Mirror Image Twins with Congenital Venous Malformations and Associated Vestibulocochlear Dysfunction

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Abstract

Objective: To demonstrate the effects of a congenital venous malformations on hearing and balance through a case study of mirror twins.

Methods: A retrospective chart review of twin sisters who presented to a tertiary children’s hospital with hearing loss and vertigo was performed. Imaging and audiometry results were compared between patients.

Results: Our first patient was an 11 year old female who presented with complaints of chronic hearing loss, dizziness and migraine headaches. On audiometry she was found to have unilateral mild sloping to severe mixed hearing loss on the left. Imaging with CT, MRA, and angiogram demonstrated aplasia of the right sigmoid sinus. The left sigmoid sinus and jugular bulb were greatly enlarged and had expansile erosion into the IAC, basal turn of the cochlea, inferior limbus of the posterior canal, and the vestibular aqueduct.

Her twin sister presented with complaints of chronic hearing loss and episodic vertigo. Audiometry showed unilateral mild sloping to severe mixed hearing loss on the right. A CT of the temporal bones demonstrated dysplasia of the cochlea bilaterally and an enlarged right jugular bulb that was dehiscent into the area of the vestibular aqueduct.

Conclusion: Vascular malformations, such as high rising jugular bulb, have been described in the setting of vestibulocochlear dysfunction. However, these cases are unusual in that they are uncommon examples of vascular malformations, and are found on contralateral sides in a set of identical twins.

Introduction

The jugular bulb is a dilated segment of vein where the sigmoid sinus within the temporal bone takes a relatively sharp turn and travels through the jugular foramen to become the internal jugular vein. It is a dynamic structure that forms after 2 years age and stabilizes in adulthood with the precise location and size varying considerably within the general population. The etiology of these variations is poorly understood and is likely multi-factorial with contribution related to variations in blood flow. With the proximity of these structures to the middle ear, various pathologies have been described in cases of mal-placed jugular bulbs.

A high-riding jugular bulb (HRJB) is the most common vascular anomaly of this portion of the temporal bone. The inappropriately high position results in subsequent thinning of the bony canal which separates the vein from the mesotympanum. This can result in dehiscence of the jugular bulb and disruption of adjacent vestibulocochlear structures with anticipated symptoms such as hearing loss, imbalance, vertigo, facial paralysis, and pulsatile tinnitus. It is estimated that nearly half of all jugular bulb abnormalities are clinically silent making it difficult to estimate prevalence. However, Friedman et al, have performed radiographic and histopathologic evaluations of 1579 temporal bones which found HRJB in 8.2%-8.5% of specimens.

Mirror-image morphological discordance between monogyzotic twins is a well described phenomenon present in up to 25% of monogyzotic twins and associated with a wide variety of pathologies. To our knowledge, we present here the first case of mirror-image HRJB in a pair of monogyzotic twins.

Case Report

Our patients are twin sisters, born via C-section at 36 weeks for fetal distress. They did not require an ICU stay and apparently passed newborn hearing screenings. The first sister was referred for evaluation at age 10 for evaluation of hearing loss, dizziness, and headaches. She suffered from recurrent vertigo attacks from the time she was 15 months. Her symptoms included throbbing pain exacerbated by noise and light and associated with nausea and vomiting, alleviated by sleeping in a dark room. These episodes happen 3 to 4 times per year. Her hearing loss has been present since the age of 6, but the patient had good speech and school performance so no intervention had been undertaken up to that point.

Her otologic exam was normal. Her audiogram showed a moderate left sided mixed hearing loss. Her Jerger type C tympanogram. A CT scan was performed, which showed an expansile process in the left temporal bone involving the region of the jugular foramen and sigmoid sinus with effacement of the posterior wall of the IAC and erosion into the vestibulocochlear aqueduct. MRI and MRA showed the patient to have an arteritic right transverse and sigmoid sinus, with associated aneurysmal expansion of the left jugular bulb to erode into the IAC and basal turn of the cochlea. See Figure 1 for images.

The second twin also had similar, but milder, symptoms. However, her hearing loss was on the right side. Her otologic exam was normal. Her audiogram shows mild to moderately severe SNHL on the right with normal OAEs and absent acoustic reflexes for the right ear. The left ear had normal thresholds, OAEs, and reflexes. A CT was obtained of the temporal bones that showed enlarged right vestibulocochlear aqueduct that was at least partially compressed by an enlarged jugular bulb. The left sided vestibulocochlear aqueduct and jugular bulb appeared normal. See Figure 2.

Both patients were fitted with hearing aids for their affected ears, and treated conservatively for their episodic vertigo symptoms. Both are doing well, with stable hearing and their vestibular symptoms have lessened with time.

Discussion

High-riding jugular bulb occurs in 8.2-8.5% of the population with varying degrees of clinical severity. The precise mechanism is suspected to be a combination of genetic and environmental forces. We present the first case of mirror image HRJBs associated with contralateral aplasia/hypoplasia of the sigmoid sinus in monogyzotic twins. Only one other case with the same pathology has been reported in a single patient.¹¹

The phenomenon of mirror image twinning clearly has a strong genetic component. Hypotheses have been developed that suggest cleavage of the late blastostat may play a role.¹² The incidence of mirror-image features in monogyzotic twins is as much as 25% and many case reports exist that describe various phenotypes.¹³ Some have even found evidence for the mirror-imaging to expand beyond physical dysmorphia to affect the entire biopsychosocial model.¹⁴

In the current case report, the patients were each found to have a HRJB that was dehiscent into the middle ear and temporal bone resulting in vestibulocochlear dysfunction that would be anticipated from such a pathology. However, the severity of the jugular bulb dehiscence and contralateral malformation of the sigmoid sinus varied between twins. One twin had a complete aplasia of the right transverse and sigmoid sinus, which resulted in the dilation and high-riding position of the left jugular bulb. In the other twin, the right HRJB was present with a normal left sigmoid sinus and jugular bulb. These variations suggest another force upon the development of these structures beyond purely genetic influence.

The jugular bulb develops after two years of age and remains dynamic until adulthood when it stabilizes. Flow dynamics show a right sided flow predominance of the major cerebral venous structures in 70% to 80% of patients with concordant size discrepancies.² Postural changes and resultant venous pulsations have been credited with the development of the jugular bulb beginning around two years old. Such environmental effects, if different between patients, could explain the variation in size of jugular bulb and contralateral sigmoid sinus in the presence of genetically determined HRJB.

References


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Figure 1: Photo documentation from Twin A. Audigram showing left sided moderate hearing loss (a). MRA showing the absence of right sided transverse and sigmoid sinuses (b). A CT temporal bone showing the dilated jugular bulb on the left side ending into the otic capsule (c and d).

Figure 2: Photo documentation from twin B. Audigram showing right sided moderate hearing loss (a). CT scan showing a high rising jugular bulb on the right that is partially effacing the vestibular aqueduct (b).

Figure 2: Photo documentation from twin B.