## Opis choroby *

Definicja

Distal trisomy 20q is a rare chromosomal anomaly syndrome, resulting from the partial trisomy of the long arm of chromosome 20, with high phenotypic variability mostly characterized by neurodevelopmental delay, cardiac malformations (e.g. ventricular septal defect, coarctation of aorta) and facial dysmorphism (incl. large/high forehead, microphthalmia, upslanting palpebral fissures, epicanthus, large, long, low-set ears, anteverted nares, protruding upper lip, cleft lip/palate, micro/retrognathia, dimpled chin). Skeletal (brachydactyly, scoliosis, pectus excavatum) and cerebral anomalies have also been reported.

Dane

**Klasyfikacja**
Zespół wad wrodzonych

Synonimy
Distal duplication 20q
Duplikacja dystalna 20q
Duplikacja telomerowa 20q
Trisomia 20qter
Telomeric duplication 20q
Trisomy 20qter

**Kod ORPHA**
96107

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

**Kod ICD11**
LD41.K0

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*Źródło

orphanet