Carnitine-Acylcarnitine Translocase (CACT) Deficiency
Ages 0-6 months

Clinic Visits:

- ☑ Every 2 months

Laboratory studies:

- ☑ At diagnosis
  - o Acylcarnitine profile
  - o DNA analysis of CACT gene
  - o Rule out CPTII with DNA analysis
  - o Carnitine levels
  - o Comprehensive metabolic panel
  - o Blood ammonia
  - o Serum CK (muscle health)
  - o Possible urine organic acids
- ☑ Monitoring of diet
  - o Possible acylcarnitine profile
- ☑ At clinic visits:
  - o Carnitine levels
  - o Serum CK
  - o Liver function tests
  - o Possible acylcarnitine profile

During illness:

- ☑ Call Metabolic Clinic to alert them that your baby is sick
  - o 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, serum CK, and B-type natriuretic peptide (heart health)
- ☑ Please alert the Metabolic Clinic if your baby needs surgery
- ☑ **Consider medic alert bracelet!**

Evaluations:

- ☑ Cardiology evaluation at time of diagnosis
- ☑ Abdominal ultrasound
- ☑ Head CT scan

Topics:

- ☑ Biochemistry and genetics of carnitine-acylcarnitine translocase deficiency
- ☑ Diet/management
  - o Avoid fasting

- Management during illness
- Low fat diet
- Medium chain triglyceride (MCT) supplementation (formula or oil)
- Carnitine supplementation
- Cornstarch if needed

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