TEXAS Department of State Health Services

Newborn Screening FACT Sheet

3-Methylglutaconic Aciduria
(3MGA)

What is 3MGA?
3-methylglutaconic aciduria (3MGA) is the name for a group of five different conditions. All of these conditions affect the body's mitochondria, the part of a human cell that produces energy. The causes, symptoms, and treatment of the five different types of 3MGA vary. However, all types of 3MGA are classified as organic acid conditions because they can result in a build-up of harmful amounts of organic acids and toxins in the body.

What Causes 3MGA?
When we eat food, enzymes help break it down. Some enzymes help break down proteins into their building blocks, called amino acids. Other enzymes break down the amino acids.

TYPE I
The enzyme 3-methylglutaconyl-CoA hydratase is an enzyme that helps break down the amino acid leucine. When your baby has 3MGA type I, his or her body does not make enough or makes non-working 3-methylglutaconyl-CoA hydratase. When this happens, your baby's body cannot break down leucine. This causes a build-up of harmful substances in the body.

Type II (Barth Syndrome)
When your baby has 3MGA type II, his or her body is not making enough of a protein called tafazzin. Tafazzin helps balance the levels of a type of fat called cardiolipin in cells. Cardiolipin helps cells make energy. When your baby's body does not have tafazzin, the cells have trouble making energy.

Type III
When your baby has 3MGA type III, his or her body does not make enough of a protein known as the OPA3 protein. We do not yet know what the role of this protein is in the body.

Type IV
Currently, we do not understand what causes 3MGA type IV.

Type V
When your baby has 3MGA type V, his or her body does not make enough of a protein known as the DNAJC19 protein. Currently, we do not know what this protein does, but some researchers think that it might help make, move, and break down other proteins in the cells.

All types of 3MGA are genetic conditions, but they are not all passed down in the same way. Types I, III, and V are autosomal recessive genetic conditions. This means that a child must inherit one copy from each parent of the non-working gene for that type of 3MGA in order to have the condition. The parents of a child with an autosomal recessive condition each carry one copy of the non-working gene, but they typically do not show signs and symptoms of the condition. While having a child with 3MGA is rare, when both parents are carriers, they can have more than one child with the condition. Learn more about autosomal recessive inheritance at **www.babysfirsttest.org/genetics.**

Type II is an X-linked recessive genetic condition. This means that a male must inherit one copy of the non-working gene from his mother to have the condition. A female must inherit two copies of the non-working gene, one from each parent, in order to have the condition. In X-linked conditions, the gene is carried on the X sex chromosome, and the condition affects males more than females. While having a child with 3MGA is rare, when one or both parents carry the non-working gene for 3MGA type II, they can have more than one child with the condition. Learn more about X-linked recessive inheritance at **www.babysfirsttest.org/genetics.**

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What Symptoms or Problems Occur with 3MGA?
[Symptoms are something out of the ordinary that a parent notices.]

The signs of 3-methylglutaconic aciduria (3MGA) differ depending on type, but they all begin in utero or at birth.

3MGA type I signs include:
- delays in reaching developmental milestones (such as speech or motor skills)
- irregular, uncontrolled muscle movements (called dystonia)
- muscle spasms and weakness in the arms and legs (called spastic quadriparesis)

3MGA type II (Barth syndrome) signs include:
- frequent infections
- weak muscles
- delayed growth
- heart problems

3MGA type III (Costeff optic atrophy syndrome) signs include:
- vision loss
- trouble with balance
- weak muscle tone (called hypotonia)
- involuntary and irregular movements

3MGA type IV signs vary from individual to individual. Type IV has signs similar to types I, II, and III.

3MGA type V (dilated cardiomyopathy with ataxia) signs include:
- difficulty coordinating voluntary muscle movements (called ataxia)
- delayed growth
- undescended testes or an opening in the urethra on the underside of the penis

What is the Treatment for 3MGA?
The best way to care for your baby is to monitor your baby's heart for any cardiac complications by regularly visiting a cardiologist and metabolic specialist. A cardiologist can identify changes in your baby's heart and decide if any additional treatments are necessary. Each baby with 3MGA experiences unique signs and symptoms, so it is important to talk to your baby's doctor to decide which treatment is right for your baby.

Other than following up with a cardiologist as necessary, there are no specific treatments available for 3MGA.

Things to Remember
Work with your baby's doctor to determine the next steps for your baby's care. Your child's health care provider will help you coordinate care with other medical resources in the community. Care depends on what type of 3MGA your child has and on your child's symptoms.

Some children will need specialized care from a cardiologist, a metabolic specialist, a dietician, and/or an optometrist. Some children with 3MGA have developmental delays.

If you think that your baby is not meeting his or her developmental milestones, ask your child's health care provider about the next steps in accessing a developmental evaluation and care.