

## Opis choroby \*

### Definicja

Colobomatous microphthalmia-rhizomelic dysplasia syndrome is a rare, genetic developmental defect during embryogenesis characterized by a range of developmental eye anomalies (including anophthalmia, microphthalmia, colobomas, microcornea, corectopia, cataract) and symmetric limb rhizomelia with short stature and contractures of large joints. Intellectual disability with autistic features, macrocephaly, dysmorphic features, urogenital anomalies (hypospadias, cryptorchidism), cutaneous syndactyly and precocious puberty may also be present.

### Dane

#### Klasyfikacja

Zespół wad wrodzonych

#### Synonimy

Microphthalmia-coloboma-rhizomelic skeletal dysplasia  
Małocze - szczelina oka - dysplazja rizmeliczna

#### Kod ORPHA

424099

#### Kod OMIM

615877

#### Kod ICD10

Q87.5

#### Kod ICD11

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#### \*Źródło

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