Carnitine palmitoyl transferase 1A deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Carnitine palmitoyl transferase 1A deficiency. ORPHA:156

Carnitine palmitoyltransferase 1A (CPT-1A) deficiency is an inborn error of metabolism that affects mitochondrial oxidation of long chain fatty acids (LCFA) in the liver and kidneys, and is characterized by recurrent attacks of fasting-induced hypoketotic hypoglycemia and risk of liver failure.