Opis choroby *

Definicja

16p13.3 microduplication syndrome is a rare chromosomal anomaly syndrome resulting from a partial duplication of the short arm of chromosome 16 and manifesting with a variable phenotype which is mostly characterized by: mild to moderate intellectual deficit and developmental delay (particularly speech), normal growth, short, proximally implanted thumbs and other hand and feet malformations (such as camptodactyly, syndactyly, club feet), mild arthrogryposis and characteristic facies (upslanting, narrow palpebral fissures, hypertelorism, mid face hypoplasia, bulbous nasal tip and low set ears). Other reported manifestations include cryptorchidism, inguinal hernia and behavioral problems.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Distal duplication 16p
Dup(16)(p13.3)
Duplikacja dystalna 16p
Duplikacja telomerowa 16p
Trisomia 16pter
Trisomia dystalna 16p
Distal trisomy 16p
Dup(16)(p13.3)
Telomeric duplication 16p
Trisomy 16pter

Kod ORPHA
96078

Kod OMIM
613458

Kod ICD10
Q92.3

Kod ICD11
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*Źródło

orphanet