

Homocystinuria Ages 6-12 months

Clinic Visits:

- At least every 6 months

Laboratory studies:

- Monitoring of diet
 - Plasma amino acid levels at least monthly
 - Plasma total homocysteine at least monthly
 - 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

Topics:

- Biochemistry and genetics of Homocystinuria
- Complications of Homocystinuria
 - Dislocated lens in the eye
 - Risk for thrombosis (blood clotting)
- Diet/management
 - Methionine-restricted diet
 - Importance of medical formula
 - Introduction of low protein food
 - Possible pyridoxine if responsive (most babies 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

