

An Introduction to Homocystinuria (HCU)



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Important information

This is an introduction to HCU following a positive newborn screening for your child.

This guide is for information only and should not be a substitute for informed medical advice.

Consult your metabolic team for diagnosis and treatment for HCU.

If you are unclear about anything written in this booklet, please ask your dietitian for further information.

This booklet was written with Allyson Terry, Paediatric Dietitian - Metabolic. Alder Hey Children's Hospital, Liverpool



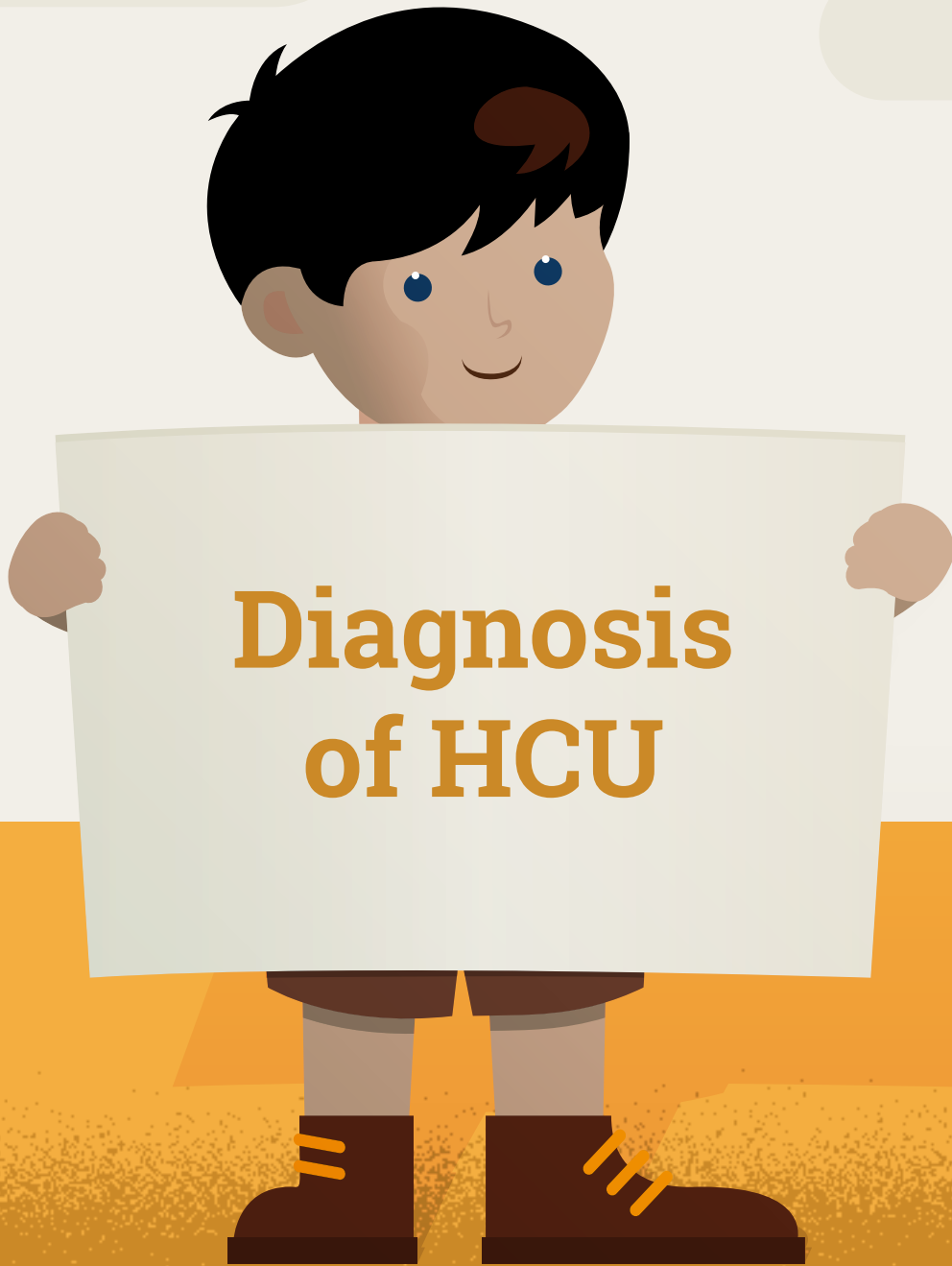
Homocystinuria (sounds like **Homo-cyst-in-u-re-ah**)
or HCU for short

This can also sometimes be known as cystathionine
beta-synthase (CBS) deficiency.

HCU is a manageable condition which affects the way the body breaks down protein.



Children with HCU have a defect in the process which breaks down part of a protein called methionine.



HCU is diagnosed by a blood test called the “newborn screening test”



This blood test (heel prick test) is carried out by a healthcare professional around 5 days after birth.

Under the national newborn screening programme, every baby in the UK and Ireland is screened for this condition.

There are two main forms of HCU



'Pyridoxine responsive HCU' can be treated with large doses of Vitamin B6 (pyridoxine). It is the milder form of HCU.

'Pyridoxine non-responsive HCU' does not respond to large doses of vitamin B6.

Initially all patients newly diagnosed with HCU are given a trial of pyridoxine treatment to establish whether they are responsive or not.

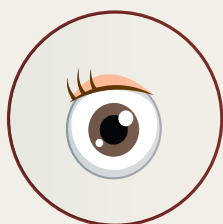
Your child has been diagnosed with pyridoxine non-responsive HCU.

This is managed by following a strict lifelong special low protein diet alongside medication.

If left untreated, substances in the blood build up to toxic levels and cause problems to several parts of the body which include:



Brain - Developmental delay and behavioural problems.



Eyes - Short sightedness, dislocation of the lens of the eye and glaucoma.




Blood vessels - Blood clots and risk of strokes.



Skeleton - Tall stature with long arms and legs and increased risk of broken bones.

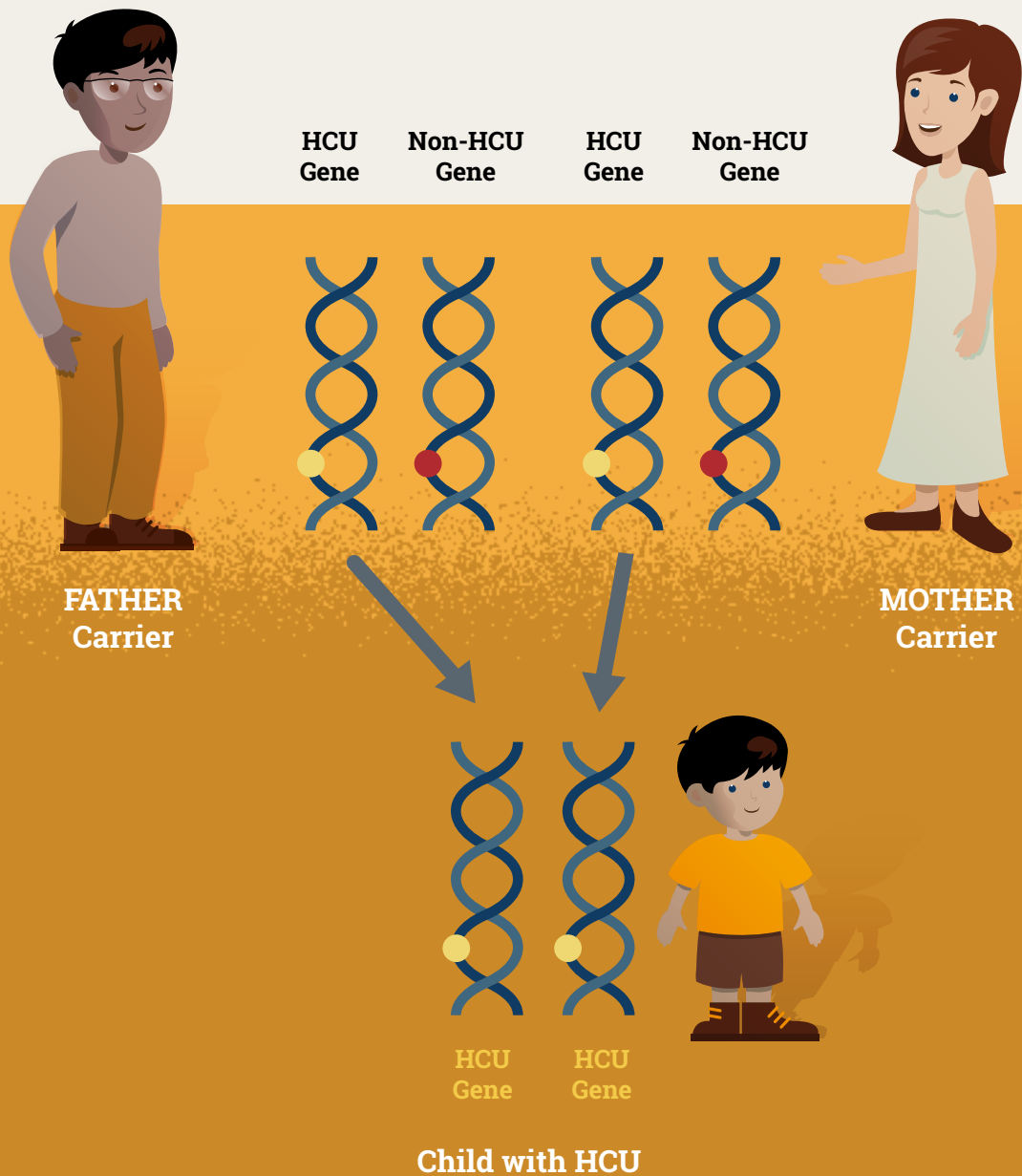
In newborn babies diagnosed with HCU, early treatment can prevent many of these symptoms from occurring and with life long management your child can reach their full potential.

