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

Kod ORPHA	**Kod OMIM**	**Kod ICD10**
363432	616204	G11.1

Kod ICD11
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*Źródło

orphanet