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Case Presentation: Hereditary Multiple Exostosis in a Pediatric Patient

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Abstract: ***Hereditary Multiple Exostosis (HME) is a rare, autosomal dominant disorder marked by the development of multiple osteochondromas, primarily at the metaphyseal regions of long bones. The disorder may result in growth disturbances, deformity, and in rare cases, malignant transformation. This case involves an 8-year-old male presenting with gradually enlarging, painless swellings and forearm deformity. Clinical evaluation, radiographic findings, and surgical considerations are discussed. The case emphasizes the need for early diagnosis, routine surveillance, and timely intervention to prevent long-term complications.***

Keywords: Hereditary Multiple Exostosis, Osteochondroma, Pediatric Orthopedics, Forearm Deformity, Genetic Skeletal Disorder, Surgical Excision

1. Introduction

Hereditary Multiple Exostosis (HME), also known as Multiple Osteochondromas, is a genetic disorder affecting approximately 1 in 50,000 individuals globally. It is caused by mutations in the **EXT1 or EXT2** genes, which play a role in the biosynthesis of heparan sulfate. HME usually manifests during the first decade of life and presents with multiple bony outgrowths near the metaphyses of long bones. These lesions are typically benign but can cause skeletal deformities, limb length discrepancies, restricted joint mobility, nerve compression, and rarely, malignant transformation into chondrosarcoma.

Diagnosis is primarily clinical, supplemented by radiography. Management is symptomatic, with surgical excision indicated for functional impairment, pain, or significant deformity. This case discusses the presentation, diagnosis, and management considerations for a child with classic HME features.

The condition is caused by mutations in the **EXT1** or **EXT2** genes, which are involved in the biosynthesis of heparan sulfate—a critical component for normal endochondral bone growth. The clinical presentation of HME is variable, ranging from asymptomatic cases to severe skeletal deformities, limb length discrepancies, joint restriction, and in rare instances, malignant transformation into secondary chondrosarcoma.
Though benign in nature, the exostoses can cause significant functional limitations and cosmetic concerns. Forearm involvement is common and may result in ulnar shortening, bowing of the radius, and restricted range of motion.

This case highlights a classic presentation of HME in an 8-year-old male, emphasizing the need for early diagnosis, individualized treatment planning, and long-term follow-up to monitor for complications and progression of deformity.

2. Case Presentation

Patient Details:
Name: Master Diyan Kirk
Age/Gender: 8 years / Male
Informant: Mother

Parental Information:
Father's Name: Mr. Sarvesh (Age: 33, Occupation: Welder, Education: 8th std)
Mother's Name: Mrs. Bincy (Age: 32, Occupation: Housewife, Education: 12th std)

Chief Complaints:
Complaints of multiple swellings all over the body, gradually increasing in size and causing pain.

History of Present Illness:
- Firstborn child of a non-consanguineous marriage.
- No maternal illness or complications during pregnancy.
- Delivered at full term via LSCS due to breech presentation.
- Birth weight: 3.4 kg
- No NICU admission
- Patient noticed to have multiple swellings over the body gradually increasing in size with pain.
- No history of trauma.
- Admitted for further evaluation and management.

Developmental History:

Milestone	Age Achieved
Social smile	—
Head control	5 months
Crawling	6 months
Sitting	7 months
Walking with support	12 months
Walking without support	1.5 years
Self-feeding	—
Writing	—

Birth History:
- Place of Birth: Hospital
- Birth Order: 1st
- Presentation: Breech
- Mode of Delivery: LSCS
- Gestational Age: 37 weeks
- Birth Weight: 3.4 kg
- Perinatal Complications: None

Family History:
- One sibling: 17-year-old female, normal
- No similar illness in the family
- No history of hospitalization or drug allergies

Physical Examination:

General:
- Pulse: 102 bpm
- Height: 115 cm
- Weight: 18.6 kg
- Abdomen: Soft
- Nutritional status: Normal
- Gait: Bipedal, biphasic unassisted gait

Local Examination:

a) Back:
- 2×1 cm swelling over medial border of right scapula, stony hard, immobile, non-tender
- Bilateral scapula prominent

b) Chest:
- Multiple small hard swellings over ribs

c) Shoulders:
- Right: 2×3 cm swelling over lateral proximal humerus
- Left: 4×3 cm swelling over proximal ⅓rd humerus

d) Forearm:
- ~5×4 cm swelling over distal ⅓rd forearm
- Hard, immobile, skin pinchable
- Active finger movements normal
- Radial/Median/Ulnar nerve: normal
- Radial pulse: felt

e) Knee:
- Genu valgum noted bilaterally
- Diffuse swelling over medial condyle of tibia & fibula head
- ROM: -10° to 110°
- Hyperextension: 10°

Radiology Report:

a) Broad-based eccentric lesion over:
- Proximal humerus – cortex and matrix continuous with bone
- Distal ⅓rd radius – involving distal ulna

Provisional Diagnosis:

Hereditary Multiple Exostosis (HME)

Plan of Care:
- Under evaluation by Dr. VK
- Planned: **Bony excision of bilateral forearm lesions**

Treatment Consideration:
Master Diyan Kirk, 8-year-old male with known case of Hereditary Multiple Exostosis (HME) and bilateral forearm deformities.

In view of deformity, plan made to do **excision of bilateral forearm exostosis**.

Counselled:
- Possibility of neurovascular injury
- Risk of recurrence
- Development of future symptoms
- Persistence or worsening of deformity
- Informed consent taken from parents.

3. Discussion

HME arises due to loss-of-function mutations in the **EXT1** (chromosome 8q24.11) and **EXT2** (chromosome 11p11) genes. These genes encode glycosyltransferases essential for heparan sulfate biosynthesis, impacting chondrocyte proliferation and endochondral ossification. This disruption leads to abnormal bone growth and formation of osteochondromas.

In this case, the child displayed typical HME features: early-onset, symmetrical swelling in long bones, and forearm deformity. Though genetic confirmation was not reported, the clinical and radiological presentation strongly supports the diagnosis.

Surgical excision is reserved for lesions causing pain, deformity, or functional limitation. The decision to operate on forearm exostoses in this patient is based on both functional concern and cosmetic deformity. Regular follow-up is essential to monitor for complications including angular deformities, limb length discrepancy, and malignancy (1–5% risk).

Early orthopedic referral and genetic counseling are crucial, especially as this is a lifelong disorder with potential implications for family planning and long-term orthopedic health.

4. Conclusion

Hereditary Multiple Exostosis is a rare but significant skeletal dysplasia with potential to impair function and quality of life. This case highlights the clinical spectrum of HME and underscores the importance of a multidisciplinary approach for optimal management. Surgical treatment should be individualized based on symptoms and deformity, and long-term surveillance is crucial to address potential complications.

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