SYNDROME OF SYSTEMIC CARNITINE DEFICIENCY: CLINICAL, MORPHOLOGIC, BIOCHEMICAL, AND PATHOPHYSIOLOGIC FEATURES

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An 11-year old boy had had recurrent episodes of hepatic and cerebral dysfunction and underdeveloped musculature. Overt weakness developed at age 10. Lipid excess, especially in type I fibers, was found in muscle. Hypertrophied smooth endoplasmic reticulum and excessive microbodies were present in liver. Marked carnitine deficiency was shown in skeletal muscle, plasma, and liver. Ketogenesis was impaired on a high fat diet, but omega oxidation of fatty acids was enhanced. There was excessive glucose uptake and essentially no oxidation of labeled long-chain fatty acids by perfused forearm muscles in vivo. Oral replacement therapy restored plasma carnitine levels to normal, but not liver or muscle carnitine levels, and was accompanied by clinical improvement.

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Comment from Jonathan W. Mink, MD, PhD, FAAN, Associate Editor: This is the original description of systemic carnitine deficiency, a disorder that has subsequently been shown to result from a defect in the high-affinity carnitine transporter. This results in impaired fatty acid oxidation and impaired ketogenesis.

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