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
Zespół wad wrodzonych

Synonimy
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