Homocystinuria (HCY)

**What is Homocystinuria?**
Homocystinuria is a condition in which the body is unable to break down certain proteins. Homocystinuria is considered an amino acid condition because individuals with homocystinuria cannot process certain amino acids, small molecules that make up proteins. These amino acids build up in the body and can cause serious health problems. However, if homocystinuria is detected early and treatment is begun, children with homocystinuria can often lead healthy lives.

**What Causes Homocystinuria?**
Enzymes help start chemical reactions in the body. Homocystinuria happens when an enzyme called “cystathionine beta-synthase” (CBS) is missing or not working right. This enzyme breaks down methionine. When the CBS enzyme is not working right, methionine and homocysteine (another amino acid) build up in the blood and cause problems.

**What Symptoms or Problems Occur with Homocystinuria?**
Symptoms are something out of the ordinary that a parent notices.

Babies look healthy and normal at birth. If untreated, homocystinuria can cause growth and learning delays. It can also affect the eyes, bones, heart, and blood vessels.

**Growth, learning and behavior** – Delays in growth and learning are often noticed between ages one and three. Common problems include:

- poor growth
- problems gaining weight
- delays in crawling, walking, and talking
- behavior and emotional problems (such as crying for no reason)
- serious learning or intellectual disabilities

**Eyes** – Children with homocystinuria usually start to develop severe nearsightedness after age one. If this isn’t treated, the lens of the eye can get loose and move out of place. This is called “lens dislocation.” It often happens between ages two and eight. Glaucoma is a condition caused by increased eye pressure. This can happen over time if the lens dislocation is not treated. Untreated glaucoma can cause blindness.

**Bones and skeleton** – Teens and adults with homocystinuria are often tall and slender. They may have very long arms, legs, and fingers. About half have thinning of the bones, called osteoporosis. Some children will have muscle weakness, especially in the legs.

**Heart and blood vessels** – Homocystinuria can cause blood clots resulting in heart disease or stroke if not treated. Stroke and heart disease are the main causes of early death in people with untreated homocystinuria.

**Other** – Children who are not treated often have pale hair and skin. Some will have periods of pancreatitis (inflammation of the pancreas gland), which causes severe pain.

**What is the Treatment for Homocystinuria?**
Your baby’s primary doctor will work with a metabolic doctor and dietitian to care for your child. Dietitians know what the right foods are to eat.

The following are treatments often used for children with HCY:

- Low-methionine diet
- Special medical formula
- Blood and urine tests
  - Vitamin B6
  - Betaine
  - Vitamin B12
  - Folic Acid
  - L-cysteine

**Things to Remember**
With treatment all life long, many children have normal growth and intelligence. Treatment may lower the chance for blood clots, heart disease, and stroke. Treatment also lessens the chance of eye problems. Even when treated, some people still develop lens dislocation. This can often be fixed by surgery or other methods.

Children who begin treatment later in life may have intellectual disabilities and behavior problems.

Disclaimer: This information is adapted from the American College of Medical Genetics and Genomics (ACMG) 01/2015