# Help and support for families living with PKU

NSPKU

The National Society for Phenylketonuria (United Kingdom) Limited

The National Society for Phenylketonuria (NSPKU) is a registered charity. It offers support to people with PKU and their families by producing various publications, including a quarterly newsletter, organising formal and informal meetings and conferences.

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# An introduction to Phenylketonuria (PKU)

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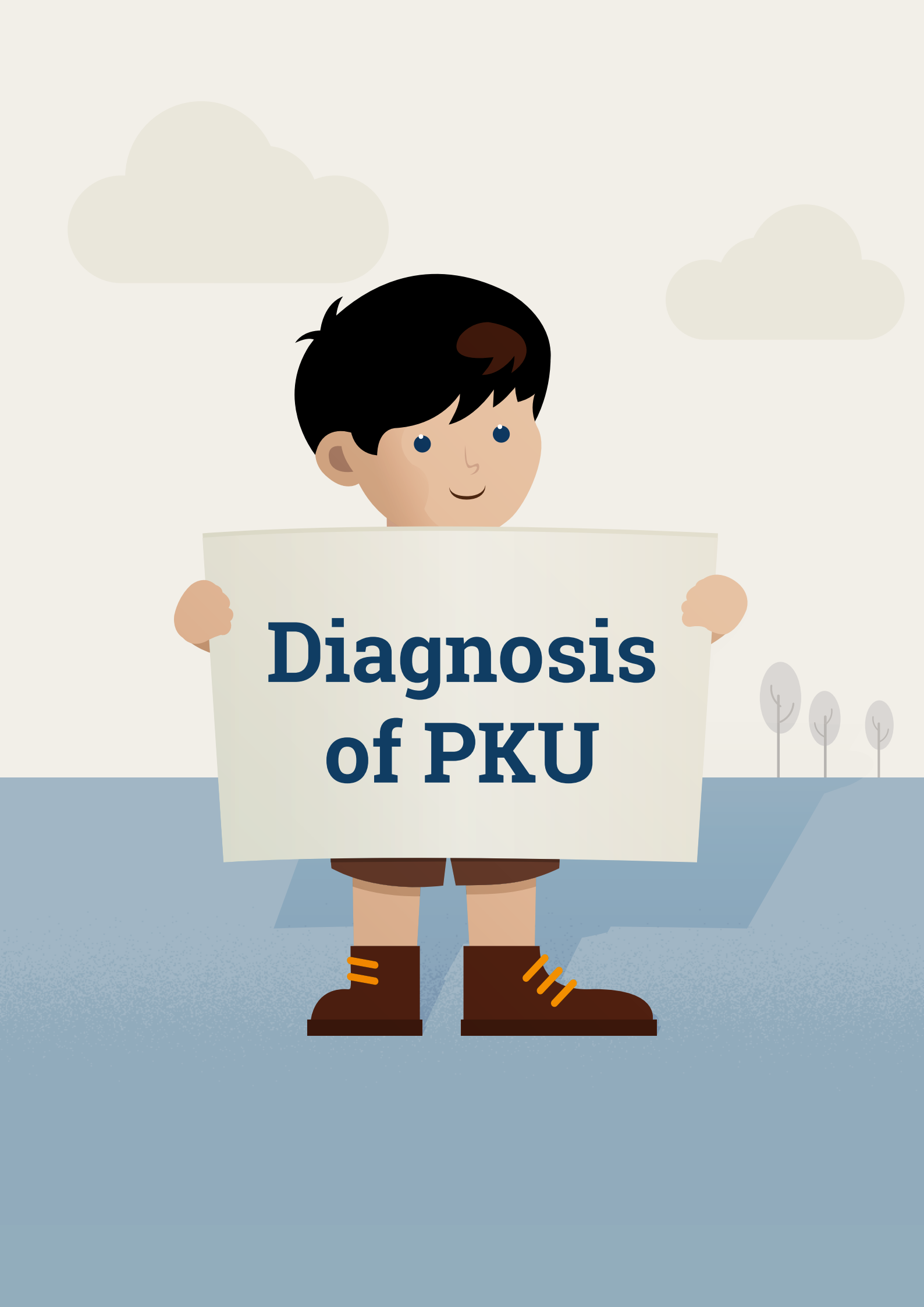
**Phenylketonuria**
(sounds like feen-il-ke-ton-u-re-ah)
**or PKU for short**

# PKU is a manageable condition which affects the way the body uses protein.

- Children with PKU cannot use a part of protein called Phenylalanine (Phe).
- If left untreated, Phe builds up in the body and causes brain damage.
- This will be avoided by following a special diet.
- With proper management, people with PKU have every chance of attending university, building a successful career and enjoying a happy family life as everyone else.

Finding PKU early by screening, special dietary management and careful monitoring means that a child is able to reach their full potential.


Diagnosis
of PKU

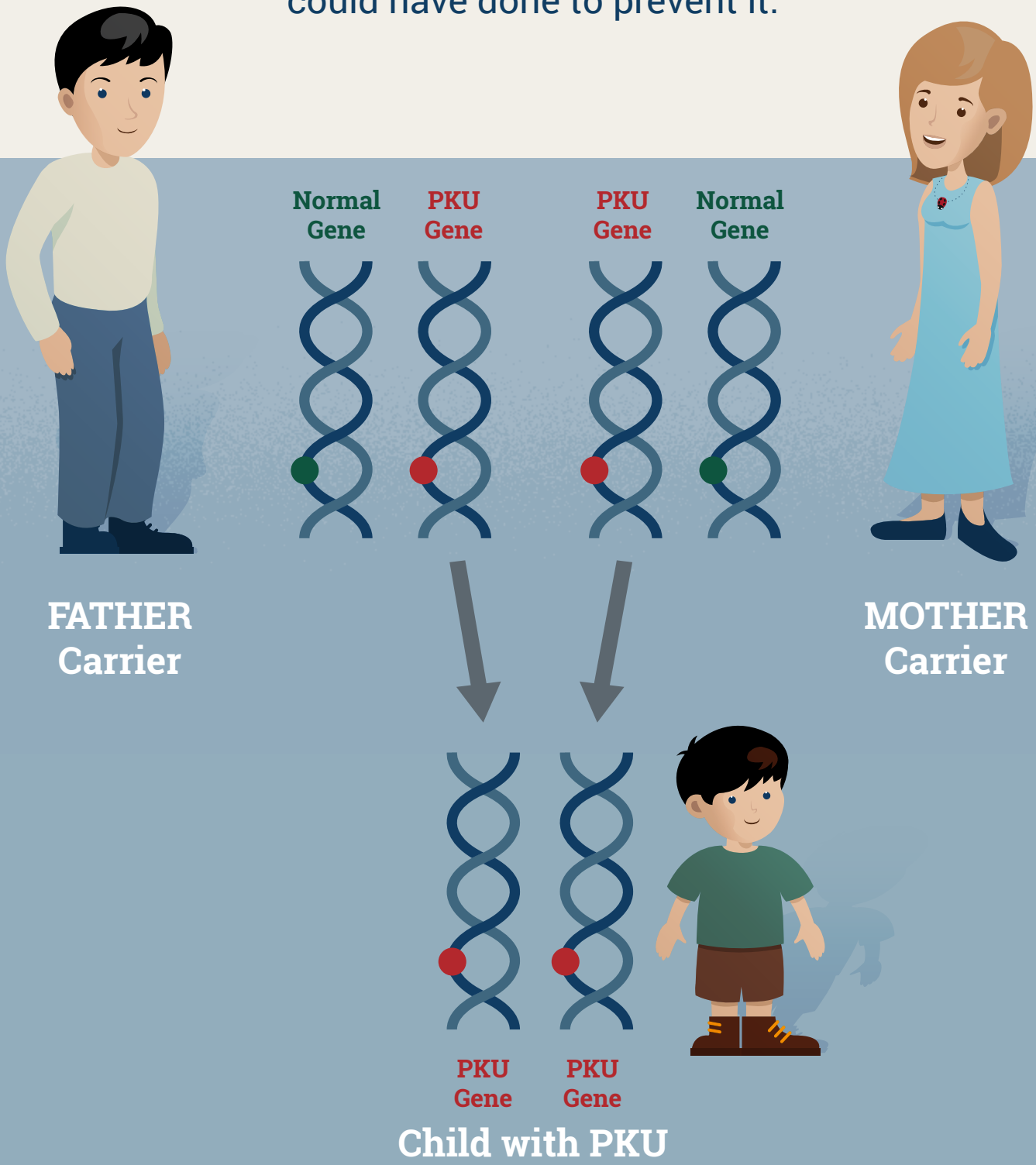
# PKU is diagnosed by a blood test called “the heel prick test”

This blood test is carried out by the midwife around 5 days after birth. Under the national newborn screening programme every baby in the UK and Ireland is screened for this condition. PKU is then picked up before any harm is caused.


