Beta-Ketothiolase Deficiency

National Cancer Institute

Source

National Cancer Institute. Beta-Ketothiolase Deficiency. NCI Thesaurus. Code C98841.

A rare autosomal recessive inherited disorder caused by mutations in the ACAT1 gene. It is characterized by the reduction or elimination of the enzyme mitochondrial acetoacetyl-CoA thiolase which is responsible for the metabolism of the amino acid isoleucine and ketone-body metabolism. Signs and symptoms appear early in life and include vomiting, dehydration, breathing difficulties, seizures, lethargy, and coma.