Newborn Screening FACT Sheet

Guanidinoacetate Methyltransferase (GAMT) Deficiency

What is GAMT Deficiency?
Guanidinoacetate methyltransferase (GAMT) deficiency is an inherited condition that affects the body's ability to produce creatine. Without an adequate supply of creatine, the body is unable to use and store energy properly. This can cause severe neurological problems such as intellectual disability, limited speech development, recurrent seizures, behavioral problems, and involuntary movements. Fortunately, early diagnosis and treatment can lead to improved health and development in children affected by GAMT deficiency.

What causes GAMT Deficiency?
Guanidinoacetate methyltransferase deficiency is caused by changes (mutations) in the *GAMT* gene. This gene provides instructions for making an enzyme that helps the body produce creatine from a compound called guanidinoacetate. Mutations in the *GAMT* gene cause lower levels of functional enzyme which impairs the body's ability to create creatine. Because creatine is needed for the body to store and use energy, reduced levels may prevent certain organs and tissues from working properly.

It is an inherited in an autosomal recessive pattern, meaning both parents are genetic carriers of the disease but do not have the disease itself.

What symptoms occur with GAMT deficiency?
The effects of GAMT deficiency are most severe in organs and tissues that require large amounts of energy, such as the brain and muscles.

Children affected by GAMT deficiency can develop symptoms anytime from infancy (age 3-6 months) to age three years.

Early signs of GAMT deficiency may include:

- Developmental delay
- Poor muscle tone
- Seizures
- Involuntary movements
- Respiratory insufficiency.

What to expect with GAMT Deficiency care:
People affected by guanidinoacetate methyltransferase (GAMT) deficiency require lifelong treatment with creatine monohydrate and ornithine supplements.

The goal of creatine monohydrate supplementation is to ensure that all parts of the body (especially those that require large amounts of energy, such as the brain) have an adequate supply of creatine to function properly.

Ornithine supplementation may be prescribed to reduce the high levels of guanidinoacetate that accumulate in the blood and tissues. This treatment is important because elevated levels of guanidinoacetate can be toxic to the nervous system.

Dietary treatment
With the guidance of a metabolic specialist, people with GAMT deficiency may follow a natural-protein restricted diet. This special diet, in combination with ornithine supplementation, can help prevent the accumulation of guanidinoacetate in the nervous system.

Medical foods and formulas may be recommended to ensure that affected children have the nutrients necessary for proper growth and development.

Disclaimer: This information is adapted from American College of Medical Genetics website ACT sheets.

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Things to remember

Guanidinoacetate methyltransferase (GAMT) deficiency is a rare condition so there is limited information regarding the long-term outlook for affected people. However, studies suggest that children who are diagnosed early and receive treatment before they develop symptoms often remain healthy with normal development.

Resources:

MedlinePlus
https://medlineplus.gov/genetics/condition/guanidinoacetate-methyltransferase-deficiency/
Creatine Deficiency Disorders Gene Reviews
https://www.ncbi.nlm.nih.gov/books/NBK3794/
NIH
https://rarediseases.info.nih.gov/diseases/2578/guanidinoacetate-methyltransferase-deficiency
HRSA Newborn Screening Clearinghouse
https://newbornscreening.hrsa.gov/conditions/guanidinoacetate-methyltransferase-deficiency
Navigate Life Texas
https://www.navigatelifetexas.org/en/diagnosis-healthcare/children-with-multiple-disabilities-rare-conditions-undiagnosed

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TEXAS Health and Human Services | Texas Department of State Health Services

Where do I go for more information?

Use your phone's camera to scan the QR code.

Next steps after a positive newborn screening for GAMT Deficiency

Newborn screening is a blood test performed on every baby that identifies those at risk for rare disorders. Your baby's results show increased guanidinoacetate. This means there is a chance that your baby may have a condition called Guanidinoacetate methyltransferase (GAMT) deficiency. More testing is needed <u>as soon as possible</u> to see if your child has GAMT deficiency.

What happens next?

- It is important to have the recommended laboratory tests drawn. These results will help confirm or rule out the diagnosis.
- Your baby's doctor will help arrange a visit to a specialist clinic that is familiar with GAMT deficiency. The specialist will check your baby's health and discuss the test results in more detail at the visit. The team may also include a genetic counselor because this is a genetic disease. It is important to keep this appointment. A quick diagnosis and early treatment may lessen the impact of the disease.

What is GAMT deficiency?

GAMT deficiency is an inherited condition that affects the body's ability to produce creatine. Without an adequate supply of creatine, the body is unable to use and store energy properly. This can cause severe neurological problems.

What are the symptoms of GAMT deficiency?

Children affected by GAMT deficiency can develop symptoms anytime from infancy (age 3-6 months) to age three years. Early signs of GAMT deficiency may include:

- Developmental delay
- Poor muscle tone
- Seizures
- Involuntary movements

What is the treatment?

GAMT deficiency is a treatable condition. Oral supplements of creatine and ornithine and a protein-restricted diet are often prescribed. Prompt and uninterrupted treatment helps children with GAMT deficiency live the healthiest lives possible.

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