NINDS CDE Project

Working group: Co-morbidities (Chiari I malformation)

Clinical Summary:

Chiari I malformation is in need of better clinical understanding, because its identification can be elusive and its outcomes variable and frequently unsatisfactory for many patients. Improved preoperative understanding and identification of underlying pathophysiologic mechanisms of cerebellar tonsillar descent should lead to better treatment outcomes and lower complication rates for this condition. The unmet needs, unanswered questions, and suggested approaches to improve care and understanding of Chiari I malformation will be advanced by such approaches. Broadening our approach to include the mechanisms of cerebellar tonsillar herniation will allow a better definition of the clinical subtypes of Chiari I malformation and their response to various methods of potential medical and surgical management (Milhorat et al., 1999).

As a result of our collaboration with many clinical sites specializing in the management of patients with Chiari I malformation, it became increasingly clear that the phenotype of cerebellar tonsillar herniation arose from a variety of other primary and co-morbid conditions of the brain, skull, spine, and connective tissue through a variety of pathophysiological mechanisms. In response to these observations, there has been increased focus on defining these other primary and co-morbid conditions. One pathophysiological group included hydrocephalus, pseudotumor, Paget's disease, arachnoid cysts, and other conditions that produced downward pressure upon the posterior fossa structures. Another group comprising about 10% of our patient population had hypermobile connective tissue disorders leading to cranio-cervical instability associated with less dramatic tonsillar descent, but very similar symptomatic complaints. These conditions could also produce symptoms independent of cerebellar tonsillar descent associated with spinal instability and poor wound healing. Cerebellar tonsillar descent was also appreciated in a subset of patients with tethered cord syndrome, a related neural tube closure deformity, leading to downward traction and tonsillar descent as well as syrinx formation. Independent observations of cerebellar tonsillar descent related to syndromes of cerebrospinal fluid (CSF) leakage or lumbar CSF shunt drainage uncovered the potential of an underlying draw or suck down mechanism. It became increasingly clear that the outcome of surgical treatment of Chiari I malformation depended upon which comorbid or potentially other primary pathophysiologic mechanism might underlie discovery of tonsillar herniation in any specific patient. The presence of such comorbid conditions could therefore influence and complicate the ultimate surgical results of Chiari I malformation treatment. Conversely, an understanding and exploration of such conditions would be necessary to inform appropriate medical and surgical treatment decisions. The appreciation of comorbid conditions and mechanisms of cerebellar tonsillar herniation led to an approach to Chiari I malformation

treatment that was tailored to underlying conditions and disease mechanisms (Milhorat et al., 2010). We believe that Common Data Elements (CDEs) that record associated co-morbidities and explore potential pathophysiologic mechanisms provide a unique and essential source of clinical information that defines the clinical syndromes of tonsillar descent and will lead to better comparisons of treatment outcomes among subgroups of patients with Chiari I malformation and other causes of cerebellar tonsillar herniation.

These co-morbidity CDEs were chosen to ferret out some of the possible underlying processes responsible for cerebellar tonsillar descent which must be taken into account when assessing the clinical presentation, natural history, and outcome of different types of medical and surgical management of cerebellar tonsillar herniation. They include relevant symptoms and comorbid conditions that differentiate the clinical subtypes of Chiari I malformation.

Chiari I malformation is most commonly diagnosed by the finding on magnetic resonance imaging (MRI) of cerebellar tonsils located below their normal position, with their tips at least 5 mm or more inferior to the foramen magnum. Symptoms of Chiari I malformation may occur in patients with tonsils positioned less than 5 mm below the foramen magnum, especially if there is associated narrowing of CSF pathways at the foramen magnum. The proportion of patients showing characteristic Chiari I malformation symptoms grows as tonsil position below the foramen magnum increases beyond 5 mm, supporting definition of the biologic deformity using the 5 mm threshold. Although this level of tonsillar position may represent perhaps the most prevalent and commonly appreciated, it accounts for only one of several underlying anatomical abnormalities within the clinical entity commonly referred to as Chiari I malformation. Some investigators contend that narrowing or obliteration of CSF pathways at the foramen magnum is more closely related to symptom production than the level of the tonsils below the foramen magnum. Continuing clinical research is needed to develop diagnostic MRI criteria that better predict the development of typical signs and symptoms of Chiari I malformation than present MRI diagnostic guidelines of the disease.

Milhorat TH, Nishikawa M, Kula RW, Dlugacz YD. Mechanisms of cerebellar tonsil herniation in patients with Chiari malformations as guide to clinical management. Acta Neurochir (Wien). 2010;152(7):1117–1127.

Milhorat TH, Chou MW, Trinidad EM, Kula RW, Mandell M, Wolpert C, Speer MC. Chiari I malformation redefined: clinical and radiographic findings for 364 symptomatic patients. Neurosurgery. 1999;44(5):1005–1017.