Multiple Epiphyseal Dysplasia

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A 50 year-old rickshaw puller presented with weakness of all four limbs for 15 days. He could not stand and walk, hold any object or button his shirt. He had lost bladder and bowel sensation. He had pins and needles of the extremities and burning sensation in palms and soles for last one year. Family history was non-contributory. He had multiple skeletal deformities since childhood. He had pectus excavatum, prominent costovertebral junctions and bilateral large ears (Figure 1). His higher mental function and cranial nerves were normal. Distal muscle weakness (4/5) was more than proximal muscle weakness (3/5) in both upper and lower extremities. No truncal weakness was present. Tone was increased in all four limbs. Fasciculation was present in both thighs. Wasting of lumbricals

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and interosseii were present. Asymmetric and patchy sensory loss was evident. Posterior column sensations were intact. Planter was extensor bilaterally. Upper limb jerks were exaggerated. Cremasteric and abdominal reflexes were absent. Widespread compressive myeloradiculopathy was suggested. His height was 5 ft 5½ inch and arm span was 5 ft 5 inch. Upper segment and lower segment ratio was equal. He had osteoarthritis of hand, knee and hip joints. He had cervical and lumber spondylosis. Few bony swellings (exostosis) were present on hands and feet. He had large second and third toes and short fourth and fifth toes bilaterally. He had brachydactyly of bilateral little and right ring fingers (Figures 2 and 3).

Straight x-ray spine revealed degenerative disc disease involving cervical, lumber and dorsal vertebrae; and loss of cervical and lumber lordosis. Chest x-ray showed degeneration of costochondral junctions with increased angulations (Figure 4). Straight x-ray hand showed bilateral small fifth metacarpals, small right fourth metacarpal, and degeneration of distal epiphysis and interphalangeal joints (Figure 5). Foot x-ray also showed degenerated joints, shortening of fourth and fifth metatarsals with degenerated distal epiphysis (Figure 6). Patient had normal serum calcium and parathyroid hormone levels. MRI confirmed degenerative disc disease with spinal cord compression involving cervical, lumber and dorsal vertebrae (Figure 7). Genetic study revealed mutation in SLC26A2 gene. The patient was finally diagnosed to have recessive multiple epiphyseal dysplasia (rMED) with quadriplegia due to myeloradiculopathy caused by degenerative disc disease.

Multiple epiphyseal dysplasia (MED) is a disorder of skeletal development affecting the epiphyseal region of bones. It may be autosomal dominant (ADMED) or autosomal recessive (rMED). Superti-Furga et al described the first case of rMED in 1999.1 Mutations in COMP, COL9A1, COL9A2, COL9A3, or matrilin 3 (MATN3) gene cause ADMED. Mutations in the sulphate transporter, DTDST (SLC26A2) gene cause rMED. Some people with MED may not have mutations in a known gene.

Children with ADMED present with joint pain and fatigue after exercise and waddling gait. Small and irregular ossification centers are seen in X-rays especially of hips and knees. X-ray of spine may show few irregularities. Adults have short stature or near normal height. They have short limbs relative to their trunk. Articular cartilage becomes secondarily damaged due to underlying bone deformities, resulting in early degenerative arthritis. Movements may be limited at major joints. Mild proximal myopathy may be present.

Ballhausen et al described phenotypic features of 18 rMED patients.2 rMED is associated with malformations of hands, feet and knees, and scoliosis. Individuals may born with clubfoot, cleft palate, clinodactyly, or ear swelling. Hand or foot deformities include brachydactyly, clinodactyly, clubfoot, and broadening of the space between first and second toes. Chest-wall deformities (funnel-shaped or pigeon-shaped chest) may occur. Genu valga or vera may present. Height is within normal range before puberty. Adults are slightly diminished in stature. Facies and body proportions are usually normal. Arthralgia usually appears at late childhood. Functional disability is mild or absent in children. Joint disease progresses in adults. Radiologic findings include flat or distorted epiphysis and brachydactyly. Ossification centers are late in appearance, small and irregularly mineralised. Double layered patella (i.e., presence of separate anterior and posterior ossification centers) may be seen on lateral x-ray of knee. Osteoarthritic changes are frequent in adults. Carpal and tarsal bone deformities are also evident. Epiphyseal regions of arm and leg long bones are often deformed. Intervertebral disc spaces may be narrowed.

Treatment of MED includes muscle strengthening exercises and analgesic medications. Occupational or recreational activities or sports causing joint overload should be avoided. Orthopedic surgery is sometimes used to relieve symptoms.

MED is rarely diagnosed because of mild symptoms in majority of cases. But apparently mild cases may present with serious morbidity due to myeloradiculopathy from degenerative spine disease like our case.

References