A RARE CAUSE OF INCREASED BONE MINERAL DENSITY IN ADULTHOOD: OSTEOPETROSIS

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OBJECTIVES

Osteopetrosis is a rare genetic disorder of reduced osteoclastic bone resorption. Defective bone remodelling induces skleletal sclerosis and abnormally dense but brittle bones. Three distinct forms of the disease (infantile, adult and intermediate forms) have been recognised. We present here a case report of autosomal dominant osteopetrosis type II as a cause of high bone mineral density (BMD) diagnosed at adult age.

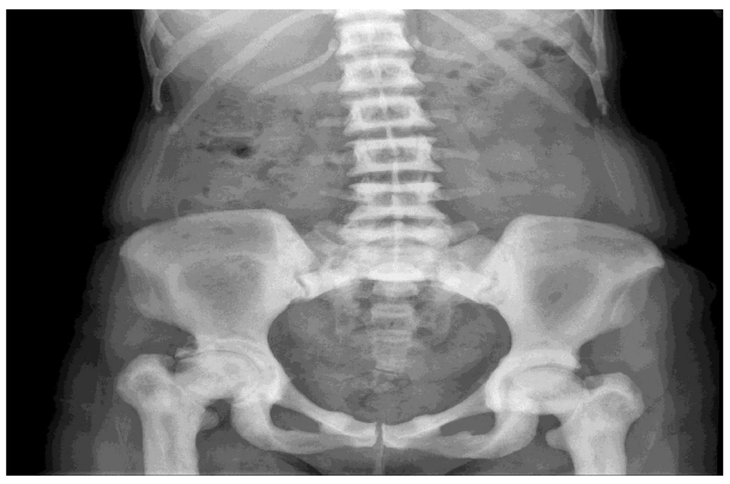

Figure-1: Bone within bone appearance in pelvis

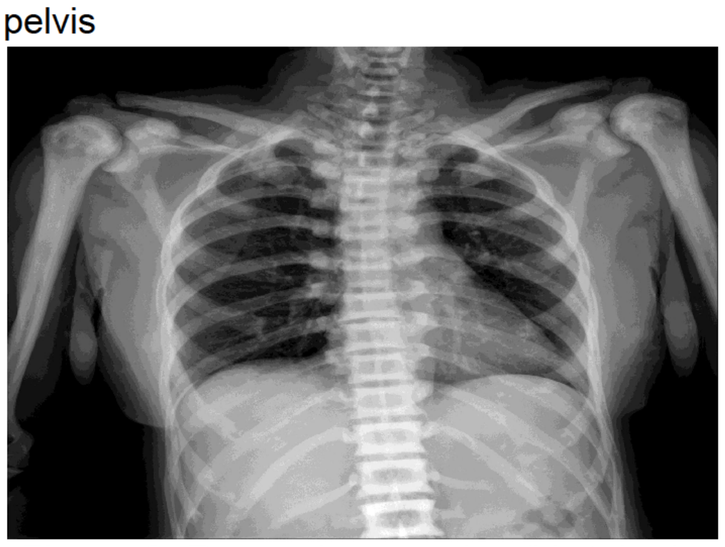

Figure-2: Rugger-jersey spine (Vertebral end plate thickening)

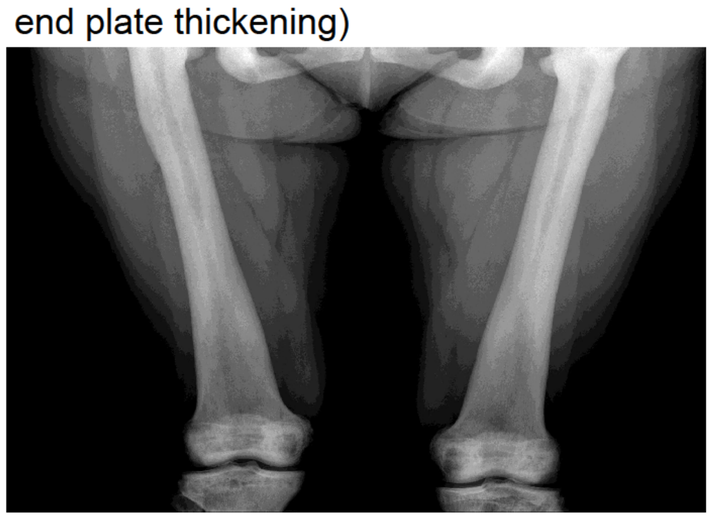

Figure-3:Transverse sclerotic bands within distal femur

Table-1: Biochemical evaluation of the patient

Ca (8,6-10,2 mg/dl)	9,52	**WBC (4,5-10,5x10³/uL)**	4,56
PO4 (2,6-4,5 mg/dl)	4,1	**Hb (11,5-15,5 gr/dl)**	12,4
PTH (15-65 pg/ml)	35	**Htc (35,5-48%)**	35,9
25OHD3 (ng/ml)	7,9	**Plt (150-400x10³/uL)**	306
ESR (mm/hr)	30	**Tryptase (<11,5mg/L)**	3,6
Urea (0-50 mg/dl)	27	**Fluoride (0-4 mmol/L)**	<1
Cr (0-0,95 mg/dl)	0,74	**IGF-1 (87-238 ng/ml)**	109
ALT (0-33 U/L)	27	**Acid phosphatase (0-5,5 U/L)**	17,6
Anti-HCV	negative		

CASE REPORT

A 52 year old woman referred to Endocrinology Outpatient Clinic for the evaluation of high BMD. She had a complaint of bone pain and headache. Her medical history and physical examination were unremarkable. Biochemical evaluation showed normal serum calcium (Ca) and phosphorous (PO4) with elevated serum parathyroid hormone (PTH) and decreased 25OH vitamin D3 concentrations (25OHD3). Increased BMD in DXA scanning affecting both spine and hip was classified as generalized and acquired causes were evaluated initially. Her serum fluoride concentration, renal function tests, serum IGF-1 level, serological tests for hepatitis C and liver function tests were all normal excluding fluorosis, renal osteodystrophy, acromegaly, hepatitis C associated osteosclerosis. There was no splenomegaly or abnormality of full blood count for miyelofibrosis. Serum tryptase level was normal excluding mastocytosis. She denied any usage of oestrogen implants. Excluding the acquired causes, we looked for the specific charactheristic features suggesting monogenic disorders such as osteopetrosis or sclerosing bone dysplasias. Skull sclerosis, rugger-jersey spine due to vertebral end plate thickening, bone within bone in pelvis and transverse sclerotic bands within distal femur, which are the classic signs of osteopetrosis, were recognized at plain radiographs. She denied any history of fracture and also, the whole body radiographic survey showed no pathological fractures. Her serum acid phophatase was high with 17,6 U/L (reference:0-5,5 U/L). She was diagnosed as adult type osteopetrosis type II. Complication screening revealed only mild decrease in high-pitched voice in audiology.

CONCLUSION

Autosomal dominant osteopetrosis is the most common form of the disease. It exhibits a heterogeneous trait with milder symptoms, often at later adolescence or adulthood. Two distinct types (type I and II) have been described on the basis of radiographic, biochemical and clinical features. The disease requires no treatment although the complications may require intervention.

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Calcium, vitamin D and bone

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