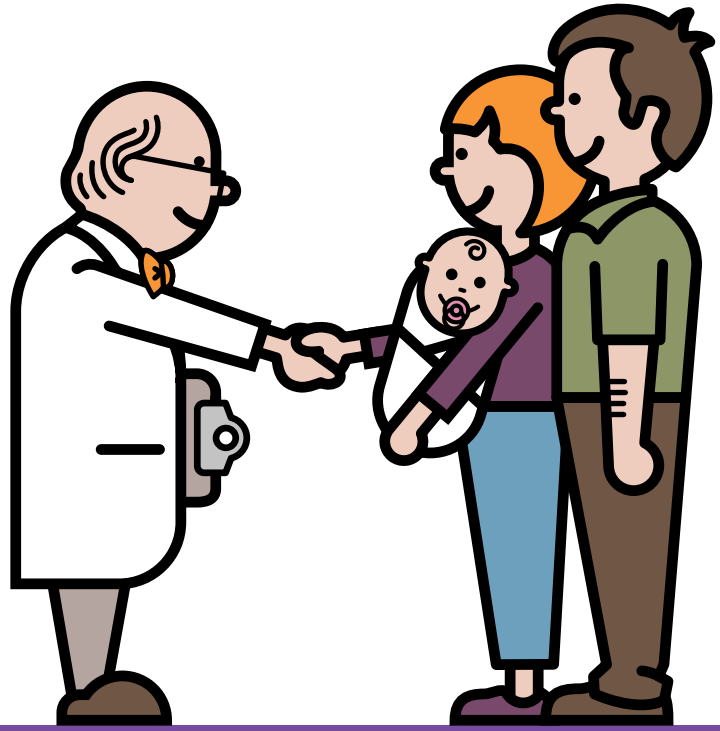
Phenylketonuria

PKU

Information for families after a positive newborn screening



Adapted by the Dietitians Group BIMDG

BIMDG

British Inherited Metabolic Diseases Group

BASED ON THE ORIGINAL TEMPLE WRITTEN BY BURGARD AND WENDEL

Reviewed & revised for North America by: A. Huber

This version of the TEMPLE tool, originally adapted by the Dietitians group of the BIMDG for use within the UK and Ireland, has been further adapted by Nutricia North America for use within United States and Canada. This version no longer represents clinical or dietetic practice in the UK or Ireland.

TEMPLE

Tools **E**nabling **M**etabolic **P**arents **LE**arning

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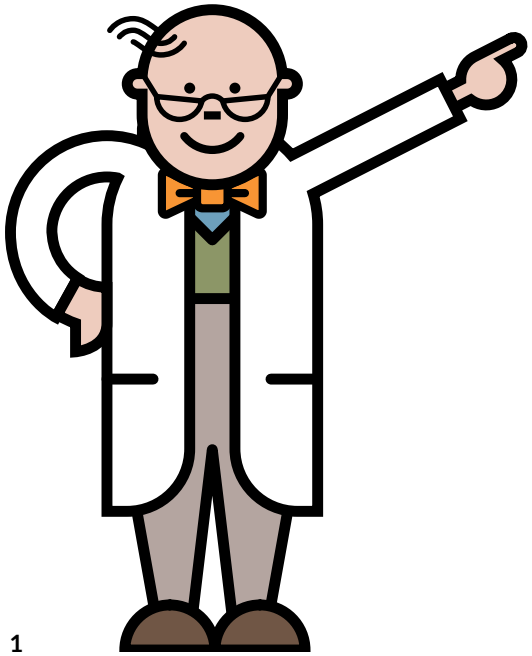
What is PKU?

PKU stands for Phenylketonuria.

It is pronounced fee-nile-keton-urea.

It is an inherited metabolic condition.

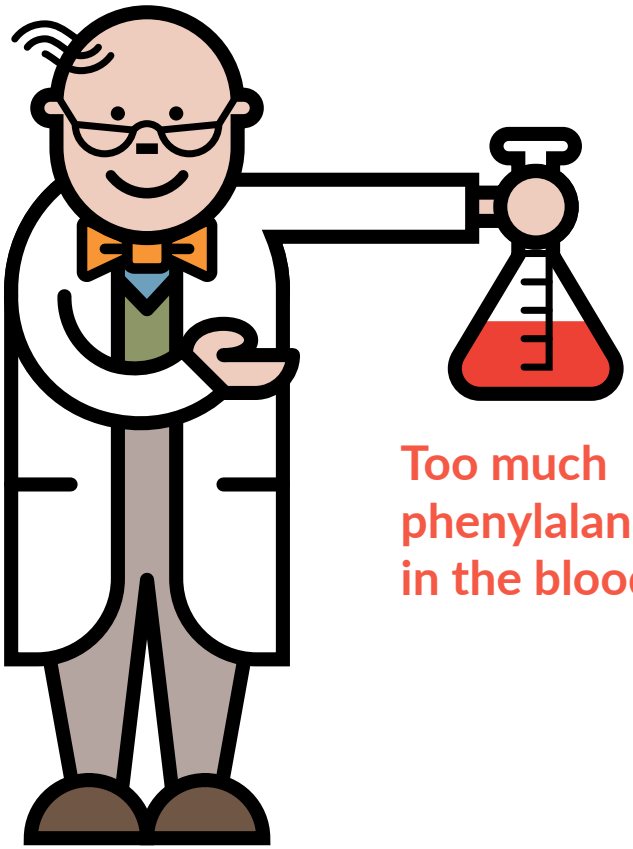


Phenyl keton uria

PKU

What is PKU?

Too many phenylketones in the urine



Too much phenylalanine in the blood

How does PKU affect the body?

PKU affects the way the body breaks down protein.

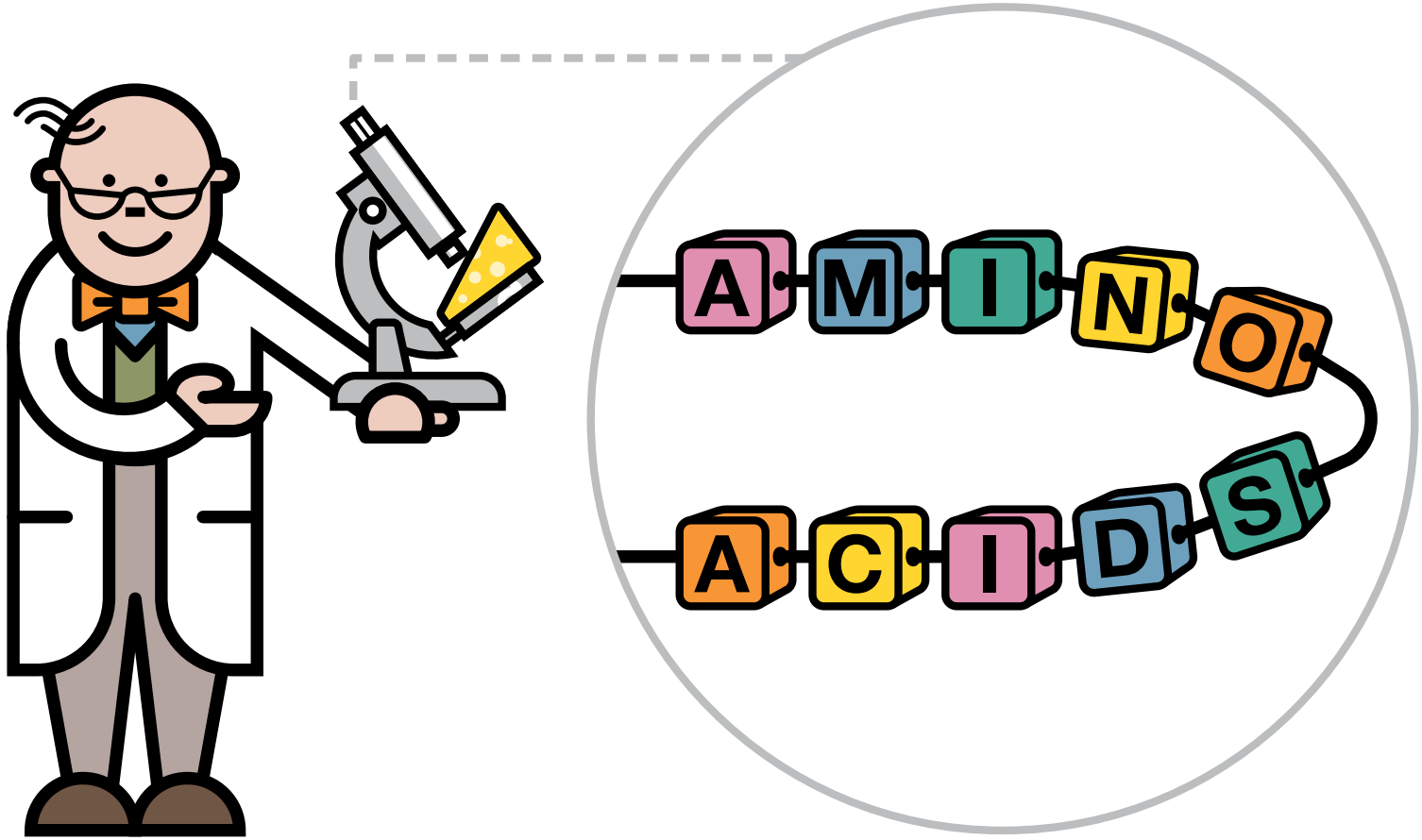
Protein is found in our bodies and in many foods. The body needs protein for growth and repair.





