

Citrullinemia

Ages 0-6 months

Clinic Visits:

- Every 2 months

Laboratory studies:

- At diagnosis
 - Plasma amino acids
 - Ammonia
 - Comprehensive metabolic panel
 - Possible urine organic acids
 - Possible urine orotic acids
 - Possible PT/PTT (clotting)
 - Possible blood gases
 - Possible skin biopsy (enzymatic studies for ASS enzyme)
 - Possible DNA analysis of ASS gene
- Monitoring of diet
 - Plasma amino acid levels at least every 2 weeks
- Clinic visits
 - Comprehensive metabolic panel with liver function test
 - Possible ammonia, PT, and prealbumin

During illness:

- Call the metabolic clinic to alert them immediately when your baby is sick
 - Poor feeding, fever, vomiting, and/or lethargy
- Give your baby glucose gel or polymer solution if needed (or instructed to do so)
- If your baby is not able to keep down glucose containing fluids, you will need to take him or her to the emergency room for IV 10% dextrose (glucose)
- Be sure to bring Emergency Room letter with you to the hospital
- Labs during illness include: comprehensive metabolic panel, ammonia, and plasma amino acids
- Medication or dialysis may be used to lower your baby's ammonia levels
- Please alert the metabolic clinic if your baby needs surgery

Evaluations:

- Head CT scan or brain MRI at time of diagnosis if there are concerns of neurological problems
- Possible referral to Liver, Renal (kidney), or neurology if needed

Topics:

- Biochemistry and genetics of Citrullinemia
- Complications
 - Elevated ammonia levels
- Diet/management
 - Low protein diet

- Importance of medical formula
- Medications to control ammonia levels
- Possible liver transplant
- Avoid fasting
- Management during illness

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. If you urgently need to reach a Metabolic physician, call _____ and ask to page the Metabolic physician on call.

Thank You,
Your Metabolic Team

