

Carnitine Palmitoyltransferase II (CPT II) Deficiency

Ages 6-24 months

Clinic Visits:

- Every 6 months

Laboratory studies:

- At clinic visits:
 - Carnitine levels
 - Serum CK (muscle health)
 - Liver function studies
 - Possible acylcarnitine profile (diet management)
- Yearly
 - Comprehensive metabolic panel
 - Acylcarnitine profile
 - B-type natriurectic peptide (heart health)
 - Fatty acid profile at 6 months then yearly

During illness:

- Call Metabolic Clinic to alert them if your child is sick
 - 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 CK, acylcarnitine profile and B-type natriurectic peptide
- IV carnitine may be given
- Possible cardiac monitoring when in emergency room
- Possible abdominal ultrasound
- Please alert the Metabolic Clinic if your child needs surgery
- Consider medic alert bracelet**

Evaluations:

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

Topics:

- Review of biochemistry and genetics of CPT II deficiency
- Diet/management
 - Avoid fasting
 - Management during illness
 - Possible continuous feeds during the night
 - Low-fat diet with limited long-chain triglycerides
 - Medium chain triglyceride (MCT) oil
 - Importance of medical formula

- Carnitine
- Cornstarch if needed

Please make sure your child has clinic visits and laboratory studies are 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

