Atlantoaxial Instability in Down Syndrome

Some issues related to participation in certain sports by persons with Down syndrome require clarification.

Since 1965 there have been occasional reports about a condition described at various times as instability, subluxation, or dislocation of the articulation of the first and second cervical vertebrae (atlantoaxial joint) among persons with Down syndrome.\(^1\)\(^\text{-}\)\(^15\) This condition has also been found in patients with rheumatoid arthritis,\(^16\)\(^,\)\(^17\) abnormalities of the odontoid process of the second cervical vertebra,\(^4\)\(^,\)\(^5\)\(^,\)\(^12\)\(^,\)\(^13\)\(^,\)\(^15\) and various forms of dwarfism.\(^18\)

Atlantoaxial (C-1, C-2) instability has not attracted general attention because clinical manifestations are rare and the condition is limited to a small portion of the population. The incidence of atlantoaxial instability among persons with Down syndrome has been reported by various observers to be 10% to 20%.\(^2\)\(^,\)\(^8\)\(^,\)\(^15\) When atlantoaxial instability results in subluxation or dislocation of C-1 and C-2, the spinal cord also may be injured. This is a rare but serious complication.

In March 1983, the Special Olympics, Inc, sponsors of a nationwide competitive athletic program for developmentally disabled persons, without prior announcement, mandated for participants with Down syndrome special precautions to prevent serious neurologic consequences from stress on the head and neck in sports competition.\(^19\) Although thousands of persons with Down syndrome have taken part in sports events during the 15-year history of the Special Olympics without a known occurrence of neurologic complications due to participation, the new directive requires all persons with Down syndrome who wish to participate in certain sports that might involve stress on the head and neck (gymnastics, diving, pentathlon, butterfly stroke in swimming, diving start in swimming, high jump, soccer, and warm-up exercises that place undue stress on the head and neck muscles) to have a medical examination, lateral-view roentgenograms of the upper cervical region in full flexion and extension, and certification by a physician that the examination did not reveal atlantoaxial instability or neurologic disorder. Failure either to comply or to have medical certification would result in exclusion from the above-specified sports.

Parents, physicians, and sports authorities were understandably surprised by the immediacy of the edict. Many parents were resentful because of the short time for screening, the cost of the examinations, and discovery that most physicians did not know about the directive or were not aware of the atlantoaxial syndrome. Some radiologists were not familiar with exact procedures for screening. In general, physicians were perplexed by the sudden concern about a condition that had never been a problem among the largest group of disabled participants during 15 seasons of the Special Olympics.

There are no national statistics to confirm the extent of screening in 1983, but valiant efforts were made to comply with the directive during the 6-week interval allowed for the procedures. It has been stated that there were no reported casualties due to atlantoaxial instability in the Special Olympics last year. However, some participants were barred from the specified events.

Atlantoaxial (C-1, C-2) instability is a manifestation of the generalized poor muscle tone and joint laxity commonly found in persons with Down syndrome. The instability is due to (1) laxity of the transverse ligament that holds the odontoid process of the axis (C-2) in place against the inner aspect of the anterior arch of the atlas (C-1), maintaining integrity of the C-1, C-2 articulation or (2) abnormalities of the odontoid, such as hypoplasia, malformation, or complete absence.\(^4\)\(^,\)\(^5\)\(^,\)\(^8\)\(^,\)\(^13\)\(^,\)\(^15\) These conditions allow some leeway between the odontoid and the atlas, especially during flexion and extension of the neck. This results in a "loose joint." In extreme cases, the first cervical vertebra slips forward and the spinal cord is vulnerable to compression by the odontoid process of C-2 anteriorly or by the arch of C-1 posteriorly.

Measurement of the distance between the odontoid process of C-2 and the anterior arch of C-1 is the most reliable test for the diagnosis of atlantoaxial instability. This measurement is possible only in a lateral roentgenogram of the head, neck, shoulders, and upper chest. A normal distance of the sum of approximately 15 mm between the tip of the odontoid and the arch of the atlas implies normal alignment of the atlantoaxial articulation. When this measurement becomes less than 15 mm, the atlantoaxial joint is considered unstable. Radiographic evaluation of the atlantoaxial region should be performed if there is suspicion of atlantoaxial instability or neurologic disorder in persons with Down syndrome who wish to participate in certain sports.
toid process and the anterior arch of the atlas on lateral roentgenograms in the neutral, flexion, and extension positions is the only way to detect atlantoaxial instability.\textsuperscript{9,14,20}

Although simple laxity and instability seldom lead to subluxation or dislocation, it has become apparent, as physicians learn more about atlantoaxial instability, that the latent condition must be viewed as a factor predisposing to neurologic complications. Detection of an abnormal space between the odontoid and the anterior arch of the atlas is a signal for precautionary measures to avoid hyperflexion or hyperextension of the neck and extreme rotation of the head.

