Osteochondromas

16

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16.1 Solitary Osteochondroma

Definition: Benign cartilaginous neoplasm arising from the surface of the bone, histologically mimicking abnormal epiphyseal plate which grows and matures according to normal enchondral ossification.

It originates from a misplaced, subperiosteal island of physeal cartilage. It may also be seen in children secondary to radiation therapy. Presumably irradiation favors the exclusion of a cartilaginous island, by partially arresting and disorganizing proliferation of the physeal plate. Chomosomal aberrations involving 8q22-24.1 (where the *EXT1* gene is located) or 11p11.2 (where *EXT2* gene is located), causing biallelic inactivation of the *EXT1* and *EXT2* genes in the cartilaginous cap support the neoplastic nature of these lesions.

Epidemiology: Osteochondroma is very frequent. It prefers male sex by 1.5–2 : 1. Originating in early infancy, it is usually first noticed between 6 and 20 years of age.

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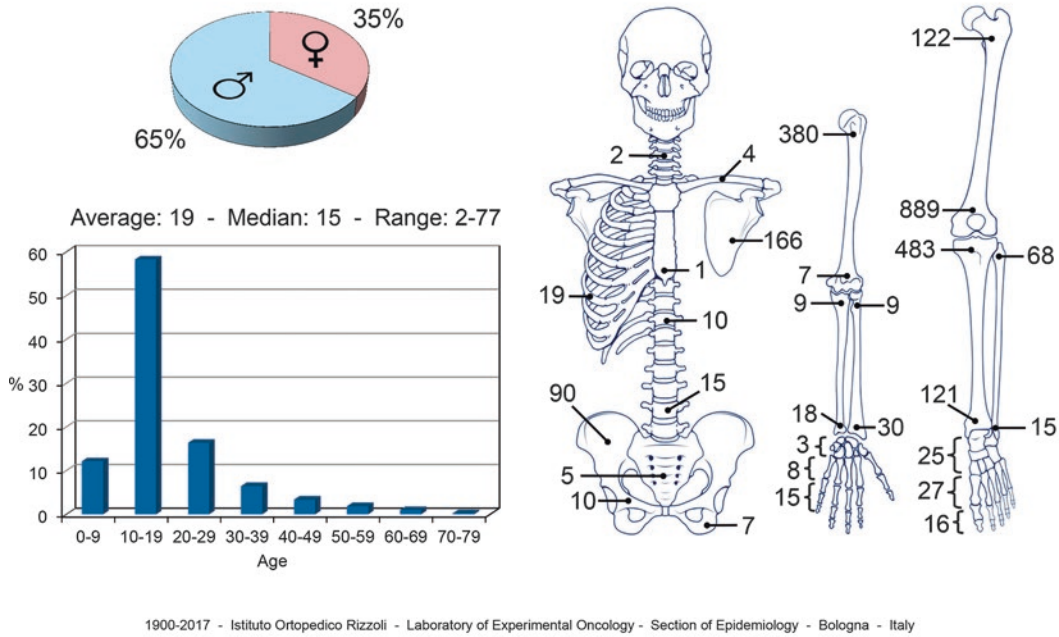


Localization: The most frequent localization is in the long bones: distal femur, proximal humerus, and proximal tibia. It originates from the metaphysis, but, with skeletal growth, it tends to move toward the diaphysis (growing away from the physis). The most frequently involved sites in the trunk are the scapula and ilium. Osteochondroma is exceptional in the hands and feet, and it does not occur in bones originating from membranous ossification (skull), nor in the epiphyses, carpal, and tarsal bones (except the calcaneus).

Clinical: A hard swelling is the main symptom, slowly increasing during skeletal growth. Osteochondroma is usually painless. Sometimes, pain is caused by an overlying bursitis or tendinitis. Occasionally, the overlying bursa, giving rise to increased volume and pain, may arise the suspicion of a malignant change. Exceptionally, osteochondroma compresses a peripheral nerve, causing neurological symptoms; or it rubs against a large artery thus producing a false aneurysm (femoropopliteal artery). Also exceptionally, as a result of traumatic fracture of its stalk, osteochondroma becomes painful and mobile, clinically simulating a muscular ossification or a loose articular body. An osteochondroma close to the physis may interfere with skeletal growth and cause axial deviations or a limb length discrepancy.

