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

Synonimy
3p- syndrome
3p- syndrome
Dystalna delecja 3p
Monosomia 3pter
Telomerowa monosomia 3p
Monosomy 3pter
Telomeric monosomy 3p
3p deletion syndrome
Distal monosomy 3p

Kod ORPHA
1620

Kod OMIM
613792

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet