

## 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](#)

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