Why does my baby have PKU?

# PKU is an inherited condition

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

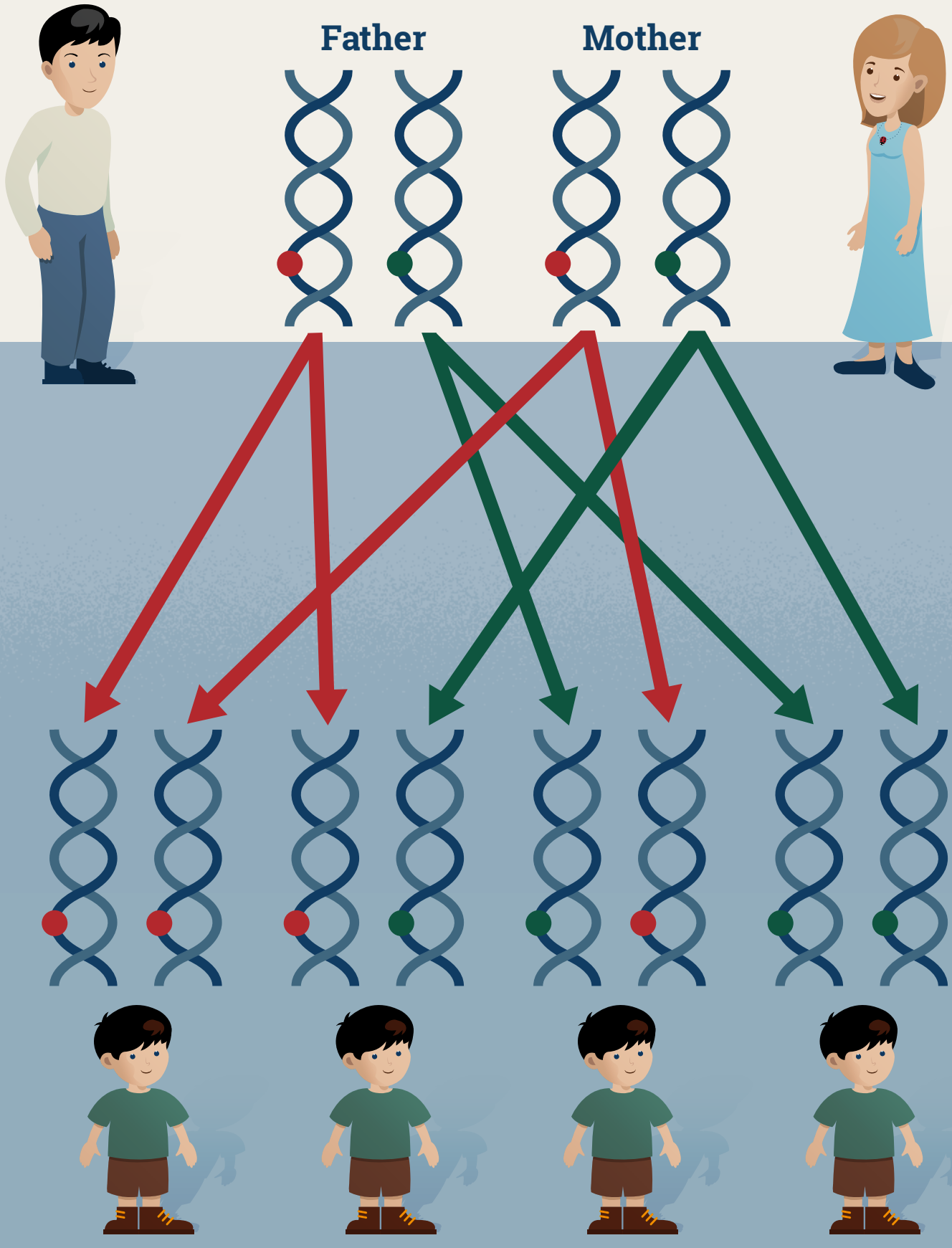



As a parent of a child with PKU, you have one PKU gene and one normal one. This is known as being a carrier.

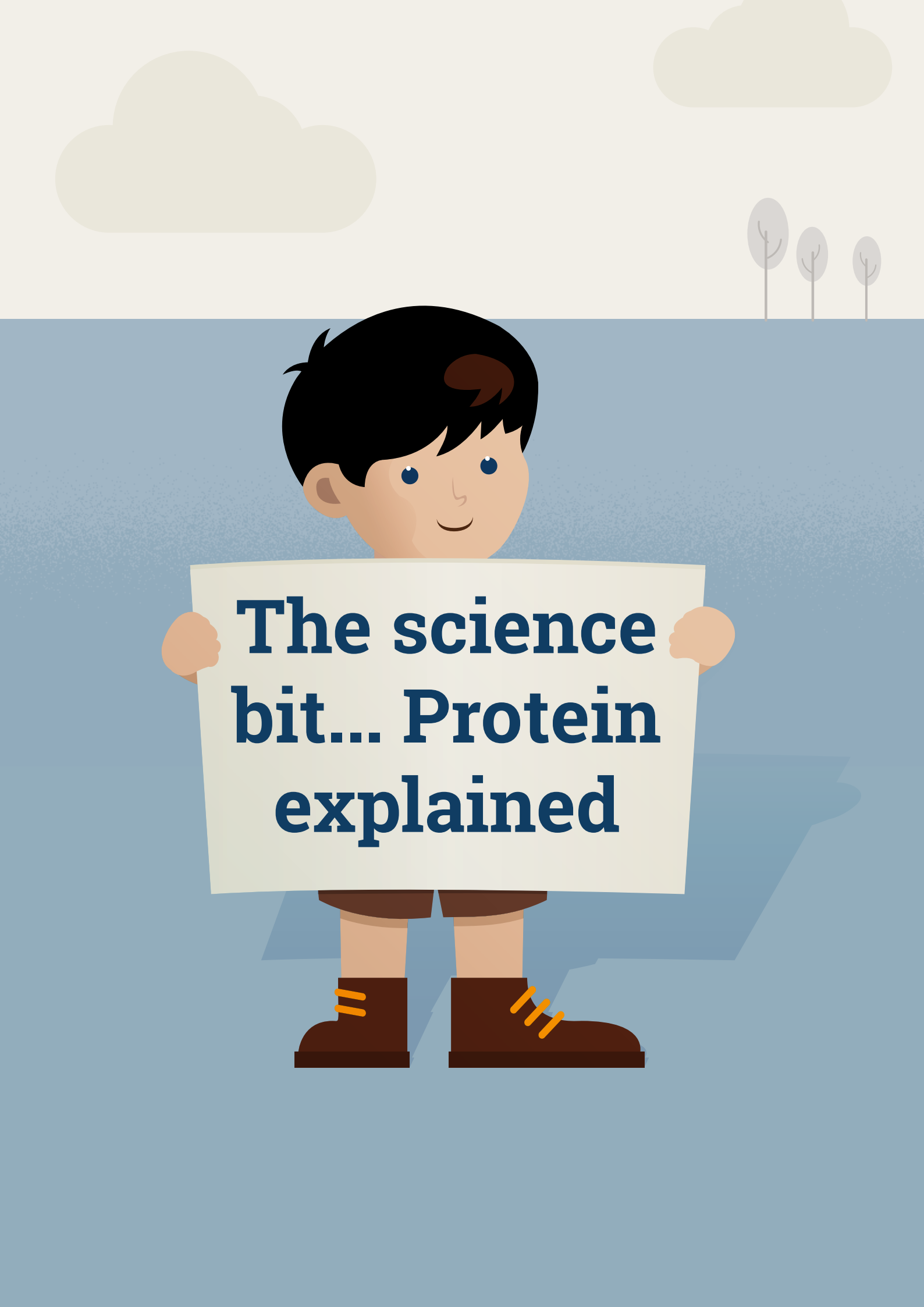
Your child has inherited 2 PKU genes, one from mum and one from dad.

