

Definitions

cerebellar tonsils - portion of the cerebellum located at the bottom, so named because of their shape

cerebellum - part of the brain located at the bottom of the skull, near the opening to the spinal area; important for muscle control, movement, and balance

cerebrospinal fluid (CSF) - clear liquid which surrounds, and protects, the brain and spinal cord

Chiari malformation - condition where the cerebellar tonsils are displaced out of the skull area into the spinal area, causing compression of brain tissue and disruption of CSF flow

magnetic resonance imaging (MRI) - diagnostic test which uses a large magnet to create images of internal body parts

posterior fossa - depression on the inside of the back of the skull, near the base, where the cerebellum is normally situated

Chiari In The Family

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Introduction: Evidence continues to mount that the existence of Chiari in certain families is not due to chance, but rather to a genetic component. This report describes three families with multiple members who have Chiari. However, what percent of Chiari cases are genetic, and the type of transmissibility is not yet known.

Family 1: A 19-month old boy was evaluated by neurology for potential cerebral palsy. The mother did not think he sat correctly and he walked late with his feet turned out. The doctors noted he had an unusually large head with wide set eyes, but the neurological exam was mostly normal. The mother stated she had a mild case of cerebral palsy, but had been suffering from headaches and back pain since her son's delivery. The mother also had a large head with wide set eyes. She remarked that her father also had had a large head and suffered all his life with chronic headaches. The mother's neurological exam revealed some minor problems. MRIs of both the mother and son showed Chiari malformations, with the mother's herniating 9mm with evidence of crowding. The mother did not want to see a neurosurgeon and the doctors lost touch with the family.

Family 2: An identical twin was evaluated at 15 months of age for trouble swallowing, severe reflux, and failure to thrive. Swallowing study showed abnormal results. Neurological exam showed normal gait and reflexes, but an MRI revealed a 7mm Chiari. Two weeks later, he came back with worsening swallowing problems and trouble breathing. He underwent decompression surgery which resulted in significant improvement in his symptoms. The other twin also had swallowing problems, but not as severe. An MRI showed an 11mm herniation. Because of the previous successful surgery, he also underwent decompression surgery and showed marked improvement.

Family 3: An 11 year old boy was evaluated for decline in athletic ability and problems urinating, plus left hand numbness. Spine MRI revealed a thoracic syrinx, but no Chiari malformation. A year later, the boy returned with worse back pain and tingling in his hands and feet. Repeat MRI's revealed the syrinx had expanded, and a second syrinx had formed. Surprisingly, the MRI also showed that the cerebellar tonsils had descended consistent with Chiari. He underwent surgery and experienced some relief from his symptoms. During the surgery, the doctors noted an arachnoid cyst in the posterior fossa region.

His half-sister was evaluated at 9 years of age for frontal headaches, neck pain, and blurred vision. Neurological exam did not show anything and an MRI showed mild crowding but no tonsillar descent and she was treated conservatively. She started to experience severe shoulder pain and her family chose an elective Chiari surgery. The surgery was successful, but again, the doctors noted arachnoid cysts in the posterior fossa region.

In addition, their first cousin was evaluated at age 7 for pain in the back of the head and neck and occasional nausea. MRI showed mild Chiari with no syrinx. A short time later she developed severe choking episodes and worsening headaches. Repeat MRI revealed a syrinx had formed and she underwent surgery which resolved her symptoms.

Authors' Discussion: The authors point out that evidence has been accumulating for more than 15 years now that Chiari has a genetic component. They suggest that doctors consider Chiari when there is a family history of the disease.

Editor's Discussion: It is unfortunate that despite the evidence, patients often encounter doctors who insist that Chiari can not be inherited. Hopefully time, and continued medical publications, will help this situation. People with Chiari in the family should be extra vigilant for any neurological signs and symptoms that might indicate Chiari or syringomyelia in themselves or relatives. If Chiari is suspected, insist on an MRI.

--Rick Labuda

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