

## 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 epicanthic 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