Opis choroby *

Definicja

A rare autosomal recessive cerebellar ataxia characterized by onset of dystonia and other extrapyramidal signs, ataxia, oculomotor apraxia, and progressive sensorimotor polyneuropathy in the first decade of life. Patients present distal muscle weakness and atrophy, decreased vibratory sensation, and areflexia, and usually become wheelchair-bound by the third decade. Variable cognitive impairment may also be seen.

Dane

Klasyfikacja
Choroba

Synonimy
AOA4
AOA4

Kod ORPHA
459033

Kod OMIM
616267

Kod ICD10
G60.2

Kod ICD11
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*Źródło

orphanet