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Juvenile Idiopathic Osteoporosis

M.L. Kulkarni

K.S. Keshavamurthy

Juvenile idiopathic osteoporosis (JIO) is a very rare condition of primary bone demineralisation that presents in childhood. Most pediatricians are unfamiliar with this condition owing to the difficulty in recognition and a long list of differential diagnosis. We report an 8-year-old girl who presented with generalized severe osteoporosis. Diagnosis of JIO was made by excluding other common causes of osteoporosis in this age.

Key words: *Childhood osteoporosis, Juvenile idiopathic osteoporosis.*

Juvenile idiopathic osteoporosis (JIO) is a rare condition of unknown etiology,

From the Department of Pediatrics, JJM Medical College, Davangere 577 004, Karnataka, India.

Correspondence to: Dr. M.L. Kulkarni, Prof & Head of the Department of Pediatrics, JJM Medical College, 2373, M.C.C. 'A' Block, Davangere 577 004, Karnataka, India

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characterized by prepubertal onset and spontaneous remission with progression of puberty(1). It is an important differential diagnosis for conditions causing generalized osteoporosis in childhood and is diagnosed by excluding other causes. In the present communication, we report a case of JIO along with an approach to generalized osteoporosis in childhood.

Case Report

An 8-year-old girl born to a noncon-sanguineous married couple presented with complaints of pain in both feet on walking and inability to get up from sitting position since the age of 3 years. Child was apparently normal till that age, when she had an accidental fall and sustained fracture of left femur, following which her movements were restricted for 6 months. The examination of the child revealed scoliosis, angulation deformity of the right arm and tenderness over both feet. Examination of other systems was essentially normal. Routine blood counts, urine examinations were normal. Creatinine-phosphokinase (CPK) level was within normal limits. A radiological skeletal survey was done which showed pencil-thin cortices of long bones, angulation of right humerus

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(Fig. 1), biconcave vertebral bodies with denser end plates and increased intervertebral spaces. All bones had severe osteoporosis. Serum calcium (8.1 mg/dL), phosphorus (4.3 mg/dL), alkaline phosphatase (318 IU/L), blood urea (10 mg/dL), serum creatinine (0.6 mg/dL), urine pH(6), and blood gases were in the normal range. Hormonal assays showed normal pituitary, adrenal, and parathyroid functions. Serum ceruloplasmin (17 mg/dL) and copper values (83 μ g/dL) were also normal. A diagnosis of JIO was considered.

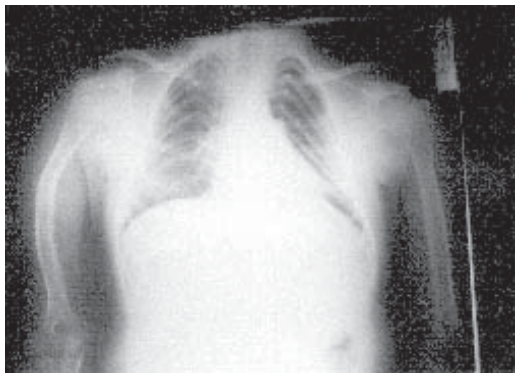


Fig. 1. Right humerus: Osteoporosis with pencil thin cortex and angulation deformity.

Discussion

Juvenile idiopathic osteoporosis is a rare form of bone demineralization disorder. As such osteoporosis itself is a relatively uncommon condition in childhood, and when occurs is usually secondary to other well known causes like rickets, endocrinopathies, malabsorption syndrome, immobilization, tumor induced, Wilson's disease, osteoporosis pseudoglioma syndrome, and inborn errors of metabolism like homocystinuria. In the present case, we excluded rickets by normal values of serum calcium, phosphorus, alkaline phosphatase, and absence of characteristic radiological changes. Endocrinal causes were ruled out by hormonal assays. Normal levels

of serum urea, serum creatinine, blood gases and urine pH excluded renal and metabolic causes. Wilson's disease was made unlikely by normal ceruloplasmin and copper values. Normal eye examination ruled out osteoporosis pseudoglioma syndrome, another rare condition causing generalized osteoporosis. After excluding these conditions, we considered two primary demineralization disorders occurring in the childhood viz osteogenesis imperfecta (OI) and juvenile idiopathic osteoporosis. The former is a heritable condition occurring in 4 types. Absence of blue sclera, deafness, dentogenesis imperfecta, wormian bones in the skull excluded OI types 1, 2, and 3. But type 4 cannot be differentiated easily. This difficulty of differentiating the two conditions is very well recognized by earlier workers as well(1). In our case, the features which go against type 4 OI are absence of other involved members in the family, presence of white sclera since birth, and absence of severe progressive deformation. Further, normal width of long bones in the X-rays makes osteogenesis imperfecta a distant diagnosis(2). Another aid which unfortunately was not available and which could have helped us in resolving the issue to some extent was the radiograph taken at the time of first fracture. If it had showed normal bone density, then type 4 OI was more likely(3). One more investigation which we have not done due to non availability is ratio of a_1 (III) to a_1 (I) collagen in pepsin-digest of skin. An increased ratio would strongly suggest mild osteogenesis imperfecta, but a normal ratio would not definitely exclude it(1). Thus, with this clinical exercise along with relevant investigations to exclude many other conditions with osteoporosis, we arrived at a diagnosis of JIO, which has been recognized as a diagnosis of exclusion(4). The child is on regular follow up.

The exact pathogenesis of this disorder is not known but available evidence points toward disturbed bone remodeling which predominantly affects surfaces that are in contact with the marrow cavity and results in a very low bone formation rate and decreased cancellous bone volume(5). The age of onset of the disease varies from one to thirteen years (mean 7 years). The disease shows no sex predilection(6). The main presenting symptoms include repeated long bone fractures, pain in the back, and difficulty or inability to walk. Particularly, the latter symptom has been stressed by many authors(7-8) and was seen in our case as well. This fact emphasizes inclusion of juvenile idiopathic osteoporosis in the differential diagnosis of a child presenting with walking difficulty after ruling neuromuscular etiologies. Typical radiological changes include generalized osteoporosis, compression of vertebral bodies and metaphyses of the long bones. Measurement of bone mineral density will show strikingly low values. In majority of cases, the disease remits during or after puberty. However, exaggerated bone resorption causing vertebral fractures during pregnancy in a recovered patient with JIO has been observed recently(9).

Although natural remission is the rule, currently affected children should be protected from developing permanent deformities of the spine and long bones by restricting activities. Many drugs like calcitriol, bisphosphonates, fluorides and calcitonin have been used with equivocal results(4). In one study, 3 out of 4 affected children treated with calcitriol showed significant improvement in bone mineralization after 12 months(10). The untreated child did not show any improvement in the same period of time. Another study on effect of calcitonin therapy did not show any effect on the disease(11). At the moment,

experience is insufficient to advocate any treatment other than activity restriction till natural remission.

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