

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	Synonimy
Zespół wad wrodzonych	Distal duplication 2q Duplikacja telomerowa 2q Dystalna duplikacja 2q Trisomia 2qter Telomeric duplication 2q Trisomy 2qter

Kod ORPHA	Kod OMIM	Kod ICD10
96094	-	Q92.3

Kod ICD11

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*Źródło

orphanet