

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

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|----------------------------|--|-----------------------|
| Klasyfikacja | Synonimy | |
| Grupa fenomenów | OPD spectrum disorder OPSD Zaburzenia spektrum OPD OPSD | |
| Kod ORPHA 364541 | Kod OMIM - | Kod ICD10 - |
| Kod ICD11 - | | |

[*Źródło](#)

orphanet