KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: INCREASED METHIONINE

CONDITION: HOMOCYSTINURIA (CBS DEFICIENCY)

DIFFERENTIAL DIAGNOSIS: Classical homocystinuria (cystathionine β-syntase (CBS) deficiency); liver disease; hyperalimentation.

METABOLIC DESCRIPTION: Methionine from ingested protein is normally converted to homocysteine. In classical homocystinuria due to CBS deficiency, homocysteine cannot be converted to cystathionine. As a result, the concentration of homocysteine and its precursor, methionine, will become elevated.

ACTION TO BE TAKEN **IMMEDIATELY:**

- Contact family to inform them of the newborn screening result and ascertain clinical status.
- Consult with pediatric metabolic specialist.
- Evaluate the newborn with attention to liver disease and refer as appropriate.
- Initiate confirmatory/diagnostic tests in consultation with metabolic specialist.
- Educate family about homocystinuria and its management, as appropriate.
- Report findings to newborn screening program.

CONFIRMATION OF DIAGNOSIS: Quantitative plasma amino acids will show increased **homocystine** and **methionine** in classical homocystinuria but only increased methionine in other disorders. Plasma homocysteine analysis will show markedly increased homocysteine in classical Homocystinuria and normal or only slightly increased homocysteine in the other disorders. Urine homocysteine is markedly increased in classical homocystinuria.

CLINICAL EXPECTATIONS: Homocystinuria is usually asymptomatic in the neonate. If untreated, these children eventually develop intellectual disabilities, ectopia lentis, a marfanoid appearance including arachnodactyly, osteoporosis, other skeletal deformities and thromboembolism.

REPORTING: Report diagnostic result to family and Kansas NBS program.

SPECIALISTS:

Bryce Heese, MD
Biochemical Genetics
Children's Mercy Hospital- Kansas City, MO

Clinic phone: 816-234-3771
Hospital Operator: 816-234-3000
Office Fax: 816-302-9963