

# 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

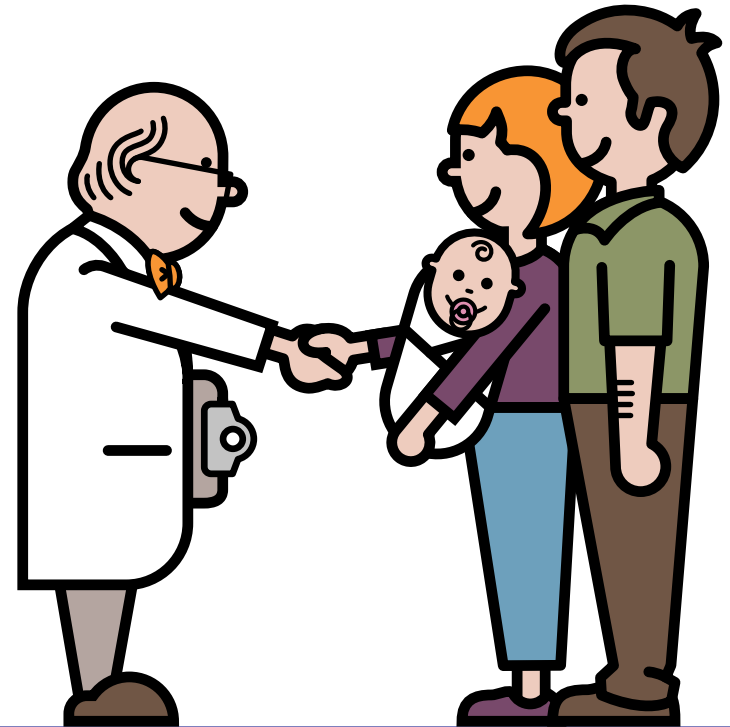
VERSION 2, FEBRUARY 2017

PKU

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

# PKU

Information for families following  
a positive newborn screening



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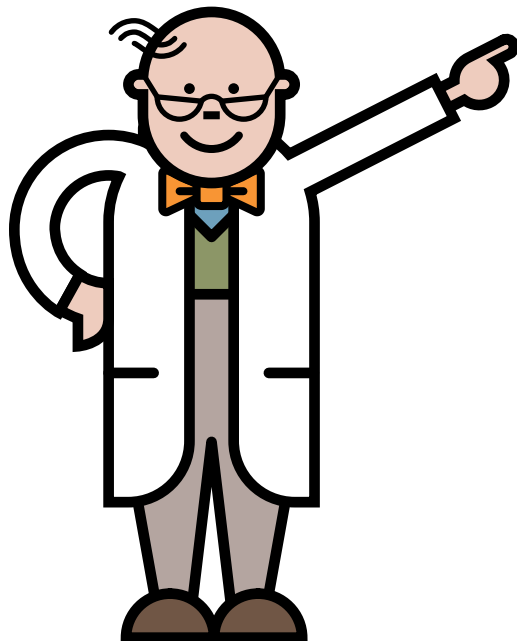
Tools Enabling Metabolic Parents LEarning

Supported by  **NUTRICIA**  
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# What is PKU?

PKU stands for Phenylketonuria

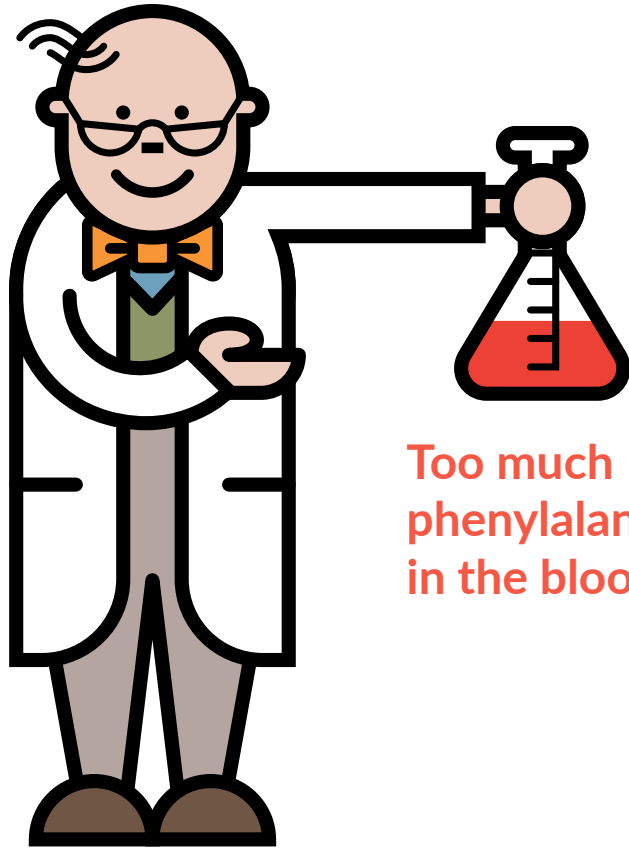
It is an inherited metabolic condition



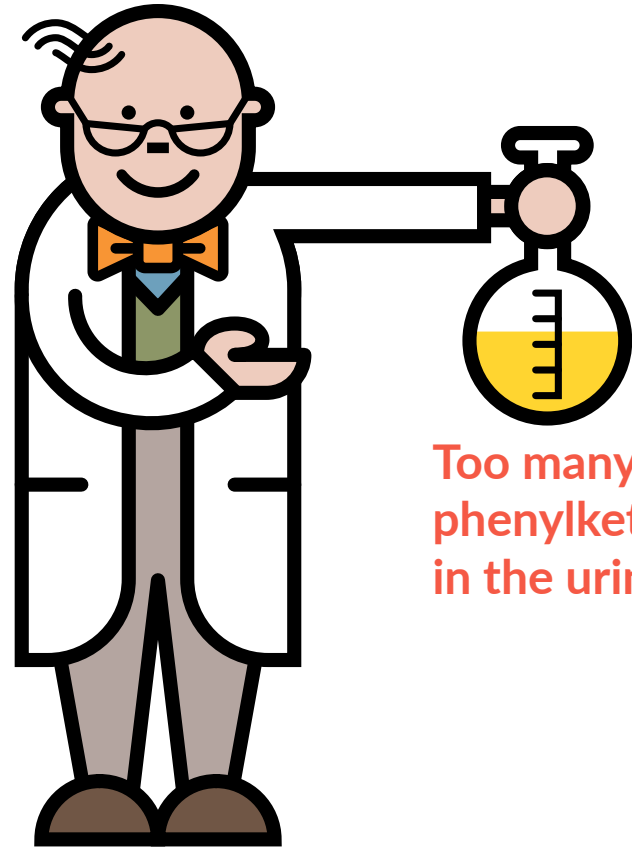
Phenyl Keton Uria

PKU

# What is PKU?



Too much  
phenylalanine  
in the blood



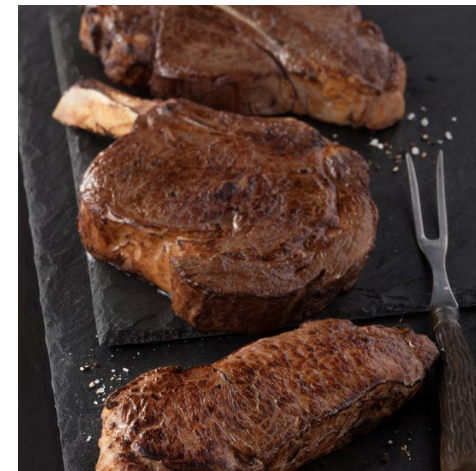
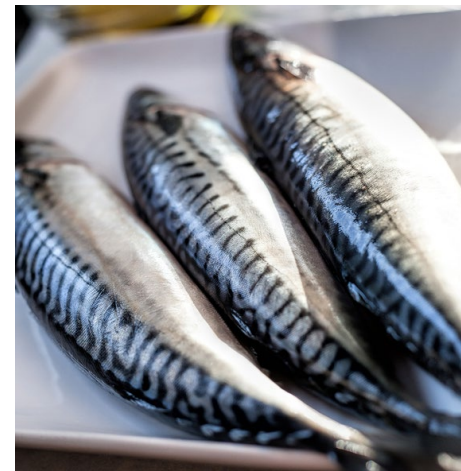
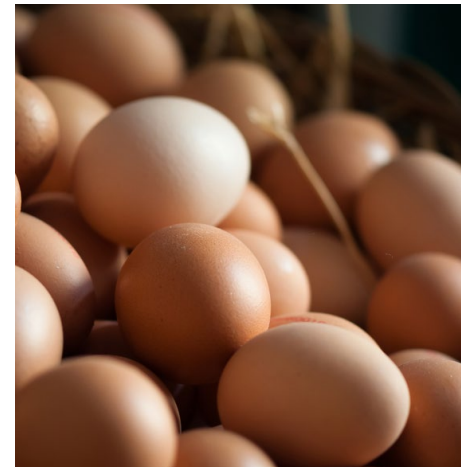
Too many  
phenylketones  
in the urine

