

Opis choroby *

Definicja

Distal trisomy 1p36 is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the short arm of chromosome 1, characterized by borderline to mild intellectual disability, mild developmental delay, metopic craniosynostosis and mild craniofacial dysmorphism (incl. sloping forehead, bitemporal narrowing, blepharophimosis). Other associated abnormalities may include growth retardation, microcephaly, large hands, syndactyly, supernumerary ribs, rectal stenosis and/or anterior displacement of anus. Congenital heart malformations (e.g. atrial septal defect, patent ductus arteriosus) have also been reported.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Distal duplication 1p36

Duplikacja dystalna 1p36

Duplikacja telomerowa 1p36

Trisomia 1pter

Telomeric duplication 1p36

Trisomy 1pter

Kod ORPHA

96069

Kod OMIM

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Kod ICD10

Q92.3

Kod ICD11

LD41.01

*Źródło

orphanet