

TEMPLE



Tools **E**nabling **M**etabolic **P**arents **L**Earning

ADAPTED BY THE DIETITIANS GROUP

BIMDG

British Inherited Metabolic Diseases Group



BASED ON THE ORIGINAL TEMPLE WRITTEN BY
BURGARD AND WENDEL

VERSION 4, NOVEMBER 2021

HCU

Supported by **NUTRICIA**
as a service to metabolic medicine

TEMPLE foreword

TEMPLE (Tools Enabling Metabolic Parents LEarning) are a set of teaching slides and booklets that provide essential information about different inherited metabolic disorders that require special diets as part of their management. These teaching tools are aimed at parents who may have an infant or child that has been recently diagnosed with a disorder. They are also useful when teaching children, extended family members, child minders, nursery workers and a school team.

They have been developed by a team of experienced clinical and research metabolic dietitians from the UK who are members of the British Inherited Metabolic Disease Group (BIMDG).

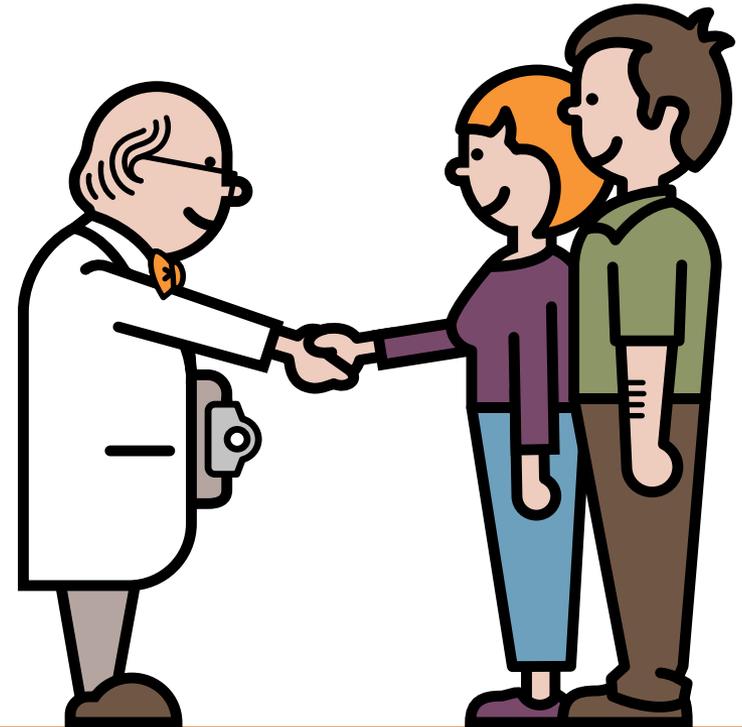
The team are Rachel Skeath, Karen van Wyk, Pat Portnoi and Anita MacDonald. The group is facilitated by Heidi Chan from Nutricia.

Each module produced is reviewed by a consultant clinician who is a member of the BIMDG.

This teaching tool is not designed to replace dietary information that may be given by a dietitian in clinic.

HCU

Information for families following
a positive newborn screening



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VERSION 3, APRIL 2020

TEMPLE



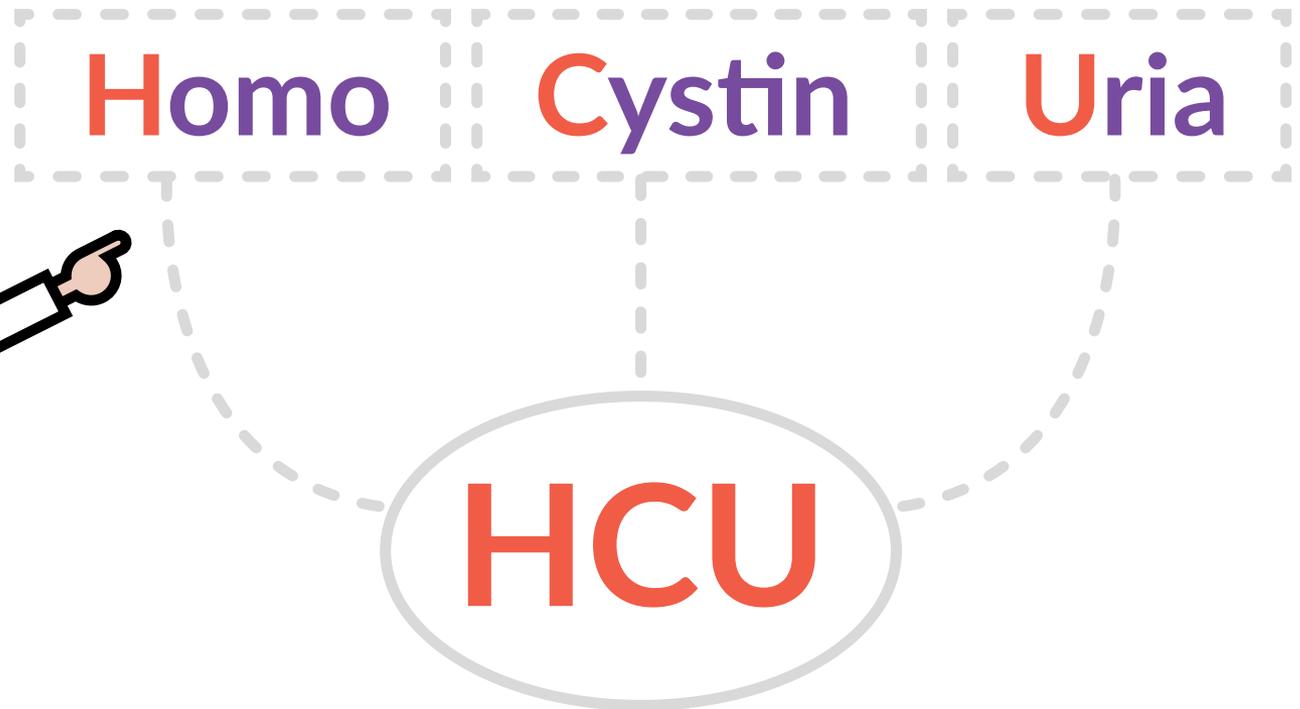
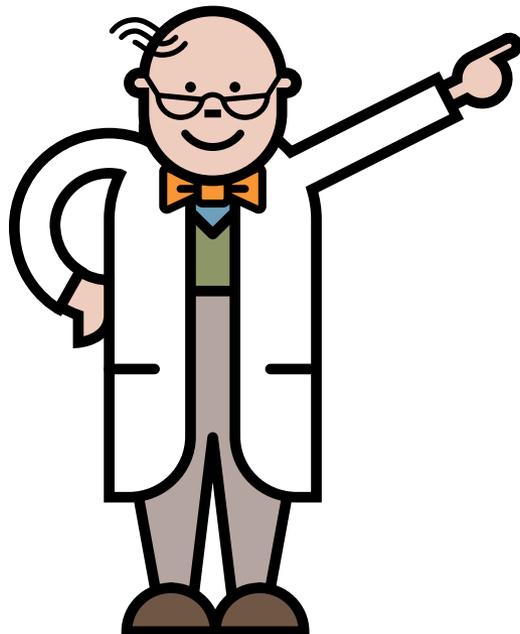
Tools Enabling Metabolic Parents LEarning

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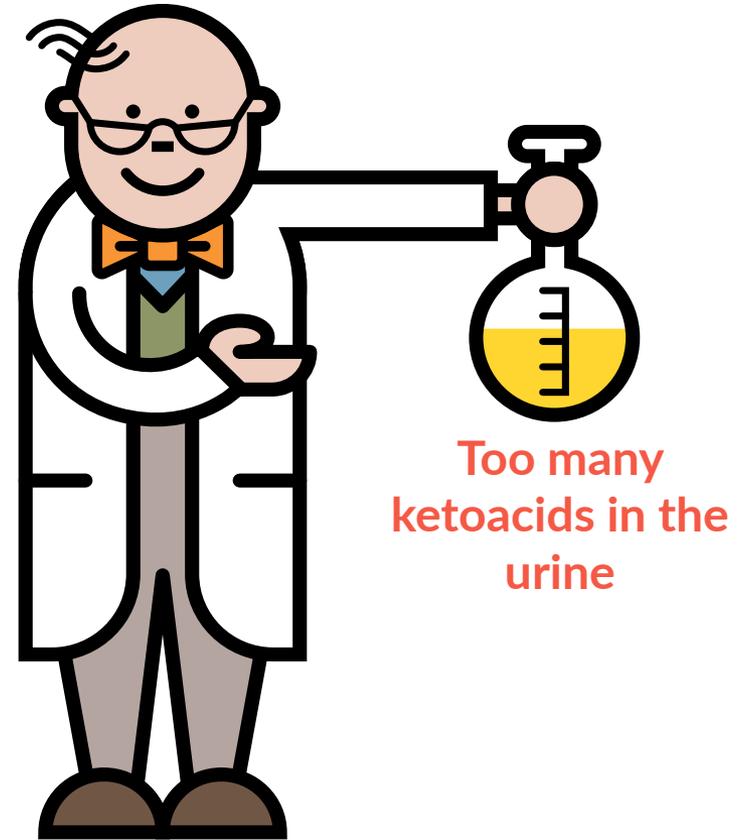
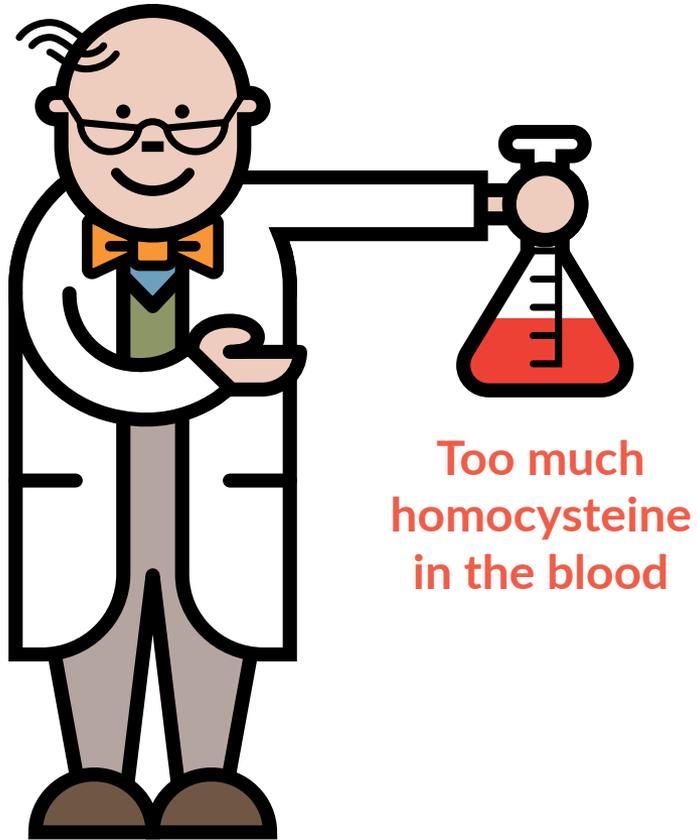
What is HCU?

