Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency

National Cancer Institute

Source

National Cancer Institute. Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency. NCI Thesaurus. Code C129929.

An inherited condition caused by mutation(s) in the HADHA gene, encoding trifunctional enzyme subunit alpha, mitochondrial. It is characterized by hypoglycemia, hypotonia, neuropathy, cardiomyopathy, pigmentary retinopathy and may be associated with sudden death.