Osteopetrosis

aka Albers-Schönberg/Marble Bone disease

- First described by German Radiologist Albers-Schönberg 1904
- Very rare inherited disorder
- Incidence 1 in 100,000 – 500,000
- Failure of osteoclasts to resorb bone, modelling and re-modelling impaired.

- Bone is thick but structurally weak and brittle.
- Can also cause haematopoietic insufficiency, nerve entrapment syndromes, growth impairment.

Infantile autosomal recessive

- Failure to thrive
- Anaemia, leukocytopenia, thrombocytopenia (can cause SAH)
- Cranial nerve compression
- Hepatosplenomegaly
- Lymphadenopathy
- Poor prognosis; death usually by second decade (recurrent infection, massive bleed, terminal leukaemia)

Benign adult autosomal dominant

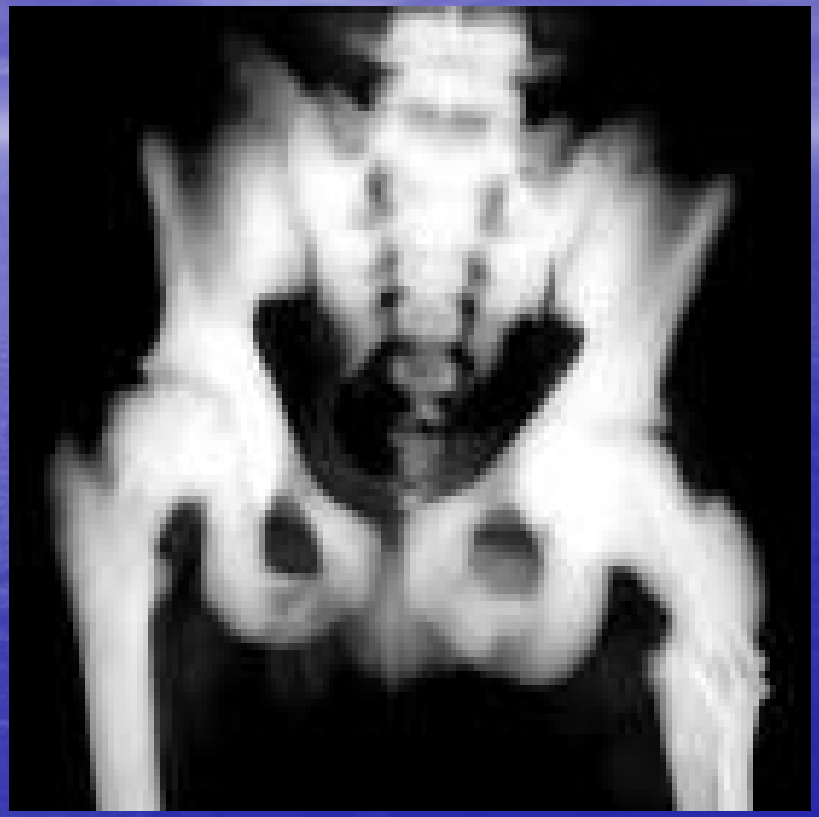
- 50% asymptomatic
- Recurrent fractures, mild anaemia, occasionally cranial nerve palsy
- Prognosis: Normal life expectancy

