Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency. ORPHA:5

Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD) is a mitochondrial disorder of long chain fatty acid oxidation characterized in most patients by onset in infancy/ early childhood with hypoketotic hypoglycemia, metabolic acidosis, liver disease, hypotonia and frequently cardiac involvement with arrhythmias and/or cardiomyopathy.