Diaphyseal Aclasis

Synonym: hereditary multiple exostoses

An inherited autosomal dominant disorder in which multiple osteochondromas are seen throughout the skeleton. The long bones of arms and legs are most commonly affected. It is associated with short stature and asymmetrical growth at the knees and ankles, which may lead to deformities.

Epidemiology

- Affects 2 per 100,000 population.
- More frequently found in Caucasians than in other races.

Risk factors

- Inheritance is autosomal dominant, with near-complete penetrance.
- It is genetically heterogeneous and has been associated with mutations in at least three different genes called EXT genes.

Presentation

- Usually presents during the first decade of life.
- The number of multiple osteochondromas varies from two to hundreds. The number of osteochondromas may vary significantly within and between families, with the mean number of locations being about 15-18.
- Bones may be broad and badly modelled.
- Most of the osteochondromas are located at the periphery of the most rapidly growing ends of long bones, but they also often involve the medial borders of the scapulae, ribs and iliac crests.
- Osteochondromas tend to grow while the growth plates are open, but growth ceases with skeletal maturity.
- Most of the osteochondromas are painless, and the main concern is often cosmetic. Pain may follow trauma to surrounding soft tissues or may be due to malignant transformation.
- Impaired body growth (symmetrical and asymmetrical) is common and results in:
  - Short stature.
  - Limb-length discrepancies.
  - Valgus deformities of the knee and ankle.
  - Asymmetry of the pectoral and pelvic girdles.
  - Bowing of the radius with ulnar deviation of the wrist.
  - Shortening of the metacarpals and finger phalanges.
  - Subluxation of the radial head.
  - Symptoms of peripheral nerve compression.

Differential diagnosis

- Dysplasia epiphysealis hemimelica (Trevor’s disease): osteochondromas arise in the epiphyses and involve the joint.
- Multiple epiphyseal dysplasia: autosomal dominant; irregular epiphyseal ossification with intracapsular or periarticular chondromas of the knees and ankles.
- Enchondromatosis (Ollier’s disease): intra-osseous benign cartilaginous tumours.
- Fetal alcohol syndrome.
- Turner's syndrome: associated with exostoses of tibia.
- Tuberous sclerosis: associated with exostoses of long bones.
- Radiation-induced osteochondroma.
- Trauma: fractures.

Investigations

- Plain X-ray - may be the only imaging study required.
- CT scan - useful in the assessment of osteochondromas in the pelvis, shoulder or spine.
- Ultrasound - useful in the assessment of complications associated with osteochondromas, eg thrombosis, aneurysm and bursitis.
MRI scan - useful in the assessment of malignant transformation, and for evaluating compression of the spinal cord, nerve roots and peripheral nerves.\[4\]
Arteriography - for demonstrating vascular occlusion, and aneurysm and pseudoaneurysm formation.

Associated diseases
Diaphyseal aclasis is associated with other genetic syndromes, such as Langer-Giedion syndrome, trichorhinophalangeal syndrome and DEFCETT 11 syndrome.

Management
Asymptomatic individuals may not require treatment.\[5\]

- Individual osteochondromas may grow large enough to require surgery.
- Surgical treatment for limb-length discrepancy.
- Corrective osteotomy for valgus deformity at the knee.
- Supramalleolar osteotomy of the tibia for severe valgus ankle deformity.
- Surgery for deformities of the forearm.
- Urgent surgery may be required for vascular ischaemia.
- Most associated chondrosarcomas can be treated with wide excision.

Complications
- Entrapment of tendons.
- Fractures.
- Bony deformities.
- May interfere with normal childbirth and lead to a higher rate of Caesarean deliveries.
- Neurological - compression of the spinal cord\[8\] nerve roots and peripheral nerves.
- Vascular injuries - arterial or venous thrombosis; aneurysms and false aneurysms may occur.
- Bursa formation - bursae may become inflamed and painful.
- Malignant transformation - estimated to occur in 1-5% of osteochondromas.\[4\]
- Internal injuries or obstruction have been reported, eg causing dysphagia, haemothorax, and both urinary and intestinal obstruction.

Prognosis
- The prognosis depends on the development of complications.
- Spontaneous resolution of osteochondromas during childhood and puberty is rare but has been described.

Further reading & references

1. Exostoses, Multiple, Type I, Online Mendelian Inheritance in Man (OMIM)
2. Multiple Cartilaginous Exostoses (Hereditary Exostosis), Wheeless’ Online Textbook of Orthopaedics
5. Gupta PP, Agarwal D; A middle-aged man with persisting chest opacity and multiple bony swellings. CMAJ. 2006 Nov 7;175(10):1206.

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