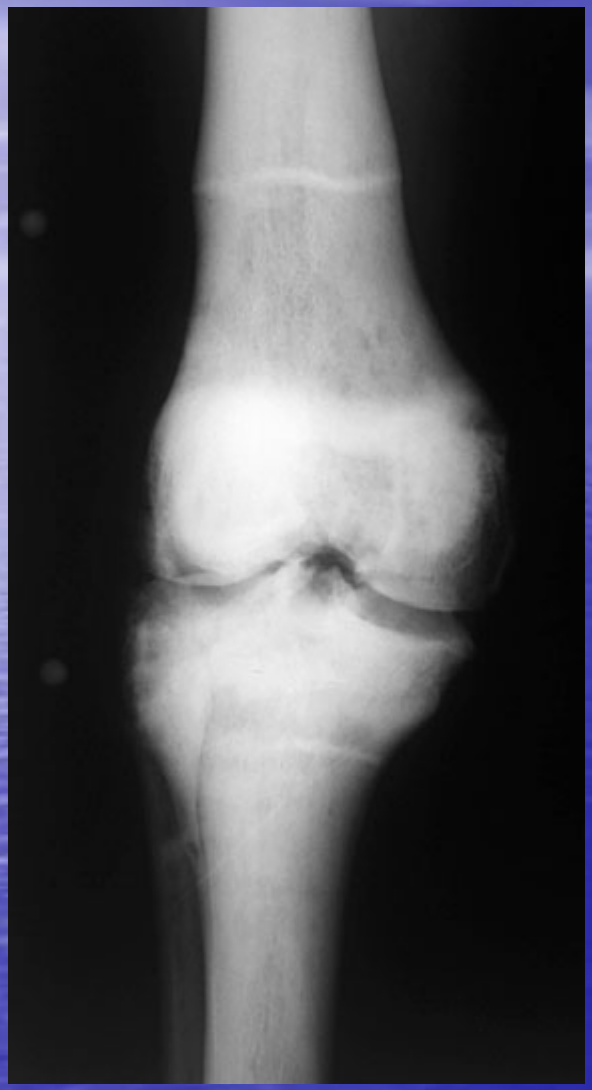
Radiological appearances

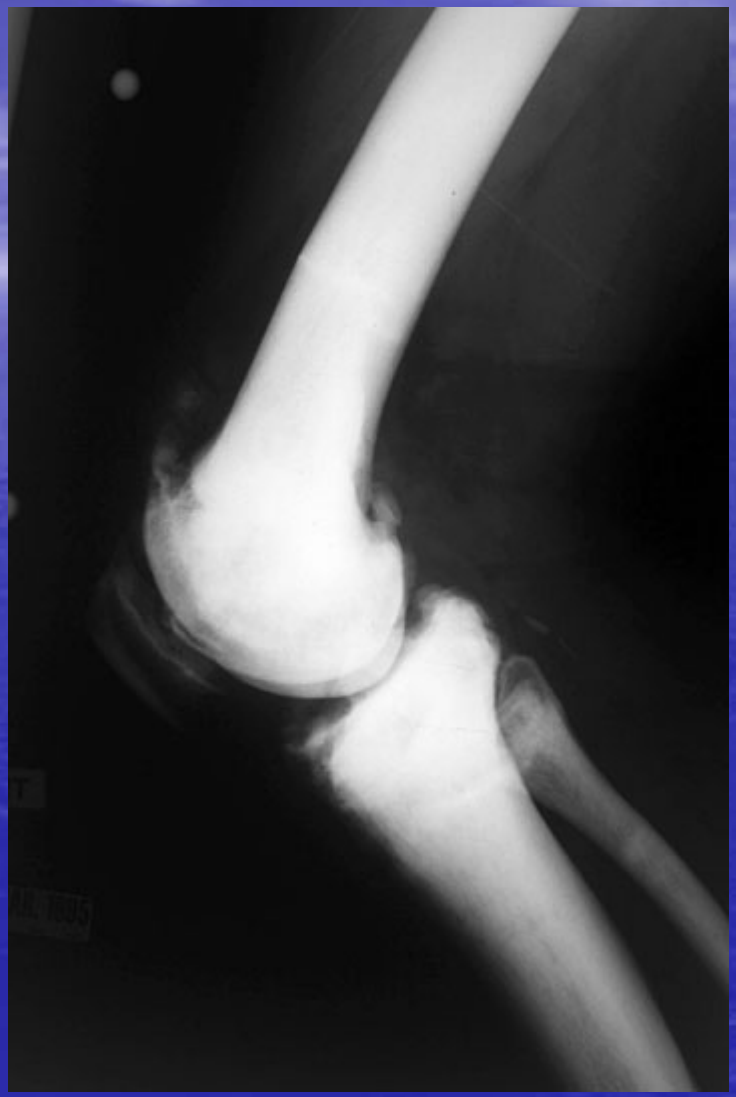
- Diffuse osteosclerosis, medullary compartment often obliterated by immature unresorbed bone. Mandible least commonly involved
- Erlenmeyer flask deformity – clublike long bones and flaring of ends
- Bone within bone appearance (endobone)
- Rugger-jersey spine
- Alternating sclerotic + radiolucent transverse metaphyseal lines indicate fluctuating course of disease.
- Obliteration of mastoid cells, paranasal sinuses, basal foramina by osteosclerosis
- Transverse fractures + lots of callus

