

Opis choroby *

Definicja

A rare, life-threatening, mitochondrial DNA depletion syndrome disease characterized by severe, progressive sensorimotor neuropathy associated with corneal ulceration, scarring or anesthesia, acral mutilation, metabolic and immunologic derangement, and hepatopathy (which can manifest with fulminant hepatic failure, a Reye-like syndrome or indolent progression to liver cirrhosis, depending on clinical form involved), present in the Navajo Native American population. Clinical presentation includes failure to thrive, distal limb weakness with reduced sensation, limb contractures with loss of function, areflexia, recurrent metabolic acidosis with intercurrent illness, immunologic anomalies manifesting with severe systemic infections, and sexual infantilism.

Dane

Klasyfikacja

Choroba

Synonimy

Navajo neuropathy

Neuropatia Navajo

Kod ORPHA

255229

Kod OMIM

256810

Kod ICD10

E88.8

Kod ICD11

5C53.20

[*Źródło](#)

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