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Case Report

ABNORMALITY IN ENCHONDRAL OSSIFICATION ENCHONDROMATOSIS

(A Rare Severe Clinical case with Embryological and Radiological perspective)

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ABSTRACT

In humans sometimes abnormal intracartilaginous mode of ossification can lead to formation of a bony lesion referred to as Enchondroma. Enchondroma can present as a solitary lesion that involves metacarpals, metatarsals, and phalanges in most of the cases or as multiple lesions. We present a case of a 28 year old male who presented with pain in his forearm and digits on both sides. The pain was of mild nature .Rest of the general and skeletal survey was normal. He was followed with radiographs which revealed multiple enchondromata bilaterally. On history there was no familial history of similar or related disease.

KEYWORDS: Enchondroma; Ossification; Ollier; bone; Histological.

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BACKGROUND

They appear as deforming masses or become apparent when they induce pathologic fractures. Radiographs show circumscribed areas of rarefied bone with thinning and often bulging of the cortex and stippled calcification. Lesions in the hands or feet are generally benign; those in the large tubular bones have greater malignant potential and may be difficult to separate histologically from malignant lesions. Treatment consists of curettage for well-contained lesions and autologous bone grafting for extensive lesions in the metacarpals or phalanges. The presence of multiple enchondromas is known as Ollier disease.

DISCUSSION

These lesions commonly occur in the phalanges and metacarpals but can also be found in the femur, tibia, and iliac crest. They have got malignant potential.

The Cartilage in humans is seen to develop from the mesenchyme . In a developing embryo the cartilage is seen to proliferate and start developing during the fifth week of intraembryonic life. The area where future cartilage is to develop, the mesenchyme condenses to form chondrification centers. The mesenchymal cells differentiate into chondroblasts that secrete collagenous fibrils and the ground substance.

Endochondral ossification is a special type of ossification process where the hard bone formation occurs from a preexisting cartilaginous models. Especially In a long bone, the primary center of ossification appears in the diaphysis where the chondrocytes increase in size , the matrix becomes calcified, and the cells die [1].

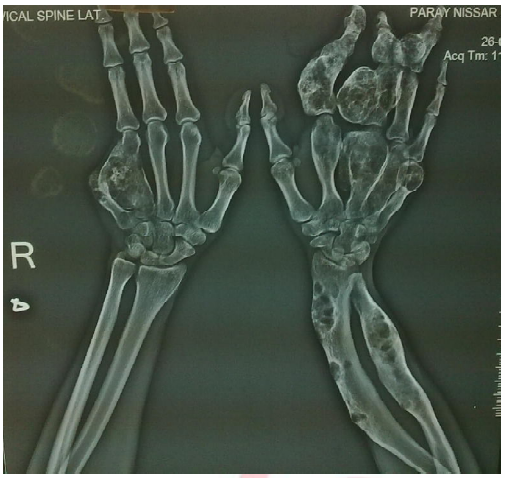

Fig 1: Radiograph of a 18 year old male showing Enchondromas.

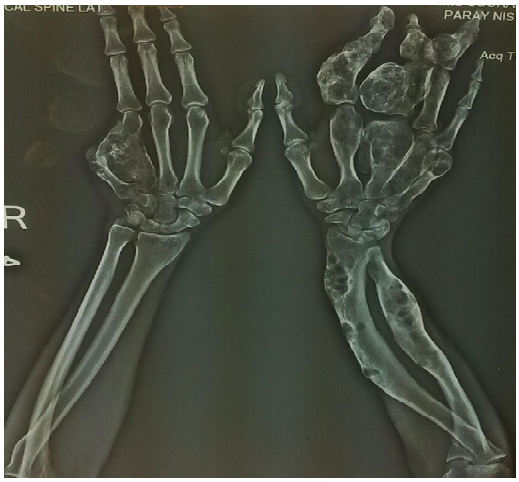

Fig 2: Radiograph of a 18 year old male showing Enchondromas.

Concurrently, a thin layer of bone is deposited under the perichondrium surrounding the diaphysis; thus, the perichondrium becomes the periosteum. Invasion by vascular connective tissue from blood vessels surrounding the periosteum also breaks up the cartilage. Some invading cells differentiate into hemopoietic cells, blood cells, of the bone marrow. This process continues toward the epiphyses (ends of the bone). The spicules of bone are remodeled by the action of osteoclasts and osteoblasts. In case this process of enchondral ossification is not completed. Enchondromas are formed. The behaviour of growth plate chondrocytes is tightly regulated at all stages of endochondral ossification by a complex network of interactions between circulating hormones (including growth hormone and thyroid hormone), locally produced growth factors (including Indian hedgehog, bone morphogenetic proteins and fibroblast growth factors) and the components of the extracellular matrix secreted by the chondrocytes (including collagens, proteoglycans, thrombospondins and matrilins) [2].

