



www.mi.gov/newbornscreening

Michigan Resources & Support

Children's Hospital of Michigan Metabolic Clinic

Toll-free: 1-866-44CHMMC

Children's Special Health Care Services

Family Phone Line

Toll-free: 1-800-359-3722

Early On® Michigan

Toll-free: 1-800-EARLY ON

www.1800earlyon.org

Michigan Genetics Connection

www.migeneticsconnection.org

Michigan Newborn Screening

Follow-up Coordinator

Toll-free: 1-866-673-9939

E-mail: MDCH-newbornscreening@michigan.gov

@michigan.gov

Michigan NBS Parent Liaison

Toll-free: 1-866-673-9939

E-mail: NBS-parent@michigan.gov

National Resources & Support

Family Village

www.familyvillage.wisc.edu

GeneReviews

www.genetests.org

Genetic Alliance

www.geneticalliance.org

National Coalition for PKU and Allied Disorders

Toll-free: 1-877-996-2723

www.pku-allieddisorders.org

STAR-G Project

www.newbornscreening.info/Parents/facts.html

What is Homocystinuria?

Homocystinuria is an inherited disorder in which the baby is unable to digest part of a protein found in food and milk. It occurs in about 1 in 200,000 newborns. Without treatment, babies with homocystinuria will have problems with bone development, learning, vision and blood clotting.

How may Homocystinuria affect my child?

Developmental Delay and Mental Retardation

Developmental delay is often the first sign of homocystinuria in the untreated child. Newborn screening and early treatment shortly after birth will offer a child the best outcome.

Eye (ocular) Problems

A dislocated lens in the eye (ectopia lentis) is usually seen in untreated children by 8 years of age. Near-sightedness (myopia) is also common.

Bone (skeletal) Problems

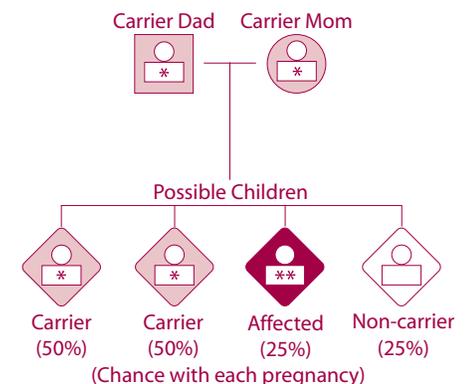
An untreated child may be taller than family members. Bones may be longer than usual. About one-half of untreated children will also develop weak bones (osteoporosis) by their teens.

Blood Clotting Problems

Untreated children may develop a blood clot (thromboembolism) in any blood vessel. In untreated young adults, these blood clots may lead to an early death.

How does Homocystinuria occur?

Homocystinuria is a genetic disorder. Parents of an affected child carry a genetic trait causing homocystinuria. Both parents pass the trait to a child with homocystinuria. There is a 1 in 4 chance that each child will have homocystinuria when both parents carry the trait for the disorder.



How is Homocystinuria treated?

Newborns are placed on a special formula to restrict protein. Children must have frequent metabolic monitoring. Special formulas and vitamin supplements including vitamin B6 may be given. The Metabolic Clinic and your pediatrician will help you begin and keep your child on a diet that meets his or her metabolic needs.

For more information contact the Newborn Screening Program toll-free at 1-866-673-9939 or e-mail NBS-Parent@michigan.gov

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