

1. What is homocystinuria?

Homocystinuria is a disorder caused by excess of the methionine derived amino acid homocysteine. The defect is in the enzyme responsible for metabolizing homocysteine to compounds that can be excreted in the urine. This enzyme is known as cystathionine β -synthase. Excess homocysteine damages connective tissue leading to problems in the eyes with myopia and lens dislocation, skeletal system (particularly osteoporosis) and cardiovascular system (marked predisposition to atherosclerotic disease). Mental retardation and behavioral problems also occur.

2. How and when will we know if my baby has homocystinuria?

If your baby's newborn screening result showed a Met level > 2 mg / dl, he or she probably has homocystinuria or MAT deficiency (which is probably not at all harmful). If the result was 1-2 mg /dl your baby could still have homocystinuria but the finding might also be transient (false positive). The newborn screening test will be repeated and additional tests will be undertaken to help determine if your baby has homocystinuria or not. Typically the results of these tests take up to 4 days to come back. Depending on the test results, additional testing can take a variable amount of time to confirm the diagnosis. If the diagnosis of homocystinuria is confirmed, a trial of treatment with vitamin B6 will be undertaken to determine if your baby has the B6 responsive variant or not.

3. How did my baby get this?

Homocystinuria is an autosomal recessive disorder. This means that your baby has two mutated genes, one from the mother and one from the father. Having only one mutated gene (a carrier) does not affect a person at all.

4. What does it mean for my child?

If your baby has homocystinuria, he or she should stay on a special low methionine diet, or other treatment, throughout life. This will help to prevent or minimize the complications of this condition.

5. What is the treatment? Does it work? Is the diet difficult to do/expensive?

Homocystinuria is primarily treated by either a low methionine diet, a large amount of vitamin B6 or both. Children with homocystinuria cannot eat as much protein as other children and must have their feeds supplemented with a special methionine-free formula. This diet is very effective at preventing the complications of homocystinuria. Most babies and children get used to this diet. Later on, additional therapies such as the drug betaine may be used.

6. What about my other children/future children?

Since homocystinuria is an inherited condition it is important to have your other children tested. Children from the same father and mother as the affected infant have a 1 in 4 (25%) chance of having homocystinuria and, though it is less likely if they are well, some individuals remain relatively asymptomatic or very mildly affected for some time. Since there is a risk for having a future child with homocystinuria it is important to let your obstetrician and pediatrician know that you have a child with homocystinuria if you are planning future pregnancies so that they may discuss the options with you and prepare accordingly.