

## 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 it a [carrier](#) like its parents. These chances are the same in each pregnancy for the same parents.

### Is there a test for organic acidemias?

Yes. Babies are tested (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](#).

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 (IBCD)
- 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  
13210 - 35th Avenue North  
Plymouth, MN 55441  
Phone: 763-559-1797  
Fax: 763-694-0017  
Email: [oaanews@aol.com](mailto:oaanews@aol.com)  
[www.oaanews.org](http://www.oaanews.org)

Association for Neuro-Metabolic Disorders  
5223 Brookfield Lane  
Sylvania OH 43560-1809  
Phone: 419-885-1497  
E-mail: [VOLK4OLKS@aol.com](mailto:VOLK4OLKS@aol.com)

Propionic Acidemia Foundation  
1963 McCraren  
Highland Park, IL 60035  
Phone: 763-559-1797  
Email: [PAF@pafoundation.com](mailto:PAF@pafoundation.com)  
[www.pafoundation.com](http://www.pafoundation.com)

**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.geneclinics.org>
- National Newborn Screening & Genetic Resource Center (2004) US National Screening Status Report – MS/MS <http://genes-r-us.uthscsa.edu>