

Opis choroby *

Definicja

The newly described 2q23.1 microdeletion syndrome includes severe intellectual deficit with pronounced speech delay, behavioral abnormalities including hyperactivity and inappropriate laughter, short stature and seizures.

Dane

Klasyfikacja

Zespół wad wrodzonych Del(2)(q23.1)

Synonimy

Del(2)(q23.1)

Monosomia 2q23.1

Rzekomy zespół Angelmana

Monosomy 2q23.1

Pseudo-Angelman syndrome

Kod ORPHA

228402

Kod OMIM

156200

Kod ICD10

Q93.5

Kod ICD11

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*Źródło

orphanet