

Opis choroby *

Definicja

A rare mitochondrial disease characterized by adult onset of the triad of sensory ataxic neuropathy, dysarthria, and ophthalmoparesis. Additional signs and symptoms are highly variable and include myopathy, seizures, and hearing loss, among others. Brain imaging may show cerebellar white matter abnormalities and/or bilateral thalamic lesions.

Dane

Klasyfikacja

Choroba

Synonimy

SANDO

SANDO

Kod ORPHA

70595

Kod OMIM

607459

Kod ICD10

G71.3

Kod ICD11

5C53.21

*Źródło

orphanet