

Opis choroby *

Definicja

A rare neurologic disease characterized by progressive sensorineural deafness, progressive sensory neuropathy and gastrointestinal abnormalities, including progressive loss of gastric motility and small bowel diverticulosis and ulcerations, resulting in cachexia. Additional neurological manifestations may include dysarthria and absent tendon reflexes, as well as ptosis and external ophthalmoplegia. There have been no further descriptions in the literature since 1985.

Dane

Klasyfikacja

Choroba

Synonimy

Groll-Hirschowitz syndrome

Zespół Grolla i Hirschowitza

Hearing loss-small bowel diverticulosis-neuropathy syndrome

Kod ORPHA

3217

Kod OMIM

221400

Kod ICD10

G60.8

Kod ICD11

LD2H.Y

*Źródło

orphanet