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Case Report

A RARE CASE OF HEREDITARY MULTIPLE EXOSTOSES

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ABSTRACT

Objective: A rare genetic condition, hereditary multiple exostoses (HME), follows the autosomal dominant pattern of inheritance. It is evident by multiple bony elevations on the appendicular skeleton by the age of 2 years.

Case Report: Here, we present the 42-years-old patient known case of CKD presenting with uncontrolled hypertension and breathlessness; however, incidentally, it was noted that there is bilateral swelling in the knee and further investigated for X-ray and blood parameters. On assessing the similar pattern in family members, it was noted such swelling over the shoulder in the daughter and swelling in the wrist joint in the younger brother.

Conclusion: Hereditary multiple osteochondromas are a hereditary ailment that should be studied. Despite tremendous advances in recent years, the intricacy and unresolved difficulties associated with HME remain a challenge for academics and physicians.

Keywords: Hereditary multiple exostoses, Deformity, inherited, Autosomal dominant.

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INTRODUCTION

Several benign osteochondromas (exostoses) are seen in a rare genetic condition known as hereditary multiple exostoses (HME), which follows an autosomal pattern of inheritance. It affects around one in every 50,000 persons and does not appear to be sexually predominant. This disease has been addressed by multiple names in literature which are multiple cartilaginous exostoses, hereditary deforming dyschondroplasia, diaphyseal aclasis, and hereditary multiple osteochondromas. It has nearly full penetrance, especially in males [1-3]. In the majority of the patients, the condition is passed down by the father who is affected by the disease. The possibility of transfer of disease by an unaffected male is rare, but a suppressed or dormant form of the disease can be transmitted by the unaffected female.

CASE REPORT

A 42-years-male patient presented to the emergency department with complaints of breathlessness, raised blood pressure, and complain of vomiting 3 days back. There was no history of known diabetes mellitus and tuberculosis. He was a bidi smoker for the past 15 years and an alcoholic for the past 5 years. There was a similar episode 15 days before the present episode, and diagnosed as hypertensive. On examination, there was swelling around the knee, which was present bilateral present. The patient was screened for the blood cell picture, serum lipid profile, X-ray joint, and USG abdomen. KFT of the patient was deranged urea – 150 mg/dl and creatinine – 7 mg/dl; USG KUB was advised, and it revealed loss of cortical medullary differentiation. The patient was labeled as CKD with hypertension. Blood parameters for lipid profile, the serum cholesterol, HDL, LDL and triglycerides, CBC, and thyroid profile were found to be within normal limits. There is an elevation of the ESR inpatient. Initially, the size of the swelling was small, but over the course of time, it grew in size. The swelling was initially small in size. The patient did not complain of any weight loss, fever, or cough. He also denied any history of trauma at the site of the swelling. On further inquiry and examination of his daughter, she had similar signs of swelling in the joints of the left shoulder, and the patient's brother had the presence over the wrist joint as shown in Fig. 1. A diagnosis of multiple hereditary exostoses, on examination of the patients, family history of similar signs and symptoms and also supported with the X-ray of the limb involved as shown in Fig. 2.

DISCUSSION

When compared to solitary exostosis, the condition occurs in only approximately 5% to 10% of instances, it is 1.5 times more common in men compared to women, and it is detected in the first decade of life in majority of cases [2,3]. Osteochondromas can develop from any segment except for the facial bones. Any bone formed as a result of endochondral ossification can be affected by the disease. Frequently, exostosis involves the ribs, scapulae, vertebrae, and pelvis; the met diaphyseal segments are the most afflicted. There has been no mention of the cranium, carpal, or tarsal bones being involved. Exostoses can create a variety of clinical difficulties due to their wide range of location, size, and number [1,4].

Exostosis is the most common sign of HME. Osteochondromas, which are formed by theprogenitor cells of the chondrocytes in the growth plate have recently been discovered in the perichondrium's inner layer [5]. Understanding how osteochondromas develop in HME patients requires genetic, molecular biology, and clinical research. EXT1 gene was discovered while gene sequencing in HME patients, later EXT2 gene was discovered which are situated on Chromosome-8 in 28%–65% and Chromosome 11, in 21%–61% of the afflicted patients, respectively [6], EXT1 and EXT2 mutations were found [7-9]. Mutations are more commonly associated with EXT1 genes, when compared to EXT2, as EXT2 before the disease onset can tolerate a larger mutation [10]. EXT1 gene mutations are associated with a shorter stature, more incidences of limb malalignment, more pelvic involvement, and greater number of exostoses [8]. Surprisingly, 5%–34%patients did not have either mutation [7-9,11]. Since, at birth patients are asymptomatic, routine screening for genetic mutation in afflicted family can lead to early diagnosis. The condition in often diagnosed during median age, that is, third decade of life [3]. By the age of 5 years, half the cases are diagnosed as they develop a visible tumor, and by the age of 10, it can be detected in 80% of the affected individuals. In general, all patients are

