

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 3, APRIL 2020

PKU

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).

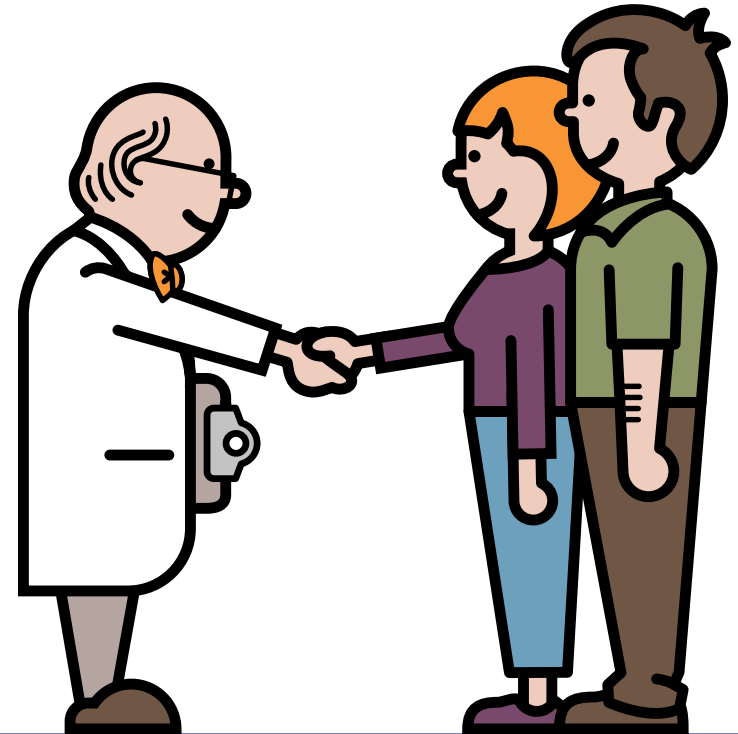
The team are Rachel Skeath, Karen van Wyk, Pat Portnoi and Anita MacDonald. The group is facilitated by Heidi Chan from Nutricia.

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.

PKU

Information for families following
a new diagnosis



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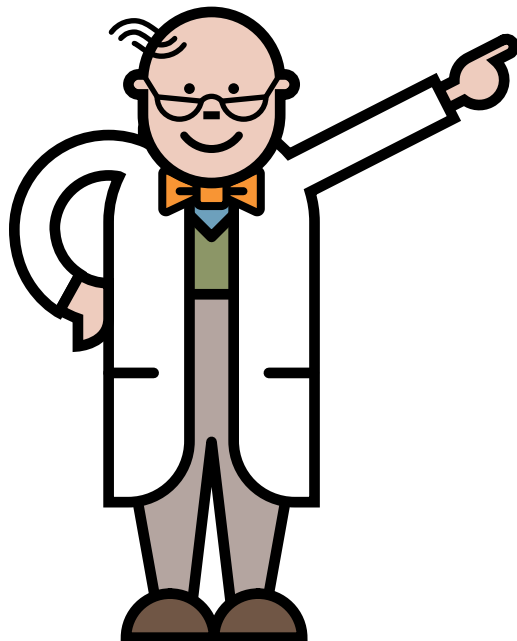
Tools Enabling Metabolic Parents LEarning

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as a service to metabolic medicine

What is PKU?

PKU stands for Phenylketonuria

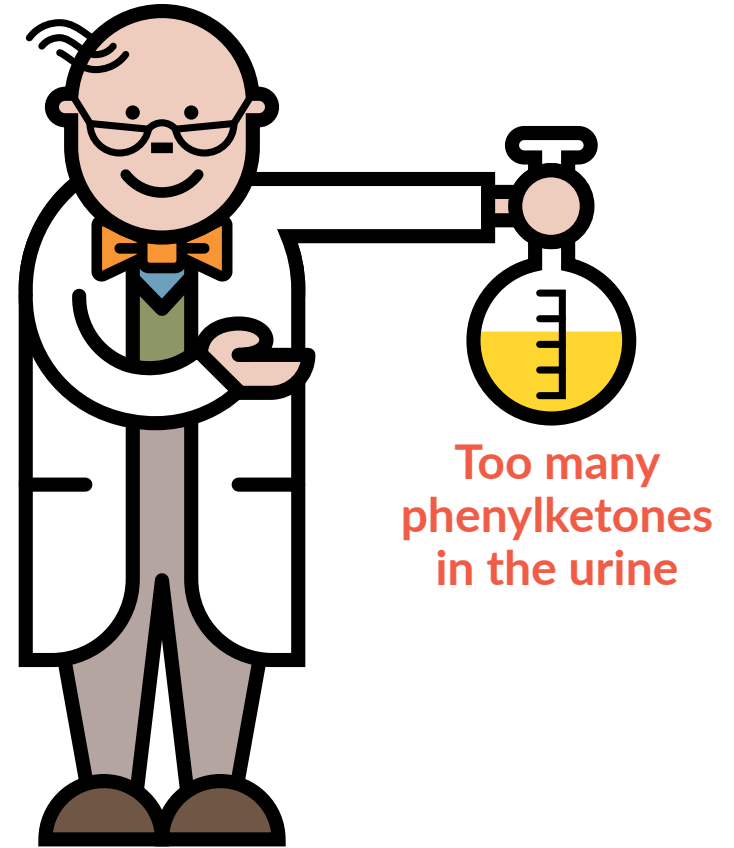
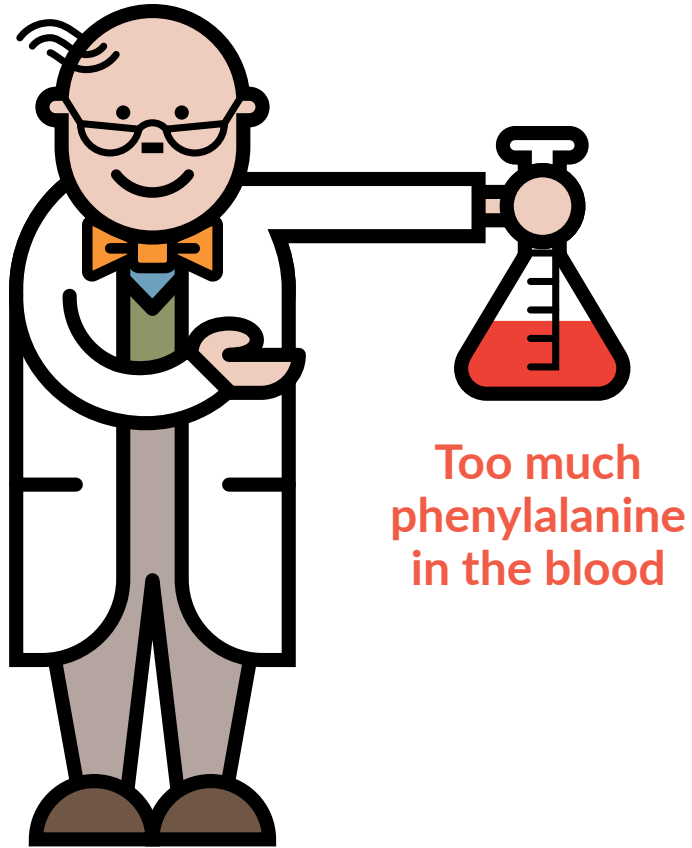
It is an inherited metabolic condition



Phenyl Keton Uria

PKU

What is PKU?

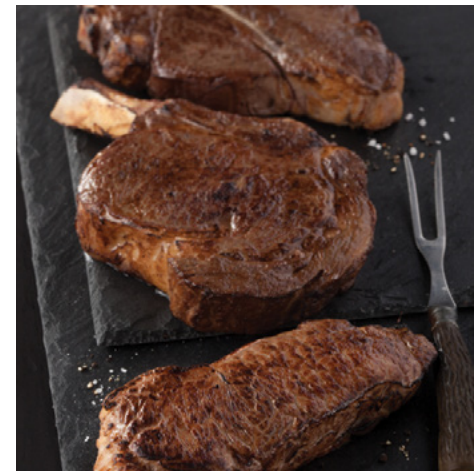
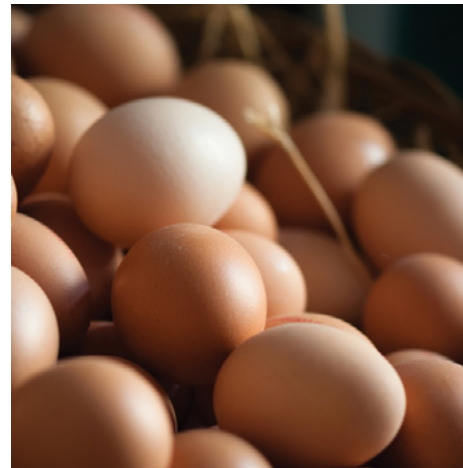