# Around 1 in 50 people are carriers of the PKU gene

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



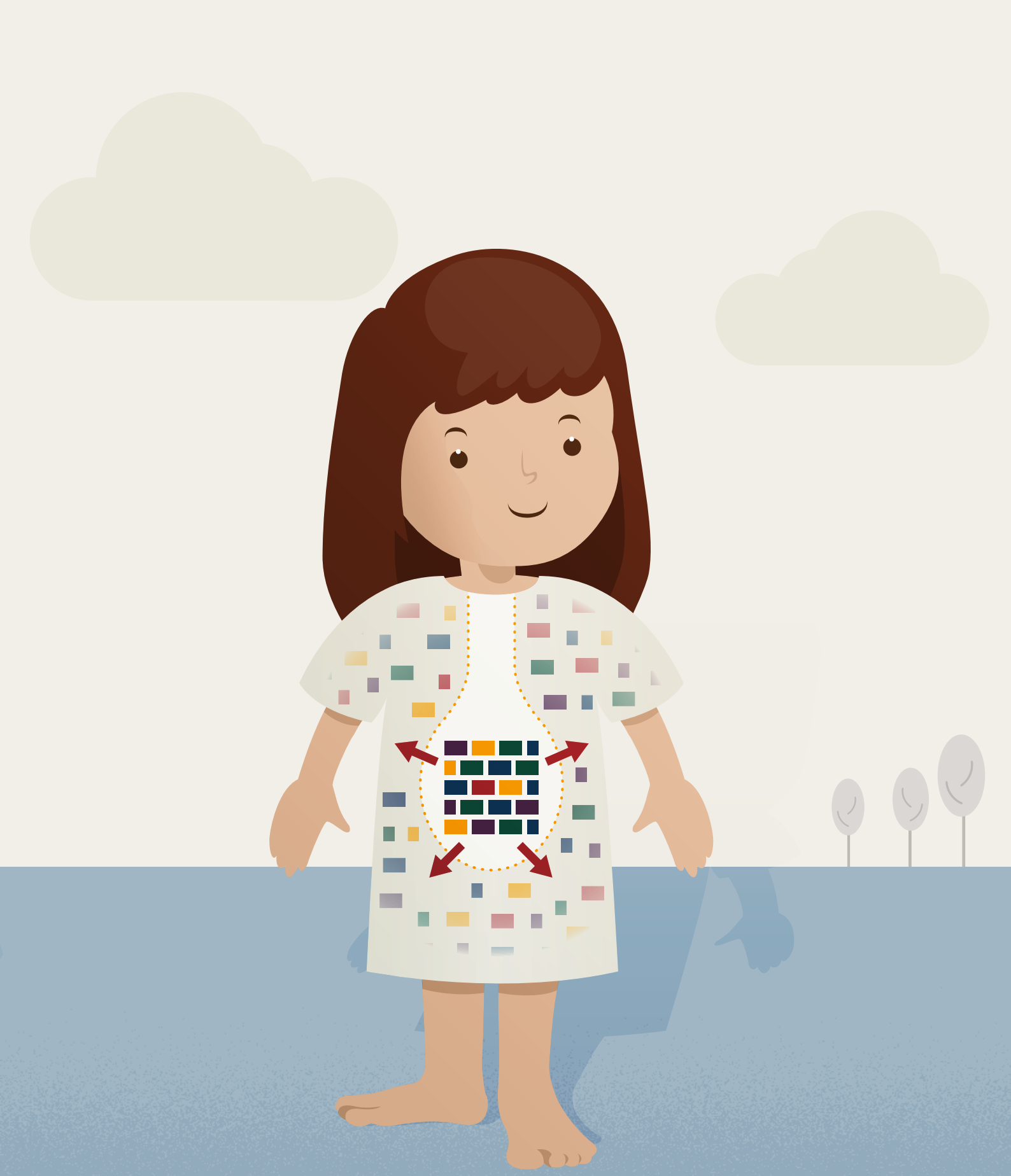

When 2 people who are carriers of the PKU gene conceive a child there is a 1 in 4 chance of that child having PKU.


The science bit... Protein explained

# Why do we need protein?

Protein is a nutrient needed by the body.

It helps to build, repair and maintain body cells and tissues, like your skin, muscles, organs, blood and even bones.



When protein is eaten, it is broken down in the body (during digestion) into smaller pieces (like building blocks).

These smaller pieces are called amino acids.

# Protein is made up of many building blocks called amino acids


There are 20 amino acids (building blocks) that make up protein.

Some of these are ESSENTIAL and cannot be made in the body - so they must come from the food we eat.

Phenylalanine (Phe) is one of these essential amino acids. It is this particular amino acid that a person with PKU cannot process correctly.

# What normally happens

## The conversion of protein into amino acids

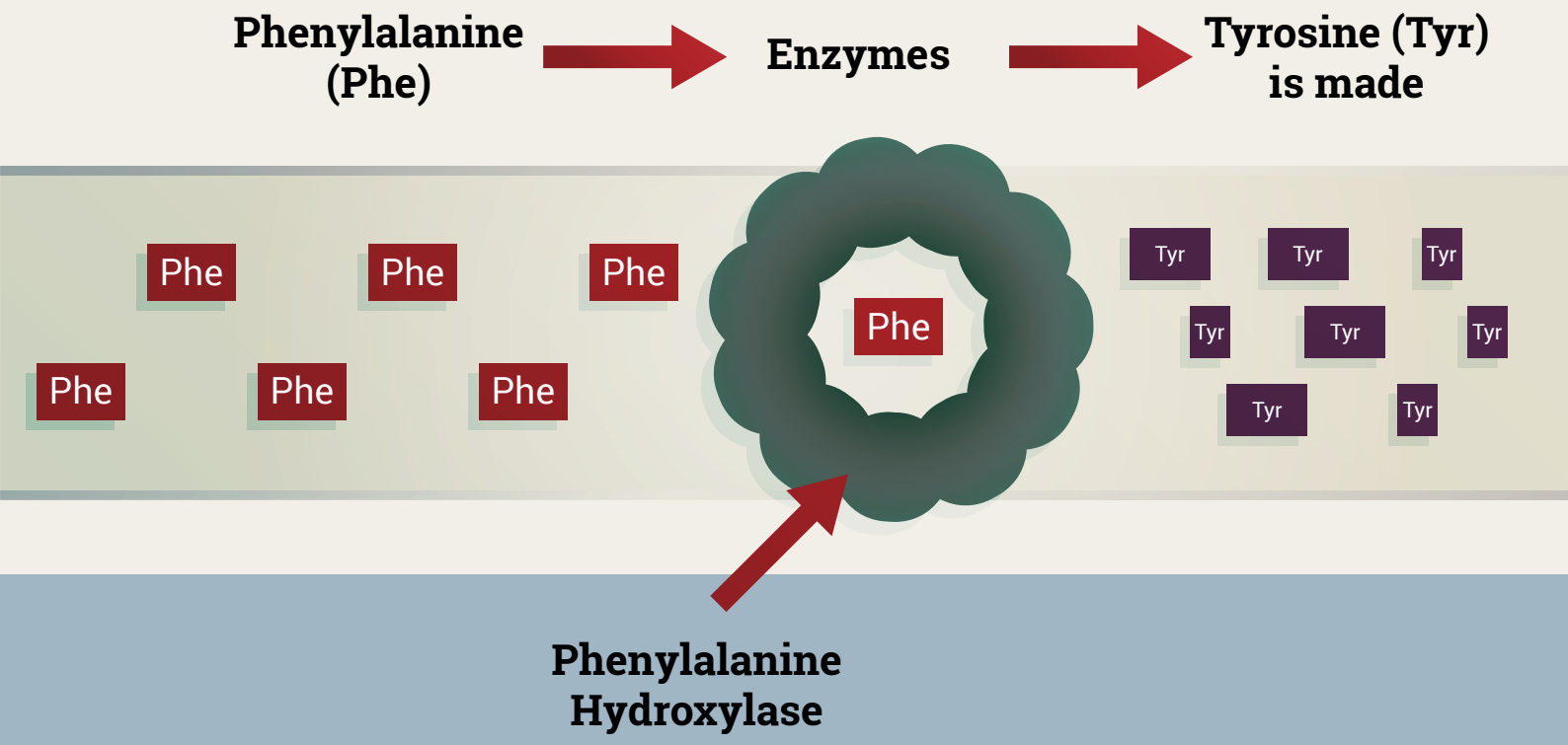
**Protein** → **Enzymes** → **Amino acids (Building Blocks)**

Enzymes are like chemical scissors needed for the breakdown of protein into amino acids.

