

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

96096

Kod OMIM

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Kod ICD10

Q92.3

Kod ICD11

LD41.30

*Źródło

orphanet