

DISEASE:
Carnitine palmitoyl transferase 1A deficiency

NAME:	Carnitine palmitoyl transferase 1A deficiency
DESCRIPTION:	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.
ORPHACODE:	156
SYNONYMS:	CPT1A deficiency Carnitine palmitoyl transferase 1A deficiency Hepatic carnitine palmitoyl transferase 1 deficiency Hepatic carnitine palmitoyl transferase I deficiency L-CPT1 deficiency L-CPTI deficiency
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	CPT1A
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: <http://gentest.healthdata.be/disease/577>

RELATED CONTENT

Related Genetic Tests

- [Metabolic diseases with hepatic disorders \(20 genes\)](#)

Related Laboratories

- [Centre de Génétique Médicale UCL](#)

Related Analytes

- [carnitine palmitoyltransferase 1A](#)

Related Gene Panels

- [Metabolic diseases with hepatic disorders \(20 genes\) - UCL](#)

Source URL: <http://gentest.healthdata.be/disease/577>