Fibrous Dysplasia

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12.1 Related Conditions: McCune–Albright Syndrome, Mazabraud Syndrome

Definition: Intramedullary lesion consisting of a peculiar fibro-osseous tissue; it may be either monostotic or polyostotic.

Epidemiology: Monostotic fibrous dysplasia is frequent, polyostotic uncommon, and McCune–Albright syndrome is rare. Difficult to assess true incidence because often asymptomatic. Slight female predominance. Usually diagnosed between age 10 and 30. When asymptomatic, it can be discovered at any age. The polyostotic forms and McCune–Albright syndrome manifest in early childhood.

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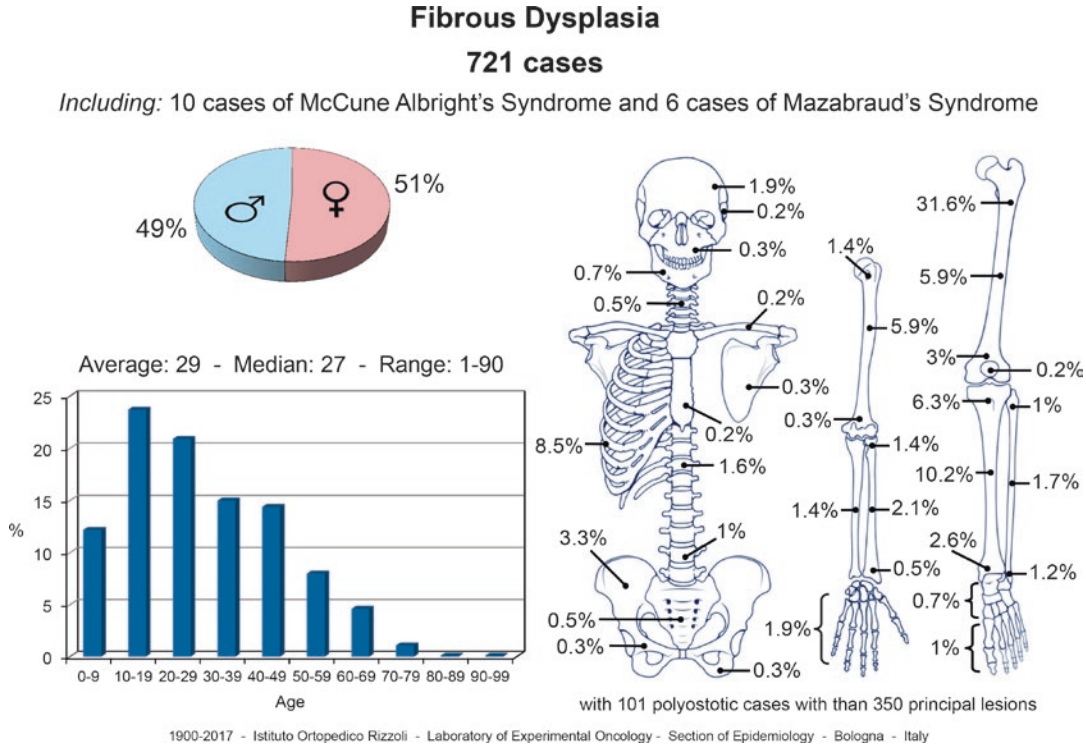


Location: Femur (proximal), tibia, craniofacial bones, and ribs; then humerus, forearm, and pelvis. Same sites in multicentric and polyostotic disease. Frequently more areas in the same long bone or two to three adjacent bones are affected. Lower limb more frequent than upper limb. Hand and foot are involved almost only in extensive polyostotic forms. Spine, scapula, and clavicle are rarely affected. Poliostotic type is usually prevalent in one body side.

Clinical: Monostotic is usually asymptomatic representing an incidental finding. Polyostotic: discontinuous pain (fatigue fractures), bony expansion in superficial bone, pathologic fracture, deformity, and lower limb length discrepancy. In polyostotic forms also cafè au lait spots ("coast of Maine"), multiple endocrine abnormalities (McCune–Albright syndrome), and intramuscular mixomas (Mazabraud syndrome).

Imaging: Standard X-rays show defined defect involving cortical and cancellous areas. Margins are well defined, sometimes marked by a rind of bone sclerosis. The cortex is sometimes thinned and expanded but continuous. No periosteal reaction. Radiolucency of "ground glass" appearance depends upon the amount of intratumoral trabeculae of woven bone. Severe "shepherd's crook" deformity of the proximal femur, usual in polyostotic form. Isotope scan: rather hot (diffuse dysplastic bone formation) and corresponding to radiographic extent. CT: homogeneity of ground glass radiolucency; cystic cavities and cartilaginous areas (sometimes calcified) when present. MRI: fairly homogenous low signal in T1.