PKU and protein

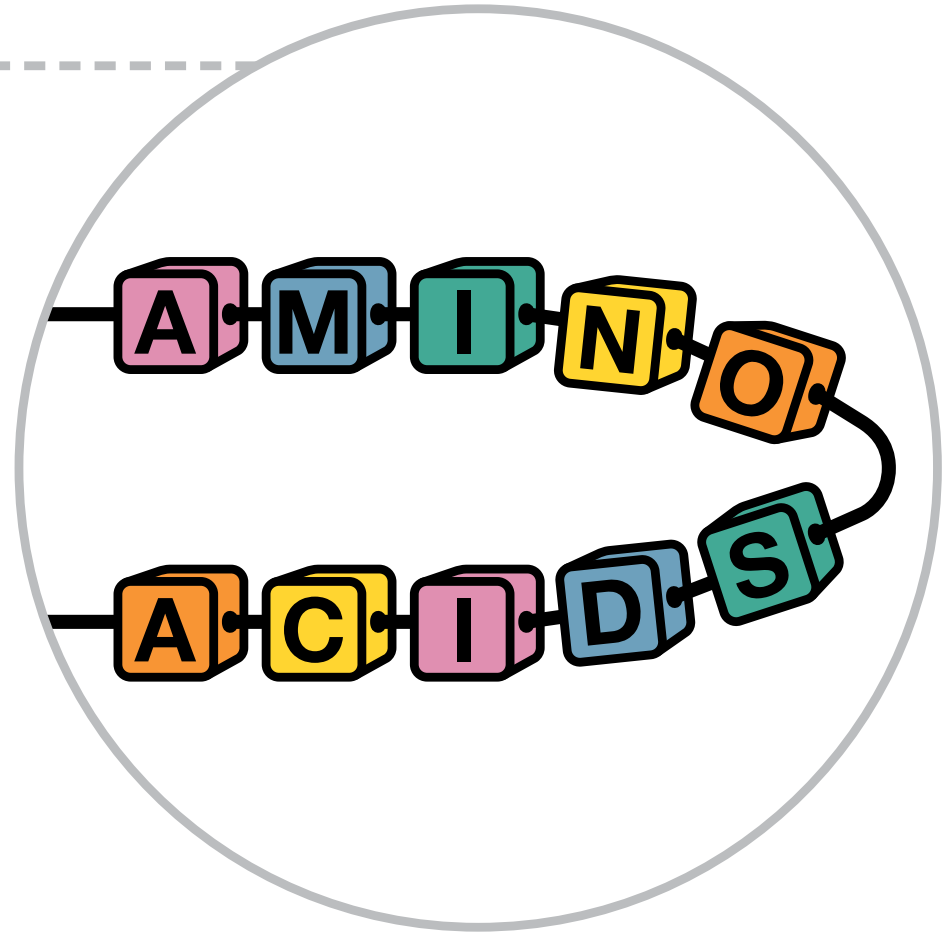
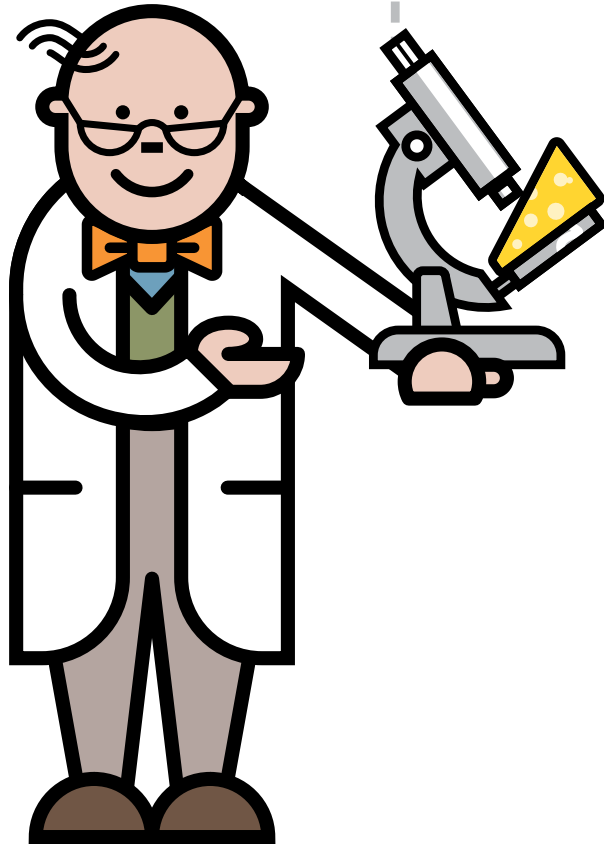
PKU affects the way your baby breaks down protein.

Many foods contain protein.

The body needs protein for growth and repair.



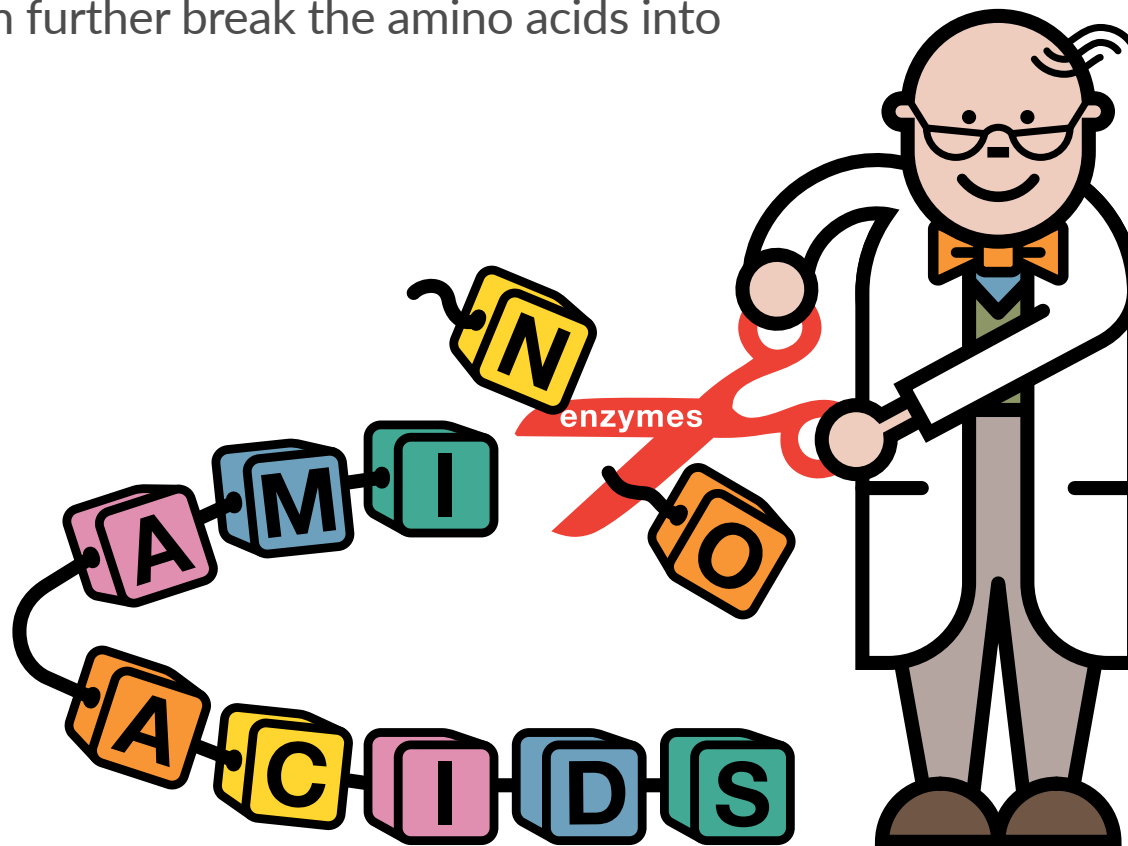
What is protein?



