Opis choroby *

Definicja

20q11.2 microduplication syndrome is a rare chromosomal anomaly syndrome, due to partial duplication of the long arm of chromosome 20, characterized by psychomotor and developmental delay, moderate intellectual disability, metopic ridging/trigonocephaly, short hands and/or feet and distinctive facial features (epicanthus, hypoplastic supraorbital ridges, horizontal/downslanting palpebral fissures, small nose with depressed nasal bridge and anteverted nostrils, prominent cheeks, retrognathia and small, thick ears). Growth delay and cryptorchidism are often associated features.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Dup(20)(q11.2)
Dup(20)(q11.2)

Kod ORPHA
363659

Kod OMIM
-

Kod ICD10
Q93.5

Kod ICD11
-

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*Źródło

orphanet