CSI: AUDIOLOGY

WELCOME BACK to an ongoing series that challenges the audiologist to identify a diagnosis for a case study based on a listing and explanation of the nonaudiology and audiology test battery. It is important to recognize that a hearing loss or a vestibular issue may be a manifestation of a systemic illness. Being part of the diagnostic and treatment “team” is a crucial role of the audiologist. Securing the definitive diagnosis is rewarding for the audiologist and enhances patient hearing and balance health care and, often, quality of life.

—Hillary Snapp, Investigator-in-Chief


No Running
Is BPPV Blocking the Path to a Carefree Childhood?
By Hillary Snapp

Case History
OH is an 8–year-old female who presented to the clinic complaining of severe headaches with vertigo. She was accompanied by her mother, who reported that the patient cannot swing, swim, or tolerate sudden direction changes. OH reportedly closes her eyes and braces herself for turns when riding in the car and dislikes running-based sports. OH reported that she enjoys gymnastics, but can no longer participate in this activity. Her symptoms appeared to be motion induced, although she also reported associated headaches with nausea and vomiting. The episodes last hours and occur 7 out of every 10 days. The patient’s mother reported that OH’s primary care physician diagnosed the patient with benign paroxysmal positional vertigo (BPPV) and prescribed at-home Epely maneuvers. At time of presentation, OH had been consistently performing Epely maneuvers at home for more than a year with no improvement in symptoms.

Are the patient’s reported symptoms consistent with a diagnosis of BPPV?

Past Medical History
The patient’s mother reported that she was hospitalized during pregnancy at 28 weeks gestation and given steroids and multiple medications to stop pre-term labor. OH’s mother remained in the hospital for the last month of pregnancy, ultimately delivering at 37 weeks gestation. OH passed her newborn hearing screening at birth. At 4 years of age, OH was diagnosed with bilateral chronic otitis media treated with pressure-equalization tubes. She later was diagnosed with bilateral hearing loss (FIGURE 1), although her mother believes the hearing loss existed from birth. OH was observed to have noticeable speech production errors. Her mother reported that OH has bilateral hearing aids, although she rarely uses them. The patient presented with visual deficit, requiring the use of glasses since she was a toddler. The mother reported that OH never
crawled and did not take her first steps until 16 months of age. The mother reported that when OH was an infant/toddler she would often vomit with different movements. She stated she thought that it was due to a weak stomach at the time, but as OH became old enough to express her symptoms, headaches always accompanied nausea and dizziness. OH has a brother with normal hearing and no reported developmental delays. Family history is significant for maternal aunt with severe migraines and maternal grandfather with episodic vertigo. The patient’s mother provided an MRI report from the primary care physician indicating normal findings. The ENT ordered a full vestibular evaluation.

Findings
Ocular and cervical VEMP responses were robust and symmetrical, suggesting normal otolith function and intact inferior and superior vestibular nerve function.

Dynamic posturography resulted in falls for eyes-closed sway reference and visual surround and sway reference (FIGURE 2), suggesting a vestibular disturbance pattern. These conditions remove visual and proprioceptive input, forcing the patient to rely on vestibular input alone to maintain balance. Individuals with impaired vestibular systems are left with no reliable input to maintain balance and often display abnormal deviations in balance function.

Rotational chair resulted in phase lead with normal gain and symmetry on sinusoidal harmonic acceleration (SHA) tests. SHA testing consists of rotating the patient in the dark in a sinusoidal motion at frequencies from 0.01 HZ to 0.64 Hz. This activates both the right and left horizontal semicircular canal, which in turn generates the vestibulo-ocular reflex (VOR) response. For this

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<td>WRS at MCL</td>
<td>80% @ 90 dBHL</td>
<td>88% @ 80 dBHL</td>
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<td>Tympanometry</td>
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FIGURE 1. Results from audiometric evaluation revealing bilateral mild- to severe-mixed hearing loss, right poorer than left.

