

## Presentation, Natural History, Signs and Symptoms: NINDS CDE Project

**Working group:** Presentation, Natural History, Signs and Symptoms

The Common Data Elements (CDEs) covering the clinical presentation of patients with Chiari I malformation are based on the classical symptoms described in patients with the established diagnosis of Chiari I malformation as documented in the medical literature over the course of many years. In particular, this includes headache, symptoms of imbalance, and visual symptoms. When syringomyelia is associated with Chiari I malformation, symptoms of altered sensation in limbs or trunk, as well as weakness of one or more limbs, may be among the presenting complaints. Many of these symptoms are at first glance quite general, as for example, headache. The CDEs aim to define these symptoms more specifically. In the example of headache, the CDE would specify onset, frequency, duration, location and other factors. Reliable quantification of symptoms such as headache is generally not available.

The natural history of Chiari I malformation is not clearly described inasmuch as many, if not most, symptomatic patients undergo treatment once the diagnosis is confirmed by imaging. It is one of the aims of this project to describe the natural history of individuals with tonsillar descent who, for various reasons, are not treated surgically, but are observed with or without medical therapy.

CDEs detailing the findings on examination cover the standard neurological examination of cranial nerves, motor and sensory systems, with an emphasis on those modalities likely to show abnormalities in Chiari I malformation patients, with or without syringomyelia. This includes manifestations of increased intracranial pressure, brainstem dysfunction (e.g., nystagmus, imbalance), weakness of limbs, abnormalities of reflexes and sensory impairment. It is recognized that absolute quantification of findings such as limb strength and limb tone is not possible, but reasonable quantification has been performed over the course of years for limb strength (Compston, 2010) and limb tone (Bohannon and Smith, 1987). These have been used for many years and therefore have the benefit of validation by common acceptance and usage.

Recognized co-morbidities, such as hereditary connective tissue disorders and tethered cord and pseudotumor cerebri, occur in a relatively small percentage of the overall Chiari I malformation population. The symptoms and findings associated with these conditions are broken down and detailed separately under the section co-morbidities.

Bohannon RW, Smith MB. Interrater reliability of a modified Ashworth scale of muscle spasticity. *Physical Ther.* 1987;67:206–207.

Compston A. Aids to the investigation of peripheral nerve injuries. Medical Research Council: Nerve Injuries Research Committee. His Majesty's Stationery Office: 1942; pp. 48 (iii) and 74 figures and 7 diagrams; with aids to the examination of the peripheral nervous system. By Michael O'Brien for the Guarantors of Brain. Saunders Elsevier: 2010; pp. [8] 64 and 94 Figures. *Brain.* 2010;133(10):2838–2844.