Homocystinuria

National Cancer Institute

Source

National Cancer Institute. Homocystinuria. NCI Thesaurus. Code C84765.

An autosomal recessive inherited metabolic disorder caused by mutations in the CBS, MTHFR, MTR, and MTRR genes. It is characterized by abnormalities in the methionine metabolism and is associated with deficiency of cystathionine synthase. It results in the accumulation of homocysteine in the serum. It may affect the cardiovascular, musculoskeletal and the central nervous systems.