Musculoskeletal case 14. Diagnosis

Multiple enchondromatosis

The lesions present throughout the patient's hands (Fig. 1) are multiple enchondromas. In this distribution, the disease is known as Ollier's disease, a condition that was first described by the French surgeon Ollier (1830–1899). This rare disorder of skeletal development is characterized by multiple enchondromas that tend to be primarily located within the metaphyseal regions of tubular bones.[1]

An enchondroma is a benign, intramedullary cartilage tumour that is usually found in the short tubular bones of the hands and feet. Most enchondromas are solitary. They are the most common primary tumour in the hand. On gross examination, an enchondroma consists of bluish-grey lobules of fine translucent tissue. The degree of calcification determines if the consistency is gritty.

Ollier's disease is a nonhereditary disorder of enchondral ossification. The characteristic appearance is one of multiple, central, well-circumscribed enchondromas,[2] which are typically radiolucent and stippled with calcification as demonstrated in Fig. 2. Linear or columnar lucencies in the metaphyses, representing columns of growing cartilage may also be seen. Frequently enchondromas cause thinning and bulging of the cortex. The diagnosis is usually made in childhood. Skeletal involvement tends to be greater on one side of the body than the other. Males are affected more than females. The involved bones are also more prone to fractures, and this is a common presentation. Painful lesions in the absence of a fracture should suggest the possibility of malignant change. Biopsy should be considered in such cases.

Although single enchondromas undergo malignant transformation in less than 1% of cases, Ollier's disease is associated with a 30% risk of malignant transformation.[3] The commonest malignant lesion encountered is chondrosarcoma. This likelihood of malignant transformation is even higher in patients having Maffucci's syndrome (multiple enchondromas in association with soft-tissue hemangiomas). Microscopically, a thin layer of lamellar bone surrounding the cartilage of nodules is a positive sign that the lesion is benign. At low power, lobules of different sizes can be seen. Blood vessels are surrounded by osteoid. Enchondromas have chondrocytes without atypia inside hyaline cartilage. The nuclei are small, round and pyknotic. The cellularity varies among lesions and within the same lesion. The pathological diagnosis can be difficult, particularly the distinction between chondrosarcoma and benign enchondroma, and it must be made in conjunction with the radiologist and the surgeon.

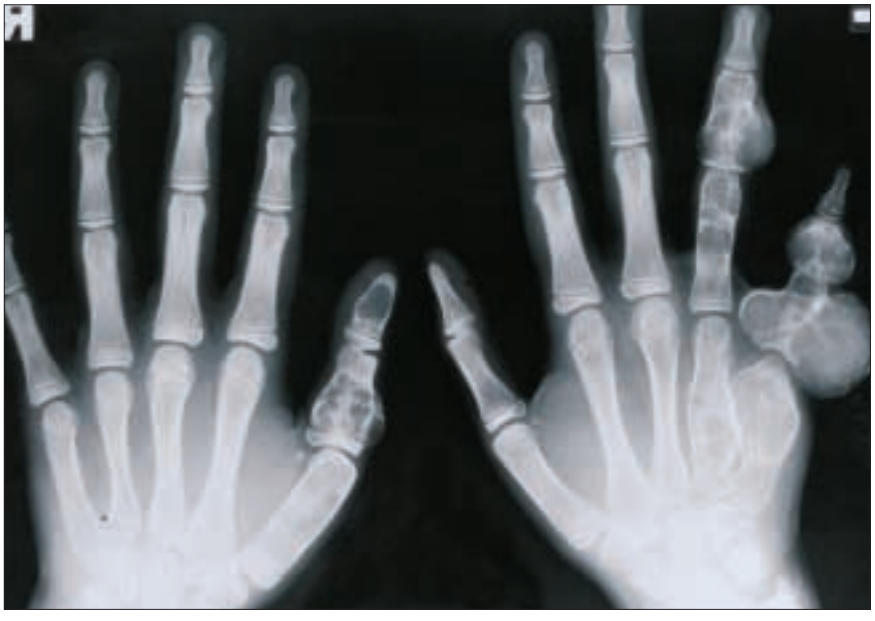

FIG. 1.

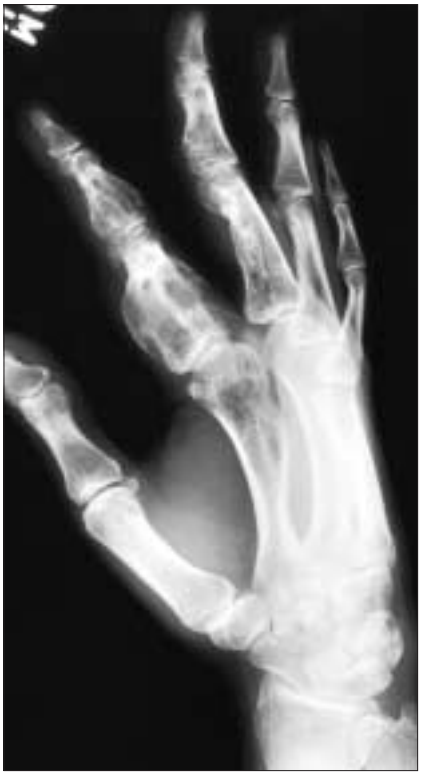

FIG. 2.

