Opis choroby *

Definicja

Trisomy 1q is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the long arm of chromosome 1, with a highly variable phenotype principally characterized by intellectual disability, short stature, craniofacial dysmorphism (incl. macro/microcephaly, prominent forehead, posteriorly rotated, low-set ears, abnormal palpebral fissures, microphthalmia, broad, flat nasal bridge, high-arched palate, micro/retrognathia), cardiac defects and urogenital anomalies. Patients may also present cerebral (e.g. ventriculomegaly) and gastrointestinal malformations, as well as dystonic tremor and recurrent respiratory tract infections.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Duplication 1q
Duplikacja 1q

Kod ORPHA
261344

Kod OMIM
-

Kod ICD10
Q92.2

Kod ICD11
-

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*Źródło

orphanet