

Carnitine-Acylcarnitine Translocase (CACT) Deficiency

Ages 24+ months

Clinic Visits:

- Every 6 months

Laboratory studies:

- Monitoring of diet
 - Possible acylcarnitine profile
- At clinic visits:
 - Carnitine levels
 - Serum CK (muscle health)
 - Liver function tests
 - Possible acylcarnitine profile
- Yearly
 - Comprehensive metabolic panel
 - Acylcarnitine profile
 - B-type natriuretic peptide (heart health)
 - Fatty acid profile

During illness:

- Call Metabolic Clinic to alert them that your child is sick
 - Poor feeding, fever, 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, and B-type natriuretic peptide
- Please alert the Metabolic Clinic if your child needs surgery
- Consider medic alert bracelet!**

Evaluations:

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

Topics:

- Biochemistry and genetics of CACT
- Diet/management
 - Avoid fasting
 - Management during illness
 - Low fat diet
 - MCT supplementation (formula or oil)

- Carnitine supplementation
- Cornstarch if needed

Please make sure your child 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