

Homocystinuria >18 years

Clinic Visits:

- At least yearly

Laboratory studies:

- Monitoring of diet
 - Plasma amino acid levels at least yearly
 - Plasma total homocysteine at least yearly
 - 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 if you are sick or if you need surgery
- Emergency letter for thrombosis (blood clotting) risks with surgery and illness

Evaluations:

- Ophthalmology evaluation yearly
- DEXA spine scan every 5 years

Topics:

- Review of biochemistry and genetics of Homocystinuria
- Complications of Homocystinuria
 - **Risk for thrombosis!**
 - Dislocated lens in the eye
 - Risk of psychiatric problems
 - Thrombosis in pregnancy
 - Anticoagulation medications
- Diet/management
 - Methionine-restricted diet
 - Importance of medical formula
 - Low protein food
 - Possible pyridoxine if responsive (most individuals detected by NBS are not)
 - Folate
 - B12
 - Betaine
 - Possible L-cysteine if levels are low
 - Possible aspirin therapy to reduce risk of blood clots

Please make sure you have 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