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