



3MCC Deficiency (3-Methylcrotonyl CoA Carboxylase Deficiency)

What is it?

3MCC deficiency is one type of organic acid disorder. People with this condition have problems breaking down an amino acid called leucine from the food they eat.

What causes it?

In order for the body to use protein from the food we eat, it is broken down into smaller parts called amino acids. Special enzymes then make changes to the amino acids so the body can use them.

3MCC deficiency occurs when an enzyme, called 3-methylcrotonyl CoA carboxylase (3MCC), is either missing or not working properly. This enzyme's job is to help break down leucine. When a child with 3MCC deficiency eats food containing leucine, harmful substances may build up in the blood and cause problems. Leucine is found in all foods with protein.

If 3MCC deficiency is not treated, what problems occur?

Each child with 3MCC deficiency may have somewhat different effects. In fact, some children with this condition never have symptoms and may not ever need treatment.

Babies with 3MCC deficiency are healthy at birth. If symptoms occur, they often start after 3 months of age. Some babies do not have their first symptoms until 6 months to 3 years of age. Others do not have symptoms until adulthood. Some people will never develop symptoms.

3MCC deficiency can cause episodes of illness called metabolic crises. Some of the first symptoms of a metabolic crisis are:

- 1) Poor appetite.
- 2) Extreme sleepiness or lack of energy.
- 3) Behavior changes.
- 4) Irritable mood.
- 5) Muscle weakness.
- 6) Nausea.
- 7) Vomiting.

Common blood and urine findings are:

- 1) Low blood sugar, called hypoglycemia.
- 2) Increased levels of acidic substances in the blood, called metabolic acidosis.
- 3) High levels of ammonia in the blood.
- 4) Low levels of carnitine in the blood.
- 5) Increased ketones in the urine.
- 6) Liver problems.

If a metabolic crisis is not treated, a child with 3MCC can develop:

- 1) Breathing problems.
- 2) Seizures.
- 3) Liver failure.
- 4) Coma, sometimes leading to death.

If a metabolic crisis is not treated, it could result in death. In surviving babies and children, repeated episodes of metabolic crisis can cause brain damage. This can lead to life-long learning problems or mental retardation.

Episodes of metabolic crisis can be triggered by:

- 1) Illness or infection.
- 2) Going without food for long periods of time.
- 3) Eating large amounts of protein.

When a child is ill or goes without food for too long the body breaks down its own protein and fat to use for energy. In some people with 3MCC deficiency, this can trigger a metabolic crisis.

Between episodes of metabolic crisis, children with 3MCC deficiency are usually healthy.

Some children do not ever have metabolic crises. However, they may have other symptoms. These can include:

- 1) Poor growth and development.
- 2) Either low muscle tone or spasticity.

Some people do not have any symptoms until adulthood. Some of the symptoms seen in adults are:

- 1) Weakness.
- 2) Fatigue.

Some people with 3MCC deficiency never have symptoms and are found to be affected only after a brother or sister is diagnosed.

What is the treatment for 3MCC deficiency?

Your baby's primary doctor will work with a metabolic doctor and a dietician to care for your child.

In some children, prompt treatment is needed to prevent metabolic crises and the health effects that follow. Certain treatments may be advised for some children but not others. Children who do not show symptoms may not need treatment. The following are treatments that are used for some babies and children with 3MCC deficiency.

Low-Leucine Diet, Including Medical Foods and Formula

A food plan low in leucine with limited amounts of protein is sometimes needed. Most food in the diet will be carbohydrates (bread, cereal, pasta, fruit, vegetables, etc.). Carbohydrates give the body many types of sugar that can be used as energy. Eating a diet high in carbohydrates and low in protein can help prevent hypoglycemia and metabolic crises.

Foods high in protein that may need to be avoided or limited include:

- 1) Milk and dairy products.
- 2) Meat and poultry.
- 3) Fish.
- 4) Eggs.
- 5) Dried beans and legume.
- 6) Nuts and peanut butter.

Many vegetables and fruits have only small amounts of protein and can be eaten in carefully measured amounts. Do not remove all protein from the diet. Children with 3MCC deficiency need small amounts of protein to grow properly. If needed, your dietician will create a food plan that contains the right amount of protein, nutrients and energy for your child. Some children may be on a special food plan throughout life.

Medical Foods and Formula

There are medical foods such as special low-protein flours, pastas and rice that are made especially for people with organic disorders. If necessary for your child, your dietician will tell you how to use these foods.

In addition to a low-protein diet, some children are given a special leucine-free medical formula. Your metabolic doctor and dietician will decide whether your child needs this formula.

Medications

Some children may benefit by taking L-carnitine. This is a safe and natural substance that helps the body cells make energy. It also helps the body get rid of harmful wastes. Your doctor will decide whether your child needs L-carnitine. Unless you are advised otherwise, use only L-carnitine prescribed by your doctor. Do not use any medication without checking with your doctor.

