Carnitine-Acylcarnitine Translocase (CACT) Deficiency
Ages 6-24 months

Clinic Visits:

- ☑ Every 3 months

Laboratory studies:

- ☑ Monitoring of diet
  - Possible acylcarnitine profile
- ☑ At clinic visits:
  - Carnitine levels
  - Serum CK (muscle health)
  - Liver function tests
  - Possible acylcarnitine profile
- ☑ Yearly
  - Comprehensive metabolic panel
  - Acylcarnitine profile
  - B-type natriuretic peptide (heart health)
  - Fatty acid profile at 6 months and then yearly

During illness:

- ☑ Call Metabolic Clinic to alert them that your child is sick
  - Poor feeding, fever, vomiting, and/or lethargy
- ☑ Give your child glucose gel or polymer solution if needed (or instructed to do so)
- ☑ If your child 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
- ☑ Please alert the Metabolic Clinic if your child needs surgery
- ☑ **Consider medic alert bracelet!**

Evaluations:

- ☑ Cardiology evaluation every 2 years
- ☑ Yearly developmental questionnaires (to be completed by parents)

Topics:

- ☑ Biochemistry and genetics of carnitine-acylcarnitine translocase deficiency
- ☑ Diet/management
  - Avoid fasting
  - Management during illness
  - Low fat diet
  - 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