Case Report

Diaphyseal Aclasis — A case report

Soumik Das[1], Debalina Sarkar[2], Avishek Bhadra[3]

Diaphyseal aclasis is a rare genetic skeletal condition due to developmental abnormalities of the growth plate causing multiple cartilage covered exostoses to form on the surface of the metaphysis or the adjacent diaphysis region of long bones. A diagnosis of diaphyseal aclasis can be made when radiologically at least two osteochondromas of the juxta-epiphyseal region of long bones are observed. Radiological examination of the case revealed multiple exostoses involving the metaphysis of proximal end of both humeri, medial border of left scapula, distal end of both radii and both ulnae, distal end of both femora, proximal end of both tibiae and both fibulae, and distal end of right tibia and right fibula associated with shortening of the 4th and 5th metacarpals of left hand. A diagnosis of diaphyseal aclasis was made. No treatment was recommended at that time and the relatives of the patient were counselled regarding the future potential complications and what actions may be taken.

[*J Indian Med Assoc* 2019; **117(12):** 22-3]

Key words **: Diaphyseal aclasis, exostosis, osteochondroma.**

Diaphyseal aclasis, also known as hereditary multiple exostoses or osteochondromatosis is a rare autosomal dominant condition, characterized by the development of multiple osteochondromas. Here we present a classical case of diaphyseal aclasis in a 14-year-old boy who presented with swelling of the wrist and knee, which had appeared at the age of 2 years, and since then had slowly increased in size, associated with asymmetry in the length of the upper limbs. The diagnosis was confirmed on radiological examination.

Case Report

A 14-year-old boy presented with swelling of the wrist and knee, which had appeared at the age of 2 years, and had slowly increased in size, associated with asymmetry in the length of the upper limbs. The patient did not complain of any pain or paraesthesia distally. There was no history of fracture or pulsatile mass in the popliteal fossa.

Family history — A similar condition was present in his mother but these swellings had not shown any increase in size since she stopped growing.

Examinations — Examination of the patient revealed a normally developed child with short stature (height – 141 cm. which is below the 3rd percentile line representing height-for-age of boys between 5 to 19 years as per WHO growth chart 2007) and multiple, non-tender and non-mobile bony swellings over both knee, both wrists, and proximal end of both humeri. There was no evidence of any neuro-vascular deficit distal to the swellings.

Radiological examination revealed multiple exostoses involving the metaphysis of proximal end of both humeri, medial border of left scapula, distal end of both radii and both ulnae, distal end of both femora, proximal end of both tibiae and both fibulae, and distal end of right tibia and right fibula associated with shortening of the 4th and 5th metacarpals of left hand.

A diagnosis of diaphyseal aclasis was made. No treatment was recommended at that time and the relatives of the patient were counselled regarding the future potential complications and what actions may be taken.

Discussion

Hereditary Multiple Exostoses (HME), alternatively called diaphyseal aclasis or osteochondromatosis, is a highly penetrant, autosomal dominant trait characterized by slightly stunted growth of long bones and multiple osteochondromas[1].

Osteochondroma (osteocartilaginous exostosis), according to the 2002 WHO definition, is a cartilage capped benign bony neoplasm on the outer surface of bones containing a marrow cavity that is continuous with that of the underlying bone[2]. A diagnosis of diaphyseal aclasis can be made when radiologically at least two osteochondromas of the juxta-epiphyseal region of long bones are observed.

The prevalence of diaphyseal aclasis is estimated at 1:50,000 persons within the general population and seems to be higher in males (male-to-female ratio 1.5:1). Approximately 62% of the patients with multiple osteochondromas have a positive family history[1].

Two genes, EXT1 and EXT2 located respectively at 8q24 and 11p11-p12, have been isolated to cause diaphyseal aclasis. In osteochondromas in which EXT expression is decreased due to mutation or deletion, the heparan sulphate proteoglycans seem to accumulate in the cytoplasm of the cell, instead of being transported to be expressed at the cell surface, resulting in abnormal proliferation of growth plate cartilage.

The chief complaint is the discovery of single or multiple hard, painless masses near joints. Osteochondromas develop and increase in size in the first decade of life, ceasing to grow when the growth plates close at puberty but may recur during pregnancy. The

Department of Radiodiagnosis, Medical College & Hospital, Kolkata 700073

[1]MBBS, Postgraduate Trainee and Corresponding Author

[2]MBBS, Postgraduate Trainee, Department of Paediatrics, Medical College & Hospital, Kolkata 700073

[3]MBBS, MS (Obstet & Gynaecol), RMO-*cum*-Clinical Tutor, Department of Obstetrics and Gynaecology, Calcutta National Medical College, Kolkata 700014

distribution is usually bilateral and may be symmetrical.

In diaphyseal aclasis patients a variety of orthopaedic deformities can be found like deformities of the forearm (shortening of the ulna with secondary bowing of radius) giving the characteristic "bayonet hand" deformity, inequality in limb length, varus or valgus angulation of the knee, deformity of the ankle and disproportionate short stature. Complications include osseous and cosmetic deformities, bursa formation, arthritis and impingement on adjacent tendons, nerves, vessels or spinal cord.

Malignant degeneration occurs in 5%-25% of cases into chondrosarcoma[3,4]. The most common sites are the pelvis and shoulder girdle[5].

Vascular complications of the popliteal vessel occur due to ossification of the previously protective cartilage cap, around the 2nd decade and relative immobility of the artery in the popliteal fossa between exiting Hunter's canal and the trifurcation[6]. The artery undergoes chronic abrasion on the sharp exostosis forming a defect in the adventitia resulting in pseudoaneurysm formation[7].

