

Opis choroby *

Definicja

Distal trisomy 6q is a rare chromosomal anomaly syndrome resulting from the partial duplication of the long arm of chromosome 6, with highly variable phenotype, typically characterized by growth and developmental delay, intellectual disability, craniofacial dysmorphism (microcephaly, flat facial profile, frontal bossing, hypertelorism, downward-slanting palpebral fissures, flat nasal bridge, anteverted nares, bow shaped mouth, micrognathia), short webbed neck and joint contractures. Cardiac, urogenital, ophthalmologic and hand and foot anomalies, as well as umbilical hernia, spasticity, and seizures, are other features that have been reported.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Distal duplication 6q

Duplikacja dystalna 6q

Duplikacja telomerowa 6q

Trisomia 6qter

Telomeric duplication 6q

Trisomy 6qter

Kod ORPHA

96098

Kod OMIM

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Kod ICD10

Q92.3

Kod ICD11

LD41.50

*Źródło

orphanet