

Homocystinuria (HCU) (Home o sis tin your ee a)

What is HCU?

Homocystinuria (HCU) is a metabolic disorder. This means the body has a chemical imbalance. HCU is a condition in which an enzyme needed to break down certain amino acids does not function properly. An increase level of methionine results from this improper function. HCU is inherited. It is present at birth.

What type of problems occurs with HCU?

A baby who has this condition may have a delay in their development, poor bone structure, and a greater risk for blood clots. Early diagnosis and treatment will help prevent these problems.

What is the chance my baby will have HCU?

This condition occurs in less than 1 in every 100,00 births. Babies born with this condition have a changed gene from each parent. A person who has one changed gene is called a carrier. A person who is a carrier does not have symptoms. If both parents are carriers, either parent can pass on the changed gene to their baby. If both parents pass on the changed gene, the baby will have the condition. If both parents are carriers, for each pregnancy:

- There is a 25% chance that the baby will be born with this condition.
- There is a 50% chance that the baby will be a carrier for this condition.
- There is a 25% chance that the baby will not be born with this condition and will not be a carrier.

What is the treatment of HCU?

The treatment of HCU is to maintain a diet low in methionine and include medications. Your baby's metabolic doctor will help you make sure that your baby gets the right diet and medical care.

Where in Virginia can I take my baby for care?

Please speak to your baby's pediatrician about obtaining a referral to a pediatric metabolic specialist in your area. If you want to know more about this condition, please contact Virginia Newborn Screening Services, Virginia Department of Health. The Web site is <http://www.vahealth.org/gns>.

