Definitions

cerebellar tonsils - portion of the cerebellum located at the bottom, so named because of their shape

cerebellum - part of the brain located at the bottom of the skull, near the opening to the spinal area; important for muscle control, movement, and balance

cerebrospinal fluid (CSF) - clear liquid which surrounds, and protects, the brain and spinal cord

Chiari malformation - condition where the cerebellar tonsils are displaced out of the skull area into the spinal area, causing compression of brain tissue and disruption of CSF flow

enzyme - a protein that speeds up chemical reactions

Fabry disease - genetic syndrome which causes a deficiency in a certain enzyme; leads to kidney problems, skin problems, and burning in the hands

genetic - inherited

magnetic resonance imaging (MRI) - diagnostic test which uses a large magnet to create images of internal body parts

posterior fossa - depression on the inside of the back of the skull, near the base, where the cerebellum is normally situated

Chiari Linked To Fabry Disease

March 20, 2006

Chiari I Malformation In Four Unrelated Patients Affected With Fabry Disease

Authors: Dominique Germain, Karelle Benistan, Philippe Halimi
University/Hospital: Hopitaux de Paris
Journal: European Journal of Medical Genetics, on-line Feb. 2006

Introduction: Fabry disease is genetic disorder which results in lowered production of a key enzyme. This in turn leads to problems with the kidneys, skin problems, and possible mental retardation. This case highlights how doctors found an apparent connection between Fabry disease and Chiari.

Patient 1: 30 year old man with Fabry went to the emergency room for dizziness and balance problems. He was given enzyme replacement therapy to treat the Fabry disease, but an MRI was ordered to look for cerebellar lesions. The MRI revealed a 5 mm Chiari with no syrinx.

Patient 2-6: After this finding, the doctors began to run MRI's on all Fabry patients they encountered before they were treated with enzyme therapy. Out of 54 total Fabry patients six were found to have Chiari >5mm

Author's Discussion: The authors believe that this is the first documented case of Fabry being linked to Chiari. Even though they found that greater than 10% of the Fabry patients had Chiari, they state that further investigation is required to confirm a link. While the connection between Fabry and Chiari is not clear, the authors do note that Fabry patients have been shown to have abnormally high blood flow in the cerebellar area. Interestingly, they reviewed the medical literature to identify other genetic disorders in which Chiari has been reported to co-exist and found 17 such disorders, including (but not limited to): Crouzon syndrome, growth hormone deficiency, kabuki syndrome, klippel-feil, and marfan syndrome.

Editor's Discussion: Besides the powerful results they found when they screened their Fabry patients for Chiari, I thought seeing the list of 17 genetic disorders with which Chiari has been reported in the medical literature was very interesting. It should be noted that their list did not include Ehler-Danlos, the connective tissue disorder whose link to Chiari is being investigated by the Chiari Institute and NIH.

--Rick Labuda