Opis choroby *

Definicja

A rare, genetic, slowly progressive neurodegenerative disease characterized by delayed psychomotor development beginning in infancy, mild to profound intellectual disability, gait and stance ataxia, pyramidal signs (hyperreflexia, extensor plantar responses), dysarthria, and ocular abnormalities (e.g. nystagmus, oculomotor apraxia, abduction deficits, esotropia, ptosis). Brain imaging reveals progressive, generalized cerebellar atrophy, mild ventriculomegaly and, in some, retrocerebellar cysts.

Dane

Klasyfikacja
Choroba

Kod ORPHA
363429

Kod OMIM
616204

Kod ICD10
G11.1

Kod ICD11
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*Źródło

orphanet