

# 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, JANUARY 2025

**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.

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

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.

# PKU

Information for families following a new diagnosis



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**TEMPLE**

Tools Enabling Metabolic Parents LEarning

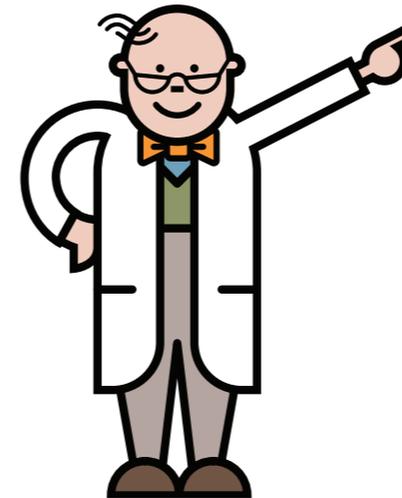


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# What is PKU?

PKU stands for Phenylketonuria

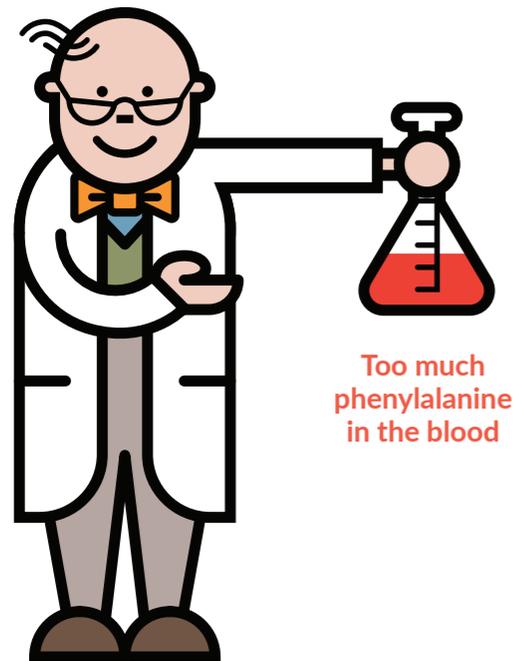
It is an inherited metabolic condition



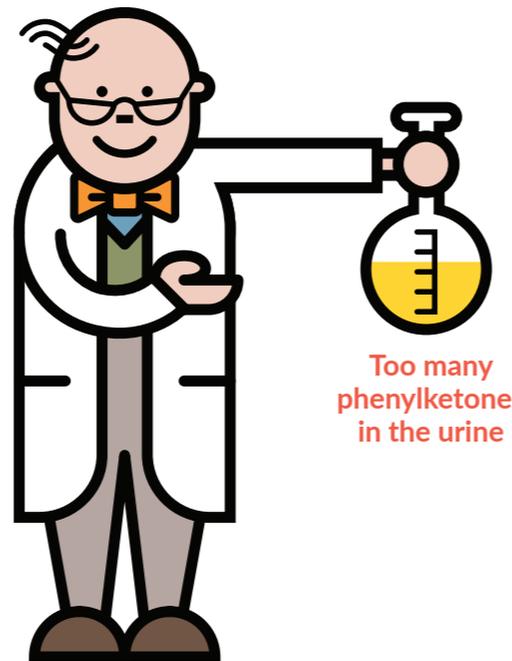
Phenyl Keton Uria

PKU

# What is PKU?



Too much  
phenylalanine  
in the blood



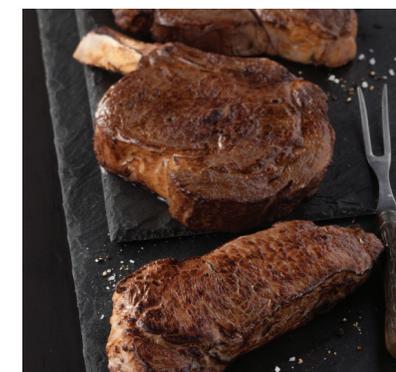
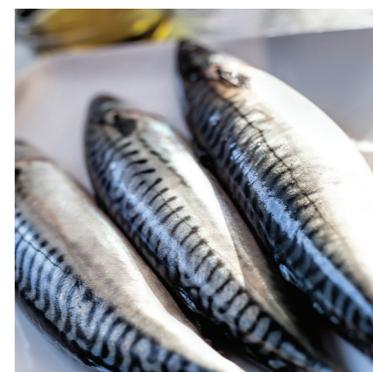
Too many  
phenylketones  
in the urine

# PKU and protein

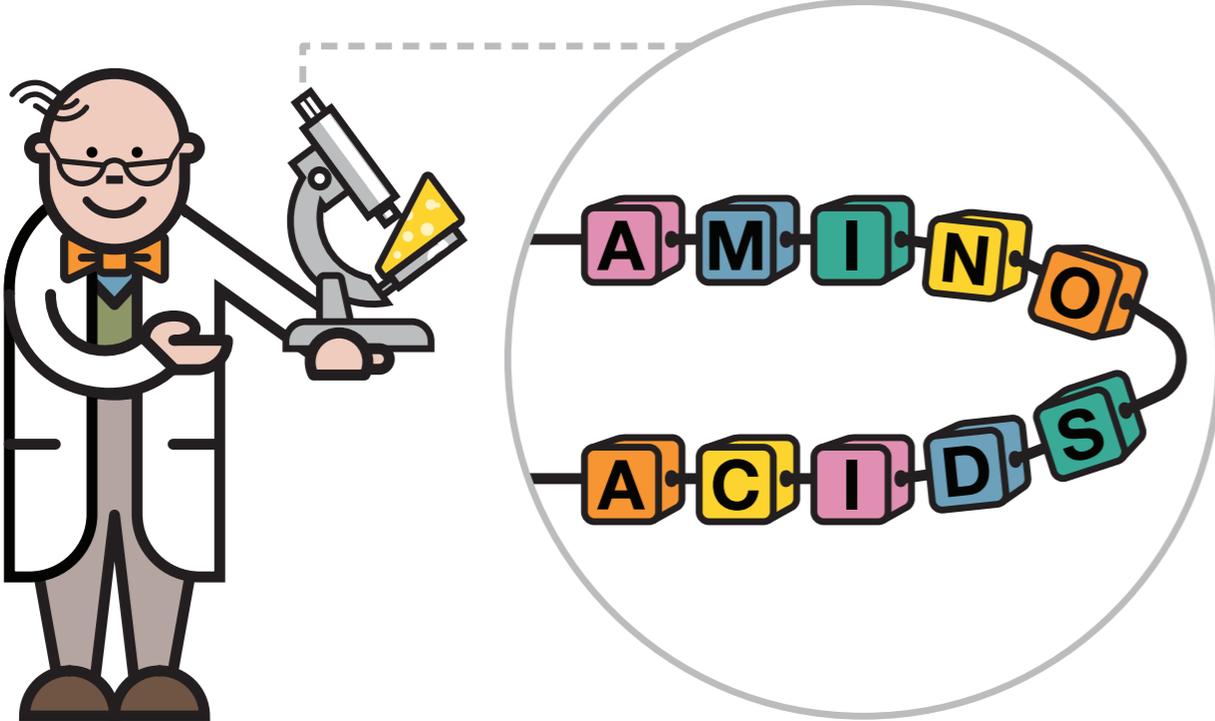
PKU affects the way your baby breaks down protein.

Many foods contain protein.

The body needs protein for growth and repair.



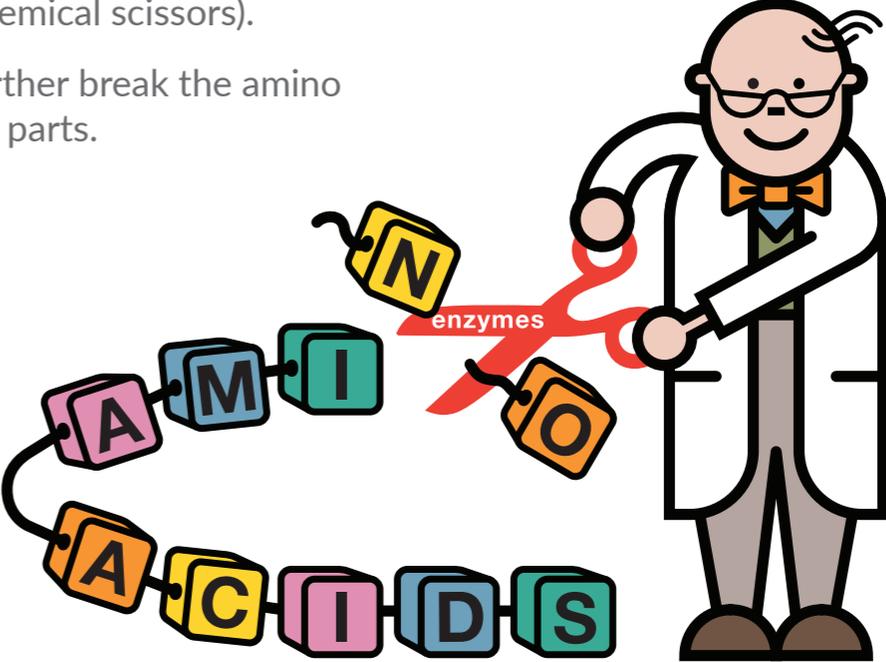
# What is protein?



# Protein and enzymes

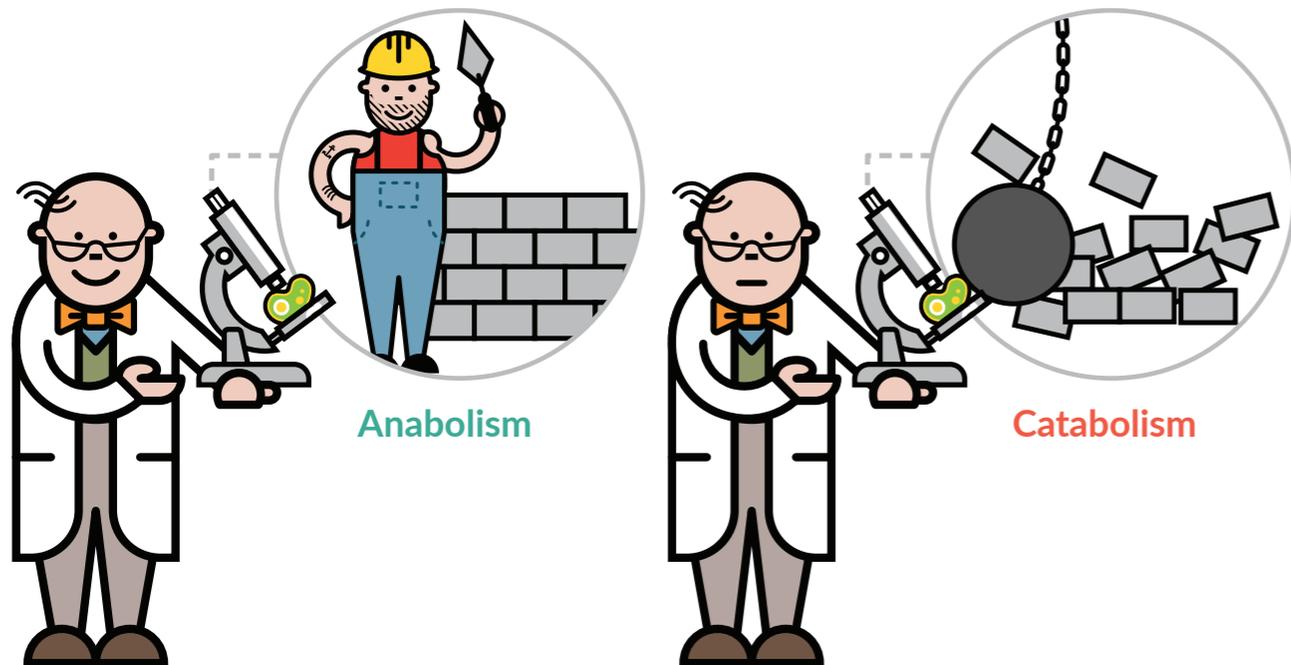
Protein is broken down into amino acids (building blocks of protein) by enzymes (which are like chemical scissors).

