Opis choroby *

Definicja

8p23.1 duplication syndrome is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the short arm of chromosome 8, with a highly variable phenotype, principally characterized by mild to moderate developmental delay, intellectual disability, mild facial dysmorphism (incl. prominent forehead, arched eyebrows, broad nasal bridge, upturned nares, cleft lip and/or palate) and congenital cardiac anomalies (e.g., atrioventricular septal defect). Other reported features include macrocephaly, behavioral abnormalities (e.g., attention deficit disorder), seizures, hypotonia and ocular and digital anomalies (poly/syndactyly).

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Dup(8)(p23.1p23.1)
Dup(8)(p23.1)
Trisomia 8p23.1
Trisomy 8p23.1

Kod ORPHA
251076

Kod OMIM
-

Kod ICD10
Q92.3

Kod ICD11
LD41.71

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*Źródło

orphanet