Incidental Cerebellar Tonsillar Ectopia in Children Raises Questions About Chiari

(*Note: The author of this update – Rick Labuda – is one of the authors of the journal paper being discussed.)*

As has been discussed previously in these Research Updates, the widespread adoption of MRI technology has raised some difficult questions about Chiari. First and foremost is the fact that many people meet the imaging definition of Chiari – meaning tonsillar herniation of at least 5mm – but do not and likely will not have Chiari symptoms. These cases are often referred to as incidental, because Chiari was not being looked for, and are being identified more frequently. In fact, studies of hospital imaging databases have found that up to 1% of adults and 3% of children actually have tonsillar herniation of 5mm with the vast majority presumed to be incidental in nature. However, there is some question as to whether these cases are truly symptom free as they were in the hospital for a reason. For example, in one such study of incidentally identified pediatric cases more than a third actually showed symptoms such as seizures, headaches, and developmental delays, and another 10% were evaluated after a trauma. Now, researchers from the Conquer Chiari Research Center (CCRC) have leveraged a National Institutes of Health project to identify and study truly incidental cases of tonsillar ectopia (another word for herniation) in adolescents. The Adolescent Brain Cognitive Development (ABCD) study is the largest and most extensive study of brain development in the US. Nearly 12,000 9 and 10 year olds were recruited through schools, screened for major health issues, and will be given regular MRIs and battery of health assessments as they are followed into adulthood. The CCRC researchers developed specialized software to automatically identify over 100 cases of Incidental Cerebellar Tonsillar Ectopia (ICTE) from the MRIs of over 11,000 ABCD participants. The ICTE cases all had tonsillar descent of at least 5mm as measured on MRI. From the same dataset they also created a control group that matched the ICTE group exactly by age, gender, race, ethnicity, and BMI but who did not have tonsillar herniation. Next they compared 22 MRI measurements of the brain, skull and spine, and 22 health instruments between the two groups. They found that the morphometrics of the ICTE group actually looked very similar to what is seen in symptomatic Chiari (versus healthy controls) with reduced size of the posterior fossa, sagging of the hindbrain, and crowding around the cerebellum and cerebellar tonsils. However, there were absolutely no differences in the health instruments between the two groups, including items that you would expect to see with Chiari such as headaches, sleep problems, vision problems, etc. This means that even though these children looked like Chiari cases on MRI they showed absolutely no effects from it in terms of their physical, mental, or behavioral health. It should be noted that in the ICTE group, the alignment of the upper cervical spine appeared normal and in adult Chiari cases it often isn't. However it is not clear if this is an important distinction between who gets symptoms and who doesn't, or if this is a reflection of the young age of the study subjects. It is also not clear how many (if any) of the ICTE children may develop symptoms as adults. But since around 1% of the ABCD group had ICTE and symptomatic Chiari is estimated at 1 in 1,000, it seems likely that the vast majority will never experience symptoms. What is clear is that this study calls into question the prevailing theory that Chiari symptoms arise from tissue crowding and disruption of the flow of cerebrospinal fluid. The ICTE cases showed similar levels of crowding as to what is seen in symptomatic Chiari but the children showed no apparent effects from it. It appears likely that crowding and spinal fluid blockage are only part of the equation and that there is another component as to what makes some people with tonsillar herniation symptomatic and others not. Fortunately, the CCRC researchers can follow the same subjects and see how their brains, skulls, and spines develop as they age and also watch for any indications of underlying Chiari symptoms.

Source: Imaging and health metrics in incidental cerebellar tonsillar ectopia: findings from the Adolescent Brain Cognitive Development Study (ABCD). Nwotchouang BST, Ibrahimy A, Loth DM, Labuda E, Labuda N, Eppleheimer M, Labuda R, Bapuraj JR, Allen PA, Klinge P, Loth F. Neuroradiology. 2021 Jul 11. Online ahead of print.

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 at www.conquerchiari.org.