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Chiari Malformations and Syringohydromyelia in Children

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Chiari malformations are a heterogeneous group of disorders with distinct clinical anatomical features all of which involve the hindbrain. Our understanding of Chiari malformations increased tremendously over the past decades, and progress in neuroimaging was instrumental for that. Conventional and advanced neuroimaging of the brain and spine play a key role in the workup of children with suspected Chiari malformations. In addition, neuroimaging studies in Chiari malformations may guide the management, serve as a predictor of outcome, and shed light on the pathogenesis.
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Introduction

In 1896, Hans Chiari,[1] an Austrian pathologist who worked at the universities of Prague and Strasbourg, described 3 different morphologic abnormalities of the posterior fossa characterized by downward herniation of part of the cerebellum and the brainstem into and through the foramen magnum. To date, these 3 forms of hindbrain herniation are known as the Chiari malformations types 1-3.[2,3] Since the original description and the advent of neuroimaging, particularly magnetic resonance imaging (MRI), the diagnosis of Chiari malformations has become "easy" and our understanding of their embryology and pathophysiology improved. In addition, other types of Chiari malformations (types 0, 1.5, and 4) have been described.[3-5] However, many gaps still exist in our knowledge of the Chiari malformations. The increasing knowledge and experience revealed that the classification proposed by Chiari does not represent a continuum of the same disease but includes different entities. In addition, the same type of Chiari malformation (particularly Chiari type 1 malformation [C1M]) represents a heterogeneous group of diseases or etiologies that share a neuroimaging finding on a single midline sagittal MR image.[6] More and more children are being diagnosed with C1M, including an increasing number of children without appropriate symptoms for the diagnosis.[7] The increasing number of children in which C1M is diagnosed raises questions about the management and indications for surgical intervention.

In this review article, we discuss the various Chiari malformations focusing on the conventional (anatomical) and advanced (functional) MRI findings. We also include embryologic, pathophysiologic, and clinical information that are essential for the understanding and management of Chiari malformations.

Chiari Malformation Type 0

Chiari malformation type 0 (C0M) is defined by (1) presence of syringohydromyelia that resolves after posterior fossa decompression and (2) absence of tonsillar ectopia.[5] The term C0M was coined because syringohydromyelia may be due to an alteration of cerebrospinal fluid (CSF) flow dynamics at the level of the craniocervical junction as for other Chiari malformations. C0M and idiopathic cervical syringohydromyelia refer to the same pathology. We prefer the second term because it better describes the underlying anatomical problem.

An isolated syringohydromyelia may have numerous other causes including infections (any kind of meningitis may cause arachnoid scarring and syringohydromyelia), inflammatory diseases (eg, transverse myelitis, sarcoidosis, and multiple sclerosis), traumatic injuries, spinal cord tumors (particularly

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ependymomas and hemangioblastomas), extramedullary tumors and arachnoid cysts, and spinal canal stenosis.[8] It is important to exclude secondary causes of syringohydromyelia before making the diagnosis of idiopathic syringohydromyelia.

Chiari Malformation Type 1

C1M is defined by caudal displacement of one of the cerebellar tonsils (>5 mm) or both tonsils (3-5 mm) below the virtual line connecting the basion with the opisthion of the foramen of magnum on conventional sagittal brain or cervical spine MRI (Fig. 1).[9-11] The prevalence of C1M in children is approximately 1% and is not gender dependent.[7]

Pathogenesis

The pathophysiology of C1M is complex and heterogeneous and our knowledge is based on anatomical observations obtained using neuroimaging techniques as well as prospective presurgical and postsurgical physiological studies. The pathogenesis of C1M can be classified into 4 main general pathomechanisms: (1) overcrowding caused by underdevelopment of the posterior fossa and skull base bony structures, (2) CSF hydrodynamic disorders, (3) excess tissue in the posterior fossa, and (4) downward movement or displacement of the central nervous system (CNS) by events that lower intrathecal pressure.[12] Pathomechanism 1 is the primary cause of C1M, whereas pathomechanisms 2-4 result in secondary herniation of the cerebellar tonsils (for pathomechanisms 2-4, the term C1M is not accurate, and secondary tonsillar herniation reflects better the underlying pathomechanism).

