Opis choroby *

Definicja

A rare, genetic, chromosomal anomaly syndrome resulting from a partial deletion of the long arm of chromosome 21 characterized by pre- and post-natal growth delay, short stature, intellectual disability, developmental delay with severe language impairment, thrombocytopenia, and craniofacial dysmorphism which may include microcephaly, downslanted palpebral ﬁssures, low-set ears, broad nose, thin upper vermillion, and downturned corners of the mouth. Brain MRI abnormalities (such as agenesis of the corpus callosum), behavioral problems and seizures may be associated.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Del(21)(q22.11q22.12)
Del(21)(q22.11q22.12)
Monosomia 21q22.11q22.12
Monosomia 21q22.11-q22.12
Zespół mikrodelecji 21q22.11-q22.12
Monosomy 21q22.11q22.12

Kod ORPHA
261323

Kod OMIM
-

Kod ICD10
Q93.5

Kod ICD11
-

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*Źródło

orphanet