Opis choroby *

Definicja

A rare inherited mitochondrial disease characterized by the clinical features of Leber hereditary optic neuropathy in combination with other systemic or neurological abnormalities. These abnormalities include: postural tremor, motor disorder, multiple sclerosis-like syndrome, spinal cord disease, skeletal changes, Parkinsonism with dystonia, anarthria, motor and sensory peripheral neuropathy, spasticity, mild encephalopathy, and cardiac arrhythmias.

Dane

Klasyfikacja
Choroba

Synonimy
LHON plus disease
LHON plus disease

Kod ORPHA
99718

Kod OMIM
500001

Kod ICD10
H47.2

Kod ICD11
8C73.Y

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*Źródło

orphanet