Opis choroby *

Definicja

Trisomy 9p is a rare chromosomal anomaly syndrome, resulting from a partial or complete trisomy of the short arm of chromosome 9, with a wide phenotypic variablility, typically characterized by intellectual disability, craniofacial dysmorphism (e.g. microcephaly, large anterior fontanel, hypertelorism, strabismus, downslanting palpebral fissures, malformed, low-set, protruding ears, bulbous nose, macrostomia, down-turned corners of mouth, micrognathia), digital anomalies (brachydactyly and clinodactyly), and short stature. Less frequently patients present with cardiopathy and renal, skeletal, and central nervous system malformations.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Duplication 9p
Duplikacja 9p
Duplikacja krótkiego ramienia chromosomu 10
Trisomia krótkiego ramienia chromosomu 9
Duplication of the short arm of chromosome 9
Trisomy of the short arm of chromosome 9

Kod ORPHA
236

Kod OMIM
-

Kod ICD10
Q92.2

Kod ICD11
LD41.81

---

*Źródło

orphanet