ejpmr, 2018,5(12), 264-266

SJIF Impact Factor 4.897

EUROPEAN JOURNAL OF PHARMACEUTICAL AND MEDICAL RESEARCH

www.ejpmr.com

Case Report

ISSN 2394-3211

EJPMR

METACHONDROMATOSIS – A RARE IMAGE FOR CLINICAL RADIOLOGISTS

Dr. Eesha Rajput*

Consultant Radiologist, INHS Kalyani, Vizag, AP 530005.

***Corresponding Author: Dr. Eesha Rajput**

Consultant Radiologist, INHS Kalyani, Vizag, AP 530005.

Article Received on 24/09/2018 **Article Revised on 14/10/2018** **Article Accepted on 4/11/2018**

INTRODUCTION

Metachondromatosis is a rare hereditary condition that displays a combination of multiple enchondromas,[1] with multiple osteochondroma-like lesions involving the iliac crests and extremities. We present one such case where diagnosis was difficult at initial presentation and was achieved only after skeletal survey.

KEYWORDS: Enchondroma Osteochondroma hereditary.

CASE REPORT

A 3 year old female child presented to our department with complaints of progressive deformity of second toe of right foot. The deformity was painless, progressive, not associated with trauma or swelling of the limb. No systemic symptoms like fever, rash were reported.

On clinical examination, there was lateral deviation of the right second toe and the second digit appeared longer than the expected length. There was no redness, tenderness, local rise in temperature, swelling or features s/o trauma. On close inspection of other extremities, similar but milder deformity was also noted in both hands of the child.

In view of above findings, skeletal survey was done. Xrays of both hands and feet, both upper and lower limbs, pelvis and spine were carried out.

These revealed multiple enchondromas in the small bones of both feet causing lateral deviation of right second toe. Enchondromas were also noted in the small bones of the hand. In addition on skeletal survey, multiple osteochondromas were noted affecting left radius, right ulna (distal metaphysis), distal metaphysis of both femora, proximal metaphysis of right tibia and left fibula. No changes were observed in the spine and pelvis.

In view of the above findings, it was inferred that this is a case of metachondromatosis. As the condition does not require any active intervention, the patient was counselled accordingly about the prognosis of the condition. Skeletal survey of the elder male sibling was also advised. However, it did not reveal any significant findings.

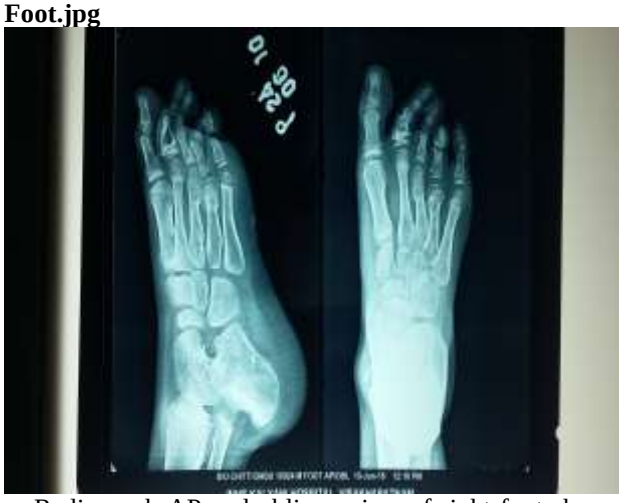
Clinical pic 1

• Clinical picture of feet shows lateral deviation of II and III toes of right foot with IV toe appearing shorter and curved - deformity.

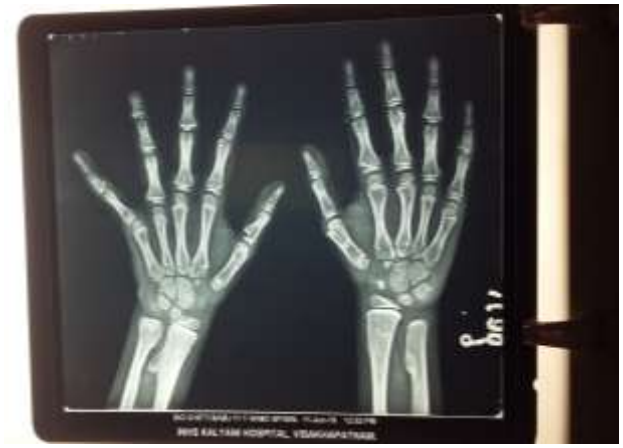
Clinical pic 2

• Clinical picture of left hand shows lateral deviation of II digit with prominence at the lateral aspect of wrist

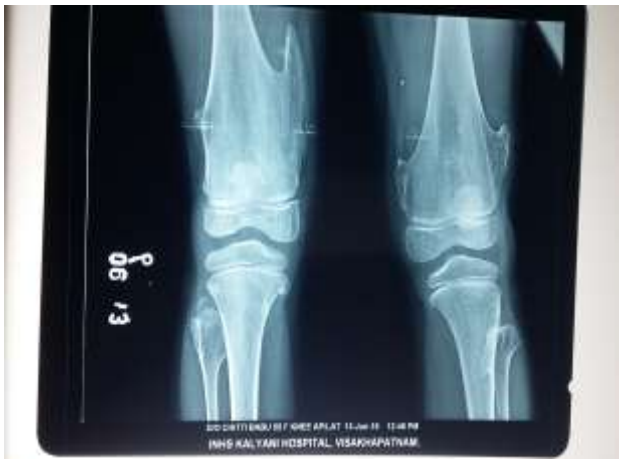
Foot.jpg



• Radiograph AP and oblique view of right foot shows enchondromas in proximal phalanx of right II digit and head of right IV metatarsal. There is fusion of middle and distal phalanx of IV digit noted with lateral deviation of II and III digits as seen on the clinical picture Both hands.jpg

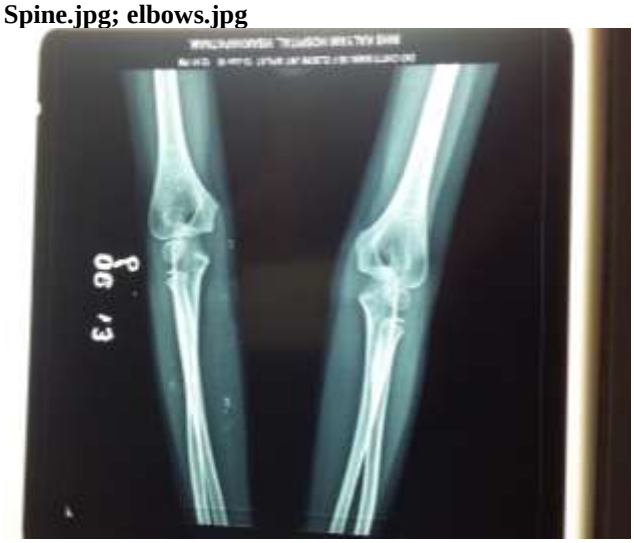


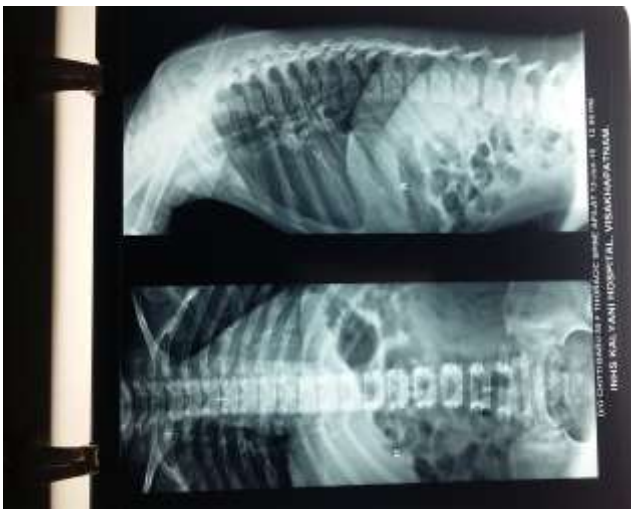
• PA view - Multiple enchondromas are noted. Osteochondromas are noted at distal end of left radius (growing away from the joint), right I metacarpal, proximal phalanx of right middle finger Both knees.jpg



AP view - osteochondromas are noted in both femora, left fibula. Enchondroma is noted at proximal end of right fibula

Spine.jpg; elbows.jpg





Screening images of spine AP and lateral view and both elbows AP view do not reveal any abnormality

DISCUSSION

Cartilage tumor syndromes are characterized by multiple cartilaginous bone tumors,[3] that develop in childhood, often causing significant morbidity and predisposing to chondrosarcoma.

Tumors can form as exostoses (on the surface of bone), as in the autosomal dominant multiple osteochondroma MO (hereditary multiple exostoses HME/ diaphyseal aclasis) syndromes or as endosteal tumors (within bone), as in the sporadically occurring multiple enchondromatosis disorders Ollier disease[2] and Maffucci syndrome.[4]

MO /HME – is an autosomal dominant condition characterized by development of multiple osteochondromas. An osteochondroma is a chondroid neoplasm of the growth plate. It can either be sessile or pedunculated. It is noted in the metaphyseal region typically projecting away from the epiphysis, associated with broadening of the metaphysis and has a variable

cartilage cap, sometimes showing arc and ring type of calcification.

