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Radiology

OLLIER'S DISEASE- A NON-HEREDITARY, SPORADIC SKELETAL DISEASE

Dr Pratiksha Kapadia	3rd Year Resident, Department of Radiology, B J Medical college, Civil hospital, Ahmedabad, Gujarat, India
Dr S K Turakhia	Associate Professor, Department of Radiology, B J Medical college, Civil hospital, Ahmedabad, Gujarat, India
Dr Sahil N. Shah*	Senior Resident Doctor, Department of Radiology, Smt. N.H.L. Medical College, Ahmedabad *Corresponding Author
Dr Akshay Patel	2nd Year Resident, Department of Radiology, B J Medical college, Civil hospital, Ahmedabad, Gujarat, India

ABSTRACT Ollier's disease, also known as Enchondromatosis, is a non-hereditary, sporadic, skeletal disorder characterized by multiple enchondromas that are principally located in the metaphyseal regions. Diagnostic approach is based on clinical and radiological investigations. Biopsies of the enchondromas are also used to look for features that are characteristic of Ollier's disease. Here we present a case of Ollier's disease. In this case report, we would like to highlight the radiological features of this disorder which help in the diagnosis of the disease.

KEYWORDS : Enchondromatosis, Rings and arcs calcification, Radiology, Chondroid matrix.

INTRODUCTION

Ollier's disease is a rare nonhereditary sporadic disorder where intraosseous benign cartilaginous tumors (enchondroma) develop close to growth plate cartilage. Prevalence is estimated at around 1 in 100,000. Nominally, the disease consists of multiple enchondromas which usually develop in childhood. The growth of these enchondromas usually stops after skeletal maturation. The affected extremity is shortened (asymmetric dwarfism) and sometimes bowed due to epiphyseal fusion anomalies. Persons with Ollier's disease are prone to breaking bones and normally have swollen, aching limbs. Enchondromas have a predilection for unilateral distribution and frequently affect the long tubular bones, particularly the tibia, the femur, and/or the fibula; flat bones, especially the pelvis, can also be affected. A case of Ollier's disease located in the anterior ribs has been reported. The enchondromas can grow large and can be disfiguring.

CASE REPORT

Here we present a case of 18 year old female patient who presented with the complain of left knee pain and swelling since 6 years associated with difficulty in walking and reduced movements of left knee joint. On clinical examination, genu valgum was present on left side [Figure 1]. In this case report, we would like to highlight the radiological features of this disorder which help in the diagnosis of the disease.

Radiographs of bilateral lower limbs revealed:

- Bowing of shaft of left femur.
- Multiple expansile lytic lesions causing thinning of cortex and endosteal scalloping with a distinct zone of transition.
- Rings and Arcs calcification in the matrix of the lesions.
- No evidence of any periosteal reaction.
- No evidence of any soft tissue mass. [X-ray features in figure 2 and figure 3]

MRI of left femur was performed which revealed the following features:

- Diffuse altered signal intensity lesions in medullary cavity of left ischium, pubic bone, left femur, left patella and visualized left tibia.
- The lesions were diffusely hypointense to muscle on T1WI [Figure 4], iso to slightly hyperintense on T2//STIR images with multiple patches of rings like increased signal hyperintensities [Figure 5 and 6].
- There was evidence of cortical thinning and medullary expansion with lengthening and bowing of the femur.
- On post contrast study the lesion shows heterogenous contrast enhancement.

CT Scan of left femur revealed:

- Multiple expansile lytic lesions causing thinning of cortex and endosteal scalloping with a distinct zone of transition [Figure 7].
- Rings and Arcs calcification in the matrix of the lesions.

DISCUSSION

Ollier disease is a rare skeletal disorder characterized by abnormal bone development (skeletal dysplasia). While this disorder may be present at birth (congenital); it may not become apparent until early childhood when symptoms, such as deformities or improper limb growth, are more obvious. Ollier disease primarily affects the long bones and cartilage of the joints of the arms and legs, specifically the area where the shaft and head of a long bone meet (metaphyses). The pelvis is often involved; and even more rarely, the ribs, breast bone (sternum), and/or skull may also be affected.

Ollier disease manifests as greater than normal growth of the cartilage in the long bones of the legs and arms so that growth is abnormal and the outer layer (cortical bone) of the bone becomes thin and more fragile. These masses of cartilage are benign (non-cancerous) tumors known as enchondromas. Enchondromas may occur at anytime. After puberty these growths stabilize as cartilage is replaced by bone. In rare cases, the enchondromas may undergo malignant changes (e.g., chondrosarcomas). The exact cause of Ollier disease is not known, although in some cases it may be inherited as an autosomal dominant genetic trait.

When the enchondromas of Ollier Disease are accompanied by substantial, most often benign, proliferation of blood vessels (hemangiomas), the array of symptoms is known as Maffucci Syndrome.