HCU stands for Homocystinuria

It is an inherited metabolic condition



What is HCU?

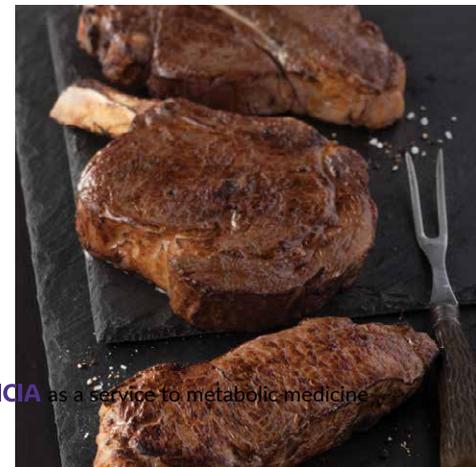


HCU and protein

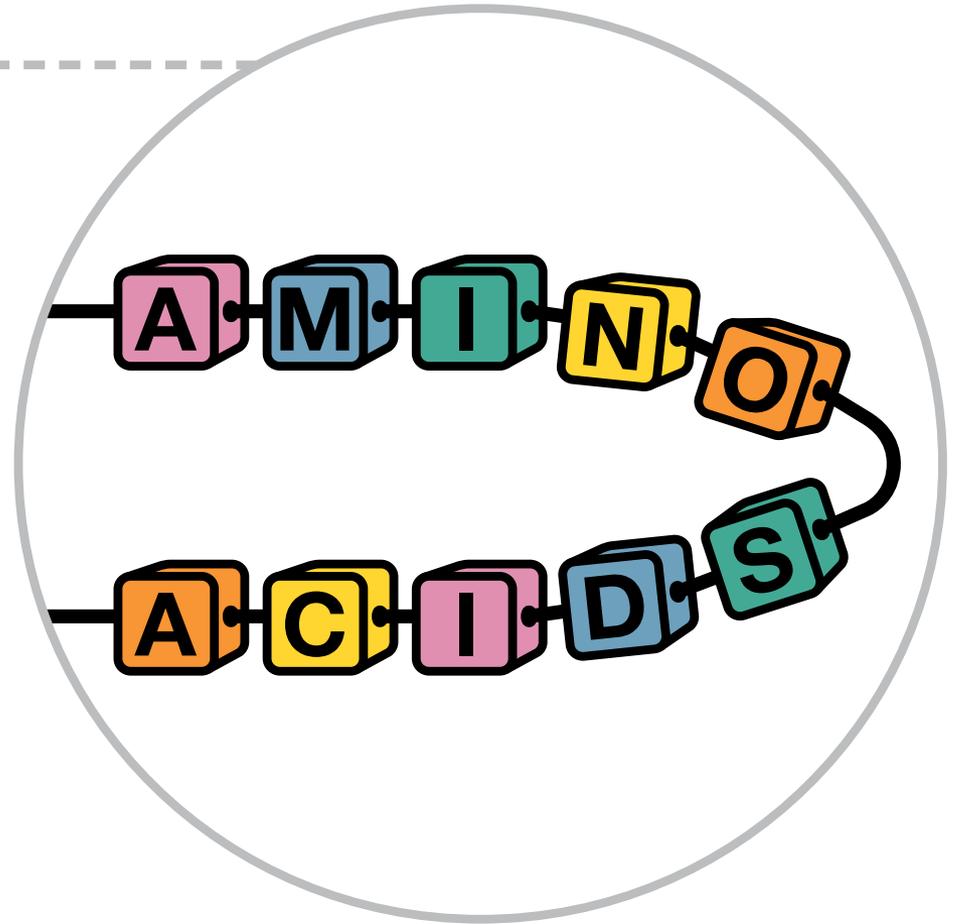
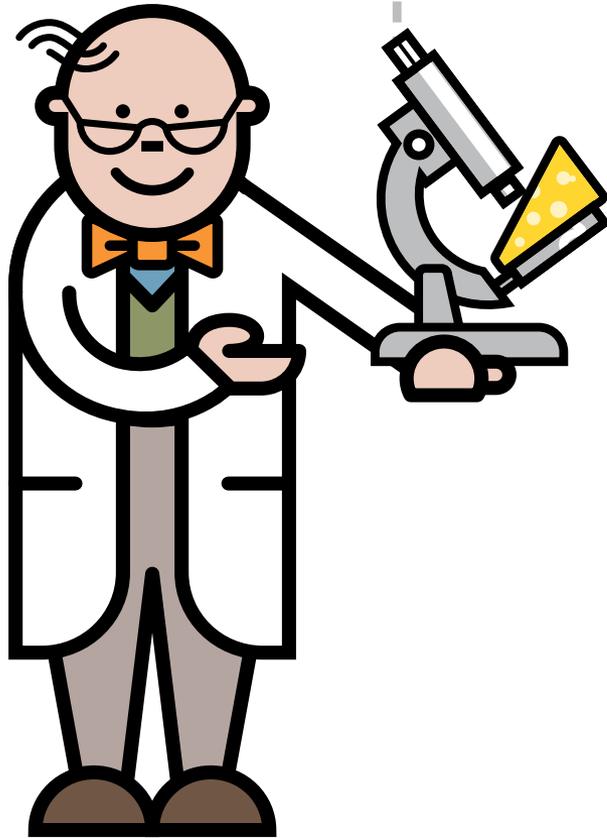
HCU affects the way your baby breaks down protein

Many foods contain protein

The body needs protein for growth and repair



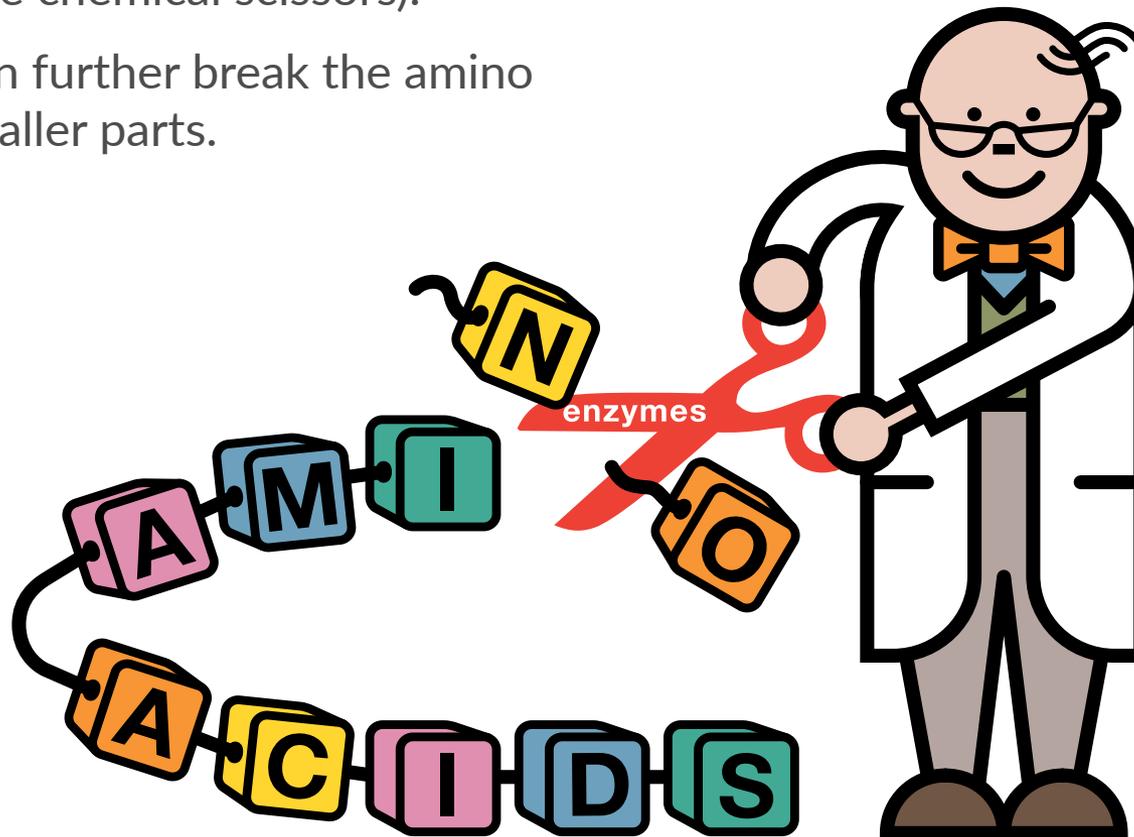
What is protein?



