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Case Report

Radiology Section

Multiple Enchondromatosis: Olliers Disease- A Case Report

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ABSTRACT

Olliers disease is a rare disease characterized by widespread enchondromas with a unilateral predominance, in early childhood. The diagnosis is based on clinical and conventional radiological evaluations. The prognosis for olliers disease is difficult to assess. Enchondromas in olliers disease present a risk of malignant transformation into chondrosarcomas. Due to its rarity literature focusing on olliers disease is limited. This case report discusses the case of a 12-year-old female who presented with difficulty in walking, leg pain and multiple painless swellings in hands and legs for a period of 2 years.

Keywords: Chondrosarcomas, Enchondromas, Scoliosis

CASE REPORT

A 12-year-old female patient presented to our hospital with difficulty in walking, leg pain and multiple swellings in hands and leg for 2 years. There was no such history in her family.

On examination, multiple discrete, hard swellings were palpable in both legs and hands. Overlying skin was normal. There was shortening of right leg which was noticed by the parents since 3 years of age. An informed consent was taken and plain x-rays of legs, feet, hands, forearms, hip with pelvis, and thoracolumbar spine were obtained. X-ray of right leg (AP and lateral views), pelvis with both hips (AP view) showed multiple lytic expansile lesions involving the metaphysis of femur, tibia and fibula [Table/Fig-1]; and in the metaphysic of right femur, both iliac bones and both pubic bones [Table/Fig-2] respectively. X-ray both hands AP view showed multiple enchondromas involving bilateral metacarpal bones and phalanges [Table/Fig-3a]. X-ray of right forearm showing short distal end of ulna-reverse Madelung deformity [Table/Fig-3b]. Similar lesions were also observed in X-ray (AP view) of both feet involving metatarsal bones and phalanges [Table/Fig-4a]. X-ray thoracolumbar spine AP view showed scoliosis [Table/Fig-4b].

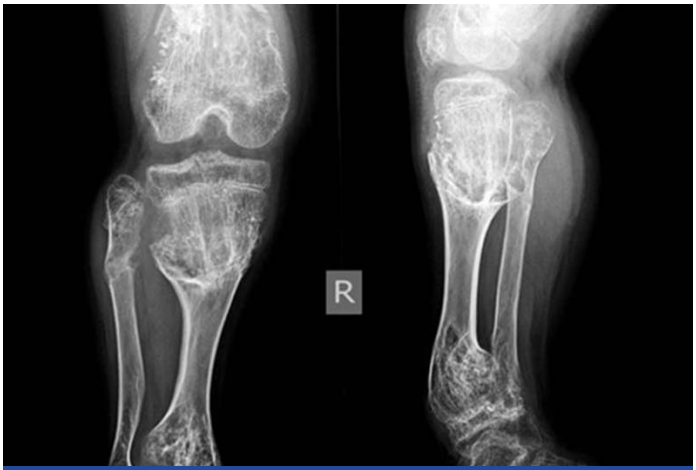
A diagnosis of multiple enchondromatosis (olliers disease) was made based on the morphology and location of bone lesions on plain radiographs. However, a differential of hereditary exostosis needs to be ruled out. No other tests were performed apart from x-rays as it was not affordable for the patient and she did not visit us for further follow up or treatment even though advised.

DISCUSSION

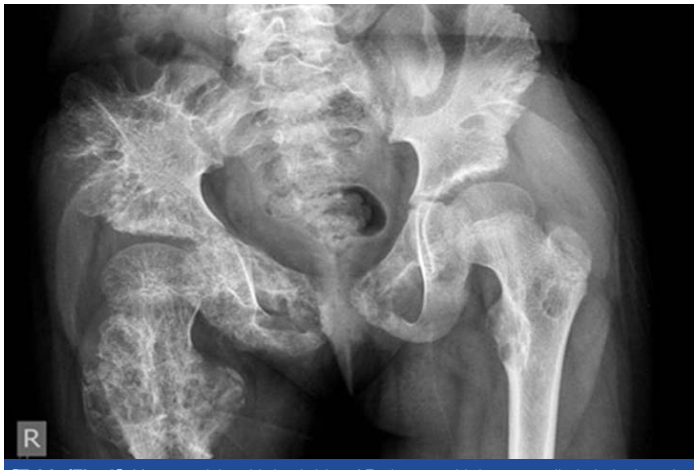
Olliers disease is characterized by the presence of at least three enchondromas [1]. The pathogenesis of olliers disease is unknown [2]. It was proposed that they result from abnormalities in signalling pathways controlling the proliferation and differentiation of chondrocytes, leading to the development of intraosseous cartilaginous foci [3].

Olliers disease and maffucci syndrome are usually non-familial disorders [3]. The non-hereditary asymmetrical polyostotic distribution of the lesions might suggest a somatic mosaic mutation. PTH1R mutations may contribute to the disease in a small subset of olliers patients but is probably not causative for the disease [1]. There is no marker that would indicate progression towards malignancy, thus there is a vital need to understand the genetics of these tumours which may help to develop markers for early diagnosis [4]. Despite the recent advances in the molecular diagnosis of congenital abnormalities the final decision for genetic evaluation still relies on clinical and radiological criteria [5].

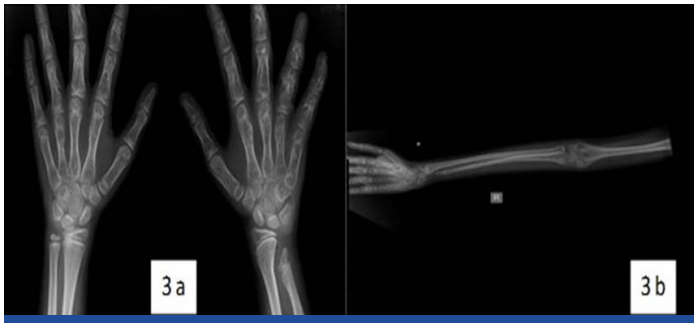
The prevalence of this disease is 1 in 100,000. Enchondromas frequently affect the long tubular bones, particularly the tibia, the femur, and/or the fibula; flat bones, especially the pelvis, can also be affected [2].