The body uses these amino acids for growing, building up muscle and helping the body stay healthy.


Why can someone with PKU not process Phe correctly?

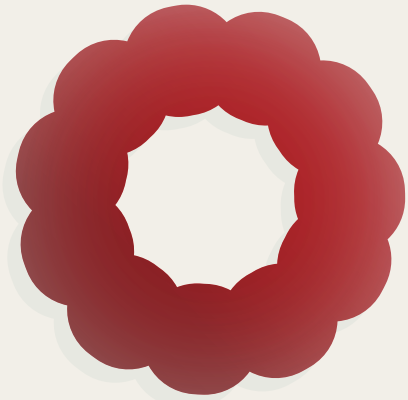
# In a person without PKU, Phenylalanine (Phe) is converted into another amino acid called Tyrosine (Tyr)



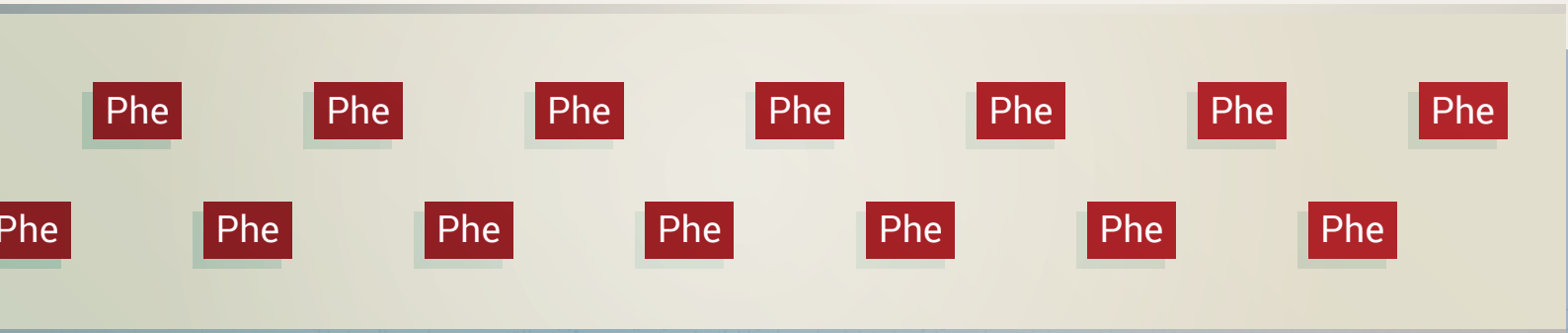

Normally, the liver produces an enzyme called Phenylalanine Hydroxylase. This enzyme breaks down Phenylalanine (Phe) to another chemical called Tyrosine (Tyr).

Tyrosine is important for many functions in the body.

# In a person with PKU, the conversion of Phenylalanine (Phe) into Tyrosine (Tyr) does not happen as it should



**Phenylalanine (Phe)** → **Faulty/No Enzyme** → **Little/No Tyrosine (Tyr) is made. This means there is a build up of Phe**

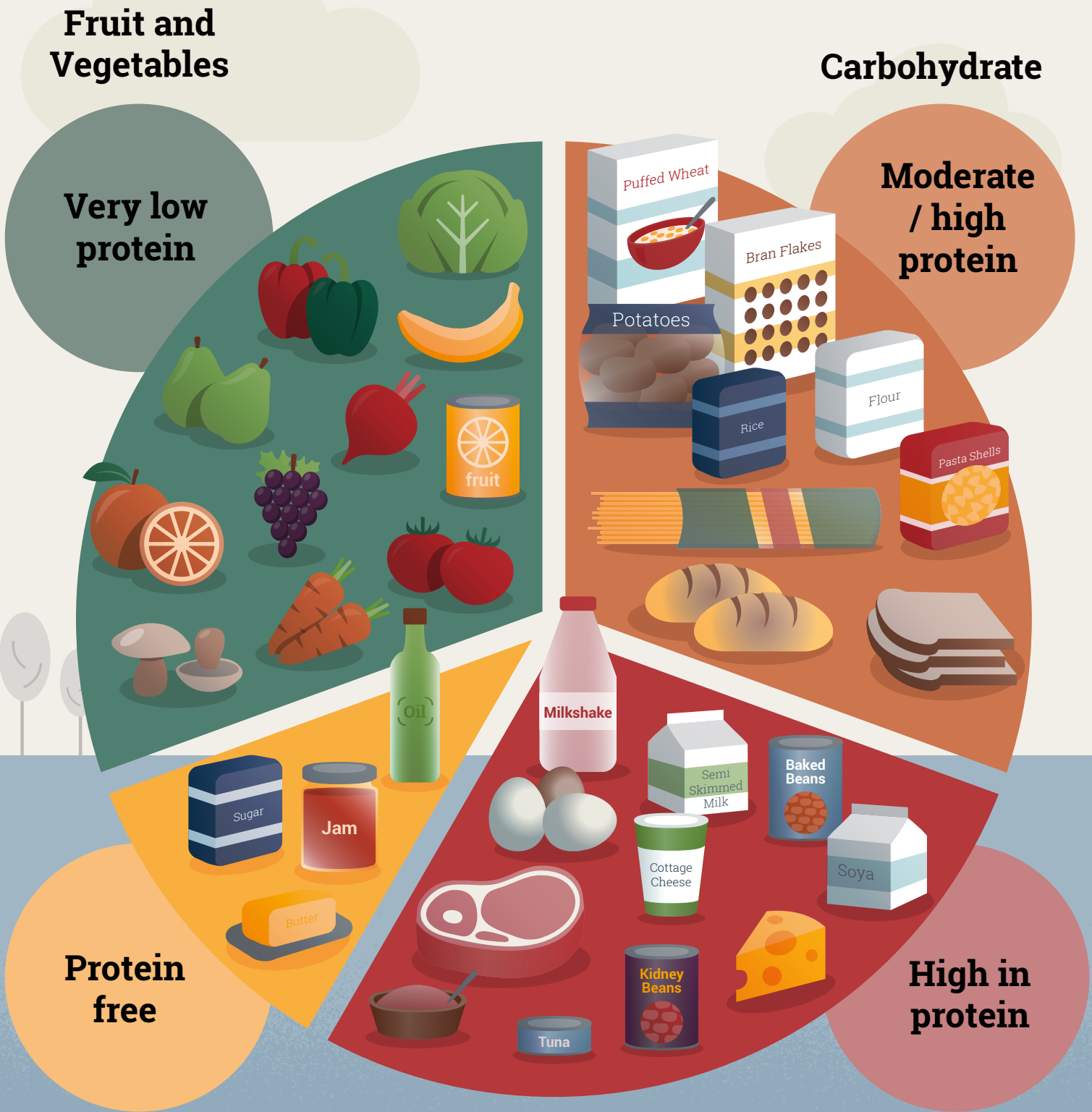



In PKU, the enzyme phenylalanine hydroxylase is not made by the liver or the enzyme does not work properly. This means that Phe is not converted to the amino acid Tyrosine and builds up in the blood. This is the reason a special diet low in protein / Phe needs to be followed.

Low levels of Tyrosine also cause problems. Extra Tyrosine is therefore needed in the diet.


