Genetic Fact Sheets for Parents

Organic Acid Disorders

Screening, Technology, and Research in Genetics is a multi-state project to improve information about the financial, ethical, legal, and social issues surrounding expanded newborn screening and genetic testing – http:// www.newbornscreening.info

Disorder name: Isobutyryl-CoA dehydrogenase deficiency
Acronym: IBD deficiency

This fact sheet contains general information about IBD deficiency. Every child is different and some of these facts may not apply to your child specifically. At present, little is known about IBD deficiency, and there is no standard treatment plan. Certain treatments may be recommended for some children but not others. All children with IBD deficiency should be followed by a metabolic doctor in addition to their primary doctor.

What is IBD deficiency?

IBD stands for “isobutyryl-CoA dehydrogenase”. IBD deficiency has symptoms that are part of two different groups of conditions: **organic acid disorders** and **fatty acid oxidation disorders**. Some children with IBD deficiency have problems breaking down an amino acid called valine from the food they eat.

Organic Acid Disorders:

Organic acid disorders (OAs) are a group of rare inherited conditions. They are caused by enzymes that do not work properly. A number of enzymes are needed to process protein from the food we eat for use by the body. Problems with one or more of these enzymes can cause an organic acid disorder.

People with organic acid disorders cannot break down protein properly. This causes harmful substances to build up in their blood and urine. These substances can affect health, growth and learning.

The symptoms and treatment vary between different organic acid disorders. They can also vary from person to person with the same organic acid disorder. See the fact sheets for each specific organic acid disorder.

Organic acid disorders are inherited in an autosomal recessive manner and affect both males and females.

Fatty Acid Oxidation Disorders:

Fatty acid oxidation disorders (FAODs) are a group of rare inherited conditions. They are caused by enzymes that do not work properly.

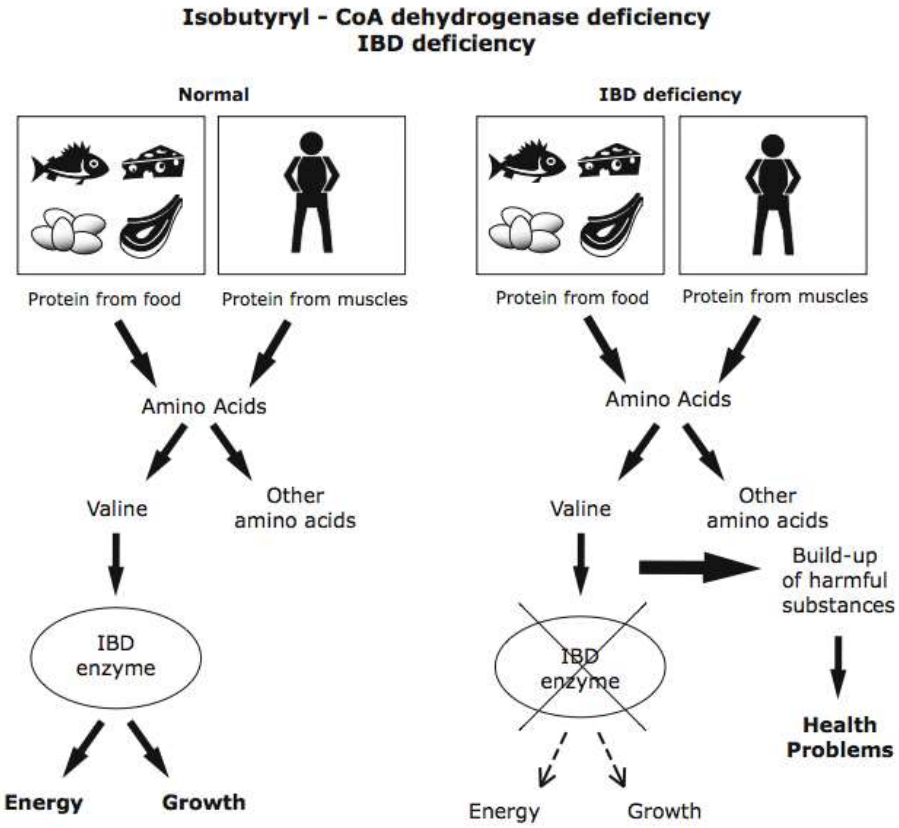
A number of enzymes are needed to break down fats in the body (a process called fatty acid oxidation). Problems with any of these enzymes can cause a fatty acid oxidation disorder. People with FAODs cannot properly break down fat from either the food they eat or from fat stored in their bodies.