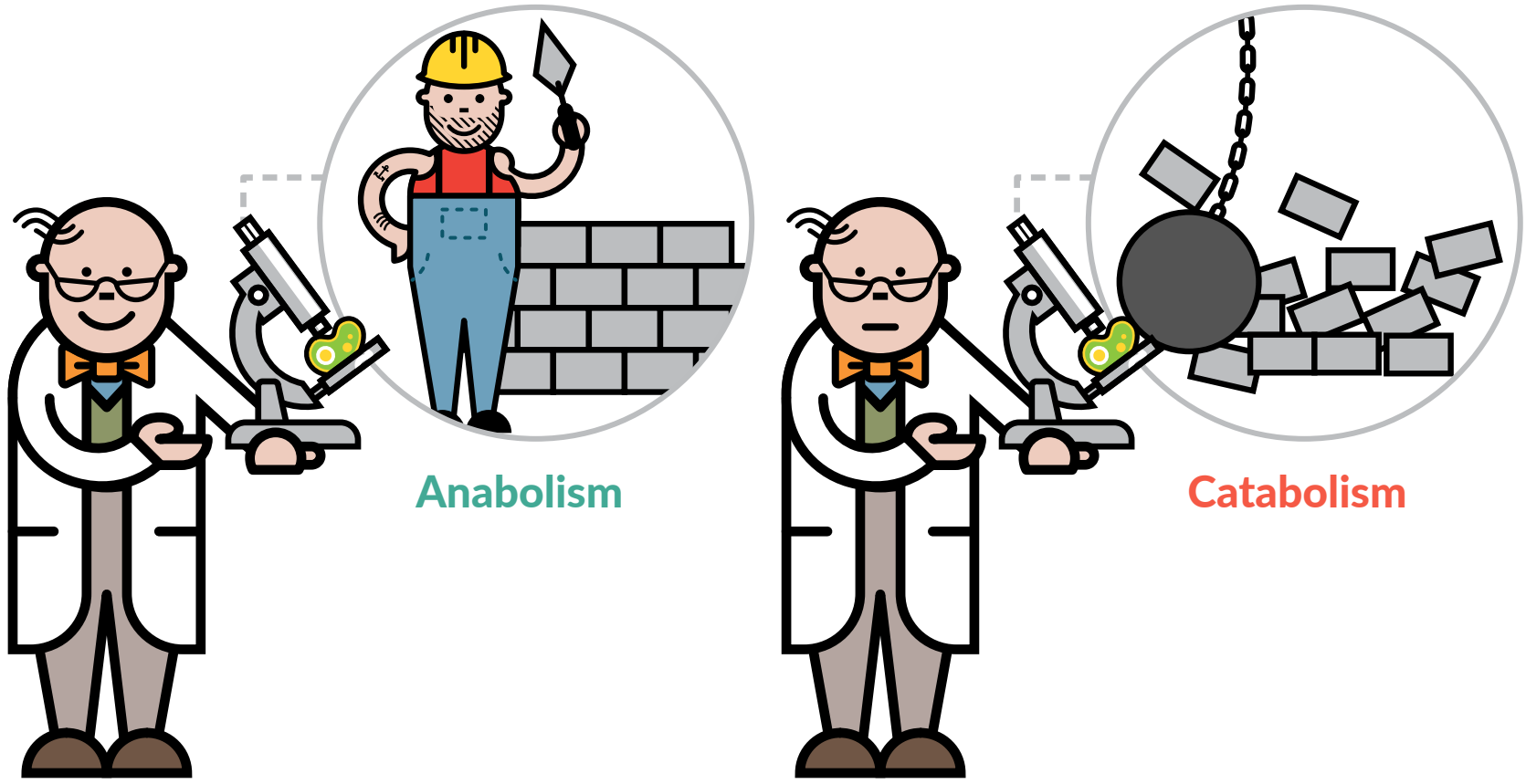

What is protein?

Protein consists of chains of many smaller units called amino acids.



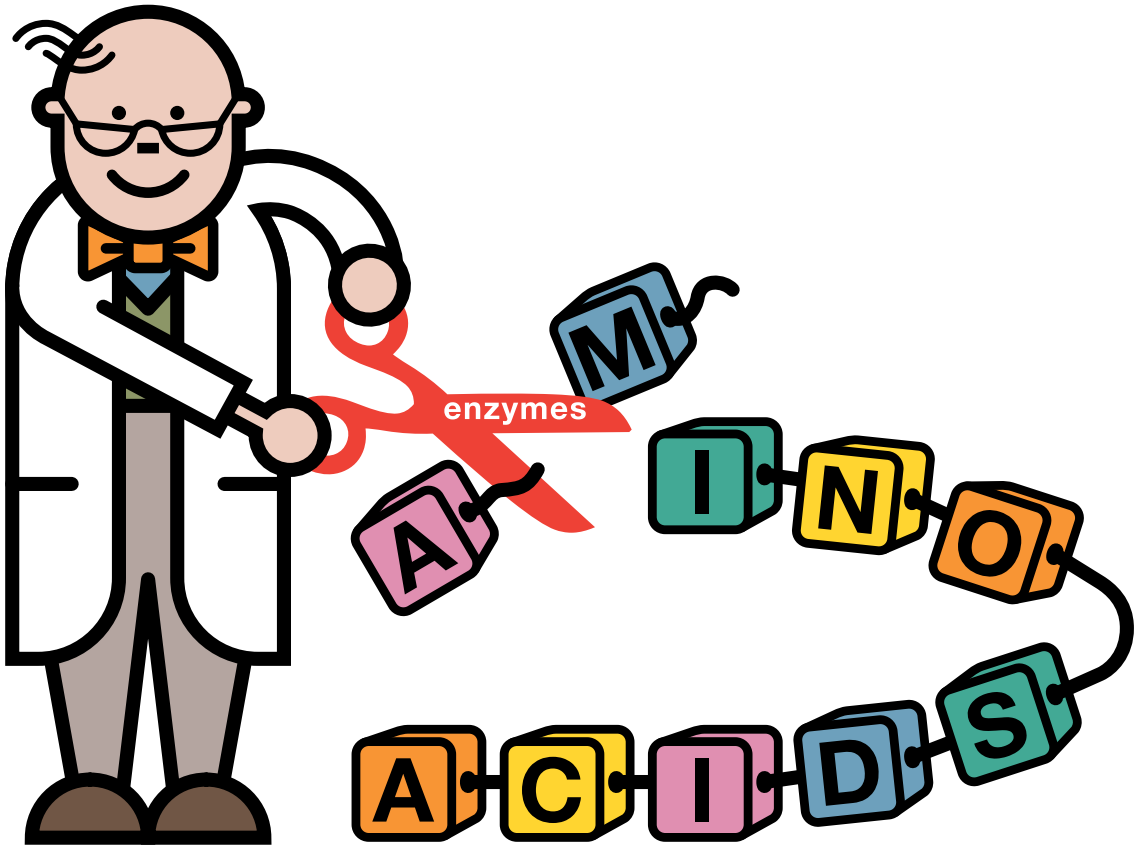
Protein metabolism

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



What do enzymes do?

Enzymes help with metabolism by functioning like scissors. They break down proteins into smaller parts, including amino acids.

