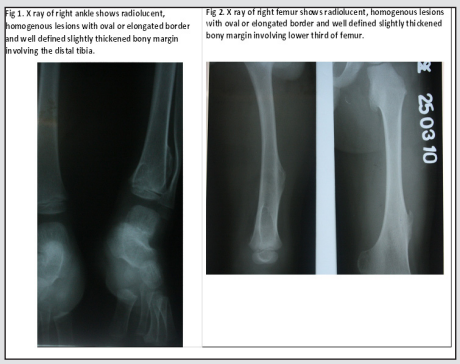
SPOT DIAGNOSIS (IMAGE GALLERY)



BONY SWELLINGS

Rahul Sinha, Vandana Negi, S S Dalal

Department of Pediatrics,167 Military Hospital, Pathankot, Army Hospital, New Delhi, Command Hospital Air Force, Bangalore, India.

Address for Correspondence: Dr Rahul Sinha, 167 Military Hospital, Pathankot, Punjab India. Email: drrahul_2000@yahoo.com

A 5 year old boy presented history of progressively increasing painless swelling in the right ankle for 2 years. The parents also noted similar swellings in lower and upper aspect of right thigh and right elbow about 3 months later. There was no history of recurrent fractures or any hemangiomas. Birth history and milestones were normal. No other family member was affected. X-ray of right ankle, right lower end of femur and pelvis revealed radiolucent, homogenous lesions with oval or elongated border and well defined slightly thickened bony margin.

What is the diagnosis?

Ollier's disease. It is a rare nonhereditary sporadic disorder where intraosseous benign cartilaginous tumors develop close to growth plate cartilage. Prevalence is estimated at around 1 in 100,000. (1) The disease consists of multiple enchondromas which usually develop in childhood. On radiographs, streaks of low density are seen projecting through the diaphyses into the epiphyses of the long bones, due to ectopic cartilage deposits. With age, the cartilage may calcify in the typical snowflake pattern. The affected extremity is shortened (asymmetric dwarfism) and sometimes bowed due to epiphyseal fusion anomalies. Differential diagnoses include Maffucci syndrome which is characterized by multiple enchondromatosis as well as multiple soft tissue cavernous hemangiomas, and less commonly lymphangiomas which was not seen in our patient. (2) Both Maffucci syndrome and Ollier;s disease are associated with an increased incidence of juvenile granulosa cell tumor of the ovary. (3) Patients with Maffucci syndrome also have an increased incidence of malignancies other than musculoskeletal malignancies, including gliomas, gastrointestinal adenocarcinoma, pancreatic carcinomas and ovarian tumor. (3,4) Another differential diagnosis to be considered is metachondromatosis which is a hereditary autosomal dominant transmitted condition consisting of multiple enchondromas and osteochondromas. In this syndrome, the multiple exostoses characteristically occur in the digits and long bones, point towards the joints and frequently regress spontaneously. (5) The most important criterion to distinguish enchondromas from osteochondromas is the localization of bone lesions: osteochondromas are located at the bone surface and enchondromas are located in the center of bones, thus allowing radiographic distinction.

Persons with Ollier disease are prone to breaking bones and normally have swollen, aching limbs. There is no medical treatment for enchondromatosis. Surgical intervention is indicated in case of complications (pathological fractures, growth defect, malignant transformation).

References

1. Khoo RN, Peh WC, Guglielmi G. Clinics in diagnostic imaging (124). Multiple enchondromatosis in Ollier disease. Singapore Med J 2008; 49: 841-845
2. Zwenneke Flach H, Ginai AZ, Wolter Oosterhuis J. Best cases from the AFIP. Maffucci syndrome: radiologic and pathologic findings. Armed Forces Institutes of Pathology. Radiographics. 2001; 21: 1311-1316
3. Tamimi HK, Bolen JW. Enchondromatosis (Ollier;s disease) and ovarian juvenile granulosa cell tumor. Cancer. 1984; 53: 1605-1608.
4. Mahafza WS. Multiple enchondromatosis Ollier;s disease with two primary brain tumors. Saudi Med J. 2004; 25: 1261-1263
5. Schwarz W, Hardes J, Schulte M. Multiple enchondromatosis: Ollier;s disease. Unfallchirurg. 2002; 105: 1139-1142

E-published: June 2012 . **Art**#38, **DOI No. :** 10.7199/ped.oncall.2012.38

Quick Response Code