Medium Chain Acyl CoA Dehydrogenase Deficiency
Ages 0-4 years

Clinic Visits:

- ☑ At least every 6 months

Laboratory studies:

- ☑ At diagnosis
  - o Acylcarnitine profile
  - o Carnitine panel
  - o Possible urine organic acids
  - o Possible urine acylgylcine profile
  - o Possible DNA analysis of MCAD gene
  - o Possible skin biopsy (fatty acid oxidation studies)
- ☑ Possible carnitine levels at clinic visit

During illness:

- ☑ Call Metabolic Clinic to alert them that your child is sick
  - o Poor feeding, vomiting, 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, possible ammonia, possible uric acid, possible urine analysis, and possible serum CK.
- ☑ Consider medic alert bracelet

Evaluations:

- ☑ Yearly developmental questionnaires to be completed by parents
- ☑ Developmental evaluation at age 3

Topics:

- ☑ Biochemistry and genetics of Medium Chain Acyl-CoA Dehydrogenase Deficiency
- ☑ Diet/management
  - o Avoid fasting
  - o Management during illness
  - o Carnitine supplementation
  - o Cornstarch after age 1 year if needed
  - o Heart healthy diet (lower fat)

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