Enzymes then further break the amino acids into smaller parts.



# Protein metabolism

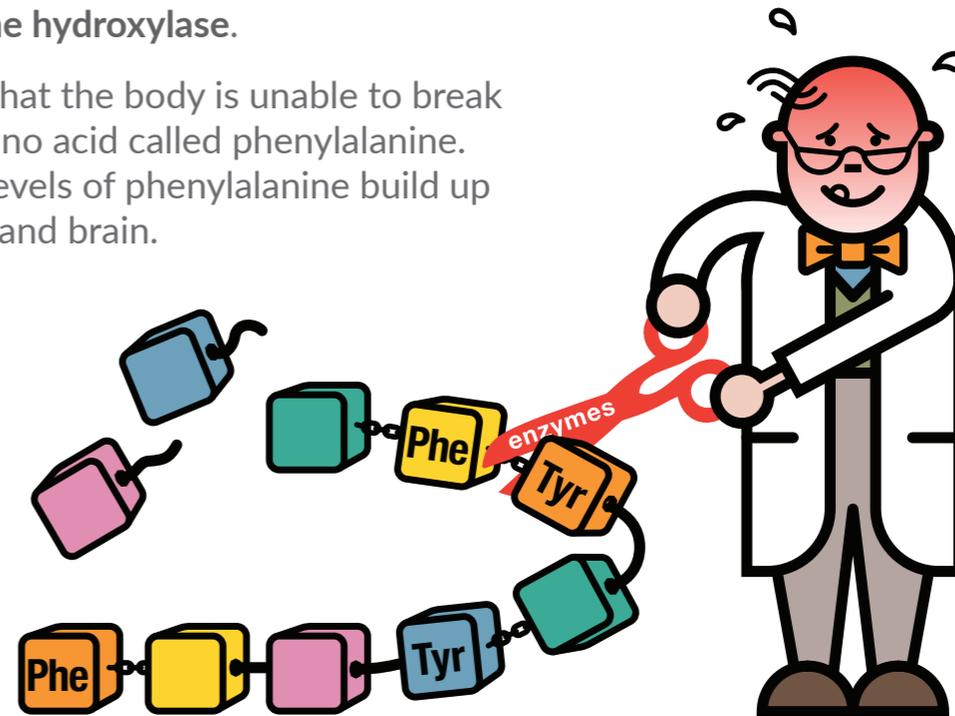
Metabolism refers to the chemical processes that occur inside the cells of the body.



# What happens in PKU?

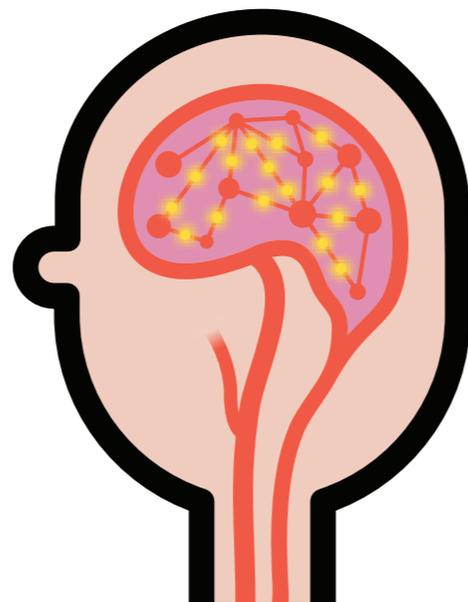
In PKU, the body lacks an enzyme called **phenylalanine hydroxylase**.

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.



