Abstract


Plasma concentration of 3-hydroxyisovaleryl carnitine is an early and sensitive indicator of marginal biotin deficiency in humans.

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BACKGROUND: Blood-based indicators of biotin status in humans were shown to be useful tools in several clinical situations, including pregnancy. We previously validated the activity of the biotin-dependent enzyme propionyl-coenzyme A carboxylase (PCC) in lymphocytes as a sensitive and specific blood-based indicator of marginal degrees of biotin deficiency. However, the measurement of PCC activity in population studies presents substantial analytic challenges. 3-Hydroxyisovaleryl carnitine (3HIA-carnitine) increases in response to the decreased activity of the biotin-dependent enzyme methylcrotonyl-coenzyme A carboxylase and might reflect biotin status.

OBJECTIVE: We sought to determine whether the plasma concentration of 3HIA-carnitine increases significantly in marginal biotin deficiency.

DESIGN: We experimentally induced marginal, asymptomatic biotin deficiency in 10 healthy adults (8 women) by having the subjects consume undenatured egg white for 28 d; biotin status was then repleted. Plasma concentrations of 3HIA-carnitine were measured on days 0, 14, 28, 35, and 50 by liquid chromatography-mass spectroscopy.

RESULTS: The mean plasma 3HIA-carnitine concentration increased with depletion (P < 0.0001) and decreased with repletion (P < 0.0001). Plasma 3HIA-carnitine concentrations were greater than the upper limit of normal concentrations in 7 of 10 subjects by day 14 and in 9 of 10 subjects by day 28 and decreased to within normal limits in 9 of 10 subjects by day 50.

CONCLUSIONS: These studies provide evidence that 3HIA-carnitine is an early and sensitive indicator of marginal biotin deficiency. The ease of sample collection, small sample volume requirement, and stability of 3HIA-carnitine during storage suggest that plasma 3HIA-carnitine concentration is likely to be a useful indicator of marginal biotin deficiency for larger population studies.

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