# PKU and protein

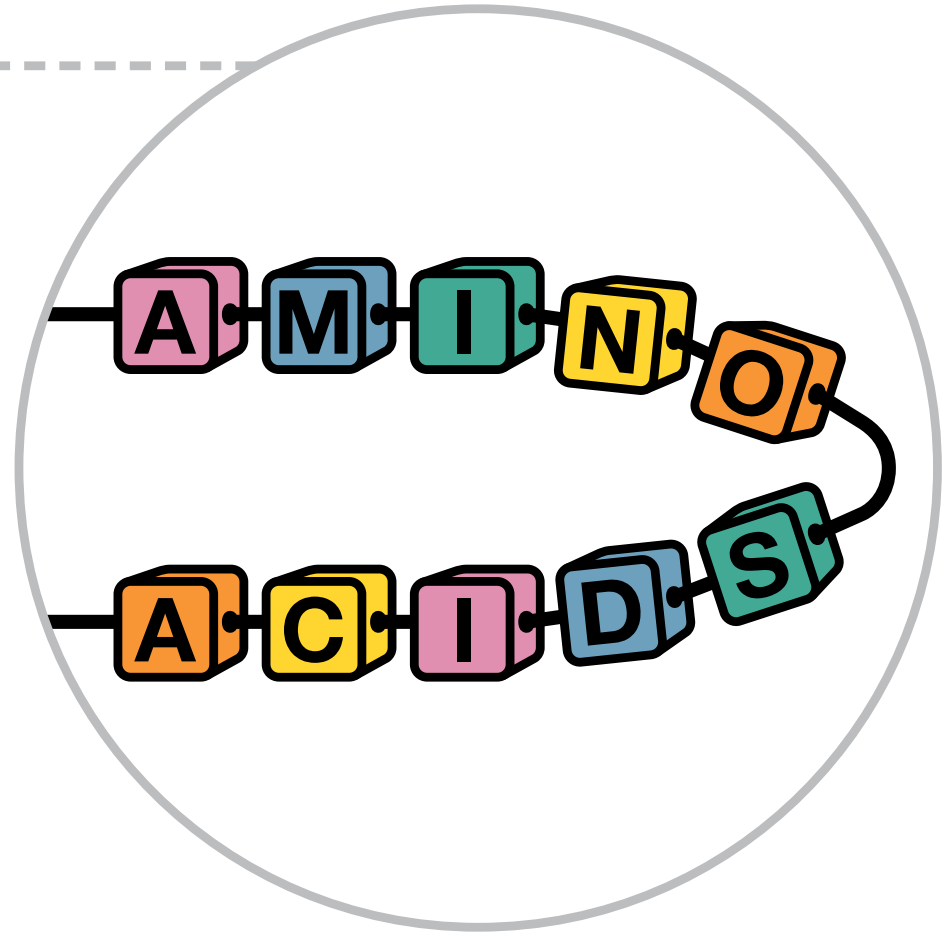
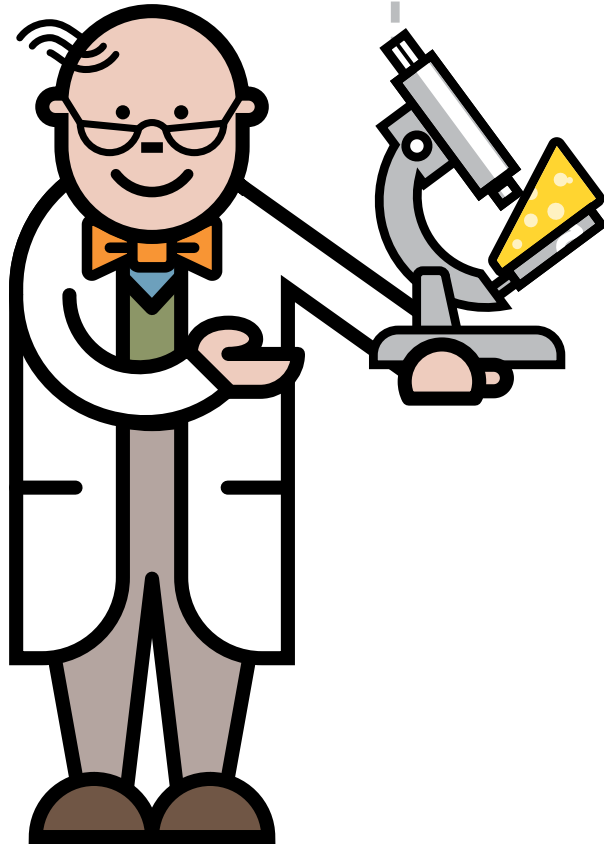
PKU affects the way your baby breaks down protein

Many foods contain protein

The body needs protein for growth and repair



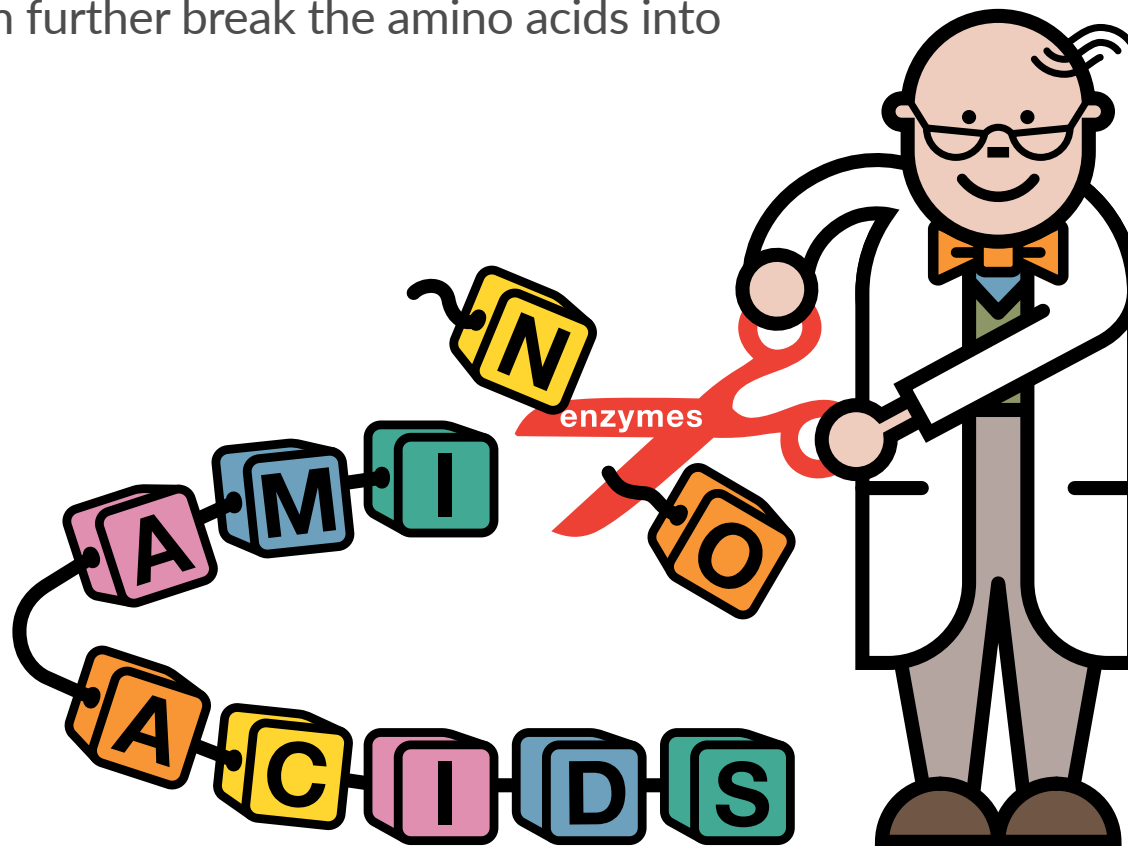
# What is protein?



