## Opis choroby *

Definicja

Distal trisomy 13q is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the long arm of chromosome 13, with variable phenotype principally characterized by intellectual disability, psychomotor delay, craniofacial dysmorphism (incl. microcephaly, bushy eyebrows, long curled eyelashes, hypotelorism, low-set ears, prominent nasal bridge, long philtrum, high palate, thin upper lip), short neck, polydactyly, and hemangiomas. Cardiac, urogenital and neural tube defects, as well as umbilical and inguinal hernias, seizures and hypotonia, have also been reported.

Dane

**Klasyfikacja**
Zespół wad wrodzonych

Synonimy
Distal duplication 13q
Duplikacja dystalna 13q
Duplikacja telomerowa 13q
Trisomia 13qter
Telomeric duplication 13q
Trisomy 13qter

**Kod ORPHA**
96105

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

**Kod ICD11**
LD41.C

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*Źródło

orphanet