FIGURE 2. Conditions 5 and 6 for dynamic posturography demonstrating how the vestibular system is isolated by removing input from the visual and proprioceptive systems.
test, the gain and the phase of the eye movement are calculated, as well as symmetry between responses for movements to the right versus the left. Gain is the ratio of the amplitude of eye movement to the head movement, where phase represents the timing relationship between head movement and eye movement. Phase leads (velocity of the eye leads the velocity of the head movement) and reductions in gain are most commonly associated with peripheral disorders. Phase lead in the presence of normal gain suggests central compensation has been initiated.

Videonystagmography resulted in normal oculomotor function. No spontaneous nystagmus was observed and Dix Hallpike was unremarkable bilaterally. Horizontal high frequency headshake resulted in down beat nystagmus. Positional testing resulted in a 5 degree/sec right beat nystagmus in the head left position, which suppressed with fixation. Bithermal calorics were robust and symmetric.

Consider the Facts
The patient presents with:
- Episodic vertigo lasting hours, accompanied by nausea, vomiting, and severe headaches
- Familial history of vertigo and migraine
- Report of complications during pregnancy requiring significant medical intervention
- Delayed developmental milestones
- Ongoing challenges with engaging in age-appropriate activities and sports
- Bilateral severe mixed hearing loss
- Vision impairment
- Normal MRI report

Differential Diagnosis
You be the detective. Possible diagnoses could be:
- Benign paroxysmal positional vertigo (BPPV)
- Vestibular migraine
- Benign paroxysmal vertigo (BPV) of childhood
- Congenital
- Central pathology

Discussion
The patient previously was diagnosed with BPPV by her primary care physician. BPPV occurs when the otoconia from the maculae become loose and are free-floating in the semicircular canals. This is typically characterized by brief momentary vertigo when the head is placed into the provoking position. It is not typically associated with headaches, vertigo lasting hours, or nausea/vomiting. Further, the patient had been engaging in therapeutic maneuvers for an extended period of time without any improvement in her symptoms.

The patient reported a long history of severe headaches associated with vertiginous episodes, consistent with vestibular migraine. Vestibular migraine can vary widely.
in presentation, making diagnosis challenging. Consistent with patient report, common complaints include nausea/vomiting during episodes and motion intolerance (Lewis, 1995). Although migraine is not typically associated with hearing loss, episodic vasospasm can lead to damage to the inner ear and subsequent hearing loss.

BPV of childhood is the most common cause of pediatric vestibular dysfunction. BPV of childhood typically presents in early childhood (Russell and Abu-Arafeh, 1999) and is characterized by vertigo, nausea, and vomiting. Children with BPV often report specific triggers including light, sound, or movement. Although similar in name, the presentation and origin of dizziness is quite different from BPPV. BPV of childhood is thought to be of central origin and is commonly used interchangeably with migraine variant vertigo (Goebel, 2001). As such, children with BPV of childhood do not respond to repositioning maneuvers as would be expected in BPPV. BPV of childhood is more commonly associated with brief attacks of vertigo lasting seconds to minutes (although this is often debated), while migraine variant vertigo can last minutes to hours. BPV of childhood is thought to be a precursor to migraine that typically presents in late adolescence or early adulthood (Russell and Abu-Arafeh, 1999; Herraiz et al, 1999).

Congenital abnormalities are present at birth and are either of genetic origin or acquired due to perinatal/birth risk factors. The mother’s report of significant complications during pregnancy is consistent with acquired congenital hearing loss and vestibular impairment, however genetic disorder is also a possibility. When a patient presents with one or more features or symptoms, a genetic syndrome is suggested.

The most common syndrome affecting both hearing and vision is Usher’s syndrome (NIDCD, 2014). Vestibular impairment also may be observed in Usher’s syndrome, although it is less commonly associated. Another possible genetic cause is Pendred syndrome, which causes early onset of bilateral hearing loss and vestibular weakness in children (NIDCD, 2015a). Additionally, children with Pendred syndrome often present with decreased vision and delayed motor milestones, specifically walking (NIDCD, 2015a). A specific feature of Pendred syndrome is enlarged vestibular aqueduct (EVA), which is the likely source of the associated vestibular impairment in this condition (NIDCD, 2015a).

