



# Newborn Screening FACT Sheet

## Homocystinuria (HCY)

### What is homocystinuria?

Homocystinuria is a condition in which the body cannot break down certain proteins. Because people with homocystinuria cannot process certain amino acids, which are small molecules that make up proteins, homocystinuria is classified as an amino acid disorder. These amino acids build up in the body and can cause serious health problems. When doctors detect homocystinuria early and begin treatment, children with homocystinuria can often lead healthy lives.

### What causes homocystinuria?

HCY occurs when an enzyme called "cystathionine beta-synthase" (CBS) is missing or not working correctly. This enzyme typically breaks down methionine. When the CBS enzyme is not working, methionine and homocysteine (another amino acid) build up in the blood and cause problems.

### What symptoms or problems occur with homocystinuria?

Babies may look healthy at birth. If untreated, homocystinuria can cause growth and developmental delays. It can also cause eye problems, abnormal bone growth, and higher risk for blood clots. However, early treatment can help prevent many of these effects.

### What is the treatment for homocystinuria?

People with this disease require a special diet. The specialist doctor seeing your baby will have you meet with a dietician. Dietitians know which foods are safe to eat. The following are treatments often used for children with homocystinuria:

- Low-methionine diet
- Special medical formula

### Things to remember

With lifelong treatment, many children have normal growth and ability. Treatment may lower the chance of blood clots, heart disease, and stroke. Treatment also lessens the severity of eye problems. Even when treated, people may still develop lens dislocation in the eye later in life. It can often be fixed by surgery or other methods. It is very possible that children with homocystinuria can go on to have healthy lives.

### Resources:

**HCU Network America** [Home - HCU Network America](#)

**MedlinePlus** [Homocystinuria: MedlinePlus Genetics](#)

**Health Resources & Services Administration**  
[Homocystinuria | Newborn Screening](#)

**Navigate Life Texas**  
[navigatelifetexas.org/en/diagnosis-healthcare/children-with-multiple-disabilities-rare-conditions-undiagnosed](https://navigatelifetexas.org/en/diagnosis-healthcare/children-with-multiple-disabilities-rare-conditions-undiagnosed)