Phosphoenolpyruvate Carboxykinase Deficiency

National Cancer Institute

Source

National Cancer Institute. Phosphoenolpyruvate Carboxykinase Deficiency. NCI Thesaurus. Code C99015.

A very rare, autosomal recessive inherited disorder caused by deficiency of the enzyme phosphoenolpyruvate carboxykinase, which is involved in gluconeogenesis. It presents with hypoglycemia, failure to thrive, metabolic acidosis, muscle weakness, and hepatomegaly.