Atlas of Genetics and Cytogenetics in Oncology and Haematology

OPEN ACCESS JOURNAL AT INIST-CNRS

Cancer Prone Disease Section

Mini Review

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)

Published in Atlas Database: July 2008

Online updated version : http://AtlasGeneticsOncology.org/Kprones/EnchondromatosisID10151.html
DOI: 10.4267/2042/44527

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.

Type of Enchondromatosis	Etiology	Characteristic features	References
Ollier disease	Non-hereditary	multiple enchondromas with a unilateral predominance, mainly affecting the small bones of the hands and feet, causing severe deformities	Spranger et al., 2002
Maffucci syndrome	Non-hereditary	multiple enchondromas combined with vascular lesions of the soft tissue	Lewis et al., 1973
Metachondromatosis	autosomal dominant	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	Maroteaux et al., 1971
Spondyloenchondro dysplasia	autosomal recessive?	Multiple enchondromas combined with spinal aberrations (generalized platyspondyly). Variable clinical features within and between the families. Type I is classic and type II also includes cerebral calcifications	Schorr et al., 1976
Dysspondylo enchondromatosis (Enchondromatosis with irregular vertebral lesions)	not known	Multiple enchondromas (asymmetrically distributed in the long tubular bones), neonatal dwarfism, unequal limb length and severe segmentation abnormalities of vertebral column	Kozlowski et al., 1994
Genochondromatosis	autosomal dominant	Type I: Main feature is thickening of clavicles. Type II:Mainly short tubular bones are involved Moderately severe hand involvement and more irregular long, dense streaks and radiolucent channels can be seen. No spinal changes	Le Merrer et al., 1991
Cheirospondylo-enchondromatosis		Flattened/abnormal vertebrae and massive enchondromas of metacarpals and phalanges. Frequent mental retardation.	Spranger et al., 2002

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 wide spread 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.

Cytogenetics

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

References

Maroteaux P. [Metachondromatosis]. Z Kinderheilkd. 1971;109(3):246-61

Lewis RJ, Ketcham AS. Maffucci's syndrome: functional and neoplastic significance. Case report and review of the literature. J Bone Joint Surg Am. 1973 Oct;55(7):1465-79

Schorr S, Legum C, Ochshorn M. Spondyloenchondrodysplasia. Enchondromatomosis with severe platyspondyly in two brothers. Radiology. 1976 Jan;118(1):133-9

Spranger J, Kemperdieck H, Bakowski H, Opitz JM. Two peculiar types of enchondromatosis. Pediatr Radiol. 1978 Dec 4;7(4):215-9

Beals RK. Metachondromatosis. Clin Orthop Relat Res. 1982 Sep;(169):167-70

Kennedy LA. Metachondromatosis. Radiology. 1983 Jul;148(1):117-8

Bassett GS, Cowell HR. Metachondromatosis. Report of four cases. J Bone Joint Surg Am. 1985 Jun;67(5):811-4

Schwartz HS, Zimmerman NB, Simon MA, Wroble RR, Millar EA, Bonfiglio M. The malignant potential of enchondromatosis. J Bone Joint Surg Am. 1987 Feb;69(2):269-74

Paterson DC, Morris LL, Binns GF, Kozlowski K. Generalized enchondromatosis. A case report. J Bone Joint Surg Am. 1989 Jan;71(1):133-40

Le Merrer M, Fressinger P, Maroteaux P. Genochondromatosis. J Med Genet. 1991 Jul;28(7):485-9

Robinson D, Tieder M, Copeliovitch L, Halperin N. Spondyloenchondrodysplasia. A rare cause of short-trunk syndrome. Acta Orthop Scand. 1991 Aug;62(4):375-8

Freisinger P, Finidori G, Maroteaux P. Dysspondylochondromatosis. Am J Med Genet. 1993 Feb 15;45(4):460-4

Mulder JD, Schutte HE, Kroon HM, Takonis WK.. Radiological Atlas of Bone tumors. 1993, Amsterdam: Elsevier.

Haga N, Nakamura K, Taniguchi K, Nakamura S. Enchondromatosis with features of dysspondyloenchondromatosis and Maffucci syndrome. Clin Dysmorphol. 1998 Jan;7(1):65-8

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

Benbouazza K, El Hassani S, Hassikou H, Guedira N, Hajjaj-Hassouni N. Multiple enchondromatosis: a case report. Joint Bone Spine. 2002 Mar;69(2):236-9

Kozlowski KS, Masel J. Distinctive enchondromatosis with spine abnormality, regressive lesions, short stature, and coxa vara: importance of long-term follow-up. Am J Med Genet. 2002 Jan 22;107(3):227-32

Spranger JW, Brill PW, Poznanski A.. Bone Dysplasias: An Atlas of Genetic Disorders of Skeletal Development. Second edition, 2002, Oxford University Press.

Bhargava R, Leonard NJ, Chan AK, Spranger J. Autosomal dominant inheritance of spondyloenchondrodysplasia. Am J Med Genet A. 2005 Jun 15;135(3):282-8

This article should be referenced as such:

Pansuriya TC, Bovée JVMG. Enchondromatosis. Atlas Genet Cytogenet Oncol Haematol. 2009; 13(7):524-526.