

## **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

Klasifikacja	Synonimy
Choroba	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