Opis choroby *

Definicja

A rare mitochondrial disease characterized by a variable clinical phenotype with the core features of optic atrophy, ataxia, and hypotonia. Additional common manifestations include global developmental delay with or without regression, neuropathy, spasticity, and microcephaly, less frequently seizures, movement disorder, hearing loss, and respiratory failure. Brain imaging may show abnormalities of the corpus callosum, basal ganglia, and midbrain, cerebral or cerebellar atrophy, or white matter abnormalities. The condition is frequently fatal at an early age.

Dane

Klasyfikacja
Choroba

Kod ORPHA
543470

Kod OMIM
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Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet