Cervical cord compression in a case of hereditary multiple exostosis

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CERVICAL CORD COMPRESSION IN A CASE OF
HEREDITARY MULTIPLE EXOSTOSIS

BY

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CERVICAL CORD COMPRESSION IN A CASE OF HEREDITARY MULTIPLE EXOSTOSIS

PURPOSE

The purpose of this paper is to describe a rare complication of a fairly common disease. The disease, hereditary multiple exostosis, was first referred to in a family by Boyer in 1814, followed by a more detailed description in Guy's Hospital Reports in 1825. Nearly 2,000 cases have been described since that time. Complications of this disease affecting the central nervous system are, however, exceedingly rare. This paper describes a patient with hereditary multiple exostosis in which an exostotic lesion compressed the cervical spinal cord resulting in death from respiratory paralysis.

CASE REPORT

This sixteen year old Sioux Indian boy was admitted to the University of Nebraska Hospital on December 16, 1968, with marked respiratory distress, severe generalized muscle wasting, and multiple skeletal deformities. He was noted in infancy to have multiple bony deformities, but, aside from these lesions and a delay in the onset of walking attributed by his parents to his being dragged behind a wagon at the age of ten months, he was an active child. Despite the accident, he learned to walk by the age of eighteen months, although he used his left arm and
leg less well than the right. Near the onset of adolescence, the patient became gradually weaker, suffering from recurrent respiratory infections, but continued to walk to school until a year before the present hospital admission. He was evaluated at that time for entrance into a school for crippled children and was found to have a generalized muscle weakness most striking on the left side and on the trunk with consequent difficulty in chest expansion. He was able to walk unaided, if somewhat unsteadily, had multiple bony deformities, and contracture deformities of the left shoulder, elbow, and knee. He was also found at this time to have a normal IQ. Shortly after entering the crippled children's school, approximately three months prior to the present hospitalization, he suffered an episode of acute respiratory distress, was hospitalized, and was placed on a respirator because of marked hypoxia. An unsuccessful attempt was made to discontinue the respirator after an initial improvement in his respiratory state.

FAMILY HISTORY

Family history was difficult to document, but there was no history of bony abnormality in either of the patient's parents, both three fourths Sioux Indians, or in any of his ten siblings and fifteen half siblings. Radiographs of the hips and knees of both parents showed no abnormality.
PHYSICAL EXAMINATION

The patient was small and emaciated, appearing older than his stated age. Height was 147.3 cm.; weight was 30 kilograms. The patient had large multiple exostoses at the knees, ankles, feet, wrists, elbows, shoulders, anterior chest, scapulae, and iliac crests bilaterally. He was confined to the respirator. Neurological examination showed the cranial nerves to be intact with absent muscle action below the level of C2. The deep tendon reflexes were hyperactive bilaterally, with bilateral ankle clonus and Babinski signs present. Pain and temperature sensation could not be adequately evaluated because of the patient's critical condition and inability to talk, but an obvious hypesthesia was present.

LABORATORY

The initial hemogram and urinalysis were normal, as were the spinal fluid, serum proteins including electrophoresis, immunoglobulins, and serum electrolytes. Numerous blood gas determinations showed a variable hypoxia, regulated and corrected by the respirator. Urine screening tests for amino acids, cystine, mucopolysaccharides, and galactose were all negative. Multiple twenty four hour urinary calcium and phosphorous levels were all lower than normal although vitamin D2 administration resulted in a normal rise of the urinary phosphorous with no concomitant change in the serum calcium levels. Multiple serum calcium, phosphorous,
and magnesium determinations were all within normal range. Alkaline phosphatase values ranged from eight to twelve King Armstrong units (Normal=2-8 K-A units). Chromosome analysis showed a normal male karyotype. Electrocardiogram suggested left atrial and left ventricular hypertrophy. Electroencephalogram done ten days after admission showed a generalized abnormality fluctuating with the state of consciousness. Electromyogram done one week after admission was consistent with a lower motor neuron lesion.

