

Zespół mikrodelecji 17q24.2

Kod Orpha: 529962 Kod OMIM:

Opis choroby *

Definicja

A rare, genetic, multiple congenital anomalies/dysmorphic features-intellectual disability syndrome characterized by developmental and speech delay, intellectual disability, feeding difficulties, failure to thrive, growth retardation, and associated malformations such as abnormality of fingers and toes (i.e. clinodactyly of the 5th finger, 2-3 toe syndactyly), microcephaly, heart defects, and upper airways anomalies. Observed facial dysmorphism includes hypertelorism, small, narrow or downslanting palpebral fissures, ptosis, epicanthus, ear malformations, broad nasal bridge, bulbous/prominent nose, short philtrum, thin lips, retrognathia/micrognathia, arched/cleft palate, and dental anomalies. Additional variable manifestations include hearing and visual impairment, seizures, joint anomalies, obesity, and behavioral/psychiatric disorders.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Del(17)(q24)
Del(17)(q24)

Kod ORPHA

529962

Kod OMIM

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Kod ICD10

Q93.5

Kod ICD11

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[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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