Chronic Atlanto-Axial Instability in Down Syndrome*

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ABSTRACT: We studied the radiographs of thirty-two patients with Down syndrome for evidence of atlanto-axial instability. One of the patients had instability in 1970 and seven had it in 1983. The interval between the atlas and the odontoid process in the patients who demonstrated motion at that interval radiographically averaged 2.78 millimeters in 1970 and 6.93 millimeters in 1983 (p < 0.005). Four patients whose radiographs showed atlanto-axial motion in 1970 lost that motion by 1983, and in seven patients who did not show atlanto-axial instability in 1970 it developed by 1983. Atlanto-axial instability was more likely to develop in boys who were more than ten years old. Accessory upper-cervical ossicles became evident in three patients, none of whom had atlanto-axial motion. However, one of these three patients had an abnormally wide atlanto-axial interval.

The atlanto-axial segments of the cervical spine in children with Down syndrome have a predilection for the development of localized anomalies that are of particular interest because of the danger of atlanto-axial dislocation. In several studies, groups of such children have been surveyed for the incidence of anomalies in the upper cervical spine that might lead to instability. However, all of these studies were limited by the fact that they were not longitudinal — that is, they only described the incidence of abnormalities in a population with Down syndrome and did not attempt to study the natural history of either the anomalies or atlanto-axial instability.

In 1983, at the Pinecrest State School, we screened 140 patients with Down syndrome for evidence of upper cervical instability. The study was prompted by an acute atlanto-axial dislocation that resulted in quadriplegia in a child at that institution (Figs. 1-A, 1-B, and 1-C). Thirty-two patients were identified as having been screened radiographically for atlanto-axial instability approximately thirteen years earlier. The cases of these thirty-two patients are described in the present report.

Methods and Materials

All of the radiographs in 1970 had been obtained as part of a screening program for atlanto-axial instability. In 1983, each patient had a physical examination in which we noted neurological status, associated physical anomalies, date of birth, sex, race, activity level, and intelligence. The radiographs that we made of the upper cervical spine included flexion-extension radiographs.

Despite the presence of hyperlaxity of all ligaments in all of the patients, we observed no correlation between the severity of the laxity and the presence of either instability or anomalies, nor was there a correlation between those abnormalities and hallux varus (present in thirteen patients), subluxating patellae (present in two patients), or cardiac disease (present in five patients).

Intelligence quotient and activity level similarly were unrelated to the presence of either instability or osseous anomalies. No patient, other than the one with quadriplegia (the index case) (Figs. 1-A, 1-B, and 1-C), showed any evidence of neurological dysfunction related to possible compression of the cervical spinal cord.

Each set of 1983 radiographs was classified in one of three categories based on the atlanto-axial measurements and stability, and the presence or absence of an ossicle in the atlas was also recorded. If the atlanto-axial interval measured more than four millimeters in a patient who was fifteen years or younger, or more than three millimeters in one who was older than fifteen, it was judged to be abnormal. Excessive motion at the atlanto-axial level meant instability. The radiographs were therefore classified as either normal, stable, or unstable. A normal classification indicated an interval of less than three or four millimeters with no motion on lateral flexion-extension radiographs and no ossicle on the atlas. A stable classification indicated at least one millimeter of atlanto-axial motion and a maximum interval of less than three or four millimeters. An unstable classification meant more than three or four millimeters of motion, or a fixed interval of more than five millimeters.

Except for the one patient who died of atlanto-axial dislocation (Case 1; Figs. 1-A, 1-B, and 1-C) all of the patients were still living at the time of writing.
Results

In 1970, the findings were classified as normal in twenty-six sets of radiographs, as stable in five, and as unstable in one. The analogous numbers in 1983 were nineteen, three, and seven. In three patients ossicles developed: two were stable and one had a fixed interval of six millimeters.

In 1970, seven of the patients had some motion at the atlanto-axial level, the mean being 2.78 ± 0.95 millimeters, but only one of them met our criterion for instability (that is, more than a four-millimeter interval when younger than sixteen). In seven other patients the average atlanto-axial motion in 1983 was 6.93 ± 4.04 millimeters (Table 1). Evaluation by the Student t test demonstrated that the difference between these two groups was statistically significant at the p < 0.005 level. In 1983 four of the patients who had some motion in 1970 had no evidence of motion, including the patient (Case 4) who had been considered to have instability on the 1970 radiographs. Of the seven patients who had instability in 1983, six had had no motion in 1970 and one had had less than a three-millimeter interval in 1970. Nineteen other patients who had normal radiographs in 1970 remained unchanged (stable).

The average age in 1970 of the group that had improvement was 9.25 years, and it was 18.16 years in the group that had deterioration. The difference in age was significant at the p < 0.05 level. Being male and older than ten years statistically increased the risk of increasing instability (Cases 2, 3, 6, and 7) (chi-square test, p < 0.05).

