

DISEASE:**Carnitine palmitoyl transferase II deficiency, neonatal form**

NAME:	Carnitine palmitoyl transferase II deficiency, neonatal form
DESCRIPTION:	The neonatal form of carnitine palmitoyltransferase II (CPT II) deficiency (see this term), an inherited disorder that affects mitochondrial oxidation of long chain fatty acids (LCFA), is the lethal form of the disease which presents with multisystem failure.
ORPHACODE:	228308
SYNONYMS:	CPT2, lethal systemic form CPT2, neonatal form CPTII, lethal systemic form CPTII, neonatal form Carnitine palmitoyl transferase II deficiency, lethal systemic form Carnitine palmitoyl transferase deficiency type 2, lethal systemic form Carnitine palmitoyl transferase deficiency type 2, neonatal form
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	CPT2
CREATED:	13 May 2019 - 01:02

CHANGED:	22 Jun 2023 - 16:14
-----------------	---------------------

Source URL: <http://gentest.healthdata.be/disease/1537>

RELATED CONTENT

Related Genetic Tests

- [Carnitine Palmitoyl transferase type II](#)
- [Carnitine Palmitoyltransferase type II](#)

Related Laboratories

- [Centre de Génétique-Institut de Pathologie et de Génétique \(IPG\)](#)
- [Centrum Medische Genetica - UZ Antwerpen](#)

Related Analytes

- [carnitine palmitoyltransferase 2](#)

Source URL: <http://gentest.healthdata.be/disease/1537>