Dysplasia Epiphysealis Hemimelica with Chondromas and Osteochondromas

Alternative Names
Osteochondromatosis, Dominant Carpotarsal
Dominant Carpotarsal Osteochondromatosis
DCO

Record Category
Disease phenotype

WHO-ICD
Neoplasms > Neoplasms of uncertain or unknown behavior

Incidence per 100,000 Live Births
0-1

OMIM Number
127820

Mode of Inheritance
Autosomal dominant

Gene Map Locus
N/A

Description
Dominant carpotarsal osteochondromatosis (DCO) is a hereditary skeletal developmental disorder, characterized by abnormal bone proliferation and osteochondromas arising from the epiphysis or epiphyseal-equivalent areas in the hands and feet. This is an extremely rare disorder, with only a handful of affected families having been reported around the world.

Molecular Genetics
The disorder is differentiated from the phenotypically similar sporadic dysplasia epiphysealis hemimelica by its hereditary nature. DCO is transmitted in an autosomal dominant manner. This differentiation is important, since genetic counseling would be very different for the two disorders.

Epidemiology in the Arab World
Kuwait
Fahmy and Pandey (2008) described a family in which three members were affected with a variant of DCO. The affected patients included a 10-year old girl, her father, and his cousin. The patients showed the presence of multiple parosteal osteochondromatous proliferations. This family had several significant differences from other previously reported families with DCO, including the absence of carpal or upper limb epiphyseal osteochondromas. Fahmy and Pandey (2008) speculated that the condition could either be a variant of DCO or a separate entity.

References
Fahmy AL, Pandey T. Epiphyseal osteochondromas with autosomal dominant inheritance and multiple parosteal bone proliferations. Skeletal Radiol. 2008; 37(1):67-70. PMID: 17938915

Related CTGA Records
N/A

External Links
N/A

Contributors
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