The one patient (Case 4) who had instability in 1970 was radiographically normal in 1983.

Discussion

Instability

Tishler and Martel reported that four of their nineteen patients with Down syndrome had atlanto-axial dislocation, but they did not report the range of atlanto-axial motion. Based on several reports on normal individuals, we defined the normal atlanto-axial interval of adults as less than three millimeters and that of children between the ages of one month and sixteen years as less than four millimeters. Martel et al. stated that five millimeters is the upper limit of normal in skeletally immature individuals with Down syndrome. They formulated this five-millimeter limit because no neurological damage was encountered with this amount of atlanto-axial motion. However, there is no record of a long-term evaluation of...
TABLE 1

DATA ON PATIENTS WITH ATLANTO-AXIAL INSTABILITY IN 1970 OR 1983

<table>
<thead>
<tr>
<th>Case</th>
<th>Sex</th>
<th>Age in 1970 (Yrs.)</th>
<th>Radiographic Classification in 1970*</th>
<th>Radiographic Classification in 1983*</th>
<th>Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>M</td>
<td>6</td>
<td>Normal</td>
<td>Unstable (&gt;1 cm)</td>
<td>Died of atlanto-axial dislocation</td>
</tr>
<tr>
<td>2</td>
<td>M</td>
<td>12</td>
<td>Normal</td>
<td>Unstable (9 mm)</td>
<td>Asymptomatic</td>
</tr>
<tr>
<td>3</td>
<td>M</td>
<td>13</td>
<td>Stable (3 mm)</td>
<td>Unstable (6 mm)</td>
<td>Asymptomatic</td>
</tr>
<tr>
<td>4</td>
<td>F</td>
<td>16</td>
<td>Unstable (4.5 mm)</td>
<td>Normal</td>
<td>Activities restricted after 1970</td>
</tr>
<tr>
<td>5</td>
<td>F</td>
<td>9</td>
<td>Normal</td>
<td>Unstable (6 mm)</td>
<td>Asymptomatic</td>
</tr>
<tr>
<td>6</td>
<td>M</td>
<td>15</td>
<td>Normal†</td>
<td>Unstable (5 mm)</td>
<td>Asymptomatic</td>
</tr>
<tr>
<td>7</td>
<td>M</td>
<td>57</td>
<td>Normal†</td>
<td>Unstable (6 mm)</td>
<td>Also had ossicle, atlanto-axial fusion performed</td>
</tr>
<tr>
<td>8</td>
<td>M</td>
<td>8</td>
<td>Normal</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

* Size of atlanto-axial interval is in parentheses.
† Radiographs made in 1975.

the patients of Martel et al., and their study was too small to evaluate statistically. From our study, we concluded that an atlanto-axial interval of more than four millimeters is at the upper limit of adequate mechanical stability of the atlanto-axial joint, and that interval should be the maximum in children who are less than sixteen years old. Our standard for instability in patients older than sixteen years is an interval greater than three millimeters. Based on these criteria, we found only one instance of instability on the original radiographs of our thirty-two patients, but there was a 22 per cent incidence (seven patients) at follow-up after thirteen years. These data agree with those of other reported series. In the present series, the magnitude of the atlanto-axial subluxation increased with age. This represents a reversal of the trend in normal individuals toward increased stability (that is, narrowing of the atlanto-axial interval) with maturation. Evaluation of the patients who showed increasing instability indicated that they usually were more than ten years old at the time of the initial evaluation. Patients who were less than ten years old at the first evaluation were more likely to improve (p < 0.05). This probably reflects the fact that younger patients have more physiological motion at the atlanto-axial joint.

Ossicles

Several authors have noted an increased incidence of osseous anomalies of the upper cervical spine associated with Down syndrome. Accessory ossicles, mostly ossicula terminale or occipital vertebrae, have been reported. Interestingly, the case described by Finerman et al. and one of the cases described by Sherk and Nicholson had previous radiographs that did not show these osseous anomalies, suggesting that these ossicles developed in late childhood.

The three patients in our series who had accessory ossicles in 1983 did not have them on the 1970 radiographs, which also suggests that these ossicles may develop late in childhood.

All three of our patients in whom an ossicle developed in the atlas by 1983 clearly had a normal apophysis of the upper part of the odontoid process in 1970 (Figs. 2-A and 2-B). Hefke demonstrated that 93 per cent of children with
Down syndrome reach skeletal maturity at or before normal
children\(^1\), so any congenital accessory ossicle should have
been visible when the initial radiographs were made. The
1983 radiographs of the three patients with an ossicle all
showed an odontoid process of normal length with an ossicle
superior to its tip; therefore, the presence of an ossicle above
the odontoid process suggests that there had been an avulsion
fracture of the upper end of the odontoid process. This
implies that there may have been instability of the atlanto-
axial level at some time, despite the fact that no atlanto-
avarial motion was evident in 1983 in any of the three patients.
It appears that the degree of atlanto-axial instability in pa-
tients with Down syndrome increases with age, as was seen
in our Cases 1, 2, 3, and 5 through 8, and that the ossicles
may be associated with transient instability.

In several case reports, the instability was reported to
be symptomatic for more than a year\(^2\). Sherker and Nicholson
reported the case of a fifteen-year-old with a non-reducible
atlanto-axial dislocation and a progressive neurological
lesion\(^27\). Other cases have been reported of defects pro-
gressing over several months\(^5\,7\,28\,31\). Nordt and Stauffer
reported neurological deficits complicating sublaminar wiring
in two patients with irreducible atlanto-axial dislocation\(^22\).
Several of their patients with neurological defects had the
osseous lesions of os odontoideum, ossiculum terminale, or
an occipital vertebra. In some, irreducible atlanto-axial dis-
location with spinal cord compression developed\(^6\,13\,20\,23\,27\).
It has been previously noted that these upper cervical os-
seous anomalies are commonly associated with gradually
progressive neurological symptoms\(^16\,18\,30\). This may be ex-
pected if the osseous lesion is the result and not the cause
of the instability.

The etiology of the upper cervical instability in patients
with Down syndrome is most likely a defect in collagen
that is manifested by generalized ligament laxity. The weak
connective tissue is susceptible to injury and stretching. The
combination of an increasing incidence of injury with minor
trauma and increasing mechanical disadvantage at a joint
because of a lengthened fibrous structure leads to progres-
sively increasing instability of the joint with minor trauma.
The collagen of the structures supporting the odontoid pro-
cess may be affected in that way. Therefore, patients with
Down syndrome in whom the odontoid process is not well
supported may be susceptible to an avulsion fracture. Also,
decreased tensile strength in the growth plate during the
adolescent growth spurt may lead to such an injury at that
time of life.

It is interesting to note that one patient (Case 4) with
an atlanto-axial interval of 4.5 millimeters in 1970 was
restricted from participation in sports and showed a stable
interval by 1983. Two patients (Cases 1 and 8) have under-
gone posterior fusion of the first and second cervical ver-
tebrae for instability, and one of them died of complications
of quadriplegia incurred at the time of atlanto-axial dislo-
cation. The dislocations that were surgically treated could
not be completely reduced. This reinforces the hypothesis
that the atlanto-axial instability in patients with Down syn-
drome is a chronic and progressive process rather than an
acute dislocation.

The chronic subluxation at the level of instability may
explain the sudden postoperative respiratory arrest that oc-
casionally is associated with reduction and fusion of the first
and second cervical vertebrae\(^1\,13\,20\,23\,27\), because impinge-
ment of the spinal canal may occur when the reparative scar
tissue narrows the spinal canal. Even minor cord compres-
sion at this level is likely to result in respiratory arrest, as
previously noted by Nordt and Stauffer\(^23\).

Because of the predisposition of patients with Down
syndrome to instability of the cervical spine and because
our data indicate that it is progressive, presumably the result
of repetitive trauma to constitutionally lax ligaments, and
most importantly because the instability can cause a cata-
trope (Figs. 1-A, 1-B, and 1-C), we recommend that all
patients with the syndrome, whether the spine is stable or
unstable, be restricted from all high-risk activities. Patients
with potential or actual instability can be identified; they
have more than four millimeters of atlanto-axial subluxation
when older than ten years, an accessory ossicle at the at-
lanito-axial level, or a fixed atlanto-axial interval of more
than four millimeters, and they should be treated with strin-
gent restriction of activity and close observation. We further
recommend that atlanto-axial fusion should be considered
in all healthy patients with instability whose activity cannot
be restricted.

The only other and absolute indication for fusion is the
presence of neurological symptoms caused by the instabil-
ity. It is important to remember that the dislocations in
question are chronic, as a rule, and therefore are often ir-
reducible. The risk of neurological injury is higher if re-
duction is too vigorous.

Conclusions

Atlanto-axial instability is a gradual, progressive lesion
in patients with Down syndrome. It probably is secondary
to an intrinsic defect in the collagen. The instability results
from cumulative minor trauma over time; therefore, periodic
radiographic evaluation is essential and the activities of a
patient who is at risk must be restricted. An accessory ossicle
in the atlanto-axial interval most likely represents an avul-
sion fracture of the apophysis of the odontoid process, and
some degree of atlanto-axial instability more than likely
precedes that lesion.

An increase in atlanto-axial motion over normal values
in children who are less than ten years old is likely to be
physiological, but in individuals who are older than ten it
is a danger signal. Progressive instability and neurological
deficits are more likely to develop in male patients who are
more than ten years old.

References


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