Managing Atlantoaxial Instability

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The release of the new guideline "Health Supervision for Children With Down Syndrome" from the American Academy of Pediatrics in July, 2011 brought about multiple new recommendations for the routine care of children with Down syndrome. It is likely that you are all very aware of these, but in case you are not, Dr. Jenny Motley gave a nice overview of them in the previous Ds Press. At first glance, most of the changes in the guideline appear very logical as they address issues in children with Down syndrome that were previously under-recognized at the time that the last guideline was written. One of the recommendations, that to no longer perform routine neck x-ray screening to look for atlanto-axial instability, has raised many questions both from parents as well as medical professionals.

The human spinal cord is supported and kept safe by our vertebral bones, which are arranged like a stack of rings in the middle of the back. The atlas is the first vertebral bone just below the skull. Just as Atlas supported the Earth in Greek mythology, the atlas supports our skull. The next vertebra is the axis, and just as the Earth rotates on its axis, the atlas bone rests and can rotate on the axis bone. The atlas and axis are shaped such that a portion of the axis sticks up and sits inside a portion of the atlas, and the two bones are held together by several ligaments. This allows us to rotate and move our heads in the many directions that we are able, while at the same time keeping the spinal cord safe as it passes through both bones.

For a variety of reasons children with Down syndrome are at risk for a condition known as atlanto-axial instability or subluxation which occurs when the structures that hold the atlas and axis together loosen and allow extra, unwanted movement of the two bones. With instability the two bones are simply "loose" which leads to pinching of the spinal cord. With subluxation the two bones truly slip on one another, causing dangerous or even life threatening pressure to be put on the spinal cord. In order to keep children with DS safe, the previous edition of the health care guidelines published in 2001 suggested that all children undergo screening with an x-ray study of the neck between the age of three to five years. The age of three years was chosen as a starting point because that is when the bones are developed and mineralized enough to make an accurate diagnosis. However, what has been found since then is that plain neck x-rays are not adequate for predicting which children are at increased risk for the condition, and may even miss the diagnosis in children who actually have the condition. Additional factors leading to this decision are that obtaining these x-rays is costly, and it exposes the children to unnecessary radiation.

The new guideline recommends monitoring children with DS carefully for myelopathic signs, or symptoms that indicate that there is early impingement of the spinal cord in this region. These signs include “new onset of symptoms or change in gait or use of arms or hands, change in bowel or bladder function, neck pain, stiff neck, head tilt, torticollis, how the child positions his or her head, change in general function, or weakness.” At first this seems rather simple as these signs would seem to be plainly evident if present. But, since children with DS demonstrate low muscle tone and generalized laxity of their joints, recognizing these signs may not be so easy. If you believe that your child is exhibiting any of
the signs mentioned, bring it to your health care providers attention as further testing is warranted. They should obtain neck x-rays in what is known as a neutral position, that is without bending the neck too far in either direction. If those x-rays are normal, additional x-rays should be obtained with the head flexed forward and extended back. If the x-rays are abnormal, the child should be referred to an orthopedic surgeon or neurosurgeon who is familiar with this condition in children in Down syndrome. Even if the x-rays are normal, the symptomatic child should be referred to a specialist.

The guidelines also emphasize the importance of careful positioning of the neck in children with DS when undergoing anesthesia, surgical procedures, and x-ray tests. Another section of the new guideline states that "Parents should be advised that participation in some sports, including contact sports such as football and soccer and gymnastics (usually at older ages), places children at increased risk of spinal cord injury and that trampoline use should be avoided by all children with or without Down syndrome younger than 6 years and by older children unless under direct professional supervision. Special Olympics has specific screening requirements for participation in some sports."

The key point is that as parents we know our children best. This makes it more likely that we will notice a subtle change in our child's behavior or function before those same signs will become apparent to a medical professional. If we all work together, we will have the best chance of keeping our children safe.