Autosomal dominant osteopetrosis
(Albers-Schönberg disease)
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Section: Musculoskeletal system
Area of Interest: Bones
Procedure: Diagnostic procedure
Technique: Conventional radiography
Special Focus: Genetic defects
Case Type: Clinical
Cases
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Patient: 57 years, male

Clinical History:

A 57 year old male patient was admitted to hospital because of fever and syncope. Initial blood samples showed pancytopaenia and the patient was initially suspected of having leukaemia.

Imaging Findings:

As the patient was predisposed to osteopetrosis, both his mother and daughter were known with this disease, x-ray of the full skeleton was performed. X-ray showed diffuse osteosclerosis and generalised hyperdense bone structure. There was cortical thickening and the tubular bones showed medullary encroachment. X-ray of the spinal column showed the bone–within–bone appearance and also the "sandwich" vertebrae appearance.

Discussion:

Osteopetrosis is a rare group of hereditary disorders of the skeleton characterised by increased bone density on radiographs. The disease is caused by failure in osteoclast differentiation or function with impaired bone reabsorption producing sclerotic and brittle bones. It is a heterogenic group of disorders varying greatly in presentation and severity (1). The disease ranges from the "malignant" neonatal form (autosomal recessive osteopetrosis, ARO) with life-threatening complications such as bone marrow failure and carries a poor prognosis to the more "benign" form of adult onset osteopetrosis (autosomal dominant osteopetrosis, ADO, also known as Albers-Schönberg disease) which can be an incidental finding on radiographs and has a normal life expectancy. The incidence of the autosomal recessive form is estimated to be 1 in 250,000 births and the incidence of the autosomal dominant form is estimated to be 1 in 20,000 births (2). Mild forms of the disease have no symptoms while the more severe forms can present with fractures, osteomyelitis, skeletal deformity, failure to thrive, bone marrow depression with anaemia, infections and hepatosplenomegaly (due to extramedullary haematopoiesis), blindness, facial paresis and deafness (due to bone compression of nerves). The diagnosis is clinical and largely depends on the radiographic appearance of the skeleton (2). A bone biopsy and genetic tests can be used to confirm the diagnosis and differentiate between different subtypes of osteopetrosis. No effective treatment of osteopetrosis exists so treatment is primarily symptomatic of complications such as fractures, osteomyelitis and arthrosis. In the severe form of ARO haematopoietic stem cell transplantation can be a last resort (2, 3, 4).

The patient from this case recovered fully and his blood samples normalised, the pancytopaenia was ascribed as a
viral infection.

**Differential Diagnosis List:** The final diagnosis was osteopetrosis (ADO), Paget’s disease, Beryllium, lead and bismuth poisoning. Other conditions which causes secondary bone sclerosis, hypoparathyroidism

**Final Diagnosis:** The final diagnosis was osteopetrosis (ADO).

**References:**


Figure 1

**Description:** Hyperdense bone structure. Bone-within-bone and "sandwich vertebrae" appearance (arrows). **Origin:** Department of Radiology, University Hospital of Herlev, Copenhagen, Denmark
Figure 2

Description: Hyperdense bonestructure. Bone-within-bone and "sandwich vertebrae" appearance (arrows). Origin: Department of Radiology, University Hospital of Herlev, Copenhagen, Denmark
Description: Hyperdense bonestructure and medullary encroachment of the femoral bones (arrow).
Origin: Department of Radiology, University Hospital of Herlev, Copenhagen, Denmark