Protein and enzymes

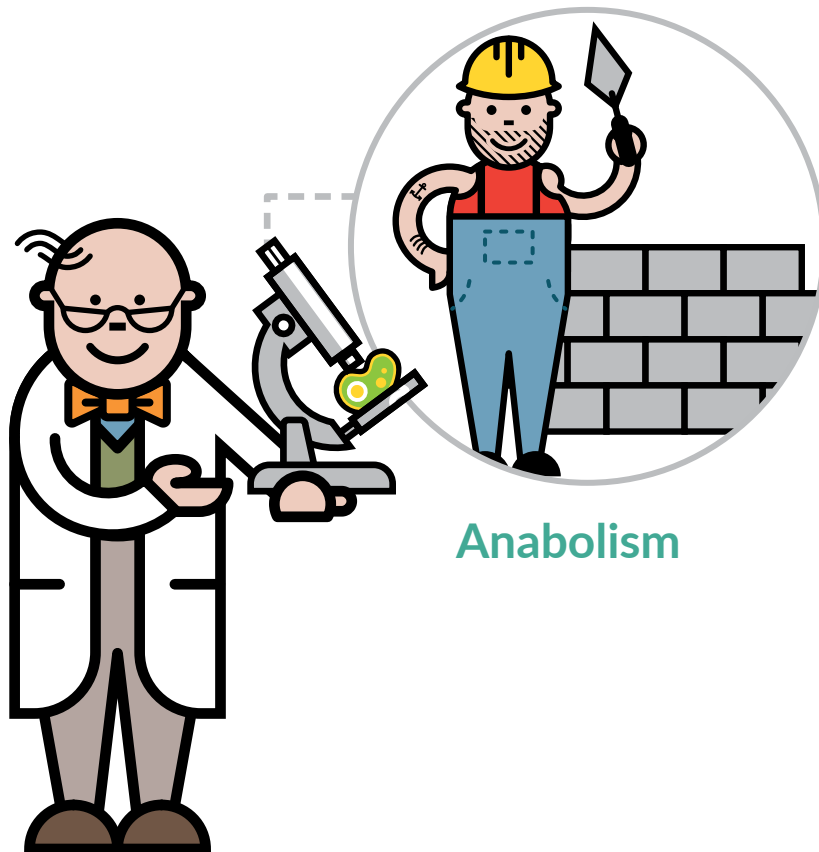
Protein is broken down into amino acids (building blocks of protein) by enzymes (which are like chemical scissors).

Enzymes then further break the amino acids into smaller parts.

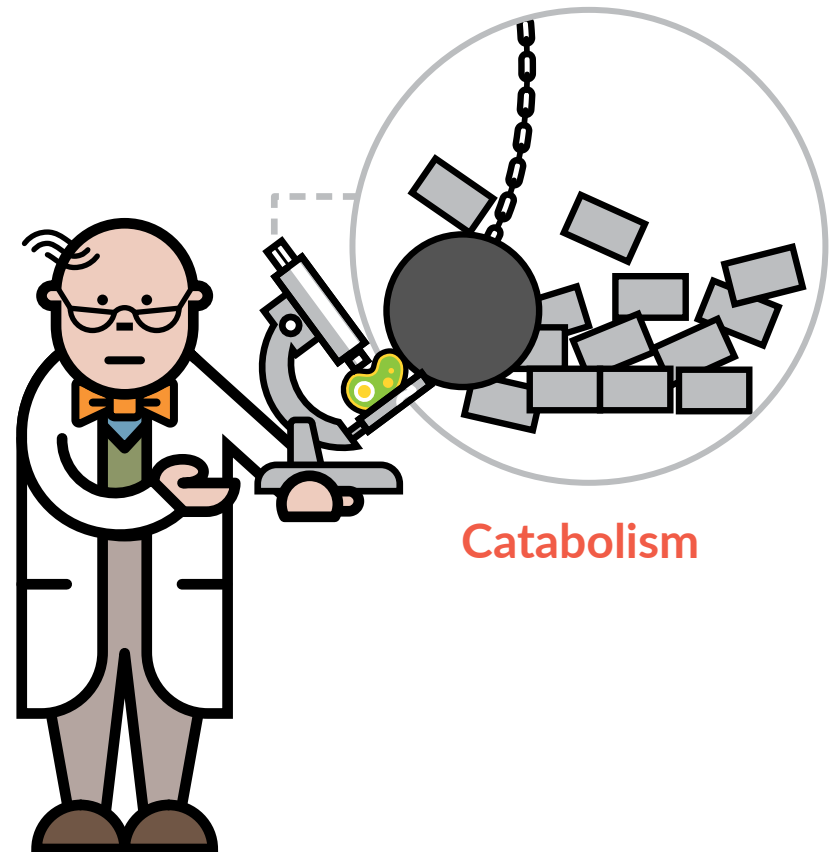


Protein metabolism

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



Anabolism

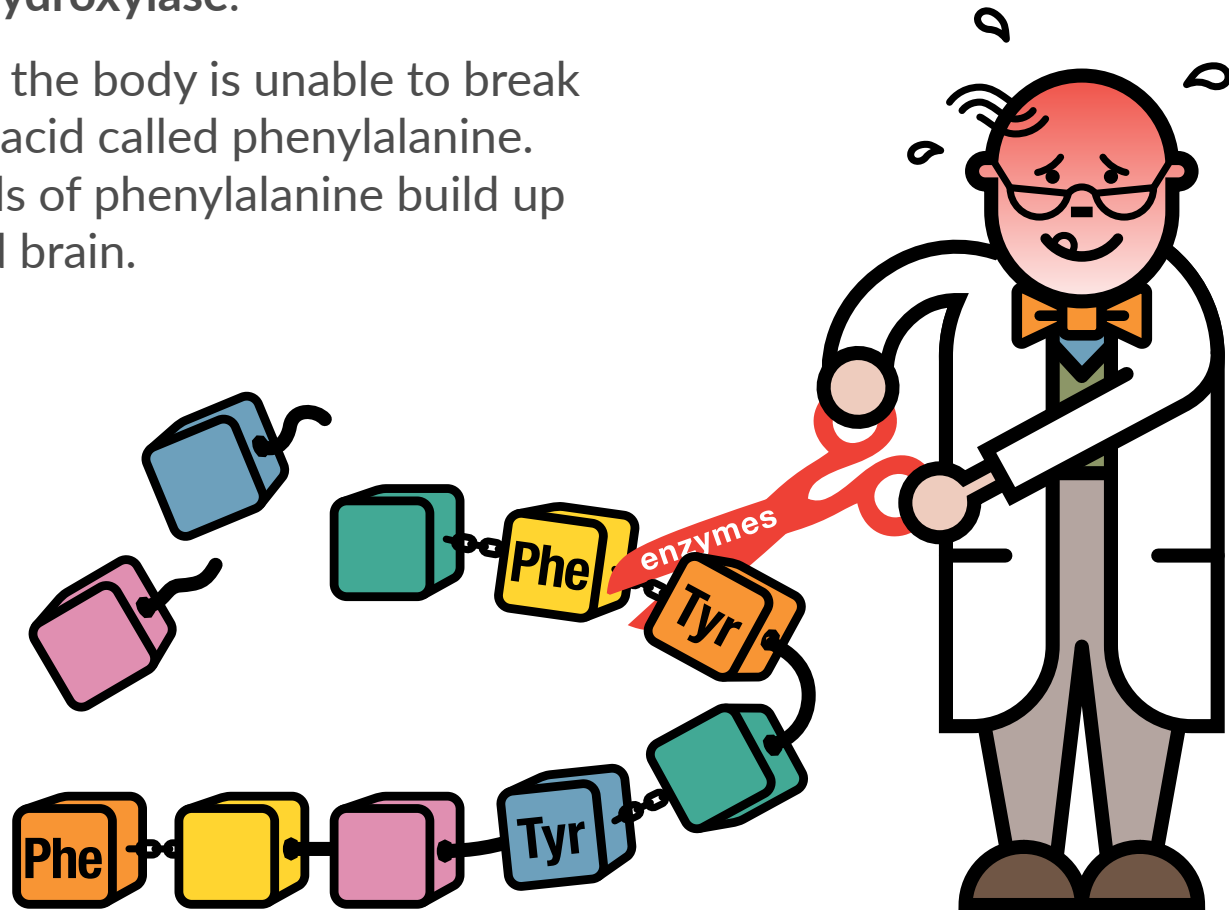


Catabolism

What happens in PKU?

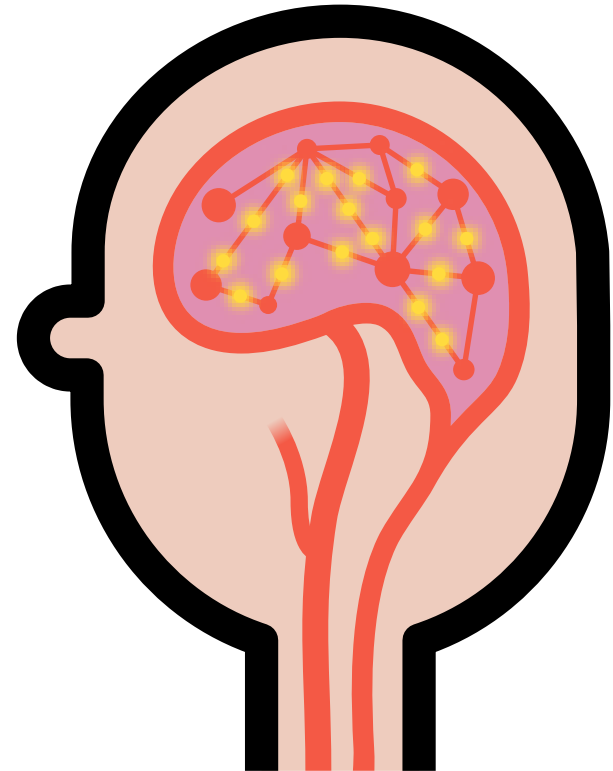
In PKU, the body lacks an enzyme called **phenylalanine hydroxylase**.

