

## 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 variability, 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

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#### Kod ICD10

Q92.2

#### Kod ICD11

LD41.81

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#### [\\*Źródło](#)

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