Natural history of the Chiari Type I anomaly

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In the study by Novegno and colleagues, the authors discuss the cases of 22 patients with MR imaging evidence of a Chiari malformation Type I (CM-I) who were selected for observation rather than surgery because the abnormality was found either incidentally or because the patient’s symptoms were sufficiently mild that surgical intervention was not pursued. During the course of their study, the authors evaluated a total of 94 patients, with the remaining 72 patients requiring surgery of one kind or another at the time of the initial diagnosis. The asymptomatic or minimally symptomatic patients were followed up for 3–19 years. Interestingly only 1 patient required surgery directed at the Chiari malformation, and only 5 patients later developed symptoms or had progressive symptoms. This work underscores 2 very important issues related to surgical decision making and our understanding of this protean condition and its pathophysiology. The first issue relates to the critical question of if and when to conduct an operation in a child with CM-I who is asymptomatic or manifesting only mild symptoms. The second question relates to nomenclature: what is actually meant by the term “Chiari I malformation?” In reading the literature does each discussion relate to the same condition? Is there a recognized definition of a CM-I? Should we use a different nomenclature?

There are multiple questions involved in making decisions regarding surgical treatment of any potentially surgical disorder. Surgeons are often faced with the need to decide with incomplete information. Sometimes the progression rate of the condition is such that a decision is necessary without further information. The results of the study by Novegno and colleagues should be interpreted to mean that in the absence of rapid clinical deterioration or lower cranial nerve dysfunction, herniation of the cerebellar tonsils below the foramen magnum can be asymptomatic, nonprogressive, or can even improve, as seen in 5 of their patients. Clinical deterioration or increasingly severe symptoms may arise, but it is more likely that the deterioration will be insidious, and careful follow-up and treatment at the time of progression can reverse it. In these situations, in the absence of compelling symptoms the proper approach should be that of “watchful waiting.”

Probably the most important piece of information required for surgical decision making is a risk–benefit analysis of the procedure. Symptoms related to the CM-I can be quite subtle and require a careful history and thorough assessment of the physical examination and imaging studies. Sometimes the problems caused by this condition cannot be assessed until after the surgical procedure is done. In the absence of lower cranial nerve deficits or syringomyelia, rapid deterioration and permanent neurological injury is unlikely. On the other hand, the surgery for this condition is low risk, and in terms of reestablishing normal dynamics at the craniovertebral junction, the likelihood of success is high. Although as many as 25% of patients with minimal symptoms may experience spontaneous improvement or resolution of the Chiari malformation, surgery is a reasonable option for those in whom symptoms such as suboccipital headaches, cough, headaches, and oral defensive-ness have an effect on quality of life.

An even more interesting aspect of this study relates to the definitions of Chiari anomaly, Chiari malformation, and hindbrain herniation. The title of the authors’ work relates to something called a “Chiari anomaly.” Within the text the authors use this term interchangeably with the more commonly used “Chiari malformation.” These terms may or may not be synonymous, but what is clear is that there is no accepted definition of CM-I, which makes both discussion of the condition and making surgical decisions very difficult. The use of the term “anomaly” may be unique. In a PubMed literature search for the keywords “Chiari” and “anomaly,” I found only 1 reference to a Chiari anomaly, and this related to the anatomical relationship between the right atrium of the heart and the superior vena cava.1 To me this terminology is new but very logical and it may be that it should be generally adopted. The Merriam-Webster dictionary defines the word “malformation” as “irregular, anomalous, abnormal or faulty formation.” This definition implies that the abnormality began before the full development of whatever is abnormal. Using this definition, an acquired malformation would be an oxymoron. The definition of “anomaly” is “something different, abnormal, peculiar or not easily classified.” What is now called the Chiari malformation Type I—involving descent of the cerebellar tonsils 5 mm below the foramen magnum with some distortion of these tonsils
and a wide variety of associated conditions and pathophysiological characteristics—is more appropriately termed an “anomaly” than a malformation because many of these problems arise after the completion of the posterior fossa development, as described by the authors of this study.

Finally, a third term that has been used in this context is “hindbrain herniation.” Hindbrain herniation is a more general name that is useful in describing the presence of the cerebellar tonsils (in CM-I) or brainstem and cerebellar vermis in the context of spina bifida and Chiari malformation Type II.2,4 The word “hernia” is defined as “a protrusion of an organ or part.” This term is useful in that it can be applied to the condition when it is associated with mass lesions, as well as in other conditions that create a pressure differential between the spinal subarachnoid space and the intracranial compartment (for example, in the context of lumboperitoneal shunts). These forms have been discussed in terms of acquired malformations as well as reducible Chiari malformations. The use of the name “hindbrain hernia” in this context would seem most appropriate.

I became acutely aware of the problem of the definition of CM-I in a report on the correlation of hindbrain herniation in patients with craniofacial syndromes.3 The text required several revisions due to the lack of agreement among reviewers concerning the nomenclature. We have not come a great deal further since that time. Hopefully this discussion will lead to a concerted attempt to create a consensus as to what to call abnormalities of cerebellar descent so that we can all speak of the same conditions.

References

Response

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We are grateful to the reviewer because his editorial emphasizes 2 points, one of which is obviously declared in our paper, and the second is only implied and not brought explicitly to the readers’ attention.

The first point relates directly to the results of our study, that is the unpredictable course of events in children with low-lying cerebellar tonsils in the absence of manifest clinical symptoms or signs and without ventricular dilation.

In our experience, the clinical condition in the majority of these patients tends to remain stable, challenging the usefulness of a surgical correction based on the mere neuroradiological evidence of the phenomenon. Should the results of our study be confirmed in other studies, the second point—namely the need to agree on an appropriate terminology to describe the low-lying cerebellar tonsils in these particular circumstances—would require a focused discussion. The discussion would not be merely semantic as the appropriate terminology should be aimed at differentiating patients who require only careful clinical and radiological observation, from those who would benefit from surgery.

In this direction, we introduced the term “Chiari anomaly” because the most used definition of “Chiari type I malformation” or “tonsillar herniation” seemed to us to carry a pathogenic connotation, which did not apply to our patients. The term “anomaly” could better describe a variant of the normal anatomy that does not necessarily play a pathogenic role. We agree with Dr. Rekate’s suggestion that the phenomenon, generically referred to as “Chiari type I malformation,” might have diverse clinical implications, ranging from the probably innocuous anomaly, which requires only a wait-and-see policy, to the acquired and often progressive cerebellar tonsil herniations that develop in infants with Crouzon syndrome or in some children with hydrocephalus. In comparison to those with the Chiari anomaly we have described, these children often require timely surgical correction and the placement of an extrathecal cerebrospinal fluid shunt. (DOI: 10.3171/PED/2008/2/9/177)