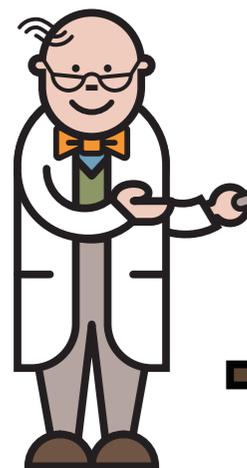
Early management can prevent brain damage and learning difficulties

## 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



## High protein foods

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

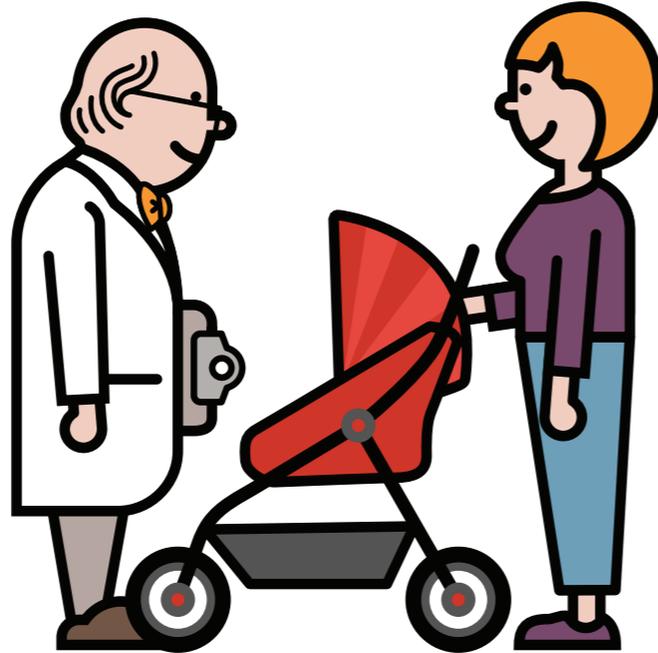
Any food or drink containing aspartame should also be avoided.



## 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.

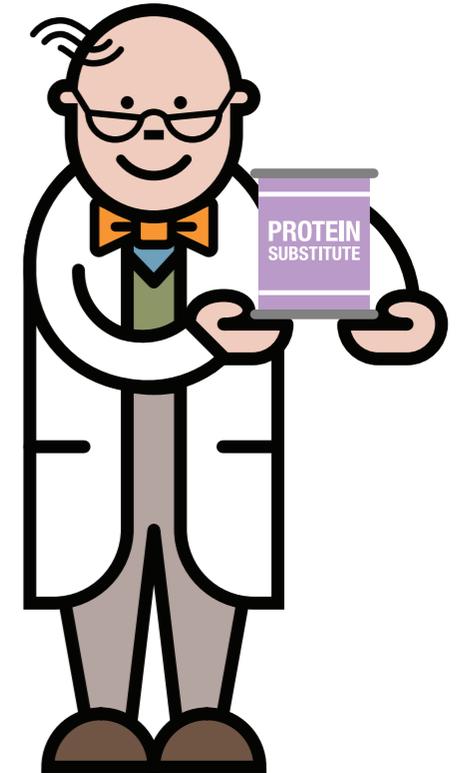


## Protein substitute

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.

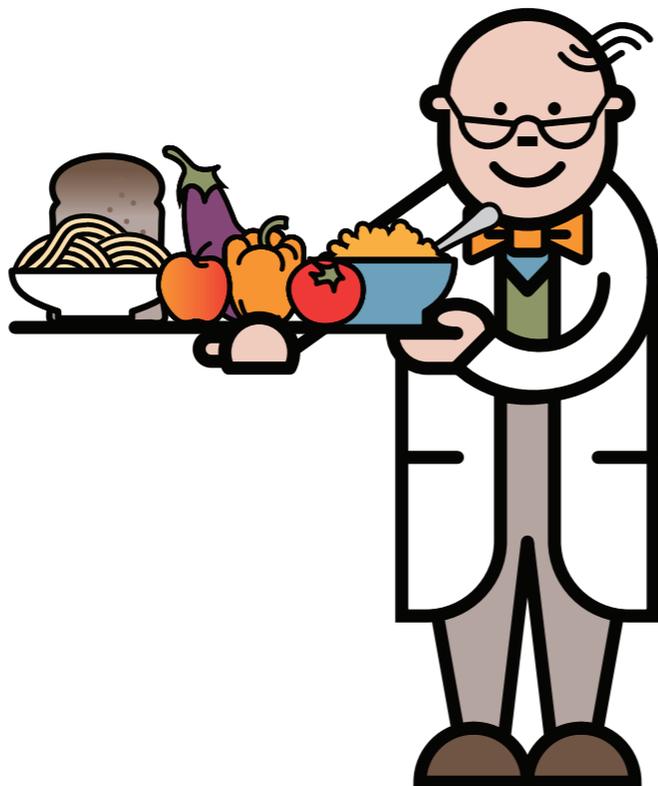


## Low protein foods

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

They provide:

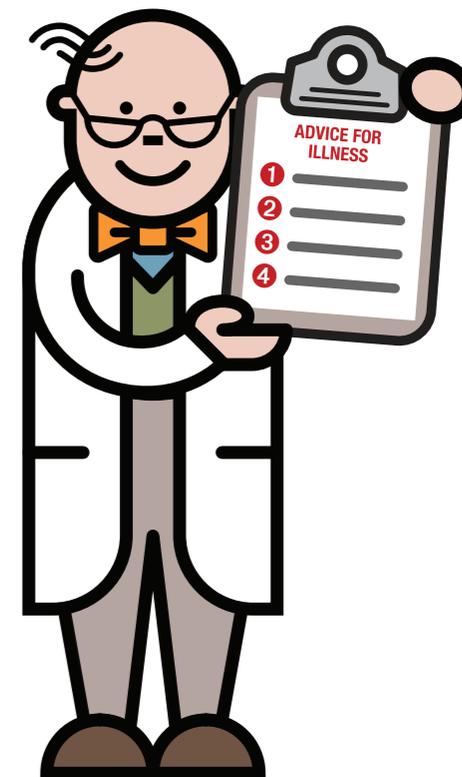
- a source of energy
- variety in the diet



