Open Peer Review on Qeios

Medium chain acyl-CoA dehydrogenase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Medium chain acyl-CoA dehydrogenase deficiency. ORPHA:42

Medium chain acyl-CoA dehydrogenase (MCAD) deficiency (MCADD) is an inborn error of mitochondrial fatty acid oxidation characterized by a rapidly progressive metabolic crisis, often presenting as hypoketotic hypoglycemia, lethargy, vomiting, seizures and coma, which can be fatal in the absence of emergency medical intervention.