

Opis choroby *

Definicja

A rare genetic syndromic intellectual disability characterized by global developmental delay, moderate to severe intellectual disability, motor and language impairment, behavioral abnormalities (with mood instability, aggression, and self-mutilation), and progressive hand tremor. Facial dysmorphism includes narrow palpebral fissures, large ears, long philtrum, and prominent chin.

Dane

Klasyfikacja

Choroba

Kod ORPHA

457212

Kod OMIM

616269

Kod ICD10

F78.1

Kod ICD11

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*Źródło

orphanet