Radiologic findings in Ollier's disease

Enchondromas are usually long and oval, having well-defined margins.[4] In larger lesions, the lucent defect has endosteal scalloping and the cortex is expanded and thinned. In some cases, the expansion may lead to pathologic fracture. Calcifications throughout the lesion can range in appearance from punctate to rings. Stippled cartilage is more common with increasing age of individual enchondromas. Cartilaginous areas, extending from the physis can interfere with growth, resulting in limb shortening and deformities. typically, the lesions involve the hands, but other bones may also be involved: Figs. 3 and 4 show marked involvement of the elbow joint in a patient with Ollier's disease. Fig. 5 demonstrates a gross example of Ollier's disease involving the pelvis. In patients with Ollier's disease, radiographs may be normal in the first few years of life and later demonstrate streaking within the metaphysis of involved bones, or in infancy they may demonstrate radiolucent masses, which can be round, triangular or linear. Enchondromas typically stabilize or regress in adulthood. Computed tomography is useful for detecting matrix mineralization and cortex integrity. Magnetic resonance imaging is helpful for describing the nonmineralized portion of the lesion and visualizing any aggressive or destructive features.[5]

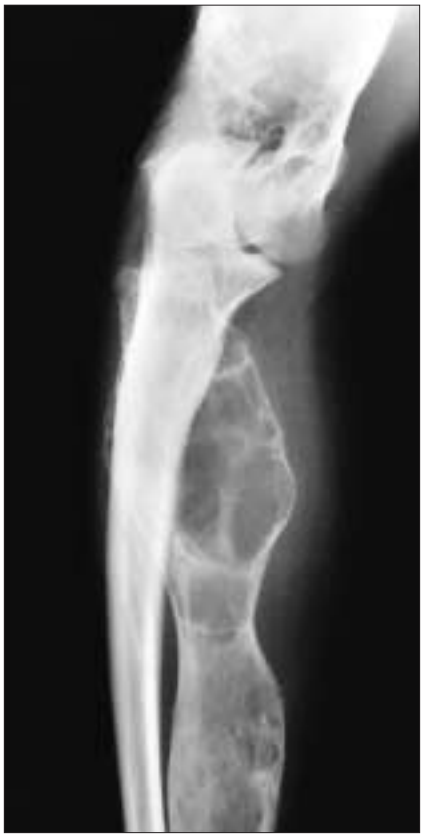

FIG. 3.

The differential diagnoses of Ollier's disease include polyostotic fibrous dysplasia and diaphyseal aclasia.

References

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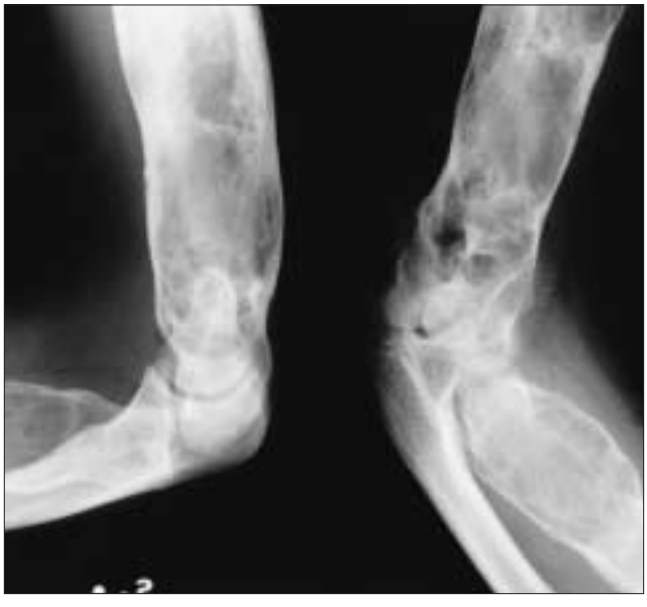

FIG. 4.

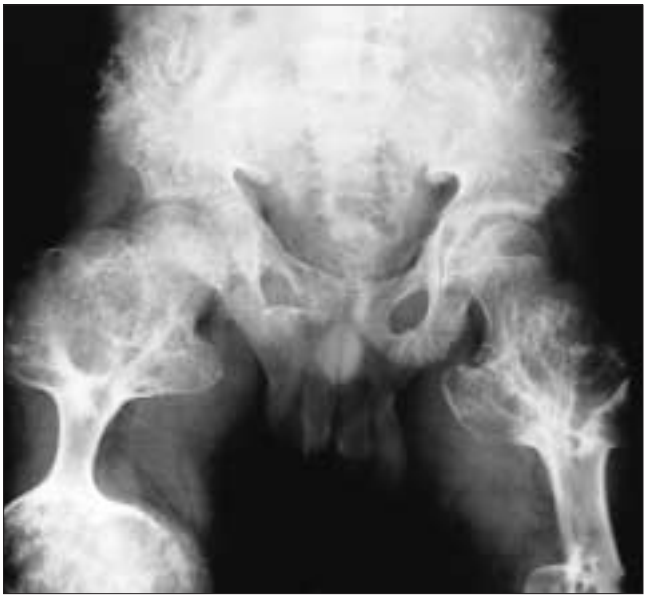

FIG. 5.