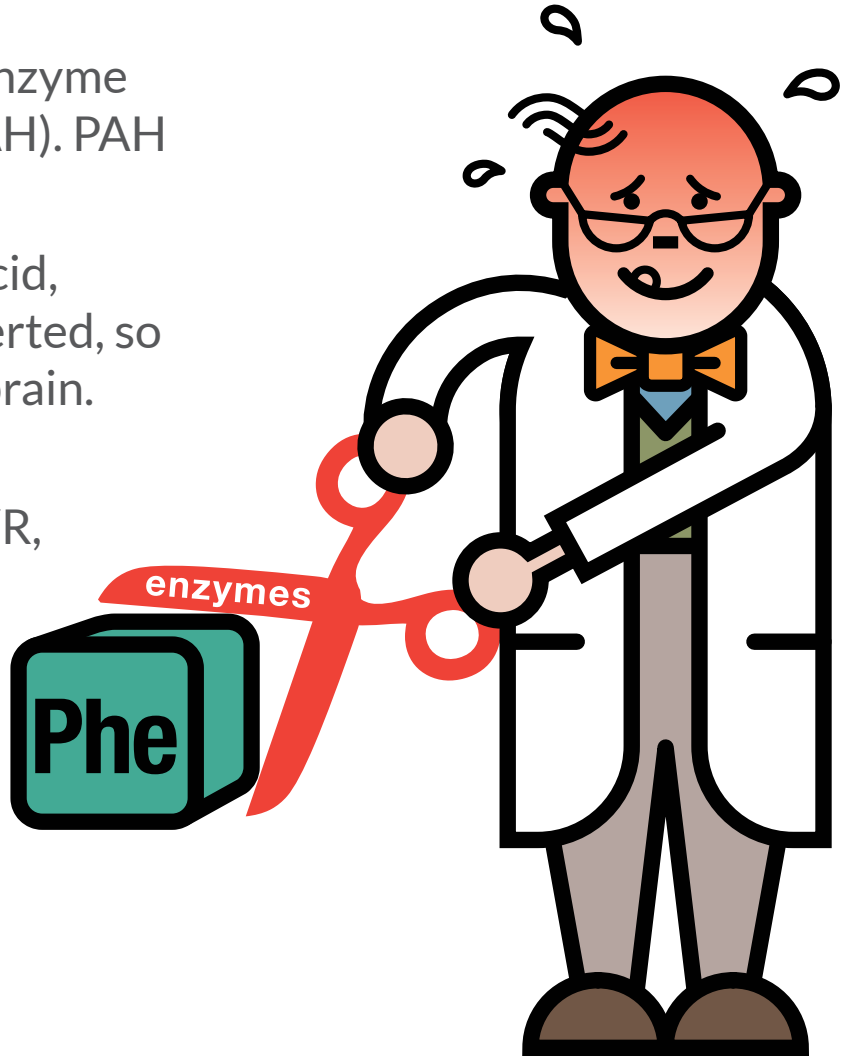


What happens in PKU?

PKU is caused by a deficiency of an enzyme called phenylalanine hydroxylase (PAH). PAH converts PHE into TYR.

The deficiency results in the amino acid, phenylalanine (PHE), not being converted, so instead it builds up in the blood and brain.

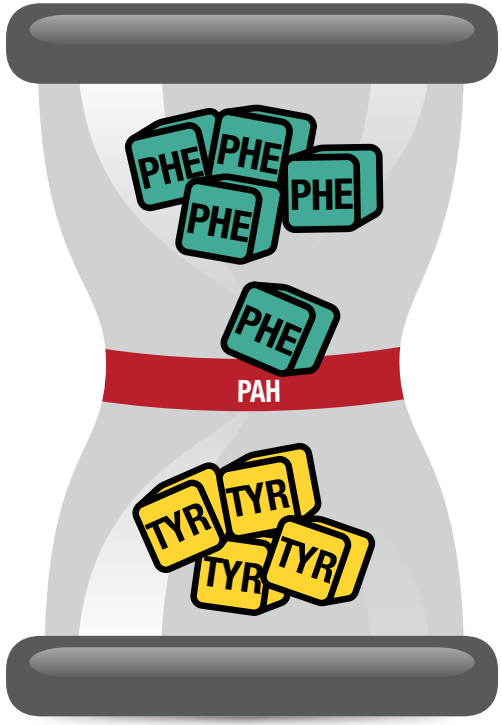
Blood tyrosine (TYR) levels are low as PHE cannot be broken down to TYR, and phenylketones are found in the urine.



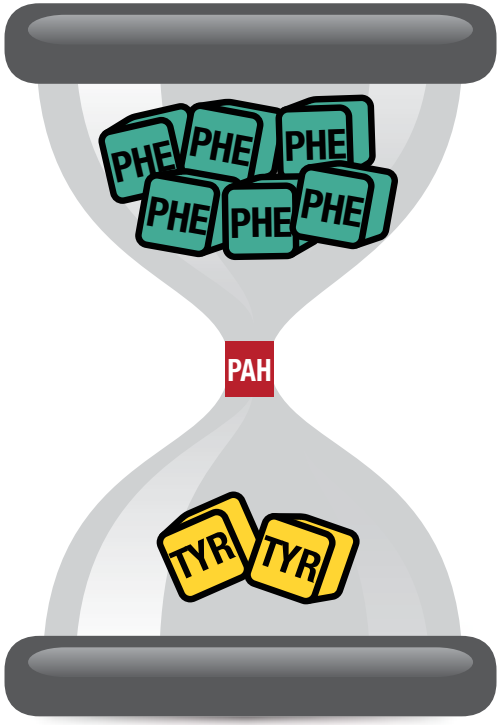
What happens in PKU?

For example:

For someone without PKU, PAH converts PHE into TYR



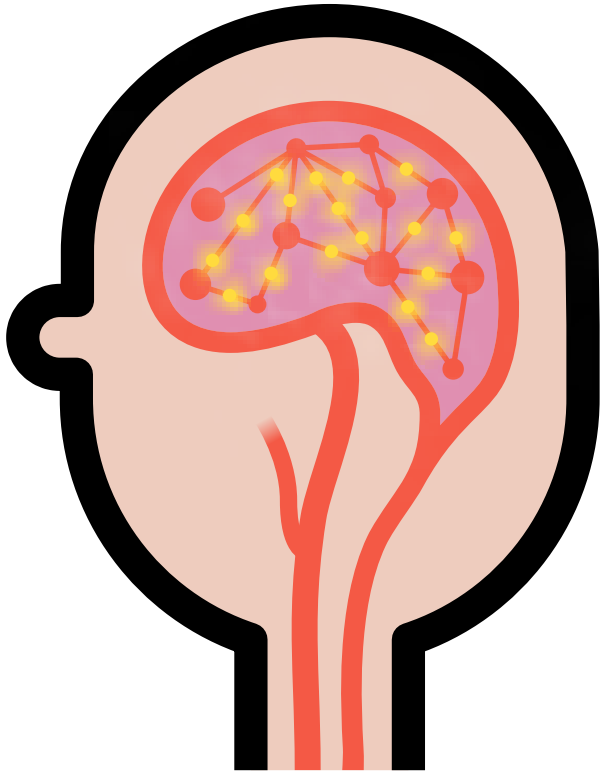
For someone with PKU, there is not enough PAH to convert PHE into TYR



What are the effects of high PHE levels?

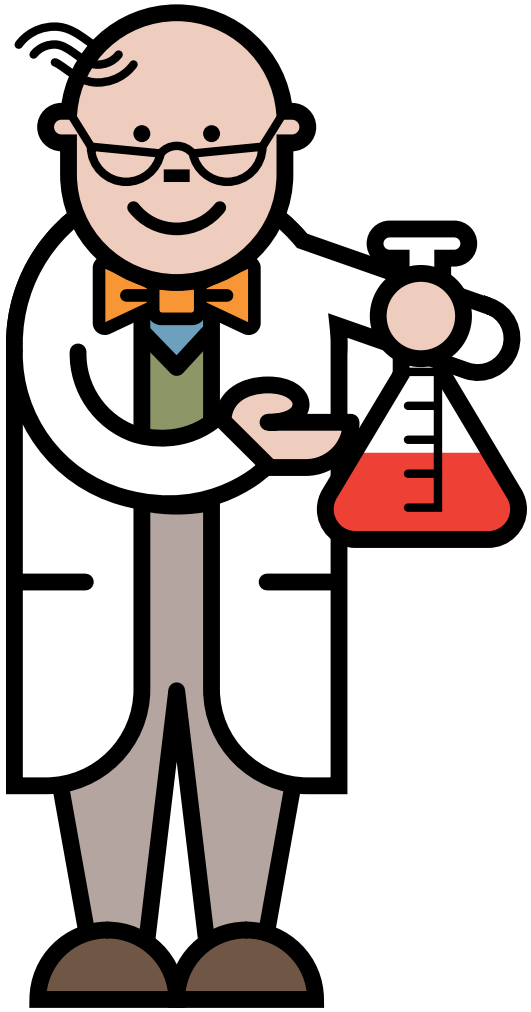
Without management, over time, the build-up of phenylalanine causes irreversible brain damage and poor intellectual development.

With management, the build-up of PHE is controlled to support normal growth and development.



What about other symptoms?

If blood levels of phenylalanine are poorly controlled, it is likely to lead to learning difficulties and behavior problems.



