

Family action sheet for guanidinoacetate methyltransferase deficiency (GAMT)

If a newborn blood screening result shows that your baby could have a serious condition called guanidinoacetate methyltransferase deficiency (GAMT). This result requires further testing.

All babies born in California have a routine blood screen shortly after birth. The goal of this newborn screening is to find those at risk for serious medical conditions. Babies can look healthy at birth and still have one of these conditions. Babies with these conditions benefit from early diagnosis and treatment.

What is guanidinoacetate methyltransferase deficiency or GAMT?

GAMT is a condition in which the body builds up a toxic chemical called guanidinoacetate (GUAC) and cannot make creatine. Without creatine, the body cannot store and use energy properly. This leads to brain, muscle, speech and behavior issues, and developmental delays. Symptoms may be seen anytime between 3 months to 3 years of age.

GAMT is an inherited condition. This means it is passed from parents to children.

Is there treatment for GAMT?

Yes, there are treatments for GAMT. Early identification and treatment can help improve the health and development of babies with GAMT. Treatments include supplements and medications. Other supportive care can include nutritional, speech, occupational, and behavioral therapies.

Newborn screening takes blood from a newborn's heel to screen for medical conditions



Next steps

- 1. Your baby's medical provider will refer you to specialists with GAMT experience.** The specialists will arrange for further testing and evaluation. More tests are needed to find what treatment or monitoring will be needed for your baby.
- 2. Work closely with your baby's medical provider and specialists and follow their recommendations.** It is important that you keep all appointments and start treatment if instructed.

Where can I get more information?

Your baby's medical provider and specialists are the best people to teach you about GAMT. Find more information at these websites:

- **Baby's First Test**
(<https://www.babysfirsttest.org>). Type "GAMT" into the "Find a Condition" Box. Follow the link. For Spanish, click on the top right black box marked "en Español."
- **Health Resources & Services Administration (HRSA)**
(<http://newbornscreening.hrsa.gov/conditions/guanidinoacetate-methyltransferase-deficiency>)

