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Ehlers-Danlos syndrome + Chiari I malformation

Overview

Ehlers-Danlos (ED) syndrome affects connective tissue in the body making joints hyper flexible. ED patients with Chiari I malformation need special expertise during diagnosis and treatment to prevent further progression. Chiari relates to the herniation of the lower part of the brain (cerebellar tonsils) through the skull, exerting pressure on the brainstem and spinal cord. Symptoms vary widely and are caused by the disruption of cerebrospinal fluid flow. When Chiari occurs with ED syndrome, symptoms related to the lower brainstem may be more likely. Patients with ED + Chiari often have loose/unstable joints and ligament laxity at the cranial-cervical junction.

Incidence of Ehlers-Danlos + Chiari

- In one study of 2800 Chiari patients, about 13% of these patients also had ED syndrome. The combination of Chiari + ED more often affects women (7:1 female:male), causes lower brain stem problems, and shows scar tissue covering the finger-shaped C2 vertebra.
- Ehlers-Danlos types I-III include the Classic (I and II) and Hypermobility (III) types. Patients may face chronic joint or limb pain, sprains or dislocations, musculoskeletal pain, and tissue fragility.

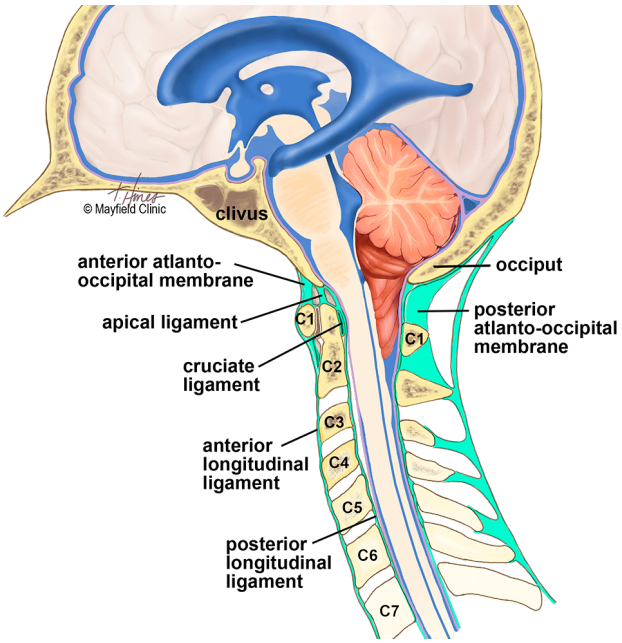
Implications for diagnosis

Chiari + ED syndrome can cause a myriad of symptoms, ranging from mild to severe. Accurate diagnosis of Chiari is made by assessment of patient symptoms, neurological exam, MRI, and cine MRI. When ED also occurs, additional imaging may be obtained. Flexion-extension x-rays and sometimes CT can determine the stability of the topmost spine, the C1-C2 joint.

Implications for treatment

ED syndrome includes a faulty collagen that weakens the strength and elasticity of the muscles, skin, and joints. Joint hypermobility and scar tissue are special concerns for ED + Chiari patients.

Neurological/spinal problems, such as neck pain, scoliosis, pain across shoulder blades, and more, can be worsened in ED + Chiari.



Patients with Ehlers-Danlos and Chiari may have hypermobility at the C1-C2 joint that can cause instability where the skull and spine join.

Chiari surgery to expand the space at the back of the skull is more complicated because of ED syndrome. Removal of the C1 arch is often avoided because of the risk of further instability. If the C1 arch must be removed, the patient will be followed closely with flexion-extension x-rays of the neck to ensure stability of the C1-C2 spine. If instability is a problem, fusion is added to stabilize the joint; most patients do not undergo fusion.

Mayfield addresses the combination of ED + Chiari with a team approach. The Chiari specialist can review the patient's imaging studies and perform a thorough exam. Neurological tests help determine the combined effects/injury to the bones, nerves, and soft tissues. Patients can then carefully weigh each treatment option, whether observation or surgery, understand what to expect from their treatment, and take steps toward health habits that may help.

https://www.ehlers-danlos.com

updated > 4.2022
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