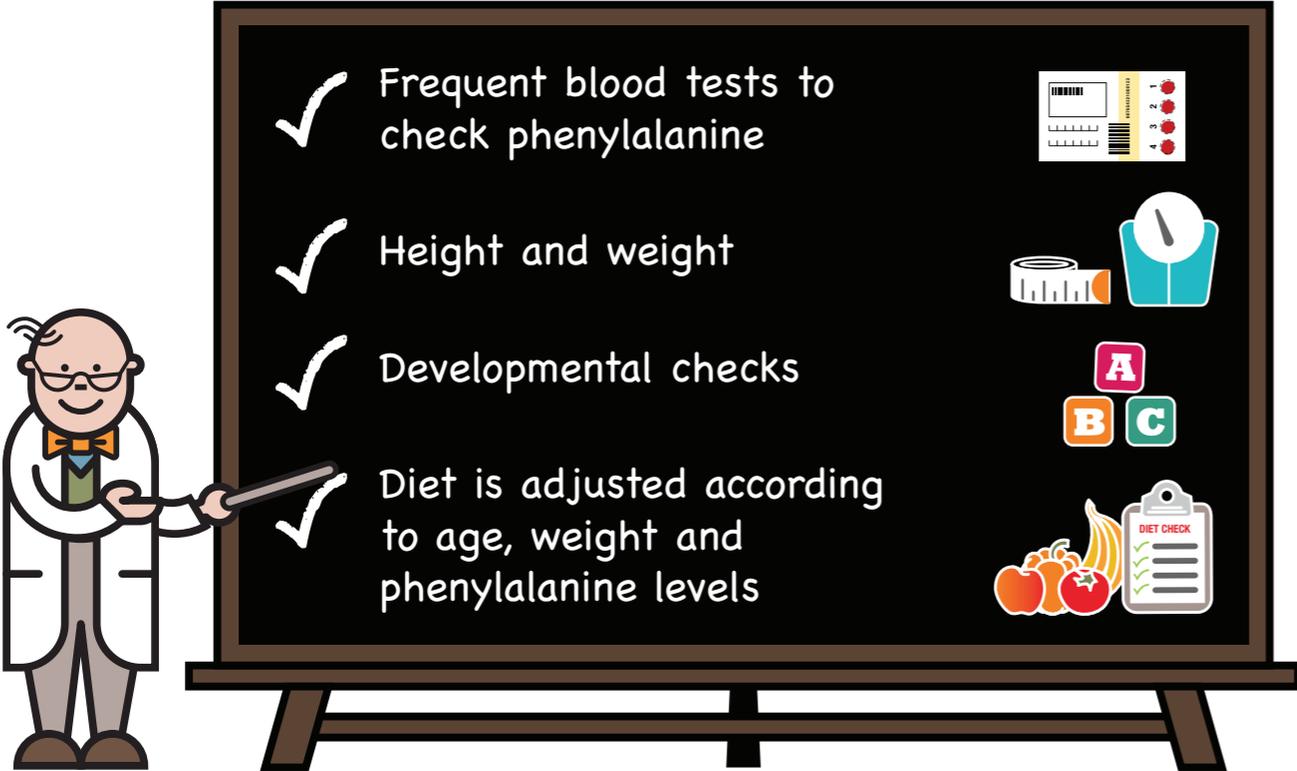
## How is PKU managed during illness?

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.

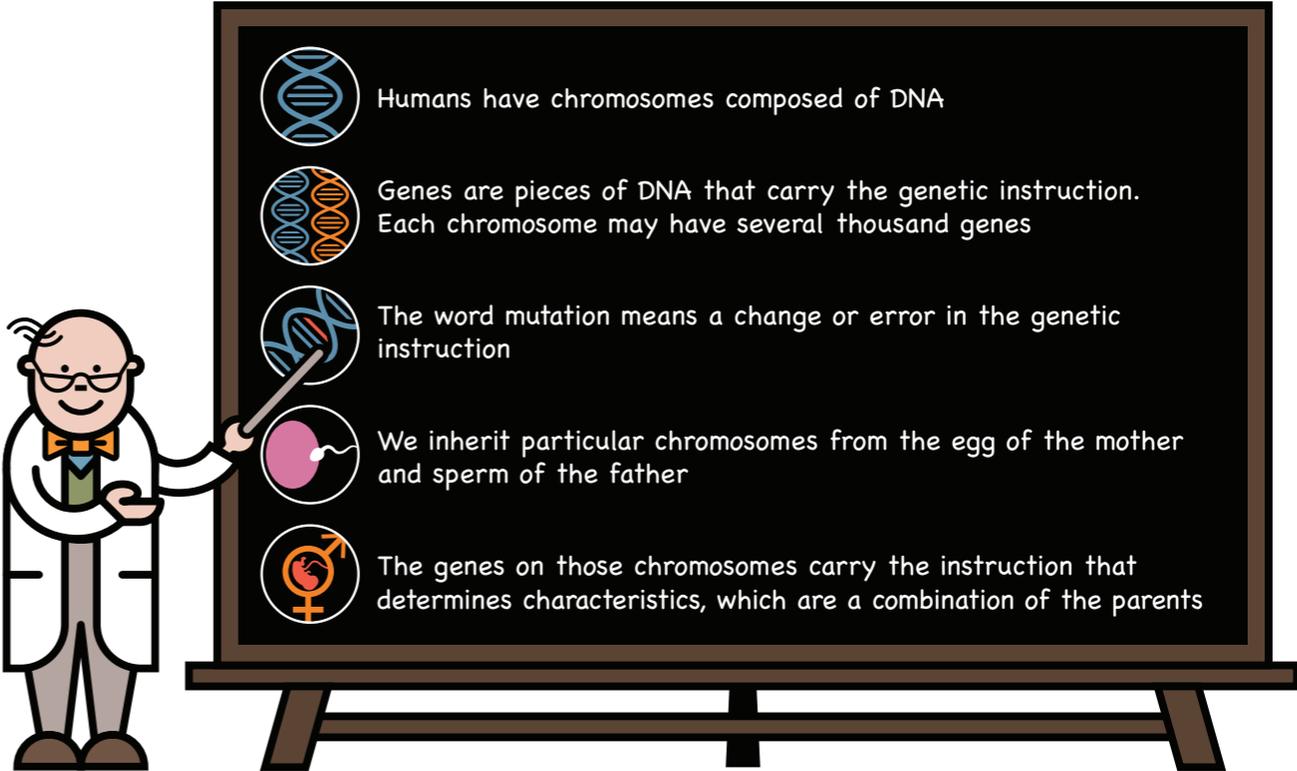


# How is PKU monitored?



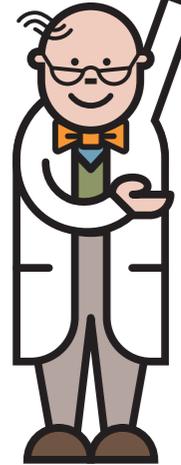
- ✓ Frequent blood tests to check phenylalanine 
- ✓ Height and weight 
- ✓ Developmental checks 
- ✓ Diet is adjusted according to age, weight and phenylalanine levels 

