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Do Chiari Symptoms Run In the Family?

One of the hallmarks of Chiari is the wide array of symptoms and related conditions (comorbidities) that patients exhibit. Against this backdrop, researchers wanted to see if family members with Chiari (about 12% of Chiari patients have one or more close relatives with Chiari) suffered from similar symptoms and comorbidities. For the study, they recruited 24 families representing 57 patients through social media and several participating medical centers. Each participant completed a clinical survey and was asked to share their medical imaging and medical records. The researchers compared the number of family members who shared a specific symptom or comorbidity to the number who didn't in order to see if there was a statistically significant difference. Using this technique meant that if a given symptom commonly affected family members, then not only the Chiari, but that specific symptom might have a genetic, heritable basis.

For the overall group, 15 of the families (63%) had two affected members, and 9 (37%) had three affected members. The average of diagnosis was 15 years, and the average tonsillar herniation was 11mm. About one-fifth of the group (21%) had a connective tissue disorder such as Ehlers-Danlos Syndrome. Similarly, 23% had syringomyelia while 5% had hydrocephalus.

The group found that headaches, neck pain, dyslexia, EDS, syringomyelia, and hydrocephalus appeared more often in the same families than would be expected. For example, more than two-thirds of patients with syringomyelia had a family member with syringomyelia, compared to the overall rate of 23%. Similarly, 86% of patients who experienced headaches had a Chiari family member who also suffered from headaches, even though only about half of the overall participants reported headaches as a symptom.

While the syringomyelia and headache findings are interesting, EDS is known to be heritable and dyslexia is highly heritable as well. Meanwhile there were only three participants with hydrocephalus overall, which isn't enough to draw any conclusions from. Unfortunately, this was true for many of the symptoms they looked at as only headaches, neck pain, and balance issues had more than ten patients. So while this was an interesting study idea, the group failed to recruit enough patients to draw any strong conclusions. Hopefully, the work will be built upon in the near future.

Source: Clinical phenotypes among patients with familial forms of Chiari malformation type 1.
Mekbib KY, Muñoz W, Allington G, Zhao S, Mehta NH, Fortes C, Shohfi JP, Fan B, Nelson-Williams C, DeSpenza T, Butler WE, Alper SL, Jackson EM, Kahle KT. J Neurosurg Pediatr. 2025 May 2:1-10. doi: 10.3171/2025.1.PEDS24187. Online ahead of print. PMID: 40315599

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