

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	Synonimy
Choroba	TFP deficiency Niedobór TFP TFPD TFPD
Kod ORPHA	Kod OMIM
746	609015
Kod ICD10	Kod ICD11
	G71.3
	5C52.01

*Źródło

orphanet