Editorial

Hypoplasia of C-1 in children with Down syndrome

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In the paper by Matsunaga and colleagues in this issue of Journal of Neurosurgery: Pediatrics, the authors investigated 102 children between the ages of 10 and 15 years with Down syndrome and matched them against 176 healthy children. They obtained plain lateral x-ray images of the cervical spine in all individuals to measure the anteroposterior (AP) diameter of the atlas and the atlas–dental interval (ADI). A cross-sectional area of the atlas was also measured on a cross-sectional computed tomography scan of the atlas. The authors thought that little had been published about myelopathy related to atlantoaxial subluxation in children with Down syndrome, especially in conjunction with spinal stenosis due to “C-1 hypoplasia.”

In their analysis, the authors were able to identify eight children with atlantoaxial subluxation who also had neurological deficits. Among these eight, four had an os odontoideum. The AP diameter of the atlas and the spinal canal area along the cross-section of the atlas was significantly smaller in children with Down syndrome than in the control group. The ADI among the children who exhibited atlantoaxial subluxation ranged from 6 to 9 mm, and all these children exhibited C-1 hypoplasia. The mean ADI in children with Down syndrome was 2.5 mm, and in healthy children it was 2.2 mm. The authors believed that the difference between the two groups was not statistically significant. Sample computed tomography scans of a child with Down syndrome and a same-age healthy child provide conclusive evidence that the cross-sectional area and the AP diameter of the atlas is smaller in children with Down syndrome.

An early description of “hypoplasia of the atlantal arch in Down syndrome” was provided by Martich et al. in 1992. In 1999 and again in 2000, Taggard and colleagues described the atlantal arch abnormalities. In an earlier description of 36 consecutive patients with Down syndrome and craniovertebral abnormalities with instability, neurological deficit was present in all. Atlantoaxial instability was a common radiographic finding observed in 23 of 36 patients. A rotary component was present in 14 others. Occipitococcygeal instability was observed in 16 of 36 patients; in 15 a coexisting atlantoaxial dislocation was also present. Twenty individuals had bone abnormalities, the most frequent of which was os odontoideum followed by atlantal hypoplasia and bifid anterior and posterior arches. Subsequently in 2005, Menezes reported a database analysis of 4800 symptomatic patients with craniovertebral junction abnormalities. Of these patients, 104 had Down syndrome.

The deformity or abnormality of the posterior atlantal arch with reduction in the spinal canal area has been described in skeletal abnormalities such as achondroplasia and in Klippel–Feil syndrome. Os odontoideum occurs in 40 to 50% of patients with Down syndrome and signifies previous trauma. The condition of these patients is unstable. Myelopathy in Down syndrome is due to structural changes (such as os odontoideum, hypoplasia of the atlas arches, assimilation, and basilar invagination) and the attendant instability. This is much worse when the patient is in a state of general anesthesia or asleep and should not be confused with the paucity of excursions that are noted on dynamic radiographs between flexion and extension when the patient is awake.

The term “C-1 hypoplasia” referred to by the authors is actually a deformity of the lateral C-1 mass and an inward bending of the posterior atlantal arch. It is regrettable that surgical intervention, despite being recommended by the authors, was not able to be done.

This is the first matched control study of atlas morphology in Down syndrome, and the authors are to be congratulated.

References

RESPONSE: I appreciate Dr. Menezes’ kind comment about our paper. He mentions that the term “C-1 hypoplasia” is actually a deformity of the lateral C-1 mass and an inward bending of the posterior atlantal arch. I agree with this statement; however, Martich used the term of “hypoplastic posterior arch of C-1” in the first reported paper so we used the term in the presented manuscript. We are familiar with the excellent works by Dr. Menezes about atlantoaxial and craniovertebral instability in patients with Down syndrome. In our original manuscript, we discussed craniovertebral instability and myelopathy in patients with Down syndrome. However, one reviewer commented that we should not discuss craniovertebral instability because we had not examined it in our study. Therefore we deleted this discussion in our revised manuscript. I also think that craniovertebral instability is an important pathological finding for occurrence of myelopathy in patients with Down syndrome. Finally, I appreciate that Dr. Menezes has mentioned that our paper is the first match control study of atlas morphology in patients with Down syndrome. (DOI: 10.3171/PED-07/12/455)

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References