Open Peer Review on Qeios

Carnitine-acylcarnitine translocase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Carnitine-acylcarnitine translocase deficiency. ORPHA:159

Carnitine-acylcarnitine translocase (CACT) deficiency is a life-threatening, inherited disorder of fatty acid oxidation which usually presents in the neonatal period with severe hypoketotic hypoglycemia, hyperammonemia, cardiomyopathy and/or arrhythmia, hepatic dysfunction, skeletal muscle weakness, and encephalopathy.