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