Imaging: Osteochondroma is a bony protuberance with well-defined limits, having a thin outer cortex and an internal cancellous structure. The pathognomonic radiographic feature is that the cortex of the host bone flares into the cortex of the osteochondroma, and the cancellous bone of the osteochondroma is continuous with the cancellous bone of the metaphysis. In large lesions, areas or rarefied bone may alternate with irregular blotches of intense radio density, due to

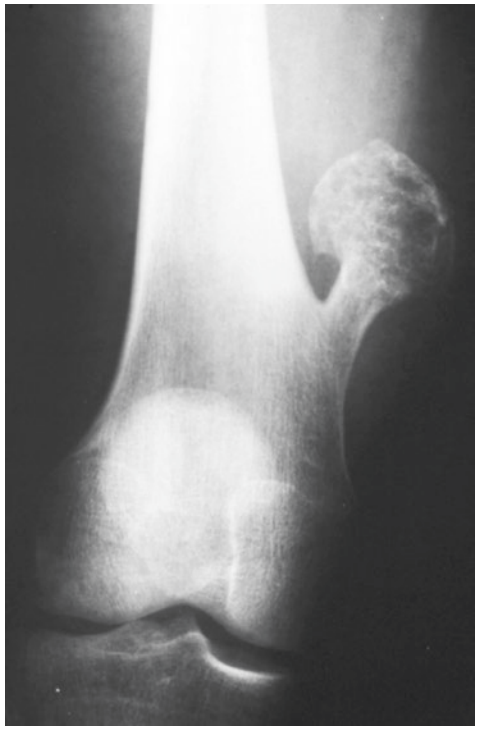
remnants of calcified cartilage, focal thickening of bone trabeculae, and bone necrosis. Some are pedunculated with a globose, cauliflower-like cap, or with a sharp horn-like extremity. Others have a broad sessile base. Pedunculated osteochondromas are usually inclined toward the diaphysis. Rarely osteochondroma becomes very large (even 15–20 cm), which is not a proof of malignancy. By chronic compression osteochondroma can cause scalloping and bowing of an adjacent bone. CT and MRI are useful (a) to confirm the diagnosis; (b) to perform preoperative planning (considering the relationship with the nearby neurovascular structures); (c) to measure the thickness of the cartilage cap or identify a reactive bursitis in case of suspicion of malignant transformation. Isotope scan is hot in active osteochondromas during childhood and adolescence and remains weakly positive or becomes negative after skeletal maturity, becoming again positive in malignant changes or when adjacent to a bursitis.

Histopathology: There is continuity between the cortex of the host bone and the cortical bone of the osteochondroma (so-called flaring of the cortex); similarly there is continuity between the medullary bone of the host bone and the medullary bone of the stalk of the osteochondroma. In children, osteochondroma is covered by a cartilage cap with a thickness ranging from a few millimeter to approximately 1.5–2 cm, and it appears as a light blue cartilage similar to that of the physeal plate. In the adult, this cap decreases in thickness, and in some areas, it disappears; residual cartilage is white and similar to articular cartilage. Limits of the cartilage with the underlying bone are well-defined. The inner part of osteochondroma is irregularly cancellous, with fatty or occasionally hemopoietic marrow. When osteochondroma is covered by a bursa, this may contain serous or hematic effusion, rarely osteo-cartilagenous loose bodies. In its active stage, the cartilage cap presents, although irregular, the same features of the normal growth plate. Some cellularities, plumpness of the nuclei, hypertrophy of the cells are to be expected in children and adolescents. The bony trabeculae of osteochondroma are originated by enchondral ossification of cartilage. Cancellous bone may include remnants of calcified cartilage and/or areas of necrotic bone.

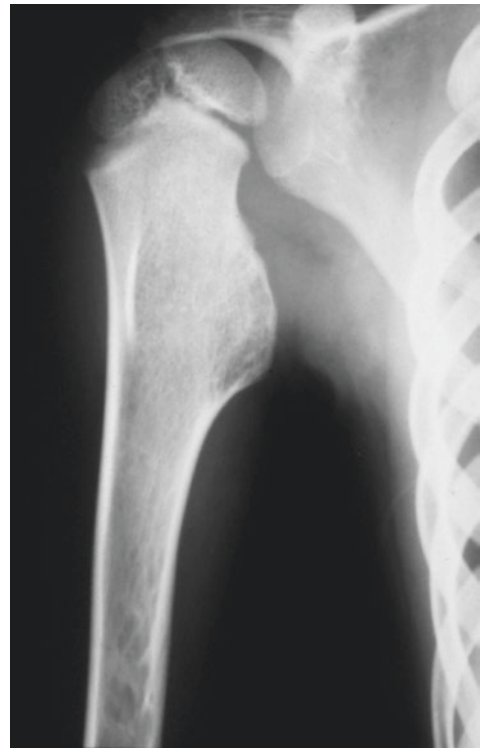
Course and Staging: Growth occurs during childhood and adolescence. After skeletal maturity, osteochondroma stops growing. Thus, it is a benign stage 2 lesion in children and adolescents, becoming stage 1 in the adult. The change of a solitary osteochondroma into a peripheral chondrosarcoma is rare (<1%) and does not occur before puberty. Risk of transformation depends on the site. It is exceptional in the more distal extremities, rare around the knee, and less rare in the trunk and limb girdles.