# Protein and enzymes

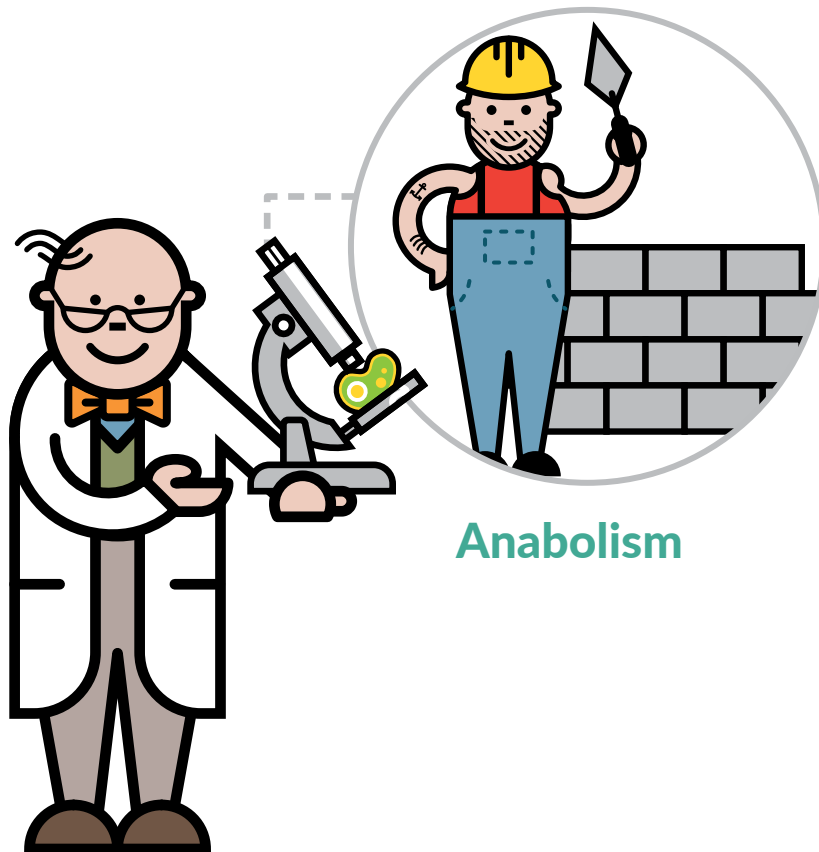
Protein is broken down into amino acids (building blocks of protein) by enzymes (which are like chemical scissors).

Enzymes then further break the amino acids into smaller parts.

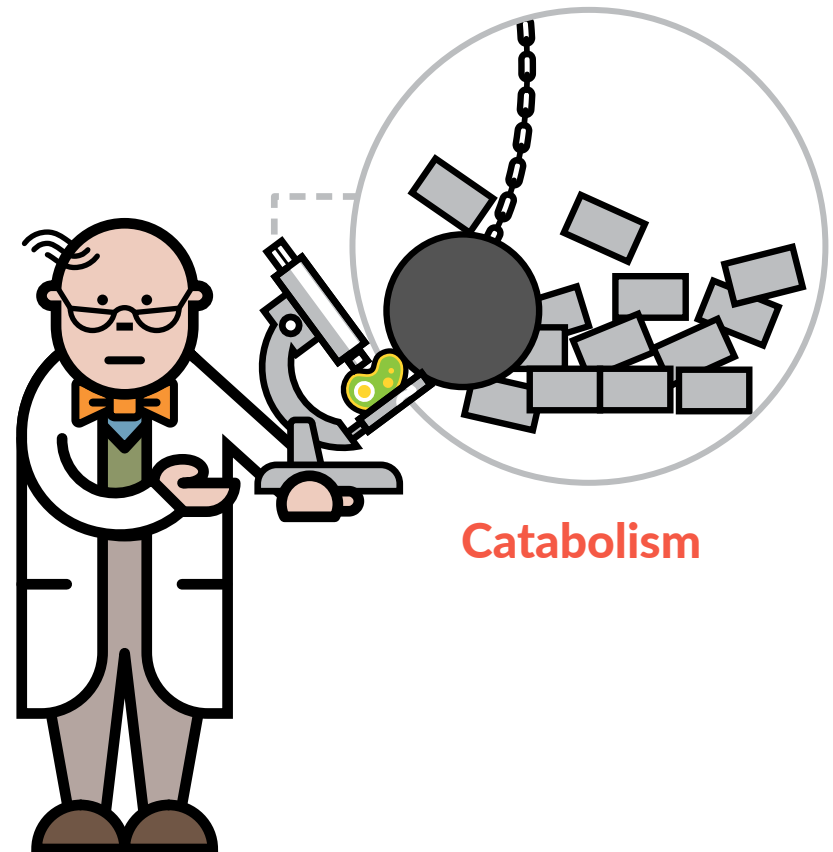


# Protein metabolism

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



Anabolism



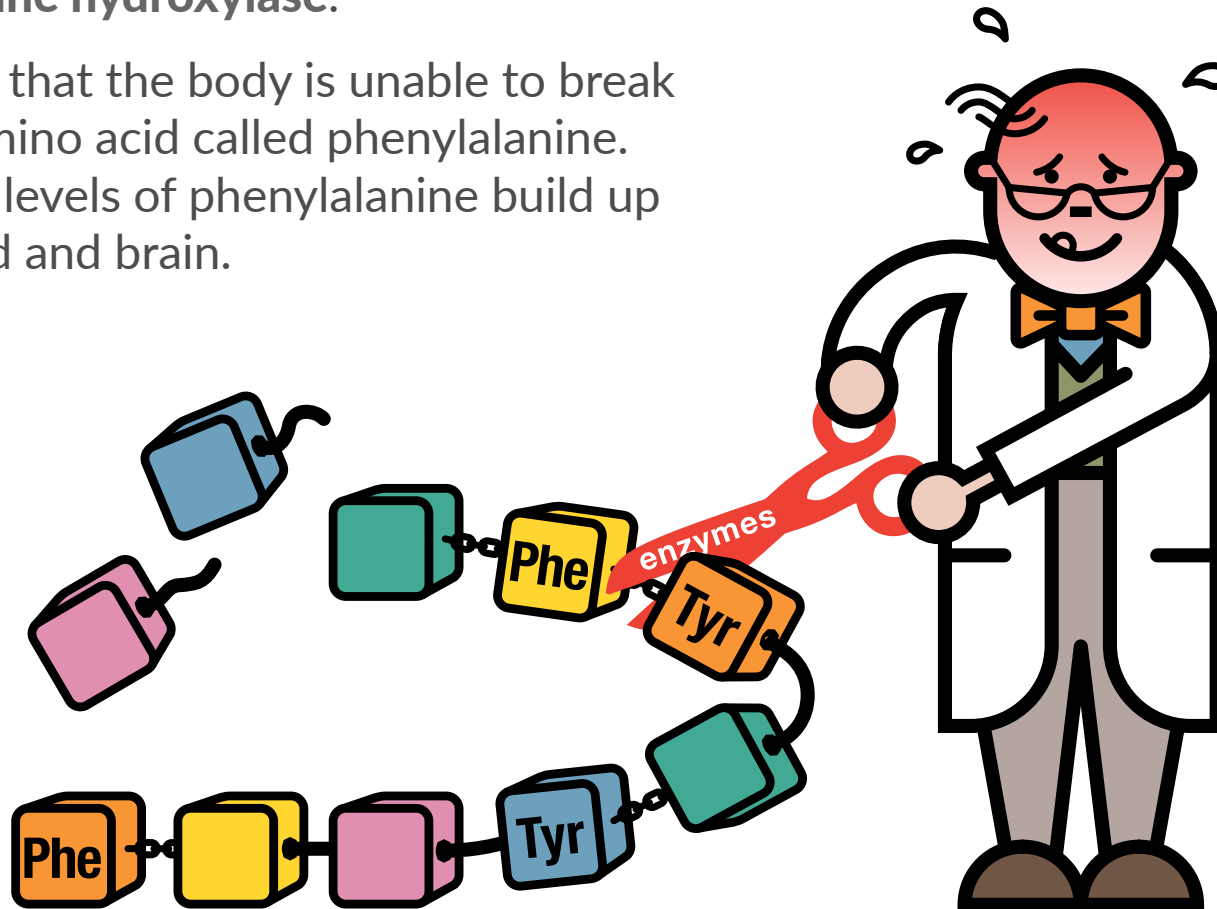
Catabolism



# What happens in PKU?

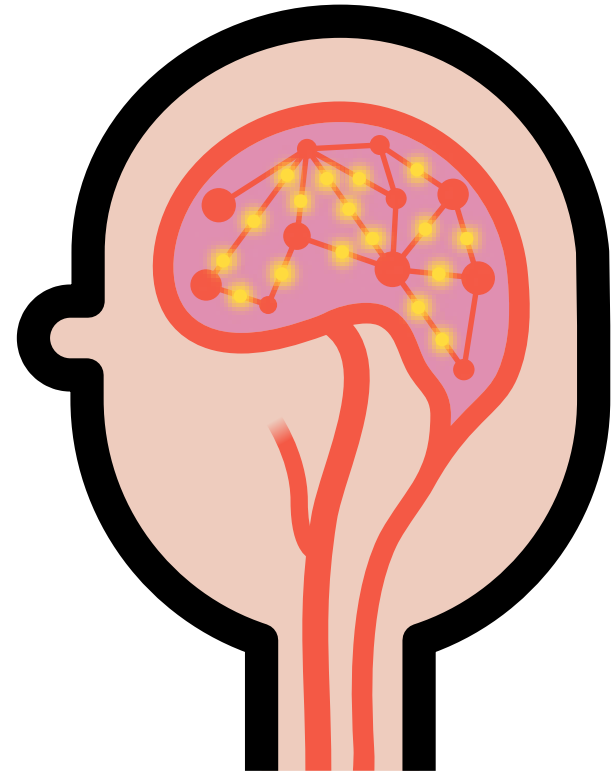
In PKU, the body lacks an enzyme called **phenylalanine hydroxylase**.