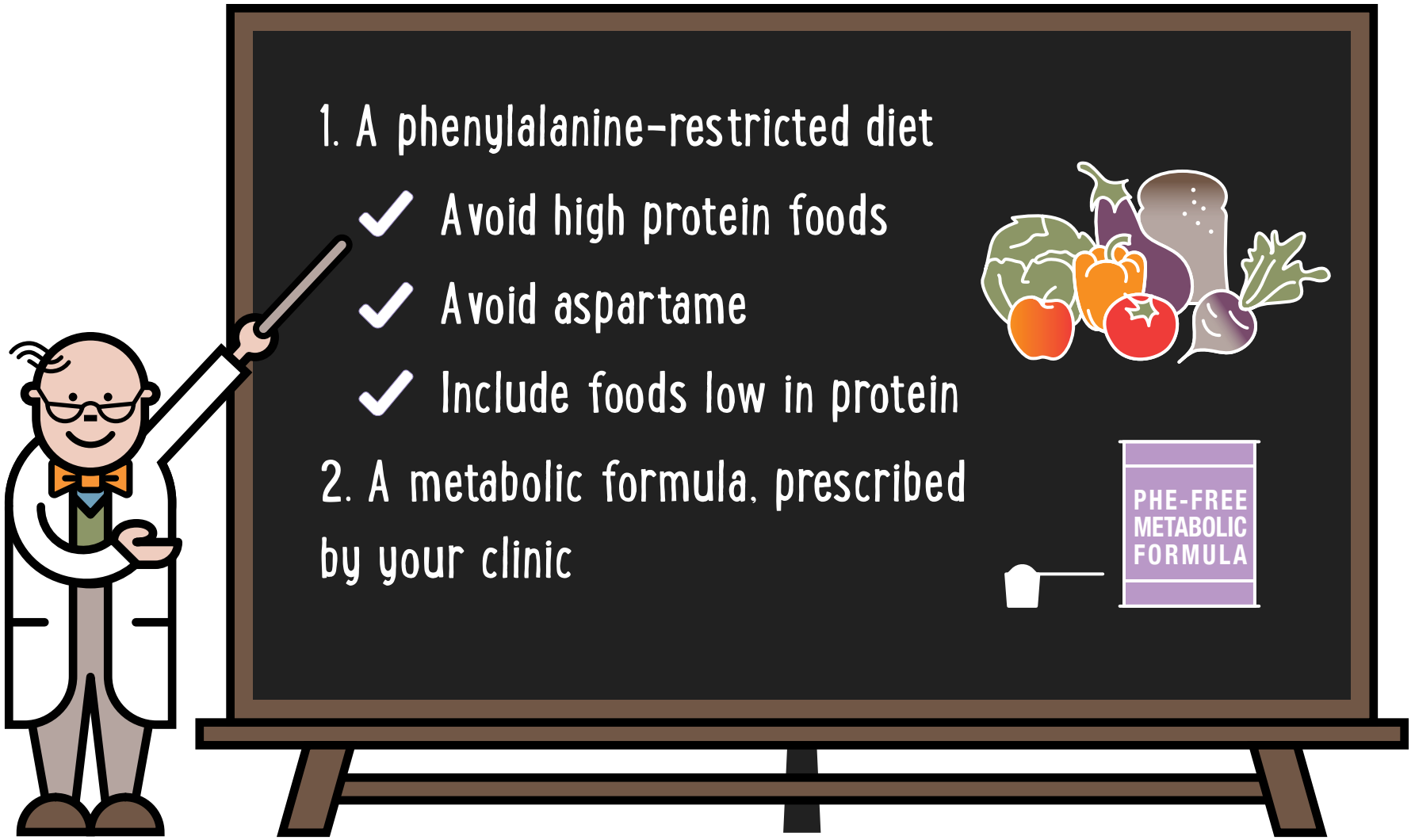
How is PKU diagnosed?

As part of newborn screening, a few drops of blood are collected.

The blood sample is then analyzed and the PHE level is measured.

A high PHE level could mean your child has PKU which will prompt your clinician to do further testing to confirm the diagnosis.

How is PKU managed day-to-day?



Avoid high protein foods

Foods rich in protein, and therefore phenylalanine, should be avoided. This includes **meat, fish, eggs, cheese, milk, bread, pasta, nuts, soy and tofu**.

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

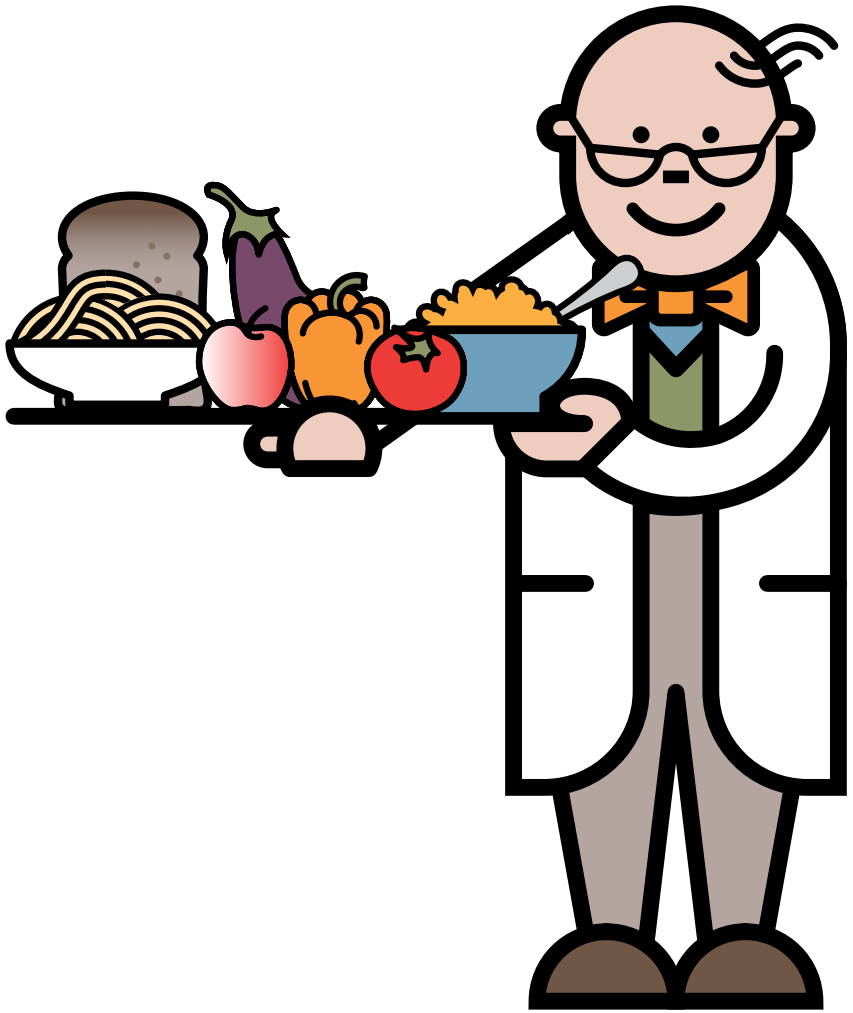
Include foods low in protein

These are foods that contain small amounts of phenylalanine which can be used in typical quantities.

They include many fruits and vegetables, and special medical low protein foods.

They provide:

- an important source of energy
- variety in the diet



Low protein cooking

Cooking low-protein meals for your child can still be appealing to the eye and taste good.

There are many low protein cookbooks, some even created by PKU families. Your dietitian may be able to recommend a few favorites.

Feeding your baby with metabolic formula

Phenylalanine is essential for normal development and therefore a limited and controlled amount must be taken daily.

Breast milk or standard infant formula will provide the phenylalanine required by your baby prior to the introduction of solids, generally around 4-6 months of age.

Your baby will also need a special metabolic formula to provide protein without PHE.

Your dietitian will determine how much breast milk or standard infant formula and metabolic formula to offer.

Phenylalanine-free metabolic formula

Phenylalanine-free metabolic formula is an essential part of meeting your baby's nutritional requirements.

Like breast milk or standard infant formula, metabolic formula has carbohydrate, fat, vitamins, minerals and protein in the form of amino acids without PHE.

Metabolic formula, with prescribed amount of PHE, allows your baby to get all the nutrients he or she needs to grow.

Tracking Phenylalanine

As your baby starts to eat solids your clinic will work with you to track phenylalanine.

Foods must be weighed or measured using household measures (1 cup, 1 Tablespoon, etc) to determine phenylalanine content.

