Opis choroby *

Definicja

A rare chromosomal anomaly syndrome resulting from the partial deletion of the long arm of chromosome 20 with a highly variable phenotype typically characterized by hypotonia, intellectual disability, cognitive and language deficits (including decreased or absent speech), pre and post-natal growth retardation, feeding difficulties, microcephaly, and malformed hands and feet. Neurodevelopmental disorders (including hyperactivity, social interactive problems and autism spectrum disorder), seizures and dysmorphic facial features (high forehead, hypertelorism, malformed ears, broad nasal bridge, bulbous nasal tip, thin upper lip, small chin) are frequently associated.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Del(20)(q13.33)
Del(20)(q13.33)
Monosomia 20q13.33
Monosomy 20q13.33

Kod ORPHA
261311

Kod OMIM
-

Kod ICD10
Q93.5

Kod ICD11
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*Źródło

orphanet