Management of PKU... the diet explained

# A Regular Diet



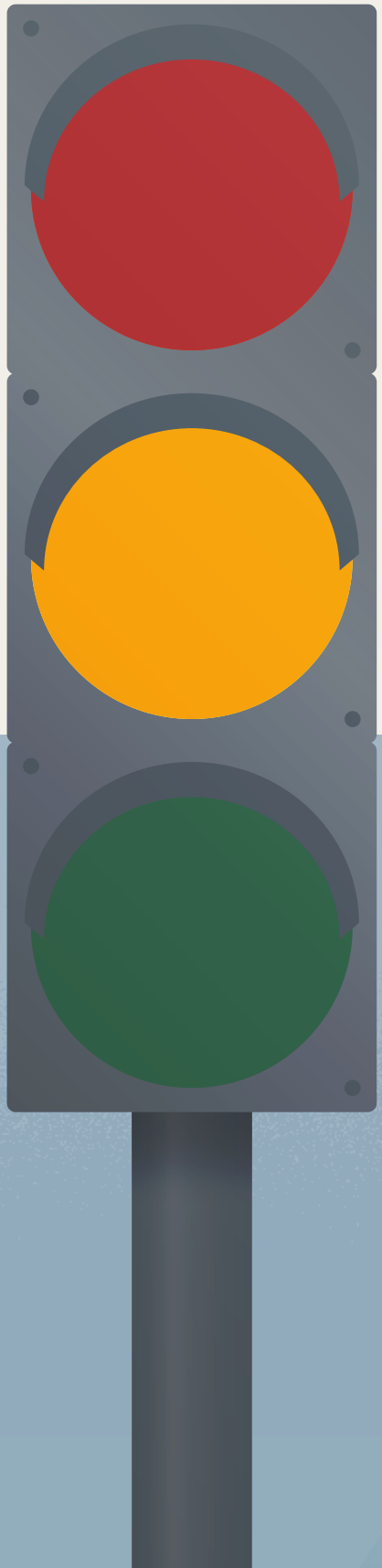

A regular diet has too much protein / Phe for a person with PKU. Therefore they must follow a special diet.

# Management of PKU is by following a low protein / Phe diet

- Phenylalanine (Phe) is found in all protein containing foods, e.g. meat.
- In order to manage PKU, all protein foods **must be restricted** - and a low protein / Phe diet must be followed.
  - All amino acids have a role or function in the body. Phe helps us with our growth, development and tissue repair.
  - It is important that someone with PKU gets some Phe, but not so much that it becomes harmful.

# Traffic Light System

The low protein / Phe diet a person with PKU must follow is best described by using the traffic light system



## RED STOP!

Do not eat these foods

**Too high in protein / Phe**

## AMBER Go Cautiously!

These foods can be eaten
in measured amounts (exchanges)

**Contains some protein / Phe**

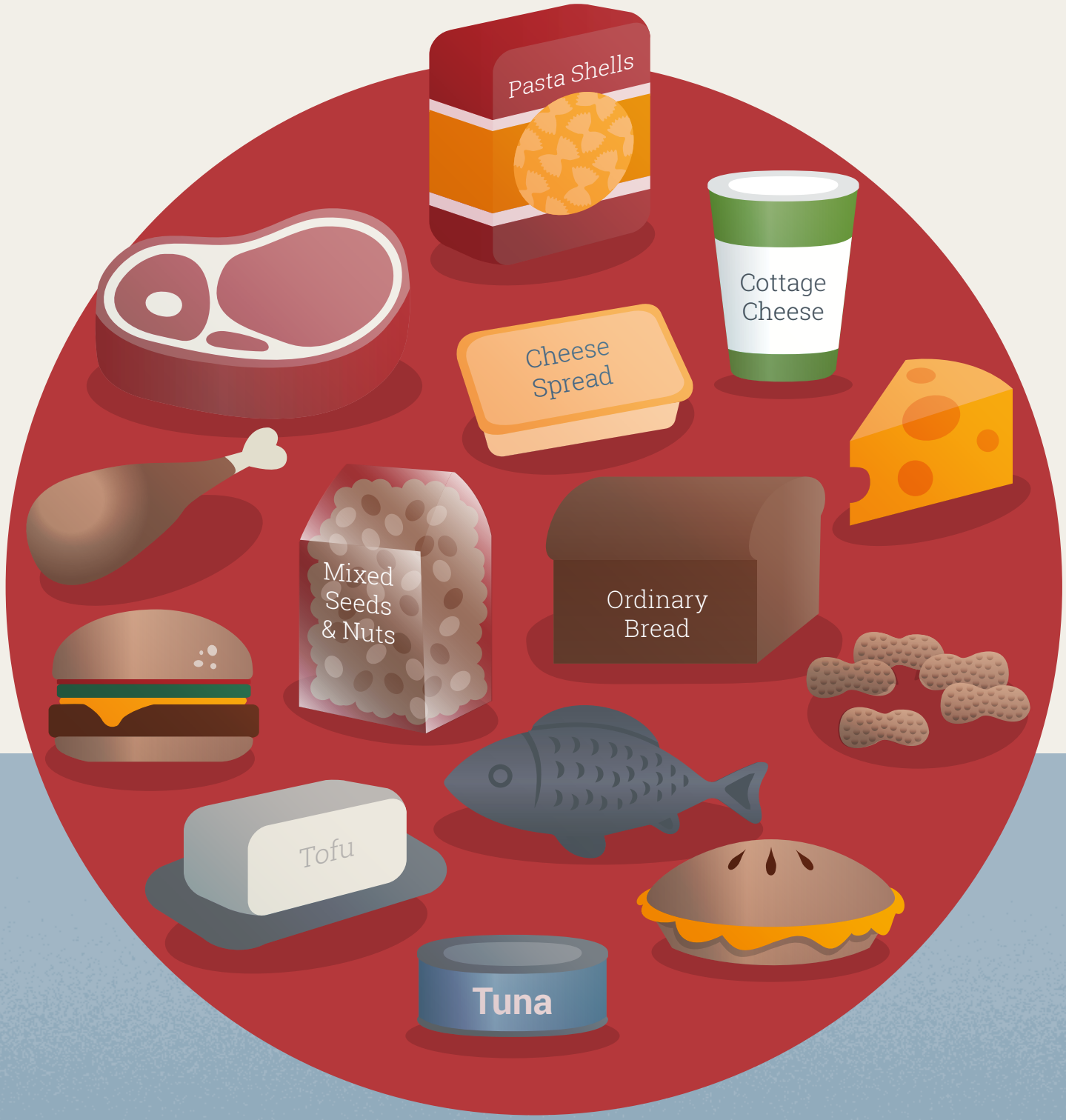
## GREEN Go!

