Atlantoaxial dislocation associated with neurofibromatosis

Report of three cases

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Atlantoaxial dislocation was found in three patients with neurofibromatosis. Roentgenographic findings
included marked reduction of sagittal diameter at the C-1 vertebral level, and cervical spine abnormalities
associated with mesodermal dysplasia, such as posterior scalloping of the cervical spinal bodies with dural
ectasia and vertebral body deformity (vertebral body dysplasia). Although the relationship of the atlas and
axis did not change with neck position, all three patients had progressive neurological deficits and were
treated by decompressive surgery combined with fusion. The pathogenesis of atlantoaxial dislocation
associated with neurofibromatosis is discussed.

KEY WORDS □9 atlantoaxial dislocation □9 neurofibromatosis □9 cervical spine □9 computerized tomography

ALTHOUGH deformities of the vertebral column
are not uncommon in patients with neurofibromatosis, the association of atlantoaxial dislocation and this disorder has been given little attention. We present three such cases and briefly discuss the pathogenesis and clinical significance of the condition.

Case Reports

Case 1

This 5-year-old boy had exhibited difficulty in walking and clumsiness of his left hand for 6 months. He had a family history of neurofibromatosis.

Examination. Physical examination showed numerous café-au-lait spots on the skin and a subcutaneous nodule on the left side of the neck. Neurological examination revealed hemiparesis on his left side and a spastic gait. Deep tendon reflexes were hyperactive in the left upper and lower extremities, and Babinski's sign was positive bilaterally.

Cervical spine tomography in the sagittal plane demonstrated that the atlas had slipped on the axis; however, no alteration in atlantoaxial distance was observed on changes of neck position. Tomography also showed basilar impression, kyphosis at C2–3, and posterior scalloping of the vertebral bodies from C-3 to C-7. On pneumomyelography, the subarachnoid space was seen to be markedly narrowed above C-2, but dilated from C-3 down to C-7 (Fig. 1).

Operation. The patient underwent laminectomy of C-1 combined with posterior fusion from the occipital bone to C-3. Thereafter, anterior fusion of C-2 to C-5 was performed for kyphosis at that level. The patient showed excellent improvement of his neurological symptoms postoperatively.

Case 2

This 21-year-old man complained of progressive difficulty in walking for 3 months. He had also experienced suboccipital pain for 9 months prior to admission. There was no family history of neurofibromatosis.

Examination. Physical examination at admission showed numerous café-au-lait spots and subcutaneous

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FIG. 1. Case 1. Lateral tomographic pneumomyelogram showing the subarachnoid space obliterated at the C-1 level due to a posteriorly displaced odontoid and the atlas. The sagittal diameter of the spinal canal measures 6 mm. The posterior aspects of the vertebral bodies from C-3 to C-7 are excavated by the distended air-filled subarachnoid space. There is kyphosis at the C2-3 level.

ods. The patient could not rotate his neck without pain. Neurological examination revealed spastic right hemiparesis. Deep tendon reflexes were exaggerated in the right upper and lower extremities. There was a decrease of all forms of sensation below the C-2 dermatome, more pronounced on the left side than the right.

Cervical spine tomography in the sagittal plane disclosed atlantoaxial dislocation (Fig. 2). Functional studies demonstrated no mobility of the atlas in relation to the axis. On Myodil myelography, the subarachnoid space was seen to be dilated from C-3 to C-7, and it was completely obliterated at the C-2 level.

Operation. Posterior decompression of the foramen magnum and C-1 was performed, followed by posterior fusion from the occipital bone to C-3. Postoperatively, the patient showed good recovery from his symptoms.

Case 3

This 32-year-old woman was admitted to the hospital with a 2-year history of pain in her left shoulder. There was no family history of neurofibromatosis.

Examination. Physical examination revealed numerous café-au-lait spots, subcutaneous nodules of the scalp, and cleft palate. On neurological examination all the deep tendon reflexes were hyperactive, more pronounced on the left than the right, and Chaddock’s sign was positive on the left side.

Cervical spine tomography in the sagittal plane (Fig. 3 left) revealed atlantoaxial dislocation, deformity of the vertebral bodies of C-2 and C-3 (vertebral dysplasia), and posterior scalloping of the bodies of C-3 to C-7. The relationship of the atlas to the axis did not alter with changes in neck position. Computerized tomography (CT) after intrathecal introduction of metrizamide (Fig. 3 center and right) demonstrated impingement of the odontoid process on the spinal cord at the C-1 level. The CT evidence of dilatation of the subarachnoid space from C-2 to C-7 and the scalloping of the vertebral bodies led to the diagnosis of dural ectasia.

Operation. The anterior arch of the atlas and the displaced odontoid were removed via a transoral approach, and C1–2 fusion was performed. Soft-tissue masses were found around the odontoid at surgery, which were diagnosed histologically as neurofibromas. The postoperative course was uneventful, but the patient died of acute nephritis 5 months later.

Discussion

Atlantoaxial dislocation has been described in only two previous patients who had the stigmata of neurofibromatosis.8,9 None of these five patients (the two
Atlantoaxial dislocation with neurofibromatosis

Fig. 3. Case 3. Left: Cervical spine tomogram, lateral view, showing atlantoaxial dislocation, vertebral body dysplasia at C-2 and C-3, and posterior scolopling of the vertebral bodies. The sagittal diameter of the spinal canal is small, measuring 8 mm, at the C-1 level, and large below C-2. Center: Computerized tomography scan with metrizamide myelogram at the C-1 level. The odontoid process (o) impinges upon the spinal cord (arrows). Right: Scan at the C-3–4 level shows that the subarachnoid space is dilated and erodes the posterior aspect of the vertebral body (arrows).

previous patients and the three cases presented here) showed agenesis or separation of the odontoid process. Four patients had vertebral dislocation, such as C-1 slipping forward on C-2, without rotatory deformity of the atlantoaxial articulation; the fifth patient had a rotatory pattern. Although pluridirectional tomography in the sagittal plane is essential for diagnosis of the condition, CT also provides good delineation of the relationship of C-1 to C-2 in the axial plane (Fig. 3 center).

Since it is widely accepted that neurofibromatosis is a maldevelopment of the neuroectoderm and mesoderm, it is not uncommon to find deformities of the vertebral column and adjacent structures in patients with neurofibromatosis. Kyphoscoliosis and vertebral dysplasia are often observed. Scalloping of the posterior aspect of the vertebral bodies or enlargement of the intervertebral foramina have also been documented and related to dural ectasia-dysplasia of the meninges. Marfan’s syndrome, which is a congenital mesenchymal disorder, is characterized by hypermobility of the joints due to flaccidity of the articular capsules and ligaments, and is also associated with atlantoaxial dislocation. It is not surprising, therefore, that in neurofibromatosis the ligaments supporting the atlantoaxial joints can also be either malformed or flaccid, and result in dislocation of the joints. In one of our patients, multiple neurofibromas were found to intervene between the odontoid and the anterior arch of C-1. Thus, tumor infiltration in the atlantoaxial joint may be another possible cause of dislocation.

References

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