Carnitine palmitoyltransferase 1A deficiency (CPT1A) is characterized by a sudden onset of liver failure and damage to the nervous system resulting from liver failure (hepatic encephalopathy), usually associated with fasting or illness. CPT1A deficiency is caused by an abnormality (mutation) in the CPT1A gene that results in the production of an abnormally functioning carnitine palmitoyltransferase 1 enzyme and decreased metabolism of long-chain fatty acids. CPT1A deficiency is inherited as an autosomal recessive genetic disorder. Three types of CPT1A deficiency have been recognized: The hepatic encephalopathy type usually occurs in children and is associated with a low level of ketones in the blood, low blood sugar (hypoglycemia), enlarged liver, muscle weakness and elevated carnitine in the blood. The adult-onset myopathy type is characterized by a sudden onset of muscle cramping associated with exercise without low blood sugar or liver dysfunction. The third type is acute fatty liver of pregnancy that occurs when a pregnant woman with one abnormal CPT1A gene carries a fetus with two abnormal CPT1A genes and is associated with liver failure in the mother. CPT1A deficiency has been reported in approximately 30-40 individuals. The incidence of this condition may be higher in the Hutterite populations in the northern United States and Canada and the Inuit populations in northern Canada, Alaska and Greenland. This condition occurs with equal frequency in males and females. CPT1A deficiency is diagnosed by a combination of physical symptoms and laboratory testing. The typical laboratory findings include low levels of ketones, elevated liver transaminases, elevated ammonia and elevated total serum carnitine. CPT1A enzyme activity on the cultured skin cells from affected individuals is 1-5% of normal. Molecular genetic testing is available to confirm the diagnosis if the enzyme test is abnormal. Some state newborn screening programs perform screening for CPT1A deficiency by measuring the ratio of free to total carnitine in blood plasma or serum. Carrier testing for relatives is available using CPT1A enzyme testing or molecular genetic testing.