These foods can be eaten freely

**Very low in / free from protein / Phe**

# RED STOP!

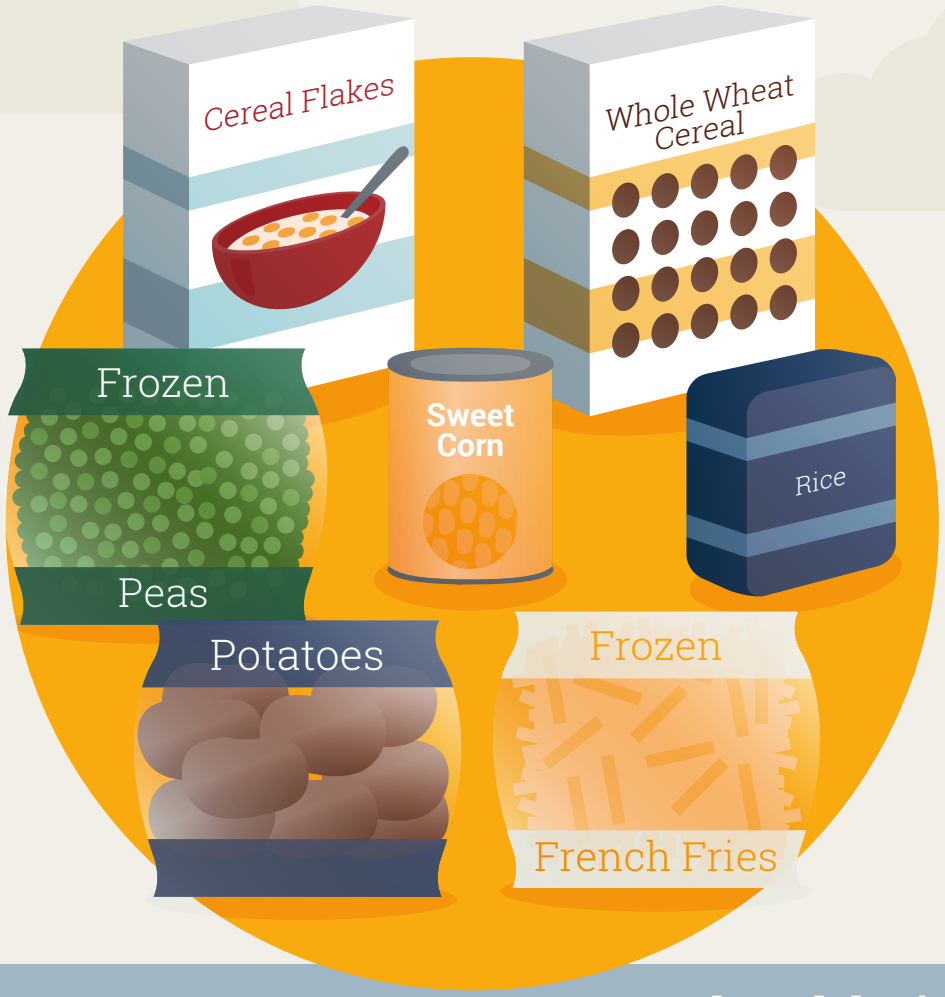
## Do not eat these foods

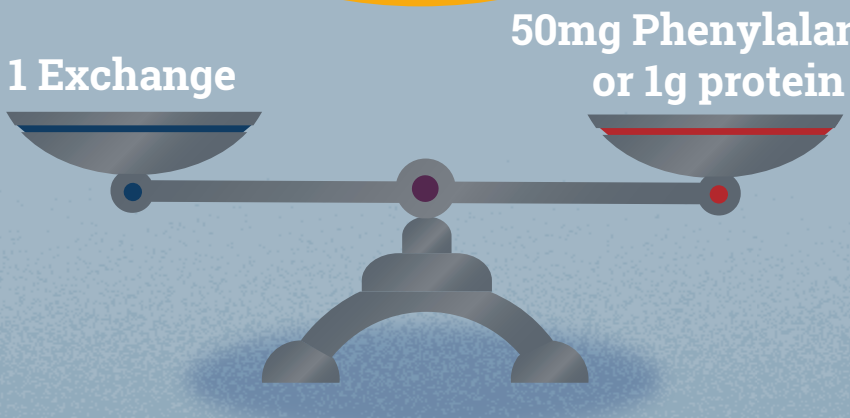



These foods are all rich in protein and therefore high in Phe so they are not allowed.

# AMBER Go Cautiously!

**These foods can be eaten in measured amounts and are known as exchanges**



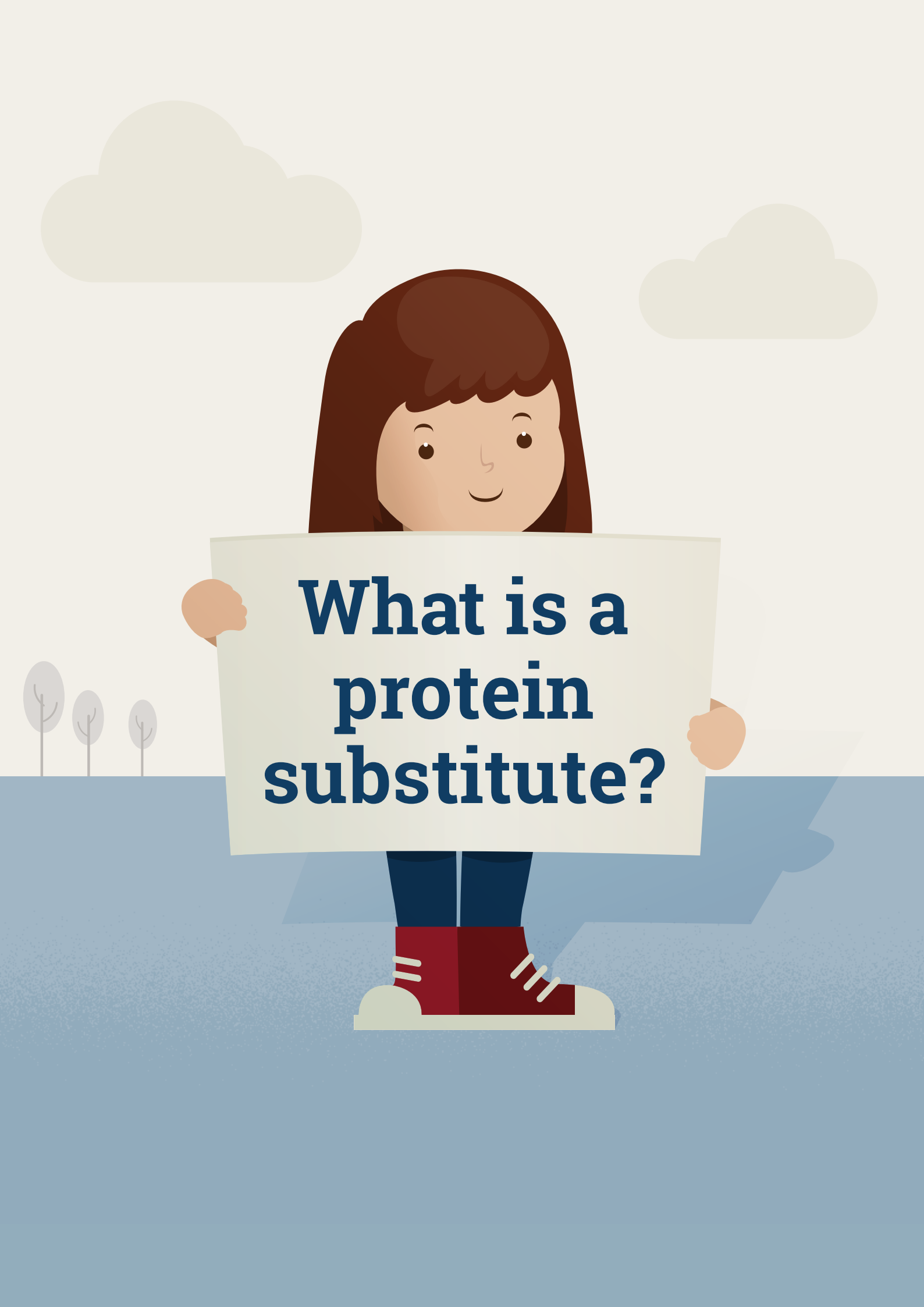

Other foods like these contain small amounts of protein. These foods can be eaten in measured amounts. The measure is called an exchange.

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

# GREEN Go!

## These foods can be eaten freely


These foods are either naturally low in protein or have been specially made to be low in protein and so are “FREE from exchanges” or do not have to be measured.


What is a protein substitute?

