Opis choroby *

Definicja

A rare mitochondrial oxidative phosphorylation disorder due to nuclear DNA anomalies characterized by onset of slowly progressive proximal lower limb weakness and exercise intolerance in the first decade of life, followed by weakness of neck flexor, shoulder, and distal leg muscles. Facial muscles become involved still later in the disease course. Additional manifestations are restrictive pulmonary function and short stature. Laboratory studies reveal lactic acidemia and increased serum creatine kinase.

Dane

Klasyfikacja
Choroba

Kod ORPHA
457050

Kod OMIM
616209

Kod ICD10
G71.3

Kod ICD11
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*Źródło

orphanet