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

References


Thomas WC Jr, Moore TH. Fibrinogenesis imperfecta ossium. Trans Am Clin Climatol Assoc. 1969;80:54-62


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