Opis choroby *

Definicja

1q21.1 microdeletion syndrome is a newly described recurrent deletion syndrome with variable clinical manifestations but without the clinical picture of thrombocytopenia - absent radius (TAR) syndrome.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Del(1)(q21)
Del(1)(q21)
Monosomia 1q21.1
Monosomy 1q21.1

Kod ORPHA
250989

Kod OMIM
612474

Kod ICD10
Q93.5

Kod ICD11
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*Źródło

orphanet