



MEDIUM and SHORT-CHAIN L-3 HYDROXY ACYL-CoA DEHYDROGENASE DEFICIENCY (M/SCHAD)

FAMILY FACT SHEET

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 36 hours after birth. The blood is tested for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called medium and short-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (M/SCHAD).

A positive newborn screen does not mean your baby has M/SCHAD, but it does mean your baby needs more testing to know for sure.

You will be notified by your primary care provider or the newborn screening program to arrange for additional testing.

What is M/SCHAD?

M/SCHAD affects a special enzyme needed to break down fats in the food we eat, so they can be used for energy and growth. In M/SCHAD, the enzyme used to break down fats is missing or not working properly.

A person who has M/SCHAD doesn't have enough enzyme to break down fat into energy. Using stored fat for energy is especially important in between meals when the body is not getting new energy from eating food.

M/SCHAD are genetic disorders that are passed on (inherited) from parents to a child. The mother and father of an affected child carry a gene change that can cause M/SCHAD. Parents usually do not have signs or symptoms, or even know they carry the gene change.

What problems can M/SCHAD cause?

M/SCHAD is different for each child. Some children with M/SCHAD have few health problems, while other children have a more severe form of M/SCHAD with serious complications.

If M/SCHAD is not treated, a child might develop:

- Low blood sugar
- Feeding problems
- Diarrhea/vomiting
- Muscle weakness
- Liver problems

It is very important to follow the doctor's instructions for testing and treatment.

What is the treatment for M/SCHAD?

M/SCHAD can be treated. Treatment is life-long and can include:

- Frequent meals/snacks and a low fat/high carbohydrate diet – a dietitian will help you learn what foods can be eaten.
- Medications to help the body make energy.
- Special approaches to routine illnesses.

Children with M/SCHAD should see their regular doctor, a doctor who specializes in M/SCHAD, and a dietitian.

Prompt and careful treatment helps children with M/SCHAD live the healthiest lives possible.

Michigan Resources and Support

Michigan Newborn Screening

Nurse Consultant

Toll-free: 1-866-673-9939

newbornscreening@michigan.gov

Michigan Metabolic Coordinating Centers

Children's Hospital of Michigan: 1-313-832-9343

Michigan Medicine: 1-734-764-0579

Children's Special Health Care Services

Toll-free: 1-800-359-3722