



2MBDH Deficiency (2-Methylbutyryl CoA Dehydrogenase Deficiency)

What is it?

2MBDH deficiency is one type of organic acid disorder. People with 2MBDH deficiency have problems breaking down an amino acid called isoleucine 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.

2MBDH deficiency occurs when an enzyme, called 2-methylbutyryl CoA dehydrogenase (2-MBDH) is either missing or not working properly. This enzyme's job is to help break down isoleucine. When a child with 2MBDH deficiency eats food containing isoleucine, harmful substances build up in the blood. Isoleucine is found in all foods with protein.

If 2MBDH deficiency is not treated, what problems occur?

This condition is very rare. Only a small number of children with 2MBDH deficiency have been reported. The symptoms have been very severe in some children and mild or absent in others. It is likely that most babies found to have this condition through newborn screening will never develop symptoms.

Babies with 2MBDH deficiency are healthy at birth. A few babies start having symptoms just a few days after birth. Others have their first symptoms later in childhood. Some children never have symptoms. In a few children, 2MBDH deficiency causes 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) Irritable mood.
- 4) Difficulty keeping warm.

Other symptoms then follow:

- 1) Fever
- 2) Nausea
- 3) Low blood sugar, called hypoglycemia
- 4) Increased levels of acidic substances in the blood, called metabolic acidosis

If a metabolic crisis is not treated, a child with 2MBDH deficiency can develop:

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

If not treated, episodes of metabolic crisis can cause brain damage. This can lead to lifelong 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 2MBDH deficiency, this can trigger a metabolic crisis.

Between episodes of metabolic crisis, children with 2MBDH deficiency are usually healthy.

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

- 1) Poor growth.
- 2) Tight, rigid muscles, called spasticity.
- 3) Involuntary movements, called choreoathetosis.
- 4) Vision problems.
- 5) Muscle weakness.
- 6) Delays in walking and other motor skills.
- 7) Learning disabilities or mental retardation.

Some people with 2MBDH deficiency never have symptoms and are found to have the condition only after a brother or sister is diagnosed.

What is the treatment for 2MBDH deficiency?

Some children diagnosed with 2MBDH deficiency through newborn screening will never have symptoms. These children may not need treatment.

Babies who do have symptoms may need lifelong treatment. If this is the case, your baby's primary doctor may work with a metabolic doctor and a dietician to care for your child.

Prompt treatment may be needed to prevent metabolic crises and the health effects that follow. Certain treatments may be advised for some children but not others. Your doctor and metabolic doctor will decide whether your child needs treatment.

The following are treatments recommended for some babies and children with 2MBDH deficiency.

Avoid Going a Long Time Without Food

Some babies and young children with 2MBDH deficiency need to eat often to avoid a metabolic crisis. They should not go without food for more than four to six hours. Some babies may need to eat even more often than this. It is important that they be fed during the

night. They may need to be awakened to eat if they do not wake up on their own.

Low-Protein Diet, Including Medical Foods and Formula

A food plan low in protein is sometimes advised. 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 legumes.
- 6) Nuts and peanut butter.

Many vegetables and fruits have only small amounts of protein and can be eaten in carefully measured amounts. It is important not to remove all protein from the diet. Children with 2MBDH deficiency need a certain amount 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 you child. It is important to follow the advice of your dietician and doctor.

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 needed for your child, your dietician will tell you how to use these foods.

In addition to a low-protein diet, some children are given special medical formula that does not contain isoleucine. Your metabolic doctor and dietician will decided 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 2MBDH deficiency is treated?

With prompt and careful treatment, children with symptoms of 2MBDH deficiency have a good chance to live healthy lives with typical growth and development.

Despite treatment, some children may have repeated bouts of hypoglycemia or metabolic crises. This can cause brain damage and may lead to lifelong learning problems or mental retardation.

What causes the 2MBDH enzyme to be absent or not working correctly?

Genes tell the body to make various enzymes. People with 2MBDH deficiency have a pair of genes that do not work correctly. Because of the changes in this pair of genes, the 2MBDH enzyme either does not work properly or is not made at all.

Is 2MBDH deficiency inherited?

2MBDH deficiency is inherited and affects both boys and girls equally.

Everyone has a pair of genes that make the 2MBDH enzyme. In children with 2MBDH deficiency, neither of these genes works correctly. These children inherit one non-working gene for the condition from each parent.

Parents of children with 2MBDH deficiency rarely have the disorder. Instead, each parent has a single nonworking gene for 2MBDH 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 2MBDH 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 2MBDH deficiency or be carriers?

Having 2MBDH Deficiency

The brothers and sisters of a baby with 2MBDH 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. Early treatment may prevent serious health problems.

Talk to your metabolic doctor or genetic counselor about whether your other children should be tested.

2MBDH Deficiency Carriers

Brothers and sisters who do not have 2MBDH deficiency still have a chance to be carriers like their parents. Except in special cases, carrier testing should be done only 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 2MBDH deficiency.

When both parents are carriers, newborn screening results are not sufficient to rule out the condition in a newborn baby. In this case, special diagnostic testing should be done in addition to newborn screening.

Can other family members be tested?

Diagnostic Testing

Brothers and sisters of a child with 2MBDH deficiency can be tested using blood, urine or skin samples.

Carrier Testing

If both gene changes have been found in your child with 2MBDH deficiency, other family members can have DNA testing to see if they are carriers.

If DNA testing is not possible or is not helpful, other methods of carrier testing may be available. Your metabolic doctor or genetic counselor can answer your questions about carrier testing.

How may people have 2MBDH deficiency?

2MBDH deficiency is very rare. The actual incidence is unknown.

Does 2MBDH deficiency happen more frequently in a certain ethnic group?

2MBDH deficiency is more common in the Hmong population from Southeast Asia and in Hmong Americans. One in every 500 babies of Hmong ancestry is born with this condition.

Does 2MBDH deficiency go by any other names?

2MBDH deficiency is sometimes also called:

- 1) 2-methylbutyrylglucosuria.
- 2) Short/branched chain acyl-CoA dehydrogenase deficiency.

Where can I find more information?

Organic Acidemia Association

www.oaaneews.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.

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North Dakota Department of Health
Newborn Screening Program
600 E. Boulevard Ave., Dept. 301
Bismarck, ND 58505-0200
800.472.2286 or 701.328.2493
www.ndhealth.gov/familyhealth