



2-Methylbutyryl-CoA Dehydrogenase Deficiency (2-MBG)

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 2-MBG.

A positive newborn screen does not mean your baby has 2-MBG, 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 2-MBG?

2-MBG affects an enzyme needed to break down proteins from the food we eat. In 2-MBG the enzyme used to break down proteins is missing or not working properly.

A person who has 2-MBG doesn't have enough enzyme to break down protein containing isoleucine, so harmful toxins can build up in the body.

2-MBG is a genetic disorder that is passed on (inherited) from parents to a child. The mother and father of an affected child carry a gene change that can cause 2-MBG. Parents usually do not have signs or symptoms, or even know they carry the gene change.

What problems can 2-MBG cause?

2-MBG is different for each child. Some children with 2-MBG have few health problems, while others may have serious complications.

2-MBG is most prevalent in newborns of Hmong ancestry.

If 2-MBG is not treated a child might develop:

- Sleepiness
- Low blood sugar
- Developmental delay

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

What is the treatment for 2-MBG?

2-MBG can be treated. Treatment is life-long and can include:

- Diet low in isoleucine, a dietician will help you with the best diet for your child.
- Medications to help the body get rid of harmful toxins.

Children with 2-MBG should see their regular doctor, a doctor who specializes in 2-MBG, and a dietician.

Prompt and careful treatment helps children with 2-MBG live the healthiest lives possible.

Michigan Resources and Support

Michigan Newborn Screening
Nurse Consultant
Toll-free: 1-866-673-9939
newbornscreening@michigan.gov

Children's Hospital of Michigan Metabolic Clinic
Toll-free: 1-866-442-4662

Children's Special Health Care Services
Toll-free: 1-800-359-3722