Glycogen storage disease due to phosphoglycerate kinase 1 deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Glycogen storage disease due to phosphoglycerate kinase 1 deficiency. ORPHA:713

Phosphoglycerate kinase (PGK) deficiency is a metabolic disorder characterized by variable combinations of nonspherocytic hemolytic anemia, myopathy, and various central nervous system abnormalities.