Opis choroby *

Definicja

A rare X-linked syndromic intellectual disability characterized by intellectual disability of variable degree, behavioral anomalies (including autism, mood disorders, obsessive-compulsive behavior, and hetero- and auto-aggression), and epilepsy. Progressive neurological symptoms like movement disorders and spasticity, as well as subtle dysmorphic features have also been reported. Heterozygous females may be as severely affected as males.

Dane

Klasyfikacja
Choroba

Synonimy
Raynaud-Claes syndrome
Raynaud-Claes syndrome

Kod ORPHA
485350

Kod OMIM
300114

Kod ICD10
F78.1

Kod ICD11
LD90

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*Źródło

orphanet