Opis choroby *

Definicja

A rare chromosomal anomaly syndrome, resulting from the partial duplication of the long arm of chromosome 22, with a highly variable phenotype principally characterized by developmental delay, intellectual disability, behavioral anomalies, and non-specific craniofacial dysmorphism. Congenital heart malformations, visual and hearing impairment, urogenital abnormalities, and seizures have also been reported. Penetrance is incomplete. In 70% of cases, the duplication is inherited from as asymptomatic parent.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Distal dup(22)(q11.2)
Dystalna dup(22)(q11.2)
Trisomia dystalna 22q11.2
Distal trisomy 22q11.2

Kod ORPHA
261337

Kod OMIM
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Kod ICD10
Q92.3

Kod ICD11
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*Źródło

orphanet