

## 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