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Review

Carnitine palmitoyltransferases 1 and 2: biochemical, molecular and medical aspects

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Abstract

Carnitine palmitoyltransferase (CPT) deficiencies are common disorders of mitochondrial fatty acid oxidation. The CPT system is made up of two separate proteins located in the outer (CPT1) and inner (CPT2) mitochondrial membranes. While CPT2 is an ubiquitous protein, three tissue-specific CPT1 isoformsâ€“â€“the so-called â€œliverâ€ (CPT1-A), â€œmuscleâ€ (CPT1B) and Â«brainÂ» (CPT1-C) CPT1sâ€“â€“have been shown to exist. Amino acid and cDNA nucleotide sequences have been identified for all of these proteins. CPT1-A deficiency presents as recurrent attacks of fasting hypoketotic hypoglycemia. Twenty four *CPT1A* mutations have been reported to date. CPT1-B and -C deficiencies have not been hitherto identified. CPT2 deficiency has several clinical presentations. The â€œbenignâ€ adult form (more than 200 families reported) is lysis triggered by prolonged exercise. The

prevalent S113L mutation is found in about 50% of mutant alleles. The infantile-type CPT2 presents as severe attacks of hypoketotic hypoglycemia, occasionally associated with cardiac damage commonly responsible for sudden death before 1 year of age. In addition to these symptoms, features of brain and kidney dysorganogenesis are frequently seen in the neonatal-onset CPT2 deficiency, almost always lethal during the first month of life. Around 40 CPT2 mutations (private missense or truncating mutations) have hitherto been detected. Treatment is based upon avoidance of fasting and/or exercise, a low fat diet enriched with medium chain triglycerides and carnitine. Prenatal diagnosis may be offered for pregnancies at a 1/4 risk of infantile/severe-type CPT2 deficiency.

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Keywords

Carnitine palmitoyltransferase 1; Carnitine palmitoyltransferase 2; Carnitine palmitoyltransferase deficiency; Fatty acids; Fatty acid oxidation; Mitochondria; Hypoglycemia; Hypoketotic

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