Opis choroby *

Definicja

Distal trisomy 17q is a rare chromosomal anomaly syndrome with variable phenotype principally characterized by intellectual disability, developmental delay, short stature, craniofacial dysmorphism (incl. microcephaly, low posterior hairline, frontal bossing, bitemporal narrowing, low-set and malformed ears, flat nasal bridge, long philtrum, wide mouth with downturned corners, thin upper lip) and a short, webbed neck, as well as skeletal anomalies (e.g. brachyrhizomelia, poly-/syndactyly) and joint hyperlaxity. Cardiac, cerebral, and urogenital anomalies are also frequently associated.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Dystalna duplikacja 17q
Telomerowa duplikacja 17q
Trisomia 17qter
Telomeric duplication 17q
Trisomy 17qter
Distal trisomy 17q

Kod ORPHA
3379

Kod OMIM
-

Kod ICD10
Q92.3

Kod ICD11
-

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*Źródło

orphanet