Opis choroby *

Definicja

A rare, genetic, syndromic intellectual disability characterized by mild to severe intellectual disability associated with variable features, including hypotonia, dyskinesia, spasticity, wide-based gait, microcephaly, epilepsy and behavioral problems. MRI imaging may show a corpus callosum hypoplasia or ventricular enlargement. Other variable features, such as joint hyperlaxity, skin pigmentary abnormalities, and visual impairment, have also been reported.

Dane

Klasyfikacja
Choroba

Kod ORPHA
457260

Kod OMIM
300958

Kod ICD10
F78.8

Kod ICD11
LD90

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*Źródło

orphanet