Key Points

1. Research has shown that idiopathic scoliosis runs in some families; this suggests a genetic component
2. Research has also shown that Chiari runs in some families, suggesting a genetic component
3. Studies have also shown that a sub-group of people with idiopathic scoliosis also have Chiari and/or syringomyelia
4. This study examined 71 children with idiopathic scoliosis, and a family history of scoliosis, for CM/SM
5. 13% of the children had CM/SM or some level of tonsillar ectopia
6. In addition 27% of the children's relatives - who had scoliosis - also had a neurological problem
7. Authors suggest that some families with idiopathic scoliosis have genetically determined Chiari/syringomyelia

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

Cobb Angle - technique used to measure the severity of a spinal curve - in degrees - from spinal images

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

Linking Chiari, Idiopathic Scoliosis & Genetics

This publication has reported on several aspects of the link between scoliosis and Chiari/syringomyelia, including when children with scoliosis should have an MRI (*Study Identifies Types Of Scoliosis That Indicate Chiari*) and the effects of decompression surgery on the progression of scoliosis (*Chiari, Syringomyelia, Scoliosis, and Surgery*, *How to treat syringomyelia related scoliosis in children*).

The research cited in these articles clearly demonstrates that a sub-group of children with so-called idiopathic scoliosis, meaning no known cause, actually have Chiari and/or syringomyelia, and that more often than not, the neurological problem is causing the scoliosis. Research has also shown that at least some cases of idiopathic scoliosis have a genetic basis - meaning that some families have several members with the condition. Research conducted at Duke University also suggests that at least some cases of Chiari have a genetic basis.

Clearly, idiopathic scoliosis and Chiari are linked at several levels, and now a study out of Japan suggests that some families with idiopathic scoliosis, may have what the researchers call genetically determined craniovertebral abnormalities - meaning some form of CM/SM that has a genetic cause.

Dr. Masatoshi Inoue and his colleagues at Chiba University, in Japan, examined 71 children with idiopathic scoliosis, and a family history of scoliosis, to determine how many of the children, and their family members, had an underlying neurological condition. They published their results in the January, 2003 issue of the journal Spine.

As part of routine screening for school, Dr. Inoue and his team identified 71 children with idiopathic scoliosis (with a curve of at least 10 degrees) and a family history of scoliosis, meaning at least one third-degree relative (see Fig. 1) had the condition. If a third-degree relative was identified as having idiopathic scoliosis, other family members were recruited into the study and screened for the condition.

Next, in order to identify underlying neurological abnormalities, the 71 children underwent thorough MRI's and neurological exams. For the purposes of the study, a Chiari malformation was defined as one or both of the cerebellar tonsils located at least 5mm below the foramen magnum. Tonsillar ectopia was defined as one or both tonsils located between 1mm and 5 mm below the foramen magnum.

Out of the 71 children, 9 (13%) had either Chiari, syringomyelia, both, or tonsillar ectopia. Interestingly, only 5 of the 9 exhibited any deficits during the neurological exam, whereas 4 children demonstrated no neurological problems. The researchers also found that the children with neurological abnormalities were more likely to have developed problems at a young age (<10 yrs), complain of headaches and neck pain, and show signs of neurological deficits upon exam than the rest of the children with scoliosis.

The researchers next turned their attention to the families of those children with positive MRI results. The nine children had a total of 56 first, second, and third-degree relatives. The researchers were able to examine 48 of the relatives for scoliosis and identified 20 with the condition. Of the 20, 15 agreed to undergo MRI's and sure enough two of the relatives had Chiari and syringomyelia, one had Chiari, and one had tonsillar ectopia, meaning that 27% of the relatives examined with scoliosis also had an underlying neurological abnormality.

The authors believe that these results suggest that some families with idiopathic scoliosis are affected by genetically determined craniovertebral abnormalities - meaning CM/SM. They also point out that it appears that the genetic trait responsible has what is known as a wide range of expression, meaning that it affects different people differently, even within the same family. Thus, in this study, some family members had Chiari, some had Chiari and syringomyelia, and some had tonsillar ectopia.

It should be pointed out that the researchers in this study chose to use a traditional definition of Chiari. It would be interesting to repeat the study using cine MRI to determine CSF flow and also look at the volume of the posterior fossa. Based on current theories, one could hypothesize that the families in this study with neurological abnormalities would on average have smaller posterior fossas than the families with no neurological abnormalities. In addition, if a small posterior fossa volume really predisposes people to Chiari, then one might expect those members within the same family who showed signs of CM/SM to have smaller posterior fossas than their relatives who didn't. Finally, it would be interesting to carefully look at the genetic differences between family members who have or don't have scoliosis, and/or Chiari.

While this study provides some tantalizing clues to the possible genetic basis of at least some Chiari cases, clearly much more research is required to solve this puzzle and answer one of the most pressing questions every parent with Chiari has, namely, "Will my children get it?"

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

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

idiopathic - due to an unknown cause

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

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

Figure 1

First-Degree Relatives - parents, children, brothers, sisters

Second-Degree Relatives - grandparents, aunts, uncles, nieces, nephews, etc.

Third-Degree Relatives - great grandparents, cousins, great uncles, great aunts, etc.