A cartoon illustration of a young girl with long brown hair and bangs, smiling. She is holding a large white sign in front of her chest. The sign contains the text 'Why does my child have HCU?'. She is wearing a dark red top and dark red boots with white laces. The background is a simple landscape with a yellow ground, a white sky, and some stylized trees and clouds.

**Why does
my child
have HCU?**

HCU is an inherited genetic condition

It is nobody's fault and there is nothing you could have done to prevent it.



As a parent of a child with HCU, you have one HCU gene and one non-HCU gene.

This is known as being a carrier.

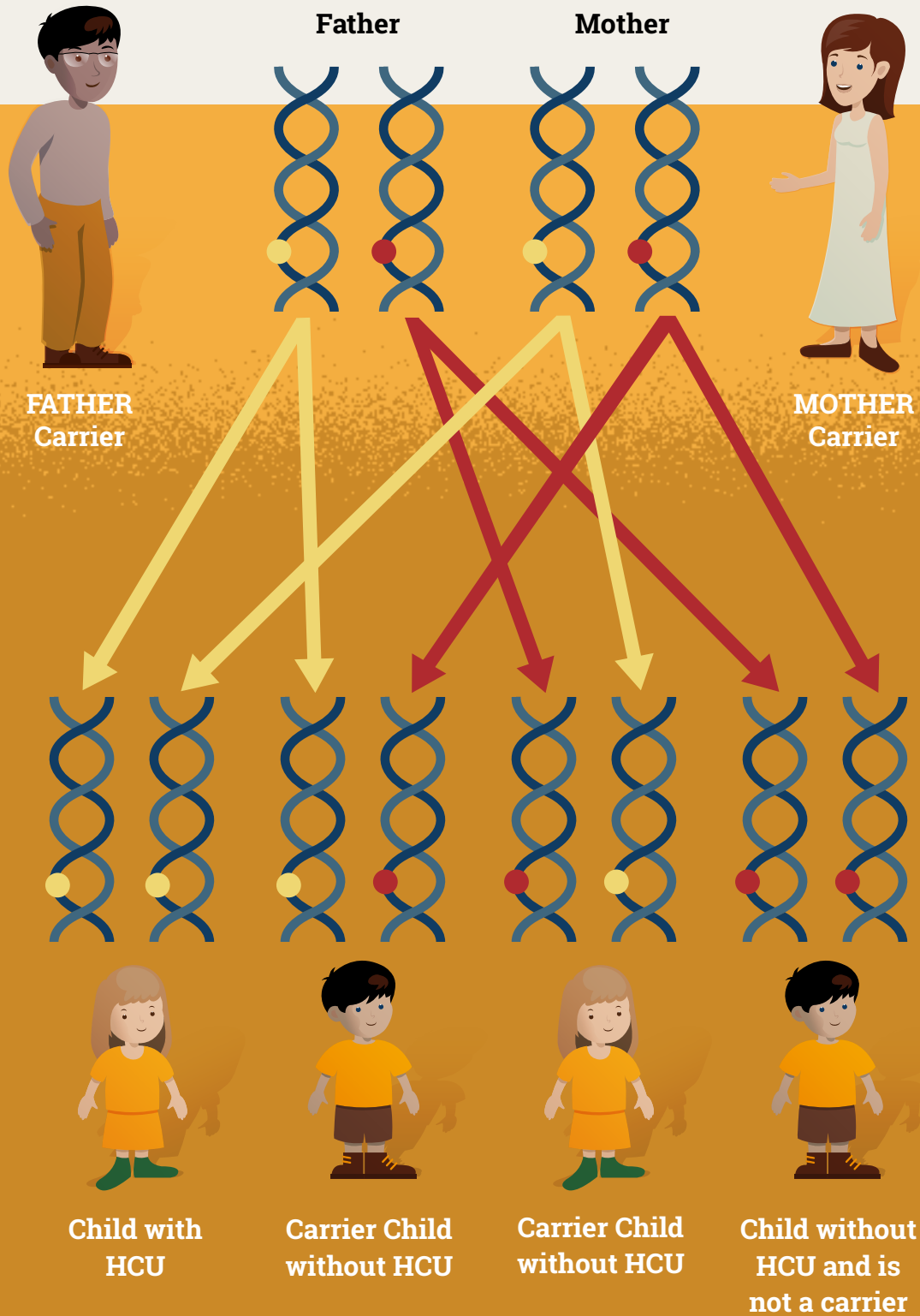
Your child has inherited 2 HCU genes, one from the father and one from the mother and so therefore will have HCU.

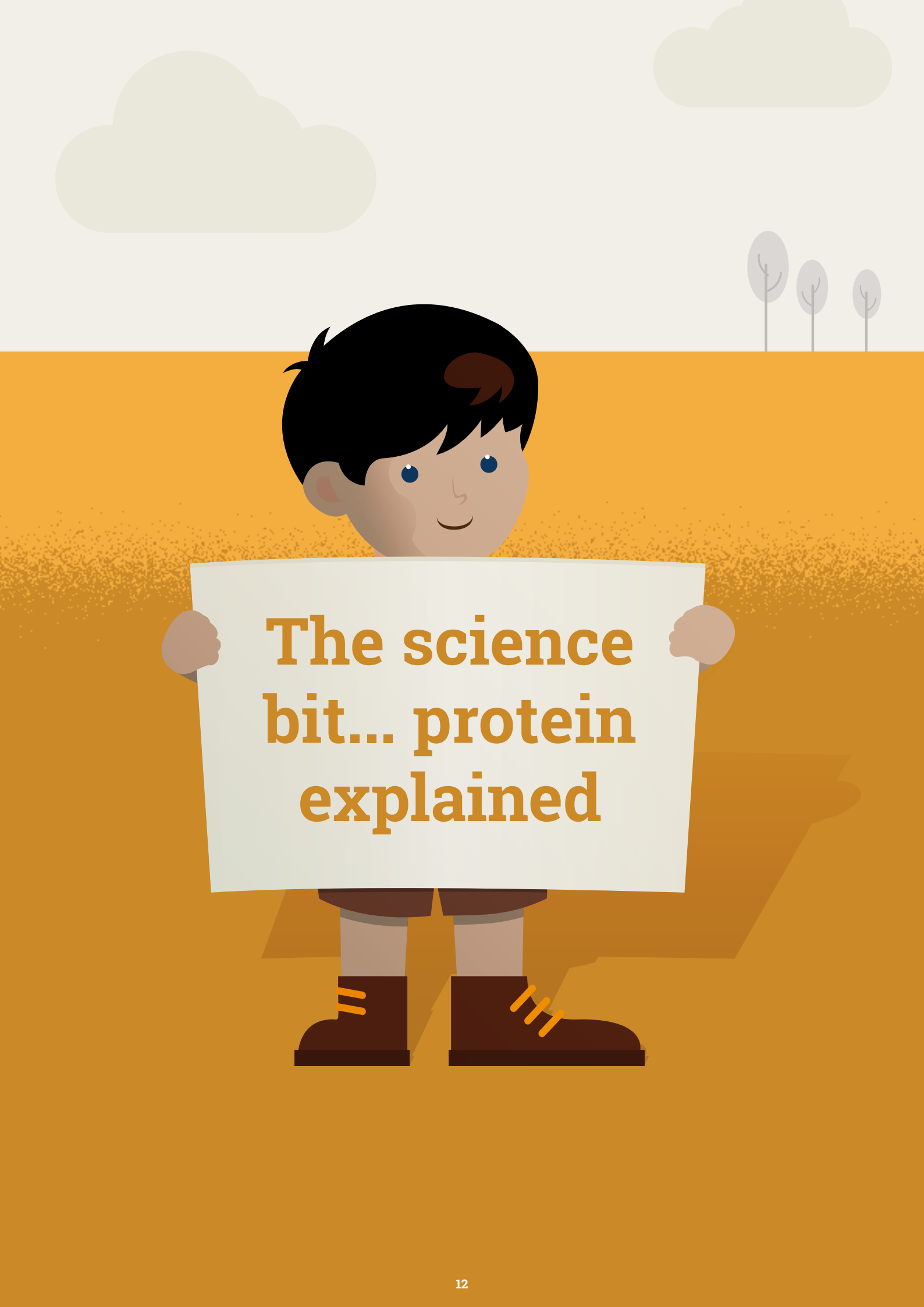
There are a small number of people who are carriers of the HCU gene



People who are carriers for HCU do not have HCU themselves and the HCU gene does not cause a problem to them.

When 2 people who are carriers of the HCU gene conceive a child there is a 1 in 4 chance of that child having HCU.