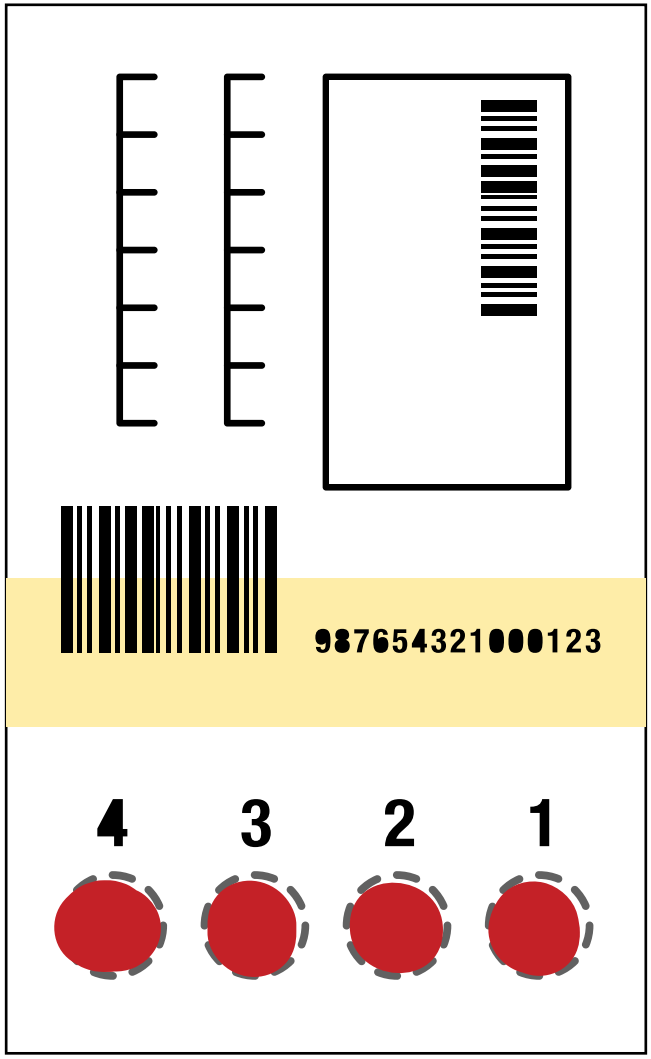
Your clinic can help you find the best tools to help determine the phenylalanine content of foods.

How is PKU monitored?

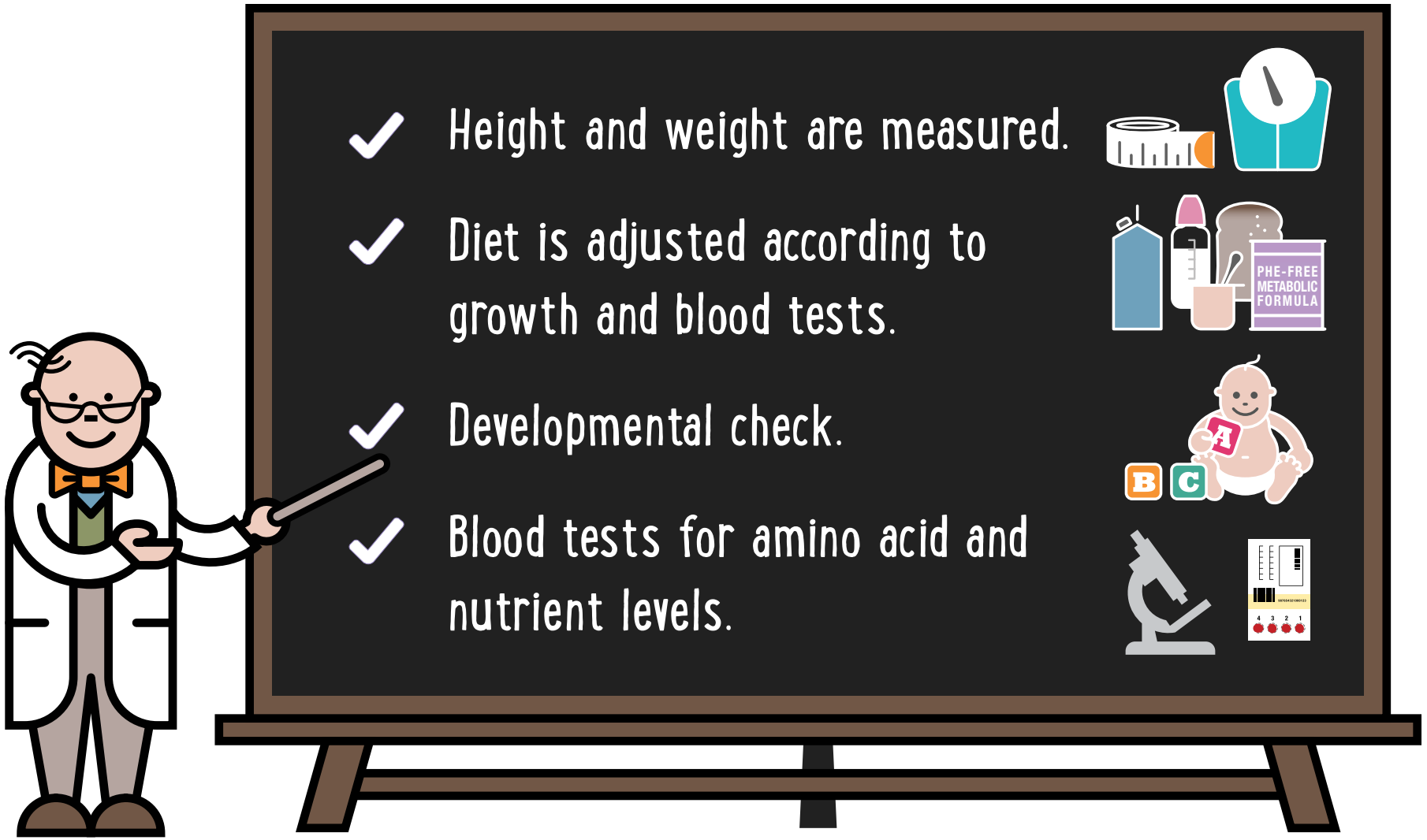
By regular blood tests taken at home or in the clinic and reviewed by a dietitian.

The sample is tested for the amount of phenylalanine it contains.

The metabolic dietitian will contact you with the result and discuss any changes in management.



What happens during a clinic check-up?



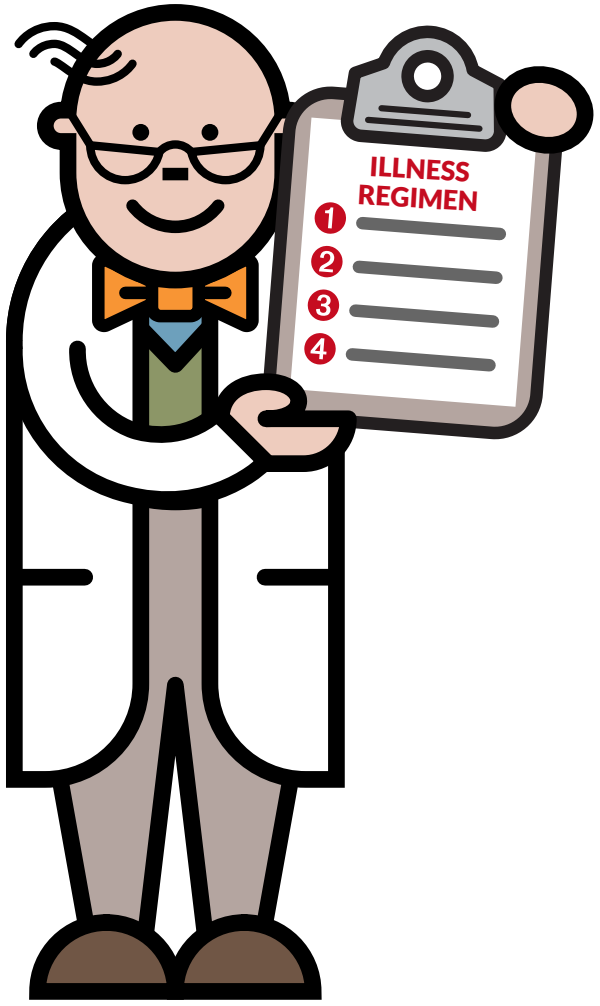
How is PKU managed during illness?

During any illness, our bodies need extra energy. The body will start breaking down body protein, causing blood PHE levels to increase. This process is also referred to as catabolism.