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:

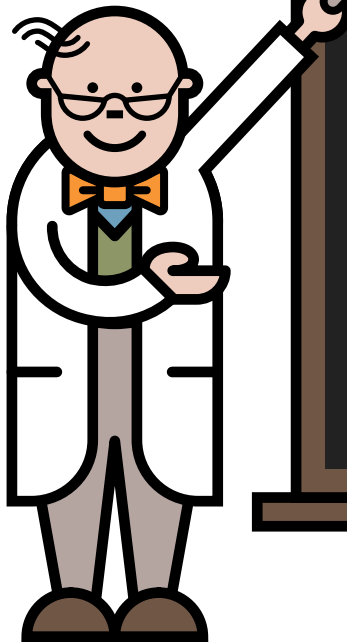
Limited high protein foods

Measured amounts of phenylalanine (protein) containing foods

A protein substitute

Low protein foods

Avoiding aspartame



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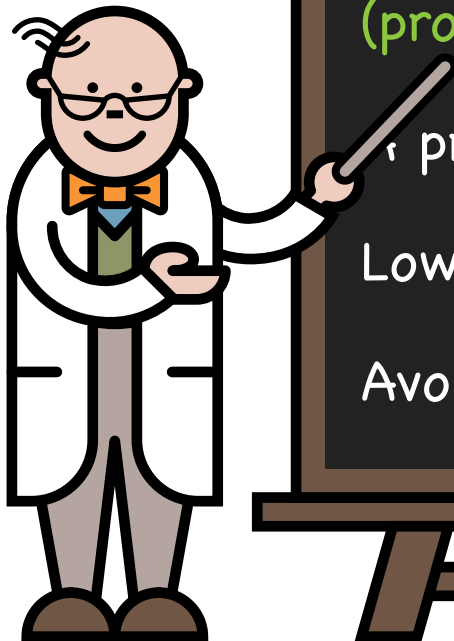
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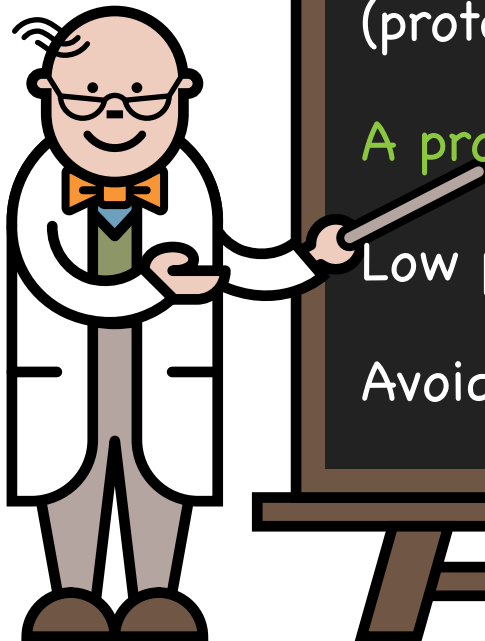
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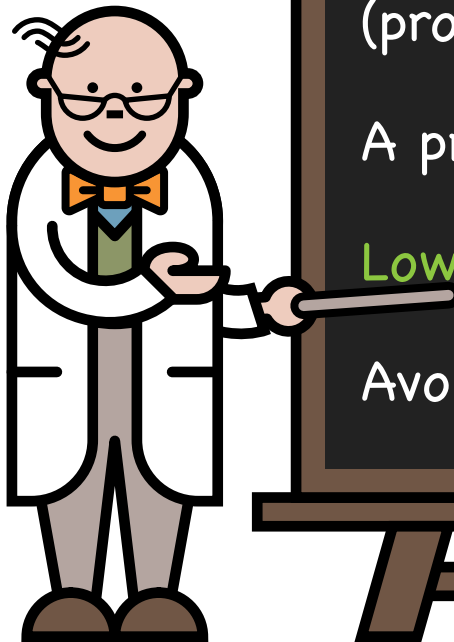
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Low protein foods



Avoiding aspartame



# 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



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, Quorn 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.

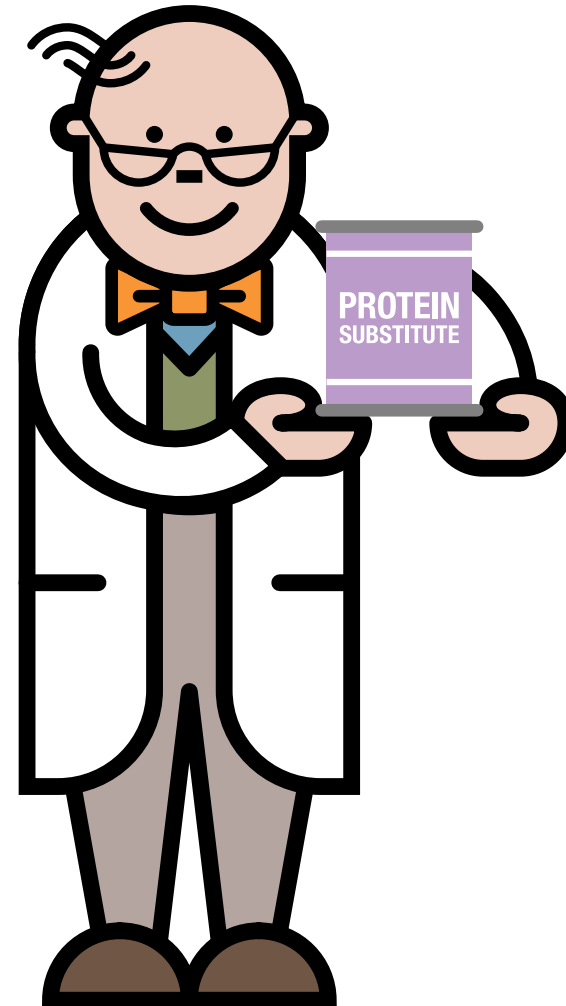


# Protein substitute

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.

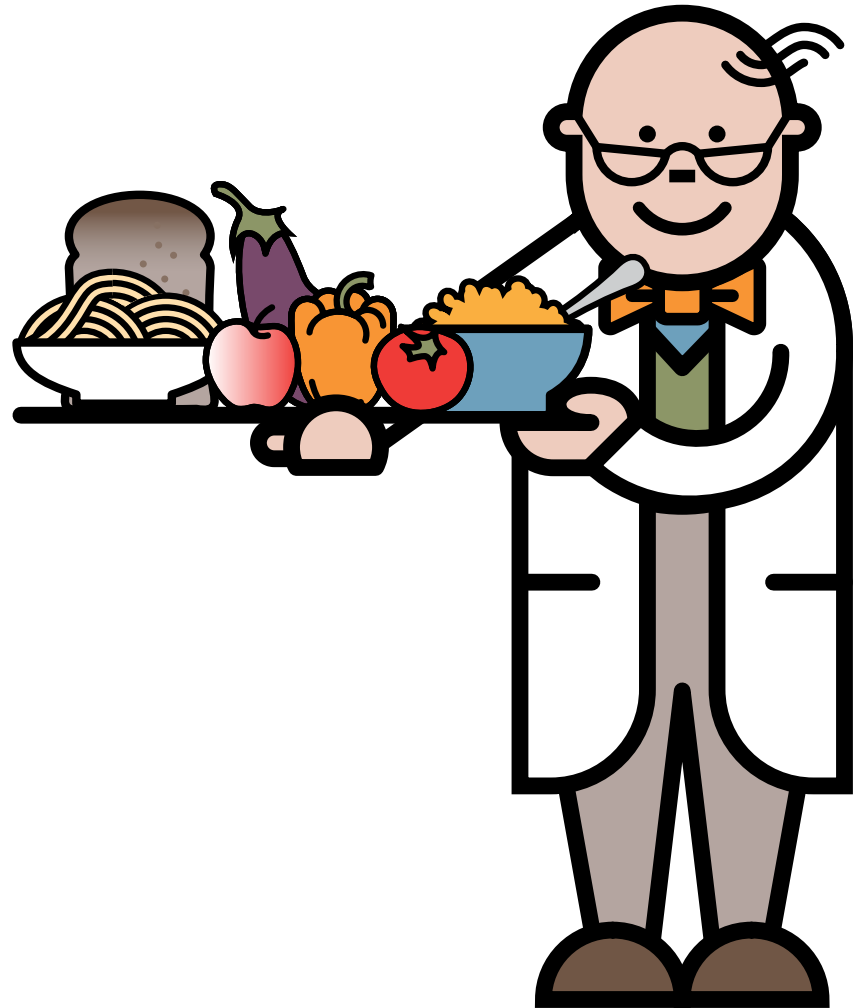


# Low protein foods

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

They provide:

