Glycogen Storage Disease Type Ia

National Cancer Institute

Source

National Cancer Institute. Glycogen Storage Disease Type Ia. NCI Thesaurus. Code C162398.

An autosomal recessive condition caused by mutation(s) in the G6PC gene, encoding glucose-6-phosphatase. It is characterized by accumulation of glycogen in the kidneys and liver resulting in hypoglycemia, hyperlipidemia, and hyperuricemia. Adults may have a high incidence of hepatic adenomas.