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

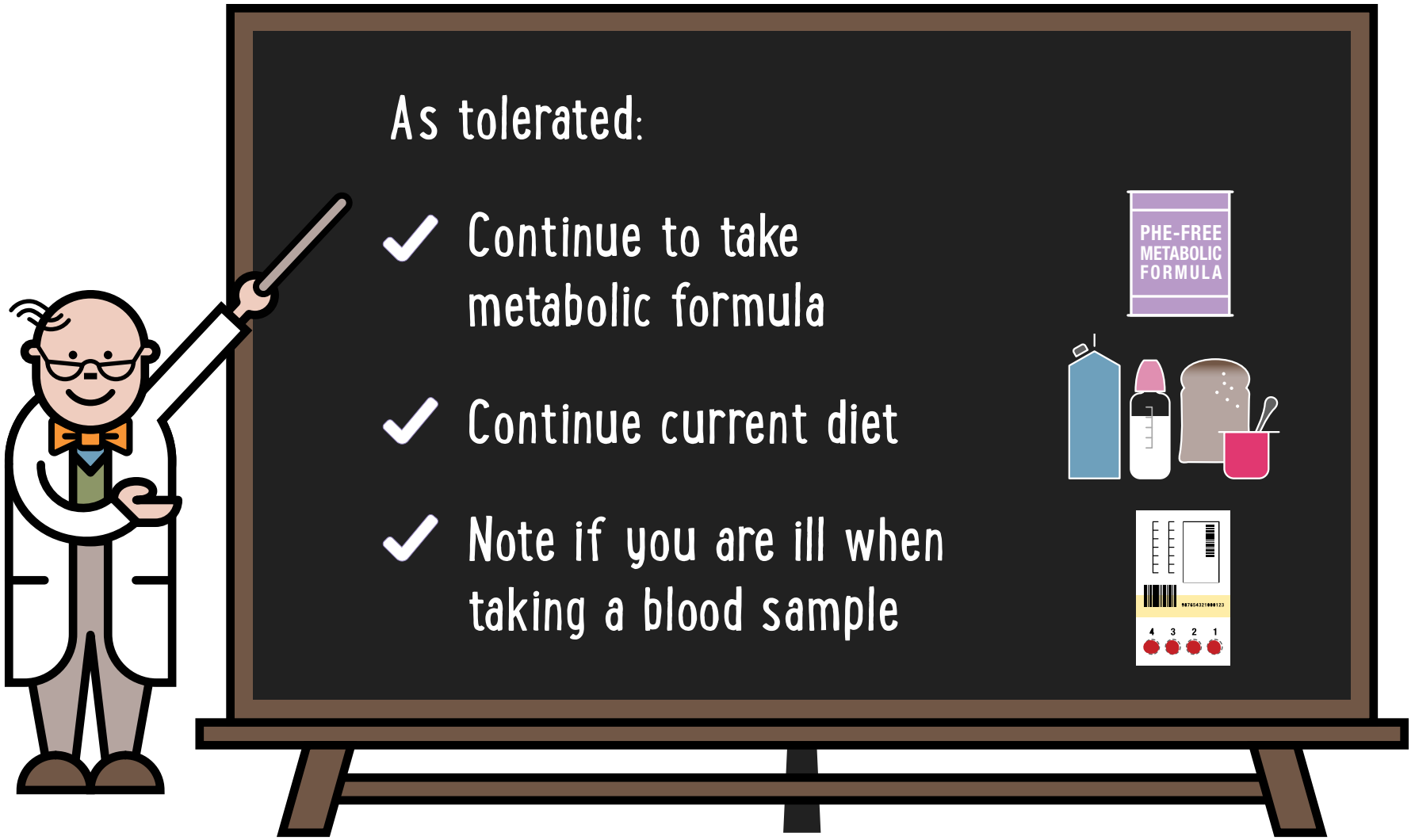
Metabolic formula can help keep blood PHE levels down by providing PHE-free protein and energy.

Your clinician will determine the best course of action.

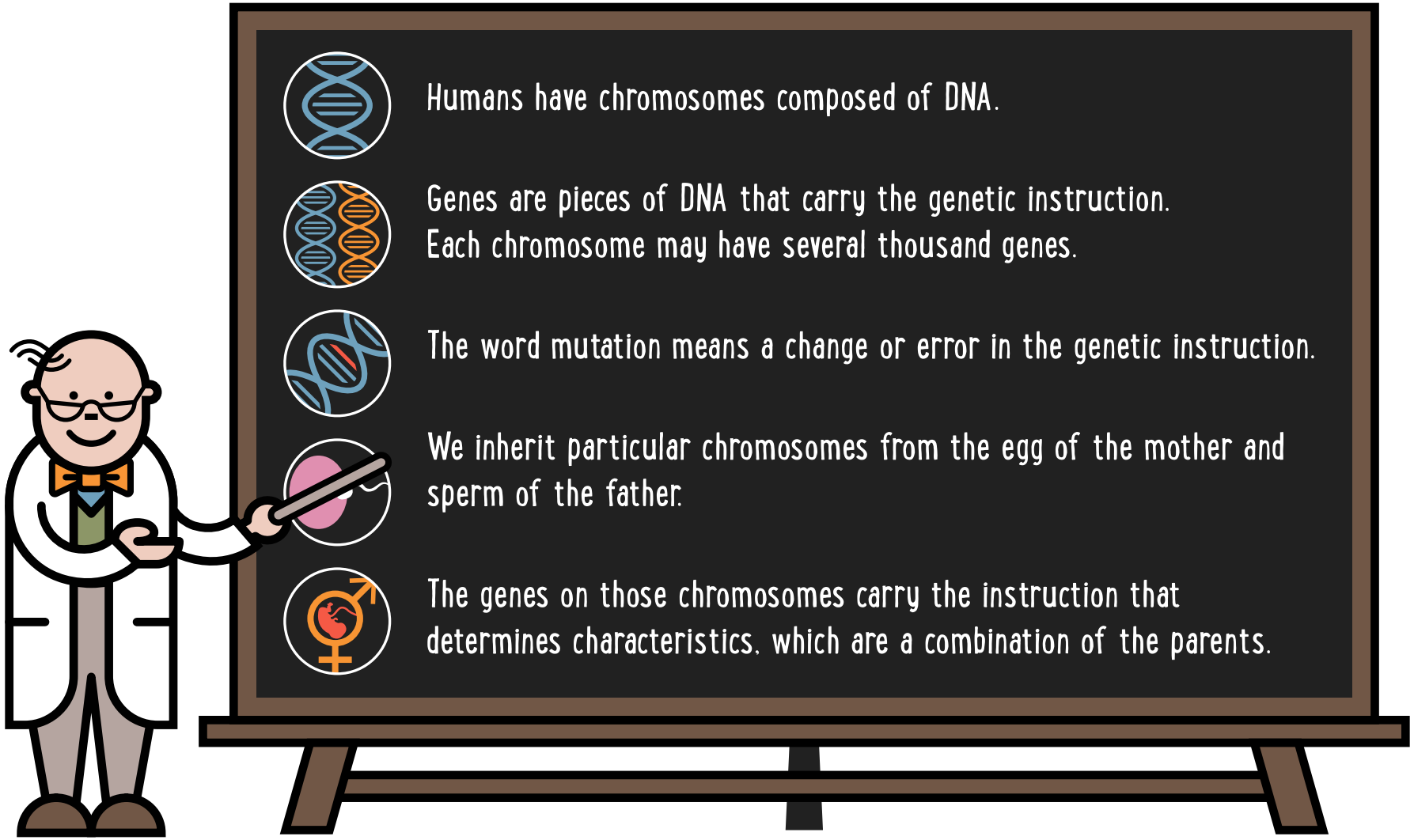


How is PKU managed during illness?

