

Positive Result:

Blood Spot Screen Result Notification



Elevated GUAC: Risk for GAMT Deficiency

What was found on the newborn screen?

The newborn screen collected at birth found your baby has high levels of guanidinoacetate (abbreviated as GUAC) compared to baby's level of creatine.

What does this mean?

High GUAC can indicate a baby has a condition called guanidinoacetate methyltransferase (GAMT) deficiency. A positive newborn screen does not mean your baby has GAMT deficiency for sure, but evaluation and more testing is needed.

What happens next?

Your baby will be referred to a specialist familiar with GAMT deficiency. They will help arrange further testing, which will give more information about if your baby has GAMT deficiency.

What causes GAMT deficiency?

People with GAMT deficiency are unable to break down GUAC into creatine, which is used by our brain and muscles to store energy. This results in GUAC building up in the body, which can be toxic. In addition, without enough creatine, the brain and muscles cannot work properly.

What health problems can it cause?

GAMT deficiency is different for each child. It is a lifelong condition that can result in serious brain and muscle problems if untreated.

GAMT deficiency symptoms typically begin before age one, but can start as late as age three. Babies are usually late sitting, crawling, walking, and speaking. They may also grow more slowly than others their age. If untreated, most children develop learning and behavioral challenges, as well as seizures.

What treatment options are available?

GAMT deficiency cannot be cured, but lifelong treatment prevents and reduces symptoms. This is especially true for children who start treatment before GAMT deficiency symptoms begin.

The most common treatments for GAMT deficiency are medications taken by mouth and a special diet.

Children with GAMT deficiency should see their regular doctor and a metabolic specialist.

Resources



Scan here for more information about newborn screening in Minnesota or visit our website:
www.health.state.mn.us/people/newbornscreening

MedlinePlus:

www.medlineplus.gov/genetics

Save Babies Through Screening Foundation:

www.savebabies.org

Newborn Screening Info Center:

www.newbornscreening.hrsa.gov