Opis choroby *

Definicja

Distal trisomy 11q is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the long arm of chromosome 11, with high phenotypic variability principally characterized by craniofacial dysmorphism (brachycephaly/plagiocephaly, low-set, posteriorly rotated ears, short philtrum, micrognathia) and intellectual disability. Short stature and seizures, as well as cardiac (e.g. atrial septal defect), skeletal (incl. brachy/syndactyly) and genital (e.g. micropenis, cryptorchidism) abnormalities may also be associated. Neurodevelopmental anomalies (pain insensitivity, sensorineural hearing loss, expressive language deficiency) and neuropsychiatric disorders (autistic features, auditory hallucination, self-talking) have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Distal duplication 11q
Duplikacja dystalna 11q
Duplikacja telomerowa 11q
Trisomia 11qter
Telomeric duplication 11q
Trisomy 11qter

Kod ORPHA
96103

Kod OMIM
-

Kod ICD10
Q92.3

Kod ICD11
-

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*Źródło

orphanet