

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	Synonimy
Podtyp kliniczny	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	Kod OMIM	Kod ICD10
284169	616708	Q87.8

Kod ICD11

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*Źródło

orphanet