Opis choroby *

Definicja

The MMEP syndrome is a congenital syndromic form of split-hand/foot malformation (SHFM; see this term). It is characterized by microcephaly, microphthalmia, ectrodactyly of the lower limbs and prognathism. Intellectual deficit has been reported. MMEP syndrome is considered to be a very rare condition, although the exact prevalence remains unknown. The etiology is not completely understood. Disruption of the sorting nexin 3 gene (SNX3; 6q21) has been shown to play a causative role in MMEP, although this was not confirmed in recent studies.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
MCOPS8
Małogłowie - małoocze - ektrodaktylia kończyn dolnych - prognatyzm
MCOPS8
Syndromiczne małoocze typu 8 8
Zespół Viljoena i Smarta
Microcephaly-microphthalmia-ectrodactyly of lower limbs-prognathism syndrome
Syndromic microphthalmia type 8
Viljoen-Smart syndrome

Kod ORPHA
3434

Kod OMIM
601349

Kod ICD10
Q87.8

Kod ICD11
LD21.0

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*Źródło

orphanet