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

Synonimy
Brown-Vialetto-van Laere syndrome
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

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*Źródło

orphanet