Atlantoaxial Instability in Down Syndrome: Subject Review

Committee on Sports Medicine and Fitness

DEFINITION OF THE PROBLEM

In 1984, the American Academy of Pediatrics (AAP) published a position statement on screening for atlantoaxial instability (AAI) in youth with Down syndrome.¹ In that statement, the AAP supported the requirement introduced by the Special Olympics in 1983 that lateral neck radiographs be obtained for individuals with Down syndrome before they participate in the Special Olympics’ nationwide competitive program for developmentally disabled persons. Those participants with radiologic evidence of instability are banned from certain activities that may be associated with increased risk of injury to the cervical spine. This policy seemed to be prudent in light of the information available at that time. However, the AAP Committee on Sports Medicine and Fitness recently has reviewed the data on which this recommendation was based and has decided that uncertainty exists concerning the value of cervical spine radiographs in screening for possible catastrophic neck injury in athletes with Down syndrome. The 1984 statement therefore has been retired. This review discusses the available research data on this subject.

BACKGROUND

AAI, also called atlantoaxial subluxation, denotes increased mobility at the articulation of the first and second cervical vertebrae (atlantoaxial joint). This condition is found not only in patients who have Down syndrome but also in some patients who have rheumatoid arthritis, abnormalities of the odontoid process of the axis, and various forms of dwarfism.¹ The causes of AAI are not well understood but may include abnormalities of the ligaments that maintain the integrity of the C-1 and C-2 articulation, bony abnormalities of C-1 or C-2, or both.¹⁻¹¹

In its mildest form, AAI is asymptomatic and is diagnosed using radiography. The instability is recognized through lateral neck radiographs in which the excessive mobility of C-2 in relation to C-1 results in an abnormally large distance between the odontoid process of the axis and the anterior arch of the atlas. Symptomatic AAI results from subluxation that is severe enough to injure the spinal cord, or from dislocation at the atlantoaxial joint.

Approximately 15% of individuals in the pediatric age group (<21 years old) who have Down syndrome also have AAI.² Almost all of the persons affected are asymptomatic. Some asymptomatic individuals with Down syndrome who have normal radiographs initially will have abnormal radiographs obtained later, and others with initially abnormal radiographs will have normal follow-up radiographs; the latter change is apparently more common. In one study, 7 of 95 (7.4%) patients with normal radiographs initially had abnormal radiographs 3 to 6 years later in follow-up, and 19 of 95 (20%) patients with abnormal radiographs initially had normal second radiographs.² In another longitudinal study of 141 patients,³ 130 (92%) had changes in the atlantoaxial interval (of less than 1.5 mm) that were judged clinically insignificant. Eleven patients (8%) had changes that ranged from 2 to 4 mm. Nine of these patients with abnormal radiographs had subsequent radiographs that were normal, whereas the two patients with normal radiographs had abnormal radiographs on follow-up.

The neurologic manifestations of symptomatic AAI include easy fatigability, difficulties in walking, abnormal gait, neck pain, limited neck mobility, torticollis or head tilt, incoordination and clumsiness, sensory deficits, spasticity, hyperreflexia, clonus, extensor-plantar reflex, and other upper motor neuron and posterior column signs and symptoms.¹⁻¹¹ Such symptoms and signs often remain relatively stable for months or years; occasionally they progress, rarely even to paraplegia, hemiplegia, quadriplegia, or death.¹⁻¹¹ Trauma rarely causes the initial appearance or the progression of these symptoms.¹⁻¹¹ Nearly all of the individuals who have experienced catastrophic injury to the spinal cord have had weeks to years of preceding, less severe neurologic abnormalities.¹⁻¹¹

The Special Olympics’ radiologic screening program to prevent catastrophic spinal cord injury has the following characteristics. Lateral spinal cord radiographs are used to screen for asymptomatic AAI, which is thought to be a risk factor for symptomatic AAI. Asymptomatic individuals are banned from
participating in several activities that are thought to have a particularly high risk of spinal cord injury—in particular, gymnastics, diving, the pentathlon, the butterfly stroke and diving starts in swimming, the high jump, soccer, and certain warm-up exercises.

For this or any screening program to be worthwhile, several criteria must be met.12 The target disease, symptomatic AAI, must be sufficiently common and severe enough to justify the work and expense of its detection in the asymptomatic phase. The screening test must have excellent sensitivity and specificity for identifying those with asymptomatic AAI, and asymptomatic AAI must be a proven risk factor for symptomatic AAI. An effective intervention must be available to prevent the progression from asymptomatic to symptomatic disease. The following discussion evaluates how well a radiologic screening program for AAI meets these criteria.

Most importantly, symptomatic AAI is apparently rare in individuals with Down syndrome. In the pediatric age group, only 41 well-documented cases have been described in the published literature (Table).5-11 New case series or reports on such patients are not likely to be published, however, because symptomatic AAI is no longer remarkable.