HOSPITAL COURSE

Maintenance on the tracheostomy and respirator was required throughout the hospital course in order to provide adequate respiration. His sensorium improved with adequate ventilation and he appeared alert, but had no movement below the level of C2. A trial of Neostigmine produced no increase in muscle tone. He was considered to have compression of the cervical cord, but myelograms were not done because of his poor clinical condition. The initial improvement in his clinical status was transient, the patient later contracting pneumonia of his left lower lobe, and the remainder of his hospital course consisted of attempts to deal with his deteriorating respiratory status. On January 16, 1969, one month after admission, the patient expired.

A postmortem cervical myelogram showed an incomplete block in the area of C1 and C2, due to a posterior compres-
sion. At autopsy a large exostosis (1 by 1.5 cm.) was found in the area of C1 and C2, with compression of the cord in that area. The cord showed marked gliosis and demyelination at the level of C1 and C2 secondary to the pressure effects of the exostosis. The exostoses from all parts of the body were similar in morphology and were typical of this disease.

DISCUSSION

Hereditary multiple exostosis has been referred to by a confusing array of names since first described. It has been variously denoted as diaphysial aclasis (common in the British literature), hereditary deforming chondrodysplasia, (formerly common in the American literature), dyschondroplasia, and simply "osteogenic disease". The term hereditary multiple exostosis is preferable to the others, however, since it alludes both to the morphologic bone changes and the hereditary nature of the condition.6

The disorder is characterized by the presence of variable numbers of bony prominences on the skeleton, and may occur in almost any degree of severity.2,4,6,11 The lesions are confined to bones preformed in cartilage, with a great propensity for the long tubular bones, especially at the region between the diaphysis and the epiphysis.6 The typical lesion is a diffuse, club-shaped thickening arising from the shaft of a long bone directed away from the epiphysis with a superimposed exostotic cartilaginous mass which can be
either pedunculated or sessile. The lesions are most noticeable at the knees, ankles, and wrists, often resulting in a synostotic-like junction for several inches between tibia and fibula or radius and ulna. The disease shows a tendency for the lesions to be bilateral, often nearly symmetrical in distribution. The long bones are characteristically shortened, often resulting in an appearance not unlike a mild achondroplasia, though the other stigmata of this disease are lacking.\textsuperscript{1,4,6,11} Bowing of the forearm, not necessarily bilateral, is a common feature.\textsuperscript{6,12}

The distribution of lesions is overwhelmingly in the long tubular bones, iliac crests, and the scapulae, with lesions less commonly located on the patellae, sternum, carpal bones, and vertebral bodies.\textsuperscript{1,4,6,11} Solomon's series found 9% of the cases to have vertebral exostoses.\textsuperscript{11}

The disease is usually noted in the first decade of life, seldom, however, before the age of two years, although Stark, et al report that early radiographic examination of the offspring of affected adults can result in earlier diagnosis.\textsuperscript{12} The earliest manifestation usually is an accidental discovery of one or more bony prominences on the skeleton. During the normal period of growth, the lesions usually increase progressively in both size and number. New exostoses rarely appear after the completion of normal growth.\textsuperscript{6,7}

The genetic nature of the disease has been well established.\textsuperscript{2,5,8}
In the large series of Stocks and Barrington, an affected parent was found in 64% of the cases. The disease appears to be due to a single autosomal dominant gene producing detectable bone lesions in the heterozygote. A second, independently segregating autosomal dominant gene may suppress the disorder in the female, resulting in the often observed higher incidence in males.

The low incidence of complications is testimony to the relatively benign nature of the disease. Complications of the disorder are often due to pressure symptoms or to interference with adjacent structures. The incidence of malignant transformation is higher in hereditary multiple exostosis than in solitary exostoses, greater than 10% in some series, and is therefore the most common serious complication.

The most frequent cause of neurological deficit with this disease is direct pressure on a peripheral nerve. Only two previous cases of cervical cord depression due to a vertebral exostosis in a case of hereditary multiple exostosis have been reported, although allusions have been made to others. Both reported cases were at the level of C2, both were successfully operated, and the patient in each case recovered with a minimum of residual.

