



**Metabolic Information:**

**Missing Enzyme & Location:** MCC is predominantly localized to the inner membrane of the mitochondria and is known to be highly expressed in kidney and liver. The enzyme involved is 3-METHYLCROTONYL-CoA CARBOXYLASE.

**MS/MS profile:** C5:1 (tiglyl or 3-methylcrotonyl carnitine)- elevated  
C5-OH (3-hydroxy-2-methylbutyryl carnitine) – elevated

**Prenatal testing:** May be possible for at risk pregnancies.

**Miscellaneous Information:**

Heterozygotes (obligate carriers) do not have abnormal metabolites in the urine. For definitive diagnosis, exclusion of multiple carboxylase deficiency, enzyme assay must show a deficit of 3-MCC activity and normal activity of at least one other carboxylase enzyme in leukocytes or fibroblasts.

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