

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

*Źródło

orphanet