

# Carnitine Transporter Deficiency (Carnitine Uptake Deficiency) Ages 4-18 years

## Clinic Visits:

- At least yearly

## Laboratory studies:

- At clinic visits:
  - Carnitine levels

## During illness:

- Call the Metabolic Clinic to alert them immediately 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 (muscle health) and B-type Natriuretic Peptide (heart health)
- Please alert the Metabolic Clinic if your child needs surgery
- Consider Medic Alert Bracelet**

## Evaluations:

- Cardiology evaluation every 2 years (with possible halter monitoring)
- Yearly developmental questionnaires to be completed by parents
- Developmental evaluation at age 6 years
- Neuropsychology evaluation at 9 years

## Topics:

- Biochemistry and genetics of Carnitine Transporter Deficiency
- Diet/management
  - Avoid fasting
  - Carnitine supplementation
  - Management during illness

*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