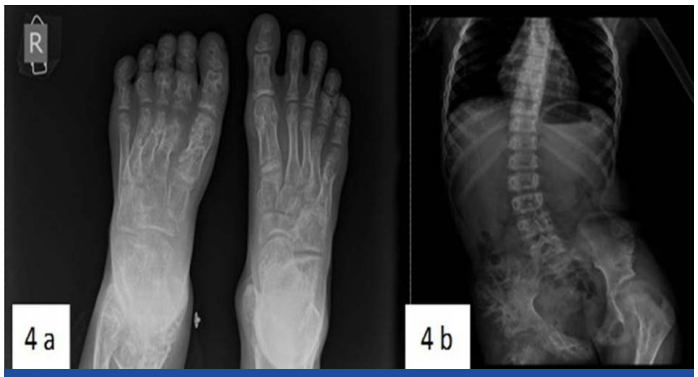
[Table/Fig-1]: X-ray of right leg AP and lateral views- multiple lytic expansile lesions in the metaphysis of femur, tibia and fibula



[Table/Fig-2]: X- ray pelvis with both hips AP view- multiple expansile lytic lesions in the metaphysis of right femur, both iliac bones and both pubic bones



[Table/Fig-3]: a) X-ray of both hands AP view - multiple enchondromas involving bilateral metacarpal bones and phalanges; b) X-ray of right forearm - short distal end of ulna-reverse Madelung deformity



[Table/Fig-4]: a) X-ray both foot AP view – multiple expansile lytic lesions involving metatarsal bones and phalanges; b) X-ray thoracolumbar spine AP – scoliosis

The patients become symptomatic usually in the first decade of life. Symptoms begin with the appearance of palpable bony masses on a finger or a toe, an asymmetric shortening of an extremity with limping, osseous deformities associated with or without pathologic fractures [3]. The lesions are usually asymmetrical, that are localized unilateral, if bilateral, and there will be one dominant side [2].

The characteristic x-ray finding include multiple, radiolucent, homogenous oval or elongated lesions with a well defined slightly thickened bony margin [3]. Magnetic resonance imaging demonstrates lobulated lesions with intermediate signal intensity on T2- weighted images and predominantly high signal intensity on T2- weighted sequences. However routine use of magnetic resonance imaging is not recommended because plain radiographs provide adequate information [2].

Malignant transformation is a major complication of enchondromatosis. Signs of malignancy should be looked for which include cortical erosion, extension of the tumour into soft tissues, and irregularity or indistinctness of the surface of the tumour. Compared to chondrosarcomas which show poor demarcation, enchondromas tend to be well circumscribed. Enchondromas normally show a uniform pattern of mineralization. Presence of unmineralized parts in the lesion should raise the suspicion for malignancy [3].

Olliers disease must be differentiated from hereditary multiple exostosis. The osteochondromas seen in hereditary multiple exostosis are located at the bone surface whereas enchondromas are located in the centre of bones [3].

There is no medical treatment for olliers disease. Surgical intervention is necessary in case of pathological fractures, growth defect and malignant transformation [1]. In patients with limb discrepancy distraction osteogenesis with the use of ilizarov instrument is indicated [2]. Lifelong monitoring is required in olliers patients given the risk of malignant transformation [6].

Of all patients diagnosed with olliers disease, malignant transformation is believed to occur in 10–20% and a recent study recorded the development of one or more chondrosarcomas in 40% of patients with olliers diseases and maffucci syndrome, respectively [7]. When malignancy is suspected, the histopathology investigation is used for grading because different grade requires different management [2].

CONCLUSION

Enchondromas are common intraosseous tumours which are usually benign and develop in close proximity to growth plate cartilage. Plain radiographs are sufficient for diagnosis. Since there is a risk for malignant transformation in these tumours a careful lifelong followup is necessary.

REFERENCES

[1] Pansuriya TC, Kroon H M, Bovée JV. Enchondromatosis: insights on the different subtypes. *Int J Clin Exp Pathol*. 2010;3(6):557-69.
[2] Wejjakul W, Pruksakorn D, Sirirungruangsarn Y, Luevitoonvechkij S, Khunsree S, Vaseenon T. The literature review of ollier disease. *Chiang Mai Med J*. 2013;52(3-4):73-9.
[3] Silve C, Jüppner H. Ollier disease. *Orphanet journal of rare diseases*. 2006;1:37.
[4] Pansuriya T, Oosting J, Krenács T, Taminiau A, Verdegaal S, Sangiorgi L, et al. Genome-wide analysis of Ollier disease: Is it all in the genes? *Orphanet Journal of Rare Diseases*. 2011;6(1):2.
[5] Vanhoenacker F, Van Hul W, Gielen J, De Schepper A. Congenital skeletal abnormalities: an introduction to the radiological semiology. *European Journal of Radiology*. 2001;40(3):168-83.
[6] Pansuriya T, Bovée J. Ollier disease. *Atlas of Genetics and Cytogenetics in Oncology and Haematology*. 2011;(8).
[7] Herget GW, Strohm P, Rottenburger C, Kontny U, Krauß T, Bohm J, et al. Insights into Enchondroma, Enchondromatosis and the risk of secondary Chondrosarcoma. Review of the literature with an emphasis on the clinical behaviour, radiology, malignant transformation and the follow up. *Neoplasma*. 2014;61(04):365-78.

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