## Opis choroby *

Definicja

Distal trisomy 5q is a rare chromosomal anomaly syndrome, resulting from a partial duplication of the long arm of chromosome 5, characterized by short stature, moderate intellectual disability, and craniofacial dysmorphism (microcephaly, flat facies, large, low-set dysplastic ears, down-slanted, almond-shaped palpebral fissures, hypertelorism, epicanthal folds, small nose, long philtrum, small mouth with thin upper lip, and micrognathia). Patients also frequently present speech and cognitive delay, cardiac (ventriculomegaly, ventricular septum defect) and skeletal abnormalities (craniosynostosis, radial agenesis, ulnar hypoplasia, brachydactyly) and genital malformations (hypospadias, cryptorchidism).

Dane

**Klasyfikacja**
Zespół wad wrodzonych

Synonimy
Distal duplication 5q
Duplikacja dystalna 5q
Duplikacja telomerowa 5q
Trisomia 5qter
Telomeric duplication 5q
Trisomy 5qter

**Kod ORPHA**
96097

**Kod OMIM**
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**Kod ICD10**
Q92.3

**Kod ICD11**
LD41.40

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*Źródło

orphanet