

## 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 MCOPS8

#### Synonimy

Małogłowie - małocze - ektrodaktylia kończyn dolnych - prognatyzm

MCOPS8

Syndromiczne małocze 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