Proceedings S.Z.M.C. Vol: 35(3): pp. 78-80, 2021. PSZMC-814-35-3-2021

Hereditary Multiple Exostoses with Ulnar Hemimelia

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ABSTRACT

Hereditary Multiple Exostoses is a skeletal dysplasia that is very rare and defined by formation of numerous cartilage capped benign tumours either pedunculated or sessile known as osteochondromas throughout skeleton especially around the growth plates of ribs, vertebrae, pelvis and long bones. Rarely it can present forearm problems such bowing deformity of radius, ulnar shortening and radiocapitellar dislocation or subluxation. We are presenting a case of 20 year old female who presented with left distal ulnar exostosis resulting in ulnar shortening and radial bowing with restricted supination and pronation range of movement. Other complaint was of multiple non tender bony hard lumps in both upper and lower limbs. Excision of distal ulnar exostosis was done which resulted in marked improvement in pronation and supination range of movement. Hereditary multiple exostoses with forearm deformities though very rare but can present and the treatment is conservative except if any bony swelling manifests any complications such as pain or associated deformity.

Key words: Osteochondromas, Distal ulnar exostosis, Excision

INTRODUCTION

Diaphyseal achlasis also known as Hereditary multiple exostoses is a genetic dysplastic condition having autosomal dominant mode of inheritance where several benign cartilage capped tumors originate from the perichondrium around the physis of bones. It has a prevalence of 1:50,000 in Western countries.[1] It usually involves vertebrae, scapula, pelvis, ribs and long bones. Exostoses can result in reduction of skeletal growth, bony deformity, restricted joint movement, shortened stature, premature arthritis, and compression of peripheral nerves. It is very rare and can present with forearm deformities such as ulnar shortening, radial bowing and radiocapitellar subluxation.[2] Surgical excision of exostosis, limb lengthening and deformity correction are the available treatment options and are advised when osteochondromas become symptomatic or there is malignant transformation which rarely occurs.[3]

CASE REPORT

A 20 year old female presented to the outpatient orthopedic surgery clinic with bowing deformity and shortening of left forearm along with multiple swellings around knee and ankle joints. There as complaint of limitation of range of motion at wrist. Problems were first noticed at age of 12 years. Family history was positive for disproportionate dwarfism in a brother and an uncle. She was born by normal spontaneous vaginal delivery, with normal developmental milestones. She had no prior surgeries, was not taking any medications, and had no known drug allergies. Physical examination revealed prominences around inferomedial aspect of bilateral knee and around both ankle joints. There was unilateral genu valgum deformity of right leg. Limb length discrepancy of 2 cm noted in left leg with Galeazzi sign positive for left femoral shortening. There was no contracture at hip or knee along with normal range of movement at hip, knee and ankle. There was varus deformity, shortening and dorso-radial bowing deformity of the left forearm with flexion contracture of little finger. There was 20° to 130° flexion and extension ROM at left elbow with 10° supination and 0° pronation. The contra lateral (right) elbow had ROM from 0° to 130° of flexion-extension with 60° supination and pronation. There were no other areas of pain or deformity in the upper extremities. Antero-posterior and lateral radiographs of the left forearm were taken at first visit (Fig-A).

A distal ulnar exostosis was visualized projecting over the region of the interosseous membrane on both views. There were associated ulnar shortening and radial bowing. In addition, posterolateral radial head subluxation was seen. Given the natural history of HME and the position of the distal ulnar exostosis, it was presumed that this lesion contributed to deformity of left forearm and resulted in restricted supination and pronation. Complete skeletal survey was done which showed multiple exostosis (Fig-B & C).

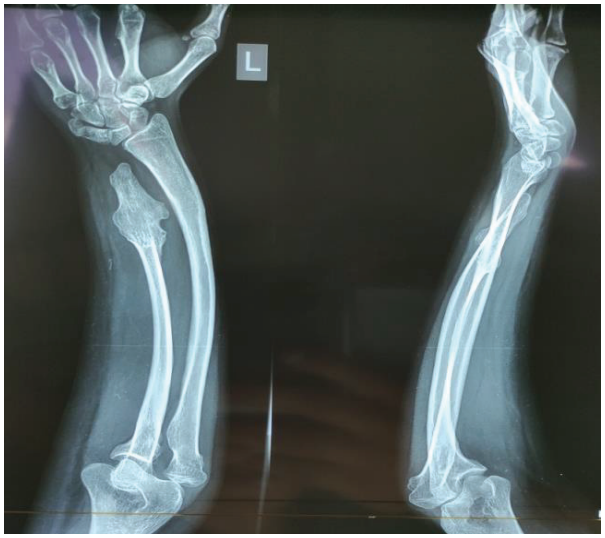

Fig-A: X-ray Left Radius/Ulna AP and Lat view showing distal ulnar Exostosis, radial bowing and radiocapitellar subluxation

