Opis choroby *

Definicja

A rare genetic disease characterized by a variable clinical phenotype which includes similar features but is typically less severe than in affected males. Patients may present with mild to borderline intellectual disability, anxiety, social phobia, selective mutism, attention deficit hyperactivity disorder, language deficit, neurologic signs and symptoms (such as seizures, hypotonia, and clonus), ophthalmologic anomalies (strabismus, refractive errors), and facial dysmorphism (including long face, prominent forehead, large, prominent ears, and mandibular prognathism).

Dane

Klasyfikacja
Choroba

Kod ORPHA
449291

Kod OMIM
300624

Kod ICD10
Q99.2

Kod ICD11
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*Źródło

orphanet