Ollier's disease: features of the hands

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DESCRIPTION

A 14-year-old boy attended our services for orthopaedic evaluation due to the development of multiple enchondromatous lesions. Clinical examination of the hands revealed painless swollen digits of asymmetric lengths (figure 1). Plain film radiographs demonstrated extensive bilateral expansile lytic lesions of metacarpals and phalanges with diffuse matrix calcification resulting in cortical thinning, which is in keeping with Ollier's disease (figure 2). With no features concerning for malignancy in the hands, this patient is being followed up annually and remained symptom free at his last review.

Ollier's disease is defined as multiple enchondromas with asymmetric distribution that are variable in size, location and number.¹ These benign cartilage tumours originate in the metaphysis and integrate into diaphysis. With a prevalence of 1/100 000, Ollier's disease is twice as likely to occur in females. Multiple enchondromas with associated haemangiomas are seen in Maffucci syndrome. The pathogenesis remains unknown; however, both are linked to mutations in the *IDH1* and *IDH2* genes.² These enchondromas, while not apparent at birth, are typically painless and can result in short stature and limb length discrepancy. Malignant transformation typically to a chondrosarcoma is reported in up to 50% of cases. Rapid increase in size and pain are features concerning for malignancy.³ Radiographic features of affected limbs demonstrate multiple lytic

Figure 2 Plain radiography; anteroposterior of left and right hands depicting multiple radiolucent expansile lesions with cortical thinning of the metacarpal and phalanges.

lesions with cartilage calcification in a typical rings and arcs pattern within larger lesions.

Conservative treatment is the primary aim as functional impairment is usually not severe. Surgical intervention is reserved for cases of severe deformity, such as limb length discrepancy, pathological fracture and malignant transformation. Poor prognosis has been identified with early age of onset, malignant transformation and gross asymmetrical distribution.

Figure 1 Topographical view of a patient's hands with Ollier's disease.

Learning points

- ▶ Ollier's disease is a benign cartilaginous tumour associated with multiple enchondromas.
- ▶ Symptoms vary depending on limb involvement.
- ▶ Conservative treatment is the primary goal with poor prognosis seen in early age of onset and malignant transformation.

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