Opis choroby *

Definicja

A rare X-linked cerebellar ataxia, characterized by a combination of upper and lower motor neuron signs, with an age of onset in the first or second decade, slow progression, and normal intelligence. Typical features of cerebellar dysfunction include gait and limb ataxia, intention tremor, dysmetria, dysdiadochokinesia, dysarthria, nystagmus, and hyperreflexia. Further phenotypic features are pes cavus, scoliosis, muscle atrophy, and peripheral sensory and motor nerve abnormalities.

Dane

Klasyfikacja
Choroba

Kod ORPHA
1175

Kod OMIM
302800

Kod ICD10
G11.1

Kod ICD11
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*Źródło

orphanet