The neurologic manifestations of spinal compression from the above causes include fatigue in walking, gait disturbance, progressive clumsiness and incoordination, spasticity, hyperreflexia, clonus, toe-extensor reflex, and other upper motor neuron and posterior column signs and symptoms from compression of the spinal cord. Onset of neck pain, head tilt, and torticollis in Down syndrome are indicative of malposition of the odontoid. Development, and particularly, progression of these neurologic signs or symptoms in a person with Down syndrome suggest atlantoaxial subluxation. Strenuous activity should be curtailed and diagnosis and management undertaken promptly.

It is very likely that many schools, recreation and rehabilitation programs, and camps in which developmentally disabled persons are enrolled will follow the example of the Special Olympics in requiring careful screening of all persons with Down syndrome before participation in activities that could result in flexion and hyperextension is permitted. Undoubtedly, pediatricians, other primary care physicians, and radiologists will be called upon to screen and authorize participation.

**RECOMMENDATIONS**

The Committee on Sports Medicine, after consultation with the Sections on Neurology, Orthopaedics, and Radiology, recommends the following guidelines:

1. All children with Down syndrome who wish to participate in sports that involve possible trauma to the head and neck should have lateral-view roentgenograms of the cervical region in neutral, flexion, and extension positions within the patient’s tolerance before beginning training or competition. This recommendation applies to all participants in the high-risk sports who have not previously had normal findings on cervical roentgenograms.

Some physicians may prefer to screen all patients with Down syndrome routinely at 5 to 6 years of age to rule out atlantoaxial instability.

2. When the distance between the odontoid process of the axis and the anterior arch of the atlas exceeds 4.5 mm or the odontoid is abnormal, there should be restrictions on sports that involve trauma to the head and neck, and the patient should be followed up at regular intervals.

3. At the present time, repeated roentgenograms are not indicated for those who have previously had normal findings. Indications for repeated roentgenograms will be defined by research.

4. Persons with atlantoaxial subluxation or dislocation and neurologic signs or symptoms should be restricted in all strenuous activities, and operative stabilization of the cervical spine should be considered.\textsuperscript{21-23}

5. Persons with Down syndrome who have no evidence of atlantoaxial instability may participate in all sports. Follow-up is not required unless musculoskeletal or neurologic signs or symptoms develop.

**COMMITTEE ON SPORTS MEDICINE, 1983–1984**

Thomas E. Shaffer, MD, Chairman
Paul G. Dyment, MD
Eugene F. Luckstead, MD
John J. Murray, MD
Nathan J. Smith, MD

Liaison Representatives
James H. Moller, MD
Section on Cardiology
David M. Orenstein, MD
Section on Diseases of the Chest
Arthur M. Pappas, MD
Section on Orthopaedics
Frederick W. Baker, MD
Canadian Paediatric Society
Richard Malacre
National Athletic Trainers Association

Consultants
E. Dennis Lyne, MD, Chairman
Section on Orthopaedics
Gerald Erenberg, MD, Chairman
Section on Neurology
Bruce R. Parker, MD, Chairman
Section on Radiology
Albert C. Fremont, Chairman
Committee on Children with Disabilities

**REFERENCES**

Atlantoaxial Instability in Down Syndrome

*Pediatrics* 1984;74:152

<table>
<thead>
<tr>
<th>Updated Information &amp; Services</th>
<th>including high resolution figures, can be found at: /content/74/1/152</th>
</tr>
</thead>
<tbody>
<tr>
<td>Permissions &amp; Licensing</td>
<td>Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at: /site/misc/Permissions.xhtml</td>
</tr>
<tr>
<td>Reprints</td>
<td>Information about ordering reprints can be found online: /site/misc/reprints.xhtml</td>
</tr>
</tbody>
</table>