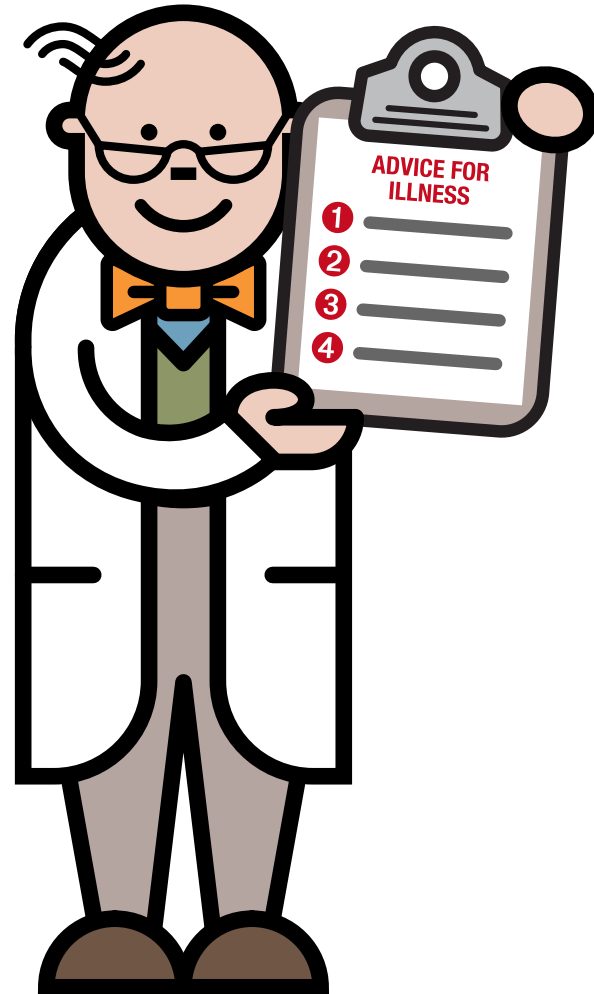
- a source of energy
- variety in the diet



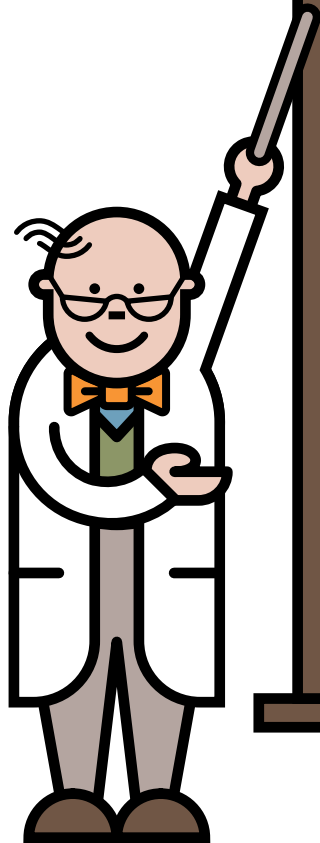
# How is PKU managed during illness?

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

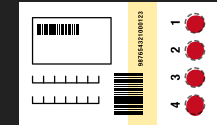


Diet is adjusted according to age, weight and phenylalanine levels



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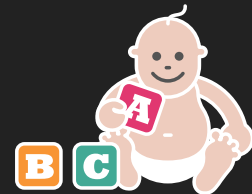
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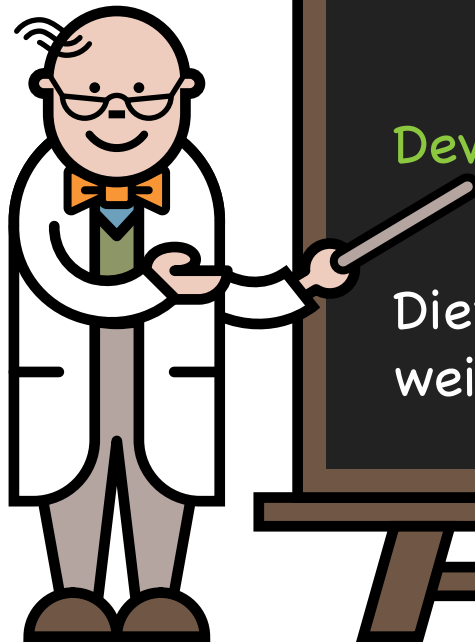
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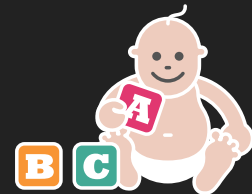
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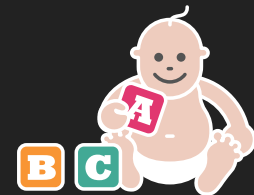
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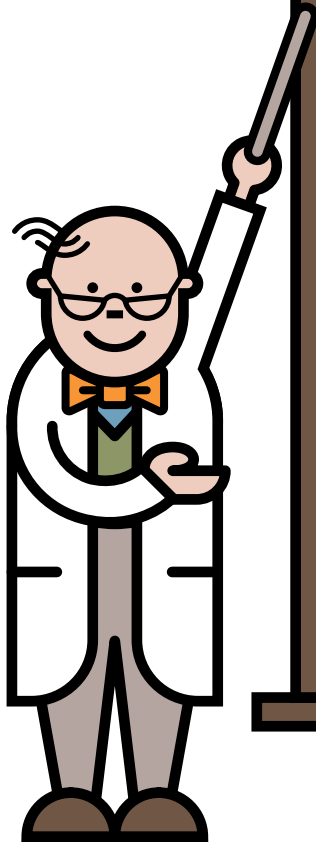
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# Chromosomes, genes, mutations



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

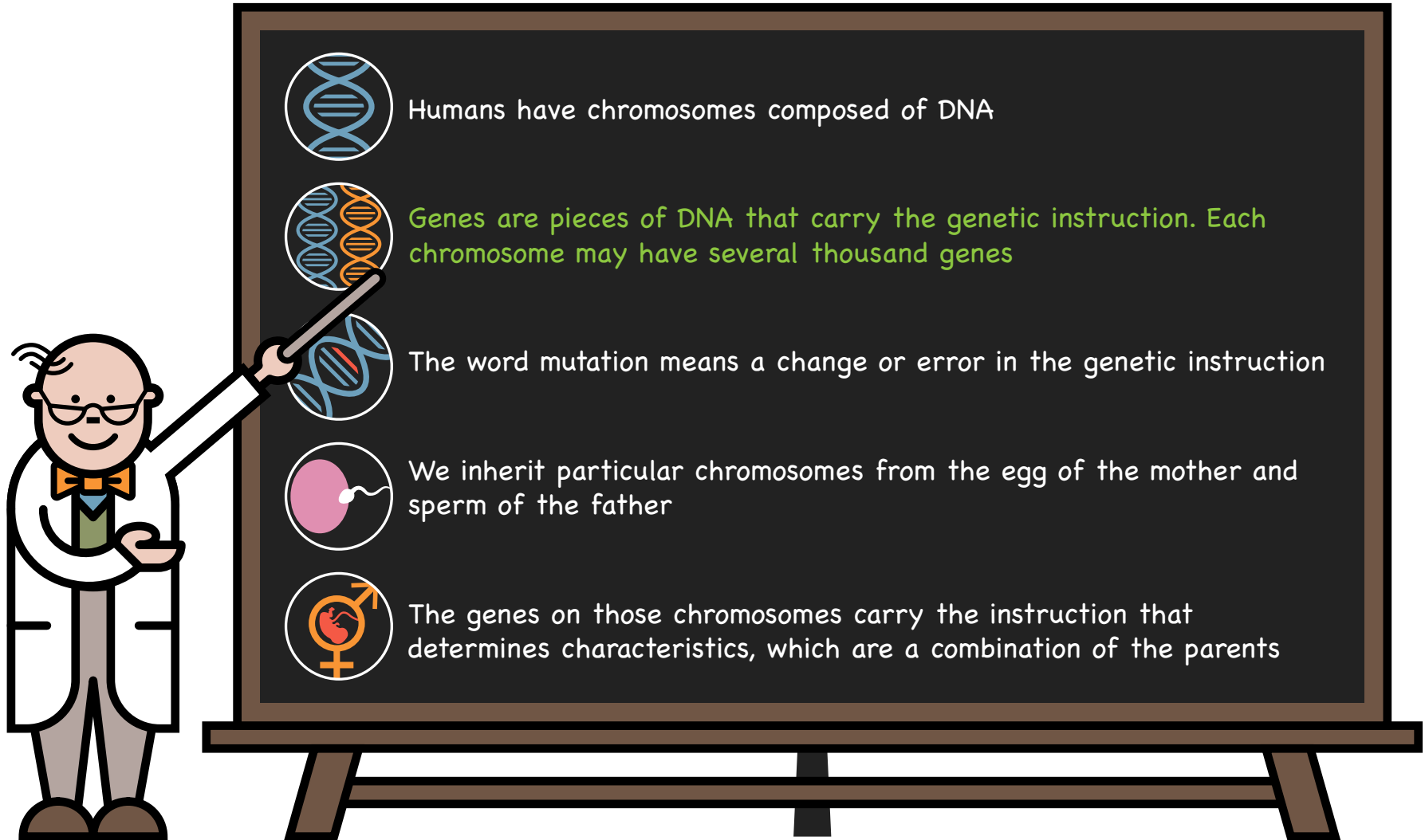


We inherit particular chromosomes from the egg of the mother and sperm of the father



