Chapter 16
Multiple Exostoses

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Multiple exostoses are infrequent (incidence 1–2/100,000). Male sex is preferred, by 2 to 1. Exostoses usually manifest before 10 years of age, earlier as compared to solitary osteochondroma. Heredity is present in 2/3 of cases. Transmission is autosomal dominant. 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. Recent studies suggest a role for other EXT genes. 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. In severe forms, limb shortening and deformity are associated. There is a large spectrum of presentation, from no deformity to severe impairment of upper and lower extremities. The relationship between type of genetic abnormality, severity of the disease, and risk of malignant transformation is under investigation in several centers. Lesions present the same pathologic features as solitary osteochondroma.

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.

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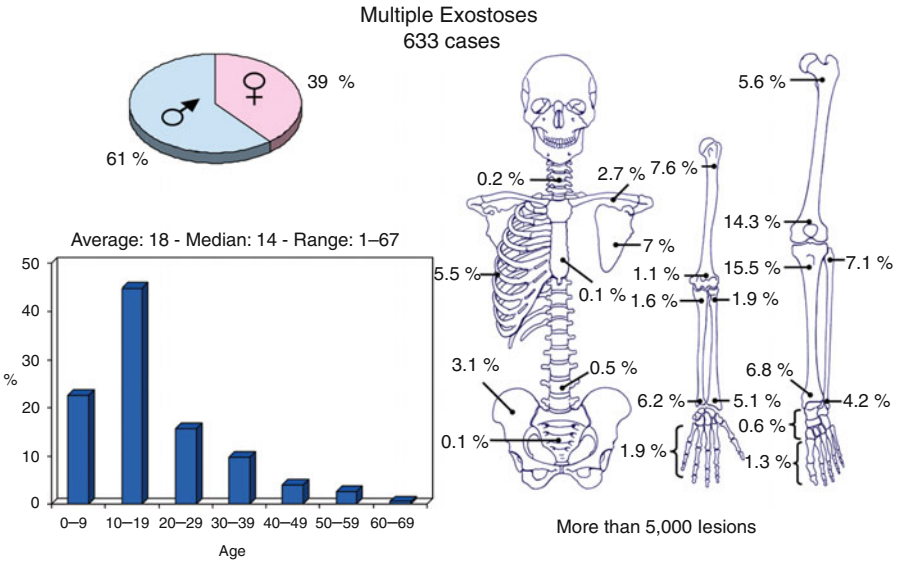
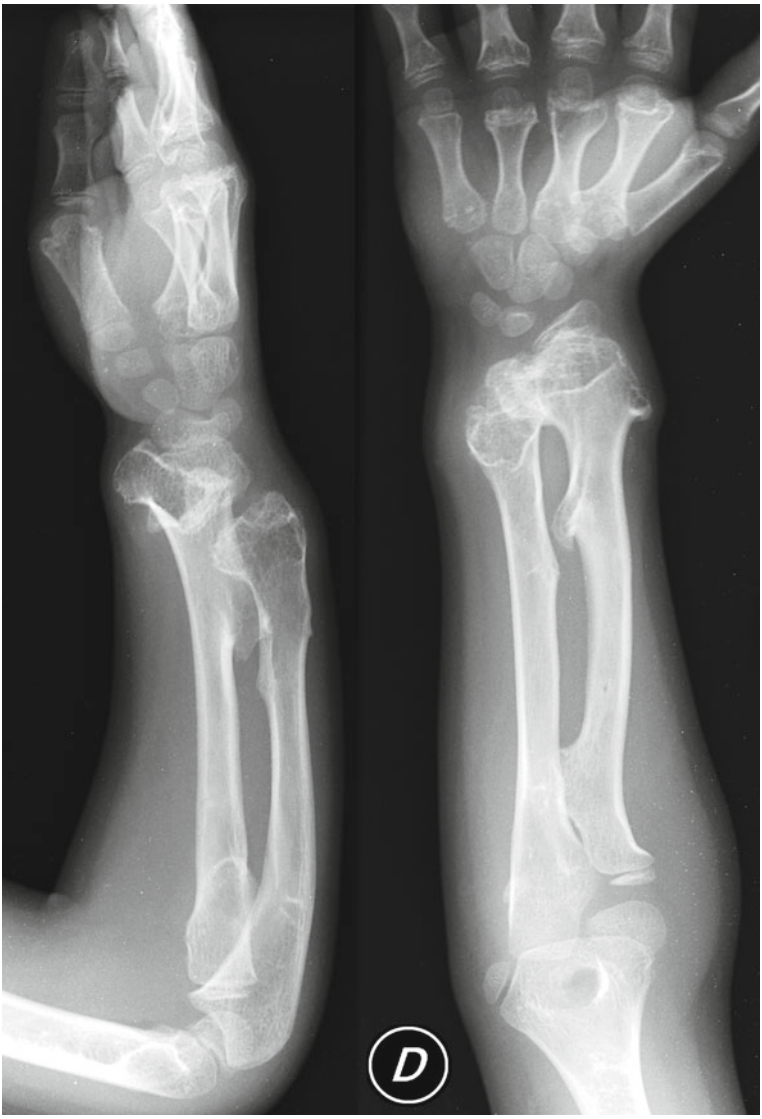
The incidence of sarcomatous change in adult patients is low, ranging about 0.5–2 %. Preferred sites for sarcoma are the trunk, limb girdles, and knees. As for solitary osteochondroma, prognosis is dependent on the risk of malignant transformation. Patients should be followed lifetime, monitoring the 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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