

## Opis choroby \*

### Definicja

A rare autosomal recessive syndromic cerebellar ataxia characterized by the association of early-onset cerebellar ataxia with hearing loss and blindness. Patients may also present demyelinating peripheral motor neuropathy. Cerebral MRI shows alterations of the cerebellar white matter without cerebellar atrophy.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Autosomal recessive spinocerebellar ataxia type

3

SCAR3

Autosomal recessive spinocerebellar ataxia-  
blindness-hearing loss syndrome

SCABD

SCAR3

#### Kod ORPHA

95433

#### Kod OMIM

271250

#### Kod ICD10

G11.1

#### Kod ICD11

8A03.1Y

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#### \*Źródło

orphanet