

## Opis choroby \*

### Definicja

Distal trisomy 9q is a rare chromosomal anomaly, resulting from the partial trisomy of the long arm of chromosome 9, with a variable phenotype mostly characterized by psychomotor and speech delay, intellectual disability, hypotonia, long narrow habitus, craniofacial dysmorphism (incl. micro/dolichocephaly, facial asymmetry, narrow palpebral fissures, deep-set eyes, strabismus, microphthalmia, abnormally shaped ears, microstomia, micro/retrognathia) and hand and feet anomalies (incl. arachnodactyly, camptodactyly, abnormal implantation of digits). Congenital flexion contractures and limited joint movements have also been observed.

### Dane

#### Klasyfikacja                      Synonimy

Zespół wad wrodzonych Distal duplication 9q  
Duplikacja dystalna 9q  
Duplikacja telomerowa 9q  
Trisomia 9qter  
Telomeric duplication 9q  
Trisomy 9qter

#### Kod ORPHA

96101

#### Kod OMIM

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#### Kod ICD10

Q92.3

#### Kod ICD11

LD41.80

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#### \*Źródło

orphanet