
SPOT DIAGNOSIS (IMAGE GALLERY)



BONE DISEASE

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Spot Diagnosis.

Osteopetrosis.

It encompasses a heterogeneous group of inherited disorders of the skeleton caused by defect in bone resorption by the osteoclasts. Several types are recognised: Autosomal Recessive Osteopetrosis {ARO} or Osteopetrosis congenita, infantile or malignant osteopetrosis is the most

severe one. Patients present with failure to thrive and growth retardation, bony defects, nasal stuffiness, short stature, frontal bossing, nystagmus and genu valgum. Hematological abnormalities include pancytopenia, hepatosplenomegaly, hypersplenism, and hemolysis. Neuropathies related to cranial nerve entrapment are deafness, proptosis, and hydrocephalus. Bones are fragile and can fracture easily. Radiologic features are usually diagnostic. There is generalized osteosclerosis. The bones might be club like or appear like a bone within bone {endobone}. The entire skull is thickened and dense, especially at the base. Sinuses are small and under-pneumatized. Vertebrae are extremely radiodense. They may show alternating bands, known as the rugger-jersey sign.

Autosomal Dominant Osteopetrosis {ADO} or Albers-Schönberg disease or ADO type II is the milder type frequently observed in adults. Life expectancy is not altered. Intermediate Severity Osteopetrosis {IRO} is less diffuse form with relatively few cases identified so far worldwide. It is diagnosed in the first decade of life. X-linked {XLO} has been recently described. It is associated with ectodermal dysplasia, lymphedema and immunodeficiency {so-called OL-EDA-ID syndrome}.

Vitamin D {calcitriol} stimulates dormant osteoclasts and thus stimulates bone resorption. It usually produces only modest clinical improvement, which is not sustained after therapy is discontinued. Interferon-gamma improves white cell function, tremendously decreasing the incidence of new infections. Erythropoietin can be used to correct anemia. Corticosteroids stimulate bone resorption and treat anemia. Bone marrow transplantation is the treatment of choice.

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