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# Idiopathic juvenile osteoporosis: A case report and review of the literature

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ABSTRACT

*INTRODUCTION:* Idiopathic Juvenile Osteoporosis is an uncommon condition that has few case reports in the literature. Reported series indicate that it is a condition classically accompanying vertebral and metaphyseal fractures during the immediate pre-puberty years but that seems to develop naturally during puberty. Current clinical treatment is complicated because of lack of understanding on the origins of Idiopathic Juvenile Osteoporosis.
*PRESENTATION OF CASE:* The 13-year-old female patient with no former complaints had pain in her left hip while walking 2 years ago. Excluding the secondary osteoporosis reasons, the patient was diagnosed with Idiopathic Juvenile Osteoporosis and after the medical treatment she was followed-up.
*DISCUSSION:* The patient was subjected to a rehabilitation program for muscle weakness. She had difficulty in walking as a result of prolonged immobilization. At the end of a two-year treatment, significant improvement was achieved in muscle strength in the extremities, walking distance, and posture.
*CONCLUSION:* With this report, we would like to raise awareness about a possible association of persistent fractures with this rare metabolic disorder, Idiopathic Juvenile Osteoporosis, which should be included in differential diagnosis of patients with persistent appendicular skeleton fractures.


## 1. Introduction

Idiopathic Juvenile Osteoporosis is a sporadic disease of children manifesting between the 2nd and 14th years of life with pain in bones, fractures and deformity of the axial and appendicular as a part of clinical evaluation skeleton after minimal traumas [1]. It is important to differentiate the diagnosis for conditions causing generalized osteoporosis in childhood. It is diagnosed by excluding the other causes [2–6].

The purpose of this study is to evaluate the clinical symptoms of a patient diagnosed with Idiopathic Juvenile Osteoporosis by using radiographic and biochemical examinations.

## 2. Presentation of case

The 13-year-old female patient with no former complaints had pain in her left hip while walking 2 years ago. After left femoral neck fracture was detected in the patient with no trauma history, she was given surgical treatment in the orthopedic clinic. She was sent to our hospital because of a 2 years' history of back pain, knee pain, feet pain and difficulty in walking. She was given conservative treatment because of radius distal fracture, humerus proximal and diaphysis fracture which developed without trauma (Figs. 1 and 2). There was no association with a trauma or additional diseases. Examination results of the other systems were normal. There was no family history of frequent fractures, childhood or adolescent osteoporosis, osteogenesis imperfecta, gross skeletal anomalies, rickets, discolored sclera, or early onset of hearing loss.

The pubertal development was as in Tanner Stage 2. The sclerae were white and the teeth were unaffected. The hearing of the patient was normal. Her Body Mass Index was 22.6 kg/m$^2$. Complete blood cell count and electrolytes were normal. Serum calcium (8.7 mg/dL), phosphorus (4.6 mg/dL), bone alkaline phosphatase 420 IU/L (normal <345 IU/L), blood urea (6 mg/dL), serum creatinine (0.5 mg/dL), urine pH (6,2), and blood gases were within normal ranges. Parameters of liver function, creatine kinase, serum protein electrophoresis, rheumatological tests, glucose, and erythrocyte sedimentation were all in the appropriate reference ranges. Twenty-four-hour urinary calcium excretion was normal. There were no proteinuria or aminoaciduria. Serum

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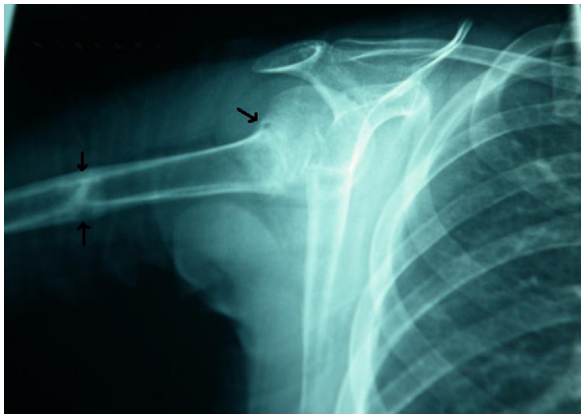

**Fig. 1.** Demonstrating the long thin diaphyses and the widespread disorganized metaphyses. The appearances on the right were almost identical (black arrows).