Differential diagnoses

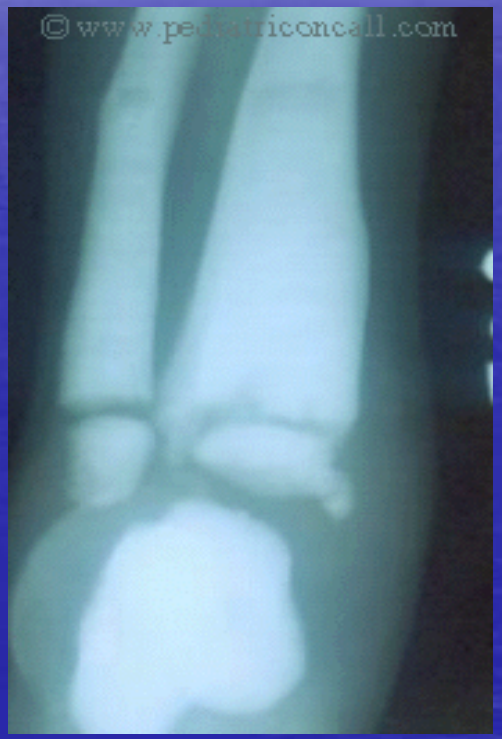
- Heavy metal poisoning (lead)
- Fibrous dysplasia of skull/face
- Melorheostosis
- Pyknodysostosis
- Hypoparathyroidism
- Pagets

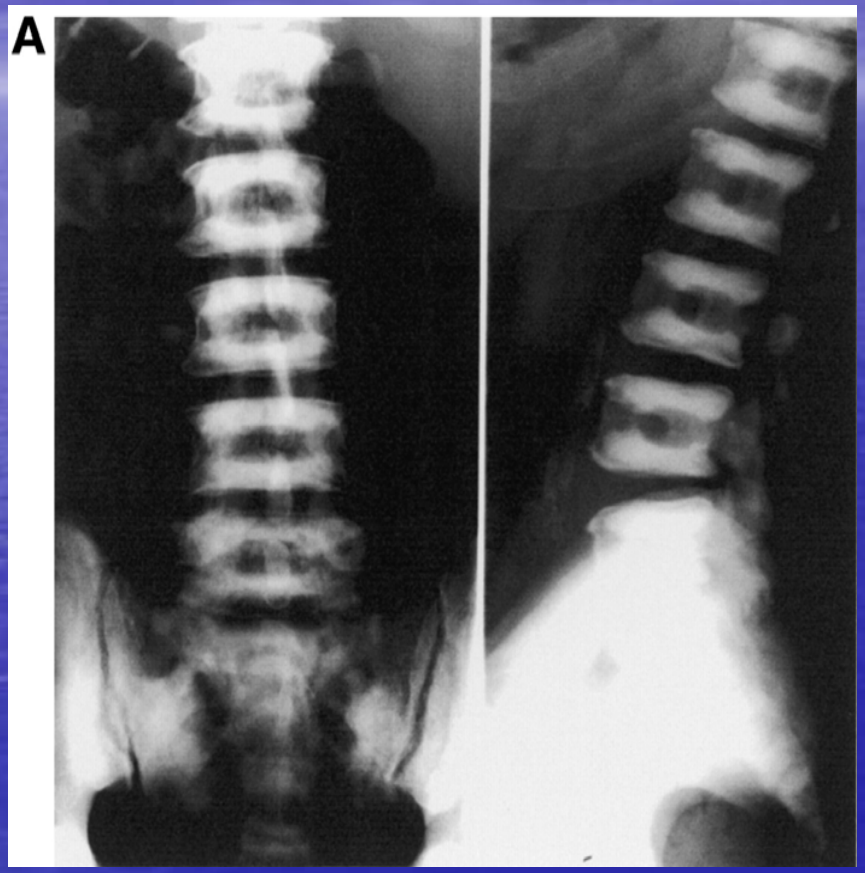






- Bone on bone appearance
- Chronic osteomyelitis of tibia

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The end!