

Key Points

1. Familial syringomyelia - without Chiari - has been reported in the medical literature
2. Research has established there is likely a genetic basis for some portion of Chiari cases
3. Authors report on identical twins, both with syringomyelia, but only one with Chiari
4. Authors believe other twin had Chiari 0, or obstructed CSF flow even though the cerebellar tonsils weren't out of position
5. Decompression surgery on both twins reduced their syrinxes and improved their symptoms
6. Authors speculate that - given the potential genetic basis of Chiari - many cases of familial syringomyelia may actually involve Chiari 0 and that decompression surgery should be considered
7. Cases also highlight environment - maybe even in the uterus and during birth - may influence how a Chiari gene is expressed

Definitions

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

cerebrospinal fluid (CSF) - clear liquid in the brain and spinal cord, acts as a shock absorber

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

cine MRI - type of MRI which can show CSF flow

craniovertebral - referring to the region of the body where the skull and spine meet

DNA - deoxyribonucleic acid,

Syringomyelia In Twins, With And Without Chiari

One of the most pressing questions people have about Chiari (and syringomyelia) involves the genetic aspects of the disease. A parent with Chiari wants to know if there's a chance their children will get it; a parent who finds out that one of their children has Chiari wants to know if their other children are at risk.

Unfortunately, like many aspects of these conditions, there are no straightforward answers. While research indicates that at least some Chiari cases have a genetic basis, it is not known how many, what genes are involved, or how the environment influences the outcome of those who might have inherited a Chiari trait. The issue is further complicated by indications that a Chiari gene, or trait, has what is known as a wide range of expression, meaning that it can affect people differently, even within the same family.

In a case which highlights the complexity of the situation, Dr. Tubbs and his colleagues from the University of Alabama at Birmingham have published a report involving identical twins with syringomyelia. What makes this case especially interesting is that only one twin had a classic Chiari malformation.

The case is described in the June, 2004 issue of the Journal of Child Neurology and details the UAB team's experience with a set of 11-year old identical twin boys. The older twin first came to the attention of the doctors because of worsening scoliosis. Despite an abnormal 30 degree curve, he didn't suffer from neck or head pain and his neurological exam was mostly normal. However, at a younger age, he had had a mass removed from his neck which turned out to be a cervical rib. An MRI showed that he had a syrinx from T2 to T8 and a 13mm Chiari malformation. An X-ray also showed some bony abnormalities involving his top vertebra. He underwent successful decompression surgery which reduced the size of his syrinx and stopped his scoliosis from progressing.

The younger twin, who did not suffer from scoliosis, only came to the attention of the doctors when during an exam of his older brother, he mentioned that his legs always felt jumpy. A neurological exam quickly revealed abnormal reflexes in his legs, and a follow-up MRI showed a syrinx from T3-T9, but no Chiari malformation - meaning his cerebellar tonsils were in the normal position.

Despite the normal positions of the tonsils, his doctors felt he had what is sometimes termed Chiari 0, meaning that he still had crowding in the area around the tonsils and that something was obstructing the natural flow of cerebrospinal fluid. The UAB doctors had had some success in using decompression surgery to treat syringomyelia associated with Chiari 0 and so surgery was discussed with the parents. Even though his symptoms were mild, given the positive outcome of his brother's surgery, the family decided to proceed. The younger twin then underwent a successful decompression which also reduced the size of his syrinx.

It should be pointed out that Chiari 0 is somewhat controversial. Some neurosurgeons have gained national prominence in using decompression surgery - when there is no evidence of Chiari - to treat fibromyalgia and chronic fatigue syndrome. These surgeons have come under a lot of fire from the established medical community and the practice remains controversial. However, several well-respected neurosurgeons have also successfully used decompression surgery to treat syringomyelia in patients with no Chiari malformation. This practice, however, is based on the theory - supported by some evidence - that even though they didn't have Chiari, these patients did have small posterior fossas, or other craniovertebral abnormalities, resulting in crowding and disrupted CSF flow.

The UAB researchers believe that this case of Chiari 0, treated with decompression surgery, indicates that in treating cases of familial syringomyelia, doctors should look carefully at the craniovertebral area for problems and should consider decompression surgery instead of shunting the syrinx directly. Given the evidence that Chiari can be genetic, and given the evidence that Chiari 0 can cause syringomyelia, the authors believe that some familial syringomyelia cases are actually genetically determined Chiari 0, and that the syringomyelia is a result of the Chiari 0.

Although the Chiari 0 aspects of this case are interesting, perhaps even more interesting are the questions it raises about the genetics of Chiari [**Ed note:** The following discussion was not addressed directly in the journal article described above]. Identical twins have identical DNA, so why did one brother have a fairly large Chiari malformation and one brother didn't?

One possible answer lies in how the environment affects the physical expression of the so-called "Chiari" genes. Despite identical DNA, identical twins are born with different fingerprints, and enough physical differences that parents can usually tell them apart. This is because the environment in the uterus is slightly different for each twin. So, in the case of fingerprints, the identical genes establish the same framework for each twin's fingerprints, but their position and movement in the womb determines the final look of their prints.

structured molecule which carries genetic information

expression - in referring to genetics, the physical manifestation of a gene or a genetic trait

familial - a disease which occurs in members of the same family

foramen magnum - large opening at the base of the skull, through which the spinal cord passes and joins with the brain

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

syringomyelia (SM) - neurological condition where a fluid filled cyst forms in the spinal cord

syrinx - fluid filled cyst in the spinal cord

tonsillar ectopia - descent of the cerebellar tonsils into the spinal area

tonsillar herniation - displacement, or descent, of the cerebellar tonsils into the spinal area

Is Chiari the same way? Is there a genetic trait which predisposes people to develop problems in the craniovertebral region? How then does the environment in the uterus affect the development of these problems? In this case, was the older twin more crowded in the womb, or did he suffer more trauma during the birth process? There are some reports (out of Japan) that birth trauma is related to the development of SM. How does the environment after we are born affect the development of these problems, e.g. what role does trauma throughout life play? Is there more than one genetic trait which can result in the same thing, or is one genetic trait expressed differently depending on the environment?

Clearly, there are many more questions than answers, and unfortunately for parents, we are only at the earliest stages of understanding and it may be years before we can answer these important questions.

Familial Syringomyelia

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