Opis choroby *

Definicja

Congenital cataract-progressive muscular hypotonia-hearing loss-developmental delay syndrome is a rare, genetic, mitochondrial myopathy disorder characterized by congenital cataract, progressive muscular hypotonia that particularly affects the lower limbs, reduced deep tendon reflexes, sensorineural hearing loss, global development delay and lactic acidosis. Muscle biopsy reveals reduced complex I, II and IV respiratory chain activity.

Dane

Klasyfikacja
Choroba

Synonimy
Congenital cataract-progressive muscular hypotonia-deafness-developmental delay syndrome
Zaćma wrodzona - postępująca hipotonia mięśni - głuchota - opóźnienie rozwoju

Kod ORPHA
330054

Kod OMIM
613076

Kod ICD10
G71.3

Kod ICD11
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*Źródło

orphanet