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
Zespół wad wrodzonych

Synonimy
Distal duplication 4q
Duplikacja dystalna 4q
Duplikacja telomerowa 4q
Trisomia 4qter
Telomeric duplication 4q
Trisomy 4qter

Kod ORPHA
96096

Kod OMIM
-

Kod ICD10
Q92.3

Kod ICD11
LD41.30

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*Źródło

orphanet