Opis choroby *

Definicja

2q31.1 microdeletion syndrome is a well-defined and clinically recognisable syndrome characterized by moderate to severe developmental delay, short stature, facial dysmorphism and variable limb defects.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Del(2)(q31.1)
Del(2)(q31.1)
Monosomia 2q31.1
Monosomy 2q31.1

Kod ORPHA
251014

Kod OMIM
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Kod ICD10
Q93.5

Kod ICD11
LD44.20

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*Źródło

orphanet