Differential diagnosis must include multiple enchondromatosis (Ollier's disease, dyschondroplasia), metaphyseal dysostosis of Jansen, and achondroplasia. Hereditary multiple
exostosis should ordinarily be distinguishable by its characteristic radiographic and clinical features.

The need to differentiate hereditary multiple exostosis from multiple enchondromatosis is more a result of the confusing nomenclature of the congenital bone diseases than any striking similarity in the diseases themselves. These two conditions were formerly thought to be different manifestations of the same osteogenic disease. It is now known, however, that these conditions are separate and independent diseases with distinct clinical and anatomical features. Multiple enchondromatosis is never hereditary, rarely do exostoses occur, the lesions are confined mainly to the interior of the bone, and the condition is usually unilateral in distribution.\textsuperscript{6,12}

Metaphyseal dysostosis of Jansen, an exceedingly rare disorder of bone growth, is similar to hereditary multiple exostosis in many ways. Its bony abnormalities are concentrated in the metaphyseal regions of the long bones with the epiphyses being spared. The disease is hereditary, bilateral in distribution, usually associated with short stature, and, classically, devoid of laboratory abnormalities. The diseases are, however, distinguishable by the notable lack of exostoses, and the sparing of the pelvic bones, bones of the trunk, and the vertebral bodies in Jansen's disease. The radiographs of a presumed case of metaphyseal
dysostosis of Jansen accompany this paper to illustrate the radiographic similarities and differences of the two diseases.\textsuperscript{3,9}

Mild cases of multiple exostosis with shortening of the limbs and few large exostoses may superficially resemble achondroplasia. However, the lack of the other signs of achondroplasia including platyspondylisis, a larger head than normal, frontal bossing, depressed nose, and square hands allow radiographic differentiation.\textsuperscript{6}

SUMMARY

Hereditary multiple exostosis is a congenital bone disorder characterized by bizarre deformities in the region between diaphysis and epiphysis. The lesions are most commonly found on the long bones and consequently rarely cause any central nervous system symptomology. This paper reports the clinical, radiographic, and postmortem findings in a patient with compression of the cervical cord caused by a vertebral exostosis in a case of hereditary multiple exostosis.
FIGURE 1: AP view of both forearms indicates severe metaphyseal abnormalities secondary to multiple exostoses. There is dislocation of the radial head at the level of the left elbow.
FIGURE 2A: A large defect is seen at the level of the posterior aspect of the C1-C2 interspace. This defect coincides with a very faintly demonstrated exostosis arising from the superior aspect of the C2 posterior neural arch.
FIGURE 2B: The large filling defect demonstrated at the level of the C1-C2 junction posteriorly is demonstrated to be central in its position. The defects in the Pantopaque column coincided exactly with the large exostosis impinging upon the cervical portion of the cord at this level. This exostosis was confirmed at the time of the post mortem examination, the cervical cord was splayed around this exostosis.
FIGURE 3: AP radiograph of the left upper arm demonstrating severe metaphyseal deformities secondary to multiple exostoses.
FIGURE 4: PA radiograph of the chest depicting numerous exostoses arising from the posterior ribs overlying the right upper thorax. The rib cage is distorted, but the rib architecture except for the exostoses appears to be intrinsically normal. No thoracic or pulmonary etiology could be established for the patient's chronic respiratory difficulties. Respiratory arrest was noted and was a prominent feature in the patient's terminal course.
FIGURE 5A: AP radiograph of right thigh indicating severe metaphyseal abnormality in a patient with metaphyseal dysostosis of Jansen.
FIGURE 5B: AP of left arm indicating metaphyseal widening of the humeral shaft proximally and of the ulnar shaft distally, characteristic of metaphyseal dysostosis of Jansen. Note lack of exostoses.
FIGURE 6: AP radiograph of both hands indicating multiple metaphyseal abnormalities with widening of metaphyses, normal epiphyses, and no exostoses, all features characteristic of metaphyseal dysostosis of Jansen.
BIBLIOGRAPHY