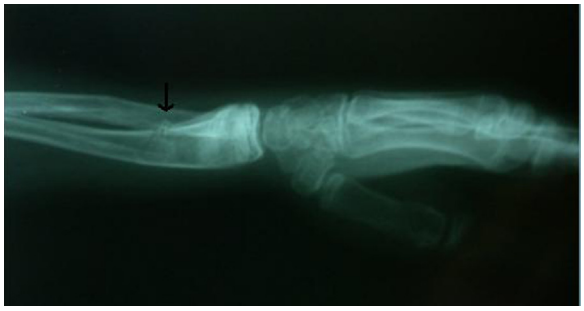

**Fig. 2.** Right radius distal angulation deformity (black arrow).

ceruloplasmin (15 mg/dL) and copper values (78 μg/dL) were also normal. The use of any medications known to affect the bone metabolism, including glucocorticoids, heparin, cyclosporine, GnRH agonist, L-thyroxin, methotrexate was excluded. The thyroid function test, parathyroid functions, somatomedin C, follicular stimulating hormone, luteinizing hormone, estradiol, plasma cortisol and urinary cortisol were normal. The bone density examination of the skeleton was carried out with dual-emission X-ray absorptiometry based on the pediatric program, which includes international reference values for children. The bone mineral density of the lumbar vertebra (L1–L4) was assessed in the "hip" and "spine" programs. The Spine Z-score was −2.63, the Hip Z-score was −2,04. Transiliac bone biopsy was carried out in our patient to exclude systemic disease. The histomorphometric parameters of the skeleton were not determined. However, consent to skin biopsy for collagen analysis, and no disorder of collagen synthesis was found. Extensive radiologic and biochemical tests were performed to exclude secondary osteoporosis. The patient was diagnosed with Idiopathic Juvenile Osteoporosis.

*Differential diagnosis*:

- osteogenesis imperfecta,
- rickets,
- Turner's syndrome,
- malabsorption syndrome,
- Wilson's disease,
- Osteoporosis,
- pseudoglioma syndrome,
- homocystinuria,
- acquired disorders such as celiac disease, medical diseases (hypothyroidism, diabetes mellitus, malabsorption disorders, anorexia nervosa, etc.),
- immobilization,
- malignancy,
- glucocorticoids [2–6].

The patient was given a treatment which included alendronate 35 mg/week, supplemental calcium (2000 mg/day) and vitamin D (400 IU/day). Since she developed spontaneous fracture despite the treatment for 6 months and her Z-score of the Bone Mineral Density was (L1–L4): −2.94, the treatment protocol was revised. The former treatment was terminated and a new one was started including 0.5 mg/kg/day pamidronate for 2 days every 3 months, calcitonin 2/week 200 Ü, calcitriol 2/week 0.25 mcg. Following the 2-year treatment Z- scores were normalized.

The patient, who had sustained immobilization due to multiple fractures and fear to fall, was subjected to rehabilitation program after she developed difficulty to walk, flexion posture in the body, limitation in joint mobility clearance and weakness in lower and upper extremity muscles.

She received programs including strengthening in joint mobility clearance in extremities, back muscles, and posture exercise and walking training. At the end of 2-year treatment, significant improvements were observed in walking distance, lower and upper extremity muscle strengths and posture.

## 3. Discussion

Idiopathic Juvenile Osteoporosis was first described by Schippers in 1938 [1]. Once all of the known causes of decreased bone mass are excluded, there remains one form of osteoporosis over IJO, which was first recognized by Dent in 1965 [1,7]. Due to the rare prevalence of Idiopathic Juvenile Osteoporosis, only approximately 100 cases have been reported in the literature, and its descriptions have been limited to case reports [1].

Idiopathic Juvenile Osteoporosis is characterized with a 2-fold dysfunction of cancellous bone formation. First, fewer remodeling cycles are initiated. This will not be detrimental to bone integrity in the short run, as the recruitment of osteoblast and osteoclast teams are similarly decreased. Second, the amount of bone formed in each remodeling cycle is decreased. The consequences are the thinning of mature trabeculae and possibly decreased production of secondary trabeculae in the metaphyses [8].

No heritable genetic mutations have been identified in patients with IJO and absence of family members with a history of pediatric or adolescent osteoporosis suggests IJO [9–11]. Autosomal recessive inheritance has been suggested by Hou and Wang [12]; unfortunately, they did not elaborate on this suggestion nor offer any specific scientific support [4].

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 as Osteogenesis Imperfecta and Idiopathic Juvenile Osteoporosis. The differentiation between osteogenesis imperfecta and Idiopathic Juvenile Osteoporosis may be difficult, especially in milder cases [9,13]. Extra-skeletal manifestations, including those which are typical for osteogenesis imperfecta (blue sclerae, dentinogenesis imperfecta, joint hyperlaxity, deafness, cardiac lesions), were absent [5,13].

Osteogenesis imperfecta is a group of genetically inherited disorders characterized by bone fragility. All forms of osteogenesis imperfecta involve a structural defect in the production of Type I collagen [4,13]. Thus, with this clinical exercise along with relevant investigations to exclude many other conditions with osteoporosis, we arrived at a diagnosis of Idiopathic Juvenile Osteoporosis. 25 OH D3 vitamin levels were not measured in our study and this was a limitation.

Current treatment of patients with Idiopathic Juvenile Osteoporosis is limited to bisphosphonate therapy, calcium, and vitamin D supplementation. Limited physical activity will assist in limiting fragility fractures. Furthermore, studies are indicated in the quest to seek the etiology of Idiopathic Juvenile Osteoporosis [1,4,14]. Further research is needed to learn whether the Idiopathic Juvenile Osteoporosis was recovered with the treatment given, or it was a self-limiting disease.

## 4. Conclusion

Important clues which guided us to the diagnosis of Idiopathic Juvenile Osteoporosis were:

- Absence of family history of pediatric or adolescent osteoporosis.
- Presence of osseous osteoporosis on radiography.
- Absence of collagen defect on skin biopsy.
- No other identifiable causes of bone loss.

## Conflicts of interest

We certify that there is no conflict of interest with any financial organization regarding the material discussed in the manuscript.

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## Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chiefof this journal on request.

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