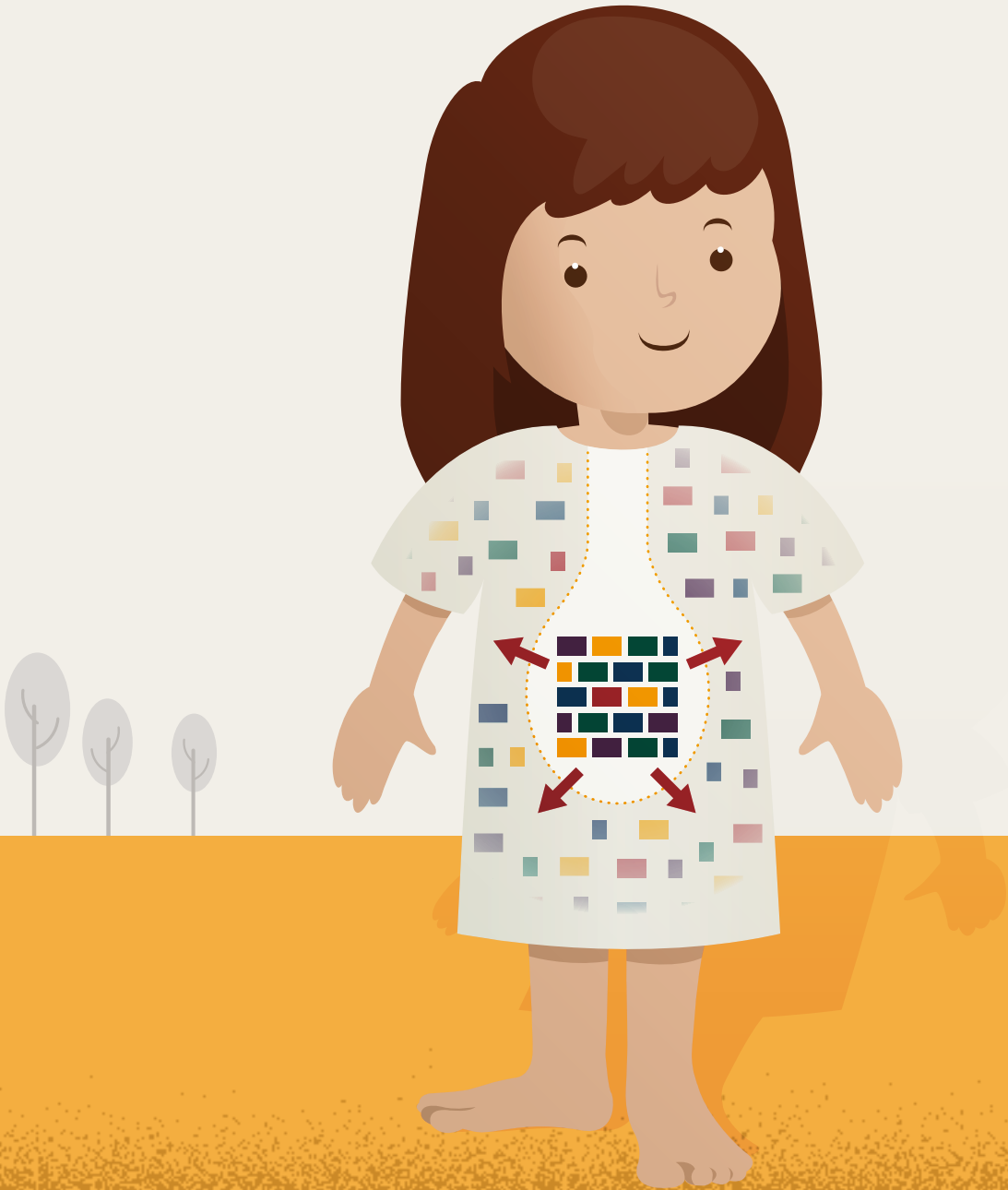


A cartoon illustration of a young boy with black hair and blue eyes, wearing a brown shirt and brown boots with yellow laces. He is holding a large white sign in front of him. The background is a bright orange field with a white sky, two light green clouds, and three stylized trees in the distance.

**The science
bit... protein
explained**

Why do we need protein?

Protein is one of the nutrients needed by the body. It helps to build, repair and maintain body cells and tissues, like your skin, muscles, organs, blood and even bones.



Protein is made up of different building blocks called amino acids. When it is eaten it is broken down into these amino acids so they can be absorbed into the blood stream and travel to where they are needed.

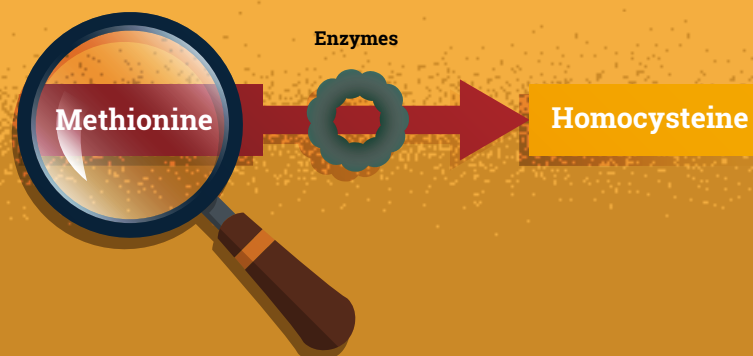
The conversion of protein into amino acids

Enzymes are special proteins which are like chemical scissors needed for the breakdown of protein into amino acids. There are 20 amino acids (building blocks) that make up protein.

Protein → **Enzymes** → **Amino acids (Building Blocks)**



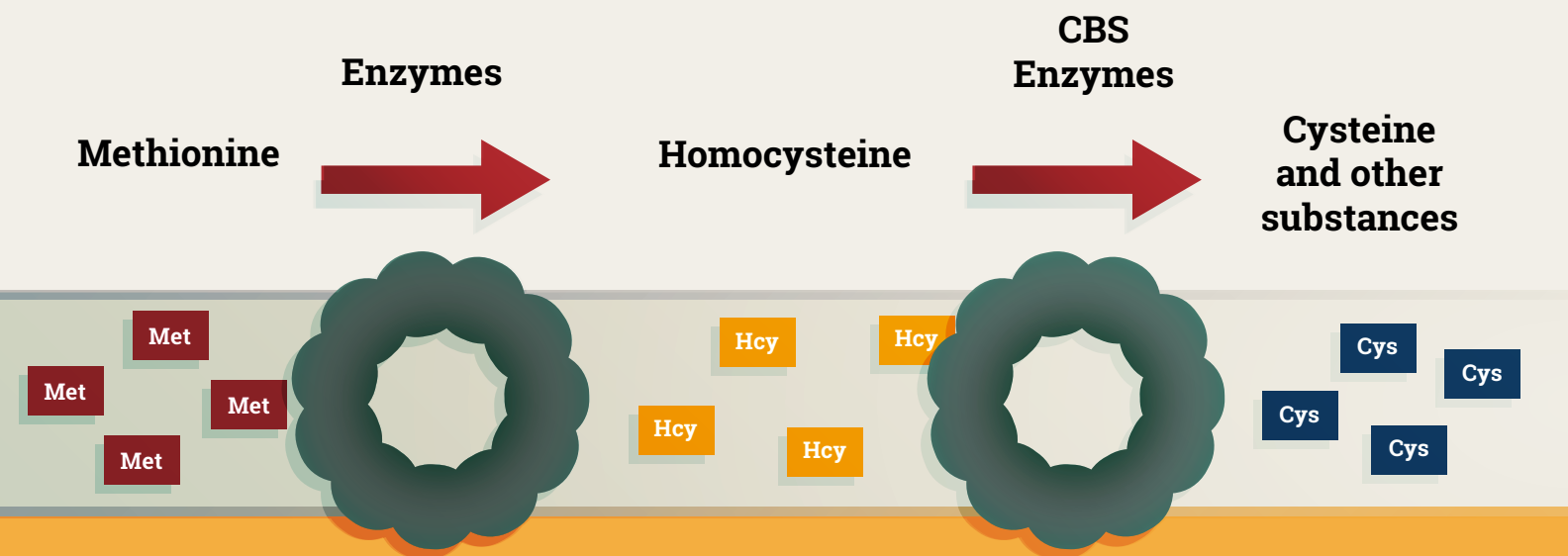
Methionine is one of these amino acids and is used by the body to make new substances, some is broken down further to form another amino acid called homocysteine.



It is homocysteine that a person with HCU cannot process correctly.

Why can someone with HCU not

A person without HCU

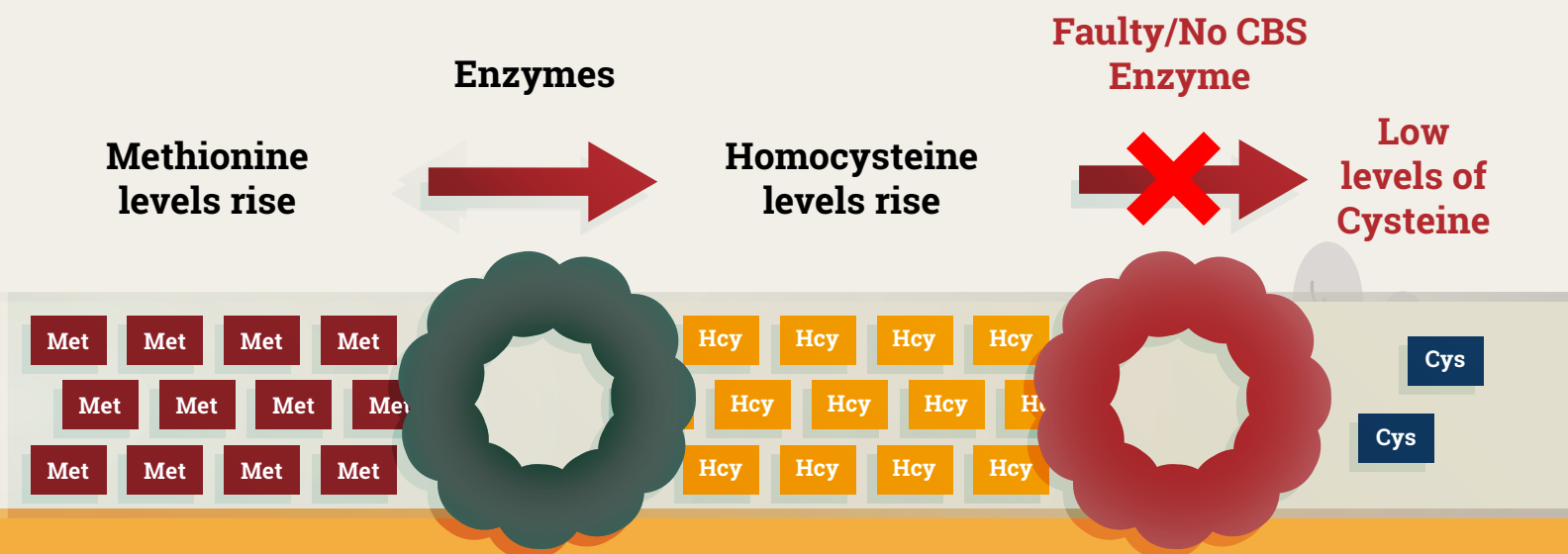


Some of the methionine is broken down further to make a new amino acid called homocysteine.