Protein and enzymes

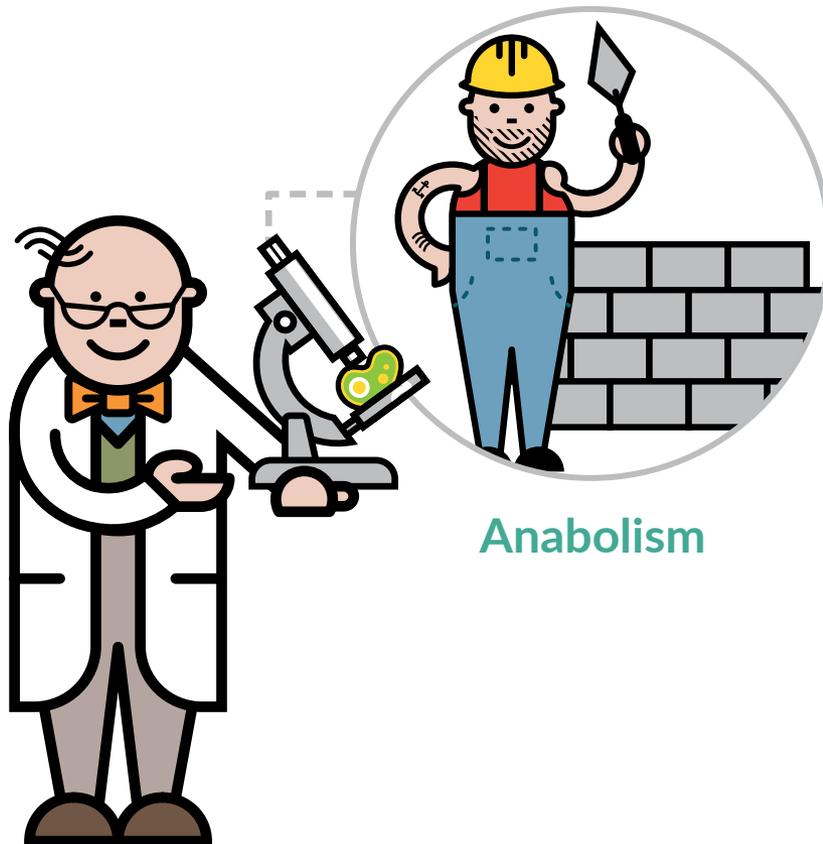
Protein is broken down into amino acids (building blocks of protein) by enzymes (which are like chemical scissors).

Enzymes then further break the amino acids into smaller parts.

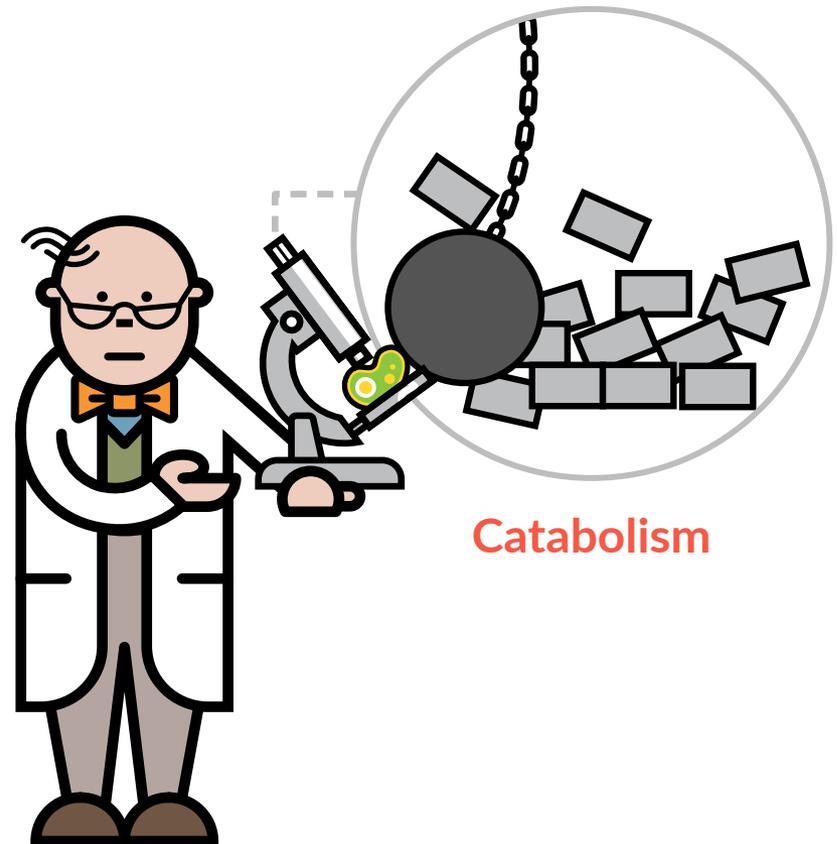


Protein metabolism

Metabolism refers to the chemical processes that occur inside the cells of the body.



Anabolism

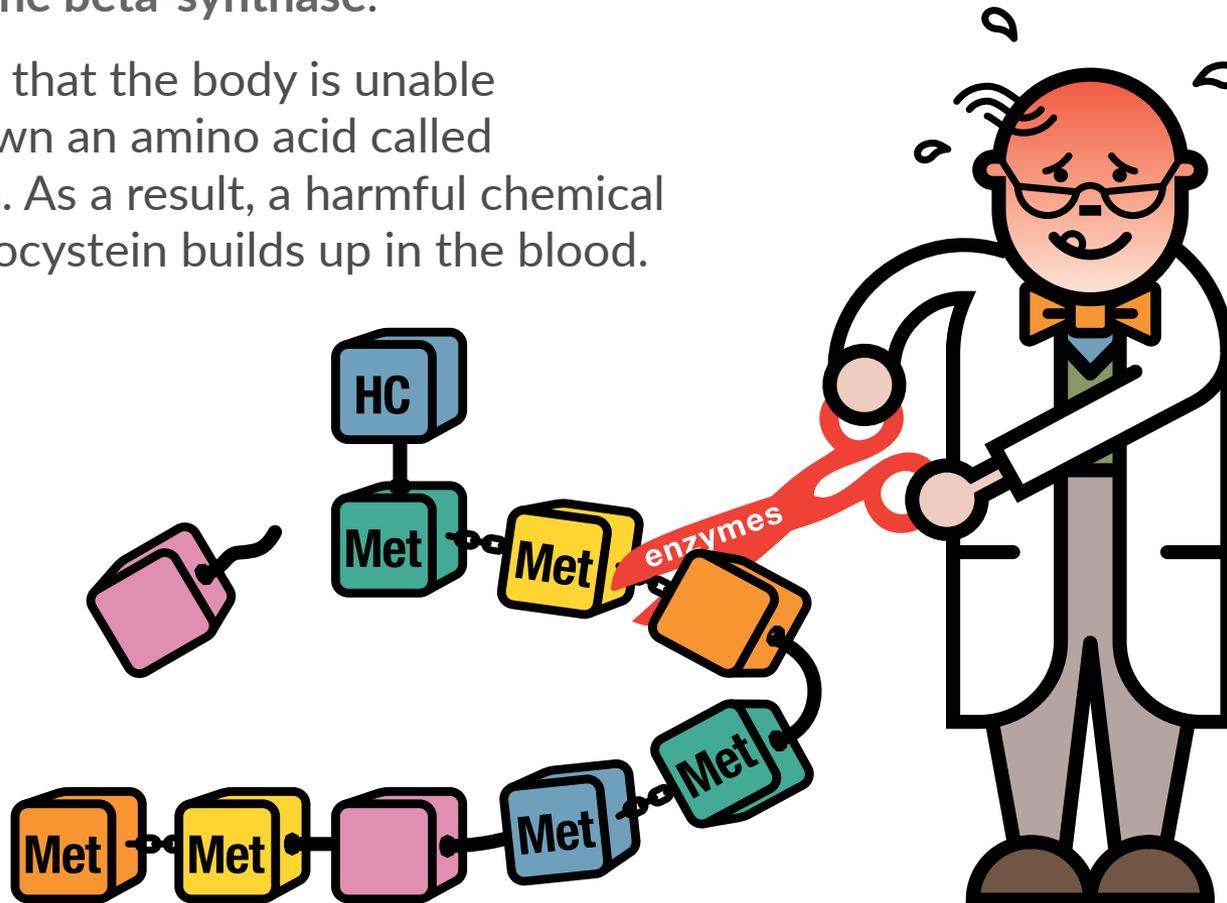


Catabolism

What happens in HCU?

In HCU the body lacks an enzyme called cystathionine beta-synthase.

This means that the body is unable to breakdown an amino acid called methionine. As a result, a harmful chemical called homocystein builds up in the blood.



What can go wrong in HCU?

