

Opis choroby *

Definicja

A rare, genetic motor neuron disease characterized by a peripheral and cranial neuropathy, neuronal loss in anterior horns and atrophy of spinal sensory tracts, causing muscle weakness, sensory loss, diaphragmatic paralysis and respiratory insufficiency, and multiple cranial nerve deficits such as sensorineural hearing loss, bulbar symptoms, and loss of vision due to optic atrophy. Depending on the transporter affected, Riboflavin transporter deficiency 2 (RFVT2) and Riboflavin transporter deficiency 3 (RFVT3) are distinguished.

Dane

Klasyfikacja

Zespół wad wrodzonych Brown-Vialetto-van Laere syndrome

Synonimy

Czuciowo-nerwowa utrata słuchu - porażenie mostowo-opuszkowe
Zespół Browna, Vialetto i an Laere'a

Kod ORPHA

97229

Kod OMIM

614707

Kod ICD10

G12.2

Kod ICD11

LD2H.Y

*Źródło

orphanet