Opis choroby *

Definicja

A rare genetic neurodegenerative disease characterized by neonatal to infantile onset of hypotonia, developmental delay, regression of motor skills with distal amyotrophy, ataxia, and spasticity, absent speech or dysarthria, and moderate to severe cognitive impairment. Optic atrophy may also be associated. Brain imaging shows cerebellar atrophy and thin corpus callosum, as well as brain iron accumulation in the pallidum and substantia nigra beginning during the second decade of life.

Dane

Klasyfikacja
Choroba

Kod ORPHA
496756

Kod OMIM
617207

Kod ICD10
G31.8

Kod ICD11
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*Źródło

orphanet