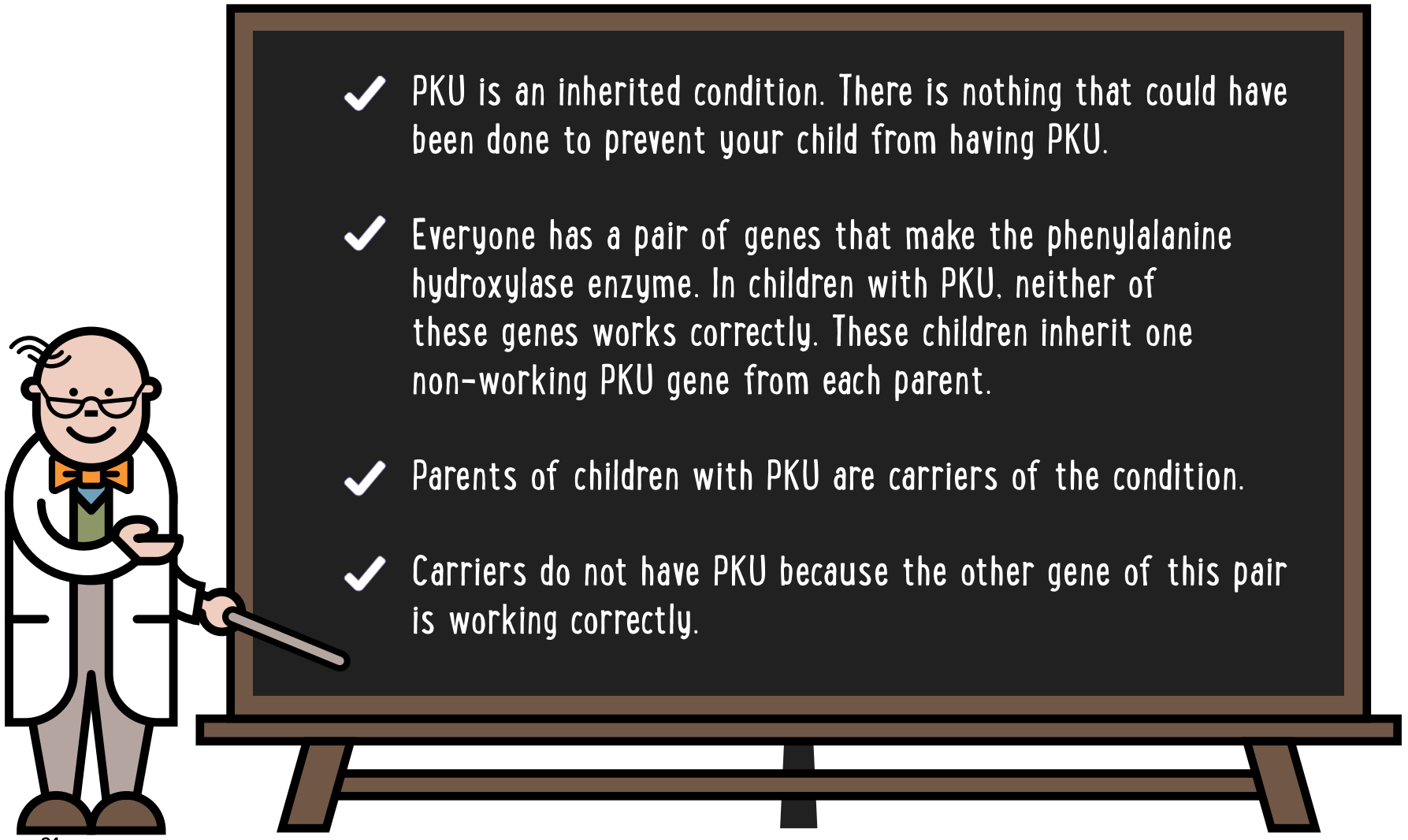
Always follow your medical team's guidance.



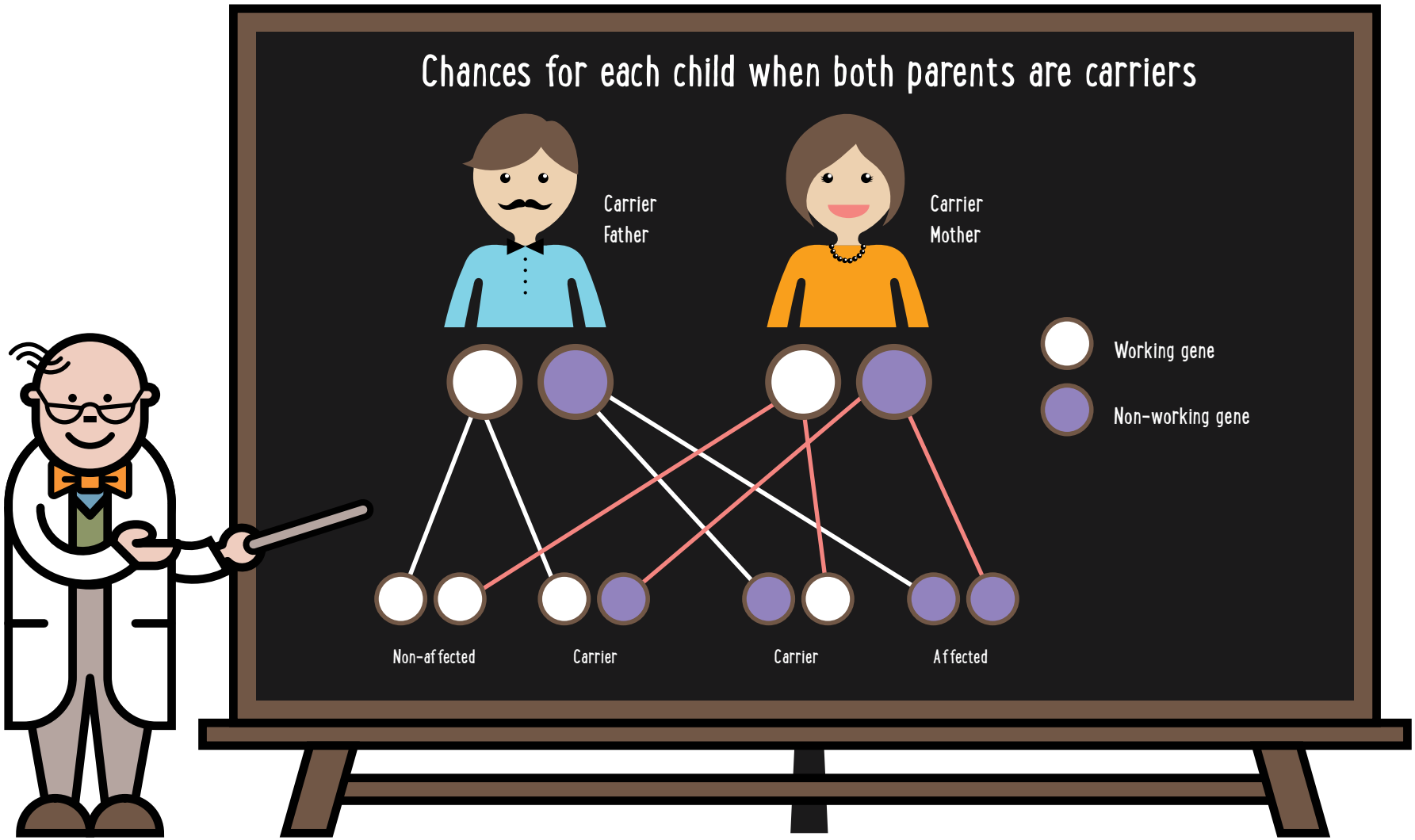
What happens in human genetics?



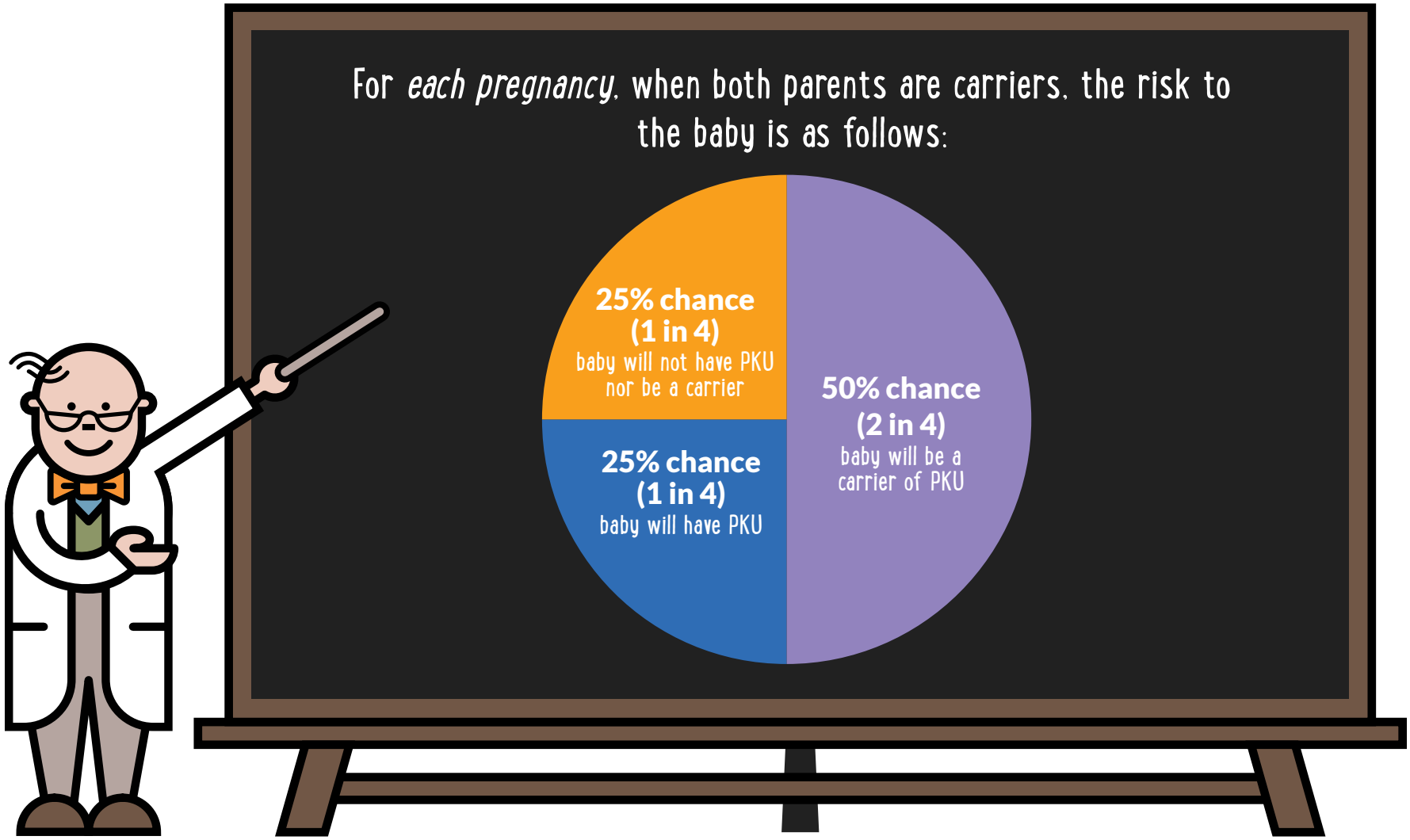
How does one inherit PKU?



