

Celiac sprue, hyperhomocysteinemia, and MTHFR gene variants.

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Abstract

Highlight Terms [?](#)

Diseases(4) Genes/Proteins(1) Chemicals(2)

GOALS: To describe the association of untreated celiac disease with hyperhomocysteinemia and variants of the methylenetetrahydrofolic acid reductase (MTHFR) gene found in clinical practice.

STUDY: Case studies with description of associated clinical, biochemical, and genetic findings and review of literature.

RESULTS: Five new cases and 7 additional cases found from literature search of hyperhomocysteinemia with celiac sprue are reported. Treatment with gluten-free diet and folic acid led to the variable improvement in homocysteine levels. MTHFR gene variants were present in each of the new patients described.

CONCLUSIONS: Untreated celiac disease may be associated with hyperhomocysteinemia caused by a combination of vitamin deficiencies and variants in the MTHFR gene. Abnormalities do not consistently improve with gluten-free diet. The abnormal findings could result from vitamin deficiencies or variant MTHFR status. Possible clinical implications for patients with celiac disease and hyperhomocysteinemia are reviewed.