AN UNUSUAL CASE OF KYPHOTIC PARAPLEgia AND HYPOPHOSPHATASIA

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Paraplegia caused by kyphoscoliosis is quite rare other than in those cases in which it is secondary to tuberculosis of the spine. In this presentation we plan to refer only to those cases of paraplegia resulting from severe kyphoscoliosis not caused by tuberculous granuloma. Scoliosis itself is a common deformity of the spine but seldom results in neurological disturbance. Some 150 patients with scoliosis without primary neurologic disease have been treated at the Springfield Unit of the Shriners Hospitals. Although many have had severe spinal deformity, no other case of spastic paraplegia has been encountered. In another large series of cases of scoliosis studied, neurological disturbance was found to occur in only 0.3 per cent.

The various conditions causing spinal deformity other than tuberculosis range from those in which the neurologic disorder is primary, such as progressive muscular dystrophy, syringomelia, Friedrich’s ataxia and poliomyelitis, and those in which it is secondary, such as von Recklinghausen’s neurofibromatosis, intraspinal extradural cyst, intraspinal neoplasms, etc.

We believe that the neurological disease in the present case is secondary to the spinal deformity and that this in turn was caused by hypophosphatasia. This occurred in a child whose scoliosis was first noted at 19 months of age but who had no neurological difficulties until aged 13, when spastic paraparesis developed.

The radiographic appearance of the skeleton in our case was typical of hypophosphatasia first described by Rathbun. This is a rare chronic disorder characterized by deficient formation of bone because of a lack of calcification of osteoid and proliferating cartilage. The basic defect appears to be low alkaline-phosphatase activity of serum, bone and other tissues. Clinically there may be dwarfism, defective ossification of the skull with premature synostosis, premature loss of deciduous teeth, hypercalcemia, renal abnormalities and skeletal deformities. Thoracolumbar kyphosis has been described by McCance. Abnormal excretion of phospho-ethane-diamine in the urine is noted often.

A genetic abnormality is present, and a low alkaline phosphatase may be found in parents and siblings who may be otherwise apparently normal. The severity of the disease correlates with the age of onset, being more severe the earlier the symptoms become manifest.

Kyphotic paraplegia was recognized as early as 1888 by MacAven and early cases were reported by Sachs and McKenzie, but a complete review of the condition was not made until 1949 by McKenzie and Dewar. Approximately 50 cases have been reported in the literature but none appears to have been associated with hypophosphatasia.

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Shriners Hospital for Crippled Children #7509. A white female was born July 12, 1944, 1 month premature. Weight at birth was 4 lbs, 8 oz., and the infant was “kept in an incubator” for 5 weeks. She sat at 8 months and began to walk at 17 months. Routine supplements of vitamins were administered from an early age. Family history was negative for orthopedic disorder. At the age of 19 months a spinal deformity was noted. She was first seen at the Shriners Hospital at the age of 20 months with a localized left dorsolumbar kyphoscoliosis. Spina bifida without meningocele was noted, but otherwise neurological findings were normal. The extremities showed no deformity at this time. The patient was fitted with a reinforced canvas corset. Despite this, increased rotatory kyphoscoliosis was apparent at T9-L1 within a year. A right genu valgum and left genu varum became apparent, and roentgenograms revealed uncalcified osteoid in the metaphyses of the knees and hips. A diagnosis of dyschondroplasia was entertained and serial films revealed progressive distortion of the vertebral bodies consistent with this condition.

In August, 1949, at the age of 5, the patient was admitted to the Shriners Hospital for corrective wedging plasters. No correction could be obtained and a reinforced corset, with bilateral auxiliary crutch extensions and an apical pressure pad, was fitted. Growth remained markedly retarded and roentgenograms of the wrist revealed retarded bone age.

Spinal deformity progressed despite continuous support and exercises. She remained active and attended school. Expansion of the chest was measured at 1/4 to 1/3 but no serious respiratory illnesses developed.

In 1958, at age 13, she complained of stiffness in both legs, which had been developing insidiously for the past year. This progressed until she had difficulty climbing stairs and even walking on level ground. There was no urinary or bowel difficulty. She complained of very slight numbness of the legs.

She was referred to the Mary Hitchcock Memorial Hospital in Hanover, New Hampshire because of progressive spasticity of the legs and difficulty in walking.

Mary Hitchcock Memorial Hospital #143283. Examination revealed her to be an exceedingly tiny, pleasant, bright, white female walking with a marked limp and spastic weakness of the legs. She was unable to climb a step and became quite exhausted after walking the length of a corridor. The cranial nerves were normal. She had a very small head. There was a very severe degree of kyphoscoliosis in the mid- and lower thoracic area. In the motor sphere there was no abnormality in the upper extremities. Reflexes were hyperactive at the knees and ankles with clonus and bilateral Babinski’s sign. Sensation of light touch and pin prick was diminished below L1.

Roentgenograms of the spine revealed not only severe kyphoscoliosis with a severe rotatory element in the thoracic spine, but also a dysplasia of the metaphyses of all the long bones of the arms and legs with no involvement of the epiphyses or diaphyses, characteristic of hypophosphatasia. Her alkaline phosphatase was 5.6 Bodansky units, normal being 10–14 at this age. Lumbar puncture revealed a complete block to Queckenstedt’s maneuver and a spinal fluid protein of 114 mg. per cent. Myelography was not done.

Decompressive laminectomy and opening of the dura mater over the kyphos was carried out on June 19, 1958, with no sectioning of dentate ligaments and no spinal fusion. There was no manipulation of the spinal cord whatever at operation but postoperatively the weakness of the legs was greatly increased, progressing to almost complete paraplegia in 48 hours at which time, fearing a postoperative hematoma, the wound was reopened but no bleeding whatsoever was encountered.

After approximately a week the paraplegia cleared, and within 2 weeks the patient was walking. She was discharged home 4 weeks postoperatively. Since that time she has had progressive improvement in her paraparesis. She has grown 2½ inches in height whereas in the previous 2 years she had grown only 1½ inches. Her pyramidal tract signs have disappeared. Clonus and Babinski’s sign no longer are present. Sensation has returned to normal. She has returned to school and now, 27 months postoperatively, is steadily improving.
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DISCUSSION

Most authors in discussing paraplegia from severe kyphoscoliosis of nontuberculous origin have agreed that decompressive laminectomy and opening the dura mater, which is stretched tightly over the cord compressing it from the dorsal aspect, is the treatment of choice.\textsuperscript{1} Anterolateral decompression through a costotransversectomy type of approach was considered but ruled out in this patient, whereas it is considered to be the treatment of choice for tuberculous paraplegia.\textsuperscript{2} Most previous cases reported occurred when the patients were in their teens.\textsuperscript{1} Only 3 were under 10 years of age and 6 were between the ages of 20 to 23. No previous case of this type of metaphyseal dysplasia caused by hypophosphatasia has been encountered in the literature.

SUMMARY

A 13-year-old girl with progressive paraplegia caused by kyphoscoliosis secondary to a diffuse metaphyseal dysplasia and hypophosphatasia has been reported. Improvement has followed decompressive laminectomy.

REFERENCES