Call Your Doctor at the Start of Any Illness

In some children, even minor illnesses such as a cold or the flu can lead to a metabolic crisis. In order to prevent problems, you may be told to call your doctor right away when your child has any of the following:

- 1) Loss of appetite
- 2) Vomiting
- 3) Diarrhea
- 4) Infection or illness
- 5) Fever

Some children need to eat more carbohydrates and drink more fluids when they are ill – even if they may not feel hungry – or they could have a metabolic crisis. They also should avoid eating protein during any illness.

Children who are ill often don't want to eat. If they can't eat, or if they show signs of a metabolic crisis, they may need to be treated in the hospital. Ask your doctor if you should carry a special travel letter with medical instructions for your child's care.

What happens when 3MCC is treated?

With prompt and careful treatment, children who have shown symptoms of 3MCC deficiency have a good chance to live healthy lives with typical growth and development.

Even with treatment, some children still have repeated bouts of metabolic crisis. This can cause brain damage and may lead to lifelong learning problems or mental retardation.

What causes the 3MCC enzyme to be absent or not working correctly?

Genes tell the body to make various enzymes. People with 3MCC have a pair of genes that do not work correctly. Because of the changes in this pair of genes, the 3MCC enzyme either does not work properly or is not made at all.

Is 3MCC deficiency inherited?

3MCC deficiency is inherited and affects both boys and girls equally.

Everyone has a pair of genes that make the 3MCC enzyme. In children with 3MCC deficiency, neither of these genes works correctly. These children inherit one nonworking gene for the condition from each parent.

Parents of children with 3MCC deficiency rarely have the disorder. Instead, each parent has a single nonworking gene for 3MCC deficiency. They are called carriers. Carriers do not have the condition because the other gene of this pair is working correctly.

When both parents are carriers, there is a 25 percent chance in each pregnancy for the child to have 3MCC deficiency. There is a 50 percent chance for the child to be a carrier, just like the parents. And, there is a 25 percent chance for the child to have two working genes.

Can other members of the family have 3MCC deficiency or be carriers?

Having 3MCC Deficiency

The brothers and sisters of a baby with 3MCC deficiency have a chance of being affected, even if they haven't had symptoms. Finding out whether other children in the family have this condition is important because early treatment may prevent serious health problems. Ask your doctor whether your other children should be tested.

3MCC Deficiency Carriers

Brothers and sisters who do not have 3MCC deficiency still have a chance to be carriers like their parents. Except in special cases, carrier testing should only be done in people older than 18.

Each of the parents' brothers and sisters has a 50 percent chance to be a carrier. It is important for other family members to be told that they could be carriers. There is a small chance they are also at risk to have children with 3MCC deficiency.

Can other family members be tested?

Diagnostic Testing

Brothers and sisters of a child with 3MCC deficiency can be tested using blood, urine or skin samples.

Carrier Testing

If both gene changes have been found in your child with 3MCC deficiency, other family members can have DNA testing to see if they are carriers.

How many people have 3MCC deficiency?

About one in every 50,000 babies in the United States is born with 3MCC deficiency.

Does 3MCC deficiency happen more frequently in a certain ethnic group?

No, this condition does not happen more often in any specific race, ethnic group, geographical area or country.

Does 3MCC deficiency go by any other names?

3MCC deficiency is sometimes called 3-methylcrotonylglycinuria.

The adult form of 3MCC deficiency is also called late-onset 3-methylcrotonyl CoA carboxylase deficiency.

Where can I find more information?

Organic Acidemia Association

www.oaanews.org

Save Babies Through Screening Foundation

www.savebabies.org

Children Living with Inherited Metabolic Diseases (CLIMB)

www.climb.org.uk

Genetic Alliance

www.geneticalliance.org

Children's Special Health Services (CSHS)

State Capitol Judicial Wing

600 E. Boulevard Ave., Department 301

Bismarck, ND 58505-0269

Toll Free: 800.755.2714

701.328.2436

Relay TDD: 701.328.3975

CSHS website: www.ndhealth.gov/CSHS

North Dakota Department of Health website:

www.ndhealth.gov

Family support resources available from CSHS:

- Guidelines of Care Info
- Family Support Packet
- Financial Help Packet
- Insurance Fact Sheet

Family Resources

Family to Family Network

Center for Rural Health

University of North Dakota

School of Medicine and Health Sciences

P.O. Box 9037

Grand Forks, ND 58202-9037

Toll Free: 888.434.7436

701.777.2359

Fax: 701.777.2353

E-mail: NDF2F@medicine.nodak.edu

www.medicine.nodak.edu/crh

Pathfinder Services of ND

Pathfinder Family Center

1600 2nd Ave. SW, Ste. 19

Minot, ND 58701

Toll Free: 800.245.5840

701.837.7500

Relay TDD: 701.837.7501

E-mail: ndpath01@ndak.net

www.pathfinder.minot.com

Family Voices of North Dakota, Inc.
P.O. Box 163
Edgeley, ND 58433
Toll Free: 888.522.9654
701.493.2634
Fax: 701.493.2635
www.geocities.com/ndfv

**This fact sheet has general information.
Every child is different and some of these facts
may not apply to your child specifically.
Certain treatments may be recommended for
some children but not others. All children
should be followed by a metabolic doctor in
addition to their primary-care provider.

Acknowledgement

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