# 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

# 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 — Autosomal-recessive (carriers of PKU)



Mother is a carrier of PKU



Father is a carrier of PKU



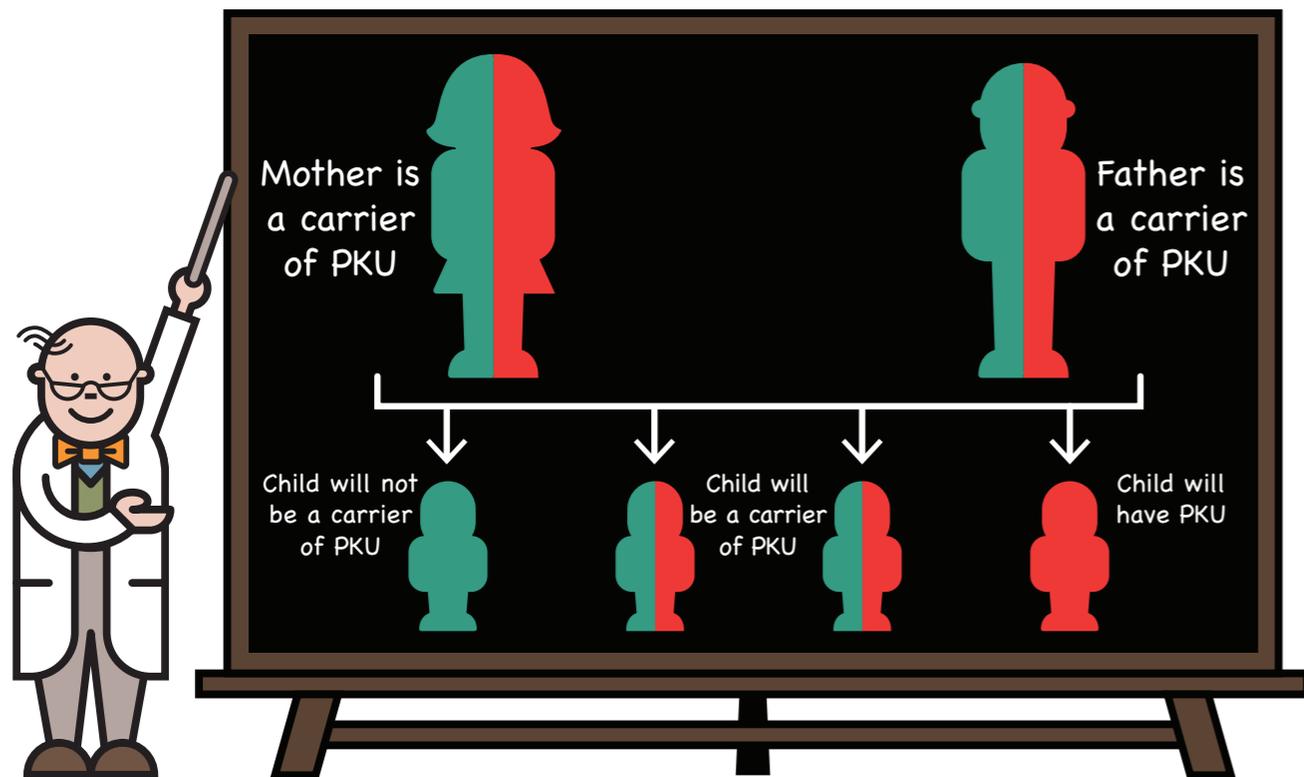
Female egg cells



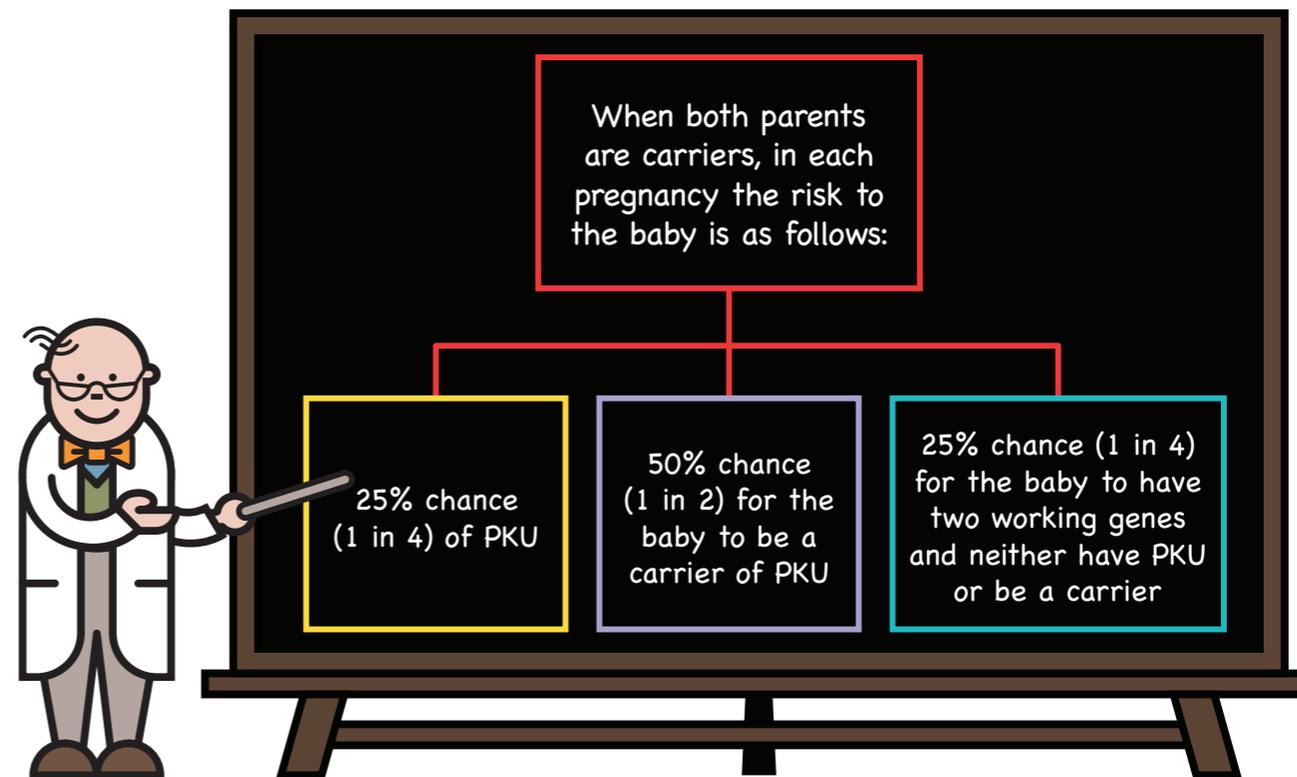
Male sperm cells



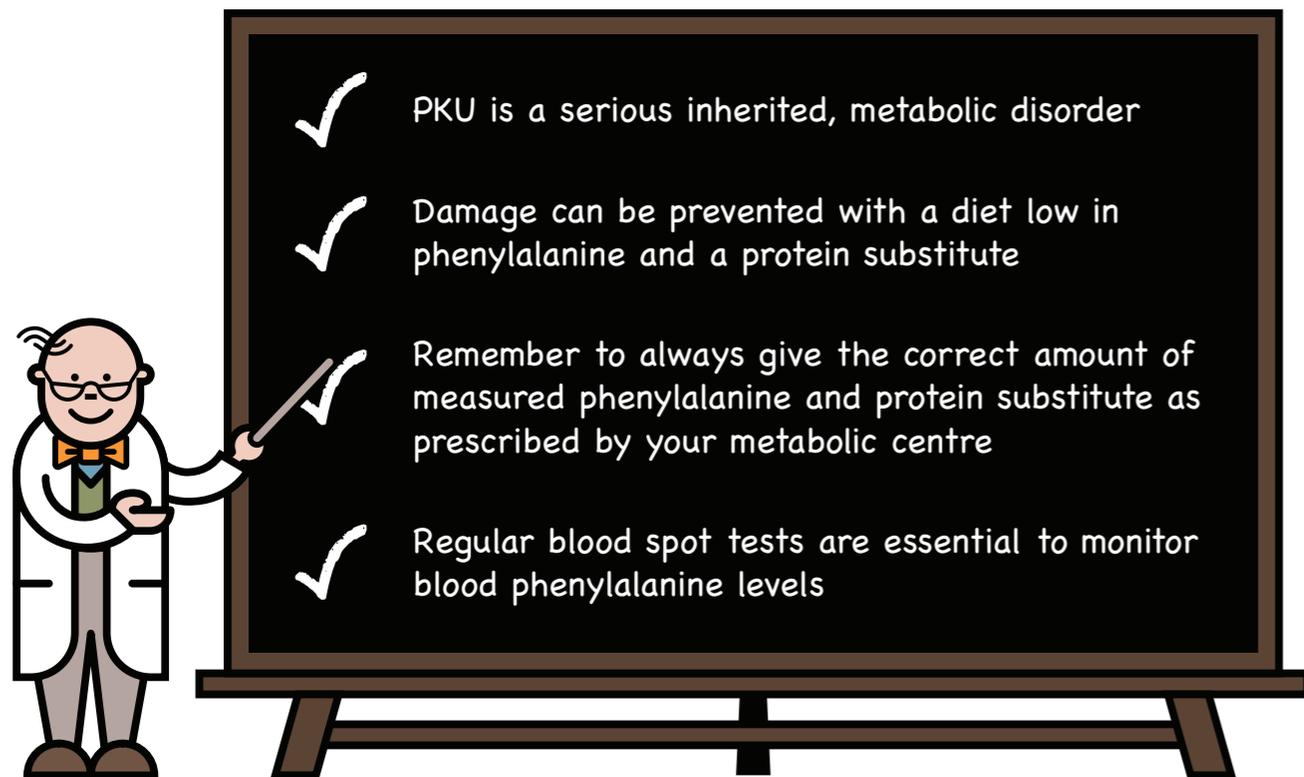
## Inheritance — Autosomal recessive — possible combinations



