

Homocystinuria

What is homocystinuria?

Homocystinuria is an inherited condition that affects the way a person's body uses a part of food called methionine (a precursor to homocystine). A person with homocystinuria cannot breakdown the methionine in food. Methionine and homocystine are [amino acids](#) needed for proper growth and development, but too much can cause serious health problems. In the case of classical homocystinuria, too much methionine builds up in the blood, which in turn causes a build up of homocystine. High levels of methionine and homocystine penetrate and damage the brain. These high levels ultimately cause mental retardation and other serious health problems.

About one baby in 340,000 is born with homocystinuria in the United States. The condition occurs in all ethnic groups.

How does homocystinuria affect a child?

Without treatment, children with homocystinuria develop permanent mental retardation and behavioral problems. Seizures, delayed development, dislocated lenses of the eye and weakening of the bones is also common. In addition, life threatening blood clots may develop and become lodged within blood vessels.

What causes homocystinuria?

Homocystinuria is a genetic condition caused by a change in the CBS (cystathionine beta-synthase) [gene](#). The CBS [gene](#) is responsible for making an [enzyme](#) called cystathionine beta synthase. Cystathionine beta synthase changes the homocystine into other needed compounds for the body. When there is an alteration in the CBS [gene](#), Cystathionine beta synthase levels go down and homocystine builds up in the blood stream.

Homocystinuria is inherited in an [autosomal recessive](#) pattern, which means two copies of the CBS [gene](#) must be changed for a person to be affected with homocystinuria. 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 percent) chance that both will pass the changed CBS [gene](#) on to a child, causing the child to be born with the condition. There also is a one-in-four (or 25percent) chance that they will each pass on a normal CBS [gene](#), and the child will be free of the condition. There is a two-in-four (or 50 percent) chance that a child will inherit a changed CBS [gene](#) from one parent and a normal CBS [gene](#) from the other, making it a [carrier](#) like its parents. These chances are the same in each pregnancy with the same parents.

Is there a test for homocystinuria?

Yes. Babies are tested (newborn screening) for homocystinuria 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 methionine.

Can homocystinuria symptoms be prevented? Yes. In most cases, the symptoms of homocystinuria can be prevented by a diet very low in methionine. This diet should begin as soon as possible following a diagnosis. Children and adults with homocystinuria require follow-up care at a medical center or clinic that specializes in this condition. Methionine content in

foods are different, so an experienced dietician or nutritionist will recommend a special diet that includes certain vegetables, fruits, grains, and a metabolic formula (food) that provides essential nutrients without methionine. Some other forms of homocystinuria also respond to prescription replacement medications.

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

Resources:

MUMS National Parent-to-Parent Network
Julie J. Gordon 150 Custer Court
Green Bay, Wisconsin 54301-1243
Phone: 1-877-336-5333 (Parents only please)
Phone: 1-920-336-5333
Fax: 1-920-339-0995
E-mail: mums@netnet.net
www.netnet.net/mums/

National Coalition for PKU & Allied Disorders
PO Box 1244
Mansfield, MA 02048
Phone: 877-996-2723
www.pku-allieddisorders.org/home.htm

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) Amino 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, 1878-1881.
- Online Mendelian Inheritance in Man (OMIM topic 236200)
<http://www.ncbi.nlm.nih.gov/Omim>
- Scriver, C.R. and Kaufman, S (2001) Disorders of Transulfuration. In: Scriver, C.R., Kaufman, S., Eisensmith, E., Woo S.L.C., Vogelstein, B. Childs, B. (eds) The Metabolic and Molecular Bases of Inherited Disease, 8th ed. McGraw-Hill, New York, Ch.8.