

Is It Chiari Or ALS? The Link With Spina Bifida

Case Studies is a feature designed to highlight interesting patient cases reported in the research. Given the lack of knowledge about CM/SM, much of the published research comes in the form of case studies - doctors describing one or two patients they have seen and treated - as opposed to rigorous scientific studies. While this type of publication doesn't advance the scientific cause as much, it does give us a window into some of the issues surrounding CM/SM, including lasting side effects and related conditions. And hopefully, some of our readers will say, "Hey, that's just like me!" and know they are not alone in what they are going through.

CASE 1: Misdiagnosis of amyotrophic lateral sclerosis in a patient with dysphagia due to Chiari I Malformation

Reported In: Journal of Neurology Neurosurgery and Psychiatry. February, 2002.

Doctors: Dr. Paulig and Dr. Prosiegel, Neurologisches Krankenhaus, Munich, Germany

Patient:

- 78-yr old woman with trouble swallowing (dysphagia) for more than a year
- She was to the point where she couldn't feed herself and needed a feeding tube
- Had been diagnosed with ALS 1 month prior
- Neurological exam showed that she was breathing in her saliva and her tongue was atrophied
- Muscles in her arms and legs were normal, no other neurological symptoms
- MRI revealed a Chiari 1 malformation with no syrinx
- Underwent successful decompression surgery
- Required therapy to learn to swallow and eat on her own

Observations:

- Chiari & Syringomyelia News has previously reported that trouble swallowing in young children may indicate a Chiari malformation
- Sometimes trouble swallowing is the only symptom of Chiari
- The problem is likely caused by the displaced cerebellum putting pressure on the part of the brain that controls swallowing

Ed Note: *This case demonstrates how Chiari - with it's myriad neurological symptoms - is often misdiagnosed or missed completely. Luckily, as MRI's become more routine, Chiari malformations are being identified sooner.*

CASE 2: Resolution of Chiari Malformation After Repair Of A Congenital Thoracic Meningocele

Reported In: Neurosurgery: Case Studies. December, 2002

Doctors: Dr. Mazzola, Dr. Pollack, Rick Madhok, B.S. Children's Hospital of Pittsburgh; Pittsburgh, Pennsylvania

Patient:

- Newborn girl of normal birth weight, normal head shape, and no obvious neurological problems
- Born with a fluid filled, skin covered mass in the middle of her thoracic area
- MRI revealed a meningocele with the spinal cord tethered and a Chiari I malformation
- The meningocele was surgically repaired the next day and the spinal cord was freed
- Follow-up MRI 3 months later showed that the Chiari malformation had completely resolved and there was adequate space around the cerebellum
- A second follow-up MRI 4 months later again showed no signs of a Chiari malformation

Observations:

Definitions

amyotrophic lateral sclerosis (ALS) - also known as Lou Gehrig's Disease; a severe, progressive neurological disease which results in loss of muscle control and function

cerebrospinal fluid (CSF) - clear liquid in the brain and spinal cord, acts as a shock absorber

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

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

craniocervical junction - area where the skull meets the spine

decompression surgery - common term for any of several variations of a surgical procedure to alleviate a Chiari malformation

dysphagia - trouble swallowing

meningocele - birth defect where the covering of the spine bulges through an opening; type of spina bifida

spina bifida - birth defect where the spinal cord doesn't develop properly and parts of the spine protrude outside the body

syringomyelia (SM) - neurological condition where a fluid filled cyst forms in the spinal cord

syrinx - fluid filled cyst in the spinal cord

tethered cord - condition where the center part of the spinal cord is abnormally attached to the bones of the spine

thoracic - the middle part of the spine; chest area

- It is well established that Chiari malformations can be acquired due to CSF leaks and other causes that result in low spinal pressure relative to the pressure in the skull
- Authors believe that some congenital (and reversible) Chiari malformations are due to low pressure in the spine; in this case because of the meningocele
- One theory postulates that the link between Chiari and Spina Bifida is a result the Spina Bifida causing low spinal pressure during development which causes the cerebellum to descend out of the skull

Ed Note: *This case highlights that it is likely that there are several "causes" of Chiari malformation. Usually, low spinal pressure is associated with acquired Chiari, but if the low spinal pressure is present (due to a meningocele for example) during development, a congenital malformation may result.*

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