Opis choroby *

Definicja

Distal trisomy 16q is a rare chromosomal anomaly syndrome, resulting from the partial trisomy of the long arm of chromosome 16, with variable phenotype principally characterized by developmental delay, severe intellectual disability, hypotonia, facial dysmorphism (incl. high, prominent forehead, epicanthic folds, dysplastic ears, broad/depressed nasal bridge, malar hypoplasia, narrow and arched palate, thin upper lip vermilion, micrognathia) and hand/feet anomalies (e.g. arachnodactyly, talipes equinovarus). Cardiac defects, genitourinary malformations and vertebral anomalies are also associated. Thrombocytopenia and recurrent infections have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Distal duplication 16q
Duplikacja dystalna 16q
Duplikacja telomerowa 16q
Trisomia 16qter
Telomeric duplication 16q
Trisomy 16qter

Kod ORPHA
96106

Kod OMIM
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Kod ICD10
Q92.3

Kod ICD11
LD41.F0

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*Źródło

orphanet