Opis choroby *

Definicja

A rare chromosomal anomaly characterized by an extremely variable clinical phenotype and may include heart defects, urogenital abnormalities, velopharyngeal insufficiency with or without cleft palate, and ranging from multiple defects to mild learning difficulties with some individuals being essentially normal.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
22q11.2 microduplication syndrome
Dup(22)(q11)
Duplikacja 22q11.2
Trisomia 22q11.2
Dup(22)(q11)
Duplication 22q11.2
Trisomy 22q11.2

Kod ORPHA
1727

Kod OMIM
608363

Kod ICD10
Q92.3

Kod ICD11
LD41.M

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*Źródło

orphanet