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
Choroba

Synonimy
Autosomal recessive spinocerebellar ataxia type 21
Autosomalna recesywna ataksja móżdżkowo-rdzeniowa typu 21
SCAR21
SCAR21

Kod ORPHA
466794

Kod OMIM
616719

Kod ICD10
G11.0

Kod ICD11
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*Źródło

orphanet