CPT1A Gene

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carnitine palmitoyltransferase 1A

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1. Normal Function

The *CPT1A* gene provides instructions for making an enzyme called carnitine palmitoyltransferase 1A, which is found in the liver. This enzyme is essential for fatty acid oxidation, a multistep process that breaks down (metabolizes) fats and converts them into energy. Fatty acid oxidation takes place within mitochondria, which are the energy-producing centers in cells. A group of fats called long-chain fatty acids cannot enter mitochondria unless they are attached to a substance known as carnitine. Carnitine palmitoyltransferase 1A connects carnitine to long-chain fatty acids so they can cross the inner membrane of mitochondria. Once these fatty acids are inside mitochondria, carnitine is removed and they can be metabolized to produce energy. During periods of fasting, long-chain fatty acids are an important energy source for the liver and other tissues.

2. Health Conditions Related to Genetic Changes

2.1. Carnitine Palmitoyltransferase I Deficiency

More than 20 mutations in the *CPT1A* gene have been found to cause carnitine palmitoyltransferase I (CPT I) deficiency. Most of these mutations change single protein building blocks (amino acids) within carnitine palmitoyltransferase 1A. Mutations in the *CPT1A* gene severely reduce or eliminate the activity of this enzyme. Without enough of this enzyme, carnitine is not attached to long-chain fatty acids. As a result, these fatty acids cannot enter mitochondria and be converted into energy. Reduced energy production can lead to some of the features of CPT I deficiency, such as low blood sugar (hypoglycemia) and low levels of the products of fat breakdown (hypoketosis). Fatty acids may also build up in cells and damage the liver, heart, and brain. This abnormal buildup causes the other signs and symptoms of the disorder.

2.2. Other Disorders

CPT1A gene mutations appear to increase the risk of a serious liver disorder that can develop in women during pregnancy. This disorder, called acute fatty liver of pregnancy, begins with abdominal pain and can rapidly progress to liver failure. Signs of acute fatty liver of pregnancy include an abnormal accumulation of fat in the liver, hypoglycemia, increased levels of ammonia in the blood (hyperammonemia), and abnormalities in liver enzymes. A small percentage of women who have a mutation in one copy of the *CPT1A* gene in each cell and are carrying a fetus with mutations in both copies of the *CPT1A* gene develop this maternal liver disease. Little is known about the relationship between *CPT1A* gene mutations and liver problems in the mother during pregnancy.

3. Other Names for This Gene

- carnitine palmitoyltransferase 1A (liver)
- carnitine palmitoyltransferase I, liver
- CPT1
- CPT1-L
- CPT1A_HUMAN
- L-CPT1

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