

Atlantoaxial Instability in Down Syndrome: Controversy and Commentary

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In 1984, the AAP issued its first position statement on Atlantoaxial Instability (AAI) in children with Down Syndrome (DS):

1. All children with DS who wish to participate in sports should have cervical spine X-rays.
 2. When the distance on X-ray between the atlas (1st vertebra) and odontoid process (2nd vertebra) is more than 4.5 millimeters (mm), restriction on sports is advised.
 3. Repeated X-rays are not indicated for children with DS who have previously had normal neck X-rays.
 4. Persons with atlantoaxial subluxation or dislocation and neurologic signs should be restricted from "all strenuous activities."
 5. Persons with DS who have no evidence of AAI may participate in all sports.
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The American Academy of Pediatrics' Committee on Sports Medicine released a revised statement in July 1995 regarding Atlantoaxial Instability (AAI) in children with DS. This was published in the journal *Pediatrics*, 96(1):151-154. Here's the text, edited for brevity's sake:

"In 1984, the Amer. Academy of Pediatrics (AAP) published a position statement on screening for AAI in youth with DS. In that statement, the AAP supported the requirement introduced by the Special Olympics (SO) in 1983 that lateral (side view) neck X-rays be obtained for individuals with DS before they participate in the SO's nationwide competitive program. Those participants with radiologic evidence of AAI are banned from certain activities that may be associated with increased risk of injury to the cervical spine....The Committee on Sports Medicine recently has reviewed the data on which this recommendation was based and has decided that uncertainty exists concerning the value of cervical spine X-rays in screening for possible catastrophic neck injury in athletes with DS. The 1984 statement therefore has been retired. This review discusses the available research data on this subject.

BACKGROUND:

AAI denotes increased mobility at the articulation of the first and second cervical vertebrae (atlantoaxial joint). The causes of AAI are not well understood but may include abnormalities of the ligaments that maintain the integrity of the articulation, bony abnormalities of the cervical vertebrae, or both.

In its mildest form, AAI is asymptomatic and is diagnosed using X-rays.....Symptomatic AAI results from subluxation (excessive slippage) that is severe enough to injure the spinal cord, or from dislocation at the atlantoaxial joint.

Approximately 15% of youth with DS have AAI. Almost all are asymptomatic. Some asymptomatic individuals who have normal X-rays initially will have abnormal X-rays later, and others with initially abnormal X-rays will have normal follow-up X-rays; the latter change is more common....

The neurologic manifestations of symptomatic AAI include easy fatiguability, difficulties in walking, abnormal gait, neck pain, limited neck mobility, torticollis (head tilt), incoordination and clumsiness, sensory deficits, spasticity, hyperreflexia...and {other spinal cord} signs and symptoms. Such signs and symptoms often remain relatively stable for months or years; occasionally they progress, rarely even to paraplegia, hemiplegia, quadriplegia, or death. Trauma rarely causes the initial appearance or the progression of these symptoms. Nearly all of the individuals who have experienced catastrophic injury to the spinal cord had weeks to years of preceding, less severe neurologic abnormalities....

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Most importantly, symptomatic AAI is apparently rare in individuals with DS. In the pediatric age group, only 41 well-documented cases have been described in the published literature....

Asymptomatic AAI, which is common, has not been proven to be a significant risk factor for symptomatic AAI....

The efficacy of the intervention to prevent symptomatic AAI has never been tested. Sports trauma has not been an important cause of symptomatic AAI in the rare patients with this disorder; only 3 of the 41 reported pediatric cases had initial symptoms of AAI or worsening of symptoms after trauma during organized sports participation. Members of the SO Medical Advisory Committee think that more such sports-related injuries occur but that they are being overlooked because of a lack of information about the association of AAI and spinal cord injury among health care providers. This claim has not been substantiated with published research....

TENTATIVE CONCLUSIONS:

....it is reasonable to conclude that lateral neck X-rays are of potential but unproven value in detecting patients at risk for developing spinal cord injury during sports participation. It seems that identification of those patients who already have or who later have complaints or physical findings consistent with symptomatic spinal cord injury is a greater priority than obtaining X-rays. Recognition of these symptomatic patients is challenging and requires frequent interval histories and physical exams, including evaluations before participation in sports, preferably by physicians who have cared for these patients longitudinally. Their parents must learn the symptoms of AAI that indicate the need to seek immediate medical care.

The SO does not plan to remove its requirement that all athletes with DS receive neck X-rays. Pediatricians will therefore continue to be called on to order these tests. The information here can be used to interpret the results for family members...." (*end excerpt*)

Dr. Siegfried Pueschel wrote his opposition to the revised AAP statement in the Jan 1998 issue of the journal *Archives of Pediatric and Adolescent Medicine*.

Dr. Pueschel is the head of the DS Clinic in Providence, Rhode Island, and the author of several studies and textbooks on Down syndrome. In his article, Dr. Pueschel's main points are:

1. While the X-ray may not be as good of a screen as we'd like, there is currently nothing better.
2. AAI is not rare: it occurs in children with DS (10-30%) and symptomatic AAI may reach up to 1 to 2% of all children with DS.
3. Symptomatic AAI is a serious disorder, which justifies the work and expense required to detect it.
4. While it isn't known if asymptomatic AAI turns into symptomatic AAI, it hasn't been disproven yet, either.
5. To date, there have been no reports of spinal cord injury from any activity associated with Special Olympics, Inc. This may mean that such an injury *is* a rare occurrence, or it may actually show that SO's precautionary measures are effective at preventing such injuries.
6. If one waits for significant neurologic signs to appear, spinal cord damage may have already occurred. By waiting, an individual at risk with no symptoms will not be detected.
7. Further, lateral neck X-rays may also detect the less common but more serious atlanto-occipital instability, or degenerative changes in the cervical spine.

Dr. Pueschel concludes, therefore, that lateral neck X-rays are still an important part of optimal care for individuals with DS.

Also in the Jan 1998 issue of the journal *Archives of Pediatric and Adolescent Medicine* is an editorial by Dr. Bill

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Cohen on the controversy.

Dr. Cohen is the head of the DS Clinic of the Children's Hospital of Pittsburgh and co-chair of the Down Syndrome Medical Interest Group, a collective of health professionals dedicated to the care of individuals with DS. Dr. Cohen summarizes both the 1995 AAP statement and Dr. Pueschel's statement, and then addresses the controversy thusly:

"If in fact asymptomatic AAI is not the precursor to symptomatic AAI, the current protocol [X-rays at 3, 12, and 18 years of age] should be abandoned. I would suggest that the devastating nature of cord compression and the technical difficulties in assessing children with developmental disabilities has led to the current quandary. Few organizations would be willing to take responsibility, however limited, for a recommendation that might lead or be perceived to lead to steps that would fail to protect individuals with DS from a spinal cord injury."

Dr. Cohen goes on to cite a recent article addressing the technical aspects of measuring for AAI, and adds that in his opinion, children with DS who have a narrowed neural canal or evidence of marked AAI should receive an MRI of the neck before restriction of activity or any surgical procedure requiring anesthesia. He ends his editorial by calling for a consensus meeting involving representatives of all medical fields that this topic encompasses.