The symptoms and treatment vary between different FAODs. They can also vary from person to person with the same FAOD. See the fact sheets for each specific FAOD.

FAODs are inherited in an autosomal recessive manner and affect both males and females.

What causes IBD deficiency?

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.



IBD deficiency occurs when an enzyme, called “isobutyryl-CoA dehydrogenase”, is either missing or not working properly. This enzyme’s job is to help break down valine. When a child with IBD deficiency eats food containing valine, harmful substances build up in the blood and cause problems. Valine is found in all foods that contain protein.

If IBD deficiency is not treated, what problems occur?

IBD deficiency is very rare and little is known about the effects. So far, symptoms have only been reported in a few children. Each child with IBD deficiency is likely to have somewhat different effects. Some children found to have IBD deficiency during newborn screening have never had symptoms.

It is likely that babies with IBD deficiency will be healthy at birth. In the first child reported to have IBD deficiency, symptoms began at one year of age.

Symptoms included:

- enlarged, weakened heart (called cardiomyopathy)

- anemia
- poor growth
- low carnitine levels (a substance needed for the breakdown of fatty acids)

What is the treatment for IBD deficiency?

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

It is important to talk with a metabolic doctor about possible treatment as soon as you know your child has IBD deficiency. Certain treatments may be advised for some children but not others. Some treatments may be needed throughout life.

The following are treatments that may be recommended for some babies and children with IBD deficiency:

1. Medications

Children with IBD deficiency may be helped by taking L-carnitine. This is a safe and natural substance that helps body cells make energy. It also helps the body get rid of harmful wastes. L-carnitine may help prevent or treat heart problems and anemia in children with IBD deficiency. Do not use L-carnitine without checking with your doctor. Unless you are advised otherwise, use only L-carnitine prescribed by your doctor.

2. Avoid going a long time without food

Some infants and young children with IBD deficiency may be advised to eat frequently to prevent health effects. Your metabolic doctor will tell you how often your child needs to be fed. In general, it is often suggested that infants be fed every four to six hours. Going without food for a long time causes the body to use its stores of fat and protein for energy. In some people with IBD deficiency, this may lead to the build up of harmful substances in the blood.

Your metabolic doctor and dietician will give you a feeding plan designed for your infant's needs.

3. Low-valine food plan (including medical foods)

Most children with IBD deficiency seem to do fine without a change in diet. However, some children may be advised to eat a diet made up of foods low in valine. Valine is found in all foods with protein.

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

- milk and dairy products
- meat and poultry
- fish
- eggs
- dried beans and legumes

- nuts and peanut butter

There are medical foods such low-protein flours, pastas, rice, and special formulas that are made especially for people with organic acid disorders. Your dietician will let you know whether you should use these foods to supplement your child's diet.

Some states offer help with payment, or require private insurance to pay for the formula and other special medical foods.

If you are advised to change your child's diet, it is important not to remove all protein from the diet. Children need a certain amount to grow properly. Your dietician will create a food plan that contains the right amount of protein, nutrients, and energy to keep your child healthy.

What happens when IBD deficiency is treated?

Although there is very little information available, it is thought that with prompt and careful treatment, children with IBD deficiency will be able to live healthy lives with typical growth and development. Treatment with carnitine may reverse the heart problems and anemia and may improve growth.

