Opis choroby *

Definicja

Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to 10p11.21p12.31 microdeletion is a rare, genetic syndromic intellectual disability characterized by developmental delay, hypotonia, speech delay, mild to moderate intellectual disability, abnormal behavior (autistic, aggressive, hyperactive) and dysmorphic facial features, including synophrys or thick eyebrows, deep set eyes, bulbous nasal tip and full cheeks. Congenital heart and brain anomalies, visual and hearing impairment are also common.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
10p12p11 microdeletion syndrome
Del(10)(p11.21p12.31)
Delecja 10p11.21p12.31
Monosomia 10p11.21p12.31
Zespół mikrodelecji 10p12p11
Del(10)(p11.21p12.31)
Deletion 10p11.21p12.31
Monosomy 10p11.21p12.31

Kod ORPHA
284169

Kod OMIM
616708

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet