From The Desk of Regina S Bland, MD

I am a board certified pediatrician practicing in West Palm Beach, FL. I was diagnosed with Chari malformation in July, 1988 and have undergone a cervico-syringoarachnoid shunt in 1990 and decompression surgery in 2003. I take medications daily to enable me to be present for my patients. I hope that in this occassional article series that I might "bridge the gap" between patient and doctor.

Dr. Bland: Connective Tissue Is The Foundation Of Our Bodies

November 20, 2006 - Connective tissue is the foundation of our bodies. It is found in the muscles, the bones, the walls of blood vessels, and the skin. Some of us with Chiari 1 malformation (CM1) have been told that we have cerebellar tonsillar herniation into our spinal column because our posterior cranial fossa is too small-an abnormality of bones in our skull. Some Chiari patients have other bony abnormalities of the skull or spine-deformities of the odontoid process (anterior part of the second cervical vertebra), bifid or posterior split in the first cervical vertebra), fusion of first cervical vertebra to the skull, fusion or abnormal bony joining of cervical vertebrae (known as Klippel Feil anomaly), cranial lacunae or unexpected holes in the cranial bones. Some Chiarians have abnormal facial structures-small sinuses with frequent infections, temperomandibular joint problems (TMJ) or small mandibles (difficulty in jaw opening).

In 2005 at the American Syringomyelia Alliance Project (ASAP) and the Ehler Danlos Syndrome (EDS) conferences, Dr. Thomas Milhorat, Director of The Chiari Institute at North Shore LIJ in Great Neck New York and Dr. McDonnell of the genetics laboratory of the National Institute of Aging at the National Institute of Health in Maryland, discussed an apparent linkage between EDS (a group of connective tissue disorders) and Chiari 1 malformation. According to their data the incidence of Chiari 1 malformation is 1:5000 and the incidence of Ehler Danlos Syndromes is 1:5000. If these two entities were random, you would expect them to occur together (multiply the two numbers) in 1 in 25 million. However Dr. Milhorat reports that he sees at the Chiari Institute in New York, an incidence of **1 in 15** patients of simultaneous CM1 and EDS.

EDS patients usually demonstrate one or more of the following components-hyper-elastic and fragile skin with frequent atrophic scarring, hyper mobility of joints (double-jointed or frequent dislocations), and/or fragility of cutaneous blood vessels (easy bruising) or abnormalities of major blood vessels (aneurysms). Collagens are the main proteins of connective tissue and defects in various collagens have been discovered for several types of EDS. Defects can happen in the quantity (amount of collagen molecules produced) or in the quality of the collagen (abnormal collagen types produced).

Dr. Milhorat at that conference discussed that the patients that presented with both EDS and CM1 appeared to have more symptoms relating to anterior brainstem dysfunction including nausea, dysphagia, hoarseness, sleep apnea, difficulty swallowing, dysautonomia (sometimes known as POTS), palpitations and shortness of breath. Their radiographic findings were more likely to show pannus formation around the odontoid and the aforementioned cranial abnormalities and facial abnormalities. Patients with both syndromes were more likely to demonstrate swallowing problems post-operatively and were more likely to demonstrate cranio-cervical instability.

Dr. McDonnell went on to discuss the known genetic defects in some types of EDS while the specific gene defects had not been identified in other types. Familial aggregation studies (families with multiple cases of illness) suggest genetic causes for EDS and CM1 even though the specific genes affected nor their mechanisms of action have not been determined.

Ed. Note: The opinions expressed above are solely those of the author. They do not represent the opinions of the editor, publisher, or this publication. Anyone with a medical problem is strongly encouraged to seek professional medical care.