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Leukaemia Section

Short Communication

Fibrogenesis imperfecta ossium

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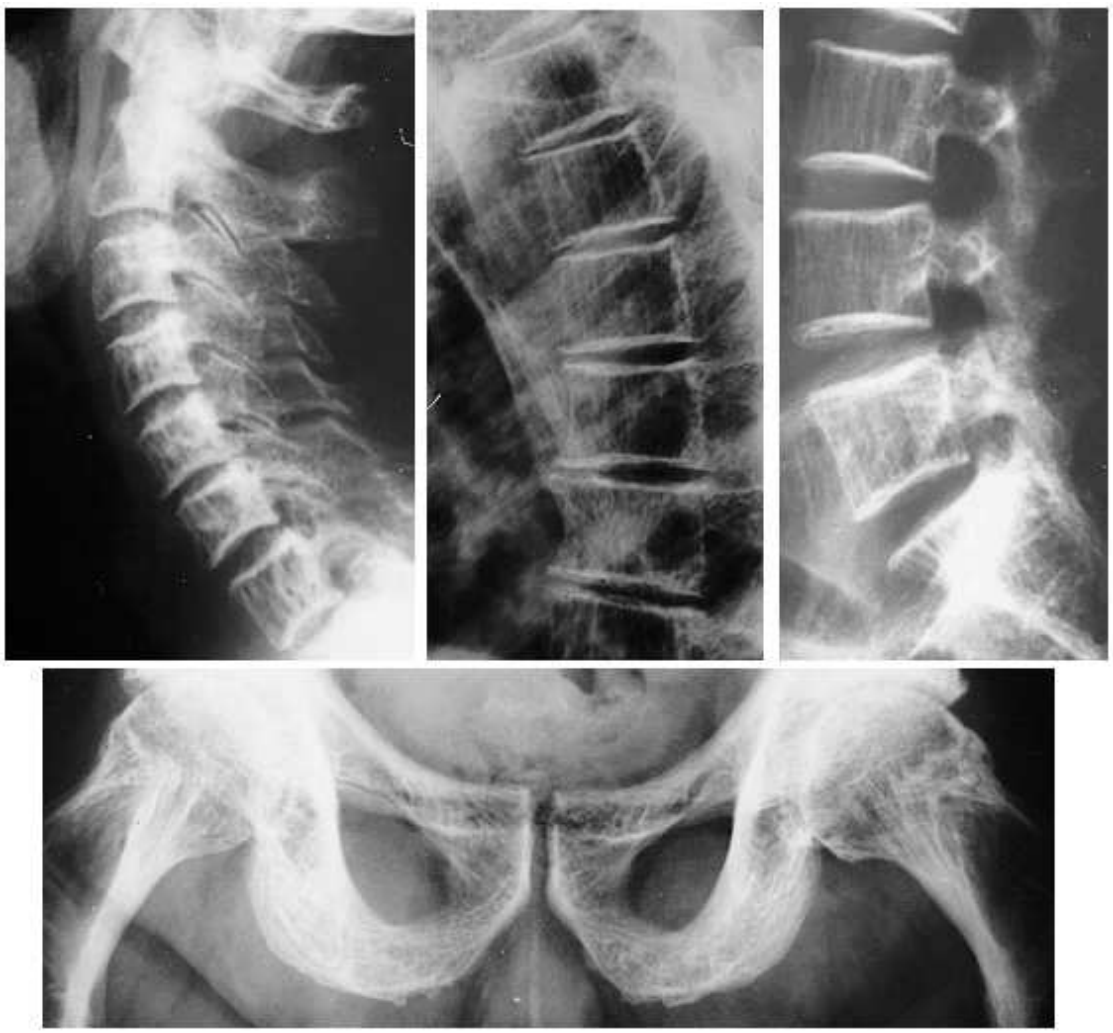
Clinics and pathology



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 daugther 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

Note

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

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