Opis choroby *

Definicja

Distal trisomy 8q is a rare chromosomal anomaly syndrome resulting from the partial duplication of the long arm of chromosome 8, with a highly variable phenotype, typically characterized by growth and developmental delay, intellectual disability, short stature, craniofacial dysmorphism (microcephaly, prominent forehead, hypertelorism, abnormal palpebral fissures, low-set, large ears, anteverted tip of nose, micro/retrognathia), congenital heart defects and skeletal and limb anomalies. Other reported features include ophthalmologic abnormalities (e.g. megalocornea), cryptorchidism, hypertrichosis, and neurologic manifestations (e.g. hypotonia, hearing loss, and seizures).

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Distal duplication 8q
Duplikacja dystalna 8q
Duplikacja telomerowa 8q
Trisomia 8qter
Telomeric duplication 8q
Trisomy 8qter

Kod ORPHA
96100

Kod OMIM
-

Kod ICD10
Q92.3

Kod ICD11
LD41.70

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*Źródło

orphanet