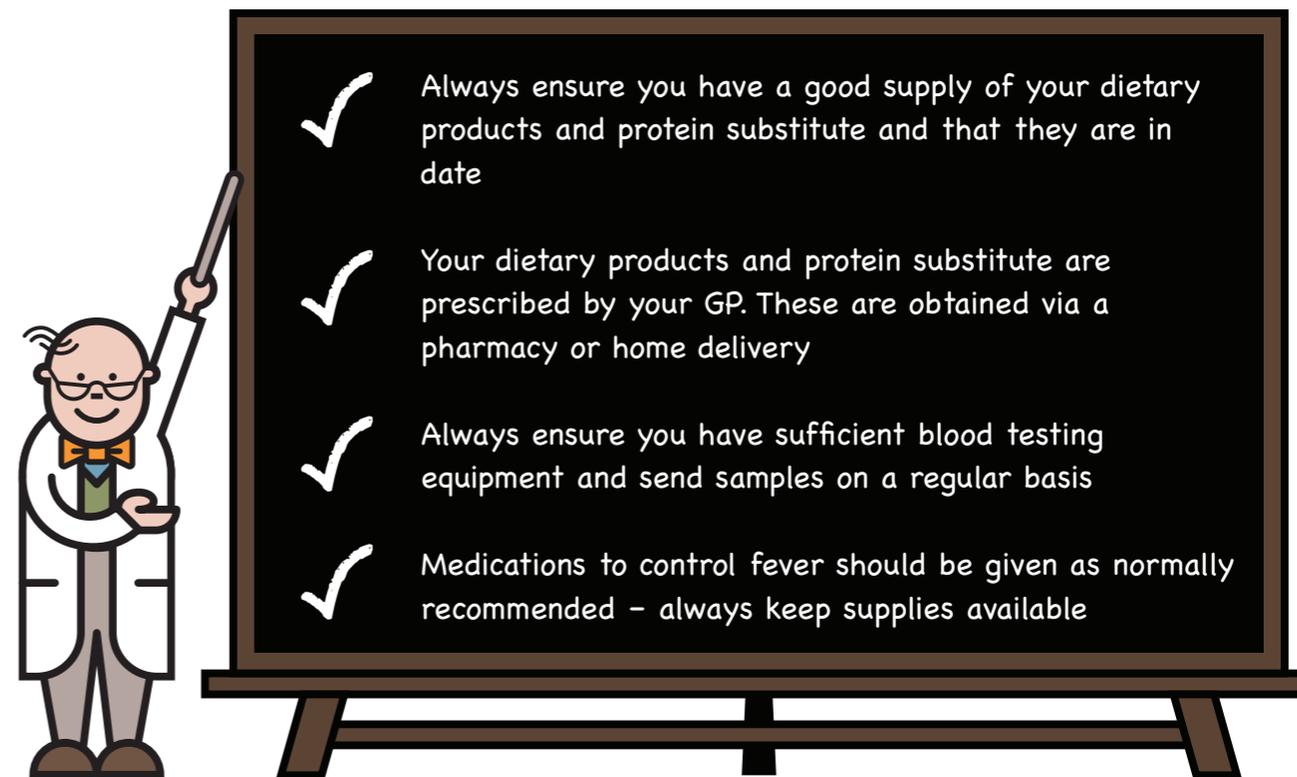
## Future pregnancies



## Take home messages



## Helpful hints





# Notes

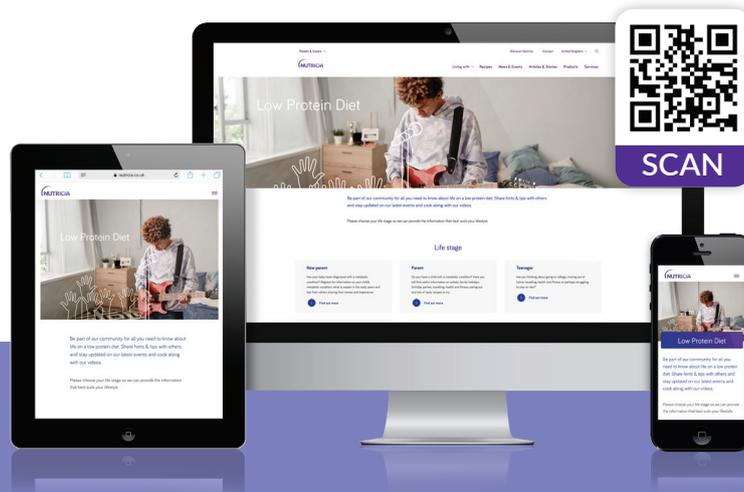
A series of 12 horizontal dotted lines for writing notes.

# Notes

A series of 12 horizontal dotted lines for writing notes.

Visit [www.nutricia.co.uk/patients-carers/living-with/low-protein-diet.html](http://www.nutricia.co.uk/patients-carers/living-with/low-protein-diet.html) 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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