Normally the liver produces an enzyme called cystathionine beta-synthase (CBS) which helps homocysteine break down to cysteine.

process homocysteine correctly?

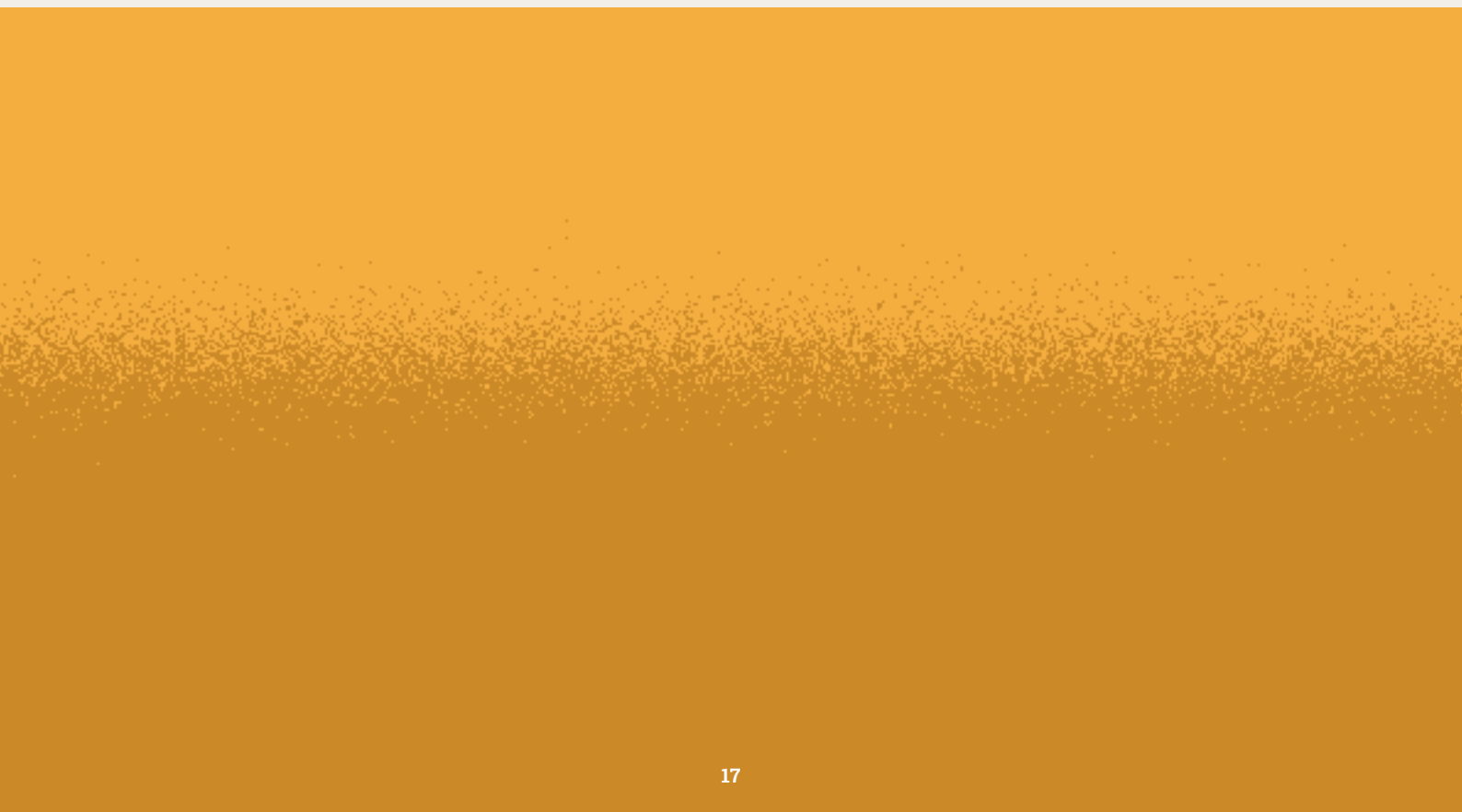
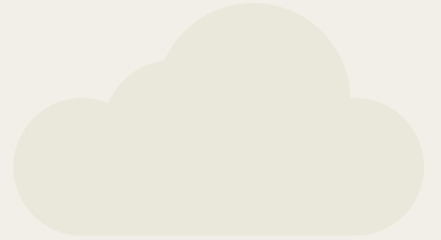
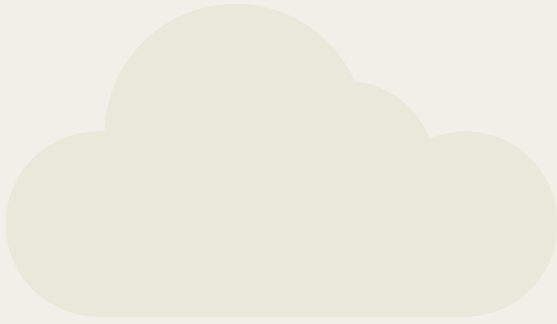
A person with HCU



In HCU the CBS enzyme is either not made by the liver or it does not work properly. This causes methionine and homocysteine to rise and cysteine levels to be low in the blood.

High levels of homocysteine are toxic in the blood and this results in damage to tissues in the body, such as the brain, eyes, blood vessels and skeleton.

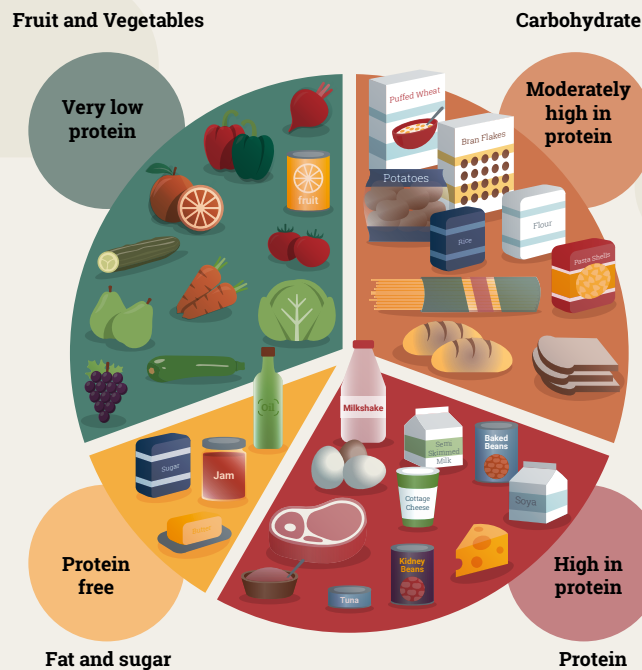
The severity of HCU varies and depends on whether the faulty CBS enzyme is completely inactive or can still break down some homocysteine.



A cartoon illustration of a young boy with black hair, blue eyes, and a slight smile. He is holding a large white rectangular sign in front of his chest with both hands. The sign has the text "Management of HCU..." written on it in a bold, orange, sans-serif font. The boy is wearing a brown shirt and brown boots with yellow laces. The background is a simple landscape with a light yellow sky, two light green clouds, and a yellow ground with a few stylized trees on the right side.

Management of HCU...

A Typical Diet



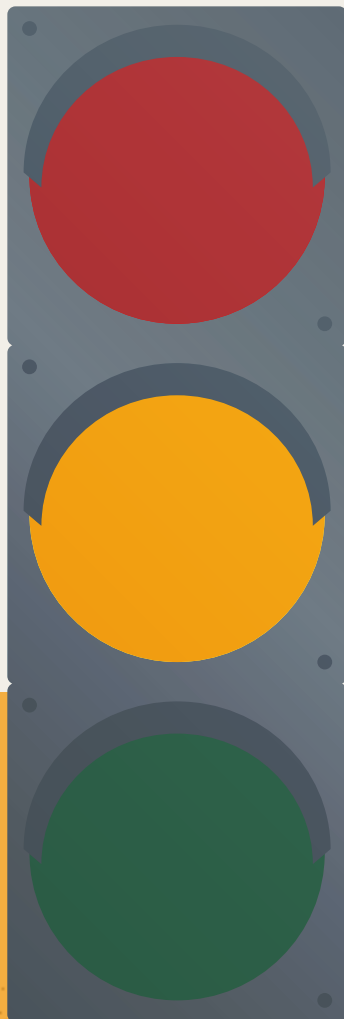
A typical diet has too much protein/methionine for a person with HCU. Therefore, they must follow a special low protein diet for life.

The HCU Diet

- HCU is managed by following a strict low protein/low methionine diet.
- If the amount of methionine in the diet is reduced, the amount of homocysteine produced by the body is also reduced. This is because homocysteine can only be made in the body from methionine.
- Methionine is found in all protein containing foods and therefore **must be restricted**
- All amino acids including methionine have a role or function in the body.
- It is therefore important that someone with HCU gets some methionine to support normal growth, but not so much that it becomes harmful.
- Extra vitamins or other medications may be prescribed, your doctor/dietitian will advise.

Traffic Light System

The low protein diet a person with HCU must follow is best described by using the traffic light system:



RED STOP!

Foods too high in protein/methionine

Do not eat these foods.

AMBER Go Cautiously!

Foods that contains some protein/methionine

These foods can be eaten in measured amounts (exchanges). Your dietitian will advise the amount your child can have.

