

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.¹⁻¹⁵ 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%.^{2,9,15} 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.^{4,5,9,13,15} 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 odon-

This statement has been approved by the Council on Child and Adolescent Health.
PEDIATRICS (ISSN 0031 4005). Copyright © 1984 by the American Academy of Pediatrics.

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.^{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.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 Malacrea

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

1. Tishler JM, Martel W: Dislocation of the atlas in mongolism: Preliminary report. *Radiology* 1965;84:904-906
2. Martel W, Tishler JM: Observations on the spine in mongolism. *AJR* 1966;97:630-638
3. Dzenitis AJ: Spontaneous atlanto-axial dislocation in a mongoloid with spinal cord compression: Case report. *J Neurosurg* 1966;25:458-460

4. Sherk HH, Nicholson JT: Rotatory atlanto-axial dislocation associated with ossiculum terminale and mongolism. *J Bone Joint Surg* 1969;51-A:957-963
5. Martel W, Uyham R, Stimson CW: Subluxation of the atlas causing spinal cord compression in a case of Down's syndrome with a "manifestation of an occipital vertebra." *Radiology* 1969;93:839-840
6. Gerard Y, Segal P, Bedoucha JS: L'instabilité de l'atlas sur l'axis dans le mongolisme. *Presse Med* 1971;79:573-575
7. Aung MH: Atlanto-axial dislocation in Down's syndrome: Report of a case with spinal cord compression and review of the literature. *Bull Los Angeles Neurol Soc* 1973;39:197-201
8. Finerman GAM, Sakai D, Weingarten S: Atlanto-axial dislocation with spinal cord compression in a mongoloid child: A case report. *J Bone Joint Surg* 1976;58-A:408-409
9. Semine AA, Ertel AN, Goldberg MJ, et al: Cervical spine instability in children with Down syndrome (trisomy 21). *J Bone Joint Surg* 1978;60-A:649-652
10. Whaley WJ, Gray WD: Atlantoaxial dislocation and Down's syndrome. *Can Med Assoc J* 1980;123:35-37
11. Shield LK, Dickens DRV, Jensen F: Atlanto-axial dislocation with spinal cord compression in Down syndrome. *Aust Paediatr J* 1981;17:114-116
12. Hungerford GD, Akkaraju V, Rawe SE, et al: Atlanto-occipital and atlanto-axial dislocations with spinal cord compression in Down's syndrome: A case report and review of the literature. *Br J Radiol* 1981;54:758-761
13. Hreidarsson S, Magram G, Singer H: Symptomatic atlanto-axial dislocation in Down syndrome. *Pediatrics* 1982;69:568-571
14. Coria F, Quintana F, Villalba M, et al: Craniocervical abnormalities in Down's syndrome. *Dev Med Child Neurol* 1983;25:252-255
15. Pueschel SM, Scola FH, Perry CD: Atlanto-axial instability in children with Down syndrome. *Pediatr Radiol* 1981;10:129-132
16. Stevens JC, Cartledge NEF, Saunders M, et al: Atlanto-axial subluxation and cervical myelopathy in rheumatoid arthritis. *Q J Med* 1971;40:391-408
17. Herring JA: Cervical instability in Down's syndrome and juvenile rheumatoid arthritis. *J Pediatr Orthop* 1982;2:205-207
18. Kopits SE, Perovic MN, McKusick V, et al: Congenital atlanto-axial dislocations in various forms of dwarfism. *J Bone Joint Surg* 1972;54:1349-1350
19. *Special Olympics Bulletin: Participation by individuals with Down syndrome who suffer from atlantoaxial dislocation condition.* Washington, DC, Special Olympics Inc, March 31, 1983
20. Locke GR, Gardner JI, Van Epps EF: Atlas-dens interval (ADI) in children: A survey based on 200 normal cervical spines. *AJR* 1966;135-140
21. Giblin PE, Micheli LJ: Management of atlanto-axial subluxation with neurologic involvement in Down syndrome: A report of two cases and review of the literature. *Clin Orthop* 1979;140:66-71
22. Spierings ELH, Braakman R: The management of os odontoideum: Analysis of 37 cases. *J Bone Joint Surg* 1982;64-B:422-428
23. Diamond LS, Lynne D, Sigman B: Orthopedic disorders in patients with Down's syndrome. *Orthop Clin North Am* 1981;12:57-71

Atlantoaxial Instability in Down Syndrome
Pediatrics 1984;74;152

Updated Information & Services

including high resolution figures, can be found at:
<http://pediatrics.aappublications.org/content/74/1/152>

Permissions & Licensing

Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at:
<http://www.aappublications.org/site/misc/Permissions.xhtml>

Reprints

Information about ordering reprints can be found online:
<http://www.aappublications.org/site/misc/reprints.xhtml>

American Academy of Pediatrics

DEDICATED TO THE HEALTH OF ALL CHILDREN®



PEDIATRICS®

OFFICIAL JOURNAL OF THE AMERICAN ACADEMY OF PEDIATRICS

Atlantoaxial Instability in Down Syndrome *Pediatrics* 1984;74;152

The online version of this article, along with updated information and services, is located on the World Wide Web at:

<http://pediatrics.aappublications.org/content/74/1/152>

Pediatrics is the official journal of the American Academy of Pediatrics. A monthly publication, it has been published continuously since 1948. Pediatrics is owned, published, and trademarked by the American Academy of Pediatrics, 345 Park Avenue, Itasca, Illinois, 60143. Copyright © 1984 by the American Academy of Pediatrics. All rights reserved. Print ISSN: 1073-0397.

American Academy of Pediatrics

DEDICATED TO THE HEALTH OF ALL CHILDREN®

