D-glyceric aciduria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. D-glyceric aciduria. ORPHA:941

D-glyceric aciduria is a metabolic disorder characterized by D-glyceric acid excretion. It has been described in several patients. Clinical findings include progressive neurological impairment, hypotonia, seizures, failure to thrive and metabolic acidosis. Some patients had hyperglycinemia secondary to the organic acidemia. However, some of the reported patients were asymptomatic. D-glyceric aciduria is caused by D-glycerate kinase deficiency. The GLYCTK gene has been mapped to 3p21.