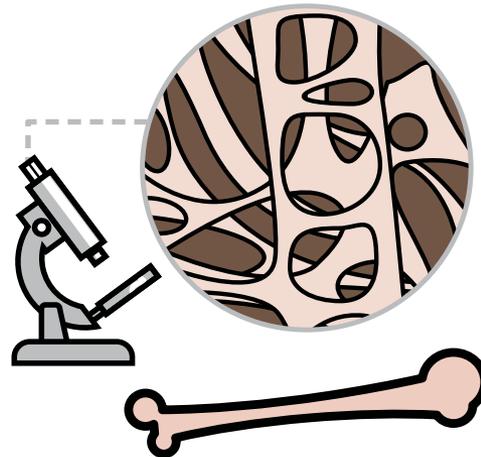
Short sightedness and dislocated eye lenses



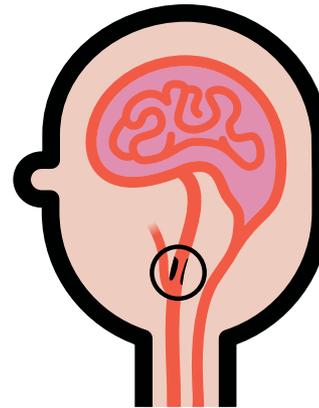
Difficulties with learning and behaviour



Long and thin bones



Blood clots and strokes



How is HCU diagnosed?

HCU is diagnosed by newborn screening. High levels of methionine and homocysteine are found in the blood.

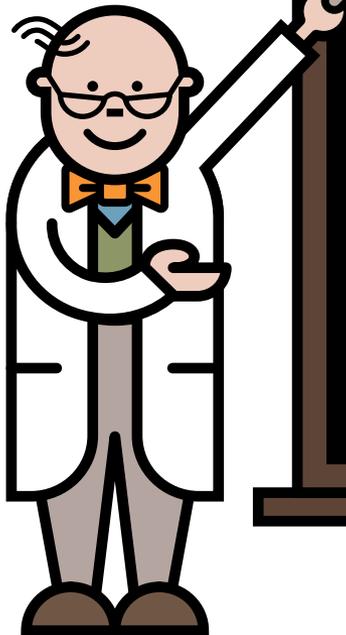


How is HCU managed?

In some people with HCU, the enzyme does not work without the help of a vitamin called vitamin B6 (together with folic acid).



The vitamin helps the enzyme work better and if successful, is the only management needed in about 10% of patients.

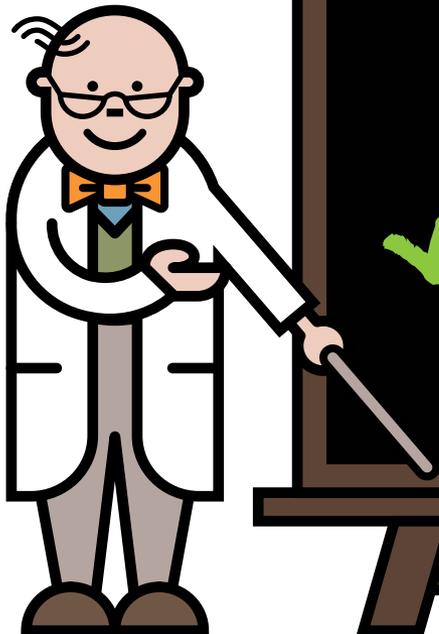


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How is HCU managed?

HCU is managed with the following special diet:

Limited high protein foods



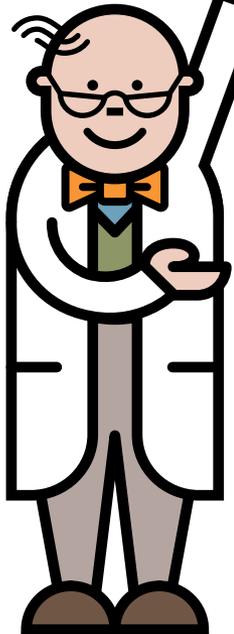
Measured amounts of methionine (protein) containing foods



A protein substitute. Sometimes extra cystine is needed



Low protein foods



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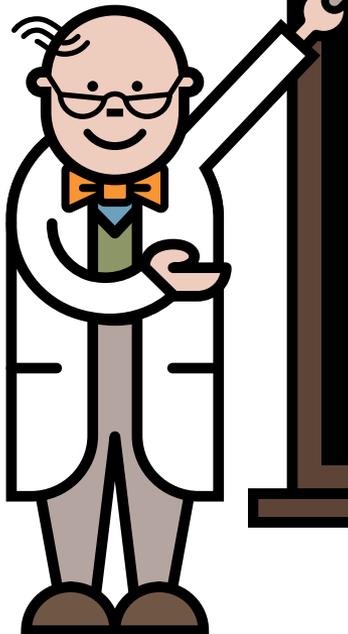
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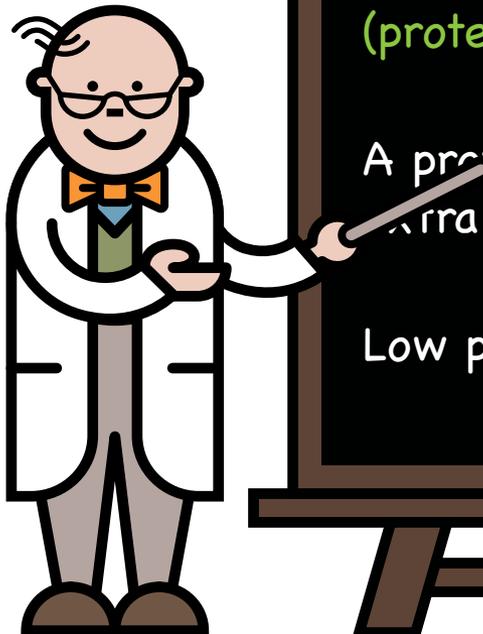
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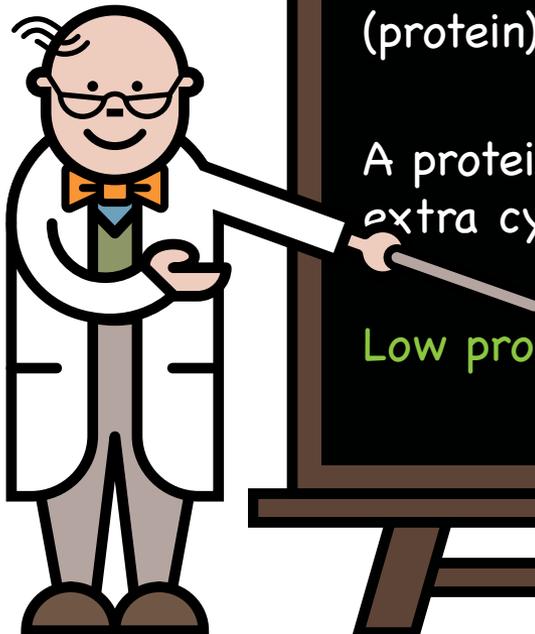
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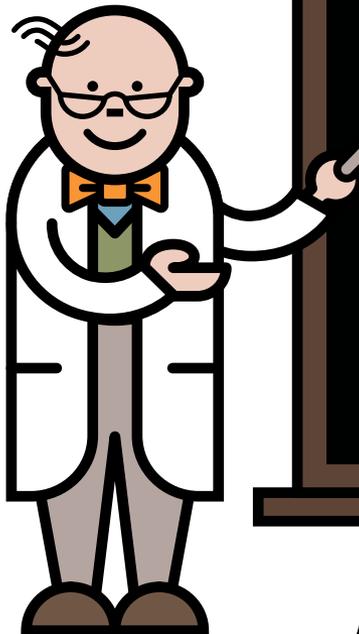
How is HCU managed?

Other forms of management may include:

✓ A drug called betaine. Betaine can help lower the amount of homocysteine in the blood



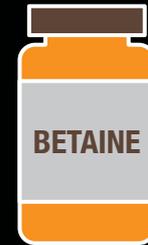
✓ Folic acid and vitamin B12 supplements



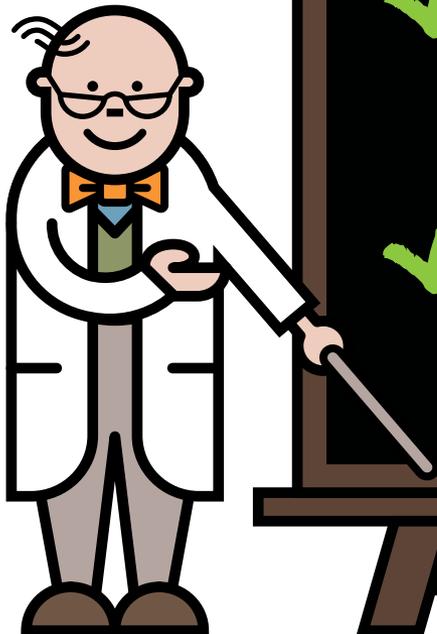
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High protein foods

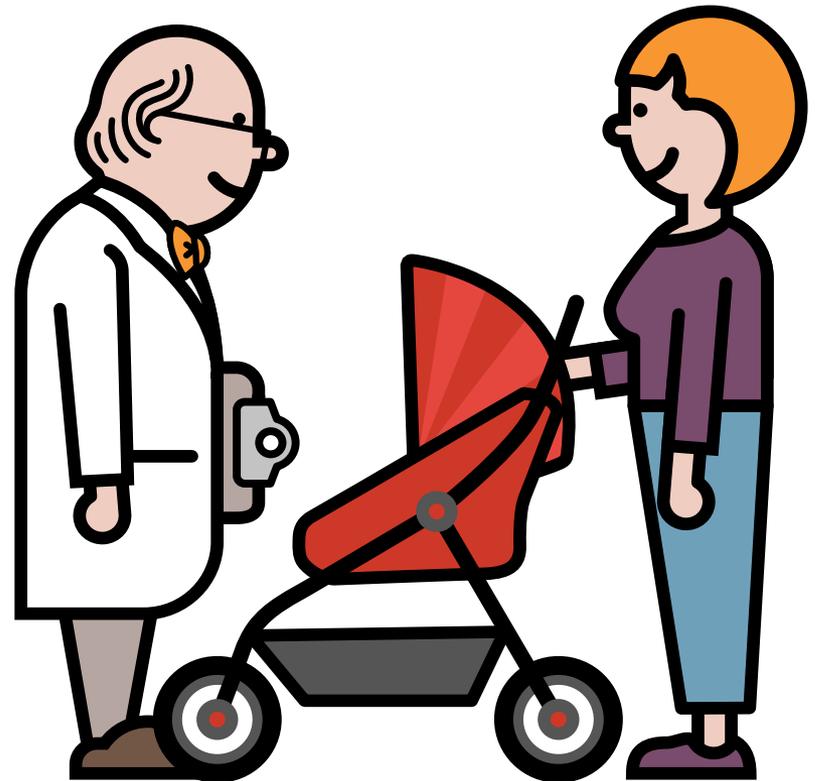
These foods are high in methionine (protein) and must be avoided: meat, fish, eggs, cheese, milk, bread, pasta, nuts, seeds, soya, Quorn and tofu.