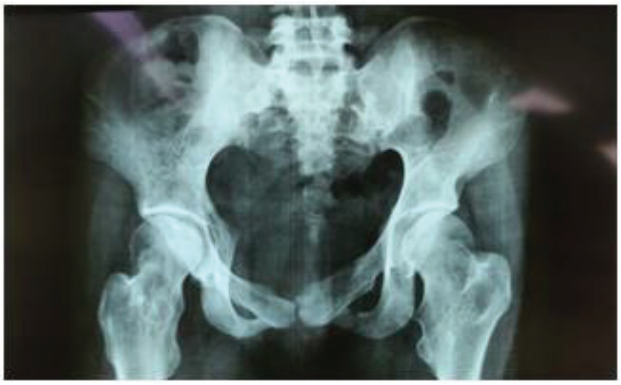

Fig-B:X-ray PELVIS showing multiple Osteochondromas

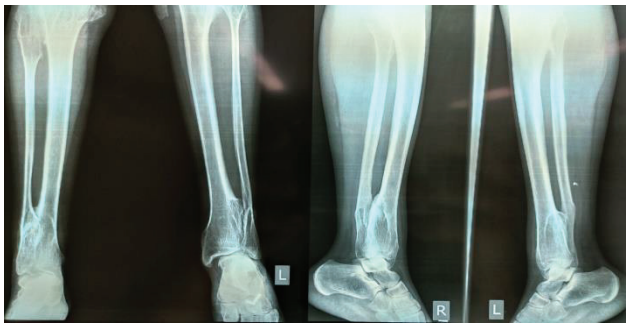

Fig-C: X-ray B/L Tibia/Fibula showing multiple exostoses

The renal anomly scan and echocardiogram were normal. Patient was operated and excision of exostosis of distal ulna was done (Fig-D) with release of interosseous membrane. Per operative assessment revealed improvement in supintion and pronation range of movement. Postoperatively, patient was followed up in OPD and at 2nd week follow up forearm range of movement was from 25° supination and 15° pronation.

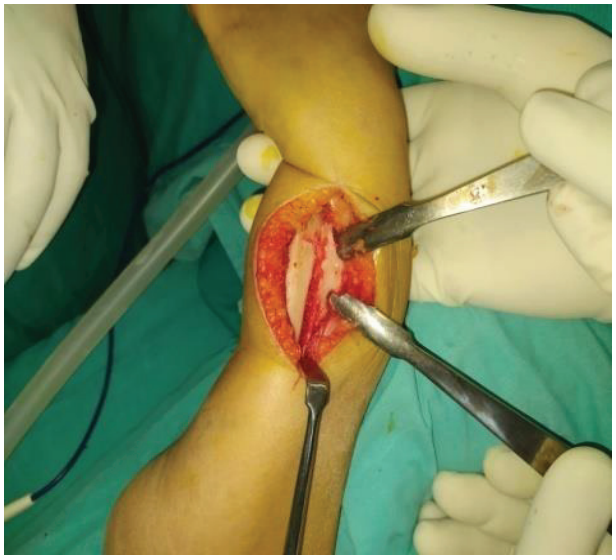

Fig-D: Per op pic showing distal ulnar exostosis

DISCUSSION

Hereditary multiple exostoses is a rare skeletal dysplasia and mutations in exostosin genes are implicated in pathogens namely, *EXT1* (8q23-q24), *EXT2* (11p11-p13) and in rare cases, *EXT3* (on chromosome 19p). These mutations lead to dysregulation of cell surface heparin sulfate proteoglycans and consequently cartilage hyperproliferation.[4] Exostoses of upper extremities may result in forearm deformities such as shortened ulna with secondary disproportionate radius bone overgrowth been frequently noticed ultimately leading to radial bowing deformity.[5] These lesions originate from perichondrium and flank the growth plate so these lesion tether the physis results in off balance growth of the bone. Radiocapitellar subluxation or dislocation is common over time complication in the forearm deformities in context of hereditary multiple exostosis. Dislocation of radial head has been associated with functional impairment such as loss of pronation and greater ulnar variance.[6]

