

Opis choroby *

Definicja

2q24 microdeletion syndrome is a chromosomal anomaly consisting of a partial long arm deletion of chromosome 2 and characterized clinically by a wide range of manifestations (depending on the specific region deleted) which can include seizures, microcephaly, dysmorphic features, cleft palate, eye abnormalities (coloboma, cataract and microphthalmia), growth retardation, failure to thrive, heart defects, limb anomalies, developmental delay and autism.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Del(2)(q24)

Del(2)(q24)

Monosomia 2q24

Monosomy 2q24

Kod ORPHA

1617

Kod OMIM

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Kod ICD10

Q93.5

Kod ICD11

LD44.20

*Źródło

orphanet