

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, that is, an amino acid from which homocystine is made). 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 buildup 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 in 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 compounds needed by 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%) 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 25%) 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%) chance that a child will inherit a changed CBS gene from one parent and a normal CBS gene from the other, making that child a carrier like the parents. These chances are the same in each pregnancy with the same parents.

Is There a Test for Homocystinuria?

Yes. Babies are tested through 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. Some states screen newborns for homocystinuria, and Tennessee is one of them.

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 specialize in this condition. Methionine content in foods is different, so an experienced dietician or nutritionist will recommend a special diet that includes certain vegetables, fruits, grains, and a metabolic formula (medical replacement food) that provides essential nutrients without methionine. Some other forms of homocystinuria also respond to prescription 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/



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.