

Opis choroby *

Definicja

Distal trisomy 4q is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the long arm of chromosome 4, with highly variable phenotype typically characterized by psychomotor delay, intellectual disability, craniofacial dysmorphism (microcephaly, low-set, prominent ears, downslanting palpebral fissures, hypertelorism, epicanthic folds, broad, prominent nasal bridge, high arched and cleft palate, micro-/retrognathia), seizures, as well as tooth and digital anomalies (clinodactyly, polydactyly). Cardiac malformations, renal anomalies, cryptorchidism, hypotonia and hearing impairment have also been reported.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Distal duplication 4q Duplikacja dystalna 4q Duplikacja telomerowa 4q Trisomia 4qter Telomeric duplication 4q Trisomy 4qter

Kod ORPHA	Kod OMIM	Kod ICD10
96096	-	Q92.3

Kod ICD11
LD41.30

*Źródło

orphanet