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Hereditary Multiple Osteochondromas - A rare accidental skeletal finding

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Abstract

Hereditary multiple exostoses is a rare genetic disorder outgrowing from bony metaphyses. Here we present an 11 year old female child, accidentally diagnosed with the condition on examination when admitted for a respiratory tract infection. Associated with short stature, restricted joint movements or sometimes bone pain, the condition can often go unnoticed as in the case discussed. Radiography of the affected bones was done and MRI to rule out malignant transformation. Here we discuss how even though it was an accidental finding, if diagnosed early, it can be prevented from resulting in malignancy or neurovascular compromise, which are the common complications of this condition.

Keywords: Hereditary multiple exostoses, osteochondromas, malignancy

Introduction

Also known as Hereditary deforming dyschondroplasia, diaphyseal achlasis and multiple cartilaginous exostoses, this rare autosomal dominant genetic disease is passed down directly from affected parents in 50% cases. 1It is characterised by growth of multiple osteochondromas, benign cartilage capped bone tumors that outgrow from the metaphyses of long bones as well as flat bones.1The disease occurs in only 5-10% of the cases when compared to solitary exostoses.

1It is associated with reduction in skeletal growth, bony deformity, restricted joint motion, shortened stature, premature osteoarthrosis, and compression of peripheral nerves, causing pain.1

Case Report:

Khushi, 11 year old female child was admitted for upper respiratory tract infection. On examination, multiple bony lesions were noticed on arms and legs. Lesions were hard, irregular, of variable sizes. No signs of neurovascular impingement were found during physical examination. Patient was investigated further, laboratory tests were within normal limit, consisted of complete blood count, urinalysis, and renal function test. A radiographic examination of the axial skeleton demonstrated multiple osteochondromas of bilateral humerus; distal ulna and radius; distal and proximal femurs, tibias, and fibulas. Lesions were most prominent along the metaphyses on long bones, the largest being on the juxtaphyseal region of the tibia, below the right knee. The lesions were not associated with pain or joint movement restriction. There was no family history of similar complaints in the parents. Lesions were differentiated from enchondromas and possibility of associated syndromes like Maffucci syndrome were ruled out due to absence of soft tissue hemangioma. On confirming the diagnosis of hereditary multiple exostosis, MRI of the largest lesion was done to rule out malignant transformation. Subsequently, contrast-enhanced MRI was performed to determine the composition, the cartilage cap of the lesion and soft-tissue extension.

Discussion:

2Hereditary multiple exostoses is caused by mutations and functional loss of the EXT1 AND EXT2 genes on chromosome 8 and 11 respectively, which encode glycosyl transferases, and enzyme family involved in heparan sulfate synthesis. Osteochondromas or osteocartilaginous exostoses or simply exostoses are not true neoplastic lesions but rather developmental lesions of bone. 3The mechanism of development is probably the result of ectopic development of growth plate of cartilage. The epiphyseal cartilage was separated from normal growth plate and herniated into the periosteal bone cuff. The enlargement of this cartilaginous fragment and its eventual enchondral ossification explained the excrescent growth of osteochondroma from bone surface and its cartilage cap. No involvement of skull, carpal and tarsal bones have been reported. HMEs usually impact quality of life and physical activity levels in children, in addition to mental health, especially in females.

Usually when osteochondroma has an early onset, lesion tends to be bigger and causes more problems, this happens because the lesion tends to grow in line with the growth plate and adjusts itself to the evolution of the diaphysis. It can produce disturbances in the growing diaphysis causing limb length discrepencies and characteristic deformities.

2Pediatricians, geneticists and orthopedic surgeons play an important role in the study and treatment of this severe pathology. Generally diagnosed before the age of 12, MRI is the ideal imaging modality for assessing osteochondroma, most common presentation being the appearance of palpable mass on knee, shoulders, ankles and wrist. Incidence is underestimated because most individuals with asymptomatic lesions are undiagnosed.2 Complications of osteochondroma were reported including fracture, deformities, bursitis, neurovascular compromise, and malignant transformation.

There is no medical treatment for the condition, surgery only recommended in symptomatic exostosis or in cases where malignant transformation is suspected. 1Painful lesions in the absence of bony deformity is corrected with surgical excision including cartilage cap and overlying perichondrium to prevent recurrence. Resection, limb shortening, ulnar lengthening procedures and corrective osteotomies are the other available treatments. Hemiepiphysiodesis, a surgical procedure affecting the growth plates, may be done in the lower extremities and wrist to correct any misalignment of the bones during future growth. Surgical resection is used to treat sarcomatous degeneration.1

Conclusion:

1Hereditary multiple osteochondromas should be suspected in individuals with multiple osteochondromas arising from the area of the growth plate in the juxtaphyseal region of long bones or surface of flat bones. Prenatal testing for a pregnancy at increased risk and preimplantation genetic diagnosis are possible if the pathogenic variant in a family is known.1 Genetic counselling is recommended for affected individuals and their families.

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