

Opis choroby *

Definicja

A rare slowly progressive autosomal recessive syndromic cerebellar ataxia characterized by late-onset cerebellar dysfunction (including gait and limb ataxia, nystagmus, and dysarthria), bilateral vestibulopathy (abnormal vestibulo-ocular reflex), and axonal sensory neuropathy. Variable features may include chronic cough and autonomic dysfunction. Brain imaging usually shows cerebellar atrophy.

Dane

Klasyfikacja

Choroba

Synonimy

CABV syndrome

CANVAS

Cerebellar ataxia with bilateral vestibulopathy syndrome

CABV syndrome

CANVAS

Cerebellar ataxia with bilateral vestibulopathy syndrome

Kod ORPHA

504476

Kod OMIM

614575

Kod ICD10

G11.2

Kod ICD11

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*Źródło

orphanet