This means that the body is unable to break down an amino acid called phenylalanine. As a result, levels of phenylalanine build up in the blood and brain.



What can go wrong in PKU?

The build-up of phenylalanine can lead to brain damage, with learning and behavioural difficulties.



Early management can prevent brain damage and learning difficulties

How is PKU diagnosed?

PKU is diagnosed by newborn screening. High levels of phenylalanine are found in the blood.



How is PKU managed?

PKU is managed with the following special diet:

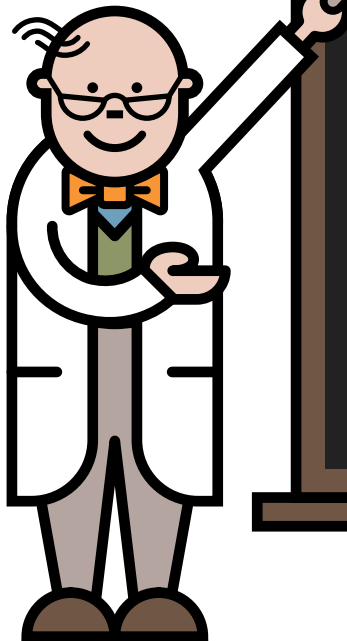
Limited high protein foods

Measured amounts of phenylalanine (protein) containing foods

A protein substitute

Low protein foods

Avoiding aspartame



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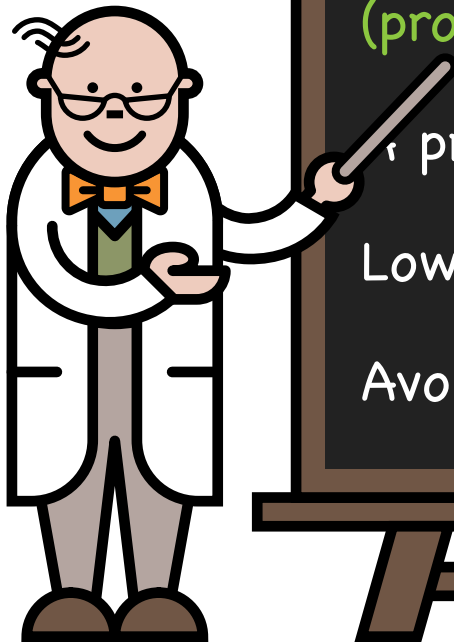
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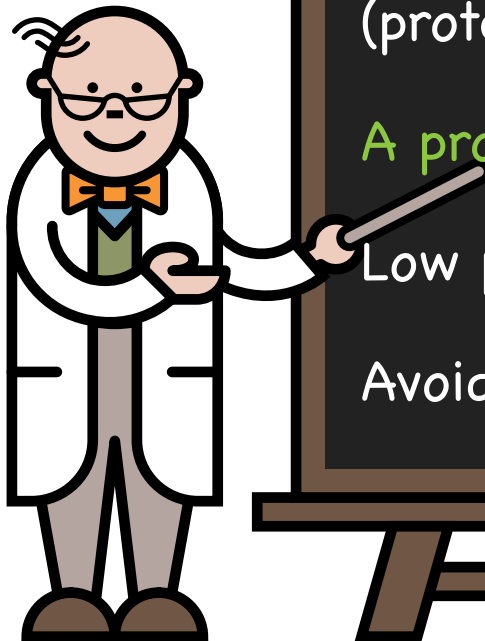
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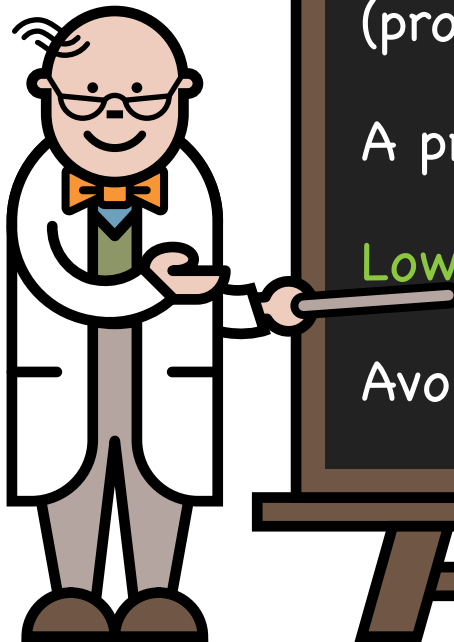
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How is PKU managed?

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Limited high protein foods



Measured amounts of phenylalanine (protein) containing foods

A protein substitute



Avoiding low protein foods



Avoiding aspartame



High protein foods

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

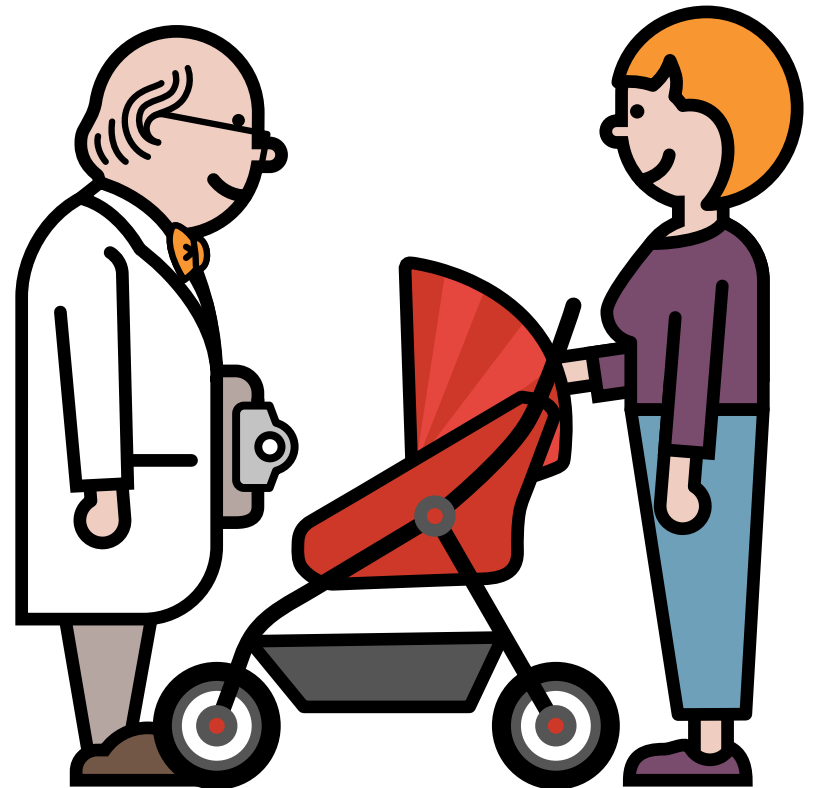
Any food or drink containing **aspartame** should also be avoided.



Measured phenylalanine intake

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

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

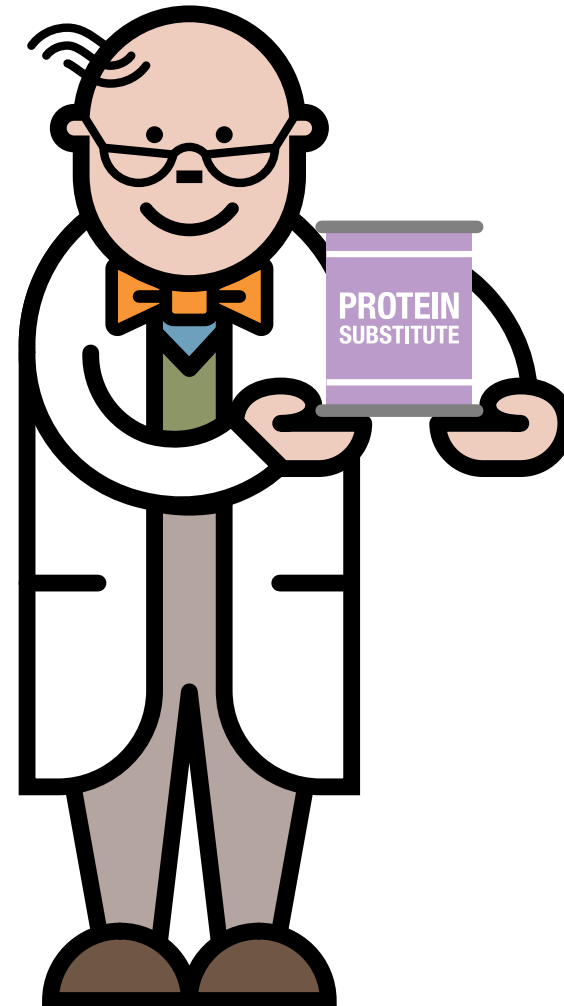


Protein substitute

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.

