

## 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, scoliosis, 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 mózdkowa z powodu niedoboru GBA2

#### Kod ORPHA

352641

#### Kod OMIM

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#### Kod ICD10

G11.8

#### Kod ICD11

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#### \*Źródło

orphanet