An enchondroma is a benign growth of hyaline cartilage lying in the medullary cavity of a bone. It arises from ectopic cartilaginous rests from the physeal plate. Enchondromas have an equal gender distribution and most frequently occur in the phalanges and metacarpals of hands. Enchondromas also appear in other long bones, as well as the pelvis, scapula, and ribs. These lesions are rarely symptomatic and are most often discovered on radiographs obtained for other purposes.

The tumor is more often recognized incidentially. However in severe cases they can present with excruciating pain or pathological fractures or severe deformities [3].

Physical examination may be normal or can reveal shortening of bones or angular deformities.

In enchondromatosis with hemangiomatosis referred to as Maffucci syndrome, multiple enchondromata and hemangiomata of bone and overlying skin develop during childhood [4].

The chromosomal changes in chromosome 1 are implicated in causation [5].

The majority of affected persons are normal at birth; the lesions develop during infancy. The most common cutaneous lesions are usually cavernous or capillary hemangiomas, with or without lymphangiomas. Their distribution in skin appears to be independent of skeletal lesions; they may be found also in mucous membranes and intra-abdominal viscera. The skeletal lesions are typical enchondromata, involving metaphyses throughout the body.

Maffucci syndrome produces a severe, cosmetically unsightly, and often painful deformation of the skeleton. The lesions can lead to short stature or, if predominantly unilateral, to leg length discrepancy and scoliosis. The most serious complication is the development of malignancy, which has a higher incidence than malignant change in this disease . Chondrosarcomatous transformation of one or more enchondromata may occur; sarcomatous degeneration of hemangiomas and lymphangiomas has been also been reported [6].

Radiographically, enchondromas in the metacarpals produce a lucent defect with well-defined margins and surrounding sclerotic reactive bone. The overlying cortex may be expanded and thinned. Varying degrees of stippled calcification may be seen within the lesion. When occurring in the metaphyses of larger long bones, the enchondroma may be less well defined. Subtle areas of stippled calcification may be the only indication of their presence. They may be better defined by CT scanning and MRI. Radionuclide scans usually show increased uptake that corresponds in extent to the size of the lesion identified on axial imaging. Endosteal erosion and expansion of the cortex are less common in enchondromas of the major long bones.

Patients present because of growth disturbance, which may be asymmetric, leading to limp, or because of swelling of the fingers and toes in infancy. The tumors may produce visible or palpable swelling, particularly in the hands or the growing ends of the long bones; they are somewhat elastic and may limit mobility of neighboring joints. Phalangeal chondromas may lead to severe deformation of the fingers.

Grossly, enchondromas are blue-white and translucent. Microscopic examination reveals lobules of hyaline cartilage with a mild degree of increased cellularity compared with normal hyaline cartilage.

Enchondromas have a potential for malignant degeneration. Any clinical evidence of growth or the development of pain in the region of an enchondroma should raise suspicion of malignant degeneration. There can be development of the chondrosarcoma. The risk for malignant transformation is low in case of solitary lesions and more in case of multiple lesions. Hence Multiple Enchondromatosis also called as Olliers disease is more prone for malignant transformation [7].

Radiographic evidence of cortical erosion or periosteal reaction is suspicious for malignant degeneration. There is a higher chance for visceral malignancies especially gastrointestinal malignancies and brain tumors such as Astrocytoma and chondrosarcomas of cranium [8]. chondrosarcoma, granulose cell tumors of ovary and precocious puberty [9.10,11].

Enchondromas may occur as multiple lesions (Ollier's disease) and may produce growth disturbance in early childhood. Distortion in both length growth and angular growth may be observed. The likelihood of malignant degeneration in multiple enchondromas is higher than for solitary lesions. Periodic radiographic survey is indicated in patients with multiple lesions. On the rare occasion when enchondromas require surgical treatment, intralesional curettage and bone grafting are appropriate.

CONCLUSION

Although a simple mechanism of incomplete enchondral ossification can lead to these localized cartilaginous tumors but many a times, incidental findings are seen while investigating a patient. One important incidental finding can be seen in the form of enchondromas which are usually solitary and conservatively managed but in certain cases especially when multiple can have malignant potential which can be dangerous. Further their association with different types of malignancies are an added danger and any person presenting with multiple enchondromas should be screened for these co morbid serious diseases.

Conflicts of Interests: **None**

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