

Methionine synthase, which is also called homocysteine methyltransferase, is responsible for the regeneration of methionine from homocysteine. A deficiency of methionine synthase can cause a buildup of homocysteine in the blood and urine.

Methylenetetrahydrofolate Reductase (MTHFR) Deficiency

[MoTher-FatheR Broken](#)

MTHFR is an enzyme in the folate cycle that catalyzes the conversion of 5,10-methylenetetrahydrofolate (THF) to 5-methyl THF, which is then used for pyrimidine synthesis. The folate cycle depends on homocysteine metabolism to function. MTHFR deficiency, therefore, leads to a build-up of homocysteine, or homocystinuria.

Signs and Symptoms

Marfanoid Body Habitus

[Marfan-the-martian Body](#)

Marfanoid habitus describes a constellation of physical findings, including tall stature, long limbs, spider-like fingers, and hyperlaxity of joints. Associated conditions include Marfan syndrome, MEN2B, homocystinuria, and spontaneous pneumothorax.

Kyphosis

[Curvy-K-spine](#)

Kyphosis is a condition of extreme curvature of the upper back. This pathologic curving of the spine can be seen in homocystinuria.

Lens Subluxation (Inferior)

[Camera-lens Sub](#)

Eye anomalies are common in homocystinuria, especially lens subluxation. Lens subluxation in homocystinuria is characterized by an inferiorly displaced or malpositioned lens in the eyes. Contrast this with Marfan's syndrome, where there is *superior* lens subluxation.

Intellectual Disability

[Tar Covered Book](#)

Intellectual Disability is characterized by significantly impaired cognitive functioning and deficits in two or more adaptive behaviors. It is common in individuals with homocystinuria.

Atherosclerosis

[Clogged Artery-guy](#)

An especially important cause of morbidity and mortality is vascular disease, including atherosclerosis. Patients exhibit extensive atheroma formation at a young age. Almost one-fourth of patients die before the age of 30 as a result of thrombotic complications.

Consideration

Dietary Deficiencies Can Elevate Homocysteine Levels

[Broken Nutritional-plate Up-arrow Homer](#)

While homocystinuria generally describes patients with a genetic disorder leading to elevated homocysteine levels, otherwise healthy patients could also have elevated levels in certain circumstances. For example, people with low dietary intake of folate, vitamin B6, or B12 can have increased levels of homocysteine. Though these patients don't have the genetic enzyme deficiencies described in classical homocystinuria, they may still be at elevated risk for atherosclerosis.