# A 13 Year-Old Boy Diagnosed As Osteogenesis Imperfecta With Normal Bone Mineral Density

**Tuba Tınastepe[1], Berna Eroğlu Filibeli[2], Gönül Çatlı[3], Bumin N. Dündar[3]**

*[1] Tepecik Training and Research Hospital, Department of Pediatrics, Izmir, Turkey*
*[2] Tepecik Training and Research Hospital, Department of Pediatric Endocrinology, Izmir, Turkey*
*[3]Katip Celebi University, Department of Pediatric Endocrinology, Izmir, Turkey*

## BACKGROUND

Osteogenesis imperfecta is a hereditary connective tissue disease developing based on the structure or synthesis impairment of type 1 collagen and proceeding with diffuse osteoporosis, fragility, fractures and deformities in bones. Bone mineral density can be at normal or even high levels particularly especially in type I and XIII. Here, a 13 year-old boy diagnosed as osteogenesis imperfecta with normal bone mineral density was presented and treatment results were shared.

**Physical examination;**
Lenght 0,57 SDS
Weight 1.31 SDS,
Tanner puberty stage: 3
No deformity finding in extremities
Other system examinations were normal

**Genetic analysis**
It was identified that there was a p.1119T (c.3355G>A) heterozygous mutation in COL1A2 gene.

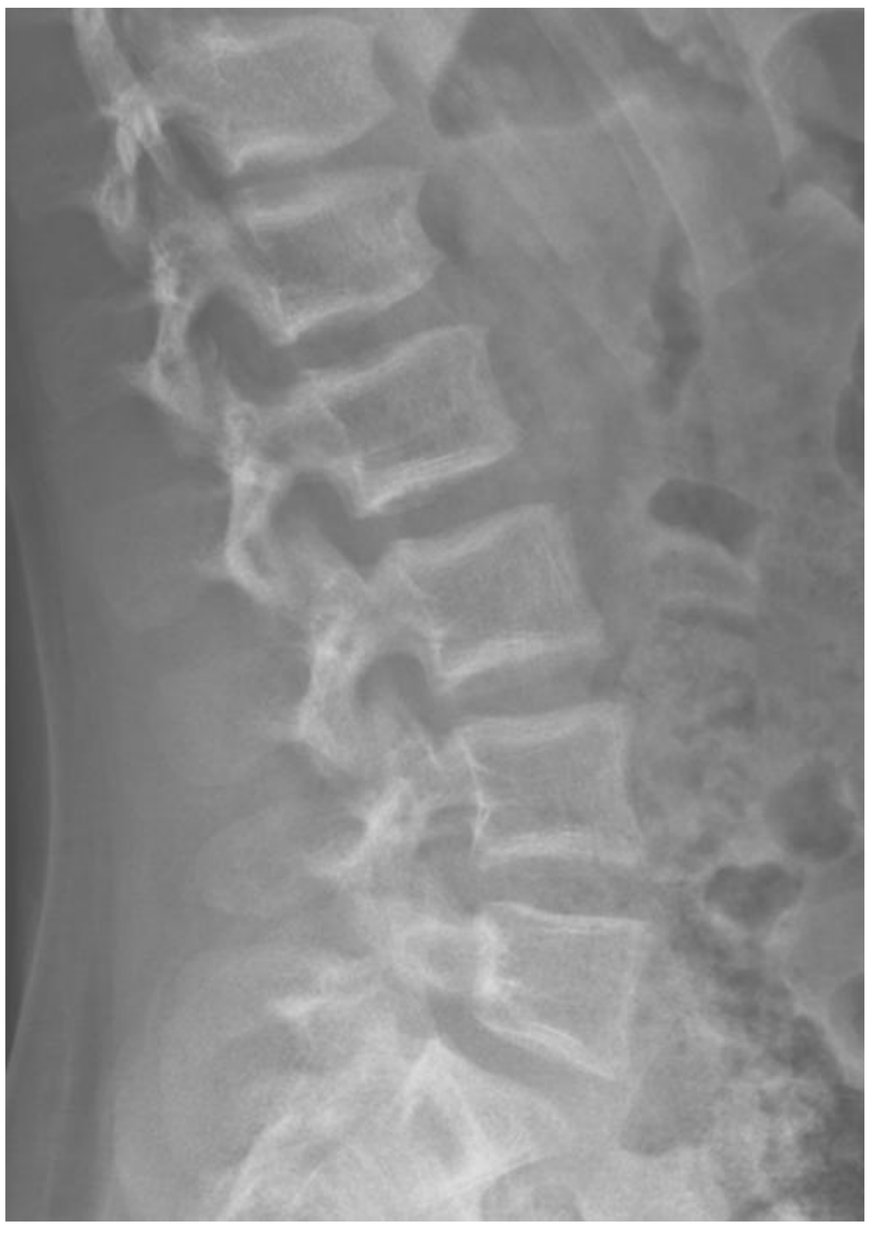

Figure 1: Lateral radiograph of lumbar

## CASE REPORT

A 13-year-old male patient was brought to hospital with complaint of frequent fracture in bones.
These fractures were caused by minor traumas in the arms and legs.
He was born as term 3100 grams and there was no known other disease histories.
In family history, there were frequent bone fractures in father, uncle, aunt, grandmother and cousins.

**Laboratory evaluation;**
Ca: 10.1 mg/dl (N, 8.8-10.6), P: 4.8 mg/dl (N, 4-7), ALP: 376 U/l (N, 74-390), PTH: 38.5 pg/ml (N, 10-69) and 25(OH) D3: 19.2 ng/ml (N, 20-100).
**Radiologic evaluation:** In extremity graphics, no radiological finding that made us think osteogenesis imperfecta or skeletal dysplasia and compression fracture was determined in vertebra graphics.
**Dual energy X-ray absorptiometry (DEXA):** Z score 1.5 SD (according to bone age)
**Audiological investigation:** Normal

Due to recurrent bone fractures, alendronat sodium and vitamin D treatment were initiated. In the first month of the treatment, only minor fracture was developed in his finger and foot following the trauma. DEXA corrected z score taken in the first year following the treatment was established as 5.76. The aunt and uncle of the patient were found out to have osteogenesis imperfecta. The treatment has been continued since there is a clinical response to alendronat sodium therapy.

## CONCLUSION

In osteogenesis imperfecta, while bone mineral density is usually determined low and it increases with the treatment, it can be normal in some types. In the treatment of these patients, treatment plan should be performed by considering parameters like fracture frequency, pain, mobilization and quality of life.