Short chain acyl-CoA dehydrogenase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Short chain acyl-CoA dehydrogenase deficiency. ORPHA:26792

Short-chain acyl-CoA dehydrogenase (SCAD) deficiency is a very rare inborn error of mitochondrial fatty acid oxidation characterized by variable manifestations ranging from asymptomatic individuals (in most cases) to those with failure to thrive, hypotonia, seizures, developmental delay and progressive myopathy.