Leukaemia Section
Short Communication

Fibrogenesis imperfecta ossium
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Clinics and pathology

X-rays of the cervical, thoracic, and lumbar spine (from left to right), and of the pelvic girdle (bottom) showing a marked demineralization with paucity of coarse, essentially vertical, trabeculae.
**Disease**
Disorder of bone mineralization with abnormal bone collagen morphology often associated with monoclonal gammopathy; may well be a clinical variant of multiple myeloma.

**Etiology**
Presents as an acquired metabolic bone disease of unknown aetiology; may also be a genetic disorder (at least in some cases), since a father and his daughter were affected.

**Epidemiology**
25 cases diagnosed to date; onset of symptoms mostly in 50-60 yr-old patients.

**Clinics**
A combination of progressive and incapacitating bone pain and spontaneous, multiple fractures typically localized at tendon insertion sites; leads to extreme bone fragility, progressive immobility and usually results in the patient becoming bedridden. Serum alkaline phosphatase can be raised; monoclonal gammopathy is found in 25% of cases; 10 to 20% atypical plasma cells can be found in the bone marrow; however, evolution towards myeloma has never been reported. No other organ involvement has yet been reported. Diagnosis on bone biopsy showing the collagen defect.

**Pathology**
Mimics osteomalacia with abnormal bone mineralization but there is complete loss of the birefringence characteristic of oriented collagen fibers; at ultrastructural level the normal lamellar pattern of collagen fibers is replaced by curved and extremely variable in thickness collagen fibrils.

**Treatment**
Treatment with melphalan and corticosteroids over years has been successful in a number (but not all) of cases.

**Prognosis**
Median survival is about 3 yrs.

**Genetics**
Genes involved in the cases possibly inherited, if any, are unknown; genes involved in the plasma cells proliferation are also unknown.

**References**


This article should be referenced as such: