Enchondromatosis

Twinkal C Pansuriya, Judith VMG Bovée

Dept of Pathology, Leiden University Medical Center, P.O. Box 9600, 2300 RC Leiden, The Netherlands (TCP, JVMGB)

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Identity

Alias: Multiple chondromatosis; Multiple enchondromatosis

Note
Most enchondromas and/or conventional central chondrosarcomas are solitary but some occur multiple in the context of a syndrome called enchondromatosis. It is rare and both sexes are equally affected. The enchondromatosis syndrome includes Ollier disease, Maffucci syndrome, spondyloenchondromatosis, metachondromatosis and generalized enchondromatosis.

In 1978 Spranger et al. summarized six different classes of enchondromatosis based on radiographic features. In 2005, Bhargava et al. further delineated some of the syndromes and distinguished non-hereditary and hereditary forms.

Inheritance
Ollier disease and Maffucci syndrome are non-inherited disorders while spondyloenchondromatosis is inherited as an autosomal recessive disorder.

<table>
<thead>
<tr>
<th>Type of Enchondromatosis</th>
<th>Etiology</th>
<th>Characteristic features</th>
<th>References</th>
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<tr>
<td>Ollier disease</td>
<td>Non-hereditary</td>
<td>Multiple enchondromas with a unilateral predominance, mainly affecting the small bones of the hands and feet, causing bone deformities</td>
<td>Spranger et al., 2002</td>
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<td>Maffucci syndrome</td>
<td>Non-hereditary</td>
<td>Multiple enchondromas combined with vascular lesions of the soft tissue</td>
<td>Lewis et al., 1973</td>
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<td>Metachondromatosis</td>
<td>Autosomal dominant</td>
<td>Combination of multiple enchondromas and osteochondroma-like lesions especially of the short bones of the hands, pointing towards the joint, no shortening of affected bones, spontaneous regression of osteochondroma-like lesions</td>
<td>Maroteaux et al., 1971</td>
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<tr>
<td>Spondyloenchondrodysplasia</td>
<td>Autosomal recessive?</td>
<td>Multiple enchondromas combined with spinal abnormalities (generalized platyspondyly), variable clinical features within and between the families, Types II is classic and IIa also includes cerebral calcifications</td>
<td>Schöber et al., 1976</td>
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<td>Dysplasia enchondromatosis (Enchondromatosis with irregular vertebral lesions)</td>
<td>not known</td>
<td>Multiple enchondromas (asymmetrically distributed in the long tubular bones), neonatal dwarfism, unequal limb length and severe segmentation abnormalities of vertebral column</td>
<td>Kostowski et al., 1964</td>
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<tr>
<td>Generalenchondromatosis</td>
<td>Autosomal dominant</td>
<td>Type I: Main feature is thickening of clavicles, Type II: Numerous short tubular bones are involved, Moderately severe hand involvement and more irregular long bones, embedded in sclerotic bone matrix, no cranial changes</td>
<td>Le Morrè et al., 1991</td>
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<td>Cheiro-spondyl-enchondromatosis</td>
<td></td>
<td>Flattened/abnormal vertebrae and massive enchondromas of metacarpals and phalanges, frequent mental retardation.</td>
<td>Spranger et al., 2002</td>
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</table>
However, there was a case reported by Robinson et al. which showed autosomal dominant inheritance of spondyloenchondrodysplasia. Metachondromatosis follows an autosomal dominant inheritance pattern. With the exception of Ollier disease, in which PTHR1 mutations are found in a very small subset of patients, the responsible genes for these extremely rare syndromes are so far unknown.

**Clinics**

**Note**
Clinical behaviour is determined by size, number, location and evolution of enchondromas, age of onset and of diagnosis. The diagnosis is mainly based on clinical, histological and radiological evaluation. Usually enchondromas are asymptomatic but in case of symptomatic enchondromas (pain, increase in size), further investigations could be indicated. The clinical features of enchondromatosis depend upon the extent of disease and ranges from few small lesions to multiple, widely distributed lesions causing marked skeletal deformation. Microscopically, the lesions can be more cellular and cytologically atypical as compared to solitary enchondroma. Macroscopic examination of enchondromas shows marked expansion and cortical attenuation in large bones. Radiographically, the lesions of enchondromatosis typically show multiple, radiolucent or mineralized homogeneous well defined lesions with oval or elongated shape.

**Phenotype and clinics**
There are several cases reported in which disease is limited to multifocal involvement of a single bone while in other cases widespread lesions and crippling deformation can be observed. The common site for development of enchondromas includes hand, foot, femur, humerus and forearm bones. Sometimes in case of severe condition, flat bones are also affected.

**Neoplastic risk**
There is an increased risk of development of malignant tumors. In Ollier disease and Maffucci syndrome 25-30% of cases undergo malignant transformation.

**Treatment**
Treatment depends on the type of enchondromatosis; it may include surgery, amputation, bone grafting and sclerotherapy.

**Prognosis**
The prognosis is dependent on the extent and severity of the disease. Cortical erosion, pathological fracture and extension of the tumor into soft tissues can be considered as a sign of malignancy.

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**Cytogenetics**

**Note**
Karyotypes of patients with Ollier disease or Maffucci syndrome are normal.

**References**


Banks RJ. Pathological fractures; a consideration with metachondromatosis and differential diagnoses. Osteochondromatosis and Gauchers disease. Australas Chiropr Osteopathy. 2002 Nov;10(2):105-10


This article should be referenced as such: