

The pathophysiology of MTHFR deficiency can be summarized as follows; 1. Impaired Methylation; The MTHFR enzyme plays a role, in converting folate into its active form called 5 methyltetrahydrofolate (5 MTHF). This active form is essential for the methylation process, which converts homocysteine to methionine. When there are gene mutations that reduce MTHFR activity the conversion of folate to 5 decreases resulting in levels of homocysteine and reduced production of methionine. 2. Elevated Homocysteine Levels; MTHFR deficiency is characterized by levels of homocysteine in the blood, known as hyperhomocysteinemia. Increased homocysteine levels can contribute to health problems such as an increased risk of disease, stroke and blood clot formation. 3. DNA Methylation; Reduced methylation capacity can affect gene expression and DNA repair processes. Changes in DNA methylation patterns may contribute to health conditions including birth defects, developmental issues and an increased susceptibility to types of cancer. 4. Neurotransmitter Synthesis; Methylation plays a role in the synthesis of neurotransmitters like serotonin, dopamine and norepinephrine. In cases of MTHFR deficiency there is a possibility that neurotransmitter levels may be affected which could potentially contribute to mood disorders such, as depression and anxiety.