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

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

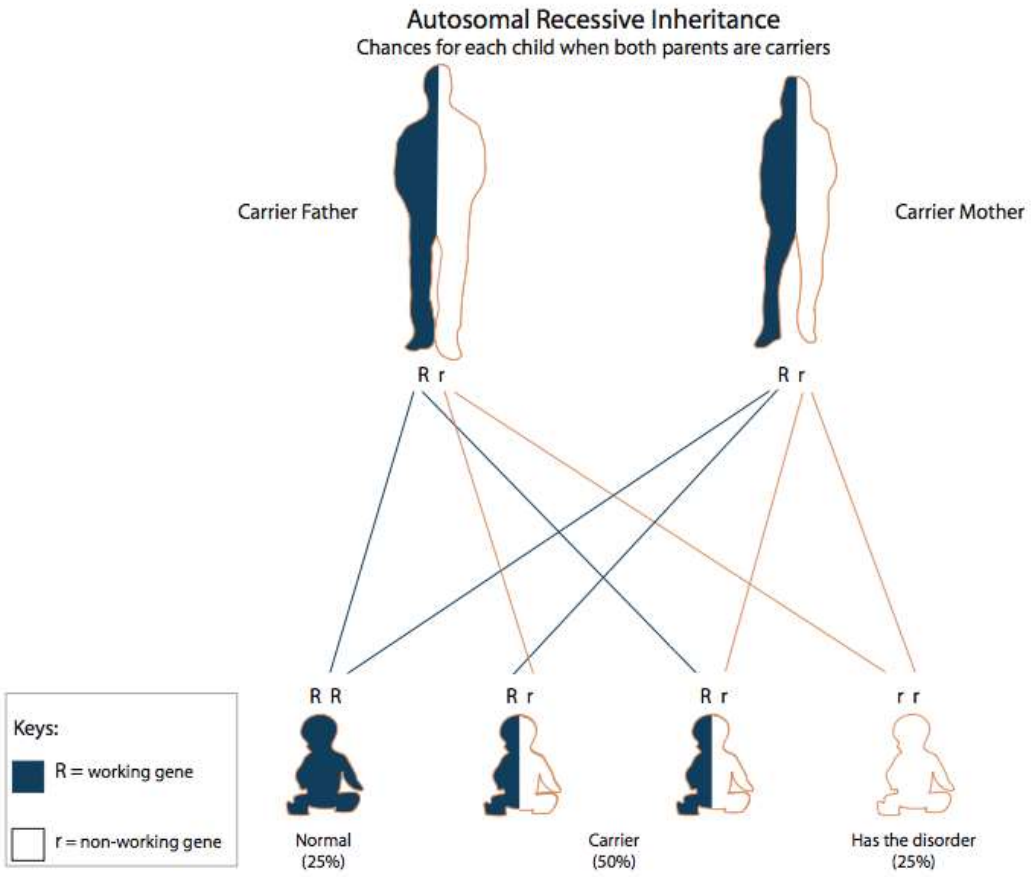
How is IBD deficiency inherited?

IBD deficiency is inherited in an autosomal recessive manner. It affects both boys and girls equally.

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

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

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



Genetic counseling is available to families who have children with IBD deficiency. Genetic counselors can answer your questions about how IBD deficiency is inherited, choices during future pregnancies, and how to test other family members. Ask your doctor about a referral to a genetic counselor.

Is genetic testing available?

Genetic testing for IBD deficiency is available. Genetic testing, also called DNA testing, looks for changes in the pair of genes that causes IBD deficiency. Talk with your genetic counselor or metabolic doctor if you have questions about DNA testing.

DNA testing is not necessary to diagnose your child. If available, it can be helpful for carrier testing or prenatal diagnosis, discussed below.

What other testing is available?

Special tests on blood, urine, or skin samples can be done to confirm IBD deficiency. Talk to your metabolic doctor or genetic counselor if you have questions about testing for IBD deficiency.

Can you test during pregnancy?

If both gene changes have been found in your child with IBD deficiency, DNA testing can be done during future pregnancies. The sample needed for this test is obtained by either CVS or amniocentesis.

Parents may either choose to have testing during pregnancy or wait until birth to have the baby tested. A genetic counselor can talk to you about your choices and answer questions about prenatal testing or testing your baby after birth.

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

Having IBD deficiency

The brothers and sisters of a baby with IBD deficiency have a small 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. Talk to your metabolic doctor or genetic counselor about testing your other children.

IBD deficiency carriers

Brothers and sisters who do not have IBD deficiency still have a chance to be carriers like their parents. Except in special cases, carrier testing should only be done in people over 18 years of age.

Each of the parents' brothers and sisters has a 50% 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 IBD deficiency.

Some states do not provide newborn screening for IBD deficiency. However, expanded newborn screening through private labs is available for babies born in states that do not screen for this condition. To learn more about expanded newborn screening, see **How to obtain MS/MS**.

When both parents are IBD 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

To make sure they do not have the condition, brothers and sisters of a child with IBD deficiency can be tested using blood, urine or skin samples.

Carrier testing

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

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

How many people have IBD deficiency?

IBD deficiency is very rare. Less than 30 people have been reported in the medical literature, and most people with IBD have no symptoms. The actual incidence is unknown.

Does IBD deficiency happen more frequently in a certain ethnic group?

IBD deficiency does not happen more often in any specific race, ethnic group, geographical area or country.

Does IBD deficiency go by any other names?

IBD deficiency is sometimes also called:

- Acyl-CoA dehydrogenase family, member 8
- ACAD8 deficiency

Where can I find more information?

Organic Acidemia Association
http://www.oaanews.org

CLIMB (Children Living with Inherited Metabolic Disorders)
http://www.climb.org.uk

Baby's First Test
http://www.babysfirsttest.org

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