CASE REPORTS

IDIOPATHIC JUVENILE OSTEOPOROSIS - A RARE CAUSE OF OSTEOPOROSIS REPORTED IN A FIVE YEAR OLD BOY

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Abstract

Idiopathic Juvenile osteoporosis (IJO) is a rare primary bone demineralization disorder that presents in childhood. Because of the difficulty in diagnosis, it is unfamiliar to most pediatricians and there is a long list of differential diagnosis. We report a five year old boy who presented with generalized osteoporosis. Diagnosis of IJO was made by excluding other common causes of childhood osteoporosis.

Key words: Childhood osteoporosis, idiopathic Juvenile osteoporosis.

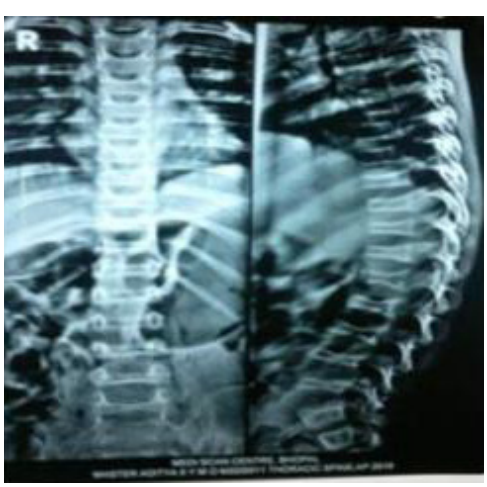
Introduction

Osteoporosis in childhood is uncommon and may be secondary to a spectrum of diverse conditions. When such causes have been excluded, the cause is either a congenital disease (osteogenesis imperfecta) or a disease of unknown etiology called as IJO (1). IJO is characterized by prepubertal onset and spontaneous remission with progression of puberty. This case illustrates the occurrence of IJO which was considered after excluding other causes of childhood osteoporosis.

Case Report

A 5-year-old male child born to a non consanguineous married couple presented with complaints of pain in back and lower limb and inability to sit up from bed, which progress to inability to stand and walk since 1 year. On examination, weight was 13.8 kg, height was 118cm. There was reduced muscle bulk of both lower limbs. The tone and deep tendon reflexes were normal. Examination of the other systems was normal. X-ray of the spine and limbs showed generalized osteoporosis (marked reduction in the normal trabecular pattern of bones). Lateral radiograph of spine showed a "cod fish" appearance (fig. 1) with biconcave vertebral bodies with denser end plates and increased intervertebral space. All the bones had severe osteoporosis without any evidence of fracture. Serum calcium (11.18mg/dl), serum phosphorous (5mg/dl), serum alkaline phosphatase (200U/l), 25 hydroxy vitamin D levels (46ng/ml), serum urea (18mg/dl), serum creatinine (0.28mg/dl) were normal. Urinary pH (6.2), arterial blood gases (pH- 7.36, HCO2- 21) were also in the normal range. Hormonal assays showed normal parathyroid (56 pg/ml) and thyroid function tests (free T3- 6 pmol/L, freeT4- 21pmol/L, TSH-2mIU/L). Serum copper (118mcg/dl) and serum ceruloplasmin (48mg/dl) were normal. His complete ophthalmological examination including slit lamp was normal. Bone densitometry showed low bone density for his age (z-score < -2 SD). A diagnosis of IJO was considered.

Fig 1: X-ray of the spine showing "cod fish" appearance (biconcave vertebral bodies with denser end plates and increased intervertebral space)



Discussion

Idiopathic Juvenile osteoporosis is a rare disorder of primary bone demineralization with no known cause, which has been recognized as a diagnosis of exclusion. (1) Osteoporosis itself is uncommon in childhood and is usually secondary to an underlying medical disorder like rickets, endocrinopathies, malabsorption syndrome, immobilization, Wilson?s disease, osteoporosis pseudoglioma syndrome, and inborn errors of metabolism like homocystinuria. (2,3) It can also be due to medications used to treat a disorder like anticonvulsants, corticosteroids or immunosuppressive agents. (4)

In the present case, rickets was excluded by normal values of serum calcium, phosphorus, alkaline phosphatase, and absence of characteristic radiological changes along with normal vitamin D levels. 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. Normal ceruloplasmin and copper values ruled out Wilson?s disease. Normal eye examination ruled out osteoporosis pseudoglioma syndrome. After excluding these conditions, we considered two primary childhood demineralization disorders viz osteogenesis imperfecta (OI) and juvenile idiopathic osteoporosis. (2) The former is a heritable condition occurring in 4 types. Type 1, 2 & 3 were excluded by absence of blue sclera, deafness, dentigenesis imperfecta, wormian skull bones and limb fractures. (3) But type 4 was difficult to differentiate. In our case, the features which were against type 4 OI were absence of involvement of other family members, white sclera since birth and absence of severe progressive deformation. This difficulty in differentiating type 4 OI and IJO was reported by earlier workers as well. (4) One investigation which would have helped us in resolving the issue but was not done due to non availability was the 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. (4) Thus, with these clinical findings along with relevant investigations to exclude many other conditions with

osteoporosis, we arrived at a diagnosis of IJO.

The exact pathogenesis of this disorder is not known but available evidence points towards 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 mean age of onset is 7 years (range 1-13 years) with no sex difference. The main presenting symptoms are long bone fractures, pain in back and difficulty or inability in walking. (6) Typical radiograph shows compression of vertebral bodies and metaphysis of long bones. Bone mineral density shows strikingly low values. In majority of cases, the disease remits spontaneously during or after puberty. Although spontaneous remission is the rule, restricting activities of the currently affected children is necessary to protect from permanent deformities of the spine and long bones. Many drugs like calcitriol, biphosphonates (7), fluorides and calcitonin have been used with equivocal results (4). Due to insufficient experience, no treatment can be advocated at the moment other than activity restriction till natural remission.

References

1. Krassas GE. Idiopathic juvenile osteoporosis. Ann NY Acad Sci 2000; 900: 409-412
2. Akler G, Pikielny PR, Kots E, Ish-Shalom S, Uziel Y. Multiple vertebral fractures in a young girl: a question of treatment. Isr Med Assoc J. 2010; 12: 116-118
3. Paterson CR, McAllion S, Miller R. Osteogenesis imperfecta with dominant inheritance and normal sclerae. J Bone Joint Surg Br. 1983; 65: 35-39
4. Smith R. Idiopathic osteoporosis in the young. J Bone Joint Surg Br. 1980; 62-B: 417-427
5. Rauch F, Travers R, Norman ME, Taylor A, Parfitt AM, Glorieux FH. The bone formation defect in idiopathic juvenile osteoporosis is surface-specific. Bone. 2002; 31: 85-89
6. Smith R: Idiopathic juvenile osteoporosis: experience of twenty-one patients. Br J Rheumatol. 1995; 34: 68-77
7. Bachrach LK, Ward LM. Clinical review 1: Bisphosphonate use in childhood osteoporosis. J Clin Endocrinol Metab. 2009; 94: 400-409

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