

Organic Acidemias

What are Organic Acidemias?

Organic acidemias are inherited conditions that affect the way a person's body uses protein. A person with an organic acidemia cannot properly break down certain components of protein for energy, growth, and development. Typically, these compounds are amino acids that are not completely broken down. Since the body cannot properly break down these amino acids, certain organic acids build up in the blood and urine. High levels of certain organic acids can cause serious health problems.

How Do Organic Acidemias Affect a Child?

The symptoms of these conditions vary and depend on the type of organic acidemia.

What Causes Organic Acidemias?

Organic acidemias are genetic conditions caused by changes in specific genes. These genes are responsible for making enzymes. These enzymes are responsible for breaking down components of protein. When there is an alteration in these genes, enzyme levels go down and organic acids build up in the blood and urine.

Organic acidemias are inherited in an autosomal recessive pattern, which means two copies of a gene must be changed for a person to be affected with an organic acidemia. Most often, the parents of a child with an autosomal recessive condition are not affected because they are "carriers," with one copy of the changed gene and one copy of the normal gene.

When both parents are carriers, there is a one-in-four (or 25%) chance that both will pass a changed gene on to a child, causing the child to be born with the condition. There also is a one-in-four (or 25%) chance that they will each pass on a normal gene, and the child will be free of the condition. There is a two-in-four (or 50%) chance that a child will inherit a changed gene from one parent and a normal gene from the other, making that child a carrier like the parents. These chances are the same in each pregnancy for the same parents.

Is There a Test for Organic Acidemias?

Yes. Babies are tested through newborn screening for organic acidemias before they leave the hospital. The baby's heel is pricked and a few drops of blood are taken. The blood is sent to the state laboratory to find out if it has more than a normal amount of organic acids. Some states screen newborns for organic acidemias. Tennessee is one of those states.

There are various types of organic acidemias. The following is a list of organic acidemias that can be screened for:

- 2-Methyl-3-Hydroxybutyryl CoA Dehydrogenase deficiency (MHBD)
- 2-Methylbutyryl CoA Dehydrogenase deficiency (2-MBCD)
- 3-Hydroxy-3-Methylglutaryl CoA Lyase deficiency (HMG)
- 3-Methylcrotonyl CoA Carboxyl deficiency (3-MCC)
- 3-Methylglutaconyl CoA Hydratase deficiency (3-MGA)
- Glutaric Aciduria Type I (GA-1)
- Isobutyryl CoA Dehydrogenase deficiency (ICBD)
- Isovaleric Acidemia (IVA)
- Malonic Aciduria (MA)
- Methylmalonic Acidemia (MMA)
- Mitochondrial Acetoacetyl CoA Thiolase – (3-Ketothiolase) (BKT)
- Multiple CoA Carboxylase (MCD)
- Propionic Acidemia (PA)

Can Organic Acidemia Symptoms Be Prevented?

In most cases, many symptoms of an organic acidemia can be prevented by diet restrictions. Each treatment depends on the specific disorder. Children and adults with organic acidemias require follow-up care at a medical center or clinic that specialize in these types of metabolic condition. In addition, regular blood tests are used to monitor an individual's health.

DISCLAIMER: The information contained on this page is not intended to replace the advice of a genetic metabolic medical professional.

Resources:



Organic Acidemia Association
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PAF
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References:

- American Academy of Pediatrics (1996): Newborn Screening Fact Sheets (RE9632). Pediatrics 98:473-501.
<http://aappolicy.aappublications.org/cgi/reprint/pediatrics;98/3/473.pdf>
- Cedaerbaum, S.D., Scott, C.R., & Wilcox, W.R. (1997) Organic Acid Metabolism In; Rimoin, D.L., Connor, J.M., Pyeritz, R.E. (eds) Emery and Romoin's Principles and Practice of Medical Genetics, 3rd ed. Churchill Livingstone, New York, 1977-1989.

- GeneTests (The Organic Acidemias) <http://www.genetests.org>
- National Newborn Screening & Genetic Resource Center (2004) US National Screening Status Report – MS/MS
<http://genes-r-us.uthscsa.edu/resources/newborn/msmstests.htm>