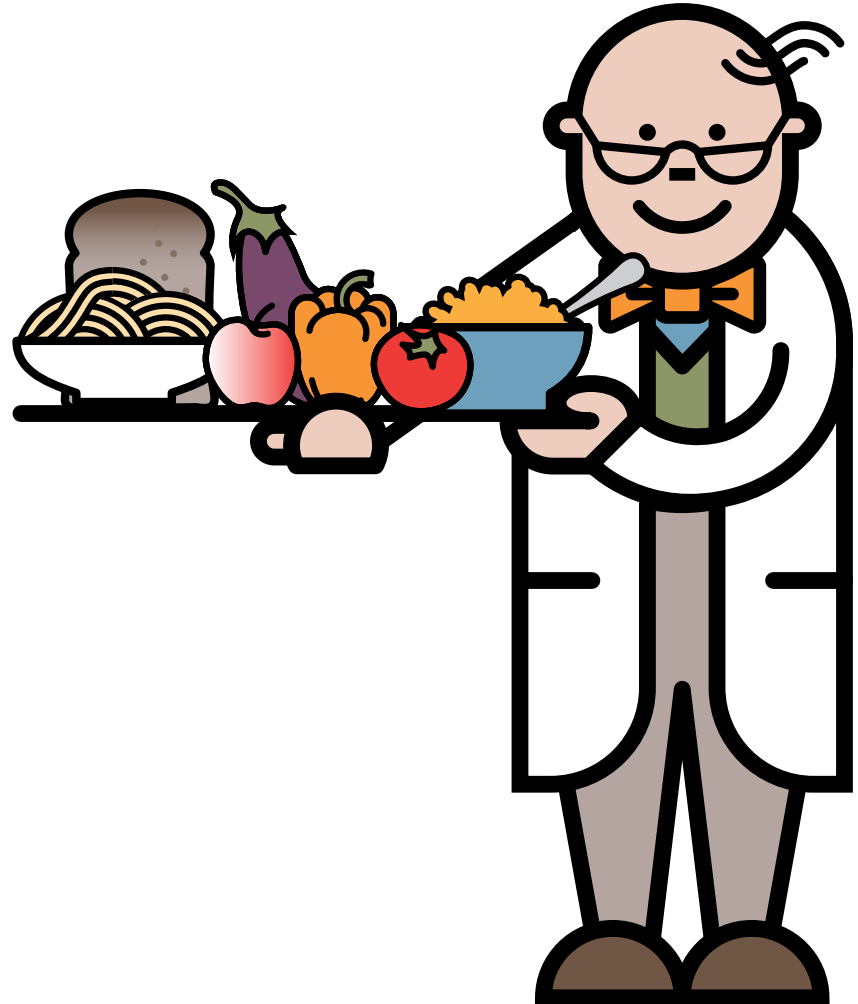


Low protein foods

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



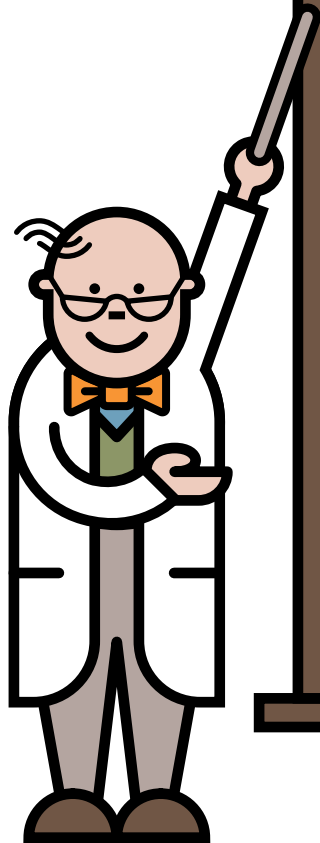
How is PKU managed during illness?

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

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



How is PKU monitored?



Frequent blood tests to check phenylalanine



Height and weight



Developmental checks

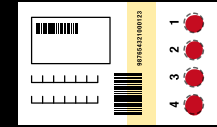


Diet is adjusted according to age, weight and phenylalanine levels

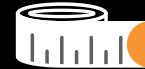


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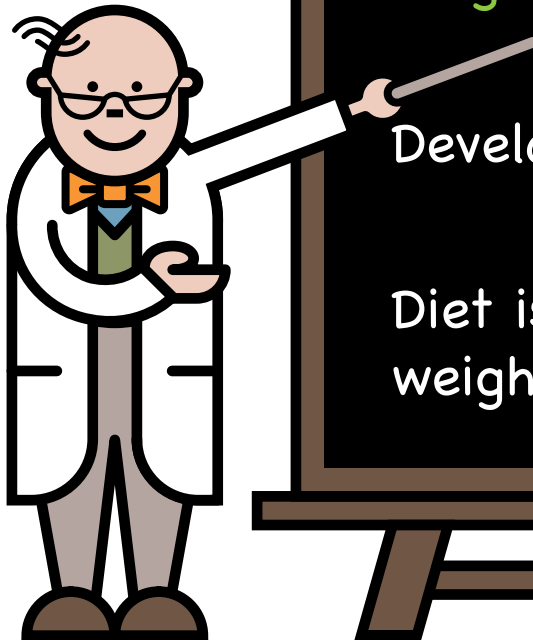
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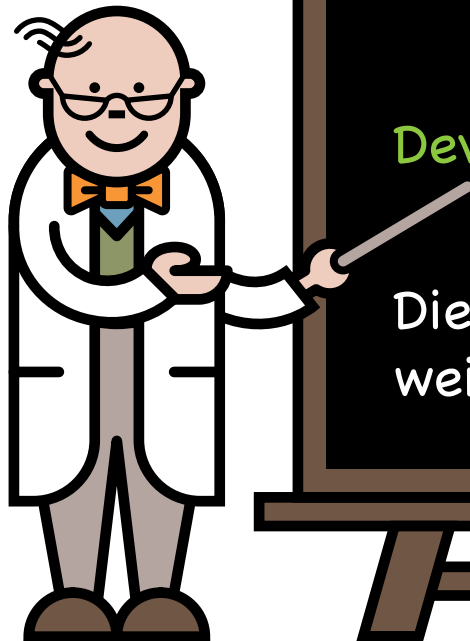
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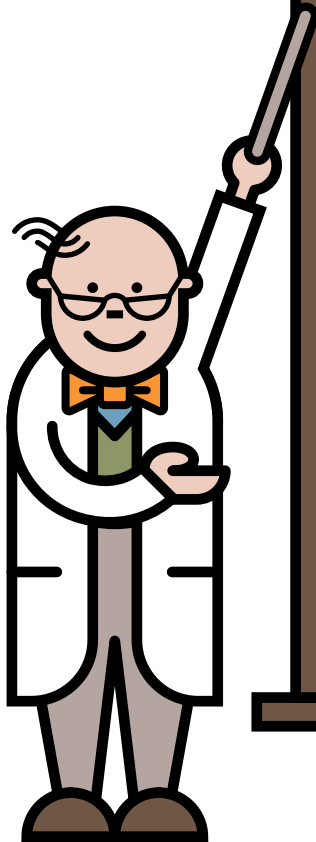
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Chromosomes, genes, mutations