Ollier's disease[2] / enchondromatosis - is a non hereditary, sporadic, skeletal disorder characterized by multiple enchondromas that are principally located in the metaphyseal regions.

Enchondroma is a benign cartilaginous lesion, commonly seen in phalanges. It is an expansile lytic lesion, with characteristic "rings and arcs " calcification, narrow zone of transition, scalloped margins; no periosteal reaction or soft tissue mass is seen.

Mafucci's syndrome is a congenital nonhereditary mesodermal dysplasia characterised by multiple enchondromas and soft tissue venous malformations (hemangiomas). On imaging, it is usually portrayed by a short limb with metaphyseal distortions due to multiple enchondromas, and soft tissue masses with phleboliths, depicting hemangiomas.

Metachondromatosis[5,6]

Genetic basis

In MO, mutations in EXT1 or EXT2, which encode heparan sulfate glycosyltransferases, affect chondrocyte orientation in the growth plate.

A small percentage of patients with Ollier syndrome have mutations in PTH1R, which encodes the receptor for parathyroid hormone and parathyroid hormone-related protein, causing altered chondrocyte differentiation in the growth plate.

The cause of Maffucci syndrome is unknown

The mode of inheritance of metachondromatosis is autosomal dominant but the underlying gene has not been identified so far, due to the extreme rarity of the disease. Fewer than 50 cases of MC have been published since Maroteaux's initial description in 1971.

Patients with MO do not develop endosteal tumors and patients with Ollier disease or Maffucci syndrome do not develop exostotic tumors. In contrast, patients with metachondromatosis form both exostotic and endosteal tumors. Exostotic lesions in MC occur frequently in the digits, involve metaphyses and epiphyses, and tend to grow toward the joint; in contrast, exostotic lesions in MO occur frequently in the long bones, involve only the metaphyses, and tend to grow away from the joint. MC exostotic lesions can also spontaneously decrease in size and completely regress. Endosteal lesions in MC are common in the metaphyses of long bones and in the pelvis.

Complications - Avascular necrosis of the femoral head, due to endosteal tumors has been a frequent complication in patients with MC.

Hand deformity due to endosteal tumors is uncommon in patients with MC. However it was found as the presenting complaint in our patient involving the hands and the feet. Whereas it is often a significant problem for patients with Ollier disease and Maffucci syndrome.

Finally, malignant transformation has only been reported in one patient with MC, whereas it has been more frequently reported in patients with MO, Ollier disease, and Maffucci syndrome.

Prognosis – Exostotic lesions in metachondromatosis may regress spontaneously and completely disappear. Malignant transformation is rare. The lesions fall in the category of touch me not lesions and only need masterly inactivity. However, sudden increase in size of the lesion, deformity or appearance of pain or inflammatory symptoms would indicate malignant transformation and require surgical intervention.

Since the mode of inheritance is autosomal dominant with variable penetrance, screening of the siblings is recommended in a family with affected child.

CONCLUSION

Metachondromatosis is a rare hereditary disorder - autosomal dominant with variable penetrance and childhood presentation. It involves development of endosteal and exostotic cartilaginous tumors in extremities and iliac bones. Spontaneous resolution is the norm and hence it falls in the category of touch me not lesions. Malignant transformation is rare and hence differentiation from other cartilaginous tumor syndromes like multiple osteochondromatosis, Olllier's disease and Mafucci's syndrome is essential as incidence of malignancy is much higher in these disorders. Experience of individual radiologist may be limited in this condition and therefore careful skeletal survey is essential before inferring the final diagnosis.

REFERENCES

1. Lucas DR, Bridge JA: Chondromas: enchondroma, periosteal chondroma and enchondromatosis. World Health Organisation classification of tumours. Pathology and genetics of tumours of soft tissue and bone. Edited by Fletcher CDM, Unni KK, Martens F. Lyon: IARC press, 2002; 237-40.
2. Silve C, Juppner H. Ollier Disease. Orphanet J Rare Dis., 2006; 1: 37.
3. Unni KK. Cartilaginous lesions of bone. J Orthop Sci., 2001; 6: 457-72.
4. Zwenneke FH, Ginai AZ, Walter OJ. Best cases from the AFIP. Mafucci syndrome: radiologic and pathologic findings. Armed Forces Institute of Pathology. Radiographic, 2001; 21: 1311-6.
5. Wittram C, Carty H. Metachondromatosis. Pediatr Radiol, 1995; 25 Suppl 1: S138-S139.
6. Bassett GS, Cowell HR. Metachondromatosis. Report of four cases.J Bone Joint Surg Am, 1985; 67: 811-4.