Masada et al.[7] devised three types morphological classification for hereditary multiple exostoses linked forearm deformities. In type I, there is shortening of ulna secondary to distal ulnar exostosis and bowing of the radial bone. The radial head is not dislocated or subluxed. In type II, there is radiocapitellar dislocation and shortened ulna. Radial bone bowing is present but in milder form as compared to type I deformity. In type IIa, the exostosis involves the proximal metaphyseal segment of radius as a result of which radial head is completely dislocated. In type IIb, there is no exostosis involving proximal metaphysis of radius but still radial head dislocation happens. In type III, there is disproportionate radial shortening relative to

ulna due exostosis involving metaphysis of distal radius. He advocated to go for radial osteotomy simple exostosis excision and ulnar lengthening in type 1 deformity all thanks to his successful results in limited patients.

Controversy exists regarding the timing of surgical intervention. Fogel et al.[8] reported that the progression of forearm deformity comes to rest by simply excising exostosis in early age but it does not permanently provides complete correction. Matsubara H et al.[9] suggested that Intervention should be performed older age after skeletal maturity as in this way a revision surgery could be evaded and satisfactory function could be obtained despite significant deformity.

Shin et al.[10] assessed the clinical results of three different procedures Sauvé Kapandji, ulnar lengthening and simple excision for addressing forearm deformities manifested in patients of hereditary multiple exostoses. He reported that the mean supination improved by 11.8 by simple excision. No significant functional and clinical outcomes were attained from ulnar distraction lengthening procedure. Results were consistent with previous studies showing that just going for simple excision was effective in improving forearm rotational range of movement.

CONCLUSION

Based on the results of this case presentation, we conclude that just performing simple exostosis excision can enhance the range of movement of the forearm in skeletally mature patients.

REFERENCES

1. G. A. Schmale, E. U. Conrad 3rd, and W. H. Raskind, "The natural history of hereditary multiple exostoses", The Journal of Bone & Joint Surgery, vol. 76, no. 7, pp. 986–992, 1994.
2. Stieber, Jonathan R. MD; Dormans, John P. MD Manifestations of Hereditary Multiple Exostoses, Journal of the American Academy of Orthopaedic Surgeons: March 2005 - Volume 13 - Issue 2 - p 110-120.
3. Porter DE, Lonie L, Fraser M, et al. Severity of disease and risk of malignant change in hereditary multiple exostoses. A genotype phenotype study. J Bone Joint Surg Br. 2004; 86:1041e6.
4. Alvarez CM, De Vera MA, Heslip TR, Casey B. Evaluation of the anatomic burden of patients with hereditary multiple exostoses. Clin Orthop. 2007; (462):73-79.
5. Stieber JR, Pierz KA, Dormans JP. Hereditary multiple exostosis: a current understanding of clinical and genetic advances. Spring 2001;14:39-48.
6. 32. Masada K, Tsuyuguchi Y, Kawai H, Kawabata H, Noguchi K, Ono K: Operations for forearm deformity caused by multiple osteochondromas. J Bone Joint Surg Br 1989; 71:24-29.
7. Jo AR, Jung ST, Kim MS, Oh CS, Min BJ. An Evaluation of Forearm Deformities in Hereditary Multiple Exostoses: Factors Associated With Radial Head Dislocation and Comprehensive Classification. J Hand Surg Am. 2017 Apr; 42(4):292.e1-292.e8.
8. Fogel GR, McElfresh EC, Peterson HA, Wicklund PT: Management of deformities of the forearm in multiple hereditary osteochondromas. J Bone Joint Surg Am 1984; 66:670-680.
9. Matsubara H, Tsuchiya H, Sakurakichi K, Yamashiro T, Watanabe K, Tomita K (2006). Correction and lengthening for deformities of the forearm in multiple cartilaginous exostoses. J Orthop Sci. 11(5): 459-66.
10. Shin EK, Jones NF, Lawrence JF (2006). Treatment of multiple hereditary osteochondromas of the forearm in children: a study of surgical procedures. J Bone Joint Surg Br. 88(2): 255-60.

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