

Carnitine Palmitoyltransferase II (CPT II) Deficiency

Ages 0-6 months

Clinic Visits:

- At least every 2 months

Laboratory studies:

- At diagnosis
 - Acylcarnitine profile
 - Carnitine levels
 - Comprehensive metabolic panel
 - Blood ammonia
 - Serum CK (muscle health)
 - B-type natriuretic peptide (heart health)
 - Possible DNA analysis of the CPT II gene
 - Possible skin biopsy for CPT II enzyme studies
 - Possible urine organic acid
- Clinic visits
 - Serum CK
 - Carnitine panel
 - Liver function tests
 - Possible acylcarnitine profile

During illness:

- Call Metabolic Clinic to alert them that your baby is sick
 - Poor feeding, vomiting, and/or lethargy
- Give your baby 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 levels, serum CK, acylcarnitine profile, and B-type natriuretic peptide
- IV carnitine may be given
- Possible cardiac monitoring when in emergency room
- Possible abdominal ultrasound
- Please alert the Metabolic Clinic if your baby needs surgery
- Consider medic alert bracelet**

Evaluations:

- Cardiology exam at time of diagnosis
- Abdominal ultrasound
- Head CT scan

Topics:

- Biochemistry and genetics of Carnitine Palmitoyltransferase 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
- Essential fatty acid supplementation
- Carnitine

Please make sure your baby 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