Opis choroby *

Definicja

Distal monosomy 13q is a rare chromosomal anomaly syndrome, resulting from a partial deletion of the long arm of chromosome 13, with a highly variable phenotype typically characterized by varying degrees of intellectual disability and developmental delay, as well as CNS malformations (e.g. holoprosencephaly, anencephaly, ventriculomegaly, Dandy-Walker malformation), ocular abnormalities (e.g. hypertelorism, microphthalmia, strabismus, aniridia, retinal dysplasia) and craniofacial dysmorphism (microcephaly, trigonocephaly, large and malformed ears, broad prominent nasal bridge, micrognathia). Cardiac, genitourinary, gastrointestinal and skeletal manifestations have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
13q32 deletion
Delecja 13q32
Dystalna delecja 13q
Monosomia 13q32
Telomerowa delecja 13q
Deletion 13q32
Monosomy 13q32
Distal monosomy 13q
Telomeric deletion 13q

Kod ORPHA
1590

Kod OMIM
602553

Kod ICD10
Q93.5

Kod ICD11
LD44.D

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*Źródło

orphanet