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

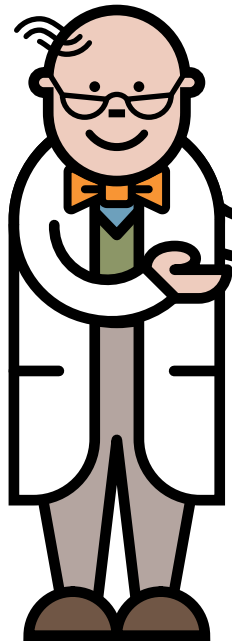
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# Chromosomes, genes, mutations



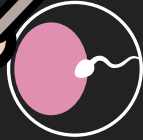
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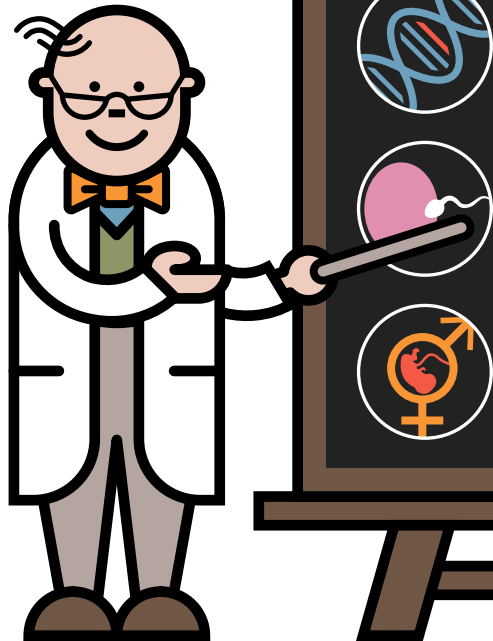


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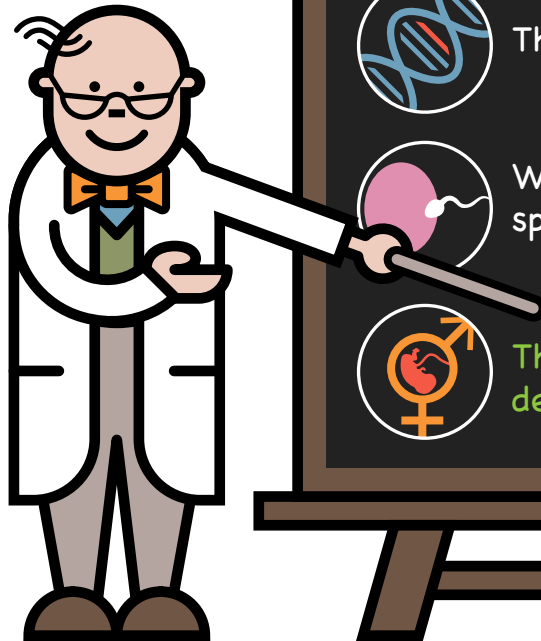


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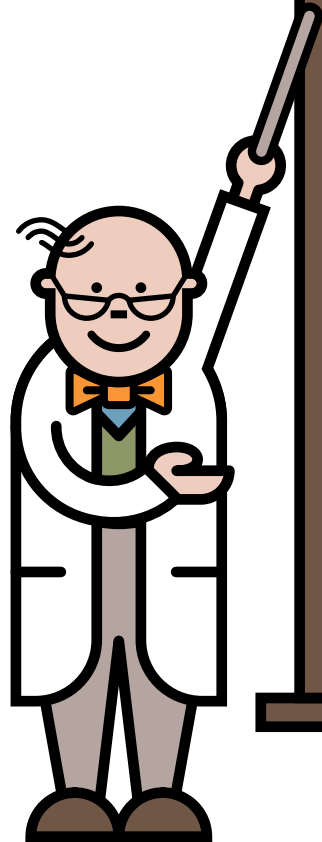


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# Inheritance



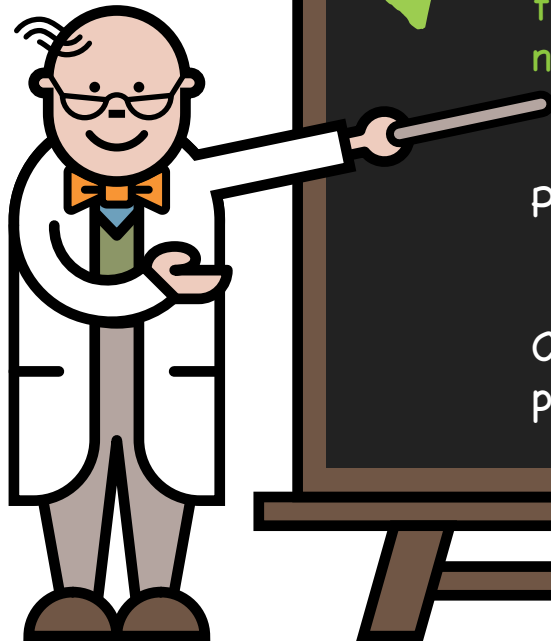
PKU is an inherited condition. There is nothing that could have been done to prevent your baby from having PKU