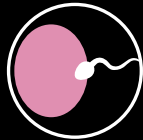
Humans have chromosomes composed of DNA



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

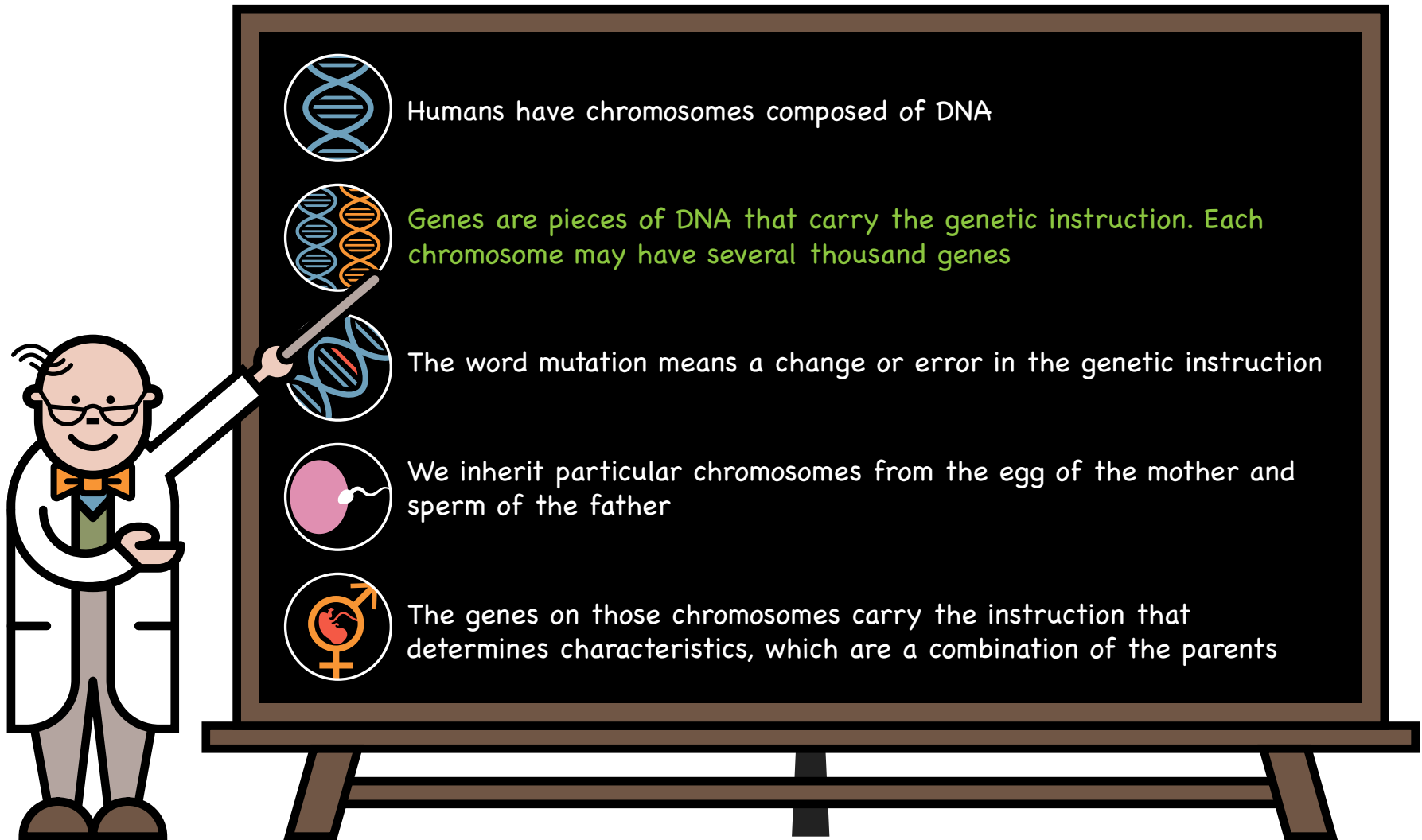



We inherit particular chromosomes from the egg of the mother and sperm of the father





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


Chromosomes, genes, mutations




 Humans have chromosomes composed of DNA

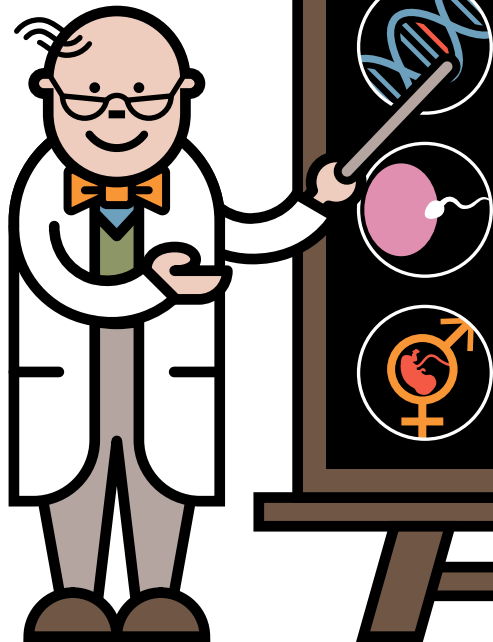
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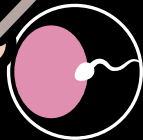
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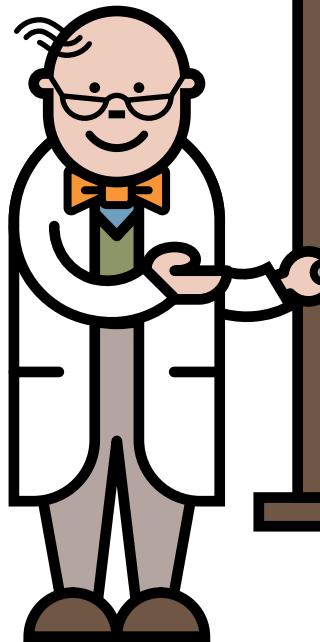


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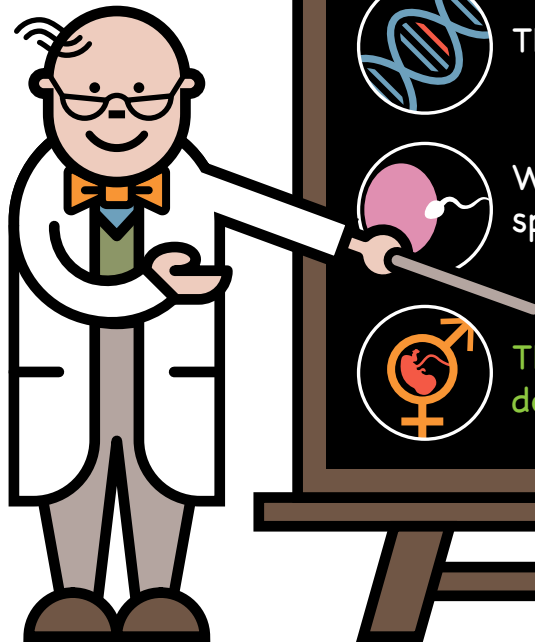


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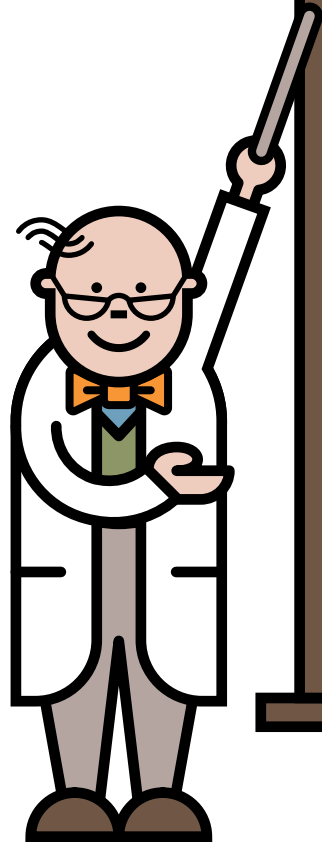


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Inheritance



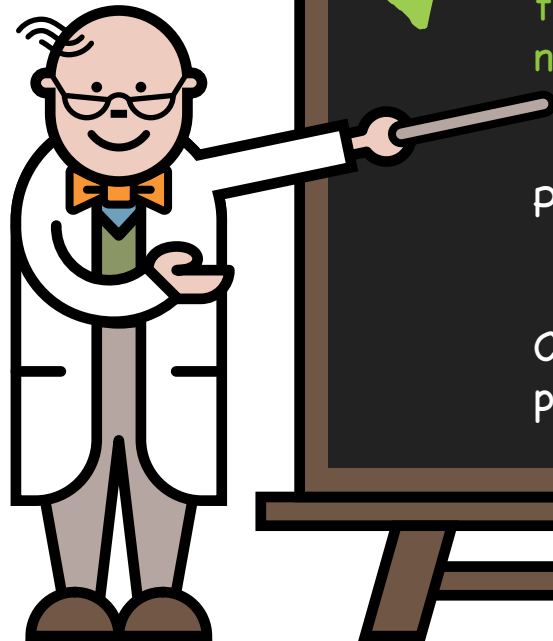
PKU is an inherited condition. There is nothing that could have been done to prevent your baby from having PKU

Everyone has a pair of genes that make the phenylalanine hydroxylase enzyme. In children with PKU, neither of these genes works correctly. These children inherit one non-working PKU gene from each parent

Parents of children with PKU are carriers of the condition

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

Inheritance



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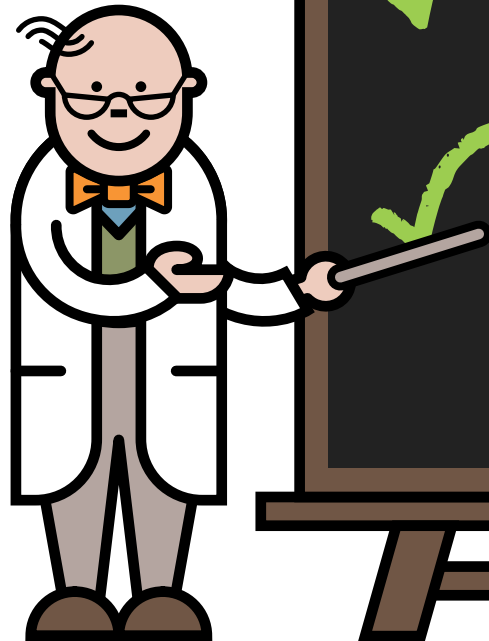