GREEN Go!

Exchange-free foods - foods very low in/free from protein/methionine

These foods can be eaten without being measured.

RED STOP!

Do not eat these foods.

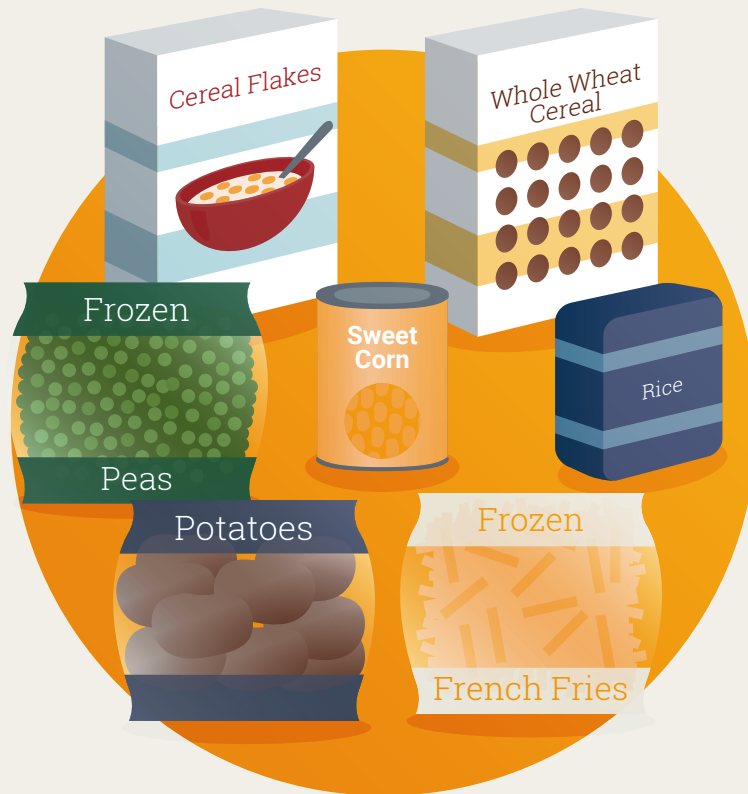


These foods are all rich in protein and therefore high in methionine, so they are not allowed.

These include meat, fish, eggs, cheese, chicken, quorn, nuts, soya, pulses and in foods such as normal bread, biscuits and chocolate.

AMBER Go Cautiously!

These foods contain a small amount of protein and therefore can be eaten in measured amounts and are known as exchanges.



Your child requires a small amount of methionine for growth, development and repair.

Your child will have a set number of exchanges allowed per day. Your dietitian will advise you how many and teach you how to calculate exchanges.

These foods are spread out between the day's meals. The quantities allowed will vary from person to person and from time to time in the same person.

* 1g protein is approximately 20mg methionine.

GREEN Go!

These foods are very low in protein and so can be eaten without being measured.

Specially manufactured foods



Exchange-free foods

These foods are either naturally low in protein and therefore methionine or have been specially manufactured to be low in protein (these foods are available on prescription) and so are exchange-free and do not have to be measured*.

These foods should make up the majority of your child's diet.
They provide a source of energy and variety in the diet.

* Please note certain specially manufactured low protein foods will need to be counted as part of methionine exchanges.
Please check with your dietitian.

A cartoon illustration of a young girl with long brown hair and bangs, smiling. She is holding a large white sign in front of her chest. The sign has the text "What is a protein substitute?" written in a bold, brown, sans-serif font. The background is a simple landscape with a yellow ground, a white sky, and a few stylized trees and clouds. The girl is wearing brown leggings and red shoes with white laces.

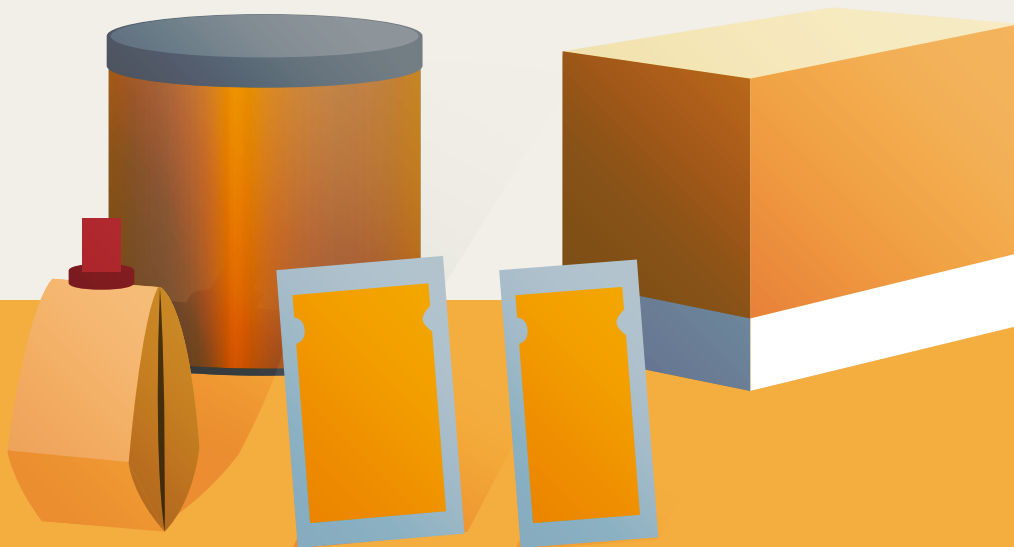
**What is a
protein
substitute?**

Why does a person with HCU need to take a protein substitute?

Due to the restrictive nature of the diet, individuals are unable to get all the nutrients required by food alone. This is why a protein substitute* is essential.

A protein substitute is a specially made medical food. It contains NO added methionine, but it contains all the other amino acids in protein that your child needs to grow and develop.

Most protein substitutes also contain vitamins, minerals and other important nutrients to ensure your child is getting what they need.



The protein substitute helps to keep the methionine, homocysteine and cysteine levels within an acceptable range throughout the day.

It is an extremely important part of the diet for a person with HCU.

It must be taken regularly and evenly spread over the day.

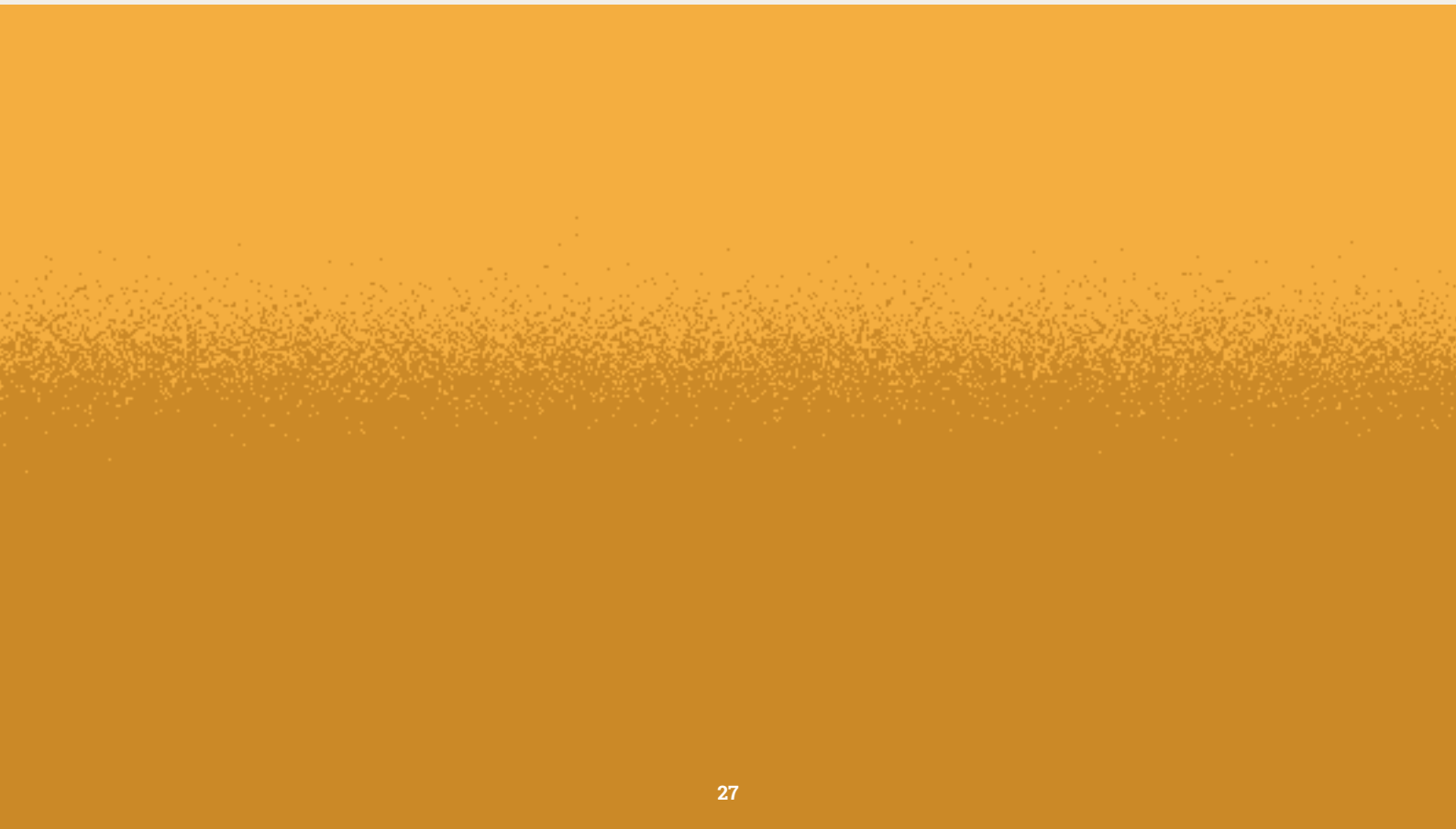
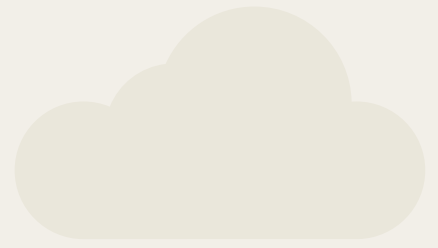
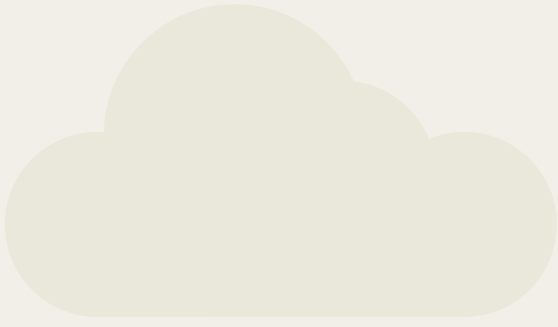
The protein substitute needs to be taken for life.


