Homocystinuria

Homocystinuria (HCU), also known as Cystathionine beta-synthase deficiency, is a rare but potentially serious inherited condition.

It means the body cannot process the amino acid methionine. This causes a harmful build-up of substances in the blood and urine.

Normally, our bodies break down protein foods like meat and fish into amino acids. These are the 'building blocks' of protein. Any amino acids that aren't needed are usually broken down and removed from the body.

Babies with HCU are unable to fully break down the amino acid methionine. This causes a build-up of methionine and a chemical called homocysteine. This can be harmful.

Diagnosing homocystinuria

In the first few days of life, babies are offered newborn blood spot screening to check if they have HCU. This involves pricking your baby's heel to collect drops of blood to test.

If HCU is diagnosed, treatment can reduce the risk of serious complications. Treatment may include high doses of vitamin B6 (pyridoxine), a special diet and advice.

With early diagnosis and treatment, most children with HCU are able to live healthy lives. But they will have to continue treatment for HCU for life.

Babies born with HCU do not usually have any symptoms in the first year of their life. But severe symptoms can develop later in life without early treatment.

These may include:

- vision problems, such as severe short-sightedness
- weak bones (osteoporosis)
- bone and joint problems
- a risk of developing blood clots and strokes

Some children with untreated HCU are also at risk of brain damage. This can affect their development.

Treating homocystinuria

Vitamin B6 (pyridoxine)

In some babies, it's possible to control the levels of homocysteine. This can be done with high doses of vitamin B6 (pyridoxine). If this works, your child will need to take vitamin B6 supplements for the rest of their life.

Diet

A baby with HCU may not respond to vitamin B6. If they do not, they will be referred to a specialist metabolic dietitian. They will be given a low-protein diet to reduce the amount of methionine they receive.

High-protein foods need to be limited, including:

- meat
- fish
- cheese
- eggs
- pulses
- nuts

Your dietitian will provide detailed advice and guidance. This is because your baby still needs some of these foods for healthy growth and development.

Your dietitian will ask you to check and measure your breast milk and baby milk. You may need to use a special formula instead. This contains all the vitamins, minerals and other amino acids your baby needs.

As your baby moves to solids, your dietitian can tell you which low-protein foods are suitable. Some of these may be available on prescription. These may include low-protein rusks, milk substitutes and low-protein pasta.

People with HCU may need to follow a modified diet for the rest of their life. As your child gets older, they'll need to learn how to control their diet. They will need to stay in contact with a dietitian for advice and monitoring.

They'll also need regular blood tests to check the amount of homocysteine in their blood.

Medication

Alongside a low-protein diet, your child may be prescribed a medication called betaine. This is to help clear some of the excess homocysteine.

Medication for HCU needs to be taken as directed by your doctor.

How homocystinuria is inherited

HCU is passed on by parents. But the parents usually do not have any symptoms of the condition. This is known as autosomal recessive inheritance.

This means a baby needs to receive 2 copies of the mutated gene to develop the condition. They need one from their mother and one from their father. If the baby only receives one affected gene, they'll just be a carrier of HCU.

If you're a carrier of the altered gene and you have a baby with a partner who's also a carrier, your baby has a:

- 1 in 4 chance of developing the condition
- 2 in 4 chance of being a carrier of HCU
- 1 in 4 chance of receiving a pair of normal genes

It's not possible to prevent HCU. But it's important to let your midwife and doctor know if you have a family history of the condition.

If you have more children, they can be tested for the condition as soon as possible.

Read about heel prick screening for babies

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