Title: Homocystinuria Caused by Cystathionine β-Synthase Deficiency *GeneReview* — Terms Used to Describe Sulfur Amino Acids

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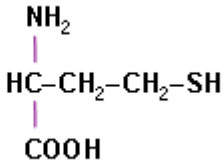
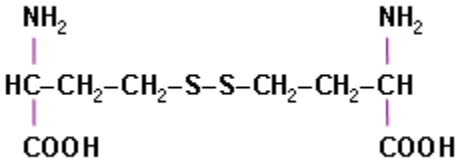
Terms Used to Describe Sulfur Amino Acids

Note: The terms used to describe the sulfur amino acids are confusing because homocysteine, the thiol within the methionine metabolic pathway (Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency; see Figure 1) with its free sulfur, readily combines with other thiols (such as another homocysteine or cysteine) to form a disulfide; it is primarily the disulfides that are measured in the standard amino acid analysis. For clarity, Mudd et al [2000] have proposed the following terminology to describe the sulfur amino acid metabolites that are important in homocystinuria and related disorders:

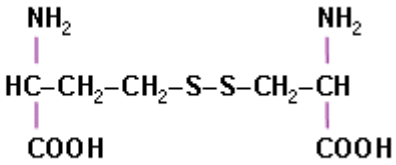
- **Homocysteine (HcyH).** A thiol compound:



- **Homocystine (Hcy-Hcy).** A symmetric disulfide:



- **Homocysteine-cysteine mixed disulfide (Hcy-Cys).** An asymmetric disulfide:



- **Total homocysteine (tHcy).** All of the Hcy that is present, including that which is bound to protein, most of which is liberated from disulfide bonding by a specific analysis that requires prior reduction.
- **Total free homocysteine (tfHcy)**. A measurement sometimes used in following individuals with homocystinuria, calculated by assigning two Hcy's to the amount of free homocystine (Hcy-Hcy), one Hcy to the amount of

homocysteine-cysteine mixed disulfide (Hcy-Cys), and adding the amounts. Total free Hcy is distinguished from tHcy, which includes the Hcy that was formerly protein bound.

References

Mudd SH, Finkelstein JD, Refsum H, Ueland PM, Malinow MR, Lentz SR, Jacobsen DW, Brattstrom L, Wilcken B, Wilcken DE, Blom HJ, Stabler SP, Allen RH, Selhub J, Rosenberg IH. Homocysteine and its disulfide derivatives: a suggested consensus terminology. Arterioscler Thromb Vasc Biol 2000;20:1704-6.