Carnitine Palmitoyltransferase II Deficiency

National Cancer Institute

Source

National Cancer Institute. Carnitine Palmitoyltransferase II Deficiency. NCI Thesaurus. Code C114766.

A rare, autosomal recessive inherited disorder of long-chain fatty-acid oxidation caused by mutations in the CPT2 gene. The disease includes three main types: a lethal neonatal form, a severe infantile hepatocardiomuscular form, and a myopathic form.