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Case Report

A Young Boy with Multiple Bony Overgrowths

MA Hannan[1], Md Rakibul Hasan[2], Sharmin Jahan[3], Md Shahed Morshed[4]
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Abstract

Hereditary multiple exostoses is a rare autosomal dominant pediatric disorder with an incidence of about 1:50000 characterized by multiple cartilage-capped bony protuberances, called osteochondromas or exostoses, projecting from the metaphyses of long bones. It is caused by loss of function mutations in exostosin-1 and exostosin-2 genes that encode glycosyltransferase enzymes involved in the synthesis of heparan sulfate which has fundamental role in extracellular matrix formation during bone development. It commonly presents with compressive symptoms due to bony overgrowth involving all bones except calvarium and rarely transformed into malignancy. No definite treatment is available, but careful screening of these exostoses with timely referral to respective surgeon prevents long term complications and improves quality of life.

Key words*: Hereditary multiple exostoses; Bony overgrowth*

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Introduction

Hereditary multiple exostoses (HME), also known as osteochondromatosis, is a rare autosomal dominant pediatric disorder with an incidence of about 1:50000[1] affecting predominantly metaphysis of the long bones or the surface of flat bones. It is mainly caused by loss of function mutations in exostosin-1 (EXT1) and exostosin-2 (EXT2) genes located on chromosomes 8 (locus 8q24.1) and 11 (locus 11p11−13) respectively[2], that encode glycosyltransferase enzymes involved in the synthesis of heparan sulfate (HS). HS is a proteoglycan which plays a fundamental role in the extracellular matrix formation during cartilage development.[3] The disease is characterized by multiple cartilage-capped bony protuberances, called osteochondromas or exostoses, projecting from the metaphyses of long bones.[4] Exostoses are usually bilateral, but may be unilateral and involve any bone in our body except the calvarium.[5] Here we report a case of a 17-year-old boy who presented in endocrine outpatient department (OPD) with short stature and multiple hard protuberances from the long bones of extremities.

Case report

A 17-year-old boy (Fig 1) presented to endocrine OPD for evaluation and management of short stature and multiple hard protuberances from the long bones of extremities. He is the only issue of non-consanguineous apparently normal parents delivered at home by normal vaginal delivery with normal birth weight. His perinatal history was uneventful. According to his mother's statement, few hard swellings were first observed around the knee joints and lateral chest wall at the age of 18 months. Since then their number has been gradually increasing over the last 15 years involving his lower end of femur, upper end of tibia-fibula (Fig 2), lower part of humerus, lower part of ulna (Fig 3) sparing the pelvic bones, calvarium,

1. *Consultant, Department of Medicine, Sherpur Sadar Hospital, Sherpur*
2. *Assistant Professor, Department of Endocrinology, Enam Medical College & Hospital, Savar, Dhaka*
3. *Assistant Professor, Department of Endocrinology, Bangabandhu Sheikh Mujib Medical University (BSMMU), Dhaka*
4. *Resident, Department of Endocrinology, BSMMU, Dhaka*

Correspondence *Md Rakibul Hasan, Email: dr.mrh46@gmail.com*

scapulae and teeth (Fig 4). No visible exostoses were present at spine, but scoliosis was present (Fig 5). These lesions produce local disfiguration and bowing of upper and lower limbs (Fig 1, 6) not associated with pain or any symptoms suggestive of nerve or blood vessel entrapment. Pseudo-madelung deformity (Fig 3) is present on both forearms predominantly on right side as a result of ulnar foreshortening with bowing of the radius, increasing interosseous space and dorsal subluxation of the distal radio-ulnar joint that produces apparently short 5th metacarpal on both sides (Fig 3). No evidence of pathological fractures or any other chronic systemic illness was present.

Table I: Anthropometry and Tanner staging

Parameters	Measurement
Height	140 cm
Weight	32 kg
BMI	16.3 kg/m^2
Growth Height for age Weight for age	 <3rd percentile <3rd percentile
Father's height	151 cm
Mother's height	137 cm
Midparental height	150.5 cm
Upper segment (US)	72 cm
Lower segment (LS)	68 cm
US:LS	1.06
Arm span	129 cm
Tanner staging	Testicular volume: 12 mL on both sides Stretched penile length (SPL): 8 cm Puberchy –P5

His milestone of development was appropriate for age including pubertal changes and school performance is average. Vital parameters are within normal limits.