Measured leucine intake

In babies, a restricted amount of methionine (protein) is given from breast milk or measured amounts of infant formula.

The amount given will be monitored regularly by your specialist metabolic dietitian.

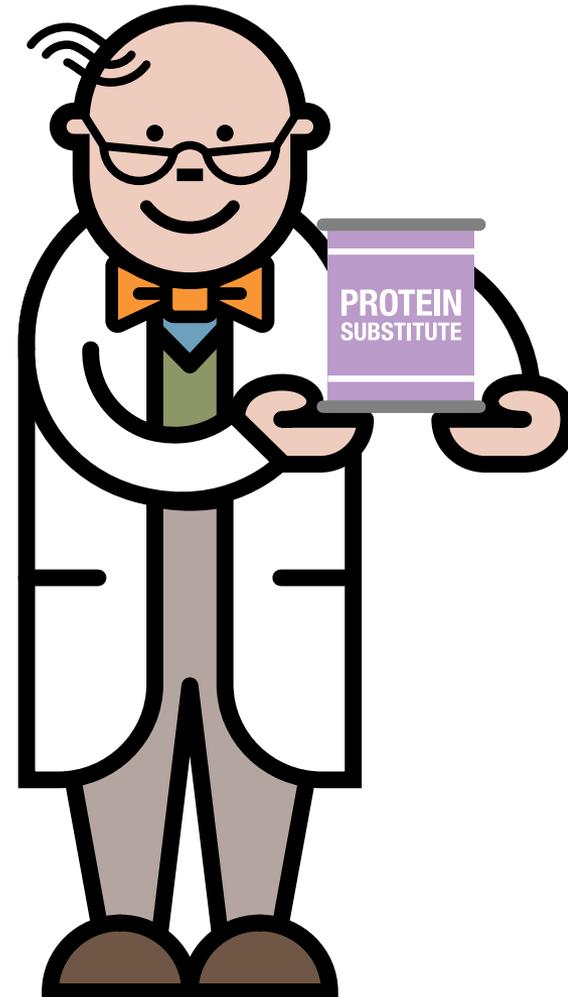


Protein substitute

Protein substitute is essential for metabolic control.

It will help to meet your baby's protein, energy, vitamin and mineral requirements.

It is available on prescription.

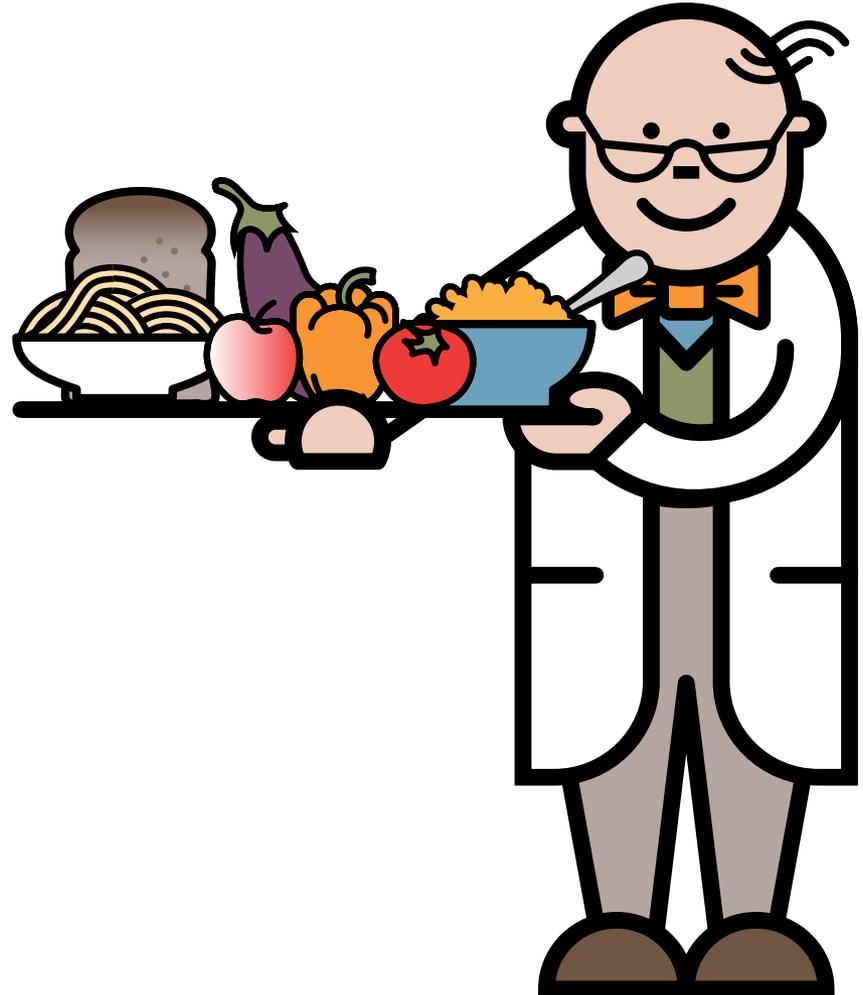


Low protein foods

There are many low protein foods. These include fruit, many vegetables and prescribable low protein foods such as bread and pasta.

They provide:

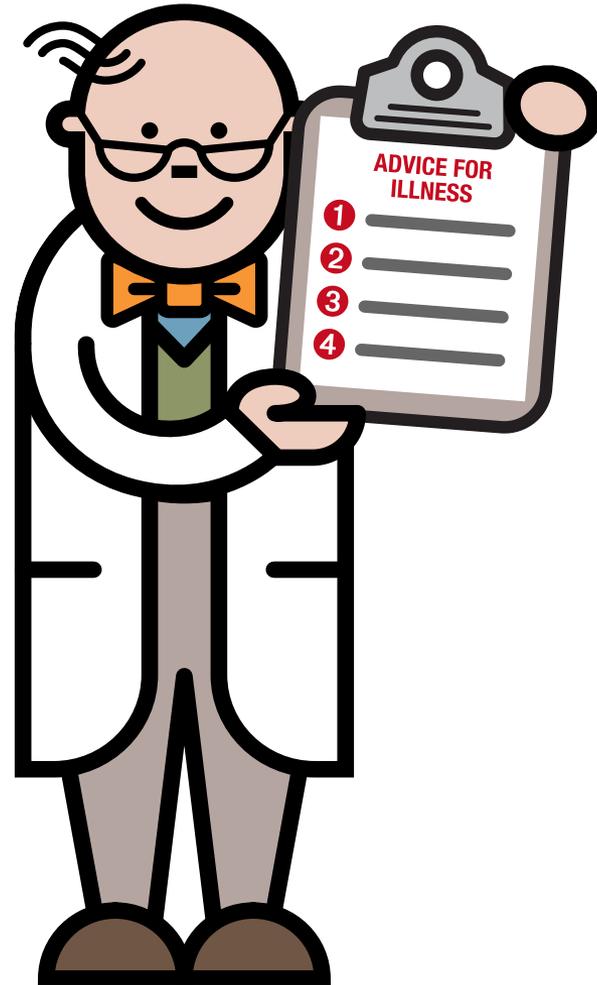
- a source of energy
- variety in the diet



How is HCU managed during illness?

During any childhood illness, catabolism or protein breakdown occurs, causing blood homocysteine levels to increase.

It is important to continue with the usual diet as much as possible.



How is HCU monitored?



Frequent blood tests to check homocysteine, methionine and cystine

Height and weight

Developmental checks

Diet and medications are adjusted according to age, weight and blood chemical levels



How is HCU monitored?

