Chiari Malformation

Chiari malformations are characterized by the herniation of the cerebellum into an opening at the base of the skull known as the foramen magnum (1,2,3,4,5,6,7,8). There are three types of malformations each based on the degree of herniation; however, the severity of symptoms is not necessarily dependent on the degree of herniation (8). Chiari I (CM1) is the most common type and is considered a congenital malformation, though cases acquired through injury have been discussed in the literature as well (2,3,4). CM1 is defined by the cerebellar tonsils extending past the foramen magnum by at least 3-5mm (8). CM2 is the most common type of CM, also known as classical CM, and involves the herniation of the cerebellum and brain tissue into the foramen magnum. CM3 is the most serious type of CM and involves the herniation of the cerebellum and brain tissue into the brain stem. CM2 and CM3 are obvious malformations that are identified at birth, while CM1 is typically not diagnosed until the teenage or adult years, if at all (3).

Unfortunately little is known about the epidemiology and natural history of CM but some research suggests that there may be a familial component (1,4). CM’s can develop when the bony space at the back of the skull, known as the posterior fossa, is smaller than normal, therefore, causing the cerebellum or both the cerebellum and brain stem to protrude into the spinal column (3). As such, the functions controlled by these areas may be impacted. Additionally, CM’s can result in the blockage of cerebral spinal fluid (CSF), which surrounds and cushions the brain, and has the potential to lead to hydrocephalus and/or syringomyelia depending on the severity of the blockage (6). Hydrocephalus occurs when there is a build-up of CSF in the brain which causes the ventricles to expand, often resulting in significant mental changes (1, 2). Syringomyelia occurs when CSF builds up in the spinal column and results in a cyst (1, 2). Adults are reportedly more likely to experience syringomyelia than children (1,2).

The prevalence rate of CM1 is estimated to be about 1%, although this number varies in the literature, and is three times higher in women than men (2). Many individuals with CM1 are asymptomatic and do not seek treatment, therefore, the actual number of cases is unknown. The increased use of diagnostic imaging, specifically magnetic resonance imaging (MRI), has led to greater identification of CM1 in individuals (7). In asymptomatic individuals, CM1 is often discovered as an incidental finding while undergoing treatment for an unrelated issue. One study found 1/3 of their sample to be asymptomatic while the majority of symptomatic patients complained of headaches (7).

Symptoms of CM1 are caused by a disruption in the flow of CSF and by the pressure exerted on the central nervous system tissue (6). Head pain of some kind is often the first presenting symptom (4,8). The clinical presentation of CM1 typically involves headaches (often made worse by coughing), neck pain, balance problems, muscle weakness, numbness, dizziness, vision problems, sleep problems, difficulty swallowing, lower cranial nerve signs, etc (3, 4, 6, 8). There is almost no report of cognitive changes or psychiatric symptoms related to CM1 in the literature, although depression may result from some of the other physical symptoms experienced. Symptoms vary dependent upon age as well. Aitkin et al., (2009) found that older age at diagnosis predicted worse neurological symptoms. Although some individuals may be asymptomatic earlier in life, they do have the potential to develop symptoms later on (3). The symptoms of CM1 are often complex and unfortunately often result in misdiagnosis (3).

Treatment of CM1 depends on the severity of symptoms (1). Monitoring and observation is typically the first line of treatment for

those with mild to no symptoms (4). Columbia University Medical Center Department of Neurological Surgery instructs parents to watch for any changes in their child’s neurological status including: breathing problems, degree of alertness, speech, feeding problems, problems walking, or uncoordinated movement. For individuals who experience pain related symptoms, medication management would typically be the first step. When the pain is severe or when syringomyelia or hydrocephalus is present, surgery may be the best treatment option. There are different surgery options including a bony decompression of the posterior fossa or the placement of a shunt (6). Overall specific treatment and surgery options will be determined by your child’s physician based on a number of factors including age, overall health, medical history, type and severity of the condition, etc. (8).

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