

Opis choroby *

Definicja

14q12 microdeletion syndrome is a recently described syndrome characterized by severe intellectual deficit, with a normal neonatal period, followed by a phase of regression at the age of 3-6 months.

Dane

Klasyfikacja

Podtyp kliniczny

Synonimy

Del(14)(q12)
Del(14)(q12)
Monosomia 14q12
Monosomy 14q12

Kod ORPHA

261144

Kod OMIM

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Kod ICD10

Q93.5

Kod ICD11

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*Źródło

orphanet