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