Diaphyseal aclasis with spinal cord compression

Report of two cases and review of the literature

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Diaphyseal aclasis, also known as hereditary multiple exostoses, is a benign condition affecting the epiphyseal plates of cartilaginous bones. It usually involves the ends of long bones, iliac crests, and scapulas. Although in 1843 Reid reported an autopsy finding of spinal cord compression of C-2 due to an exostosis, it was not until 1849 that Stanley was credited with the first description of hereditary multiple exostoses. In 1907, Ochsner and Rothstein performed the first surgical excision of an exostosis of the lamina of C-2, which had caused cord compression.

Spinal cord compression by an exostosis is an unusual complication of this disease. In addition to the cases listed in Table 1, we report two cases of cervical myelopathy that resulted from exostoses of C-2 and C-5 and review the literature to date.

Case Reports

Case 1

This 9-year-old boy with a family history of diaphyseal aclasis (Fig. 1) first presented at 4 years of age with exostoses of his knees, shoulders, and left middle finger. The exostoses remained small and gave him little bother except for the ones on the medial aspect of his tibias which were growing in size.

In June 1988, the boy was referred to the pediatric service because of “fainting” attacks. At that time he was noted to have a tendency to fall over to the right and had a mild right hemiparesis with increased reflexes and an extensor plantar response on the right. Examination at that time included a plain skull x-ray film and a computerized tomography (CT) scan of the brain, both of which were normal. He was referred for physiotherapy. After an initial improvement, the patient’s gait and balance started to deteriorate; his neck became stiff and painful and his nocturnal enuresis became worse.

Examination. In July 1989, the patient was referred to the neurosurgery service because he had developed quadripareis. Cervical spine x-ray films (Fig. 2) showed a narrowed spinal canal at C-2 caused by a large osteochondroma arising from the lamina of that vertebra. Also found was an enlargement of the spinous process of T-9. A CT scan (Fig. 3A and B) confirmed narrowing of the
spinal canal at C-2 with the effective canal diameter between the odontoid process and the exostoses measuring less than 0.6 cm. A CT scan of T-9 showed an additional osteochondroma arising from the posterior aspect of the spinous process; however, this lesion was pointing toward the skin and away from the spinal canal.

**Operation and Postoperative Course.** The patient underwent a cervical laminectomy with complete removal of the extradural osteochondroma. His postoperative recovery was uneventful with progressive neurological recovery and a return to normal function. Histopathological examination confirmed the lesion to be an osteochondroma.

**Case 2**

This 45-year-old man with diaphyseal aclasis was referred to the neurosurgical service with a 20-year history of a mass at the back of his neck. More recently, over the last 3 years, the mass had enlarged. Five months prior to admission, the patient’s gait and grip had deteriorated and he was unable to continue in his occupation as a porter because of frequent falls. He also had paresthesia and impaired sensation in his fingers. The patient’s mother was also known to have diaphyseal aclasis.

**Examination.** On examination the patient had a large, hard, lobulated, and nontender mass on the back of his neck (Fig. 4). This mass extended from the base of his skull to the level of T-1 and outward laterally well beyond the midline. Neurologically, the man had spastic quadriparesis, which was worse on the right side. His cervical spine x-ray films showed a massive osteochondroma extending from C2–T1 in a posterior direction. There were smaller lesions on his ribs, humerus, radius and ulnar, metacarpals, phalanges, and upper tibias. A CT scan of the patient’s cervical spine confirmed the massive osteochondroma extending from C2–T1 and arising from the spinous process of C-5 with involvement of its lamina (Fig. 5). There was a marked narrowing of the theca at C-5, particularly on the right with an anteroposterior diameter reduced to approximately 0.5 cm.

**Operation and Postoperative Course.** The patient underwent a cervical laminectomy with complete excision of the mass at C-2 due to a large osteochondroma. More recently, over the last 3 years, the mass had enlarged. Five months prior to admission, the patient’s gait and grip had deteriorated and he was unable to continue in his occupation as a porter because of frequent falls. He also had paresthesia and impaired sensation in his fingers. The patient’s mother was also known to have diaphyseal aclasis.

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**Operation and Postoperative Course.** The patient underwent a cervical laminectomy with complete excision of the mass at C-2 due to a large osteochondroma.
His postoperative recovery was uneventful: he made a slow and satisfactory neurological recovery and eventually returned to his job. Histopathological examination confirmed the mass to be an osteochondroma with no evidence of sarcomatous change.

**Discussion**

Diaphyseal aclasis is an anomaly of skeletal development characterized by multiple exostoses of cortical bones. Virchow\(^1\) postulated that a portion of epiphyseal cartilage is pinched off and grows independently in a transverse direction to the diaphysis. This has since been confirmed by Milgram,\(^10\) who identified chondromas that separated from epiphysis to become autonomous growth centers at right angles to the longitudinal axis.\(^2,8\) The exostoses most often arise at the metaphysis of long bones and less frequently involve the vertebrae. Histologically, the exostoses consist of cortical bone capped by cartilage. The familial nature of this condition is well established and was conclusively documented by Stocks and Barrington\(^16\) in a review of 1124 cases that elicited a positive family history in 65%,\(^2,3,9\) Other authors have confirmed these findings.\(^3,9,18\) The disease is caused by an autosomal dominant gene with variable expression. It has also been observed that a second, independently segregating autosomal gene may suppress the condition in females, resulting in a higher incidence in males.\(^2,6\) The number of exostoses tends to increase from generation to generation, particularly in families with large numbers of exostoses who, in general, tend to produce more numerous lesions in their offspring. In approximately one of every four women who transmit the disorder to their children, the mother is an unaffected carrier. Hereditary transmission of the disease has not been described in cases of solitary exostosis.\(^7\)

Diaphyseal aclasis may give rise to a number of associated deformities, especially short stature, as a result of shortening and bowing of the legs or scoliosis. It has been reported that between 1% and 20% of patients with exostoses have been found to develop malignant changes in their masses when the follow-up periods extend over a long enough time.\(^3,9\) On clinical examination, our second patient was believed to have a malignant change in his lesion due to its large size, but this proved not to be the case histologically. Neurological dysfunction is most often caused by mechanical irritation or compression of peripheral nerves. Although a significant number (7% in one series\(^3\)) of exostoses from the vertebrae have been found, spinal cord compression caused by an osteochondroma is rare.

Table 1 provides a list of case reports of patients having cord compression as a result of diaphyseal aclasis. Over 70% of these patients gave a positive family history, a higher number than that reported by Decker and Wei\(^1\) or by Madigan, et al.\(^9\) Males are affected more frequently than females, although our ratio of three males to two females is significantly lower than those specified by other reports in the literature.\(^7,12\)

The cervical spine is commonly affected, most frequently at C-2 with most exostoses located on the lamina or pedicle. In the thoracic spine, exostoses may arise from an adjacent rib or costovertebral junction.\(^3,4,6,15\) The age of presentation is commonly in the second and third decades of life, with a mean age of 20 years; our first patient and the one described by Madigan, et al.\(^9\) in 1974, are the only ones reported as presenting in the first decade of life. The results of surgical decompression in this series were generally good, with 86% showing improvement. There were two deaths, one of them occurring in the immediate postoperative period.\(^14\)

**References**

Cord compression in diaphyseal aclasis


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