* Protein substitutes designed for the dietary management of HCU are Foods for Special Medical Purposes which must be used under medical supervision.

There are many different protein substitutes which are designed to fit easily into everyday life and offer choice so that you find the right product to suit your child.



Your dietitian will advise you on which protein substitute is best for your child at each stage of their development.



A cartoon illustration of a young boy with black hair, blue eyes, and a slight smile. He is wearing a brown shirt and brown boots with yellow laces. He is holding a large white sign in front of his chest. The background is a simple landscape with a yellow ground, a white sky, and two light green clouds. In the distance, there are three stylized trees with grey trunks and light blue foliage.

**How will I
feed my new
born baby?**

Infants: breastfeeding*

Babies diagnosed with HCU will have high methionine and homocysteine levels initially.
To reduce these high levels:



Step 1: Your baby will be given a special methionine-free formula instead of breastfeeds until the levels of methionine fall back to normal levels (usually within a few days).

Step 2: Once the levels of methionine and homocysteine are under control, a measured amount of the methionine-free formula is given before a breastfeed.

This is to restrict the amount of methionine taken by the baby from the breastmilk. Breastmilk provides limited but essential methionine for normal growth. You will be advised by the dietitian on how much methionine-free formula and breastmilk to give.

Keeping a record of your baby's intake may be useful.

* Breastfeeding offers many benefits for baby and mother; it is recommended you discuss this with your health care professional.

Infants: bottle feeding

Babies diagnosed with HCU will have high methionine and homocysteine levels initially.
To reduce these high levels:

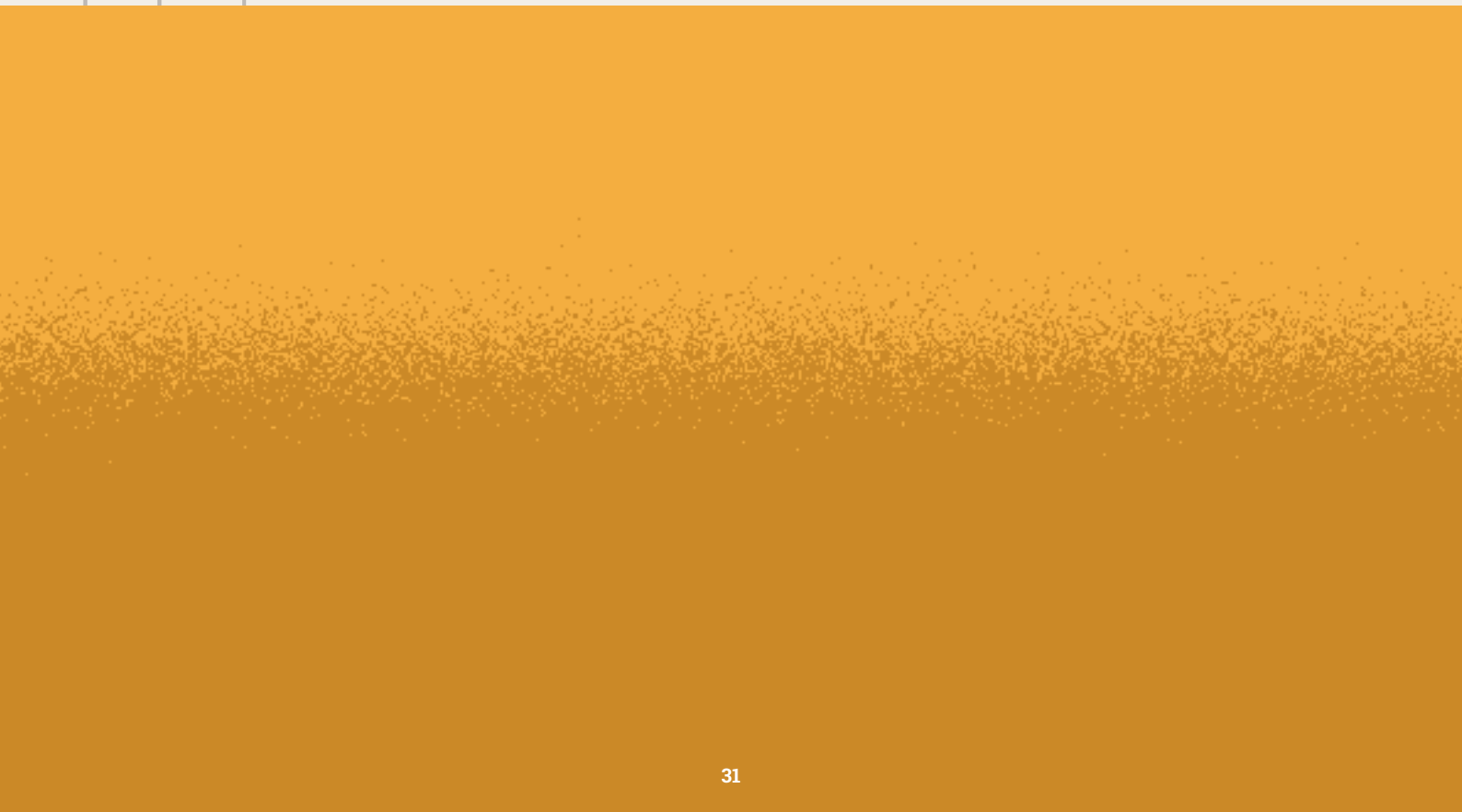
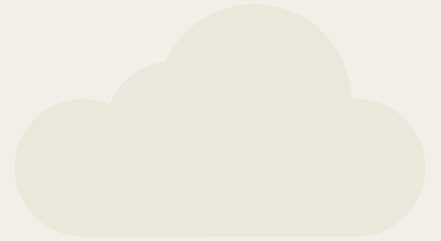
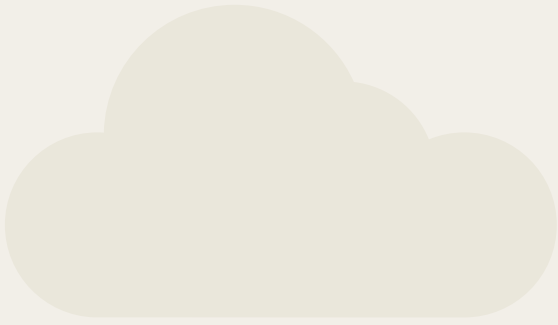


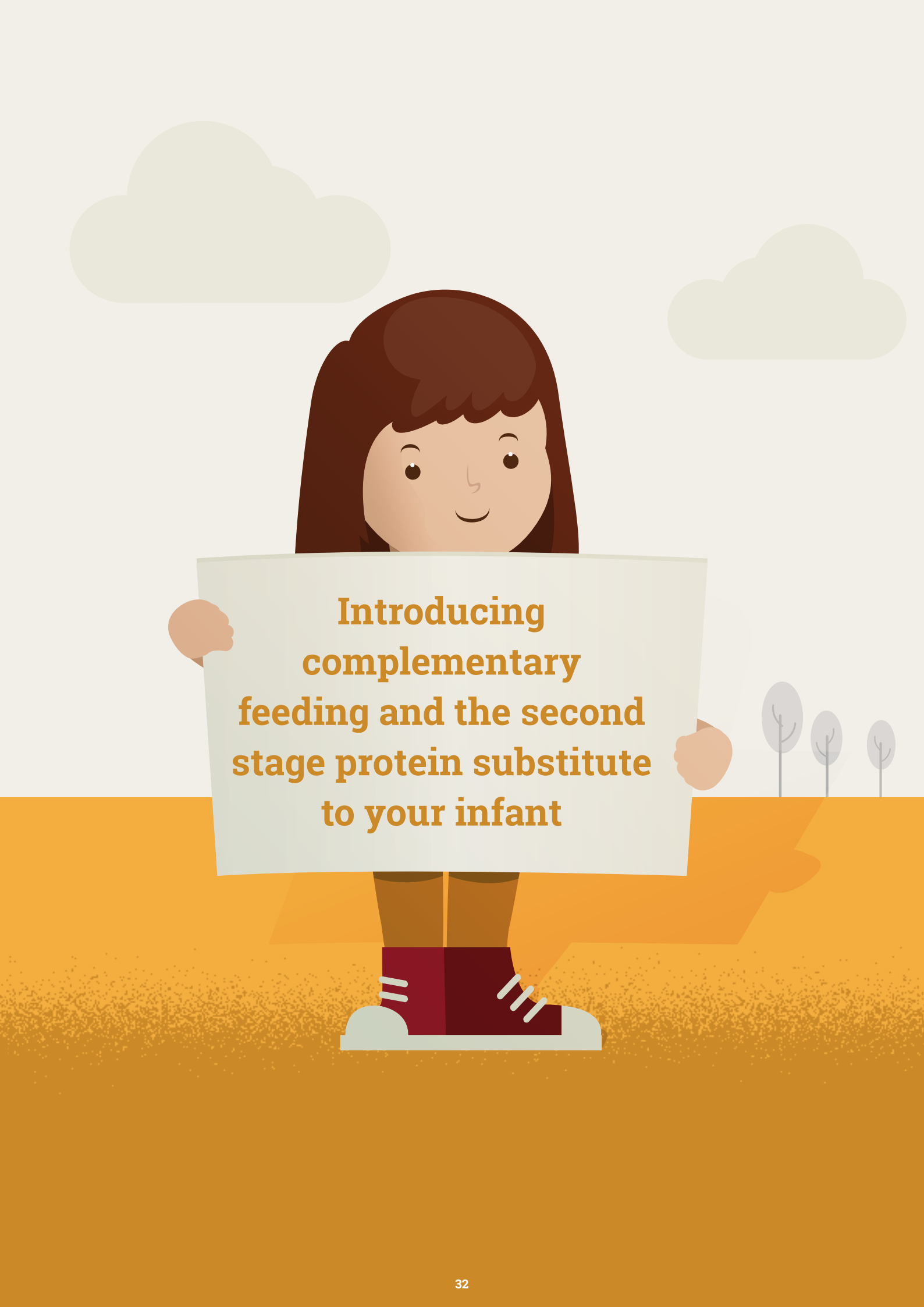
Step 1: Your baby will be given a special methionine-free formula instead of standard infant formula until the levels of methionine fall back to normal levels (usually within a few days).