Fig. 1: Clinical picture showing the joint swelling in (a) Daughter and (b) Brother of patient

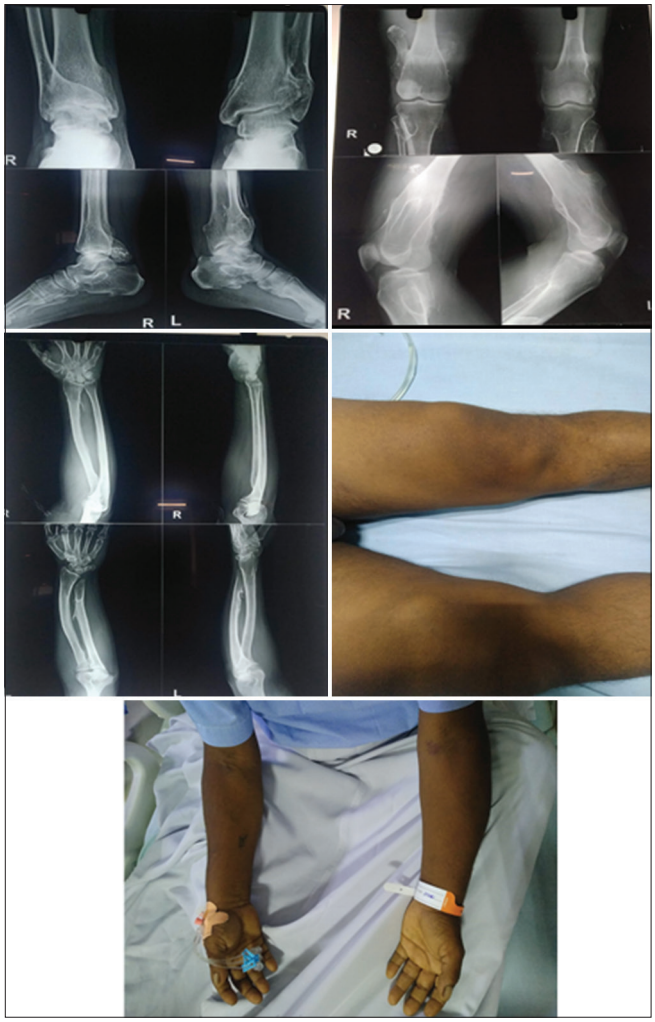

Fig. 2: Radiological and clinical picture of the patient

diagnosed between the ages of 10 and 12. In most places, conventional roentgenograms detect the shape and existence of osteochondromas with high accuracy. X-rays can also be used to detect and analyze HME-related abnormalities. In radiological studies done by Kok *et al.* ulnar shortening and pseudo-made-lung deformities have been found in 33% of cases; "Erlenmeyer flask," a deformity of the distal femur has also been found in conjunction with the disease, and in the lower limbs, X-rays can also detect axial deviations, shortenings, and limb length disparities [12].

Wicklund discovered that HME patients are shorter in stature than the general population [1].

By the age of 5

The fifth percentile is occupied by 37% of males and 44% of females. These traits are not attributable to deformities and limb length disparities, as observed in various statistical studies. Mutations in EXT1 and EXT2 also affects patients' growth. Higher risk of exostosis, exostosis-related comorbidities, and low stature have been observed with mutation of EXT1. During pregnancy, dimensional changes in exostosis have been reported by 63% of patients, and a C-section was required due to overcoming the problems encountered from exostosis during birth in 63 % of patients. Short stature, limb-length disparities, valgus deformities of the knee and ankle, asymmetry of the pectoral and pelvic girdles, lateral bowing of the radius with ulnar deviation of the wrist, and radial head dislocation are the most prevalent malformations seen in HME. Malignant transformation of a benign osteochondroma to chondrosarcoma or other sarcoma is another complication of HME.

Treatment is not recommended in non-complicated cases of HME till the patients reach the age of skeletal maturity to avoid any damage to physis. When the lesion leads to any discomfort, is in close proximity to the neurovascular bundle, or leads to deformity, then surgical removal of the lesion shall be considered. The goal of surgery is to completely remove the cartilaginous cap. Incomplete excision is associated with a recurrence probability of around 2%. Age of presentation and severity of the disease are taken into consideration while selecting the therapy for secondary deformity therapy and may include corrective osteotomy, prompt epiphysiodesis, and limb lengthening.

CONCLUSION

Hereditary multiple osteochondromas are a hereditary ailment that should be studied and treated by an orthopedician, a geneticist, and a pediatrician. Despite tremendous advances in recent years, the intricacy and unresolved difficulties associated with HME remain a challenge for academics and physicians.

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CONFLICT OF INTEREST

Nil.

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