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Mitochondrial trifunctional protein deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Mitochondrial trifunctional protein deficiency. ORPHA:746

Mitochondrial trifunctional protein (TFP) deficiency (TFPD) is a 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..