Carnitine Palmitoyltransferase I Deficiency

National Cancer Institute

Source

National Cancer Institute. Carnitine Palmitoyltransferase I Deficiency. NCI Thesaurus. Code C98871.

A rare autosomal recessive inherited disorder caused by mutations in the CPT1A gene. It is characterized by the presence of defective carnitine palmitoyltransferase 1A which is involved in fatty acid oxidation. Signs and symptoms may be exacerbated during fasting and include hypoketotic hypoglycemia, increased levels of carnitine in the blood, hepatomegaly, seizures, and coma.