Step 2: Once the levels of methionine and homocysteine are under control, a small restricted amount of standard infant formula will be given plus the methionine-free formula.

The measured amount of standard infant formula will provide the limited methionine essential for normal growth and development. You will be advised by the dietitian on how much methionine-free formula and standard infant formula to give.

Do not mix the methionine-free formula and measured amount of standard infant formula together. Give them in 2 separate bottles.
Keeping a record of your baby's intake may be useful.



A stylized illustration of a woman with long brown hair and bangs, smiling. She is holding a large white rectangular sign in front of her chest. The sign contains text in a bold, orange-brown font. The background is a light cream color with two soft, greyish clouds. To the right, there are three simple, grey trees on a yellow-orange ground. The woman is wearing a dark red top and dark red shoes with white laces. The overall style is clean and modern.

**Introducing
complementary
feeding and the second
stage protein substitute
to your infant**

When to start complementary feeding*

Complementary feeding is often referred to as weaning. It is the process of gradually introducing solid foods into your baby's diet. Your dietitian will advise you when to start solid foods, but it will occur around the same time as babies without HCU.

First foods introduced will initially be exchange-free foods.

This will help your baby to develop a healthy eating pattern while eating a variety of foods.

Your baby needs to continue to take their methionine-free formula and breast milk/standard infant formula as advised by their dietitian.



This is also around the time that a second stage protein substitute can be introduced, your dietitian will advise you about this.

* This may occur a little earlier than 6 months, depending on your baby, but it should never be started before 17 weeks.

7 to 12 months

Once your baby is accepting some exchange-free foods, **foods containing measured amounts of methionine (exchanges) are introduced.**

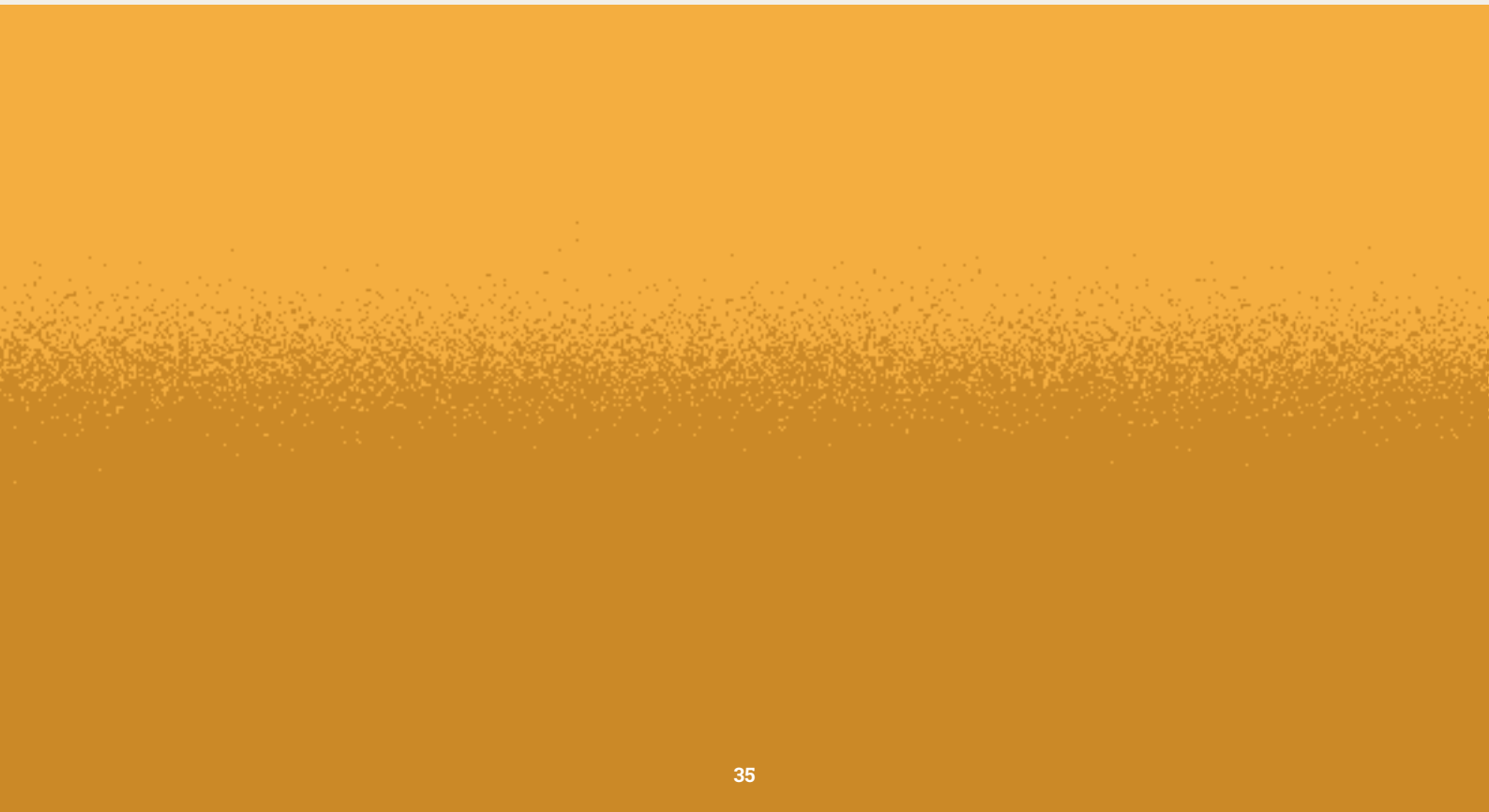
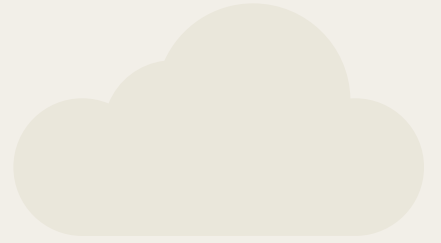
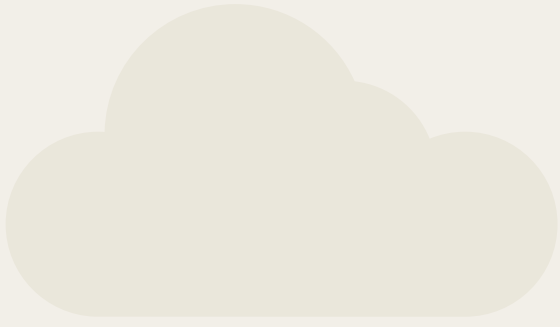
Over time, they will get more of their exchanges from food (e.g. potato or cereal) and replace the protein they were getting from breast milk or standard infant formula.

Exchange-free foods should still be included in the diet as well.

Your child will also be encouraged to move to more textured food and finger foods to encourage self-feeding. Your baby will also begin to drink from a beaker.



Your dietitian will provide more detail about introducing complementary feeding and introducing the second stage protein substitute.

