Cervical Cord Compression due to Exostosis in a Patient with Hereditary Multiple Exostoses

Case Report

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Hereditary multiple exostosis, or diaphyseal aclasis, is not a rare disorder. Almost 2000 cases have been recorded since 1849 when Stanley first described it.1 However, lesions affecting the central nervous system in this disorder are uncommon. This report describes a patient with an exostotic lesion compressing the cervical spinal cord.

Case Report

This 13-year-old girl was admitted to the Neurological Institute on June 28, 1964, with complaints of progressive weakness in the right leg of 4 months’ duration and in the right arm for 3 months, loss of sensation in the left arm, and intermittent neck pain on turning her head to the right. Multiple exostoses had first appeared when the child was 4 years of age. Her mother also had numerous exostoses, but no other relative or sibling was known to be affected.

Examination. The patient was petite, slight, and of immature appearance. Height was 144 cm (average at 13 years is 152.2 cm) and the arm span was 125 cm (average 151.8 cm). Secondary sex characteristics were poorly developed. There were large palpable exostoses at knees, iliac crests, wrists, and elbows bilaterally. The right radius was bowed, with ulnar deviation of the wrist. There was spastic hemiplegia on the right side, with hyperactive reflexes, Babinski sign, and sustained ankle clonus. Pain and temperature sensation were decreased on the left side below the upper cervical region, and touch and vibration were decreased on the left side of the neck and left upper arm. The gait was markedly ataxic.

Head turning to the right was limited and painful.

The hemogram and urinalysis were normal; alkaline phosphatase was 39 King-Armstrong units. Lumbar puncture pressure was normal with cerebrospinal fluid protein 49 mg% and 1 WBC/mm.3 A RISA scan revealed no focal collection, but the electroencephalogram showed diffuse slowing. Skeletal x-rays confirmed the bony abnormalities described above (Fig. 1). Cervical films showed a poorly calcified density at the second cervical level which was visible only on lateral views. Myelography demonstrated an extradural type of filling defect at the C-2 level, with the cord displaced towards the right; a mass lesion at this level was outlined in the lateral supine view (Fig. 2). In view of the multiple lesions, the mass was believed to be a bony exostosis.

Operation. On July 15, 1964, laminectomy was carried out on the right from C-1 through C-4. A large bony mass was seen under the hemilamina of C-2. This mass had displaced the cord from the left to the right and had compressed it to approximately one-half its normal width (Fig. 3). The bony mass was smooth in appearance and was covered with a layer of cartilage. The cartilaginous cap and underlying thin cortical bone shielded out easily, exposing soft spongy bone. Postoperative laminograms revealed the origin of the mass at the junction of the lamina and pedicle at C-2 on the left.

Postoperative Course. There was immediate and continuous improvement of right-sided motor functions, which by 5 months postoperatively were almost normal. By then there was no Babinski sign although ankle clonus persisted; the left-sided sensory defects were only slightly diminished. The patient was attending school and traveling by public conveyance unassisted.

Discussion

Diaphyseal aclasis is a disorder of bone development characterized by heaping up of bone at the region between the diaphysis and epiphysis. The disorder is also referred to as hereditary multiple exostoses.

The genetic nature of this disorder is well established. In their large series, Stock and
Hereditary Multiple Exostoses

Fig. 1. X-ray of right forearm, demonstrating typical deformity. The radius has diaphyseal thickening and exostoses. The ulnar is shortened, causing ulnar deviation of wrist.

Fig. 2. Cervical myelogram, lateral supine view. The filling defect is seen at the level of the C-2 pedicle (arrow).

Fig. 3. Operative photograph. The exostotic lesion (arrow) is seen under the lamina of C-2 (end of instrument). The cord is markedly compressed from left to right.
Barrington found evidence of an affected parent in 64% of patients.10 Similar figures are reported by others.6,11 The disease appears to be due to a single autosomal dominant gene, producing detectable bone lesions in the heterozygote.7 A second, independently segregating autosomal dominant may suppress the disorder in the female, resulting in higher incidence in males.4

Retardation of bone development in diaphyseal aclasis gives rise to a number of secondary deformities in addition to the characteristic exostotic diaphyseal thickening. Patients with this disorder are usually of short stature and proportionately shorter arm-span. A common finding seen in one-third of cases is bowing of the radius with ulnar deviation of the wrist due to shortening of the ulna.5 Genu valgum and valgus deformities of both ankles are also commonly found.

The typical exostotic lesion is a diffuse club-shaped thickening of bone in the region of the metaphysis. The mass appears in both sessile and pedunculated forms and is usually of irregular outline with surface clefts or excrescences. The cortex is heaped up by spongy bone with delicate trabeculae, and is surmounted by a cartilaginous covering.6 Exostoses are typically limited to bones developed in cartilage. Long tubular bones, iliac crests, and vertebral borders of the scapulae are most commonly involved.11 Exostoses of the tarsal and carpal bones, patellae, sternum, and vertebral bodies are found infrequently.2,4,10

Bone deformities and exostoses are rarely seen at birth but emerge as bone growth accelerates during childhood. Short stature becomes increasingly obvious towards the end of the growth period. The diminution of stature, which is more marked in males, is almost entirely due to shortness of the legs, and the head to pubis measurement is often within normal limits. New exostoses rarely appear after the completion of normal growth, although the resulting deformities may not appear until later.

Neurological deficit is most often caused by direct pressure on a peripheral nerve, or by irritation of these nerves by exostoses during movement of a major joint. Intracranial exostoses arising on the clivus or along the synchondrosis between sphenoid and occipital bones have been reported.8,10 There is no unequivocal evidence of exostosis formation in parts of the skull preformed in membrane.6 Although palpable exostoses arising from vertebrae are found in a significant number of patients (7% in Stock’s and Barrington’s series2), only a few instances of cord compression by an exostotic lesion have been recorded.1,8,13

In 1907 Ochsner and Rothstein9 described a similar case in a 23-year-old man in whom a pedunculated exostosis arising from the anterior surface of the lamina of C-2 had ruptured through the dura, causing cord compression. Ipsilateral motor and contralateral pain deficits were noted, as well as pain on rotation of the head. Dr. Ochsner opened the dura and removed the lesion. A residual mild ipsilateral hemiplegia and contralateral pain deficit were recorded following intradural excision.

Summary

Hereditary multiple exostosis (diaphyseal aclasis) is a bone disorder characterized by overdevelopment in the region between diaphysis and epiphysis. The lesions are usually found on long bones and rarely affect the central nervous system. We have reported the clinical, roentgenographic, and operative findings in a young girl whose severe neurological deficits were largely relieved by the recognition and excision of an exostosis at the C-2 level.

References

9. Ochsner, E. H., and Rothstein, T. Multiple
exostoses, including an exostosis within the spinal canal with surgical and neurological observations. *Ann. Surg.*, 1907, 46:608–616.


