

Glycogen storage diseases are a group of disorders in which stored glycogen cannot be metabolized into glucose to supply energy for the body. Glycogen storage disease type VII (GSD VII) is characterized by weakness, pain and stiffness during exercise. GSD VII is caused by abnormalities in the muscle phosphofructokinase gene that results in a deficiency of the phosphofructokinase enzyme. This enzyme deficiency leads to a reduced amount of energy available to muscles during exercise. GSD VII is inherited as an autosomal recessive genetic disorder. GSD type VII usually begins in childhood and is characterized by weakness, pain and stiffness during exercise, sometimes associated with nausea and vomiting and dark, burgundy-colored urine due to the presence of myoglobin (myoglobinuria). Destruction of muscle tissue (rhabdomyolysis) can also occur. A rare form of GSD type VII has been reported in infants that is associated with progressive loss of muscle tone (hypotonia), muscle weakness and death. A late-onset form has been reported in adults who experience only muscle weakness. Glycogen storage disease type VII is a rare disorder that occurs more often in individuals of Japanese and Ashkenazi Jewish descent. GSD type VII affects males and females in equal numbers. GSD type VII is diagnosed by a muscle biopsy for measurement of the phosphofructokinase enzyme level or measurement of the phosphofructokinase enzyme level in red blood cells. Molecular genetic testing for the phosphofructokinase gene mutations prevalent in the Ashkenazi Jewish population are available on a research basis.