

Homocystinuria is a rare metabolic condition characterized by an excess of the compound homocystine in the urine. The condition may result from deficiency of any of several enzymes involved in the conversion of the essential amino acid methionine to another amino acid (cysteine)--or, less commonly, impaired conversion of the compound homocysteine to methionine. Enzymes are proteins that accelerate the rate of chemical reactions in the body. Certain amino acids, which are the chemical building blocks of proteins, are essential for proper growth and development. In most cases, homocystinuria is caused by reduced activity of an enzyme known as cystathione beta-synthase (CBS). Infants who develop homocystinuria due to CBS deficiency (which is also known as classical homocystinuria) may fail to grow and gain weight at the expected rate (failure to thrive) and have developmental delays.