Congenital arthrogryposis associated with atlantoaxial subluxation and dysraphic abnormalities

Case report

WOLF O. LUEDEMANN, M.D., MARCOS S. TATAGIBA, M.D., SAMI HUSSEIN, M.D., AND MADJID SAMII, M.D.
Department of Neurosurgery, Medical School Hannover, Hannover, Germany

The authors report the case of a 27-year-old woman with an arthrogryposis multiplex congenita (AMC) associated with atlantoaxial subluxation. To the authors' knowledge, this is the first report of its kind. The authors review the literature with reference to dysraphic abnormalities associated with atlantoaxial subluxation and with AMC. The patient presented with severe tetraparesis following a minor traffic accident. She underwent a procedure in which transoral decompression and dorsal stabilization were performed and, postoperatively, made a good clinical outcome. The authors stress the need for diagnostic neuroimaging of the cranio cervical junction in patients with AMC.

Key Words • arthrogryposis • atlantoaxial • subluxation • dysraphism • spina bifida

Arthrogryposis multiplex congenita, a congenital syndrome characterized by multiple congenital joint contractures, refers to a large heterogeneous group of disorders. The exact pathogenesis of arthrogryposis is unknown, but all forms involve fetal akinesia (decreased fetal movement) and subsequent joint contractures. Arthrogryposis may be associated with multiple developmental defects, usually in association with other congenital abnormalities, including micrognathia, a wide flat nose, short neck, congenital heart disease, high-arched palate, hypoplastic lungs, and cryptorchism1 or it may be part of an inherited syndrome.2 We report the case of a 27-year-old woman with AMC in association with atlantoaxial subluxation and dysraphic abnormalities. To our knowledge, this is the first report of an association between AMC and dysraphic abnormalities leading to atlantoaxial subluxation.

Case Report

Presentation. When this 27-year-old woman was born, the diagnosis of AMC was made. Her development was uneventful, except for symmetrical contractures, muscle weakness, and a short stature, until a minor traffic accident 1 year prior to presentation. After the accident, the patient developed increasing tetraparesis that first appeared in the left arm. By the time of presentation, her symptoms had progressed to a severe tetraparesis with dyspneic fits and detrusor dysfunction.

Examination. At admission to our department in December 1998, the patient was in poor general condition, weighing approximately 70 lbs and measuring just taller than 4.6 ft in height. She was unable to walk and care for herself. Cranial nerve examination demonstrated no sign of dysfunction, except for the dyspneic fits that were apparent clinically. Hyperpathic dysesthesia starting below level C-4 was present. Examination of motor function revealed severe spastic tetraparesis.

Radiological examination of the skull revealed dorsal dislocation of the tip of the dens but no cranial dislocation with respect to Chamberlain's and McGregor's lines (Fig. 1 upper left). Computerized tomography scanning of the cervical spine and the CCJ revealed dorsal dislocation of the dental tip with a distance of 1.2 cm between the anterior arch of C-1 and the tip of the dens (Fig. 1 upper right). Additionally, ventral and dorsal C-1 nonunion and an odontoideum anomaly were also demonstrated (Fig. 1 lower left). Magnetic resonance imaging revealed severe compression of the brainstem and spinal cord at the CCJ (Fig. 1 lower right).

Operation. With the patient in the lateral position and under three-point head fixation, transoral resec-
Congenital arthrogryposis

tion was performed. Intraoperative somatosensory evoked potential monitoring was conducted during patient positioning and surgery, allowing continuous control of spinal cord function. As an additional indication of dysraphia, a uvula bifida was found during the transoral approach to the odontoid (Fig. 2). After the odontoid and os odontoideum were resected, a dorsal bone and internal segmental fixation construct was placed from the cranial base and to the posterior elements of C-2 and C-3 bilaterally.

Postoperative Course. After surgery the patient’s gait progressively improved. Postoperative radiological control studies demonstrated that decompression and stabilization had been successfully achieved (Fig. 3). The patient was referred to rehabilitation. For cervical immobilization a Philadelphia collar was applied. At follow-up examination 2 months postoperatively, the patient was independent, able to walk unassisted, and care for herself.

Discussion

To our knowledge, this is the first report on an atlantoaxial subluxation associated with AMC. The cause of the atlantoaxial subluxation might have been the dysraphic malformation, that is, the nonfusion of the atlas and the dorsal atlas, as has been observed in a similar case presenting with basilar impression.7 Whereas myelomeningoceles are reported to be associated with AMC,8 other spinal dysraphic symptoms are reported only in calves6 and sheep.5

The chondrous neural arches of C-1 and C-2 show gradual closure starting at 53 days in the embryonic stage, and the process is not yet finished by 77 days in the fetal stage,4 leaving a large time window for the failure of closure to occur, as is putatively suggested by this case.

Prior to traumatic injury, all the symptoms exhibited by our patient were related to the diagnosed AMC. An indicator of the early onset of the instability during intrauterine development is the os odontoideum, which may be the result of excessive movement at the time of ossification of the cartilaginous dens.3 This could result in two ossification centers, appearing as a Type I fracture (Fig. 1 upper right and lower left) during development. The trauma might have led to an aggravation of the atlantoaxial subluxation, the consequence by which the malformation was diagnosed.

The need to perform decompressive surgery was crucial in this case, because fusion in situ may cause neurological deficit to progress.4 In the present case the dorsal subluxation of the odontoid was severe and even “natural decom-

![Fig. 1. Imaging studies. Upper Left: Lateral x-ray film of the skull and the CCJ. The dislocated odontoid tip is located below the lines of Chamberlain (CH) and McGregor (Mc). Upper Right: Median sagittal reconstruction of a bone window CT scan obtained at the CCJ. The dorsal displacement of the odontoid (Od) in relation to the dorsal border of the atlas (A) is clearly demonstrated. An intermingled os odontoideum (o) with a calcified apical dental ligament (arrow) is visible. Lower Left: Transverse bone window CT scan of the CCJ at the level of the atlas. Nonunion of the anterior arch of the atlas (arrow) and the posterior arch (arrowheads) is demonstrated. The posterior displacement of the odontoid with the intermingled os odontoideum is visible. Lower Right: Sagittal magnetic resonance image obtained at the CCJ, demonstrating the severe compression of brainstem and upper spinal cord (arrows) by the posteriorly displaced dental tip.](image)
pression” by the absence of the dorsal arch of C-1 could not prevent the development of myelopathy. Therefore, a transoral decompressive procedure was mandatory.

In our case internal stabilization was achieved by segmental fixation, with the construct extending from the cranial base and to the posterior elements of C-2 and C-3 bilaterally; we used a Marburg loop (Ulrich, Ulm, Germany) and soft wires (Codman, Johnson & Johnson, Norderstedt, Germany). Autogenic iliac crest strips made up the fusion mass to provide for long-term stability.

Conclusions

The present case clearly demonstrates the need for diagnostic imaging of the CCJ in patients with AMC.

References


Manuscript received November 16, 1999.
Accepted in final form March 20, 2000.
Address reprint requests to: Wolf Luedemann, M.D., Department of Neurosurgery, Medical School Hannover, Germany, OE 7240, Carl Neubergstrasse 1, 30625 Hannover, Germany. email: Luedemann.Wolf@MH-Hannover.de.