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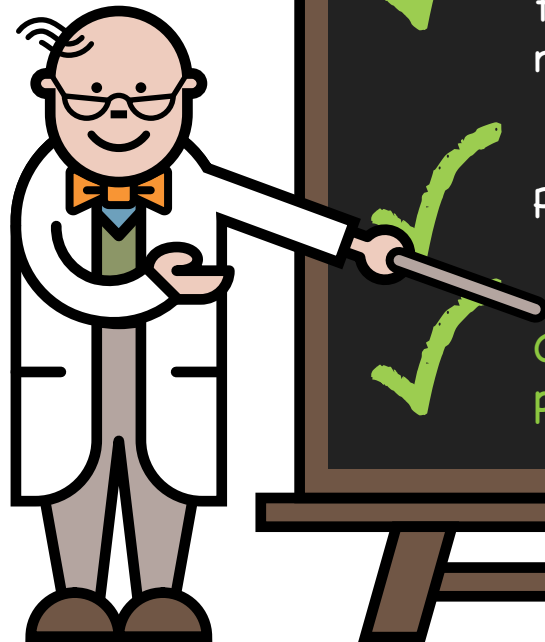
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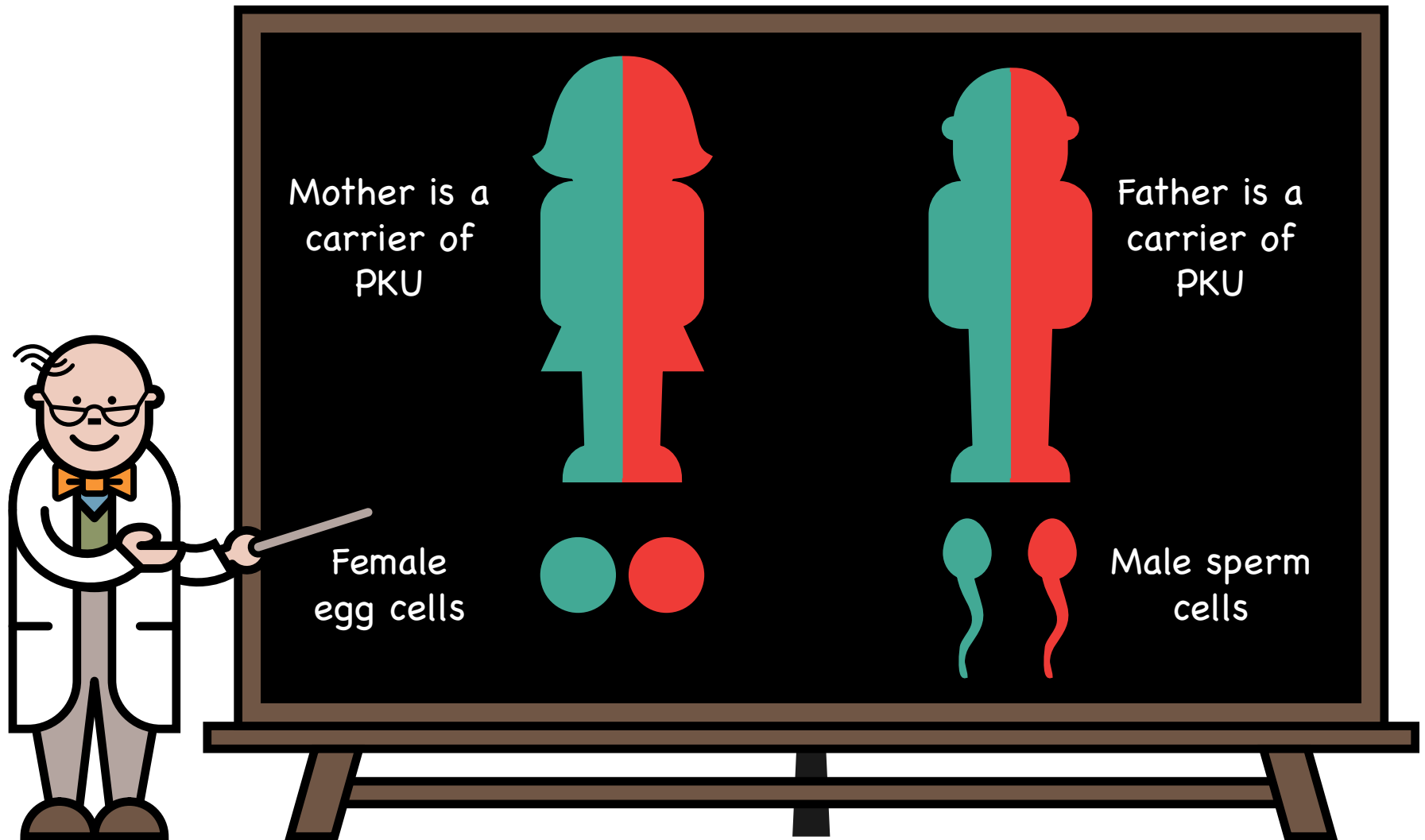


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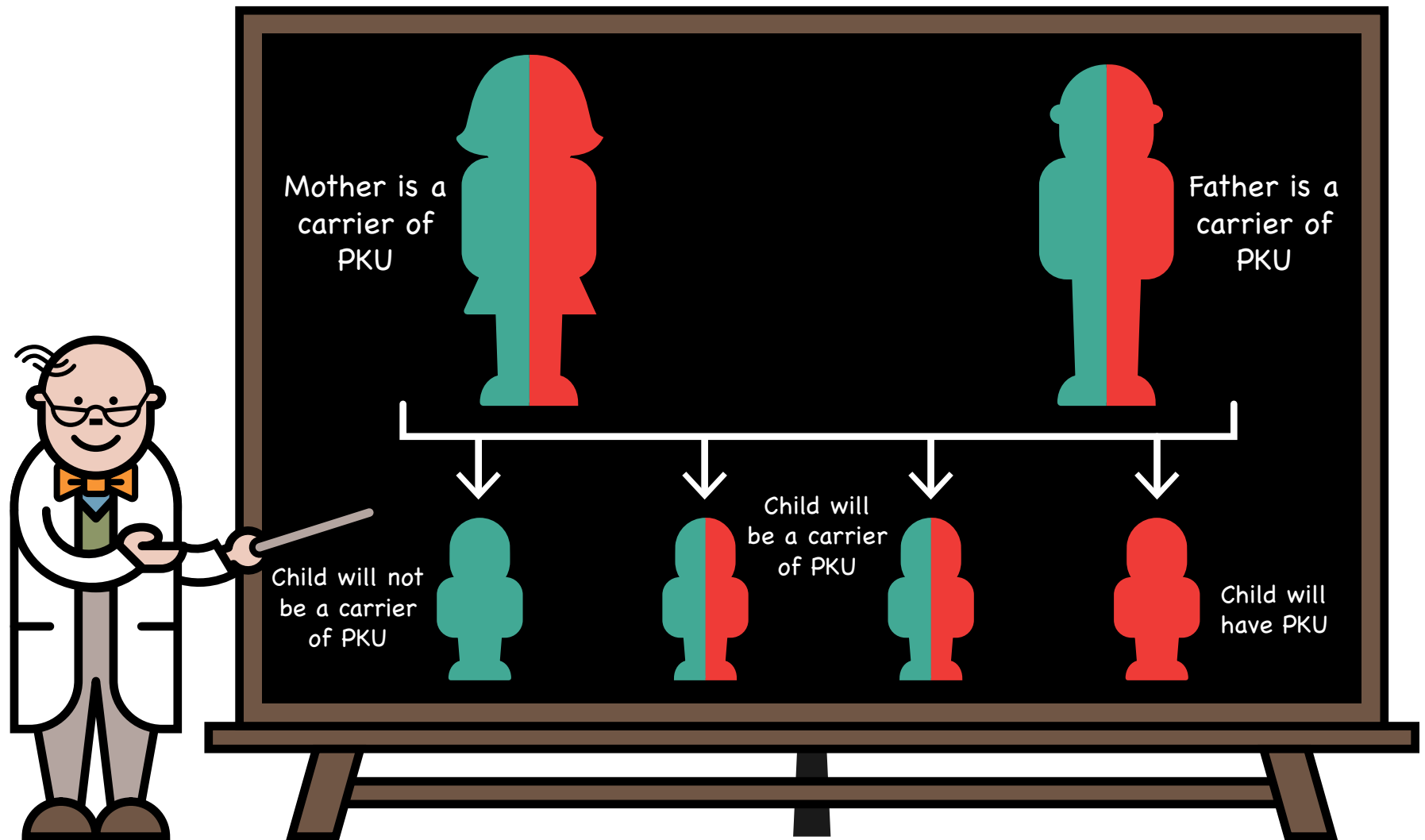


