Condition: Homocystinuria

Inheritance:

Autosomal recessive.

Genetic etiology:

Mutation in *CBS* gene, encoding cystathionine beta-synthase.

Frequency:

1/200,000 – 1/300,000.

Clinical features:

Homocystinuria is a multisystem disorder, leading to myopia, lens dislocation, tall, slender body habitus, thromboembolism, and neurological impairment (developmental delay, seizures, movement disorder).

Management:

Some are responsive to pyridoxine treatment; treatment with betaine, folate, and vitamin B12; maintenance of methionine-restricted diet; supportive care.

Genetic counseling:

Based on autosomal recessive inheritance; molecular genetic testing available. Detectable by newborn screening, allowing early institution of dietary and medical management.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.