Ollier Disease: A Case Report

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ABSTRACT

Ollier disease is a rare disease featuring multiple enchondromas mainly affecting limbs. We describe a boy who presented in our OPD with multiple painless joint deformities mostly in the both upper and lower limbs. Conventional X-ray evaluation revealed typical multiple radiolucent, homogenous oval and elongated shapes in the deformed limbs.

KEY WORDS: Ollier Disease, Enchondromatosis

INTRODUCTION

Enchondromas are common, benign, usually asymptomatic cartilage tumours that develop in metaphyses and may also involve the diaphysis of long tubular bones.² Ollier disease is defined by the presence of multiple enchondromas and characterized by an asymmetric distribution of cartilage lesions which can be extremely variable.² The prevalence of Ollier’s disease is estimated to be 1/100000. Children with symptomatic enchondromatosis cases usually present before puberty with deformity, growth disorders and recurrent pathological features.³,⁴

Enchondromas appear as well defined medullary lesions and lobulated contour, endosteal erosion ground glass appearance of matrix and sometimes maybe accompanied with expansion of bone thinning of cortex and dysmorphic calcifications within the matrix.⁴

CASE

A 10 years old boy came to the Paediatric OPD at TUTH with history of multiple joint swellings since 1 year of age. According to the patient, the joint swellings are gradually increasing in size without pain but causing deformity of limbs. There is no history of weight loss or traumatic injury. There is no similar family history. He is active, appropriate in all domains of development except delayed motor milestones, short limb gait and postural lumbar scoliosis. His vitals are stable. Weight is 20kg and height is 106cm (both less than 3rd percentile of NCHS). His head circumference is 52cm (50th percentile). Upper segment to lower segment ratio is 0.89cm. Multiple joints are swollen but nontender and non-erythematous. There are mild restrictions in range of motion of all joints. There are angular deformities of bilateral arms, legs and left wrist (cubitus varus deformity of right elbow, genu valum, cavovarum deformity of left foot, valgus deformity of right foot). Bony deformity is present at inferior pole of right scapula and right 3rd, 4th and 5th costochondral junction. Left pelvis is at higher level. Lower limb length discrepancy (true shortening of right lower limb) is 3cm. Examination of fingers and toes shows significant macrodactyly (Figure 1). Laboratory investigations shows normal hemoglobin, total leukocyte count, differential count, ESR. His liver, renal function test, serum calcium, phosphorus, 24 hour urinary calcium and phosphorous are also within normal limits. Plain radiography skeletal survey (Figure 2) shows increased lucency of the metaphyseal region with expansion at the site with multiple punctuate calcification with ring and arch like pattern noted in the conserved bones. Growth plate is partly visualized with relative
preservation of epiphysis. No obvious diaphyseal involvement is noted, with relative narrow zone of transition. There is non-visualization of the cortical margins at the metaphyseal regions of the concerned areas. Increased linear intermittent trabeculations are noted which are vertically oriented along the long axis of the bones. Skull and spine is spared. Chest X-ray is normal. In MRI (Figure 3) T2 Weighed axial and coronal images of knee shows mixed signal intensity masses in the metaphysis of distal femur, proximal tibia and fibula. Masses showed predominately high signal intensity with intermixed areas of isointense and low signal intensity. Expansion of metaphyses was also noted with thinning of the cortex. Ultrasonography of abdomen is normal. There are no evidence of haemangiomas on physical examination. Thus the case is diagnosed as Ollier Disease on the basis of clinical presentation and radiological evidence.

DISCUSSION

Ollier disease is a non-familial disease with multiple enchondromas that typically affect the metaphyseal ends of bones. In Maffucci syndrome, apart from multiple enchondromas, there are soft tissue hemangiomas. Enchondromas present on the extremities are usually visible as masses within phalanges, metacarpal and metatarsal bones. Enchondromas are frequently located in the long tubular bones, particularly the tibia, the femur and in some cases flat bones. Affected bones generally become shortened and deformed.

In our case, apart from the limb involvement, involvement of scapula, ribs and pelvis is evident clinically and radiographically with right sided predominance of lesions. The head and spine are spared. A minor delay in bone age has been observed in children affected by Ollier disease.

Ollier Disease is diagnosed on the basis of clinical findings and conventional radiological examinations. Histology evaluation done mainly when the possibility of malignancy is considered. Magnetic resonance Imaging and Ultrasound are generally done for evaluation and monitoring of disease progression in such lesions. The lesions can undergo malignant transformation such as chondrosarcomas. Ollier disease has also been associated with other tumours such as ovarian and brain tumours.

Studies have shown the association of mutations in isocitrate dehydrogenase and Parathyroid Hormone/Parathyroid Hormone related protein in cases of enchondromatosis.

There is no medical treatment for Ollier disease. Surgery is indicated in case of complications such as pathological fractures, malignant transformations, growth defects and for intracranial enchondromas.
CONCLUSION

In Ollier disease multiple enchondromas occur, mostly in the limbs. The diagnosis is based on clinical features and conventional x-ray images. Symptomatically, children present with deformities, pathological fractures and growth defects whereas there is possibility of malignant transformation of the lesions with increasing age. This case report highlights a rare bone disorder in children with important clinical implications.

REFERENCES


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