

Open Peer Review on Qeios

Cystathioninuria

National Cancer Institute

Source

National Cancer Institute. <u>Cystathioninuria</u>. NCI Thesaurus. Code C129070.

An autosomal recessive disorder caused by mutations in the CTH gene, encoding cystathionine gamma-lyase. The condition is characterized by increased concentrations of cystathionine in the plasma and urine.

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