Frequent blood tests to check homocysteine, methionine and cystine



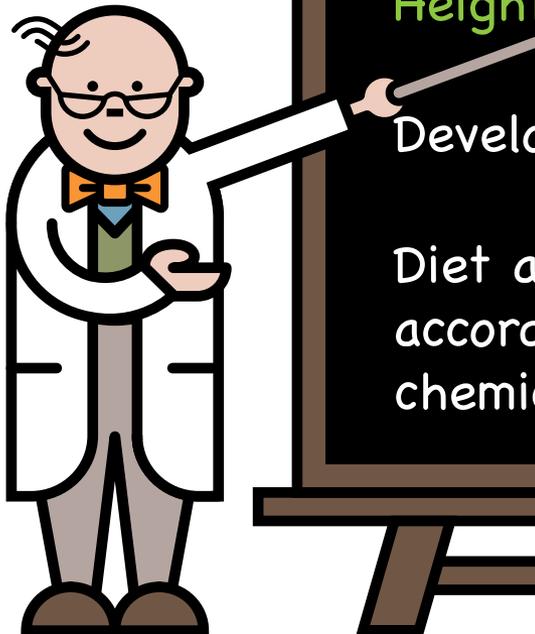
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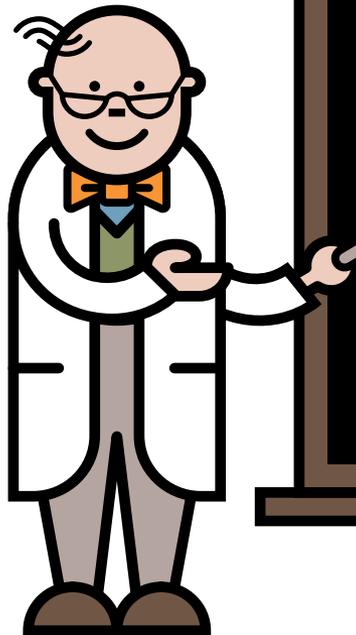
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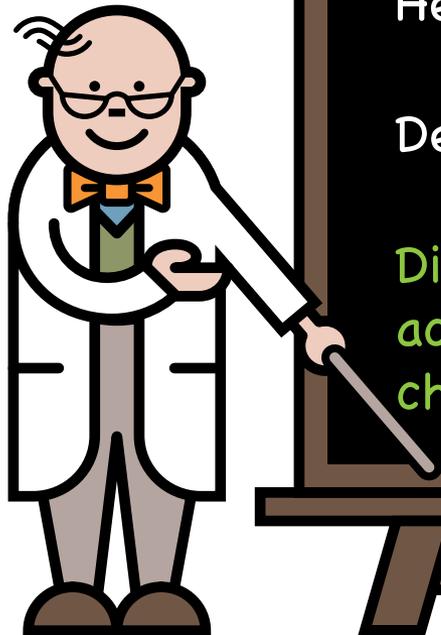
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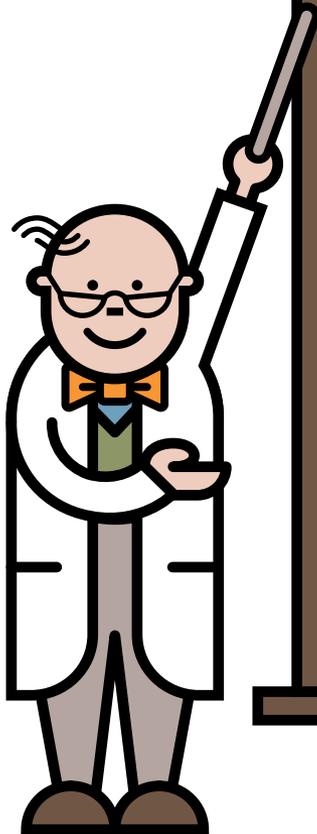
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Chromosomes, genes, mutations



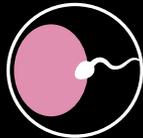
Humans have chromosomes composed of DNA



Genes are pieces of DNA that carry the genetic instruction. Each chromosome may have several thousand genes



The word mutation means a change or error in the genetic instruction

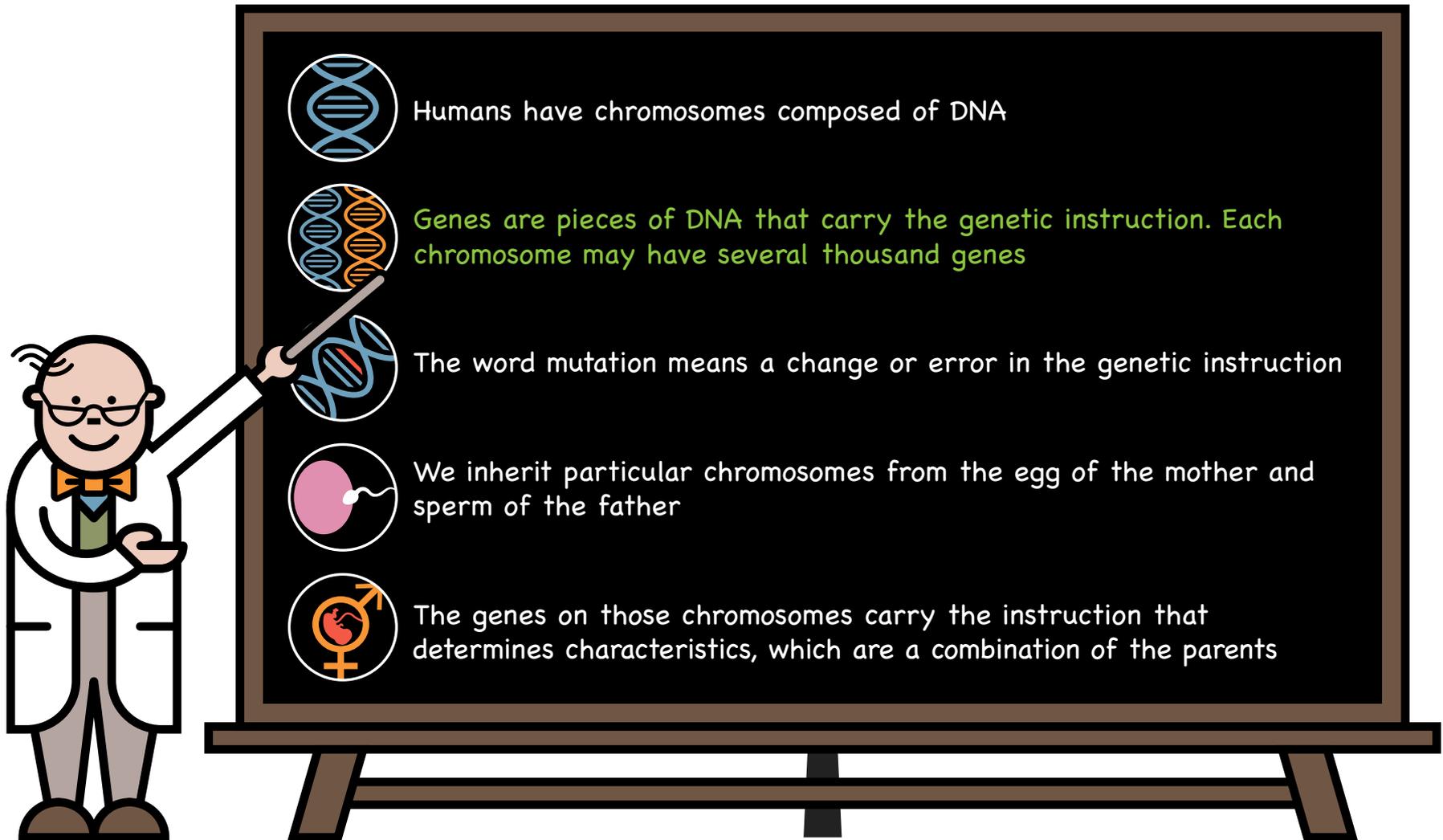


We inherit particular chromosomes from the egg of the mother and sperm of the father



The genes on those chromosomes carry the instruction that determines characteristics, which are a combination of the parents

Chromosomes, genes, mutations



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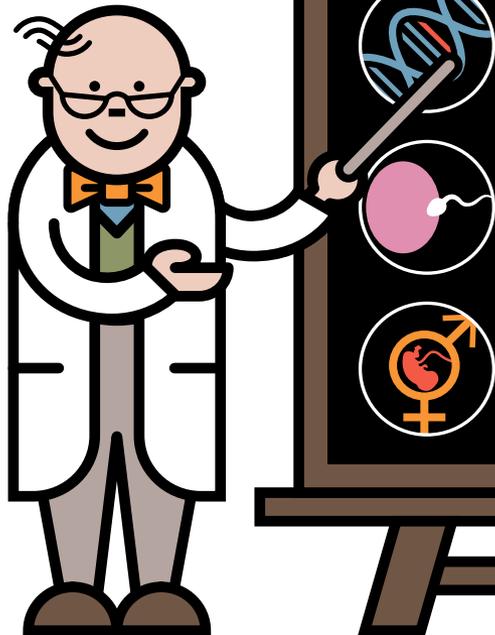
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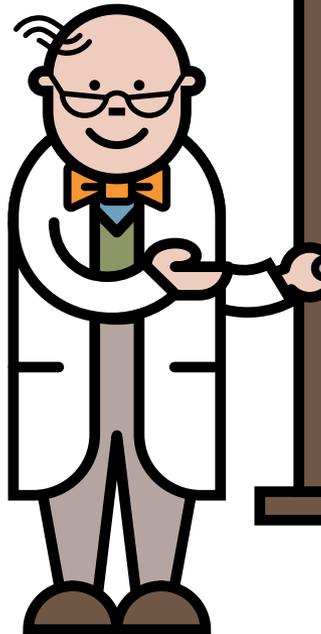


