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. slopping 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

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*Źródło

orphanet