Opis choroby *

Definicja

A rare, syndromic intellectual disability characterized by global developmental delay including severely delayed or absent speech, moderate to severe intellectual disability, behavioral issues, stereotypic behavior, febrile seizures and epilepsy, abnormal gait, vision defects, and characteristic facial features. Intrauterine growth restriction and feeding difficulties are frequently present.

Dane

Klasyfikacja
Podtyp etiologiczny

Synonimy
21q22.13q22.2 microdeletion syndrome
Del(21)(q22.13q22.2)
Monosomia 21q22.13q22.2
Monosomia 21q22.13-q22.2
Zespół mikrodelecji 21q22.13q22.2
Zespół mikrodelecji 21q22.13-q22.2
Del(21)(q22.13q22.2)
Monosomy 21q22.13q22.2

Kod ORPHA
268261

Kod OMIM
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Kod ICD10
Q93.5

Kod ICD11
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*Źródło

orphanet