Opis choroby *

Definicja

A rare neurologic disease characterized by axonal sensorimotor neuropathy, progressive optic atrophy, cognitive deficit, bulbar dysfunction, seizures, and early hypotonia and feeding difficulties. Additional possible features include dystonia, scoliosis, joint contractures, ocular anomalies, and urogenital anomalies. Brain MRI reveals variable degrees of cerebral atrophy. The disease is fatal in childhood due to respiratory failure.

Dane

Klasyfikacja
Choroba

Synonimy
ANOAC
ANOAC
Zespół neuropatii aksonalnej, zaniku nerwu wzrokowego i deficytu poznawczego
Axonal neuropathy-optic atrophy-cognitive deficit syndrome

Kod ORPHA
457205

Kod OMIM
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Kod ICD10
G60.8

Kod ICD11
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*Źródło

orphanet