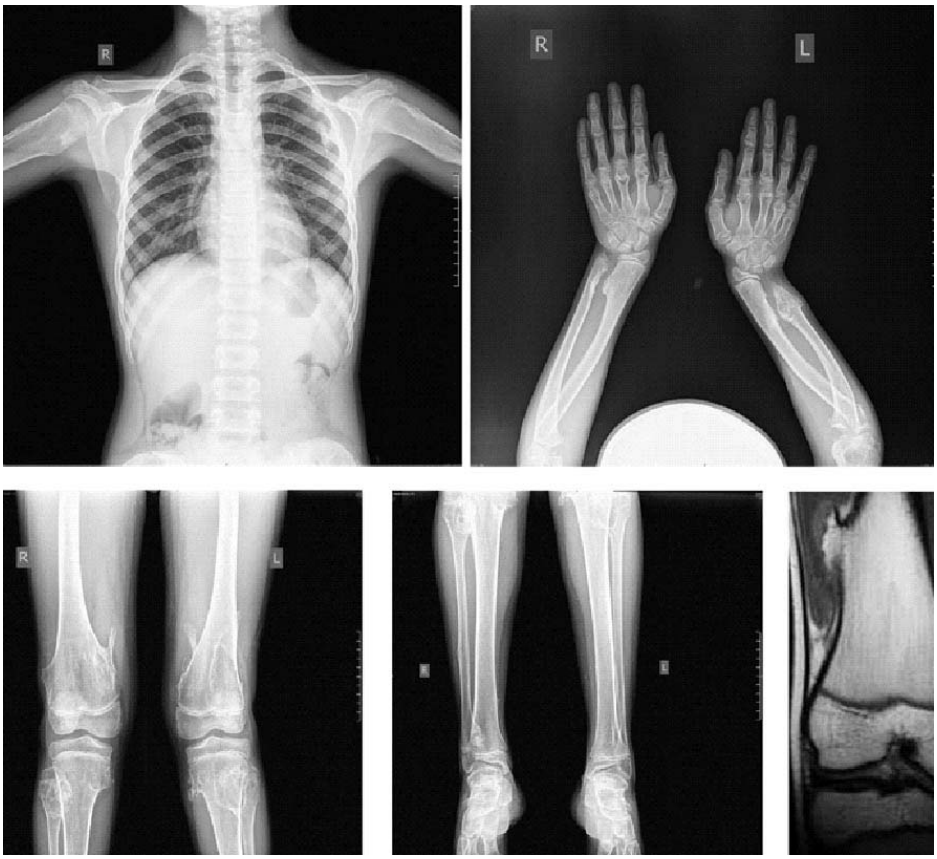

Fig 1 — Showing multiple exostoses involving the metaphysis of proximal end of both humeri, medial border of left scapula, distal end of both radii and both ulnae, distal end of both femora, proximal end of both tibiae and both fibulae, and distal end of right tibia and right fibula associated with shortening of the 4th and 5th metacarpals of left hand

Osteochondromas are only removed when they cause pain, when they give functional complaints for instance due to compression on nerves or vessels, or for cosmetic reasons. If the diagnosis of multiple osteochondromas is established and all tumours are identified, patients should be well instructed to seek earlier medical attention if their condition changes, for instance if there is pain or growth of a known lesion. In case of malignancy, en-bloc resection of the lesion and its pseudocapsule with tumour-free margins, preferably in a bone tumour referral centre, should be performed, resulting in excellent long term clinical and local results.

Osteochondromas are benign lesions and do not affect life expectancy.

Dysplasia Epiphysealis Hemimelica (DEH, Trevor's disease, tarso-epiphysial aclasis) and metachondromatosis (MC) are considered in the differential diagnosis of solitary and hereditary osteochondromas.

Moreover, diaphyseal aclasis should be distinguished from enchondromatosis (Ollier disease and Maffucci syndrome), in which multiple cartilage tumours are found in the medulla of bone, with a predilection for the short tubular bones and a unilateral predominance.

References

1 Legeai-Mallet L, Munnich A, Maroteaux P, Le Merrer M — Incomplete penetrance and expressivity skewing in hereditary multiple exostoses. *Clin Genet* 1997, **52(1):** 12-6.

2 Khurana J, Abdul-Karim F, Bovee JVMG — Osteochondroma. In: Fletcher CDM, Unni KK, Mertens F (eds) In World Health Organization classification of tumours. Pathology and genetics of tumours of soft tissue and bone. Lyon: IARC Press; 2002. 234-6.

3 Du YK, Shih HN, Wang JM, Shih CH — Dedifferentiated chondrosarcoma arising from osteochondromatosis. A case report. *Chang Keng i Hsueh-Chang Gung Med J* 1991; **14(2):** 130-5.

4 Flatt AE — Chondrosarcoma supervening on diaphyseal aclasis. *Br J Surg* 1955; **43(177):** 85-7.

5 Garrison RC, Unni KK, McLeod RA, Pritchard DJ, Dahlin DC — Chondrosarcoma arising in osteochondroma. *Cancer* 1982; **49(9):** 1890-7.

6 Greenway G, Resnick D, Bookstein JJ — Popliteal pseudoaneurysm as a complication of an adjacent osteochondroma: angiographic diagnosis. *AJR Am J Roentgenol* 1979; **132(2):** 294-6.

7 Matsushita M, Nishikimi N, Sakurai T, Nimura Y — Pseudoaneurysm of the popliteal artery caused by exostosis of the femur: case report and review of the literature. *J Vasc Surg* 2000; **32(1):** 201-4.