

Carnitine Palmitoyl Transferase, Type 1A (CPT1A)

The Arctic Variant

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Abstract

Carnitine Palmitoyl Transferase 1A (CPT1A) is a crucial enzyme needed for mitochondrial fatty acid oxidation and is fundamental for appropriate metabolic responses to prolonged fasting. Under normal conditions, the human body guarantees a constant energy supply, by metabolizing glucose in the short term and by oxidizing fatty acids into ketones during long term fasting or starvation. Fasting induces breakdown of the hepatic glycogen supply into glucose. Continued fasting eventually leads to glycogen depletion followed by a decrease in serum glucose and insulin levels, this decrease then activates hormone-sensitive lipases located in adipose tissues and causes the release of free fatty acids into the blood. Normally these free fatty acids would undergo beta-oxidation to produce energy, but CPT1A deficiency results in an 80% decrease in the activity of the CPT1A enzyme, leading to significant hepatic glycogen depletion and the inability to utilize ketones during periods of fasting. Preoperative fasting in these patients may result in vomiting, lethargy, hypoketotic hypoglycemia, seizures, liver failure, increased risk for infectious illnesses and higher rates of respiratory illnesses, placing this population at greater perioperative risk for anesthesia and surgery. Interventions include, but are not limited to, parent education and prevention techniques, recognition of symptoms, prompt treatment with glucose and even surgery cancellation. CPT1A deficiency is found in circum-arctic populations such as Alaskan Inupiat and Yupik, Canadian and Greenland Inuit, and Siberian Yupik and is thus known as the “Arctic Variant” of CPT1A. Once thought of as a rare disorder, the introduction of tandem mass spectrometry (MS/MS) to Alaska newborn screenings in 2003 revealed that the polymorphism c.1436C>T variant in the CPT1A gene has an incidence as high as 80% in specific regions of Alaska. As of 2011, an estimated 700 Alaska Native Infants born each year are homozygous for the c.1436C>T Arctic Variant. It is therefore important for anesthesia professionals in Alaska and other arctic regions to understand what CPT1A Arctic Variant is, and its implications for these patients in the perioperative period.

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Poster Presentation

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