Mitochondrial Trifunctional Protein Deficiency

National Cancer Institute

Source

National Cancer Institute. Mitochondrial Trifunctional Protein Deficiency. NCI Thesaurus. Code C98991.

A rare, autosomal recessive inherited disorder caused by mutations in the HADHA and HADHB genes. It is characterized by the deficiency of an enzyme involved in the fatty acid oxidation process. Signs and symptoms may appear early or later in life and may be triggered by periods of fasting or illnesses. They include feeding difficulties, lethargy, hypoglycemia, hypotonia, liver abnormalities, heart abnormalities, peripheral neuropathy, coma, and sudden death.