Opis choroby *

Definicja

A rare disorder of fatty acid oxidation characterized by a wide clinical spectrum ranging from severe neonatal manifestations including cardiomyopathy, hypoglycemia, metabolic acidosis, skeletal myopathy and neuropathy, liver disease and death to a mild phenotype with peripheral polyneuropathy, episodic rhabdomyolysis and pigmentary retinopathy..

Dane

Klasyfikacja
Choroba

Synonimy
TFP deficiency
Niedobór TFP
TFPD
TFPD

Kod ORPHA
746

Kod OMIM
609015

Kod ICD10
G71.3

Kod ICD11
5C52.01

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*Źródło

orphanet