Opis choroby *

Definicja

9p13 microdeletion syndrome is a rare chromosomal anomaly syndrome, resulting from a partial interstitial deletion of the short arm of chromosome 9, characterized by mild to moderate developmental delay, hand tremors, myoclonic jerks, attention deficit-hyperactivity disorder and a social personality. Patients also present bruxism, short stature and minor facial dysmorphic features (e.g., bilateral epicantic folds, broad, flat nasal bridge, anteverted nares, low-set ears micro/retro-gnathia).

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Del(9)(p13)
Del(9)(p13)
Monosomia 9p13
Monosomy 9p13

Kod ORPHA
324313

Kod OMIM
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Kod ICD10
Q93.5

Kod ICD11
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*Źródło

orphanet