

## Methylmalonic Acidemia (MMA)

(methel-ma-lawn-ic acid-ee-me-ah)

### What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called MMA (em-em-ay). There are several other conditions that can cause a similar positive result on newborn screening.

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

Your baby's doctor will help arrange for more testing by specialists in disorders like MMA.

### What is methylmalonic acidemia?

MMA affects enzymes needed to break down proteins and fats from the food we eat so they can be used for energy and growth. There are different kinds of MMA, depending on which enzyme is missing or not working properly.

A person with MMA doesn't have enough enzyme to break down protein or fats, so harmful toxins can build up in the body and cause health problems.

MMA is a disorder that is passed on, or inherited, from a child's mother and father. Because MMA is a genetic disease, family members are at risk of having MMA too, even if no one in the family has had it before.

### What problems can methylmalonic acidemia cause?

MMA is different for each child. Some children with MMA have fewer health problems, while other children have very serious complications.

If MMA is not treated, a child might develop:

- Sleepiness
- Feeding problems
- Muscle weakness
- Seizures
- Stroke
- Poor growth
- Kidney disease
- Mental retardation

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

### What is the treatment for methylmalonic acidemia?

MMA can be treated. The treatment is life-long. Treatment for children with MMA can include:

- Medications to help the body make energy
- Vitamin B<sub>12</sub>
- Low-protein diet - a dietician helps families set up the best diet for their child

Children with MMA should see their regular doctor, a doctor who specializes in MMA, and a dietician.

Children with MMA can benefit from prompt and careful treatment.

### Resources for parents

#### Genetics Home Reference

<http://ghr.nlm.nih.gov>

#### Organic Acidemia Foundation

[www.oaanswers.org](http://www.oaanswers.org)

#### MN Children & Youth with Special Health Needs

(651) 201-3650 or (800) 728-5420

#### MN Newborn Screening Program

[www.health.state.mn.us/newbornscreening](http://www.health.state.mn.us/newbornscreening)