Research Update | March, 2018

Topics: Pediatric Chiari; EDS, POTS; Syringomyelia, Charcot Joint

Conquer Chiari's monthly 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 about Chiari, plus many more research articles, can be found at www.conquerchiari.org.

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In 2003, researchers from Canada surveyed pediatric neurosurgeons world-wide on whether they would recommend surgery in hypothetical cases, and if so, what technique they would use. Recently, the same researchers conducted a similar survey to see what has changed in regards to the attitudes of practicing pediatric neurosurgeons toward treating Chiari and syringomyelia. Specifically, they contacted 300 surgeons through the International Society for Pediatric Neurosurgery, and got a response from 122, representing 34 different countries. The survey presented 3 hypothetical pediatric patients:

- Patient 1: A 12-year-old boy being investigated for learning disabilities, but with no other symptoms, a normal neurological exam, and a 12mm herniation.
- Patient 2: A 9-year-old boy with headaches for six months and a 10mm herniation
- Patient 3: An 11-year-old girl with scoliosis and 12 mm of herniation

For each case, the survey then added two extra scenarios, a small (diameter) thoracic syrinx, and then a large cervical syrinx. The respondents were asked if they would recommend surgery, and if so, whether they would open the dura, and again if so, what material they would use for a patch. The responses were compared statistically to the responses from 2003.

For the first two cases, the responses in the new survey were not significantly different from the original survey with most surgeons taking a conservative approach to patient one unless there was a large syrinx. For patient two, about half would recommend surgery even with no syrinx, and this climbed to 90% opting for surgery when a large syrinx is present. For the third hypothetical patient with scoliosis, it appears treatment attitudes have evolved. In 2003, 58% of the surgeons said they would operate even when there was no syrinx. In the recent survey, this dropped to only 32%. This trend held even with a large syrinx in the mix, with the surgical choice declining from 97% in 2003 to 83% more recently.

Significant differences also showed up in surgical technique, with more surgeons saying they don't open the dura. However, the most dramatic change in attitudes appears to be in choice of dural patches, with 98% of surgeons today saying they use either a synthetic patch or tissue from the patient. This is an increase from 58% previously. It also appears that surgeons have stopped using bovine and cadaver patches completely.

While some attitudes in the surgical community have changed over the past decade, there is still a tremendous amount of variability in when surgeons will recommend surgery and then how they will perform that surgery. The end result of this for patients and their families is if they decide to get a second opinion, there's a good chance they will hear something different.

SOURCE: *International survey on the management of Chiari 1 malformation and syringomyelia: evolving worldwide opinions. Singhal A, Cheong A, Steinbok P. Childs Nerv Syst. 2018 Mar 12. doi: 10.1007/s00381-018-3741-x.*

According to Dysautonomia International, postural orthostatic tachycardia syndrome (POTS) affects as many as three million people in the US. Diagnostically, POTS is defined as a change in heart rate of a certain value (depending on age) after standing up from a prone position. However, this clinical definition doesn't do justice to the effect that POTS can have on people. In fact, it has been estimated that the quality of life in POTS patients is comparable to those on dialysis for kidney failure. POTS is not generally considered a disease onto itself, but rather a collection of similar symptoms caused by other things that disrupt the normal functioning of the autonomic nervous system that controls heart rate and blood pressure.

In recent years, the medical literature has recognized a connection between POTS and Ehlers-Danlos Syndrome (EDS), a collection of inheritable connective tissue disorders. In fact, a researcher from Johns Hopkins and colleagues published a review of the medical literature on POTS and EDS, which showed that the link between the two is likely underestimated. Since some patients may not have a formal diagnosis of POTS, they reviewed published articles and focused on symptoms of POTS in EDS patients, such as passing out (syncope) and lightheadedness. From this analysis, they determined that the rate of POTS among hypermobile EDS patients (the type of EDS which involves extreme laxity in the joints) could be 40% or higher.

This is of interest to the Chiari community because a significant subset of Chiari patients also has hypermobile EDS. Data from the Chiari 1000 (C1000) shows that 11% of over 1100 patients in the database report being diagnosed with EDS by a physician. However, because the diagnosis of EDS is fairly involved, it is possible this number underestimates the true rate of EDS among Chiari. One of the diagnostic criteria for EDS involves the Beighton scale, which evaluates hypermobility using several tests. Although a true EDS diagnosis involves more criteria, the C1000 included the Beighton scale. The results show that 26% of over 800 adult females scored high enough on the Beighton scale to be considered clinically relevant for hypermobile EDS. While it is likely that not every single case has EDS, and there could be sampling bias in the C1000, the rate of EDS among adult women with Chiari can be estimated as somewhere between 11-20%.

Similarly, we can look at the rate of POTS among CM/EDS patients in the C1000. Here, only 3.4% of those with CM and EDS (over 130 adult women) reported being diagnosed with POTS. However, taking a similar approach to the published literature review shows that 53% of those with CM and EDS report fainting as a symptom, indicating they may have POTS. This is in line with the publication's findings among EDS patients who don't necessarily have Chiari.

To summarize, extrapolating from C1000 data indicates that 11% or more (up to 26%) of adult women with Chiari may also have hypermobile EDS and that as many as half of that group may also suffer from POTS symptoms. It is important to note that since POTS is common among EDS patients without Chiari, whether there is a causal link between Chiari and POTS is not at all clear. It is also important to note that more rigorous research is required to accurately assess the rates of EDS and POTS among Chiari patients.

SOURCE: *Postural tachycardia syndrome and other forms of orthostatic intolerance in Ehlers-Danlos syndrome. Roma M, Marden CL, De Wandele I, Francomano CA, Rowe PC. Auton Neurosci. 2018 Mar 5. pii: S1566-0702(17)30298-9. doi: 10.1016/j.autneu.2018.02.006*

Charcot arthropathy of a joint occurs when nerve damage causes a joint to degrade over time resulting in bone destruction, resorption, and deformity. Since the nerves associated with the joint are damaged, ironically the joint often eventually becomes painless. Charcot joints are caused by diseases and conditions that result in nerve damage, such as diabetes, and of interest here, syringomyelia. Because most Chiari related syrinxes are found in the cervical portion of the spine, the most common joints affected in this way in SM patients are shoulders and elbows. However, a Case Report from China demonstrates that other joints can be damaged by syrinxes as well. They present the story of a 35-year-old man with a history of hydrocephalus and Chiari which had been previously treated with shunting and a decompression. His left leg was swollen and his knee had limited movement. A CT scan of the knee showed Charcot arthropathy and an MRI of his spine revealed a syrinx. The syrinx was addressed with direct shunting and over time his knee improved enough that he could walk with a brace. This case highlights the incredible damage that syrinxes have the potential to do once nerves start to be damaged.

SOURCE: *Syringomyelia with left knee charcot arthropathy: a case report. Li G, Ding Y, Zhang C, Huang H. Br J Neurosurg. 2018 Feb 6:1-2. doi: 10.1080/02688697.2018.1429570.*

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