# Why does a person with PKU need to take a protein substitute?

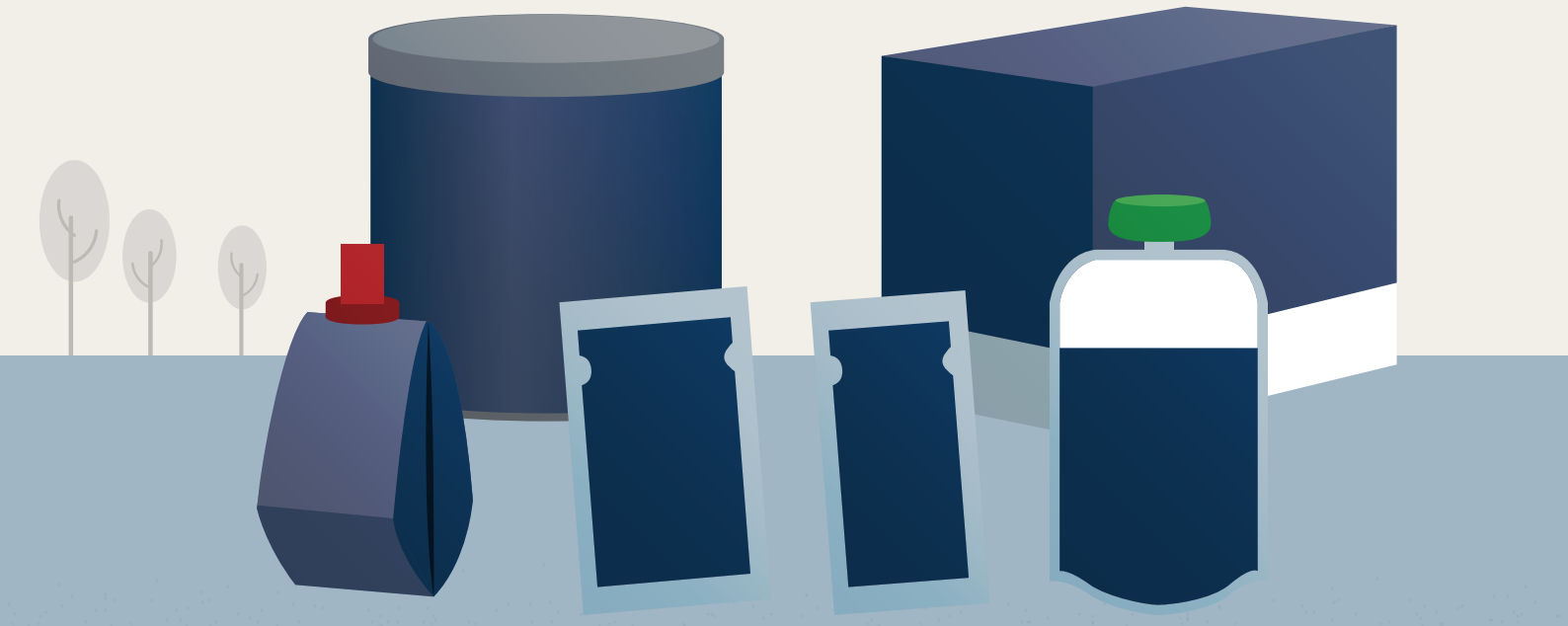
- A protein substitute is a specially made medical supplement, which can come in various formats like a drink, a gel or tablets.
- A protein substitute contains **NO** Phe... But it contains all of the other amino acids in protein that your child needs.
- Most protein substitutes also contain vitamins, minerals and other important nutrients to ensure your child is getting what they need.

The protein substitute is a very important part of the diet for a person with PKU and it must be taken regularly and evenly spread over the day. This helps to keep the Phe levels steady through the day.

# Protein substitutes

Many protein substitutes are designed to fit into everyday life.

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




How will I feed my new born baby?

# Infants: breastfeeding


**Step 1:** Once your child is diagnosed, you will be seen by a dietitian, your baby will be given a special Phe-free formula until the levels of Phe fall back to normal levels (usually within a few days).

**Step 2:** Once the levels of Phe are under control, a small amount of Phe-free formula is given before a breastfeed to restrict the amount of breastmilk taken by the baby. Breastmilk provides limited but essential Phe for normal growth.

Breastfeeding offers many benefits for baby and mother, it is recommended you discuss this with your Health Care Professional.

# Infants: non-breastfeeding (Bottle feeding)


**Step 1:** Once diagnosed, you will be seen by a dietitian, your baby will be given a special Phe-free formula until the levels of Phe fall back to normal levels (usually within a few days).

**Step 2:** Once the levels of Phe are under control a small restricted amount of standard baby formula will be given along with the Phe free formula to provide the limited Phe essential for normal growth.


Weaning

# When to start weaning*

Your dietitian will advise you when to start solid foods, but it will be at the same time as babies without PKU. These will initially be Phe free foods or foods that have a very low protein / Phe content.

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

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

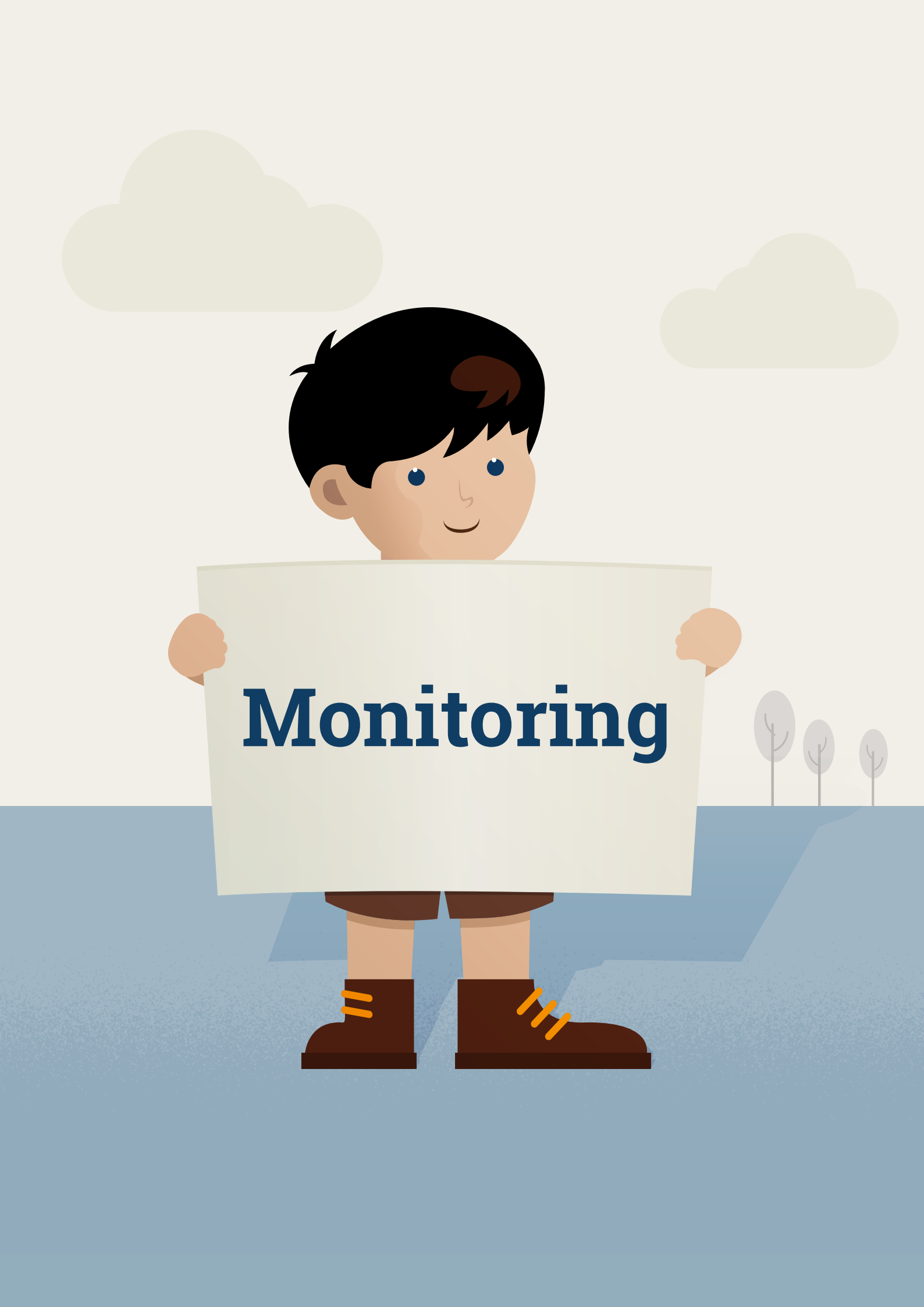
* Weaning may occur a little earlier than 6 months, depending on your baby but it should never be started before 17 weeks.

