

Natural History without treatment: While most patients have survived infancy and acute hypoglycemic episodes, some have suffered permanent neurological deficits. Death has occurred during acute episodes in some cases.

Natural History with treatment: Theoretically normal development if hypoglycemia and cardiac involvement can be prevented.

Metabolic Information:

Missing Enzyme & Location: The enzyme CPT I converts long chain acyl-CoA substrates and carnitine to respective acylcarnitines for transport into the mitochondria. CPT I deficiency results in accumulation of toxic long chain substrates and impedes mitochondrial beta-oxidation. There is no correlation between enzyme activity and clinical severity.

MS/MS profile: C0 (free carnitine)- elevated/normal
C16 with C18:1- low
C0/C16 ratio- very elevated
C0/C18 ratio- very elevated

Prenatal testing: Possible to do enzyme analysis on amniocytes or CVS in patients with known risk.

Miscellaneous Information:

Metabolite confirmation is difficult due to lack of characteristic acylcarnitine or acylglycine markers.

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