

Opis choroby *

Definicja

14q11.2 microduplication syndrome is a rare chromosomal anomaly characterized by developmental delay, mild to severe intellectual disability with speech impairment and epilepsy. Additionally, it may include dysmorphic features (such as hypo- or hypertelorism, dysplastic ears, short palpebral fissures), microcephaly or macrocephaly, behavioral abnormalities, stereotyped hand movements, ataxia, hypotonia, cleft palate.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Dup(14)(q11.2)

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Trisomia 14q11.2

Trisomy 14q11.2

Kod ORPHA

261229

Kod OMIM

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Kod ICD10

Q92.3

Kod ICD11

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*Źródło

orphanet