Opis choroby *

Definicja

A rare, genetic syndromic intellectual disability characterized by developmental delay, mild to severe intellectual disability, facial features (bulbous nasal tip, and macroglossia, macrostomia, or open mouth appearance) and a wide spectrum of other nonspecific variable clinical features.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
MED13L-related intellectual disability syndrome
MED13L-related intellectual disability syndrome

Kod ORPHA
369891

Kod OMIM
616789

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet