

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 disability 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	Synonimy
Zespół wad wrodzonych	Distal duplication 22q Duplikacja dystalna 22q Duplikacja telomerowa 22q Trisomia 22qter Telomeric duplication 22q Trisomy 22qter

Kod ORPHA	Kod OMIM	Kod ICD10
96109	-	Q92.3

Kod ICD11
LD41.M

*Źródło

orphanet