

# Carnitine Palmitoyltransferase II (CPT II) Deficiency

## Ages 2-18 years

### 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

### 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
- Wear a medic alert bracelet!**

### Evaluations:

- Cardiology evaluation every 2 years (possible halter monitor)
- Yearly developmental questionnaires to be completed by parents
- Developmental evaluation at 3 and 6 years
- Neuropsychological evaluation at 9 years

### 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