Newborn Screening Programme for
Inborn Errors of Metabolism
Information leaflet series (No. 15)

Homocystinuria

For general queries on Newborn Screening Programme for Inborn Errors of Metabolism, please call: ☎ 5741 4280 (Department of Clinical Genetics, Hospital Authority)

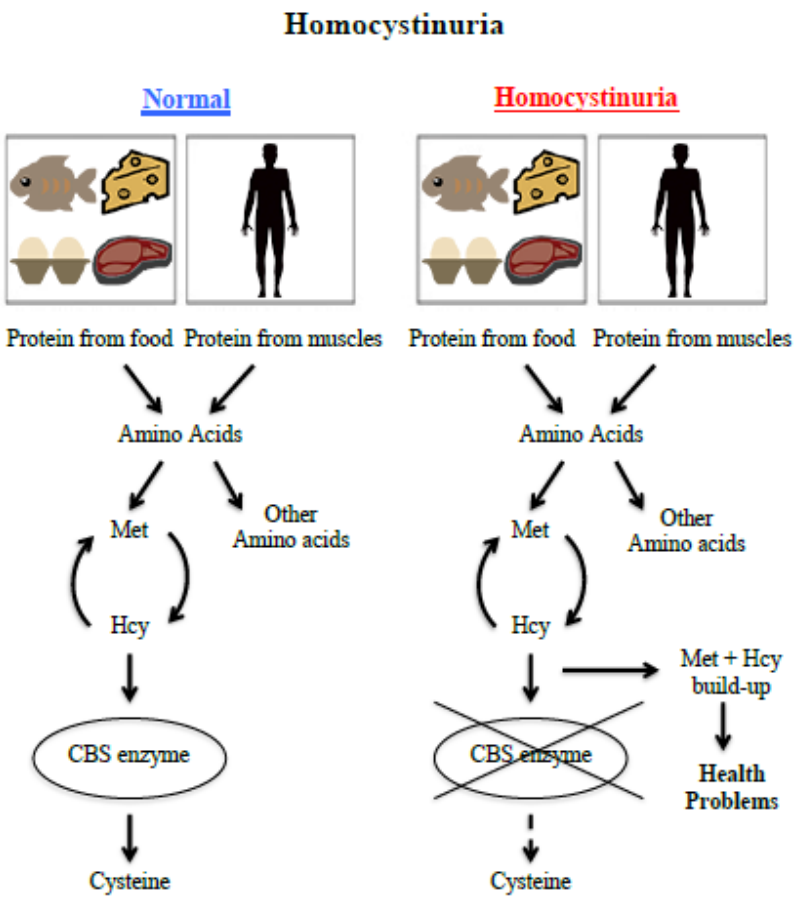
醫院管理局
HOSPITAL AUTHORITY

What is Homocystinuria (HCU)?

Homocystinuria is an amino acid disorder. People with amino acid disorders cannot process amino acids, the building blocks 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. Special enzymes then make changes to the amino acids so the body can use them.

Homocystinuria occurs when an enzyme called “cystathionine β-synthase” (CBS) is either missing or not working properly. The role of CBS enzyme is to break down methionine. When the CBS enzyme is not working correctly, its activity will be reduced. The conversion of amino acid methionine and homocysteine to cysteine will be disrupted. As a result, the harmful build-up of homocysteine and methionine in the blood cause health problems.



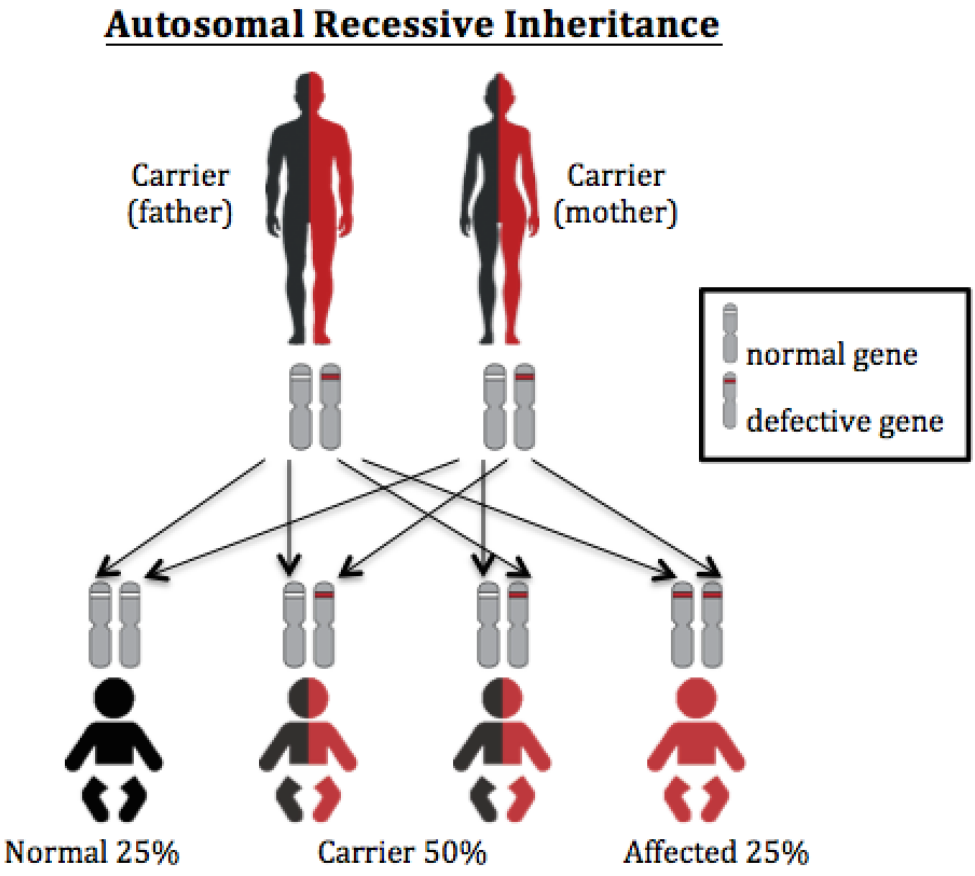
Met = Methionine

Hcy = Homocysteine

How is homocystinuria inherited?

Everybody has two copies of genes, one from each parent, which tell the body how to make specific enzymes.

Homocystinuria is an autosomal recessive disease. Only when babies inherit two faulty copies of the gene for homocystinuria from parents, the enzyme made does not work properly or is not even made at all.



What may happen if your baby has homocystinuria?

Babies with homocystinuria often appear healthy at birth. However, if the condition is not treated, symptoms can appear in early infancy. Symptoms vary from person to person – some people have very mild symptoms. The symptoms can be controlled with timely treatment.

Signs and Symptoms of homocystinuria

- Poor growth
- Learning disabilities or behavioural problems
- Bone and joint problems
- Vision problems such as short-sightedness
- A risk of developing blood clots and strokes

What is the treatment for homocystinuria?

The treatment aims to reduce and normalise homocysteine levels and prevent health problems.

- In some children, it is possible to control the levels of homocysteine with vitamin B6, also known as pyridoxine.
- Some children who do not respond to pyridoxine will require a low protein diet to reduce the amount of methionine and prevent homocysteine build up. A specialist dietician will advise and formulate a personalized meal plan on the foods of which your child will require.
- Medications may be prescribed additionally to help the body clear excess homocysteine.

When should I seek immediate help? What should I do?

If you are worried that your baby is ill, it is important to follow medical advice. Bring your baby to your local accident and emergency department immediately. Take the prescribed medication, special infant formula and any information that you have been given about homocystinuria, including this pamphlet, to the hospital with you.