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