OH’s vestibular evaluation resulted in downbeat nystagmus following horizontal headshake. The headshake test activates the horizontal semicircular canals and therefore should activate the VOR for horizontal nystagmus. The presence of downbeat nystagmus following horizontal headshake is an unexpected abnormality. This can be a result of central impairment in the cerebellum (Kim et al. 2005). This, in addition to her reported severe headaches, may support a central origin of dizziness. This is unlikely, given that the majority of her test results suggest a peripheral impairment and she presented with a normal MRI. Nonetheless, this should be considered as part of the differential diagnosis and definitively ruled out by the managing physician.

The posturography, rotational chair, and positional tests suggest peripheral origin of dizziness, while the downbeat nystagmus following headshake is a central indicator. Based on these findings, combined with the patient’s history and presence of severe bilateral mixed hearing loss, the ENT ordered a CT scan, repeat MRI, and genetic testing.

Diagnosis
And the diagnosis is... bilateral enlarged vestibular aqueduct (see FIGURE 3). Imaging studies clearly demonstrate the presence of enlarged vestibular aqueducts on both sides, with the right side enlarged more than the left. The vestibular aqueduct is a fluid-filled bony canal hosting the endolymphatic sac and duct that leads from the vestibule to the brain (NIDCD, 2015b). Approximately 25 percent of individuals with enlarged vestibular aqueduct have Pendred syndrome (NIDCD, 2015b). Pendred syndrome is still suspected as a likely cause, although it has not been confirmed due to the parents’ decision not to move forward with genetic testing. Migraine variant vertigo is also still being considered as a possible co-morbidity. Migraines are diagnosed based on symptom and are often a diagnosis of exclusion. While her symptoms and presentation may be consistent with migraine, it is hard to say if the patient’s severe headaches are a side effect of her chronic imbalance and repeated vertigo or if they are migrainous in nature. As a result, she is now being co-managed by neurology in an effort to get her vertiginous episodes under control.

The Challenges
This case represents the challenges that are often faced when diagnosing children with hearing loss and/or balance disorder. OH presented with classic symptoms early on, only to have her hearing loss confirmed at age 5 and her balance impairment at age 8. Vestibular disorders are arguably even more difficult to identify in children than hearing loss and often go under-diagnosed. Parents and physicians may deem children to be “clumsy” or “just taking their time”
when delays in crawling or walking are observed. Additionally, infants and toddlers with vestibular impairment typically catch up in motor development, resulting in further delays in diagnosis.

Of interest, OH continued to present with atypical motor function, such as the inability to swing or swim and her dislike of running-based sports. As practitioners who evaluate and manage hearing and balance disorder, it is even more critical that audiologists are alert and responsive to the signs and symptoms that may indicate vestibular dysfunction. Key indicators include delays in motor milestones, motion intolerance, significant decrease in balance in the dark, “clumsiness,” or intolerance or unwillingness to engage in quick movements. Some children will display an inordinate fear of the dark or sudden movements. It should also be noted that children with hearing loss are more likely to present with vestibular impairment (NIDCD, 2015b). Management of enlarged vestibular aqueduct often includes safety precautions to reduce chances of head trauma, as even mild head trauma has been associated with changes in hearing and vestibular function in individuals with EVA (Oh et al, 2001). Such a precaution can be challenging in the pediatric population, and patients and their parents are often subject to unexpected changes in quality of life. The presented patient enjoyed outdoor activities and gymnastics. Her family was seeking a treatment that would allow her to return to these activities and improve her ability to enjoy other activities, such as swimming and running. The suggestion to limit or reduce such activities has been difficult for the entire family.

In short, identifying the correct diagnosis early is essential, particularly in the pediatric population. This allows for implementation of the appropriate treatment plan, and also can provide the patient and the family with realistic expectations and the ability to establish a plan for the future.

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References


