Homocystinuria Ages 1-18 years

Clinic Visits:

- ☑ At least every 6 months

Laboratory studies:

- ☑ Monitoring of diet
  - Plasma amino acid levels
    - 1-6 years: at least every 3 months
    - 6-18 years: at least every 6 months
  - Plasma total homocysteine
    - 1-6 years: at least every 3 months
    - 6-18 years: at least every 6 months
  - Targeted levels:
    - Homocysteine < 50 µmol/L
    - Methionine <1000 µmol/L
- ☑ Yearly:
  - Prealbumin/albumin
  - Ferritin, transferrin, or iron studies
  - Possible CBC, hemoglobin, and hematocrit
  - Possible Folate and vitamin B12
  - Possible nutritional labs (zinc, selenium, vitamin D, essential fatty acids, and lipid profile

During illness:

- ☑ Call Metabolic Clinic to alert them that your child is sick or needs surgery
- ☑ Emergency letter for thrombosis (blood clotting) risks with surgery and illness

Evaluations:

- ☑ Ophthalmology evaluation yearly
- ☑ Yearly developmental questionnaires to be completed by parents
- ☑ Developmental evaluation at 3 and 6 years old
- ☑ Neuropsychological evaluation at 9 years old
- ☑ Psychiatric screening at 18 years old
- ☑ Bone Health
  - DEXA of spine at 6, 9, 12, and 18 years old
- ☑ Possible orthopedic evaluation
- ☑ Possible neurology evaluation

Topics:

- ☑ Biochemistry and genetics of Homocystinuria
- ☑ Complications of Homocystinuria
  - Dislocated lens in the eye
  - Risk for thrombosis
- ☑ Diet/management
  - Methionine-restricted diet
  - Importance of medical formula

- Introduction of low protein food
- Possible pyridoxine if responsive (most individuals detected by NBS are not)
- Folate
- B12
- Betaine
- Possible L-cysteine of levels are low

Please make sure your child has clinic visits and levels done on time. Call the Metabolic clinic at _____ to make an appointment, schedule a blood draw, or with any other questions.

Thank You,
Your Metabolic Team