Cause, Diagnosis and Treatments of Atlantoaxial Instability in Down Syndrome

Atlanotaxial Instability in Down syndrome - Who Should be Screened?

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Atlantoaxial instability (AAI) is a common orthopedic problem seen in people with Down syndrome. Although it has a complicated name and sounds somewhat intimidating, for the most part it causes no problems to those who have it. In order to understand AAI, it is important to understand a little about the function and structure of the spinal cord, nerves, vertebrae and ligaments.

Spinal Cord, Nerves, Vertebrae and Ligaments

The top of the spinal cord is a thick tube-like structure that starts at the base of the brain and runs all the way down the back to the lumbar region. The spinal cord contains the body's nerves or neurons. Nerves are a special type of cell that carries messages between the brain and the rest of the body. A nerve is like an electric cable that passes electric current and carries signals between parts of the body.

Vertebrae are irregularly shaped bones organized in to a column in the back of the body that runs from the base of the brain to the pelvis. The spinal cord runs through and is protected by this stack of vertebrae. There are 33 vertebrae, which are generally broken in to four regions: cervical (7), thoracic (12), lumbar (5) and the vertebra of the pelvis. Cervical vertebrae are located in the neck region and are abbreviated as C1-C7. If you bend your head forward and run your fingers down the back of your head, the first large bump that you feel is your C1 or cervical-1 vertebra. The next one down is C2 and so forth. C1 is referred to as the atlas vertebra and C2 as the axis vertebra. Misalignment between these vertebrae is referred to as atlantoaxial instability or AAI.

Vertebrae are held in place by muscles and ligaments. The functions of the vertebral column include protection of the spinal cord and internal organs, structural support for the head and maintaining both flexibility and mobility.

Since people with Down syndrome have low muscle tone and lax ligaments, their vertebrae can become misaligned. When the C1 and the C2 vertebrae are misaligned, you have AAI. In people with Down syndrome, the ligament most often involved in AAI is called the "transverse ligament."

Diagnosis of AAI

Most cases of asymptomatic AAI are made through screening x-rays. All children with Down syndrome should be screened for AAI by the age of 3. The diagnosis of symptomatic AAI is usually made either through neurological exam (physical exam that examines how the nerves are working) and/or by x-ray.

Types of AAI

There are two types of atlantoaxial instability - asymptomatic AAI and symptomatic AAI. Asymptomatic AAI means that AAI can be seen on an x-ray, but it is not causing any neurologic problems in the person who has it. Symptomatic AAI means that AAI is present on an x-ray, and it is causing some neurologic problems for the person that has it. Somewhere between 10% to 20% of people with Down syndrome have asymptomatic AAI on x-ray, and only 1% to 2% of people with Down syndrome have symptomatic AAI.

Neurologic Symptoms in AAI

One of the jobs of the vertebral column is to protect the spinal cord, which runs inside it. The spinal cord is a collection of nerves that is protected by the vertebral column. Symptomatic AAI causes a variety of different neurologic symptoms such as:

- Clumsiness
- Lack of Coordination
- Difficulty walking
- Walking with an abnormal gait (ie limping)
- Getting tired easily
- Nerve pain or limited ability to move the neck
- Spasticity - tightness in the muscles
- Clonus - muscle contractions or spasms

If someone with Down syndrome develops any of these neurological symptoms, they should immediately be evaluated by a physician. The physician will usually perform a complete neurologic exam and order imaging studies, such as x-ray, CT scans or an MRI.

Treatment of AAI

Asymptomatic AA does not require any treatment. Most often parents are simply advised what symptoms to watch for in the person with AAI.

If a person is showing signs of spinal cord compression, treatment is indicated. The goals of treating symptomatic AAI is to protect the spinal cord, stabilize the spinal column or vertebrae and decompress any trapped nerves. Depending on the extent of the problem, spinal cord stabilization can be achieved through wearing a soft collar, halter traction with pain medication and muscle relaxants and possibly surgery.

Sources:

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