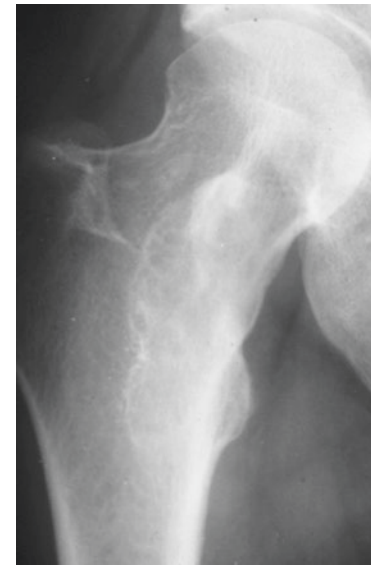
Histopathology: Periosteum is not involved; underlying cortex is regularly smooth but thin. Lesional tissue, well defined from surrounding bone, whitish to pink, from fibrous to gritty, to

hard bony. Sometimes, hemorrhagic areas or cystic spaces with serohematic content are present. Rarely, sparse lobules of hyaline cartilage are embedded in the above-described tissue. Histologically, it is characterize by a mixture of plump not atypical spindle cells and islands of woven bony trabeculae irregularly shaped, in the so-called "chinese letters" or "alphabet letters" fashion. Usually, bony trabeculae show no clear-cut osteoblastic rimming. Benign giant cells and foam cells are commonly found. Mitotic figures are uncommon and never atypical. Islands of cartilage may dominate the histologic appearance (so-called "fibrocartilaginous dysplasia"). It may show secondary aneurysmal bone cyst like areas.

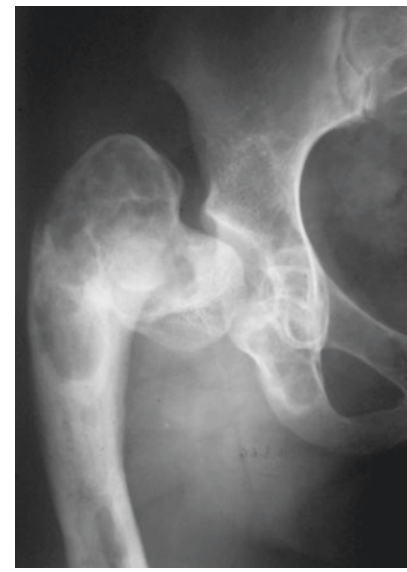
Course and Staging: Lesions are usually stage 2 in children and adolescents and stage 1 in adults. If a lesion expands and becomes symptomatic in an adult, this may be due to hemorrhage (during pregnancy). Rarely (less than 0.5% of the reported cases) a sarcoma may develop on a fibrous dysplasia. It usually occurs in adult of advanced age, both in monostotic and in polyostotic fibrous dysplasias, more frequent after radiation therapy.

Treatment: Frequently not needed. Curettage and grafting of active lesions should be avoided. Deformities may need corrective osteotomies and internal fixation, preferably by intramedullary devices.

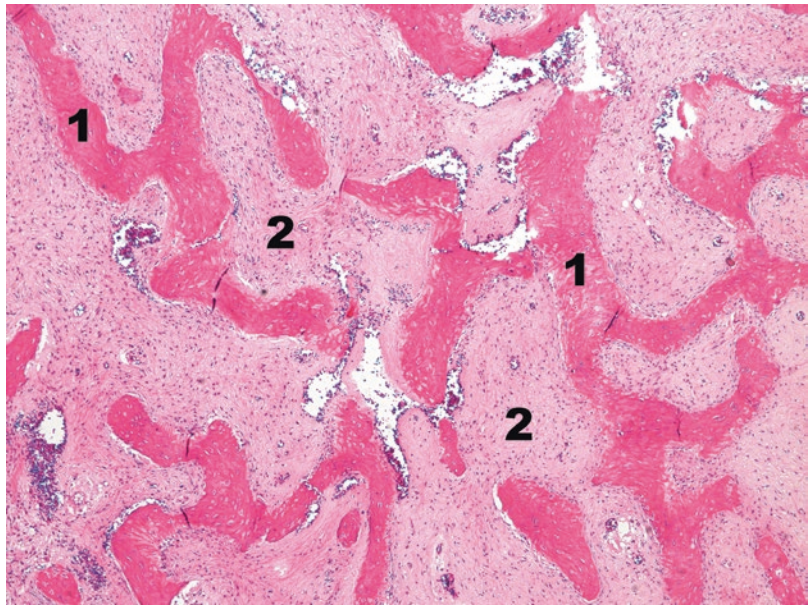
Key points		
• Clinical	Incidental findings	
• Radiological	"Ground glass" appearance	
• Histological	Benign fibroblastic lesion producing immature woven bone. No osteoblasts	
• Differential diagnosis	Low-grade central osteosarcoma	
Chromosomal translocations		
• Point mutations GNAS1	20q13.32	93%



Radiograph of the proximal femur. The lesion is metadiaphyseal, eccentric, sclerotic, and heterogeneous. It is well limited by a sclerotic ring



Radiograph of the hip. Lytic and sclerotic well-limited lesions. Typical femoral neck deformity (Shepherd's crook)



Histopathologic features are represented by immature bone trabeculae enmeshed in immature histio-fibroblastic tissue. (1) The dysplastic bone trabeculae are generally small and shaped like Chinese ideograms; they are usually not bordered by rows of osteoblasts. Trabeculae have a woven structure. (2) Undifferentiated fibrous connective tissue surrounding the trabeculae. The histio-fibroblasts are numerous and plump, with rare mitotic figures

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