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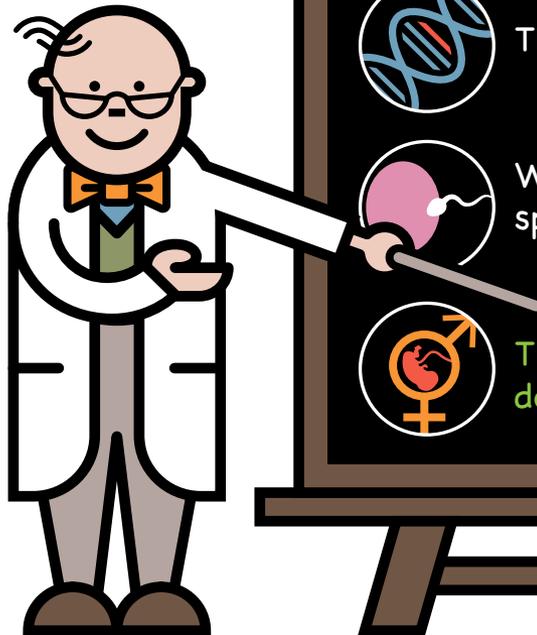


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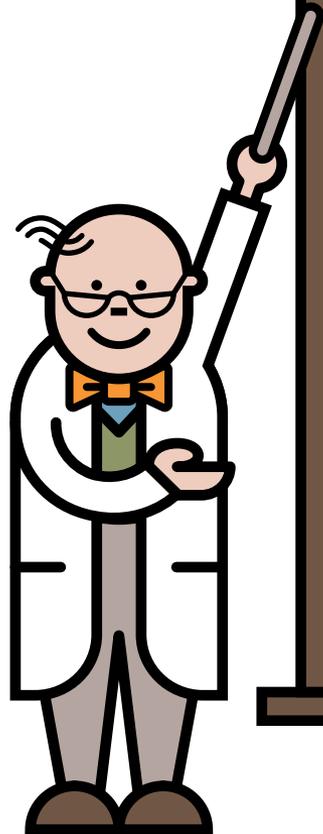


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Inheritance



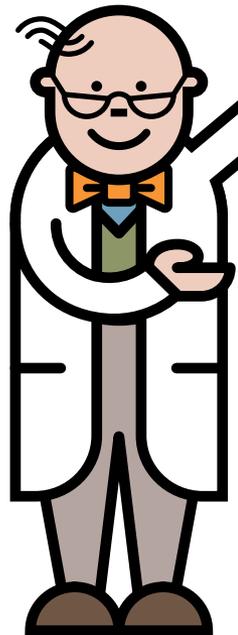
HCU is an inherited condition. There is nothing that could have been done to prevent your baby from having HCU

Everyone has a pair of genes that make the cystathionine beta-synthase enzyme. In children with HCU neither of these genes work correctly. These children inherit one non-working HCU gene from each parent

Parents of children with HCU are carriers of the condition

Carriers do not have HCU because the other gene of this pair is working correctly

Inheritance



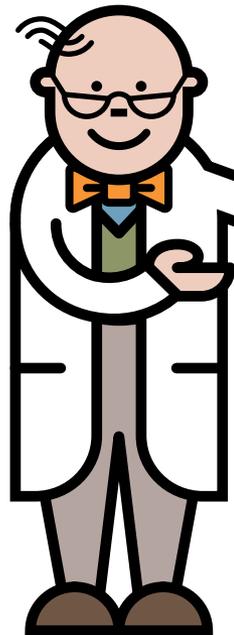
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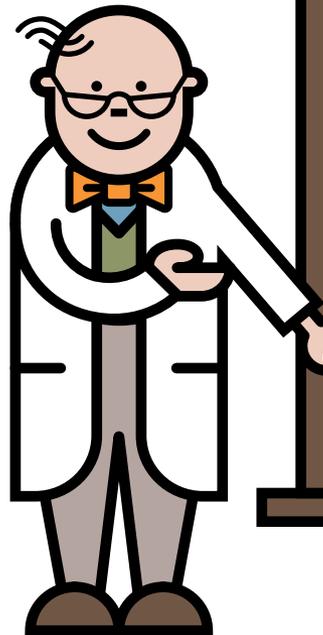
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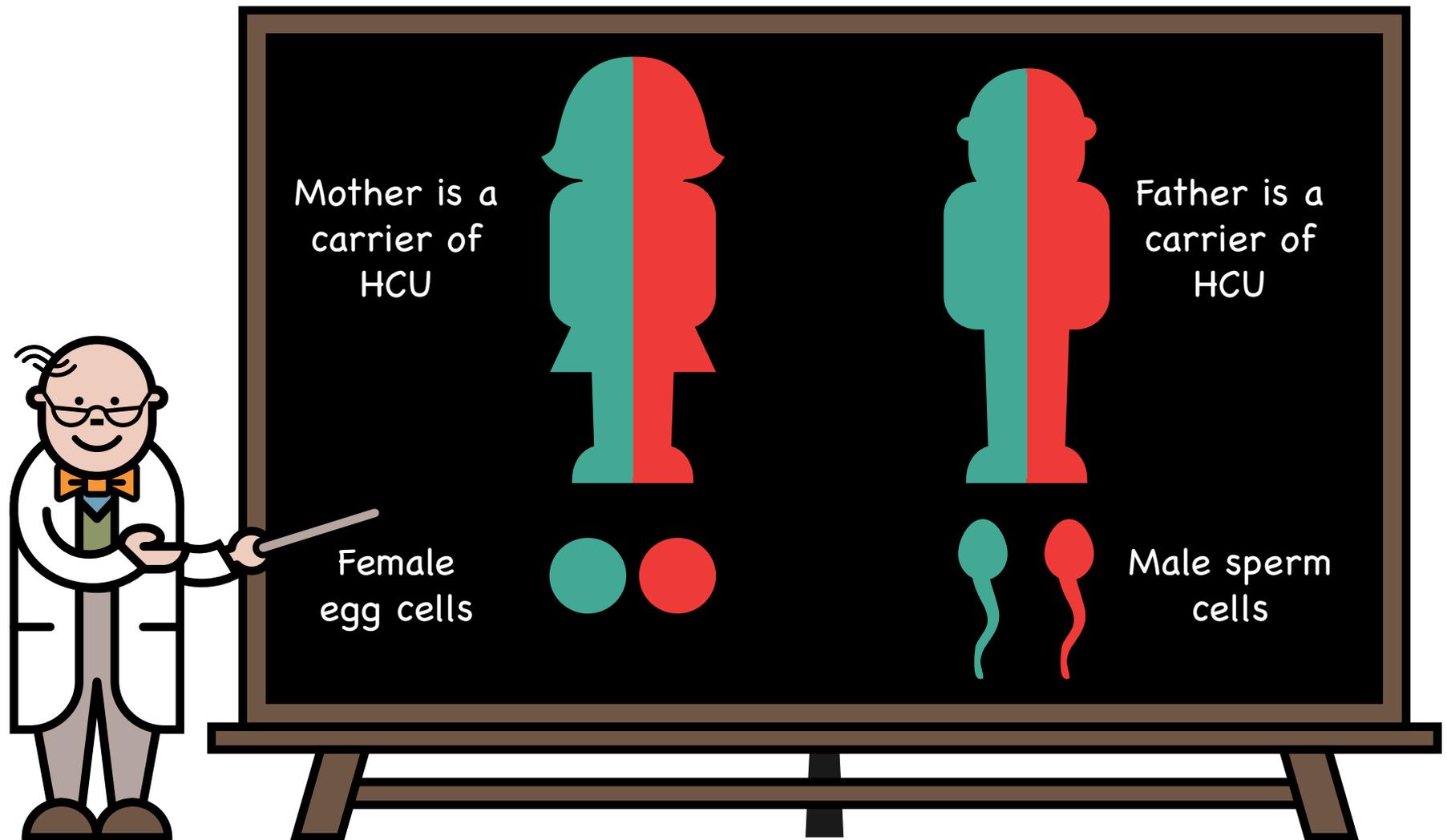
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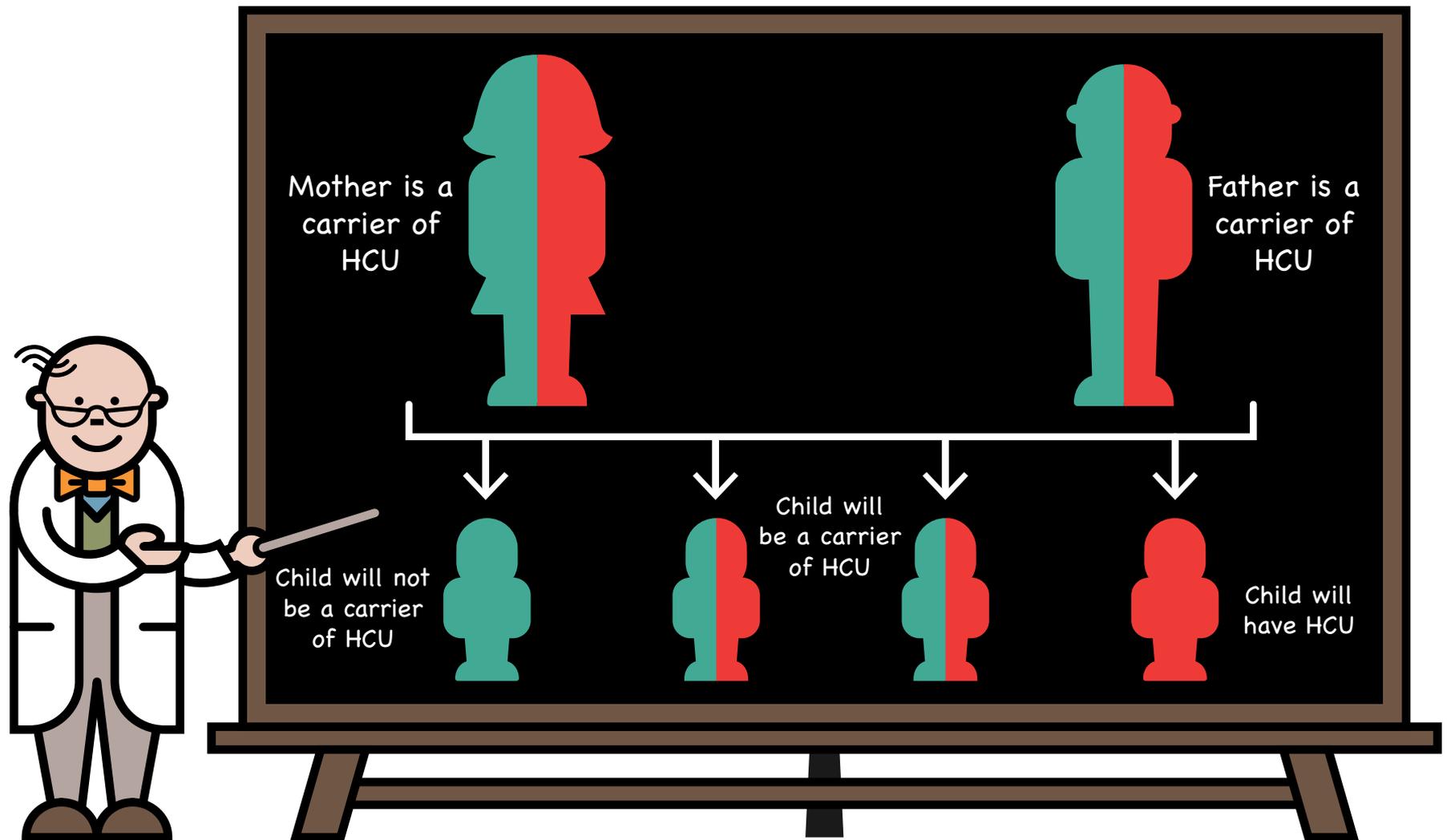
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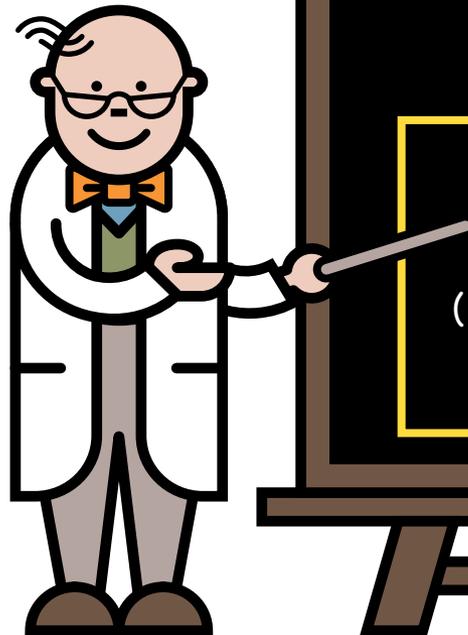
Inheritance – Autosomal recessive (carriers of HCU)