Treatment and Prognosis: There is no absolute indication for removal of an osteochondroma, and given the low risk of malignant transformation, prophylactic excision is not encouraged. Relative indications for surgery are pain, chronic bursitis/tendinitis, neurovascular symptoms, functional limitations, or growth disturbances due to the osteochondroma. In some cases, patients request excision of a large osteochondroma for cosmesis.

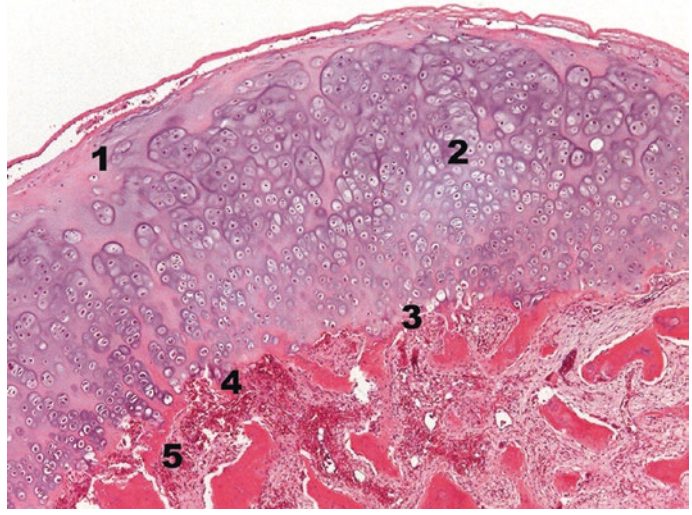
Key points	
• Clinical	Increasing swelling during growth age
• Radiological	Bony protuberance with continuity of the cortex and cancellous bone
• Histological	Cartilage cap (maximum 2 cm) covering normal cancellous bone
• Differential diagnosis	Low-grade peripheral chondrosarcoma



Pedunculated osteochondroma. Radiographs of the distal femur. The cortex of the osteochondroma is in continuity with that of normal bone



AP radiograph. Sessile osteochondroma of the proximal humerus



It commonly arises in bones formed by enchondral ossification, at the region of the edge of the epiphyseal plate. During the growth phase, the cartilage cap presents the same aspects of normal growth cartilage, although less regular. Progressive transformation of proliferating cartilage in underlying bone also mimics the epiphyseal growth mechanism, but it is much less orderly. (1) Chondrocytes arranged in clusters at the top of the cartilaginous cap. (2) Chondrocytes arranged in columns at the bottom of the cartilaginous cap. (3) Enchondral ossification at the interface between the cartilaginous cap and the underlying medullary bone. (4) Vascular mesenchymal cells invading calcified cartilage. (5) Seams of osteoid formed by the osteoblasts are laid on the framework of calcified cartilage

