

## Opis choroby \*

### Definicja

Distal trisomy 7p is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the short arm of chromosome 7, with highly variable phenotype typically characterized by severe to profound psychomotor delay, intellectual disability, dysmorphic features (incl. dolichocephaly, microbrachycephaly, high and/or broad forehead, large anterior fontanel, hypertelorism, downslanting palpebral fissures, low-set, dysplastic ears, low, broad and prominent nasal bridge, abnormal palate, micro-/retrognathia), and hypotonia. Cardiovascular, gastrointestinal, skeletal and urogenital anomalies have commonly been reported.

### Dane

#### Klasyfikacja                      Synonimy

Zespół wad wrodzonych Distal duplication 7p

Duplikacja dystalna 7p

Duplikacja telomerowa 7p

Trisomia 7pter

Telomeric duplication 7p

Trisomy 7pter

#### Kod ORPHA

96074

#### Kod OMIM

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#### Kod ICD10

Q92.3

#### Kod ICD11

LD41.61

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#### \*Źródło

orphanet