Opis choroby *

Definicja

17q12 microdeletion syndrome is a rare chromosomal anomaly syndrome resulting from the partial deletion of the long arm of chromosome 17 characterized by renal cystic disease, maturity onset diabetes of the young type 5, and neurodevelopmental disorders, such as cognitive impairment, developmental delay (particularly of speech), autistic traits and autism spectrum disorder. Müllerian aplasia in females, macrocephaly, mild facial dysmorphism (high forehead, deep set eyes and chubby cheeks) and transient hypercalcaemia have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Del(17)(q12)
Del(17)(q12)
Monosomia 17q12
Monosomy 17q12

Kod ORPHA
261265

Kod OMIM
614527

Kod ICD10
Q93.5

Kod ICD11
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*Źródło

orphanet