

Opis choroby *

Definicja

Distal trisomy 19q is a rare chromosomal anomaly syndrome characterized by low birth weight, developmental delay, intellectual disability, short stature, craniofacial dysmorphism (incl. microcephaly, midface hypoplasia, hypertelorism, flat nasal bridge, ear anomalies, short philtrum, downturned corners of the mouth, micrognathia) and a short neck with redundant skin folds. Additional features may include hypotonia, skeletal anomalies (e.g. clino/camptodactyly), seizures and congenital cardiac, urogenital and gastrointestinal malformations.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Duplikacja dystalna 19q Duplikacja telomerowa 19q Trisomia 19qter Telomeric duplication 19q Trisomy 19qter Distal trisomy 19q

Kod ORPHA 1717	Kod OMIM -	Kod ICD10 Q92.3
Kod ICD11 LD41.J0		

[*Źródło](#)

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