Opis choroby *

Definicja

8q21.11 microdeletion syndrome encompasses heterozygous overlapping microdeletions on chromosome 8q21.11 resulting in intellectual disability, facial dysmorphism comprising a round face, ptosis, short philtrum, Cupid's bow and prominent low-set ears, nasal speech and mild finger and toe anomalies.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Del(8)(q21.11)
Del(8)(q21.11)
Delecja 8q21.11
Monosomia 8q21.11
Deletion 8q21.11
Monosomy 8q21.11

Kod ORPHA
284160

Kod OMIM
614230

Kod ICD10
Q93.5

Kod ICD11
LD44.80

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*Źródło

orphanet