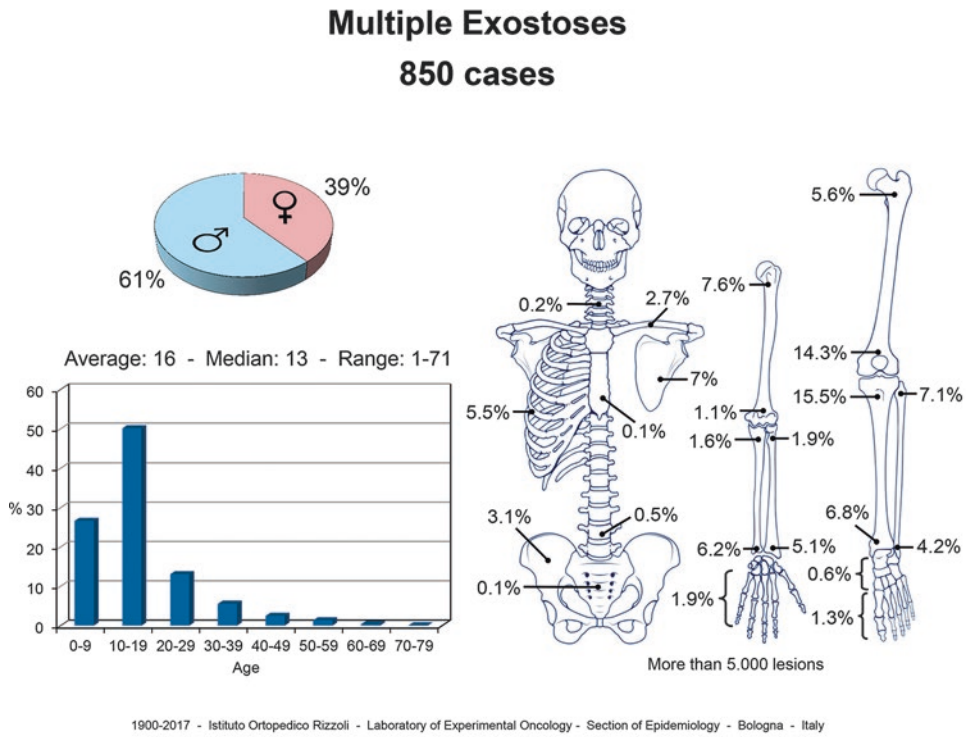
16.2 Multiple Osteochondromas

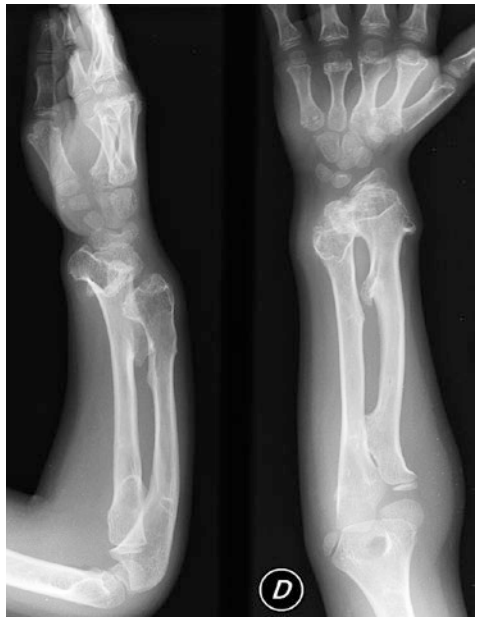
Multiple exostoses or osteochondromas are infrequent (prevalence 1/50,000). Male sex is preferred, by 1.5:1. The osteochondromas usually manifest before the age of 5–6 years, earlier as compared to solitary osteochondroma. Heredity is present in 80% of cases. Transmission is autosomal dominant. Recent studies suggest a role for other *EXT* genes. Basic research has identified several genetic abnormalities determining the disease. Most common mutations involve gene *EXT1* on chromosome 8 and gene *EXT2* on chromosome 11; a third gene named *EXT3* has been identified on chromosome 19. Cytogenetic aberration (biallelic inactivation, deletions ± mutations) of *EXT1/EXT2* genes are present in up to 90% of cases.

Multiple skeletal lesions are usually diffused and relatively symmetrical. Typically, exostoses involve the bone circumferentially, mostly surrounding the metaphyseal regions, causing swelling and sometimes limiting joint motion. Pain is a frequent symptom due to inflammation of adjacent tissues (bursitis, tendinitis). Limb shortening and deformity are frequently seen. Clinically, there is a large spectrum of presentation, from no deformity to severe impairment of upper and lower extremities, also within the same family. Relationship between type of genetic abnormality, severity of the disease, and risk of malignant transformation is under investigation in several centers. Overall, *EXT1*-positive patients seem to have a more severe clinical presentation.

Treatment principles are prevention and correction of deformity and shortening, by removal variably combined with stapling, osteotomies, and lengthening procedures. Most patients do surprisingly well and have satisfactory function without surgery.

The incidence of sarcomatous change in adult patients is low, ranging about 0.5–5%. Preferred sites for sarcoma are trunk, limb girdles, and knee. As for solitary osteochondroma, prognosis is dependent on the risk of malignant transformation. Patients should be followed clinically at regular intervals, monitoring the deeper exostoses (pelvis, spine) with serial radiograms every 2–3 years.





Radiographs of the arm and hand. Multiple exostoses. Bone deformation and shortening

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