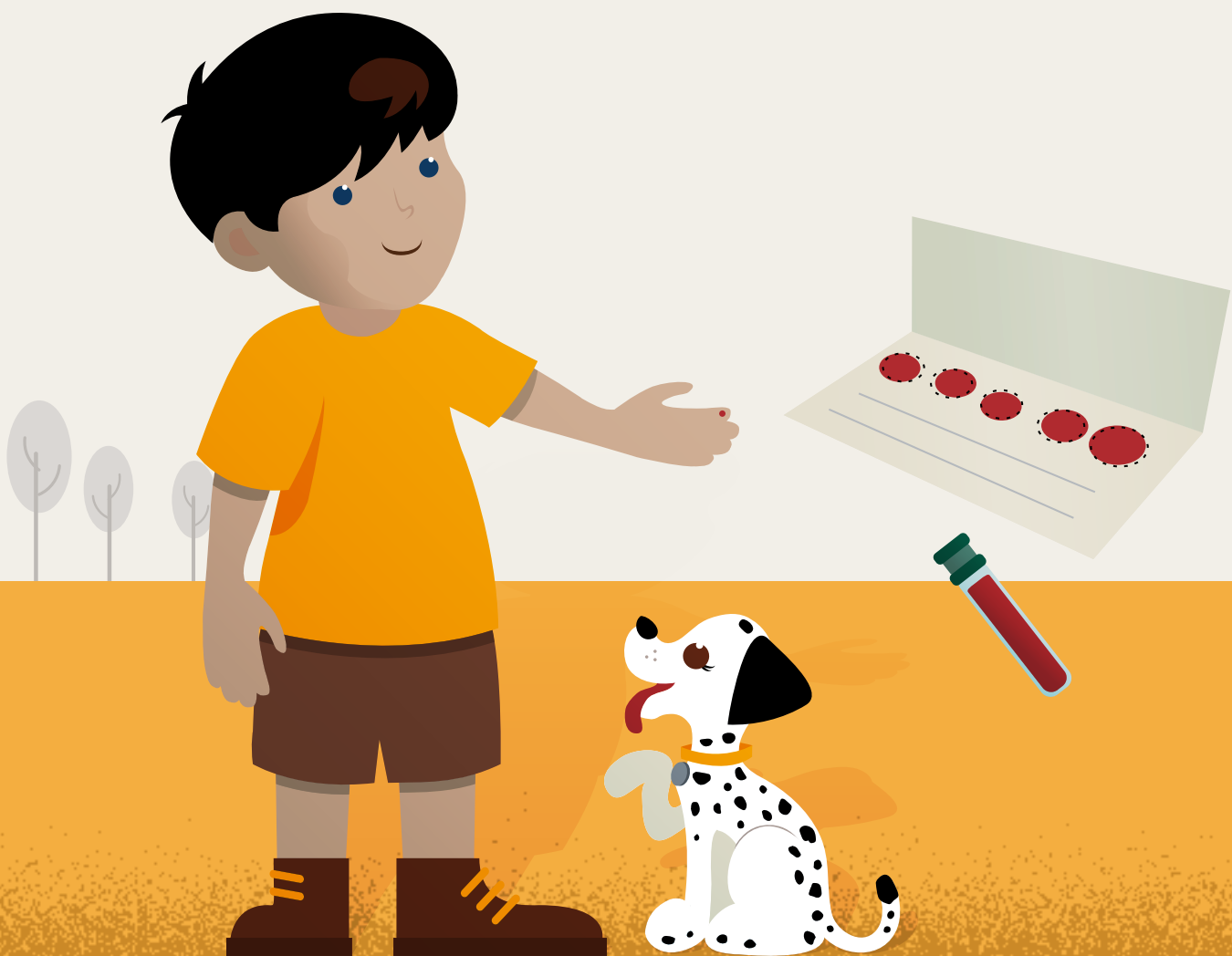




Your child will have regular blood tests and monitoring to make sure their methionine, homocysteine and cysteine levels are under control.

This is very important to ensure that they do not have too little or too much methionine in their diet.

The results of these blood levels are used to adjust the diet and medications as required.



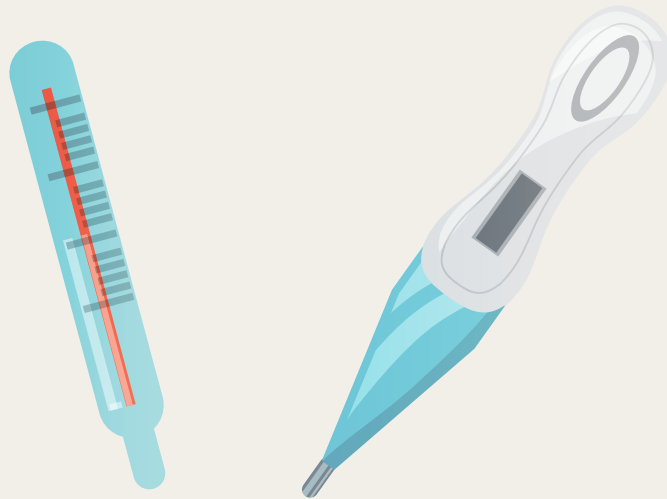
Your dietitian will contact you once the results are processed and discuss any changes that may be needed to your child's diet.



Illness

As with all babies and children illness will occur from time to time.
Contact your GP if your child is unwell to get advice on treatment and
let them know your child has HCU.

Give any medications recommended by your doctor.



Maintaining your baby's fluid intake during illness is important and extra fluid may be necessary, particularly if your baby has vomiting or diarrhoea.

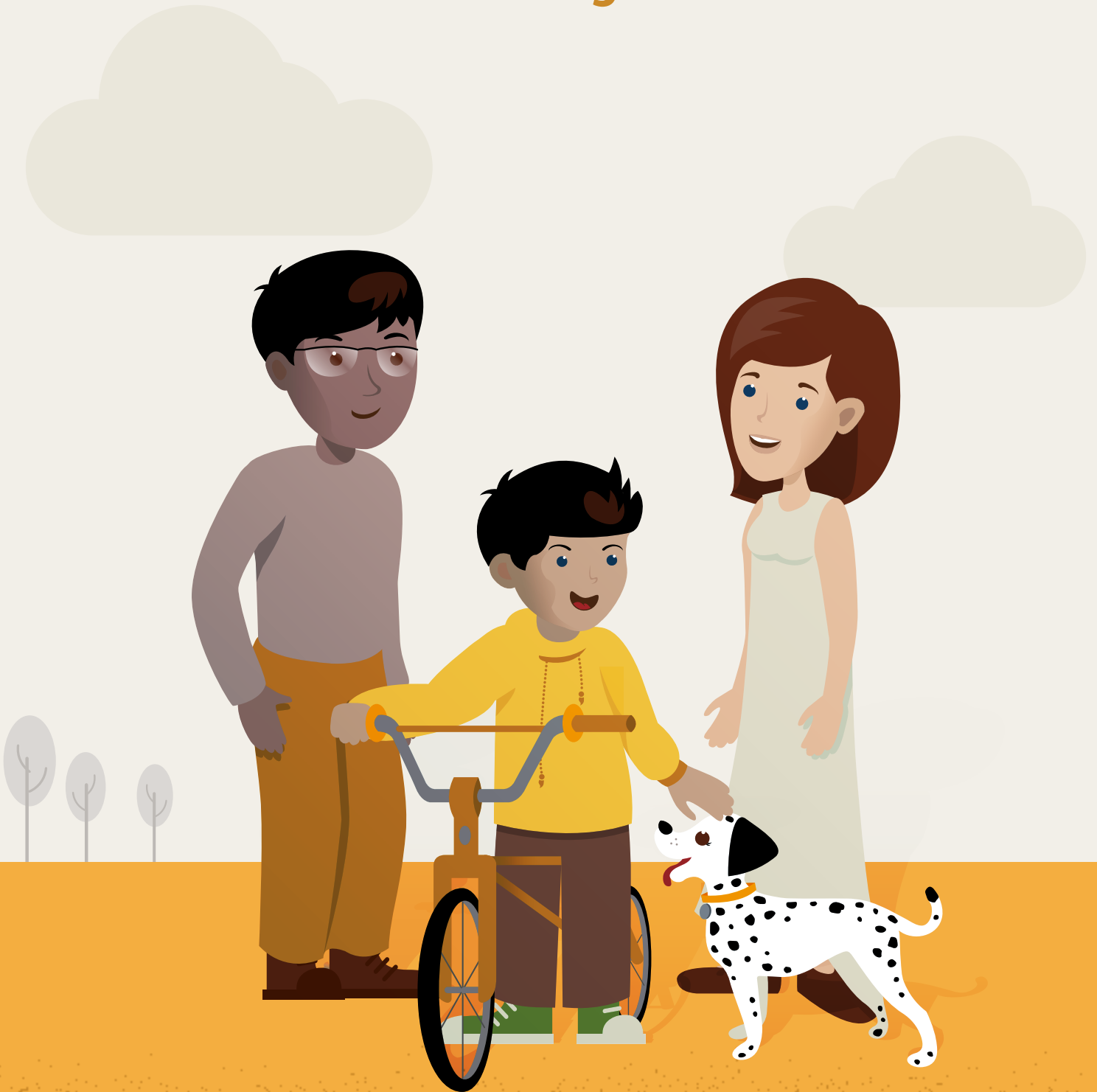
It is important that your child continues to take their protein substitute during illness to help maintain metabolic control. Speak to your dietitian for advice.

If your baby has to have any planned surgery contact your metabolic team for additional advice.

A stylized illustration of a woman with long brown hair and bangs, smiling. She is holding a large white sign in front of her chest. The sign has the text "As your child grows up" written in a bold, orange, sans-serif font. The woman is wearing a brown top and dark red shoes with white laces. The background is a simple landscape with a light yellow sky, two light green clouds, and a field of orange grass. In the distance, there are three small, grey, stylized trees. The overall style is clean and modern.

**As your
child
grows up**

Children / Teenagers / Adults:



Your child will continue to follow a low protein (low methionine) diet as they grow up into adulthood.

You, your child and dietitian will help choose the protein substitute that best suits them and their lifestyle.

Consistent good metabolic control throughout life ensures the best chance of a healthy life. HCU should not restrict your child or your family from everyday activities, for example going on holiday*, eating out or playing sport.

* If you are going on a long haul flight contact your metabolic team for additional advice.

Useful Information

Dietitian:

Nurse:

Doctor:

Other contacts:

Help and support for families living with HCU

METABOLIC SUPPORT UK

Metabolic Support UK (formerly known as CLIMB) are the leading patient organisation for Inherited Metabolic Disorders supporting thousands of patients worldwide. Its mission: to be the world's leading advocate and provider of bespoke support to families; signpost to the best experts; raise awareness amongst the medical, healthcare, social care and teaching professions; and fund research for treatments and ultimately cures. It is part of a worldwide collaborative network of groups and organisations, ensuring it is at the forefront of developments such as newborn screening and treatments.



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