Systemic primary carnitine deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Systemic primary carnitine deficiency. ORPHA:158

Systemic primary carnitine deficiency (SPCD) is a potentially lethal disorder of fatty acid oxidation characterized classically by early childhood onset cardiomyopathy often with weakness and hypotonia, failure to thrive and recurrent hypoglycemic hypoketotic seizures and/or coma.