Homocystinuria

Homocystinuria is an autosomal recessive disorder of methionine metabolism. The most common cause of homocystinuria is a deficiency of the amino acid cystathionine *B*-synthase. Due to this deficiency, elevated levels of homocystine, methionine and their metabolites accumulate in the blood and urine of these patients. Newborns appear normal and early symptoms are vague. Usually by age 3, more specific symptoms appear. An increase in visual problems leads to a diagnosis of this condition when a child on examination is discovered to have dislocation of the lens of the eye and severe myopia. Several clinical findings of homocystinuria have features similar of Marfan's syndrome including dislocation of the lens, tall thin build with elongated arms and legs and scoliosis. In addition, blood clots tend to develop and become lodged in any large or small blood vessel potentially leading to a life-threatening crisis.

Prevalence:	1: 50,000- 1: 150,000
Analyte Measured:	Methionine
Reporting Ranges:	Elevated Methionine
Timing:	Elevations may be minimal during the first 3 days of life until adequate protein intake (milk feedings).
Confirmation	A diagnosis is made in the presence of an increased homocysteine and increased methionine.
Treatment:	Referral will be made to a Metabolic Specialist. Treatment is aimed at the underlying cause of homocystinuria. A methionine-restricted, cystine-supplemented diet is indicated. Anticoagulant therapy is indicated.
Comment:	Without proper treatment, 65%- 80% of patients have developmental delays and approximately 50% of patients die by the age of 25. Death is frequently associated with arterial or venous thromboses that involve the cerebral, pulmonary, renal, and myocardial circulation. However, treatment reduces the risk of thromboembolic episodes and the incidence of mental retardation and seizures are greatly reduced.

Maine Newborn Screening Program
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