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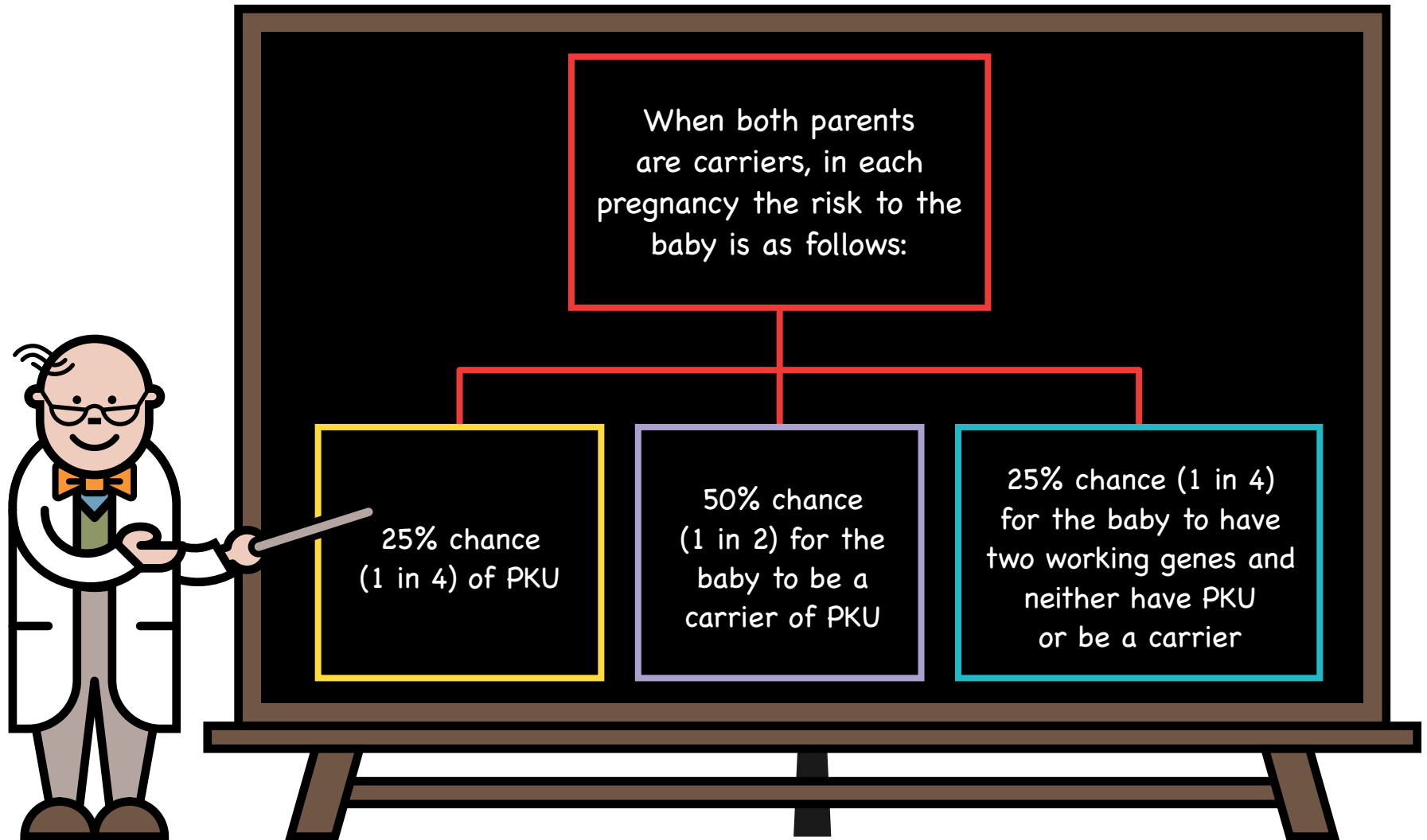
Inheritance – Autosomal recessive (carriers of PKU)



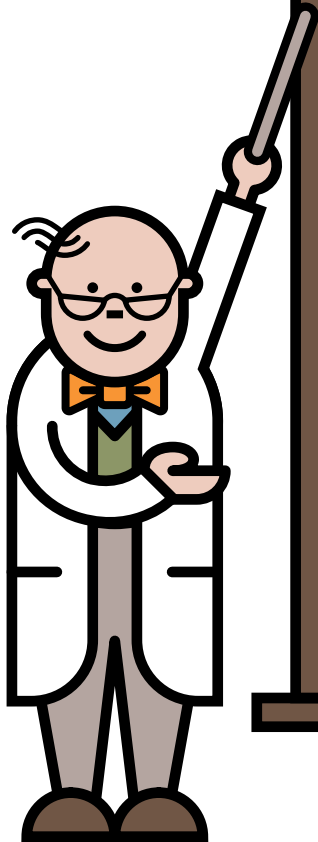
Inheritance – Autosomal recessive – possible combinations



Future pregnancies



Take home messages



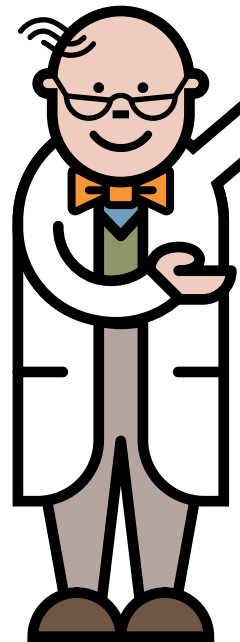
PKU is a serious inherited metabolic disorder

Damage can be prevented with a diet low in phenylalanine and a protein substitute

Remember to always give the correct amount of measured phenylalanine and protein substitute as prescribed by your metabolic centre

Regular blood spot tests are essential to monitor blood phenylalanine levels

Take home messages



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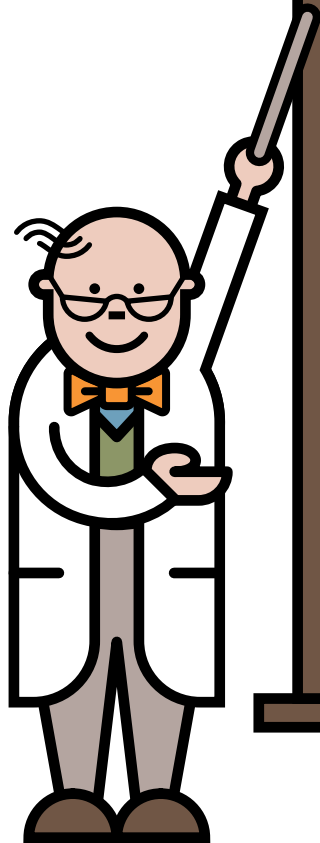


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Helpful hints



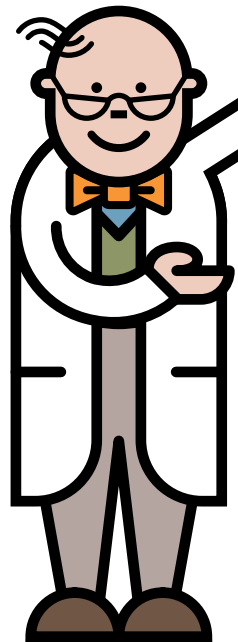
✓ Always ensure you have a good supply of your dietary products and protein substitute and that they are in date

Your dietary products and protein substitute 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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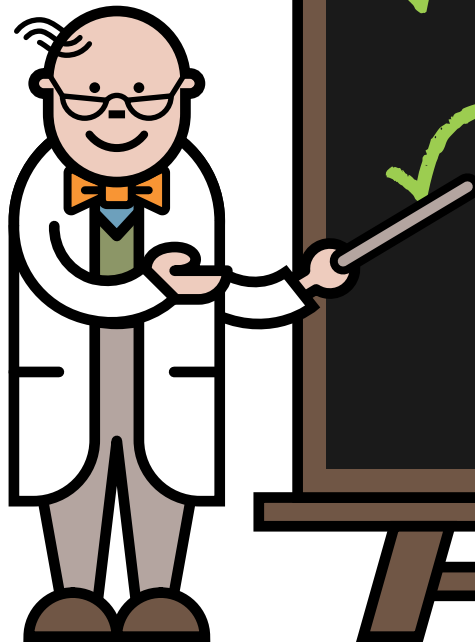


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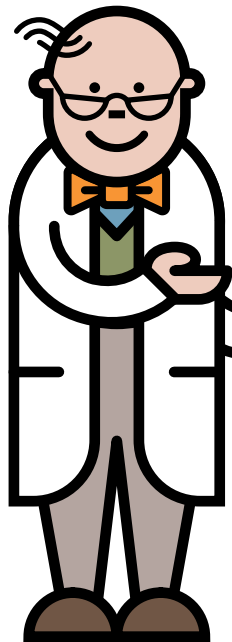
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Who's who

- 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.



www.bimdg.org.uk



www.nutricia.co.uk



www.nspku.org



www.pku.ie