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)
Dup(14)(q11.2)
Trisomia 14q11.2
Trisomy 14q11.2

Kod ORPHA
261229

Kod OMIM
-

Kod ICD10
Q92.3

Kod ICD11
-

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*Źródło

orphanet