Chiari In The Family

When Conquer Chiari first started twenty years ago, there was a lot of controversy about whether Chiari 'ran in families' or had a genetic component involved. Today, these are not controversial topics at all. Families with multiple affected members have been well documented in the medical literature; in fact, the Chiari1000 shows that about 12% of Chiari patients report at least one immediate family member who has also been diagnosed. In addition, genetic studies – including ones sponsored by Conquer Chiari – have shown that while complex, it is likely that Chiari in general has a genetic component.

Now, a recent literature review from a group in Canada profiled what Chiari *families* look like. The research group identified 29 English language articles that identified two or more family members with Chiari and where Chiari was the focus of the article. In total there were 34 families and 92 Chiari cases, with an average of 3 affected people per family.

Interestingly, nearly two-thirds of the cases were siblings (with twins included) and the Mother-Child diagnosis was nearly twice as common as the Father-Child diagnosis:

- Siblings – 35%
- Twins or triplets – 27%
- First cousins – 4%
- Father & child – 12%
- Mother & child – 23%

The average age of diagnosis was 23 years, which is younger than is normally found for adult Chiari, but likely reflects combining cases of all ages. Symptomatically, only 17% of the cases reported headaches which is extremely low for a Chiari group; however, 54% had syringomyelia which is very high compared to more general Chiari groups. Sixty-eight percent of the group had undergone decompression surgery, which is also high but could be due to the high rate of syringomyelia.

Genetic studies have shown that the genes suspected to be involved in Chiari families aren't necessarily the same genes that have been identified in more general Chiari populations, so it is not surprising that the profile of a group of Chiari families differs in certain ways as well.

There is still controversy about whether family members, especially pediatric siblings, should undergo MRIs if Chiari is found in one family member. Many physicians believe it is not necessary or even helpful unless the person has symptoms, but regardless parents can certainly be on alert for any indications of Chiari and quickly pursue imaging if an issue arises.

Source: Familial Chiari malformation: a systematic review and illustrative cases. Dhawan A, Dhawan J, Sharma AN, Azzam DB, Cherry A, Fehlings MG. J Neurosurg Spine. 2024 Apr 12:1-10. doi: 10.3171/2024.1.SPINE231277. Online ahead of print. PMID: 38608294

Conquer Chiari's research updates highlight and summarize interesting publications from the medical literature while providing background and context. The summaries do contain some medical terminology and assume a general understanding of Chiari. Introductory information and many more research articles can be found in the Conquer Chiari Library.

Conquer Chiari is a 501(c)(3) public charity dedicated to improving the experiences and outcomes of Chiari patients through education, awareness and research.