GUANIDINOACETATE METHYLTRANSFERASE DEFICIENCY (GAMT)

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 GAMT deficiency.

A positive newborn screen does not mean your baby has GAMT deficiency, 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 GAMT deficiency?

GAMT deficiency is a disease that can cause problems making creatinine in our body. Creatinine is a substance the body needs to store and produce energy, mostly for the brain and muscles. About half of our creatinine comes from food, mainly meat and dairy products. The other half is produced in our kidney and liver.

A person with GAMT deficiency does not have enough enzyme to break down a substance called guanidinoacetate (GAA) into creatinine. This results in the build-up of GAA, which can be harmful, and a lack of creatinine.

GAMT deficiency 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 GAMT deficiency. Parents usually do not have signs or symptoms, or even know they carry the gene change.

What problems can GAMT deficiency cause?

GAMT deficiency is different for each child. Some children with GAMT deficiency only have a few health problems, while other children may have very serious complications.

If GAMT deficiency is not treated, a child might develop:

- Intellectual disability, mild to severe
- Language delay
- Epilepsy or seizures ranging from occasional to severe and not responsive to anti-seizure medications
- Movement disorders involving uncontrollable jerking or slowness and delays in crawling, sitting and walking
- Behavior disorders involving hyperactivity, autism or self-injury

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

What is the treatment for GAMT deficiency?

GAMT deficiency is a treatable condition. Oral supplements of creatinine and ornithine (an important part of protein), and a protein-restricted diet are often prescribed. Early diagnosis and treatment map prevent development of intellectual disability and other symptoms of GAMT deficiency.

Children with GAMT deficiency should see their regular doctor, a doctor who specializes in GAMT deficiency, and a dietitian. Prompt and careful treatment helps children with GAMT deficiency 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 Center
C.S. Mott Children's Hospital, Michigan Medicine:
1-734-764-0579

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

MDHHS Michigan Department of Health & Human Services | Michigan Newborn Screening

Phone 1-866-673-9939
www.michigan.gov/newbornscreening

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