Opis choroby *

Definicja

Combined oxidative phosphorylation deficiency type 3 is an extremely rare clinically heterogenous disorder described in about 5 patients to date. Clinical signs included hypotonia, lactic acidosis, and hepatic insufficiency, with progressive encephalomyopathy or hypertrophic cardiomyopathy.

Dane

Klasyfikacja
Choroba

Synonimy
Fatal mitochondrial disease due to COXPD3
Śmiertelna choroba mitochondrialna spowodowana COXPD3

Kod ORPHA
168566

Kod OMIM
610505

Kod ICD10
E88.8

Kod ICD11
5C53.23

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*Źródło

orphanet