Glycogen storage disease due to phosphoglycerate mutase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Glycogen storage disease due to phosphoglycerate mutase deficiency. ORPHA:97234

Muscle phosphoglycerate mutase deficiency (PGAMD) is a metabolic myopathy characterised by exercise-induced cramp, myoglobinuria, and presence of tubular aggregates in the muscle biopsy. Serum creatine kinase (CK) levels are increased between episodes of myoglobinuria. Less than 50 cases have been described so far. The disease is due to an anomaly in one of the last steps of glycolysis. The enzymatic defect in PGAMD is caused by mutations in the cDNA coding for the M-isoform of PGAM. Residual PGAM activity in the muscles of patients (2%-6%) is due to activity of the B-isoform. Transmission is autosomal recessive. Differential diagnosis includes muscle phosphorylase deficiency (McArdle disease) and phosphofructokinase deficiency (PFKD) (see these terms).