

Opis choroby *

Definicja

A rare, genetic, neurodevelopmental disorder characterized by global developmental delay, borderline to severe intellectual disability, feeding difficulties, behavioral anomalies, vision anomalies and mild facial dysmorphism. Other associated features may include microcephaly, short stature, urogenital or palatal anomalies (e.g. cleft palate), minor cardiac defects, recurrent infections or hearing loss.

Dane

Klasyfikacja

Choroba

Kod ORPHA	Kod OMIM	Kod ICD10
363611	615502	Q87.8
Kod ICD11		
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*Źródło

orphanet