

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	Kod OMIM	Kod ICD10
96074	-	Q92.3

Kod ICD11
LD41.61

*Źródło

orphanet