

## Opis choroby \*

### Definicja

Distal trisomy 8q is a rare chromosomal anomaly syndrome resulting from the partial duplication of the long arm of chromosome 8, with a highly variable phenotype, typically characterized by growth and developmental delay, intellectual disability, short stature, craniofacial dysmorphism (microcephaly, prominent forehead, hypertelorism, abnormal palpebral fissures, low-set, large ears, anteverted tip of nose, micro/retrognathia), congenital heart defects and skeletal and limb anomalies. Other reported features include ophthalmologic abnormalities (e.g. megalocornea), cryptorchidism, hypertrichosis, and neurologic manifestations (e.g. hypotonia, hearing loss, and seizures).

### Dane

#### Klasyfikacja

Zespół wad wrodzonych

#### Synonimy

Distal duplication 8q  
Duplikacja dystalna 8q  
Duplikacja telomerowa 8q  
Trisomia 8qter  
Telomeric duplication 8q  
Trisomy 8qter

#### Kod ORPHA

96100

#### Kod OMIM

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#### Kod ICD10

Q92.3

#### Kod ICD11

LD41.70

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#### \*Źródło

orphanet