

## Opis choroby \*

### Definicja

Trisomy 4p is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the short arm of chromosome 4, with a highly variable phenotype, typically characterized by pre- and postnatal growth delay, psychomotor developmental delay and craniofacial dysmorphism (microcephaly, prominent glabella, hypertelorism, enlarged ears with abnormal helix and antihelix, bulbous nose with flat or depressed nasal bridge, long philtrum, retrognathia with pointed chin). Additional features include skeletal (rocker bottom feet, arachnodactyly, camptodactyly) and renal malformations, cardiac defects, ocular abnormalities and abnormal genitalia in males.

### Dane

#### Klasyfikacja

Zespół wad wrodzonych

#### Synonimy

Duplication 4p

Duplikacja 4p

Duplikacja krótkiego ramienia chromosomu 4

Trisomia krótkiego ramienia chromosomu 4

Duplication of the short arm of chromosome 4

Trisomy of the short arm of chromosome 4

#### Kod ORPHA

1738

#### Kod OMIM

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#### Kod ICD10

Q92.2

#### Kod ICD11

LD41.31

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#### \*Źródło

orphanet