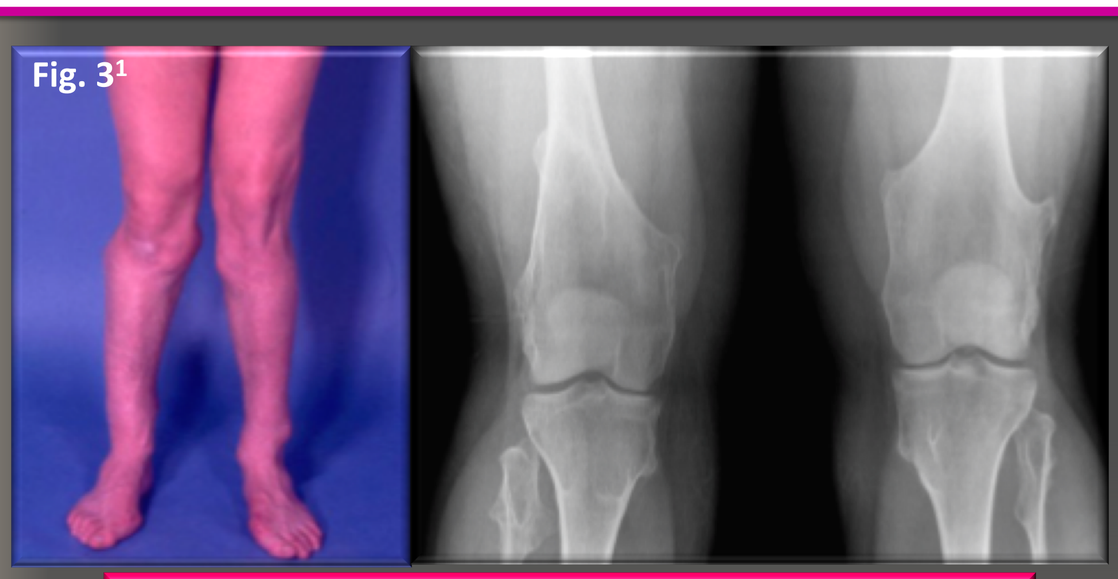
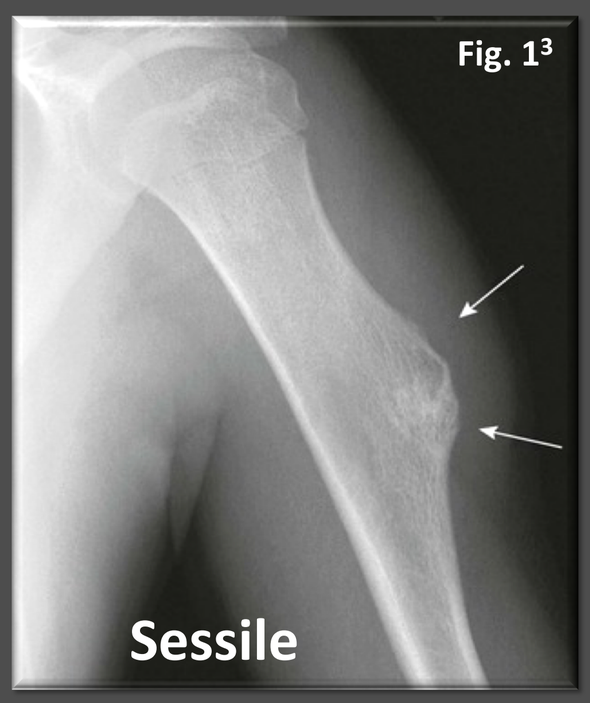
Diaphyseal Aclasis

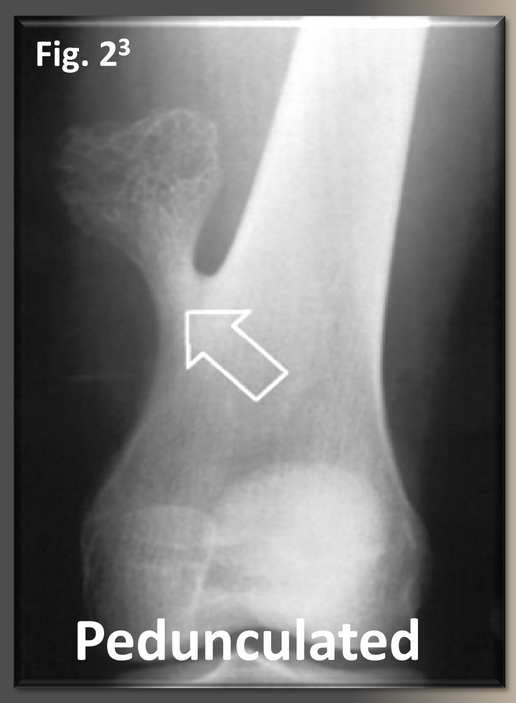
1. What is it?

