

Metachondromatosis

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Abstract

Metachondromatosis is a disorder with characteristic clinical and diagnostic radiographic features. The distinctive clinical feature is firm nodules in the hands and feet. The diagnostic radiographic features are exostoses at the metaphyses of the short tubular bones (hands and feet) pointing towards the joints coexisting with enchondromas of some large joints. The clinical course is unpredictable as there may be simultaneous growth of some of the exostoses and regression of others. Metachondromatosis is a rare autosomal dominant disorder of unknown pathogenesis. Antenatal diagnosis is unlikely. Orthopaedic intervention may be necessary.

Keywords

Metachondromatosis – exchondromatosis - enchondromatosis

Disease name/synonyms

Metachondromatosis

Definition/diagnostic criteria

Metachondromatosis is a rare exostosis disorder with characteristic clinical and diagnostic radiographic findings.

Differential diagnosis

In hereditary multiple exostoses (exchondromatosis), long bones are predominantly affected and the exostoses point away from the joints. In other exostosis disorders such as dysplasia epiphysealis hemimelica and tricho-rhino-phalangeal syndrome type II, there is a distinctive phenotype in addition to typical

exostoses. Among the disorders with enchondromatosis severe involvement of the hands and feet is observed only in Ollier disease, Maffucci syndrome and cheirospondyloenchondromatosis. In other enchondromatous bone disorders including genochondromatosis, dyspondyloenchondromatosis, spondyloenchondrodysplasia the long tubular bones are predominantly affected with little or no changes in the hands.

Etiology

Autosomal dominant inheritance.

Clinical description

Firm, non tender nodules adjacent to joints in the hands and feet, less frequently at the site of large joints, appearing during the first decade of life. The clinical course is usually benign but is unpredictable as some of the nodules may disappear spontaneously. On the other hand, nerve paralysis or vascular complications may occur (avascular necrosis of the femoral heads). Affected individuals may require surgery.

Skeletal survey reveals exostoses pointing towards the joints at the metaphyseal ends of the short tubular bones. There may be also small calcified masses separated from the metaphyses by a radiolucent zone, probably consisting of uncalcified cartilage. Exostosis at the metaphyses of the long bones, particularly in the knee region may be present. Enchondromas of the iliac crest and femoral neck are further useful radiographic clues to the diagnosis (Beals, 1982; Maroteaux *et al.*, 2002; Spranger *et al.*, 2002).

Diagnostic methods

Clinical examination may be suggestive. Skeletal survey is diagnostic.

Epidemiology

This is a rare disease. There is no geographical or racial preponderance.

Genetic counselling

Autosomal dominant inheritance.

Antenatal diagnosis

Unlikely as exostoses don't appear until postnatal life.

Management

Removal of the osteochondromas in hands and feet is necessary only in cases with severe malalignment of the fingers/toes. Orthopaedic supervision is necessary in patients with large, growing long bone exostoses (nerve and vascular compression), and in patients with femoral head involvement (Wenger *et al.*, 1991).

Unresolved questions

The genetic relationship of metachondromatosis to other exostosis and enchondromatosis disorders is uncertain.

References

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