A male Muslim patient aged 14 years presented with swelling and pain in left thigh after fall from bicycle. Patient had past history of multiple fractures in opposite limb.

Clinical examination revealed tenderness and crepitus at local site.

X-RAY left thigh (Fig.1) was performed which shows fracture in mid shaft of femur and generalized increase in bone density. A skeletal survey was performed which is given below.

What is your diagnosis?
Radiological Diagnosis

PYKNODYSOSTOSIS

Fig. No. 5 (A)-- Skiagram Right Leg Ap And Lateral View Fig. No. 5 (B)-- Skiagram Pelvis With Both Hip Joints

Discussion

Pyknodysostosis is a sclerosing skeletal dysplasia transmitted by autosomal recessive trait and characterized by increased bone density, dwarfism and skeletal fragility.

It was described first by Maroteaux and Lamy in 1962. 20% of children are born from consanguineous marriages with male : female ratio of 2:1.

Patients are typically dwarf with height less than 145 cms and characteristic facial features involving prominent forehead, beaked nose and receding jaw, proptosis of eyes and dental anomalies(4). Hands and feet are stubby with spoon shaped nails.10% patients show mental retardation. Patients are usually diagnosed in childhood and young adult life. Life expectancy is usually normal(3).

The pathological changes are non-specific; it shows abnormally dense cortical bone with tiny harvesian canals and coarse fibrous bone. Marrow is replaced by the persistent, mineralized osteochondroid material. Peculiar radiological findings noted are;

- Diffuse increase in bone density
- Spool shaped vertebrae
- Hypoplasia of mandible with obtuse mandibular angle
- Wormian bones
- Widely open cranial sutures and absence of frontal sinuses

Differential diagnosis (4).

1. Osteopetrosis:--Lack of anaemia and preservation of medullary canal in pyknodysostosis helps in distinguishing it from osteopetrosis. Loss of corticomedullary differentiation with extramedullary haemopoiesis is noted in osteopetrosis.

REFERENCES:-

1. Diagnosis of Bone and Joint disorders, Resnick. 3rd edition, volume 5 page no.4204-05
2. Radiology of Skeletal Disorders;Exercises in diagnosis by:Ronald O Murray, Harold G Jacobson, Dennis J Stoker Vol-2 part 4 page no. 1496-97
4. Yochum and Rowe's Essentials of Skeletal Radiology;Third edition Chapter -8 Skeletal Dysplasia; page no.776-778