

Opis choroby *

Definicja

12q15q21.1 microdeletion syndrome is a rare chromosomal anomaly syndrome resulting from a partial deletion of the long arm of chromosome 12, with a highly variable phenotype, typically characterized by developmental delay, learning disability, intra-uterine and postnatal growth retardation, and mild facial dysmorphism that changes with age. Nasal speech and hypothyroidism are also associated.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Del(12)(q15)(q21.1)

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Delecja 12q15q21.1

Monosomia 12q15q21.1

Deletion 12q15q21.1

Monosomy 12q15q21.1

Kod ORPHA

289513

Kod OMIM

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Kod ICD10

Q93.5

Kod ICD11

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*Źródło

orphanet