

Opis choroby *

Definicja

Severe neurodegenerative syndrome with lipodystrophy is a rare, genetic, neurodegenerative disorder characterized by progressive psychomotor and cognitive regression (manifesting with gait ataxia, spasticity, loss of language, mild to severe intellectual disability, pyramidal and extrapyramidal signs and, frequently, development of tetraplegia or tetraparesis) associated with variable degrees of lipodystrophy, hepatomegaly, hypertriglyceridemia and muscular hypertrophy. Hyperactivity, tremor and development of seizures may also be associated.

Dane

Klasyfikacja

Choroba

Synonimy

Severe neurodegenerative syndrome due to

BSCL2 deficiency

Ciężki zespół neurodegeneracyjny z niedoborem

BSCL2

Kod ORPHA

363400

Kod OMIM

615924

Kod ICD10

G31.8

Kod ICD11

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*Źródło

orphanet