

## Opis choroby \*

### Definicja

Distal trisomy 6q is a rare chromosomal anomaly syndrome resulting from the partial duplication of the long arm of chromosome 6, with highly variable phenotype, typically characterized by growth and developmental delay, intellectual disability, craniofacial dysmorphism (microcephaly, flat facial profile, frontal bossing, hypertelorism, downward-slanting palpebral fissures, flat nasal bridge, anteverted nares, bow shaped mouth, micrognathia), short webbed neck and joint contractures. Cardiac, urogenital, ophthalmologic and hand and foot anomalies, as well as umbilical hernia, spasticity, and seizures, are other features that have been reported.

### Dane

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych	Distal duplication 6q Duplikacja dystalna 6q Duplikacja telomerowa 6q Trisomia 6qter Telomeric duplication 6q Trisomy 6qter

<b>Kod ORPHA</b> 96098	<b>Kod OMIM</b> -	<b>Kod ICD10</b> Q92.3
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**Kod ICD11**  
LD41.50

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### \*Źródło

orphanet