

CARNITINE PALMITOYL TRANSFERASE-I (CPT-I) DEFICIENCY

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- most dietary fatty acids are long chain (having more than 12 carbons) but with some being medium chain (6-12 carbons)
- fatty acid oxidation:
 - ✓ fatty acids enter the cell from the blood and undergo oxidation in the mitochondria, the energy-producing compartment found in cells
 - ✓ to enter the mitochondrial compartment, fatty acids must be converted to their fatty acyl-carnitine form
 - ✓ separate enzymes transfer carnitine onto long and medium chain fatty acids
 - ✓ in the mitochondria the fatty acyl-carnitine is converted to the fatty acyl-CoA form and carnitine is recycled
 - ✓ fatty acyl-CoA is oxidized by the β -oxidation pathway to produce usable energy that can drive cell processes such as glucose biosynthesis
- **carnitine palmitoyl transferase-I deficiency**
 - ✓ transfers carnitine onto long chain fatty acids only
 - ✓ medium chain fatty acids can be supplied in the diet as a treatment to bypass this deficiency

