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
363611

Kod OMIM
615502

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet