



State of CT Genetics Newborn Screening Program Family Fact Sheet

HOMOCYSTINURIA (HCU)

Protein is an important part of our diet. Protein can help body growth and strength when enzymes break the protein down into its proper parts. Babies who have the disorder called homocystinuria (hoe-moe-SIS-tin-NU-ree-ah) cannot break down the amino acid methionine. Babies with homocystinuria (HCU) have high levels of homocysteine and methionine in their body. If HCU is not treated, the high levels of homocysteine and methionine can cause mental retardation, eye problems, blood clots and other serious physical problems.

Connecticut began screening newborns for HCU in 1993. In Connecticut, about 1 in every 50,000 to 150,000 newborn babies has HCU.

How does a baby get HCU?

HCU is an inherited condition. Both the mother and the father have one altered gene that will not allow the body to break down the amino acids, homocysteine and methionine. Your baby has to receive this gene from both the mother and the father to have this condition.

How is a baby tested for HCU?

Prior to discharge from the facility of birth, a small sample of blood is taken from the baby's heel. The sample is then sent to the State laboratory to screen for HCU. Abnormal screening results are reported to the Department of Public Health (DPH), Genetic Newborn Screening Tracking Unit nurse consultant. If the results are positive, the DPH nurse consultant will notify the baby's primary care physician of the need for a prompt referral to one of the Genetic Regional Treatment Centers at Yale or the UCONN Health Center for confirmation testing and follow-up treatment if necessary.

What is the treatment for HCU?

The Regional Treatment Center specialists will prescribe treatment for HCU if necessary, which may include supplemental vitamin B₆. If the infant does not respond to B₆ treatment, betaine therapy, a special diet, and a special formula may be necessary. Early diagnosis of HCU leads to early treatment, which is important for the health of your baby.

The information provided is offered for general informational and educational purposes only. It is not offered as and does not constitute medical advice. In no way are any of the materials presented meant to be a substitute for professional medical care or attention by a qualified practitioner, nor should they be construed as such. Contact your physician if there are any concerns or questions.



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