

## Opis choroby \*

### Definicja

Distal monosomy 3p is a rare chromosomal anomaly syndrome, resulting from a partial deletion of the short arm of chromosome 3, with a highly variable phenotype typically characterized by pre- and post-natal growth retardation, intellectual disability, developmental delay and craniofacial dysmorphism (microcephaly, trigonocephaly, downslanting palpebral fissures, telecanthus, ptosis, micrognathia). Postaxial polydactyly, hypotonia, renal anomalies and congenital heart defects (e.g. atrioventricular septal defect) may be associated.

### Dane

#### Klasyfikacja

Zespół wad wrodzonych 3p- syndrome

3p- syndrome

Dystalna delecja 3p

Monosomia 3pter

Telomerowa monosomia 3p

Monosomy 3pter

Telomeric monosomy 3p

3p deletion syndrome

Distal monosomy 3p

#### Synonimy

#### Kod ORPHA

1620

#### Kod OMIM

613792

#### Kod ICD10

Q87.8

#### Kod ICD11

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