

Results of the Newborn Blood Spot Screening

Parent Information Sheet Homocystinuria (HCU) - Confirmed Diagnosis



Your baby has been seen by the Specialist Metabolic Team and a diagnosis of HCU has been confirmed. However, each child with HCU is different and there may be some treatment differences from child to child.

This leaflet will help you to understand the condition and its treatment.

What is HCU?

HCU stands for Homocystinuria, pronounced ho-mo-sis-tin-ur-ee-a. Homocystinuria is a rare disorder that prevents the normal breakdown of protein. In order for the body to use protein from the food we eat, it is broken down into smaller parts called amino acids. In Homocystinuria one of these amino acids (methionine) does not break down in the usual way and methionine and a chemical called homocysteine builds up in the blood.

Without early treatment this can lead to long term health problems, including learning difficulties. These problems can be prevented by following the treatment advised by your Specialist Metabolic Team and your child will grow and develop normally and have a normal life expectancy.



What are the symptoms of HCU?

HCU may affect different babies in different ways. Babies with HCU are usually well in early life, although symptoms may develop later if untreated. Some children develop problems with their eyes, including severe short sightedness and dislocation of the lens.

Without early diagnosis and early start of treatment, children can develop damage to the brain, including learning difficulties. Children may also have thin bones (osteoporosis), bone and joint problems and may develop blood clots or strokes.

How is HCU Treated?

Treatment is given in children with HCU to prevent the build up of homocysteine. In a few babies with HCU, the level of homocysteine can be controlled by giving Vitamin B6 (Pyridoxine). If this does not work, HCU can be treated effectively with a special low-protein diet and extra supplements and medications. The low protein diet is controlled by the Specialist Metabolic Dietitian. The aim of the diet and other medications is to prevent the build up of Homocysteine in the blood.

Foods that provide the body with protein include milk, meat, fish, cheese, eggs, pulses and nuts. All baby milks (including breast milk) contain protein. Breast feeding is encouraged for babies. If you are breast feeding your baby who has HCU, you will be supported in continuing this as part of your baby's special diet. The amount of normal baby milk or breast milk a baby with HCU



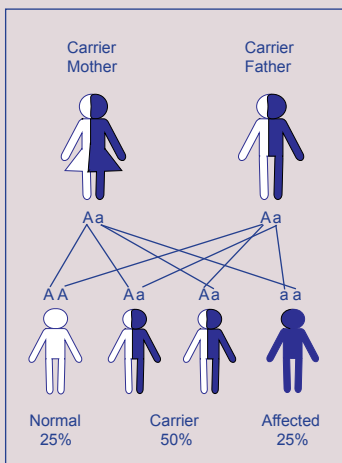
is given has to be limited. Your Specialist Metabolic Dietitian will advise you on this.

As breast feeds or normal baby milks have to be limited, a special baby milk consisting of amino acids without methionine is given to meet requirements for protein, calories, vitamins and minerals. This supplement is very important because it allows normal growth and development. Your Specialist Metabolic Dietitian will provide you with detailed information about the special baby milk and will explain how much your baby should have. Before your child starts weaning onto solids your Specialist Metabolic Dietitian will explain which foods can be used including special low protein foods such as low protein rusk, milk substitute, and low protein pasta which are available on prescription.

Your child will require regular blood tests to monitor the level of homocysteine in their blood. It is important to follow the advice regarding the low protein diet and supplements, in order to keep the levels in an acceptable range. Your child's diet will be adjusted as needed by the Specialist Metabolic Dietitian.

How will HCU affect my child's future?

Children with HCU can live full and active lives just like any other children, as long as they keep to the special low protein diet and/or medications or supplements as advised by their Specialist Metabolic Team. Children with HCU will be regularly monitored throughout their lives to check that homocysteine levels are well controlled.



Why does my baby have HCU?

HCU is an inherited condition. Everyone has two copies of the gene for HCU. A baby with HCU has inherited two faulty copies of the gene. The parents have one normal copy and one faulty copy and are said to be 'carriers'. When two HCU carriers have a baby, they have a 1 in 4 (25 %) chance in each pregnancy of having a child with

HCU. There is nothing the parents could have done to prevent their child having HCU. The diagram shows how this happens.





Will my other children need to be tested?

Your other children may be at risk of HCU even though they may not have shown any symptoms to date. It is therefore very important that they are tested if they have not been previously screened for HCU.

Who can I ask for advice and support?

The paediatric or metabolic clinician who is responsible for your child's care will be happy to discuss any queries you may have. Your Specialist Metabolic Dietitian will also be able to advise you.

Contact Details for the Specialist Metabolic Team:

Specialist Centre		
Consultant		
Metabolic Dietitian		
Clinical Specialist Nurse		
Ward (if applicable)		

Where can I find more information or support?

More information can be found at the Expanded Screening Programme website:

www.expandedscreening.org

CLIMB (The National Information Centre for Metabolic Diseases) provides information and support for people with HCU and their families.

Climb Building
176 Nantwich Road
Crewe
CW2 6BG

Telephone helpline: 0800 652 3181 (Freephone) or 0845 241 2172

Website: www.climb.org.uk

Email: info@climb.org.uk

The text and this leaflet are designed for use by Healthcare Professionals within the NHS to offer information to parents. It should not be used by others without prior permission.

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HCU - Parent Information - Confirmed

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Not to be used after August 31st 2013

Approved by Dr Andrew Morris,
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**National Institute for
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