MRI

Enchondromas tend to have lobulated borders with a cluster of numerous tiny locules of high-signal-intensity foci on T2-weighted images that appear to coalesce with one another and reflect the high fluid content of hyaline cartilage. On T1-weighted images, enchondromas demonstrate low-to-intermediate signal intensity.

When a calcific chondroid matrix is observed on radiographs, decreased signal intensity is expected to occur in those areas on images obtained with all MRI pulse sequences. These areas may become particularly pronounced on gradient-echo images.

When combined with a typical appearance on radiographs and an absence of pain, the diagnosis of enchondroma with MRI findings can be made with a high degree of confidence. Enchondromas may also be incidental findings at routine examination, such as MRI of the knee. If

the lesions have a typical appearance, a confident diagnosis may be made; however, the acquisition of correlative radiographic findings is often prudent.

Radiography

A classic pattern of calcifications, described as rings and arcs, is pathognomonic when it is seen in the hands. In the long bones, calcifications may be difficult to distinguish from the dystrophic calcifications seen in bone infarction. In addition, radiographs may not depict the rings and arcs of calcifications, particularly in the hands and feet. In Ollier's disease, enchondromas often appear to be larger than they do in other conditions. Because enchondromas occur in young patients and can be large, growth of the affected limbs may be adversely affected, and pathologic fractures may occur. Enchondromatosis can occasionally have the appearance of linear lucencies, in which the chondrocytes appear to line up in a vertical orientation along the length of the bone.

CT

The features of enchondroma on CT scan are the same as those depicted on radiographs (see the image below). Enchondromas are endosteal lesions with a lobular morphology and variable mineralization. Often, the mineralization is in the form of rings and arcs, which correspond to calcification around lobules of cartilage. A pathologic fracture may be present. Sometimes, endosteal scalloping is present, but this feature may be suggestive of degeneration of the enchondroma to a chondrosarcoma. Enchondromas should not penetrate the cortex or extend into the soft tissues. If a densely mineralized or uniformly mineralized lesion has a region that is lucent, degeneration of the enchondroma to a chondrosarcoma is suggested, and biopsy is likely necessary.

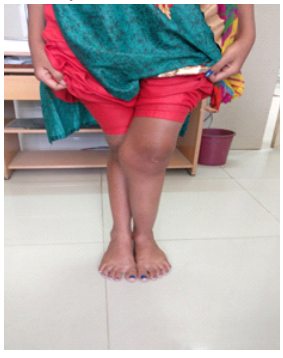
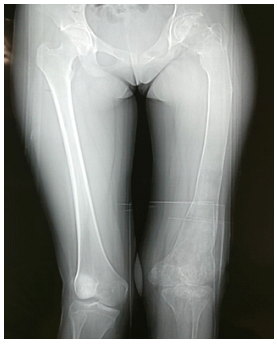


Figure 1: Genu valgum deformity of left lower limb.



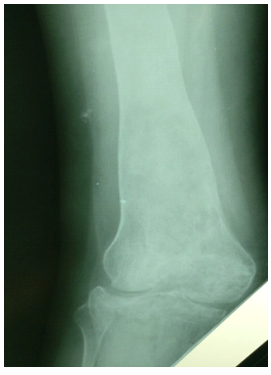


Figure 2 and 3: Xray features of ollier's disease.

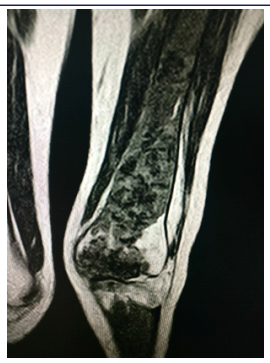


Figure 4: T1WI showing multiple hypointense lesions.

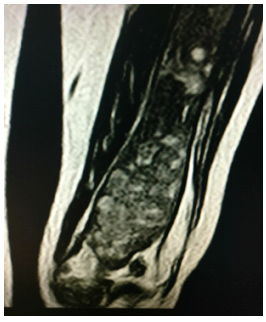


Figure 5: T2WI showing multiple hyperintense lesions.

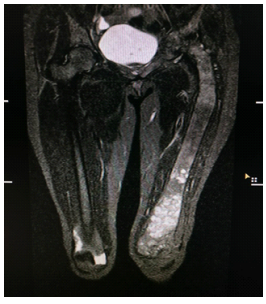


Figure 6: STIR image showing multiple hyperintense lesions with bowing of left femur.

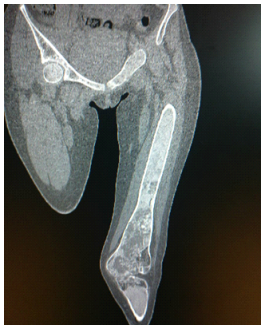


Figure 7: Multiple expansile lytic lesions with endosteal scalloping demonstrated in CT.

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