Opis choroby *

Definicja

A rare, genetic neurodegenerative disease characterized by childhood or adolescent-onset of cerebellar ataxia with dysarthria which slowly progresses and associates pyramidal signs, including lower limb spasticity, brisk reflexes, and Babinski and Hoffman signs. Patients typically present cerebellar ataxia with development of increasing asymmetric spasticity in upper and lower limbs, and variable axonal sensory or sensorimotor neuropathy. Additional heterogeneous features, including pes cavus, scoliolis, and abnormalities of the brain (e.g. cerebral atrophy), may also be associated.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive cerebellar ataxia due to GBA2 deficiency
Autosomalna recesywna ataksja móżdżkowa z powodu niedoboru GBA2

Kod ORPHA
352641

Kod OMIM
-

Kod ICD10
G11.8

Kod ICD11
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*Źródło

orphanet