

## Opis choroby \*

### Definicja

A rare, genetic, slowly progressive neurodegenerative disease resulting from GRID2 deficiency characterized by motor, speech and cognitive delay, hypotonia, truncal and appendicular ataxia, and eye movement abnormalities (tonic upgaze, nystagmus, oculomotor apraxia). Intention tremor may also be associated. Brain imaging reveals progressive cerebellar atrophy with cerebellar flocculus particularly affected.

### Dane

#### Klasyfikacja

Podtyp kliniczny

#### Synonimy

Autosomal recessive congenital cerebellar ataxia due to ionotropic glutamate receptor delta-2 subunit deficiency  
Autosomalna recesywna wrodzona ataksja mózdkowa z powodu niedoboru podjednostki delta-2 jonotropowego receptora glutaminianu SCAR18  
SCAR18

#### Kod ORPHA

363432

#### Kod OMIM

616204

#### Kod ICD10

G11.1

#### Kod ICD11

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#### \*Źródło

orphanet