medical evaluation. Estimates fall between as high as 5% to 15% in pediatric patients.^{2,3} More women than men are affected; for every two males with EVAS, there are three females who have the disorder. EVAS is associated with vestibular symptoms in a small percentage of people.²

Hearing symptoms

It is hearing loss that usually brings EVAS to the attention of a physician. Such loss can be sensorineural, conductive, or both. *Sensorineural hearing loss* (SNHL) is deafness usually related to the cochlea, but sometimes to the vestibulocochlear nerve or the brain's central auditory system. *Conductive hearing loss* involves reduced movements of the middle ear bones (malleus, incus, and stapes) which conduct external sound to the inner ear.

Some people are born with the hearing loss. However, in most cases of EVAS, a child will hear normally in the first years of life and then notice hearing loss later in childhood, or less commonly in adolescence or early adulthood. Generally, this occurs after a minor or major head impact, upper respiratory infection, or air pressure trauma, such as occurs during the rapid depressurization of an airplane. Even active play, especially jumping, can jar the head enough to result in hearing loss if an EVA is present. The loss can be progressive, fluctuating, stable, or sudden, and can involve tinnitus, a ringing in the ears. However, generally the hearing loss occurs in a series of steps. With each minor event, hearing drops one or more levels, a downward progression often culminating in profound hearing loss.

Vestibular symptoms

Vestibular symptoms sometimes related to EVAS include episodic spinning vertigo, mild unsteadiness, trouble watching revolving objects, a feeling of vague instability, rocking sensations, jumping vision, decreased visual acuity in the presence of loud sounds, instability when leaning forward, vomiting, nausea, and drunken gait. A young child may also grab his or her head and walk in circles. The symptoms of vestibular disorders are notoriously difficult for children and adults to describe; for children, the task is even more challenging. Unless well trained in recognizing vestibular disorders, a physician may not ask the questions necessary to discover them. However,

these signs and symptoms can also be seen in other types of vestibular disorders and are not unique to the diagnosis of EVAS.

Traditionally, physicians have devoted more attention and study to the effect of EVAS on hearing, but an increasing clinical awareness of the impacts of vestibular dysfunction on childhood development is starting to change that. These impacts manifest in outward signs such as reflex delays, visual-spatial problems, motion sensitivity, abnormal movement patterns, clumsiness, difficulty moving in the dark, and lingering anxiety. Many of childhood's common milestones—climbing stairs, riding bicycles—may be delayed, or simply too difficult for a child with EVAS to manage without treatment. It is also important to note that these symptoms can have other causes besides inner ear disorders, which is why a thorough medical evaluation by a pediatrician, otologist/neurotologist, and neurologist is needed.

How does EVAS cause symptoms?

The SNHL and balance symptoms associated with EVAS may occur because the enlarged endolymphatic duct and sac are unable to maintain their normal functions. These include maintaining the endolymph volume and ionic composition (concentrations of sodium, potassium, calcium, and chloride) necessary for transmitting hearing and balance nerve signals to the brain. This disrupts inner ear homeostasis, which is ionic equilibrium among the compartments of the inner ear that contain either endolymph or perilymph, fluids that have specific and different concentrations of ions. If related to head trauma, EVAS may cause symptoms when the sudden fluctuation in cerebrospinal fluid (CSF) pressure on impact forces highly concentrated (hyperosmolar) proteins into the cochlear duct which connects the CSF space to the endolymph space inside the cochlea. This is called hyperosmolar reflux.

Conductive hearing loss with EVAS may occur because the increased endolymphatic pressure reduces the ability of the stapes to move the oval window, which is the membrane separating the middle ear from the fluid-filled inner ear. Because of this dysfunction, sound waves conducted through the middle ear can't be transferred to the cochlea in the inner ear.

Clinical evaluation

The variable signs of EVAS indicate that diagnosis requires special care and attention to a person's symptoms and medical history, especially those of children. In addition to a complete medical history and physical examination, the diagnostic process for uncovering EVAS usually involves hearing and balance testing and radiologic assessment. Thyroid, renal, and cardiac function are also usually analyzed, and genetic screening is sometimes also performed.

Audiologic testing often reveals low frequency conductive hearing loss, high frequency SNHL, or both. Vestibular tests may be useful even if a person with EVAS is not experiencing active vestibular symptoms such as vertigo.⁴ When electronystagmography (ENG) is used to measure eye movements and vestibular responses to thermal (caloric) or rotational stimulation, it may reveal loss of vestibular function in one or both ears and nystagmus, the abnormal eye movements commonly associated with vestibular dysfunction.

For radiologic assessment, fast spin-echo magnetic resonance imaging (MRI) is generally considered to be the most appropriate test^{5,6} because it permits precise imaging and measurement of the endolymphatic duct and sac soft tissues. High resolution computerized tomography (CT) scans of the temporal bone are also often used to confirm the bony enlargement and other bony abnormalities that are often associated with EVAS. No standard radiographic criteria exist yet to define what size constitutes an enlargement.⁷ Some studies suggest

that a vestibular aqueduct is enlarged if it is more than 1.5 millimeters (mm) in diameter, while others define an EVA as being more than 4.0 mm in size.⁵

Treatment

Historically, medical and surgical treatments have not reversed the progression of hearing or vestibular losses from an EVA. Currently, the cornerstone of management is prevention, primarily by protecting the head from traumas that will worsen the progression of hearing loss and vestibular symptoms. People with EVAS are cautioned to avoid contact sports and must wear a helmet while bicycling and performing other activities that elevate risk of head injury. Parents are often challenged to find a middle ground between allowing their children to enjoy the typical physical activities of childhood and preventing future losses to hearing and function. Each person must negotiate his or her own decisions about risk management.

With significant hearing loss, a hearing aid may be used, but often unsuccessfully. For some people, cochlear implantation has significantly improved hearing.⁸ An audiologist can help determine whether options such as communication training in sign language or speech could be helpful. In some people with related vestibular symptoms, treatment may include vestibular rehabilitation therapy; however, as is also common for patients experiencing an active phase of Ménière's disease, EVAS may not respond well to vestibular rehabilitation.

Predicting what will ultimately happen in any one case of EVAS is difficult because the condition follows no typical course. No relationship exists between how large the aqueduct is and the amount of hearing loss a person may sustain. Some cases progress to profound deafness, some include vestibular losses or difficulties, and other cases lead to neither. It's important to note that for some people with EVAS, loss of hearing begins in childhood, but vestibular symptoms are delayed

until adulthood. Accordingly, people with EVAS should seek medical evaluation if they develop unexplained dizziness, vertigo, or other signs of vestibular dysfunction.

Additional Resources

CT images of EVA are available at www.clevelandhearingbalance.com/media.htm

Some helpful documents available from VEDA:

- Pediatric Vestibular Disorders: Recognition, Evaluation, and Treatment (Pub. E-7)
- Vestibular Rehabilitation: An Effective, Evidence-Based Treatment (Pub. F-7)

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