

Opis choroby *

Definicja

16p11.2p12.2 microduplication syndrome is a rare chromosomal anomaly syndrome resulting from the partial duplication of the short arm of chromosome 16 with a highly variable phenotype typically characterized by developmental/psychomotor delay (particularly of speech), intellectual disability, autism spectrum disorder and/or obsessive and repetitive behavior, behavioral problems (such as aggression and outbursts), dysmorphic facial features (triangular face, deep set eyes, broad and prominent nasal bridge, upslanting or narrow palpebral features, hypertelorism). Additionally, finger/hand anomalies, short stature, microcephaly and slender build are frequently described.

Dane

Klasyfikacja

Zespół wad wrodzonych
Synonimy
Dup(16)(p11.2p12.2)
Dup(16)(p11.2p12.2)
Trisomia 16p11.2p12.2
Trisomy 16p11.2p12.2

Kod ORPHA

261204

Kod OMIM

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Kod ICD10

Q92.3

Kod ICD11

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*Źródło

orphanet