Fibrous dysplasia

Definition
Disease characterised by the replacement of normal bone and marrow by fibrous tissue and irregular minute bone spicules.

Fibrous dysplasia, precocious puberty and café-au-lait patches form the McCune-Albright syndrome

Epidemiology
Patients tend to be in the second or third decade. More common in females. Polyostotic fibrous dysplasia is much more common in females.

Site
Most common site is the proximal femur. The ribs and jaw are commonly involved. Involvement of the jaw may result in leontiasis and cherubism.

The lesion is typically intramedullary and diaphyseal.

Aetiology
Failure of skeletal maturation, not hereditary.

Clinical features
Many patients are asymptomatic.

Patients may develop deformities, particularly with polyostotic fibrous dysplasia, such as the classic shepherd's crook deformity of the proximal femur. Fractures develop on the tension side of affected bones.

Hyperthyroidism, hyperparathyroidism, hypophosphataemic rickets and osteomalacia have been noted in patients with monostotic and polyostotic fibrous dysplasia, and McCune-Albright syndrome.

Radiology
Well demarcated zone of rarefaction, often surrounded by a rim of sclerotic bone.

Expansion with thinning of the cortices is particularly likely in thin bones such as the ribs.

There may be a hazy appearance, classically called a ground glass appearance.

MRI: low T1, variable T2. Cartilage is sometimes seen which will lead to increased T2.

Bone scan: may be increased or decreased.

Laboratory findings
Increased alkaline phosphatase

Pathology
Composed of dense fibrous tissue, surrounding bony trabeculae arranged in a meaningless fashion, known as Chinese alphabet.

Collections of foam cells forming little islands amongst the fibrous tissue are almost always seen. Giant cells may also be seen.

Treatment
Usually expectant. The lesions tend to stop growing at skeletal maturity, however reactivation may occur during pregnancy or during estrogen therapy.

Large lesions in weight bearing bones need curettage and grafting, often with internal fixation. The use of cortical allograft should be considered, because it provides structural support and is slower to resorb.

Sarcomas may arise from fibrous dysplasia.

McCune-Albright syndrome
Genetic, noninheritable disease.

Caused by mutations in G proteins leading to activation of cyclic AMP.

The café-au-lait spots are smaller, darker and have a more irregular contour than those seen in NF (coast of Maine). NF lesions have a smoother contour, coast of California).