Opis choroby *

Definicja

A rare, genetic, syndromic intellectual disability characterized by psychomotor delay, hypotonia, feeding difficulties, failure to thrive, anomalies of the hands and feet (clinodactyly, camptodactyly, brachydactyly, feet malposition), and craniofacial dysmorphism. Associated prenatal growth retardation, and gastrointestinal, heart and eye anomalies have been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Del(20)(q11.2)
Del(20)(q11.2)
Monosomia 20q11
Monosomy 20q11

Kod ORPHA
444051

Kod OMIM
-

Kod ICD10
Q93.5

Kod ICD11
-

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*Źródło

orphanet