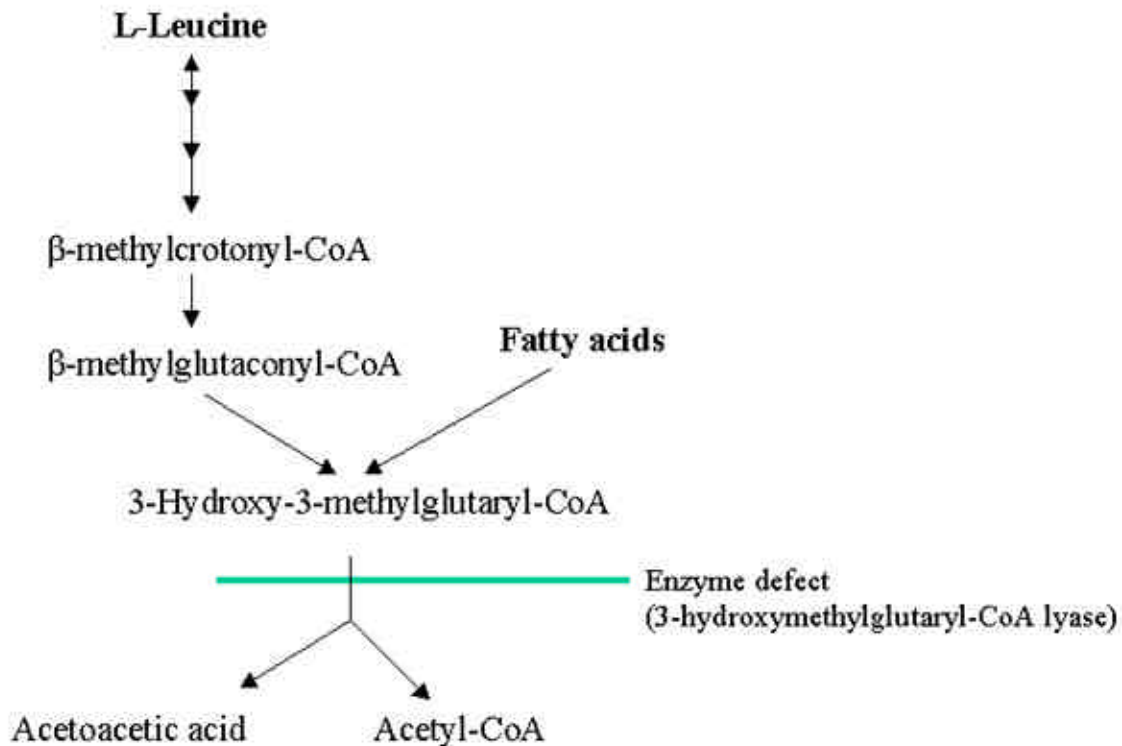


ACUTE ILLNESS PROTOCOLS

3-HYDROXYMETHYLGLUTARYL-CoA (3-HMG CoA) LYASE DEFICIENCY

PATHOPHYSIOLOGY



In the presence of catabolism or substantially reduced food intake (e.g. infection, severe exertion), the combination of an increased cellular requirement for energy and reduced glucose intake results in proteolysis with release of amino acids and fatty acids. Enhanced leucine and fatty acid degradation is an attempt by the body to produce the needed energy in the form of ketones. When 3-HMG-CoA lyase is deficient, the increased fluxes in both leucine degradation and fatty acid oxidation result in an accumulation of 3-hydroxymethylglutaryl-CoA. The accumulated substrate produces **metabolic acidosis**, inhibits gluconeogenesis resulting in **hypoglycemia**, and inhibits the urea cycle resulting in **hyperammonemia**.