

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	Synonimy
Zespół wad wrodzonych	Del(2)(q23.1)
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	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