Opis choroby *

Definicja

Otopalatodigital syndrome spectrum disorder is a primary bone dysplasia and encompasses a group of congenital anomalies that are characterized by skeletal dysplasia of varying clinical severity and an X linked dominant pattern of inheritance. This group includes otopalatodigital syndrome type 1 and 2 (OPD1, OPD2) which are characterized in affected males by cleft palate, conductive hearing loss, craniofacial abnormalities and skeletal dysplasia; Melnick-Needles syndrome (MNS) which displays skeletal deformities in females and embryonic or perinatal lethality in most males; frontometaphyseal dysplasia (FMD); and terminal osseous dysplasia - pigmentary defects.

Dane

Klasyfikacja
Grupa fenomenów

Synonimy
OPD spectrum disorder
OPSD
Zaburzenia spektrum OPD
OPSD

Kod ORPHA
364541

Kod OMIM
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Kod ICD10
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Kod ICD11
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*Źródło

orphanet