

Opis choroby *

Definicja

Distal trisomy 2q is a rare chromosomal anomaly, resulting from the partial duplication of the long arm of chromosome 2, characterized by moderate psychomotor delay, mild intellectual disability, facial dysmorphism (high hairline, prominent forehead, hypertelorism, upslanting palpebral fissures, large, low-set and/or posteriorly rotated ears, depressed/broad nasal bridge, prominent nasal tip, thin upper lip vermillion), clino-/camptodactyly and normal or increased body measurements. On occasion genital anomalies (hypospadias, cryptorchidism, shawl scrotum) and short stature may be observed.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Distal duplication 2q
Duplikacja telomerowa 2q
Dystalna duplikacja 2q
Trisomia 2qter
Telomeric duplication 2q
Trisomy 2qter

Kod ORPHA

96094

Kod OMIM

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Kod ICD10

Q92.3

Kod ICD11

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*Źródło

orphanet