

## 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

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych	Distal duplication 9q Duplikacja dystalna 9q Duplikacja telomerowa 9q Trisomia 9qter Telomeric duplication 9q Trisomy 9qter

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
96101	-	Q92.3

**Kod ICD11**  
LD41.80

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### \*Źródło

orphanet