Opis choroby *

Definicja

Distal trisomy 22q is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the long arm of chromosome 22, with variable phenotype principally characterized by varying degrees of intellectual disabilty and developmental delay, pre- and postnatal growth deficiency, hypotonia, and craniofacial dysmorphism (incl. microcephaly, hypertelorism, narrow and upslanted palpebral fissures, epicanthic folds, low-set dysplastic ears, broad and depressed nasal bridge, cleft lip an/or palate, long philtrum, retro/micrognathia). Congenital heart defects, as well as cerebral, skeletal, renal and genital anomalies, have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Distal duplication 22q
Duplikacja dystalna 22q
Duplikacja telomerowa 22q
Trisomia 22qter
Telomeric duplication 22q
Trisomy 22qter

Kod ORPHA
96109

Kod OMIM
-

Kod ICD10
Q92.3

Kod ICD11
LD41.M

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*Źródło

orphanet