Is it a Chiari or Not? The Difficult Road to Diagnosis

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Chiari I Malformation, primarily a congenital condition, is described as cerebellar tonsillar ectopia below the foramen magnum that is commonly associated with a syringomyelia, a fluid-filled cavity in the spinal cord.[2,3,5,6,7] The cerebellar ectopia occurs due to poor development of the posterior cranial fossa, and the syringomyelia occurs due to CSF flow being affected.[2,3,5,6] Although a pediatric condition, symptoms usually arise during adulthood.[5,6,7] Presentation to the ED may revolve around cerebellar dysfunction, such as truncal ataxia and dizziness, tinnitus, severe suboccipital headaches, and neck pain.[2,5,6,7] Patients may describe the occipital headaches to be orthostatic in nature that worsen upon standing. The headaches and neck pain may also worsen with straining, such as during bowel movements, or coughing, indicating that the CSF flow has been compromised.[2,5,6] Other symptoms may be secondary to brainstem compression from the cerebellar tonsillar ectopia.[2,3,5] These symptoms include dysphagia and uvular deviation from cranial nerve compression, and autonomic symptoms secondary to brainstem involvement, including palpitations and chest pain. [2,3,5] Symptoms may arise secondary to spinal cord compression from the syringomyelia.[2,3,5,7] These symptoms include numbness/tingling in extremities, pain in the arm, sudden weakness resulting in drop attacks, and temperature and pain sensation loss in the upper extremities secondary to the spinothalamic tract being affected.[2,3,5,7] Presentations to the ED may include any range of these symptoms.[2,3,4,5]

In one case report of a young healthy patient with past medical history significant for asthma, the patient presented with symptoms of tinnitus and tachycardia with shortness of breath. The patient described the tinnitus as deafening in quality especially after exertion, even with mild activity such as climbing stairs. Patient was tachycardic at baseline with maximum variation in heart rate to the 190's. Other symptoms included dysphagia, mostly with liquids, uvula deviation, drop attacks, and mild scoliosis. The patient denied symptoms of ataxia. Initial management was in consultation with Cardiology and Pulmonary, which resulted in a new medication regimen to better control asthma, since it was presumed this may be an underlying factor, and a temporary diagnosis of POTS. Following, an ENT workup was benign. Due to the pulsatile nature of the tinnitus with a deafening quality upon exertion, MRI workup was initiated, which revealed the diagnosis of a Chiari I Malformation.

Because initial symptoms may be vague with no neurologic deficits, presentations to the ED may differ.[2,3,4,5] For example, patients may primarily present with a chief complaint of chest pain that occurs with exertion but resolves with rest. [3] Although other conditions such as costochondritis may be higher on the differential due to a normal EKG and normal lab values, chest pain is also a symptom seen in Chiari patients, indicating the importance of considering non-cardiac etiologies to chest pain when in the ED.[3] In addition, another presentation may primarily revolve around scoliosis—past research has found that scoliosis has been a common finding in Chiari patients, mostly in those with a syringomyelia.[2,5,7] This is a key finding that points to a potential Chiari diagnosis and necessitates workup, especially when patients do not present with any neurologic deficits.[2,7]

Past research has also shown that symptom type may differ depending on the age at which the patient presents at.[5] This adds another layer of complexity to diagnosis, in that not only can patients present with different kinds of symptoms, but their age can affect which symptom they present with.[2,3,4,5] This can be explained by the idea that different parts of the brain can be compressed by the cerebellar ectopia, such as the cerebellum, brainstem, and cranial nerves, causing different symptoms to arise depending on which location is being affected.[5] In addition, as there is continued compression against the cerebellum and brainstem, eventually the CSF flow will be blocked, forming a syringomyelia which results in new symptoms secondary to spinal cord compression.[5] Therefore, over time, new symptoms will arise as the condition progresses, explaining why patients present with different symptoms at different ages.[5]

Presentation to the ED does not always result in surgery for these patients. Many patients may be asymptomatic.[2] Usually, patients suffering from detrimental symptoms, having a significant impact on their quality of life, will undergo surgery. Surgery may also be performed to prevent further neurologic damage.[2,3] Also, patients with an associated finding of a syringomyelia will likely undergo surgery.[2,3] Post-surgery, presentation to the ED may revolve around worsening headache and incisional drainage, worrisome for a CSF leak.[1]

Patients with Chiari malformation may present to the ED with a variety of symptoms, either neurologic or non-neurologic in nature, presenting a special challenge in correctly lining up the symptoms to the diagnosis.[2,3,4,5] One must remain vigilant as early identification and diagnosis will allow for proper intervention and prevention of disease progression.[2,3,5]