

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 22q11.2 microduplication syndrome

Synonimy

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

*Źródło

orphanet