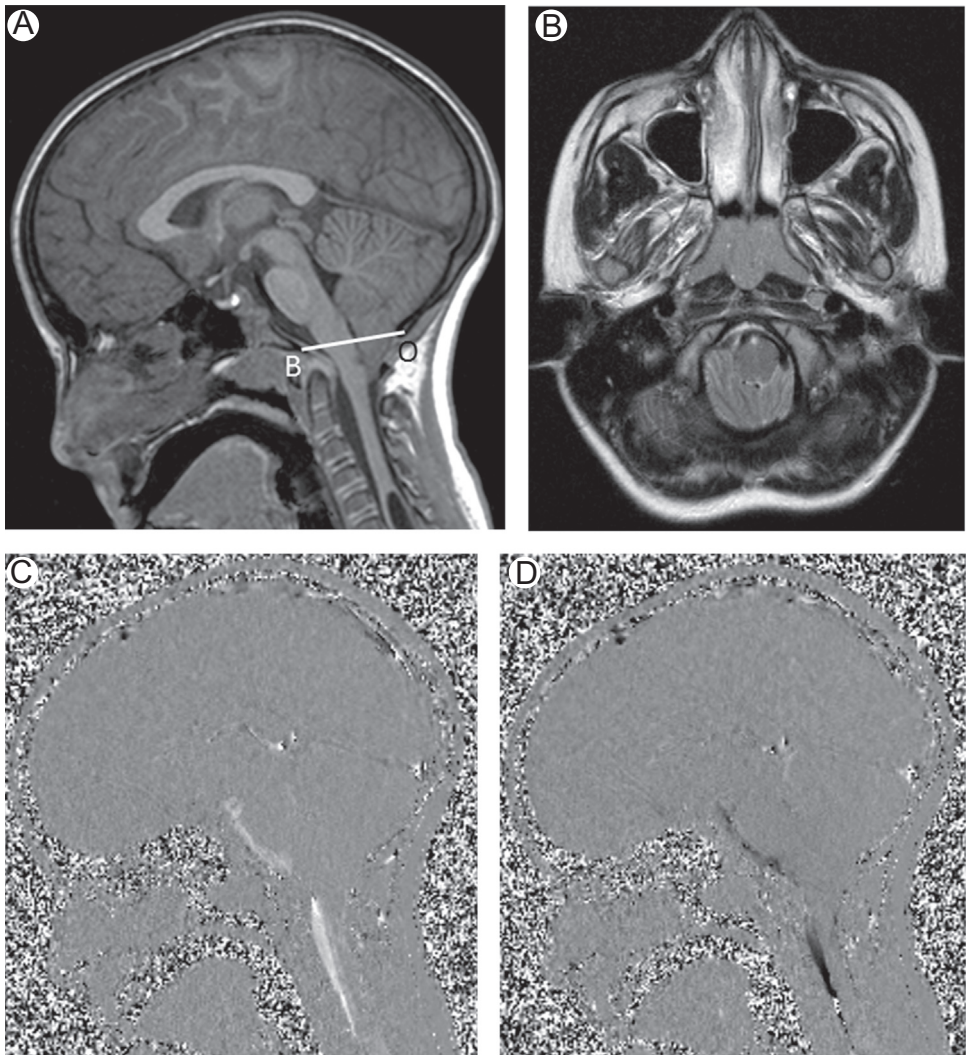

Figure 1 Chiari type 1 malformation in a 9-year-old girl. (A) A midsagittal T1-weighted MR image shows deformed, elongated, and compressed (pointed) cerebellar tonsils that extend more than 5 mm below a line connecting the basion (B) and opisthion (O). No subarachnoid space is seen around the tonsils. A hydromyelia of the cervical spinal cord is also noted. (B) An axial T2-weighted MR image at the level of the crowded foramen magnum reveals the lower medulla being compressed by the low-lying cerebellar tonsils. (C and D) Midsagittal images from cine phase contrast flow study show reduced superior-to-inferior (C, bright) and inferior-to-superior (D, dark) flow anterior to the foramen magnum and complete lack of flow posterior to the spinal cord at the level of the foramen magnum.