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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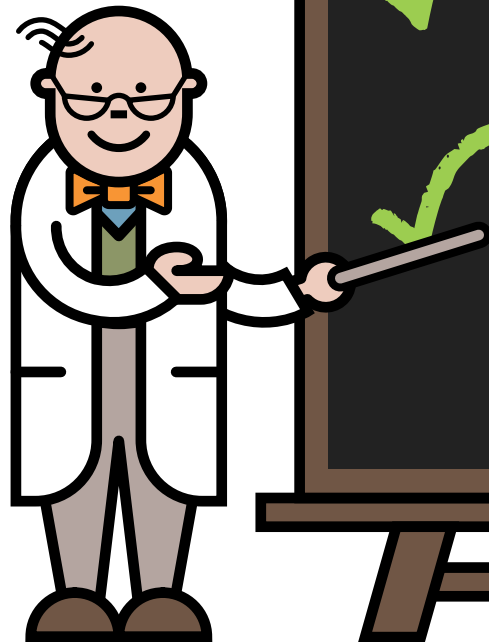
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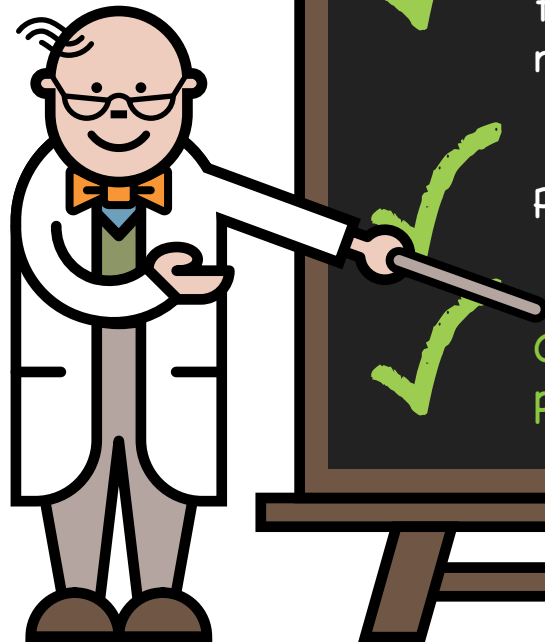
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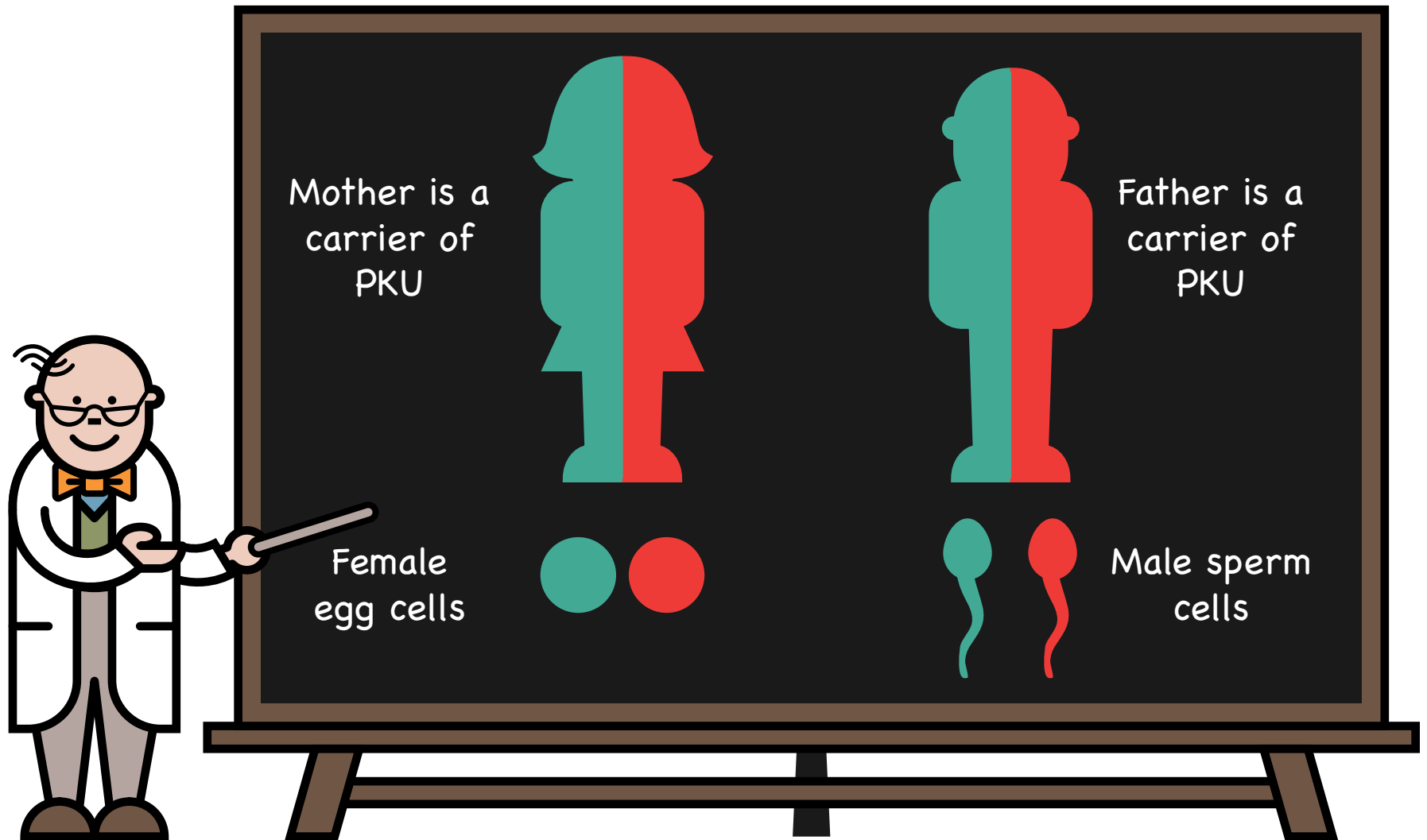
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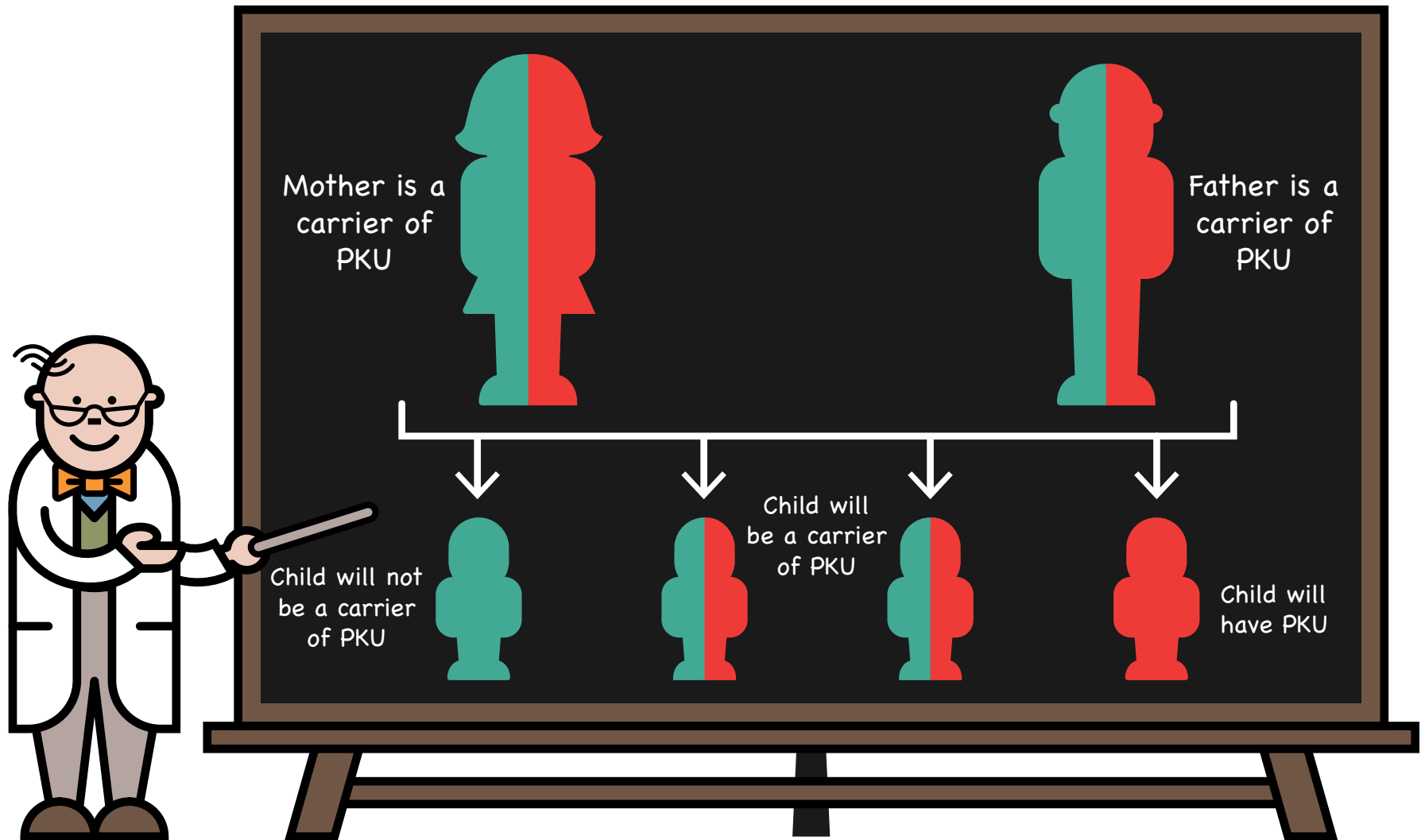
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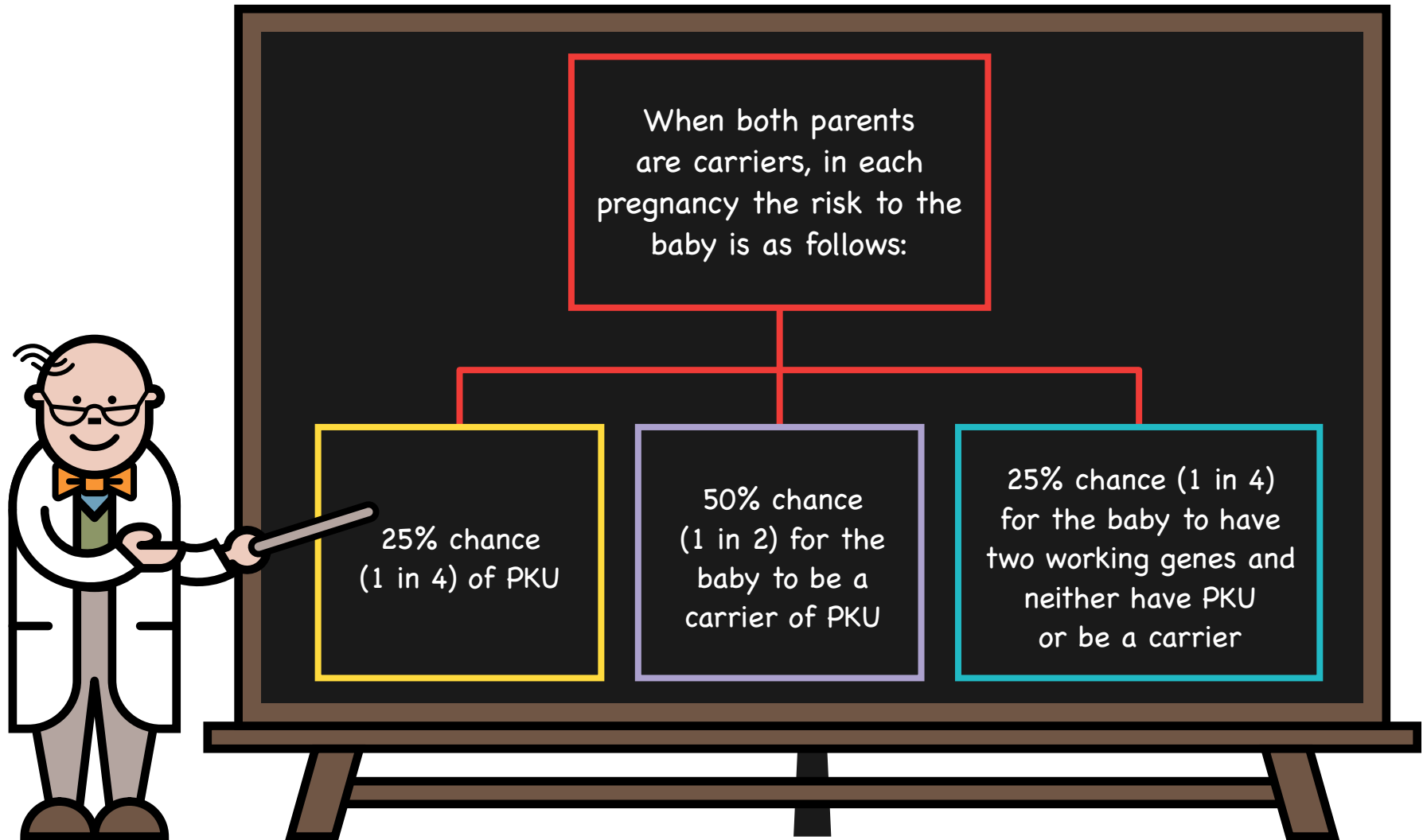
# Inheritance – Autosomal recessive (carriers of PKU)



