Opis choroby *

Definicja

Distal monosomy 9p is a rare chromosomal anomaly syndrome, resulting from a partial deletion of the short arm of chromosome 9, with a highly variable phenotype typically characterized by intellectual disability, craniofacial dysmorphism (trigonocephaly, upslanting palpebral fissures, hypoplastic supraorbital ridges), abnormal digits (long middle phalanges with short distal phalanges), as well as frequent association with genitourinary abnormalities (cryptorchidism, hypospadias, ambiguous genitalia, 46,XY testicular dysgenesis). Congenital hypothyroidism and cardiovascular defects have been reported in some cases. Patients present an increased risk for gonadoblastoma.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Dystalna delecja 9p
Monosomia 9pter
Monosomy 9pter
Telomeric deletion 9p
Distal monosomy 9p

Kod ORPHA
1642

Kod OMIM
-

Kod ICD10
Q93.5

Kod ICD11
-

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*Źródło

orphanet