Phosphoglycerate Mutase Deficiency

National Cancer Institute

Source

National Cancer Institute. Phosphoglycerate Mutase Deficiency. NCI Thesaurus. Code C131647.

A rare, autosomal recessive, inherited disorder caused by mutation of the PGAM2 gene. It is characterized by non-spherocytic hemolytic anemia, exercise-induced cramping, myoglobinuria, and presence of tubular aggregates on muscle biopsy.