

Homocystinuria

General Overview

Q. What is homocystinuria?

A. Homocystinuria is a treatable disorder that affects the way the body processes protein. Children with homocystinuria cannot use a part of the protein called methionine. If left untreated, methionine and related molecules build up in the bloodstream and lead to brain damage and other disabilities.

Q. Is there only one form of homocystinuria?

A. No, there are several forms of homocystinuria. Some people with homocystinuria are treated slightly differently than others because they respond to treatment with a specific vitamin.

Q. How does the body normally process methionine?

A. The body normally converts methionine into a different form called cysteine, which is then used by the body in other metabolic functions.

Q. What happens to methionine in a child with homocystinuria?

A. In a child with homocystinuria, methionine cannot be converted to cysteine because one of the needed enzymes does not work properly. This results in large amounts of methionine and related molecules, which are toxic to the brain and nervous system.

Q. What are the effects of having homocystinuria if it is not treated?

A. Without treatment, about half of people with homocystinuria die, usually from blood clots that block normal blood flow. Untreated homocystinuria can result in mental retardation and other problems of the nervous system. It can also result in eye problems and skeletal abnormalities.

Q. What is the treatment for homocystinuria?

A. Some people with homocystinuria respond to vitamin B6 (pyridoxine). Those who do not respond to this treatment are placed on a special diet that is low in methionine. To prevent mental retardation and developmental disability, treatment must begin shortly after birth. People with homocystinuria require specialized treatment through a clinic with experience in treating this disorder.

Q. Why would a child have homocystinuria?

A. Homocystinuria is an inherited disorder. It results when a baby receives a double-dose of a specific non-working gene involved in methionine conversion (one from each parent). For more information about this, contact your health care provider or a genetic counselor.

Q. How common is homocystinuria?

About one in every 200,000 babies in the United States is born with homocystinuria.

For more information about homocystinuria, please see the Disorders section of our website: www.doh.wa.gov/nbs.



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Revised: 04/04