Also known as Hereditary Multiple Exostoses, it is an inherited **genetic** (mutated EXT genes) disorder characterised by bone deformity and multiple **osteochondromas**[4]. These cartilage-capped bony outgrowths are commonly found at the metaphyseal region of long bones. They can be **sessile** (Fig.1) or **pedunculated** (Fig.2) and typically project away from the joint surface. They can be smooth or have a cauliflower-like appearance[2].



Multiple osteochondromas can be seen around the knee causing genu valgum (knock knee)





2. Who is affected?

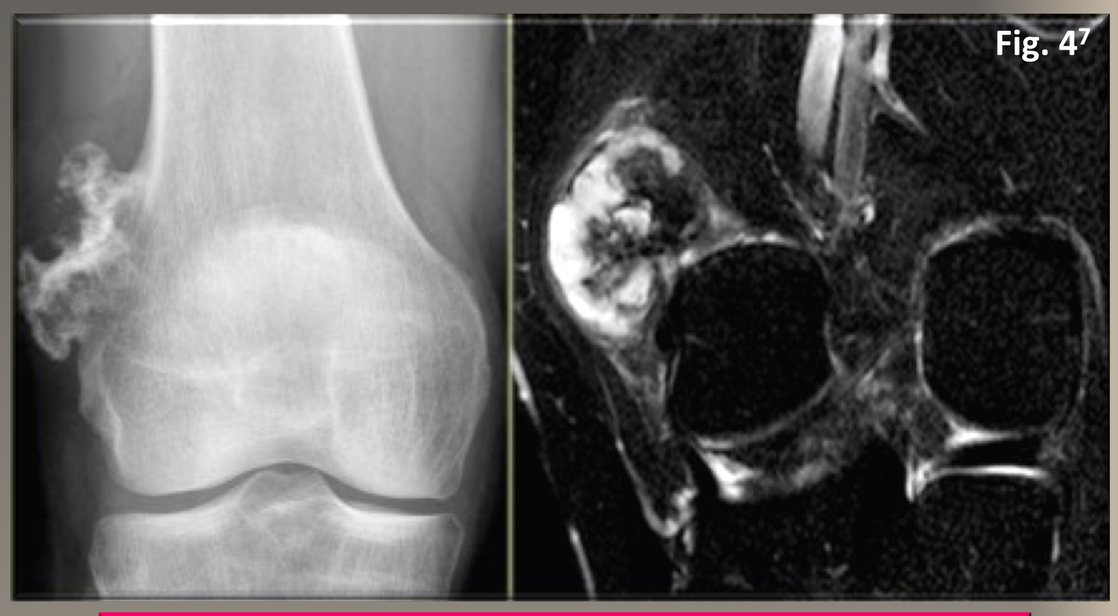
It **occurs in 1:50,000 people** and can run in families with slightly more boys diagnosed than girls[5] - although this has been more recently debated to have no gender predominance[6]. Nearly all sufferers are diagnosed by 10 years old[6]. Osteochondromas can grow anywhere except the skull and mandible, but most commonly occur around the **knee**[3]. They begin to grow as the bone growth plates form and stop growing when the growth plates close[1].

3. How is it diagnosed?

Osteochondromas are usually first discovered on the ribs and knees as these are the areas where they are most visible[6]. **Severity of symptoms often varies**, but observed manifestations are bone deformities, restricted movement of joints, slightly shorter stature and nerve compression[6]. Premature degenerative joint disease is also common due to the development pattern of the knees (Fig.3) and hips (coxa valga)[6].

Diaphyseal Aclasis is diagnosed clinically and radiologically using **plain film** but CT, MRI, US, PET and sequence analysis of EXT1 and EXT2 genes also aid confirmation[3]. Malignant transformation into **chondrosarcomas** (Fig.4) can occur in up to 5% of patients[4] and this occurrence is indicated by the thickness of the osteochondroma cartilage cap (>2cm) which is monitored using US or MRI[3].

Differential diagnoses include Trevor's Disease and Metachondromatosis, which produce similar symptoms but do not involve the EXT genes[1].



Osteochondroma viewed using plain film and T2W MRI. High signal from thick cartilage cap thought to have transformed into a chondrosarcoma

4. How is it treated?

Surgical intervention is required to ease joint deformities or due to limb length discrepancies. Osteochondroma excision is usually easy and used to relieve symptoms, cosmetic concerns or if they show signs of malignant transformation[1].

Diaphyseal Aclasis is a chronic condition but **prognosis is now good** due to regular monitoring in children to treat any growth disturbances as they occur, especially important in the spine to prevent neurological problems[6].

5. Summary

Diaphyseal Aclasis is a **complex chronic disorder** that requires paediatricians, geneticists, radiologists and orthopaedic surgeons to monitor and treat the condition for an optimal prognosis. More studies are required, but research has greatly expanded our knowledge regarding the disorder and future treatments[6].

References

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