One study has shown that the reproducibility of the radiologic test results for AAI is relatively poor.4 Of 19 children who were evaluated twice within a 10-minute interval, 6 had evidence of AAI. This instability was demonstrated in the first set of films in 5 patients and in the second set in 4 patients. Only 3 of 6 patients had abnormalities on both sets of radiographs. As described previously, other studies have shown that an individual’s radiologic status can change over time, most often from abnormal to normal.23 These changes occur either because of the lack of reproducibility of the test or because of actual variations in atlantoaxial stability over time.23

Asymptomatic AAI, which is common, has not been proven to be a significant risk factor for symptomatic AAI, which is rare. In prospective studies of 952 and 141 individuals with Down syndrome and AAI, one asymptomatic patient became symptomatic during 3 or more years of follow-up after an injury.3

In the 41 reported pediatric cases with symptomatic AAI,25-11 only one patient had had radiographs obtained before neurologic problems developed,25-11 making it impossible to know whether the other 40 had asymptomatic AAI before they became symptomatic. This individual5 had a normal radiograph at 6 years of age but became quadriplegic after a tumbling accident at 18 years of age. A radiograph obtained after the injury demonstrated 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 patients (Table) had initial symptoms of AAI or worsening of symptoms after trauma during organized sports participation.25-11 Members of the Medical Advisory Committee of the Special Olympics 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.13

### Table. Summary of Well-Described Symptomatic Cases of AAI in the Medical Literature

<table>
<thead>
<tr>
<th>Category</th>
<th>Number of Cases</th>
<th>Details</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total number of symptomatic patients with AAI in the pediatric age group (&lt;21 y):</td>
<td>56</td>
<td>Ref 5: 4 patients, 2 patients had symptoms after cardiac surgery: neck pain (1 patient) and gait problems (1 patient), 1 patient became quadriplegic while practicing for the Special Olympics (the circumstances of the injury were unclear), 1 patient had progressive gait problems after a fall, 1 patient had hemiparesis after eye surgery</td>
</tr>
<tr>
<td>Number of patients whose symptoms became worse after injury:</td>
<td>14</td>
<td>Ref 5: 2 patients, 1 patient had gradually worsening hemiparesis, made much worse after a fall, 1 patient had hyperreflexia and upgoing toes and became paraplegic after a trampoline accident</td>
</tr>
<tr>
<td>Number of patients who had radiographs obtained before they developed symptomatic AAI:</td>
<td>30</td>
<td>Refs 6-11: no patients</td>
</tr>
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</table>

Summary: Significant concern about sports-related AAI.

The arguments for continued screening of patients with Down syndrome include the theoretical possibility of preventing the rare occurrence of sports-related catastrophic spinal cord injury among individuals with asymptomatic AAI. Another purpose is to identify the very rare previously unrecognized patient with symptomatic AAI. Arguments against screening include the rarity of symptomatic AAI, and the lack of information about the association of AAI with sports trauma.
inaccuracy in the screening test, the possibility that patients with abnormal radiographs initially will have normal radiographs later on, and the absence of evidence that the screening program is effective in preventing symptomatic disease. Screening is also expensive. The expense may prevent at least some individuals from participating in the Special Olympics, and the labeling of patients as having AAI may lead to anxiety and to unnecessary medical intervention, although no research has evaluated these possibilities.

The case reports2-11 indicate that many patients with symptomatic AAI have symptoms and signs of cervical spinal cord compression for weeks or years before they are recognized as having neurologic disease. Current evidence suggests that the presence of these neurologic abnormalities may be more predictive of potential progression of injury than are abnormalities on radiographs in the asymptomatic patient.2-11 Therefore, despite the difficulties in obtaining good medical histories and physical and neurologic examinations for individuals with Down syndrome, it is clearly important that they receive these evaluations before participation in sports. If possible, these patients should be examined by physicians who have provided their care longitudinally and know their baseline status. In addition, their families must be made aware of the manifestations of symptomatic AAI and instructed to contact their physician immediately if any symptoms appear.

SUMMARY OF NEW OR EMERGING DATA

No recent studies have provided significant new information on the recognition of those asymptomatic patients with Down syndrome and AAI who are at increased risk for spinal cord injury. Research has clarified that computed tomography, as expected, gives more detailed information about bony anomalies and spinal cord compression than do plain radiographs.14 Other reports have emphasized that other abnormalities of the cervical spine, in particular atlanto-occipital instability, occur in patients with Down syndrome.15 One study found that children with Down syndrome and asymptomatic AAI who were allowed to play all sports had no serious spinal cord injuries or evidence of neurologic deterioration.16 The number of subjects was too small, and the duration of follow-up was too brief for this to be conclusive evidence of a lack of risk. In a community study of adults with Down syndrome, those with AAI shown on radiographs were not more likely to have neurologic symptoms suggesting spinal cord injury than those without evidence of AAI.17 Studies have continued to explore the effect of technique on the measurement of atlantoaxial distance.14,18

TENTATIVE CONCLUSIONS

From the available scientific evidence, it is reasonable to conclude that lateral plain radiographs of the cervical spine 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 radiographs. Recognition of these symptomatic patients is challenging and requires frequent interval histories and physical examinations, 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 Special Olympics does not plan to remove its requirement that all athletes with Down syndrome receive radiographs of the cervical spine. Pediatricians therefore will continue to be called on to order these tests. The information in this review can be used to interpret the results for family members.

Clearly, we need better research to determine what combination of symptoms, signs, and findings from imaging studies best identifies those individuals with Down syndrome who are at increased risk of a catastrophic injury to the spinal cord during sports participation. The Special Olympics and the regional centers that care for large numbers of children and adolescents with Down syndrome are in a favorable position to study this problem prospectively in a multicenter study and perhaps by establishing a national injury registry.
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