# 7 to 12 months

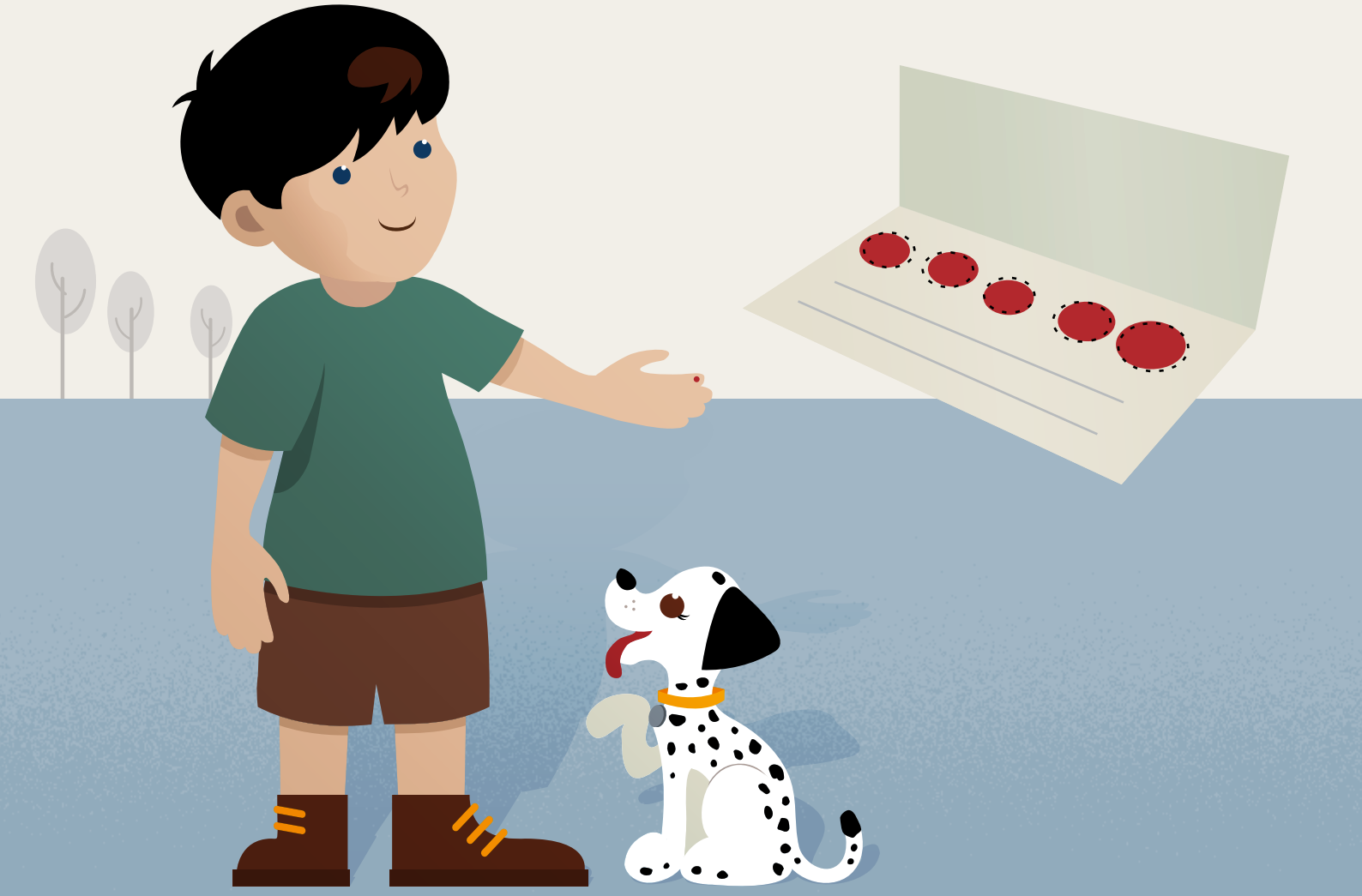
Your child will then be encouraged to move to more textured food and finger foods. Over time, they will get more of their Phe exchanges from food (e.g. potato or permitted cereal). Less exchanges will come from breast milk or formula milk.

Phe free foods should still be included in the diet as well. Your child will also continue their gradual switch from their infant Phe free formula to a second stage protein substitute.

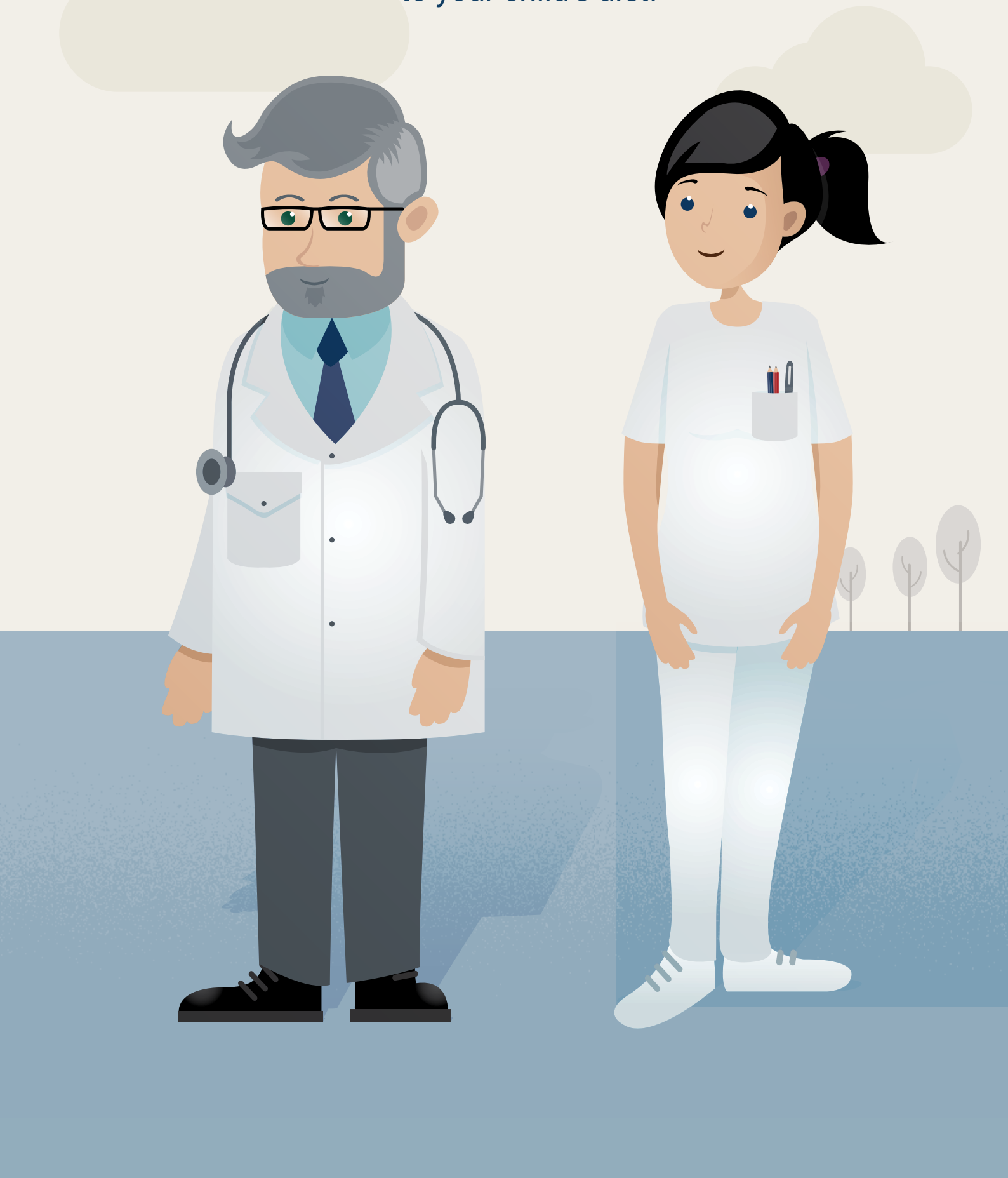
For more information on weaning in general, please consult your Health Care Professional.


Monitoring

Your child will have regular blood tests and monitoring to make sure their Phe levels are under control. This is very important to ensure that they do not have too little or too much Phe in their diet.



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




As your child grows up

# Children / Teenagers / Adults:

Your child will continue to follow a low protein / Phe 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.