



## Case Report

# Autosomal Dominant Type II Osteopetrosis in an Asymptomatic Adolescent: A Case Report

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### Abstract

Osteopetrosis is a heterogeneous group of heritable conditions in which there is a defect in bone resorption by osteoclasts. The disease has variable mode of inheritance with variable expression of severity. We are reporting a 14 year old asymptomatic girl with autosomal dominant type II osteopetrosis and then the literature is reviewed.

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### Introduction

The term osteopetrosis is derived from the Greek 'osteo' meaning bone and 'petrōs', stone. Osteopetrosis is variably referred to as 'marble bone disease' and 'Albers-Schönberg disease', after the German radiologist who described the condition first in 1904 in a 24-year-old man with generalized skeletal sclerosis.<sup>1</sup> Karschner coined the name osteopetrosis in 1922.<sup>2</sup> Osteopetrosis is a heterogeneous group of heritable conditions in which there is a defect in bone resorption by osteoclasts.<sup>3</sup> Three major clinical forms account for most of the cases: the autosomal dominant adult (benign) type; the autosomal recessive infantile (malignant) type, which if untreated is typically fatal during infancy or childhood; and the autosomal recessive (intermediate) type that presents during childhood with some of the signs and symptoms of malignant osteopetrosis.<sup>4</sup>

In 2006, detailed classification of osteopetrotic conditions have been done based on clinical pattern, inheritance, underlying gene defect, and the protein defect responsible for different types of osteopetrosis.<sup>5</sup> In the new revised classification 13 different clinical forms of osteopetrosis have been described. These

conditions are rare, and their overall incidence is difficult to estimate. Autosomal recessive osteopetrosis has an incidence of 1 in 250,000 births, with a particularly high incidence reported in Costa Rica (3.4:100,000). Autosomal dominant osteopetrosis has an incidence of 5:100,000 births.<sup>6,7</sup>

Autosomal recessive osteopetrosis, also known as "malignant" osteopetrosis, is a life-threatening condition, which classically manifests in the first few months of life. The increase in bone density can paradoxically weaken the bone, resulting in a predisposition to fractures and osteomyelitis. The longitudinal growth of bones is impaired, resulting in short stature of varying degrees. Macrocephaly and frontal bossing develop within the first year, resulting in a typical facial appearance. The skull changes can result in choanal stenosis and hydrocephalus. The expanding bone can narrow nerve foramina, resulting in blindness, deafness, and facial palsy.<sup>8</sup> The affected child has a reduced marrow space, resulting in extramedullary hematopoiesis. It is lethal within first 10 years of life if bone marrow transplantation is not performed.<sup>9</sup> Autosomal recessive osteopetrosis (ARO) has some variants i.e. Neuropathic ARO, and ARO with

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renal tubular acidosis.<sup>5</sup> Autosomal dominant osteopetrosis, also labeled as Albers-Schönberg disease in new revised classification, typically has onset in late childhood or adolescence, and classically displays the radiographic sign of “Rugger Jersey spine” (dense bands of sclerosis parallel to the vertebral endplates). This radiographic sign is also described as “Sandwich vertebra” by some authors. The main complications are confined to the skeleton, including fractures, scoliosis, hip osteoarthritis and osteomyelitis, particularly affecting the mandible in association with dental abscess or caries. Cranial nerve compression is a rare but important complication, with hearing and visual loss affecting around 5% of individuals.<sup>10</sup> This autosomal dominant form can be further subdivided into type I; where there is marked thickening of cranial vault but with an almost normal spine, and type II form; where there is a sclerotic skull base and “Rugger Jersey spine”.<sup>11</sup>

We are reporting incidentally diagnosed autosomal dominant type II osteopetrosis in an asymptomatic adolescent girl.

### Case report

A 14 year old girl attended a private hospital of Rajshahi with the complaint of low back pain. She gave history of fall while playing in the school. She did not have any physical complaints before this incident. Because of patient's eagerness to be investigated, X-ray of dorsolumbar spine was advised. There was no evidence of fracture, but to the surprise of the physician, there were dense bands of sclerosis in vertebral bodies giving the impression of “Rugger Jersey spine”. Detailed skeletal survey was advised to evaluate the condition further. The patient did not give any past history of fracture of any bone. None of her family member had any skeletal problem.

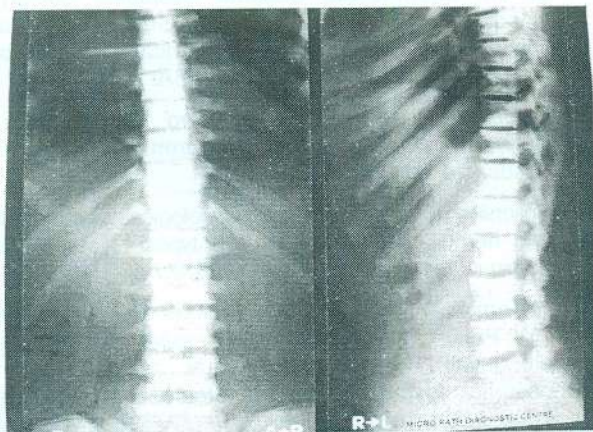


Figure 1, 2, and 3: Dorsal and lumbar spine showing “Rugger Jersey spine” appearance with horizontal sclerotic bands lying just deep to the vertebral end plates. Ribs are also sclerotic and there is “bone within a bone” appearance.



Figure 4 and 5: X-Ray pelvis and long bones showing diffuse sclerosis with almost obliterated medullary cavity. “Bone within a bone” appearance is noted in both iliac wings.





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