Gitelman Syndrome

National Cancer Institute

Source

National Cancer Institute. Gitelman Syndrome. NCI Thesaurus. Code C84730.

An inherited disorder caused by mutations in the SLC12A3 gene. It is characterized by deficient reabsorption of electrolytes in the distal convoluted tubules of the kidneys. It results in hypochloremic metabolic alkalosis, hypokalemia, hypocalciuria, and hypomagnesemia.