Hypermethioninemia due to glycine N-methyltransferase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hypermethioninemia due to glycine N-methyltransferase deficiency. ORPHA:289891

Hypermethioninemia due to glycine N-methyltransferase deficiency is a rare, genetic inborn error of metabolism characterized by a relatively benign clinical phenotype, with only mild to moderate hepatomegaly reported, in addition to laboratory studies revealing permanent, greatly increased hypermethioninemia, mild to moderate elevation of aminotransferases and highly elevated plasma S-adenosyl-methionine with normal S-adenosylhomocysteine and total homocysteine.