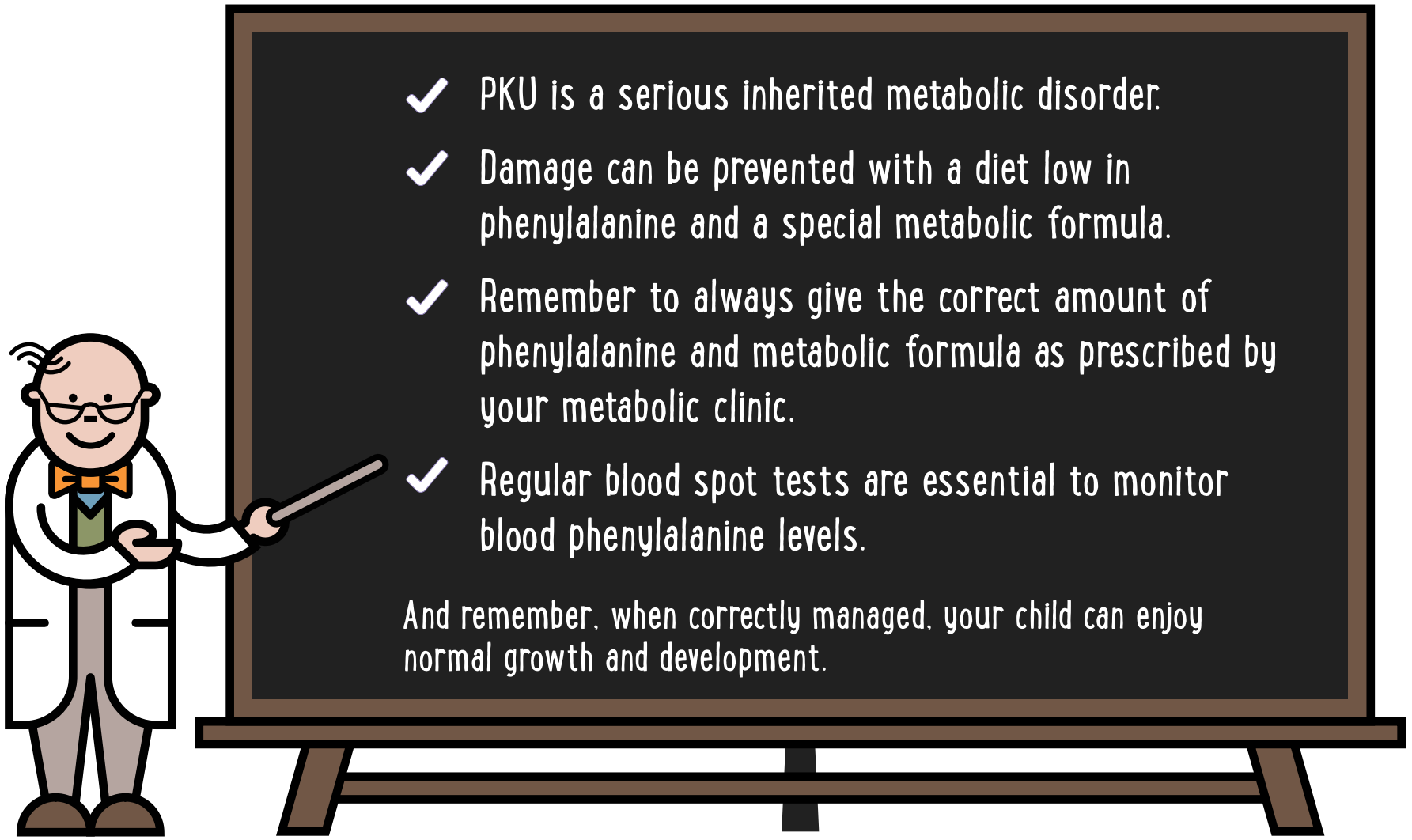
Inheritance – Autosomal-recessive – possible combinations



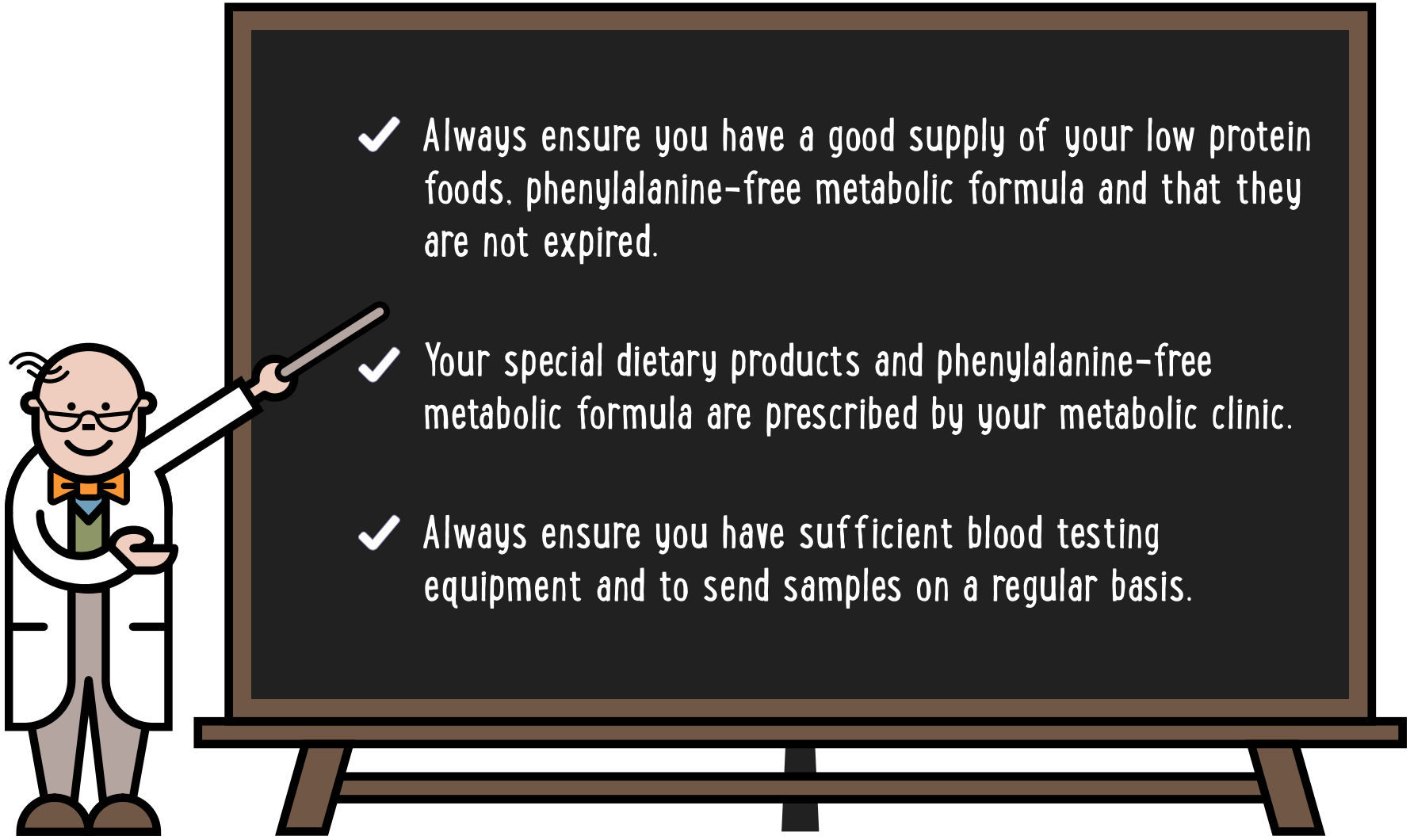
Future pregnancies



Take home messages



Helpful hints



Who's who (contact details)

My dietitian

Name: .....

Phone #: .....

Email: .....

My nurse

Name: .....

Phone #: .....

Email: .....

My doctor

Name: .....

Phone #: .....

Email: .....

Notes

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