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
Zespół wad wrodzonych

Synonimy
Duplikacja dystalna 19q
Duplikacja telomerowa 19q
Trisomia 19qter
Telomeric duplication 19q
Trisomy 19qter
Distal trisomy 19q

Kod ORPHA
1717

Kod OMIM
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Kod ICD10
Q92.3

Kod ICD11
LD41.J0

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*Źródło

orphanet