PEPCK deficiency is an extremely rare disorder of carbohydrate metabolism inherited as an autosomal recessive trait. A deficiency of the enzyme phosphoenolpyruvate carboxykinase (PEPCK), which is a key enzyme in the conversion of proteins and fat to glucose (gluconeogenesis), causes an excess of acid in the circulating blood (acidemia). Characteristics of this disorder are low blood sugar (hypoglycemia), loss of muscle tone, liver enlargement and impairment, and failure to gain weight and grow normally. PEPCK deficiency is extremely rare. One overview suggests that only 10 cases have been reported in the medical literature. Diagnosis of PEPCK deficiency can be made shortly after birth by biochemical analysis of fibroblast cells.