Table II: Baseline investigations of the patient

Investigations	Results
S. albumin	44 g/L (35–57 g/L)
S. calcium	8.8 mg/dL (8.5–10.3 mg/dL)
iPO4	4.3 mg/dL (2.5–4.9 mg/dL)
FT4	1.8 ng/dL (0.81–2.10 ng/dL)
TSH	0.9 mIU/L (0.5–5.0 mIU/L)
Intact PTH	54.69 pg/mL (15–65 pg/mL)

Fig 1. Short stature with bowing of forearm

Fig 2. Multiple exostoses around knee

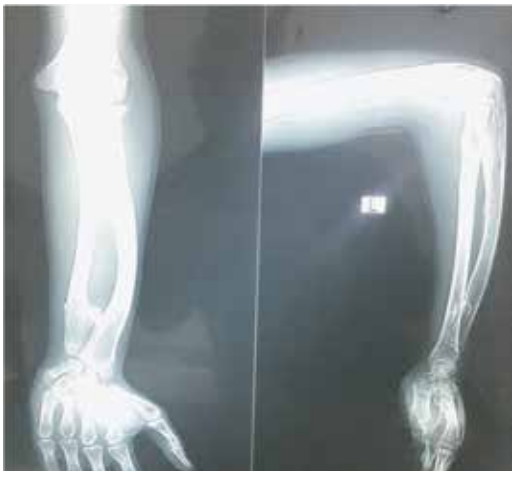

Fig 3. Exostoses and pseudo-madelung's deformity of forearm with bowing

Fig 4. Normal calvarium

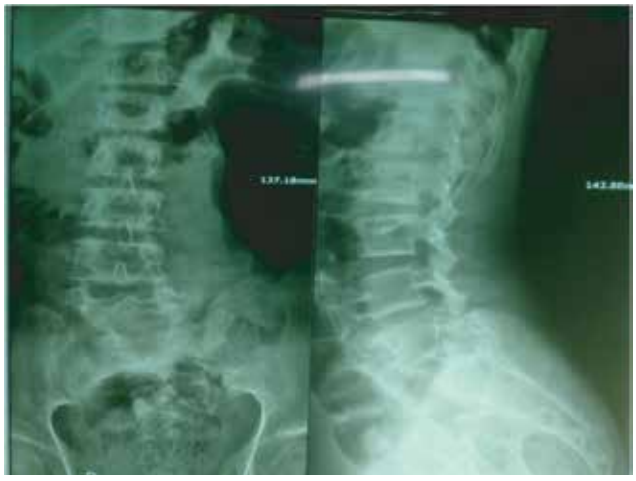

Fig 5. Scoliosis

Fig 6. Local disfiguration with bowing of legs

Discussion

Hereditary multiple exostoses is a rare autosomal dominant disease of bone. Bony protuberances or exostoses may vary in number and size. Patients are usually asymptomatic at early period, but may present with wide spectrum of physical symptoms predominantly neurovascular compression depending on their exact location.[6,7] In many case series, involvement of pelvic bones, hip joints, spine, scapula and teeth was reported.[5,8,9] It was observed that about 2.7% patient may undergo malignant transformation predominantly at pelvis and scapula.[10] One of the possible differential diagnosis is enchondromatoses which is a heterogeneous group of congenital disorder characterized by the presence of multiple enchondromas associated with musculo-skeletal malformations secondary to limb shortening, scoliosis, pathological fractures and pseudoarthrosis with high rate of malignant transformation (15–25%) to secondary chondrosarcomas.[11] Another differential diagnosis is tumoral calcinosis, but lesions are not typical in our case. Tumoral calcinosis is characterized by lesions composed of ectopic calcified tissue, most commonly seen in the large joints of the hips, shoulders, and elbows, but may involve the hand and wrist.[12] The diagnosis of HME is mainly clinical and radiological. Magnetic resonance imaging with soft tissue contrast can provide good view and suggests malignant transformation.[5] Nuclear medicine bone scan may be useful in identifying additional lesions throughout the skeleton. Genetic testing is indicated only in cases when the diagnosis is not known or it cannot be established in either of the parents. No curative treatment is available. Surgical treatment is considered in presence of complications, such as infection, synovial cysts, vascular, or nerve involvement or malignant transformation. Careful assessment of complications and timely referral improve patient survival and prevent long term complications.

Conclusion

No specific treatment is available for HME and treatment depends upon presentation of disease. Its complications are mainly associated with compression

due to bony overgrowth which should be properly identified by imaging. Early screening and referral to appropriate center is essential for all patients to prevent long term complications.

Patient consent

Written informed consent was obtained from the legal guardian of the patient for publication of the submitted article and the accompanying images.

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