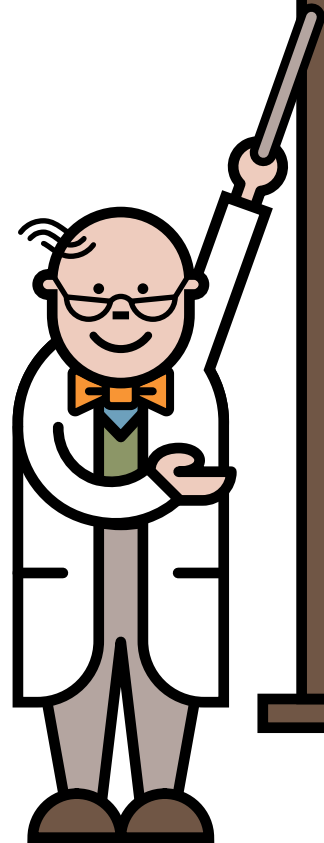
# Inheritance – Autosomal recessive – possible combinations



# 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

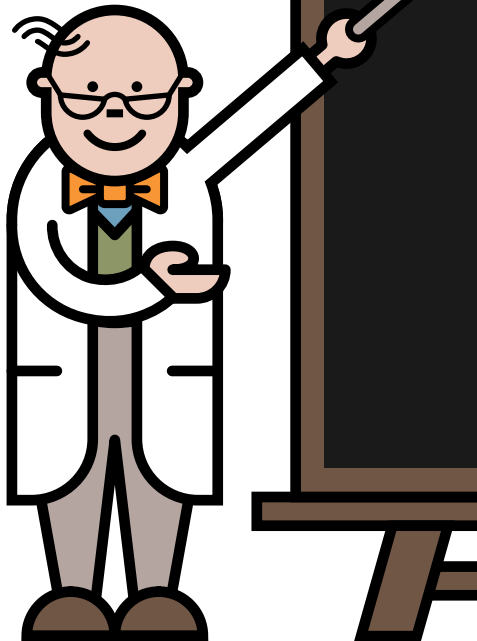
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✓ PKU is a serious inherited metabolic disorder

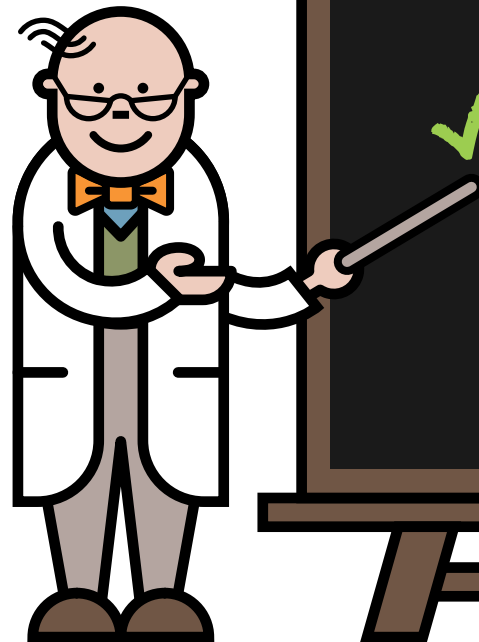
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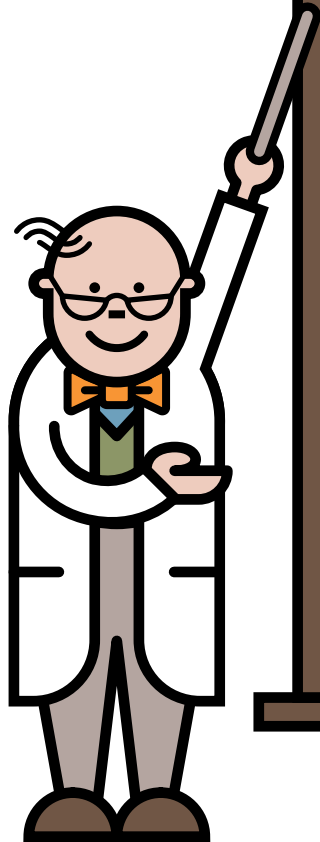


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# Helpful hints



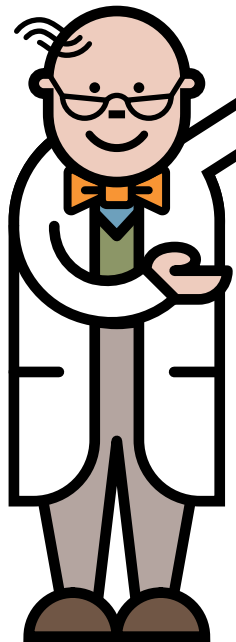
✓ Always ensure you have a good supply of your dietary products and protein substitute and that they are in date

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



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

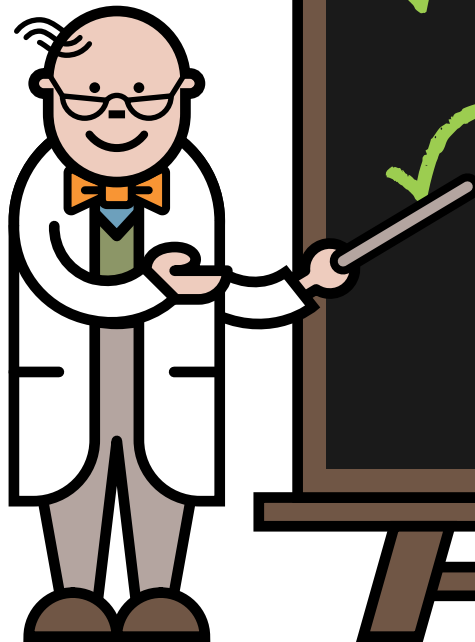


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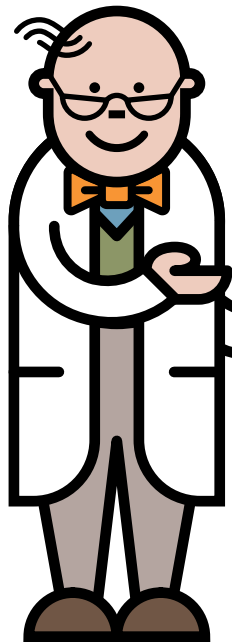
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# Who's who

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

Visit [www.lowproteinconnect.com](http://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.



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British Inherited Metabolic Diseases Group



[www.bimdg.org.uk](http://www.bimdg.org.uk)



[www.nutricia.co.uk](http://www.nutricia.co.uk)

**NSPKU**

[www.nspku.org](http://www.nspku.org)



[www.pku.ie](http://www.pku.ie)