Inheritance – Autosomal recessive – possible combinations



Future pregnancies



When both parents are carriers, in each pregnancy the risk to the baby is as follows:

25% chance
(1 in 4) of HCU

50% chance
(1 in 2) for the baby to be a carrier of HCU

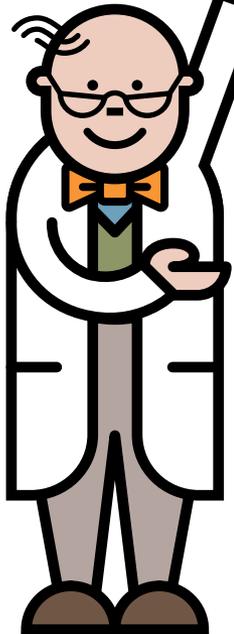
25% chance (1 in 4) for the baby to have two working genes and neither have HCU or be a carrier

Take home messages

✓ HCU is a serious inherited metabolic disorder

✓ Damage can be prevented with a protein restricted diet and a protein substitute

✓ With early management normal development is achievable. This should also prevent long term complications such as osteoporosis (thin bones), blood clots and strokes

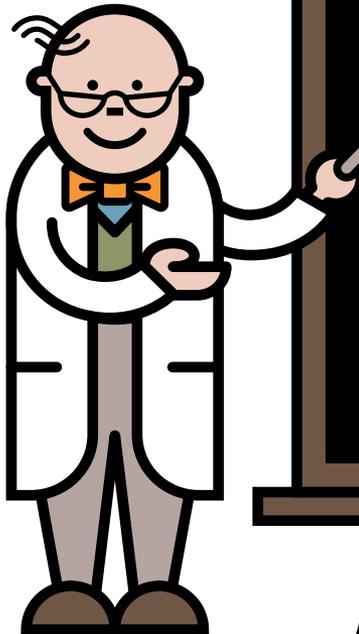


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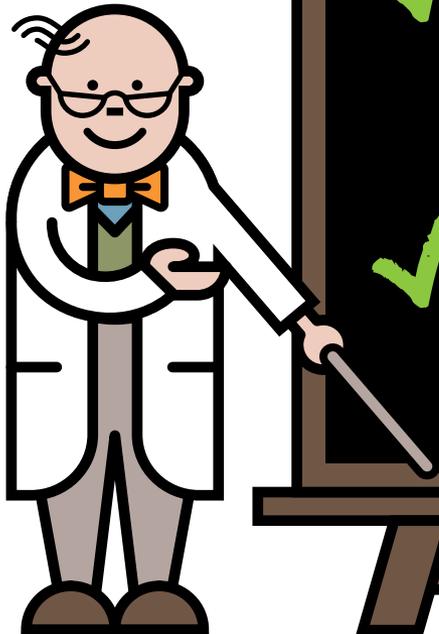


Take home messages

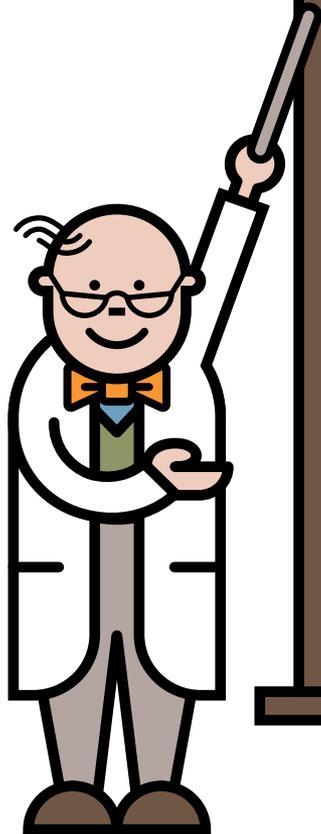
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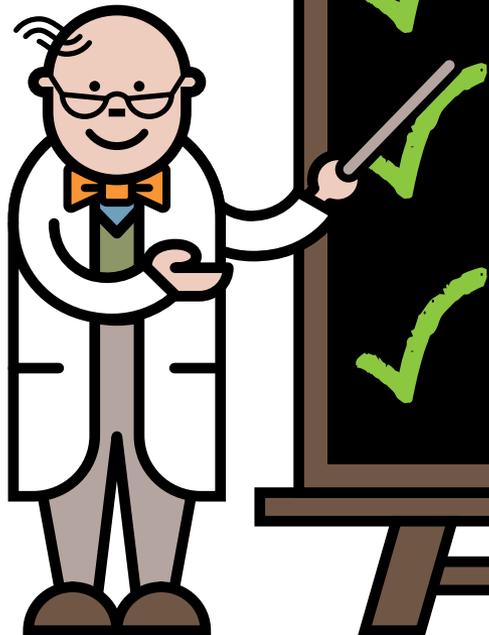


Helpful hints



- ✓ Always ensure you have a good supply of your dietary products and medicines and that they are in date
- ✓ Your dietary products are prescribed by your GP. These are obtained via a pharmacy or home delivery
- ✓ Always ensure you have sufficient blood testing equipment and send samples on a regular basis
- ✓ Medications to control fever should be given as normally recommended – always keep supplies available

Helpful hints



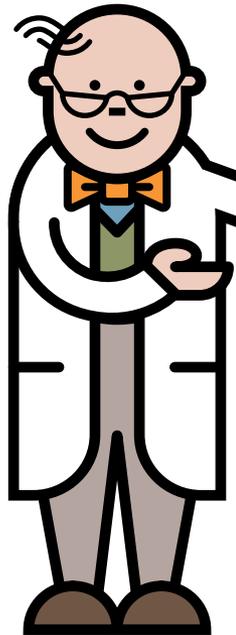
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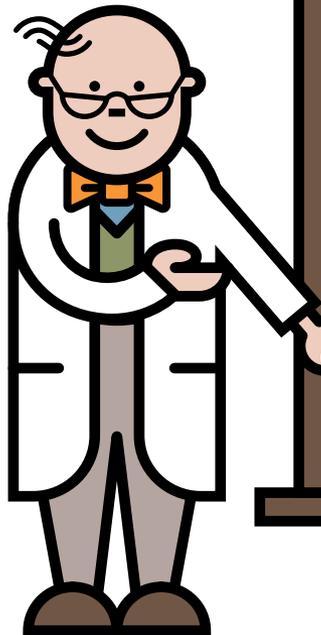
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Who's who

- My dietitians
- My nurses
- My doctors
 - Contact details, address, photos

Visit www.lowproteinconnect.com and register to get access to support and practical advice for those living on a low protein diet.

The site also provides information on upcoming events and personal stories from others on a low protein diet.



BIMDg

British Inherited Metabolic Diseases Group



NUTRICIA
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**METABOLIC
SUPPORT UK**

Your rare condition.
Our common fight.

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