Homocystinuria Ages 0-6 months

Clinic Visits:

- ☑ Every 2 months

Laboratory studies:

- ☑ At diagnosis
  - o Plasma amino acids
  - o Urine organic acids
  - o Plasma total homocysteine
  - o Possible MTHFR and factor V (coagulation disorders) testing
  - o Possible skin biopsy (enzymatic studies)
  - o Possible DNA analysis of cystathionine β-synthase (CBS) gene
- ☑ Monitoring of diet
  - o Plasma amino acid levels at least every 2 weeks
  - o Plasma total homocysteine at least every two weeks
  - o Targeted levels:
    - ▪ Homocysteine < 50 µmol/L
    - ▪ Methionine <1000 µmol/L

During illness:

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

Topics:

- ☑ Biochemistry and genetics of Homocystinuria
- ☑ Complications of Homocystinuria
  - o Dislocated lens in the eye
  - o Risk for thrombosis (blood clotting)
- ☑ Diet/management
  - o Methionine-restricted diet
  - o Importance of medical formula
  - o Possible pyridoxine if responsive (most babies detected by NBS are not)
  - o Folate
  - o B12
  - o Betaine
  - o Possible L-cysteine if 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