Carnitine Palmitoyltransferase I (CPT I) Deficiency
Ages 0-4 years

Clinic Visits:

- ☑ At least every 6 months

Laboratory studies:

- ☑ At diagnosis
  - o Acylcarnitine profile
  - o Carnitine levels
  - o Comprehensive metabolic panel
  - o Possible DNA analysis of the CPT I gene
  - o Possible skin biopsy (enzymatic studies)
- ☑ At clinic visits:
  - o Comprehensive metabolic panel
- ☑ Yearly
  - o Comprehensive metabolic panel
  - o Triglycerides
  - o Urine analysis
  - o Phosphate (blood and urine)
  - o Urine creatinine

During illness:

- ☑ Call the Metabolic Clinic to alert them immediately if your child is sick
  - o Poor feeding, 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 phosphate, and serum CK (muscle health)
- ☑ Please alert the Metabolic Clinic if your child needs surgery
- ☑ **Consider Medic Alert Bracelet**

Evaluations:

- ☑ Yearly developmental questionnaires to be completed by parents
- ☑ Developmental evaluation at age 3 years

Topics:

- ☑ Biochemistry and genetics of carnitine palmitoyltransferase I deficiency
- ☑ Diet/management
  - o Avoid fasting
  - o Management during illness
  - o Possible medium chain triglyceride (MCT) oil or formula

Please make sure your child has clinic visits and laboratory studies 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