## Opis choroby \*

## Definicja

A rare autosomal recessive axonal hereditary motor and sensory neuropathy characterized by infantile onset of recurrent episodes of acute liver failure (resulting in chronic liver fibrosis and hepatosplenomegaly), delayed motor development, cerebellar dysfunction presenting as gait disturbances and intention tremor, neurogenic stuttering, and motor and sensory neuropathy with muscle weakness especially in the lower legs, and numbness. Mild intellectual disability was reported in some patients. MRI of the brain shows non-progressive atrophy of the cerebellar vermis and thinning of the optic nerve.

Dane

Klasyfikacja Synonimy

Choroba Autosomal recessive spinocerebellar ataxia type

21

Autosomalna recesywna ataksja mózdżkowo-

rdzeniowa typu 21

SCAR21 SCAR21

Kod ORPHA 466794

**Kod OMIM Kod ICD10** 616719 G11.0

**Kod ICD11** 

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## \*Źródło

orphanet