

# 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

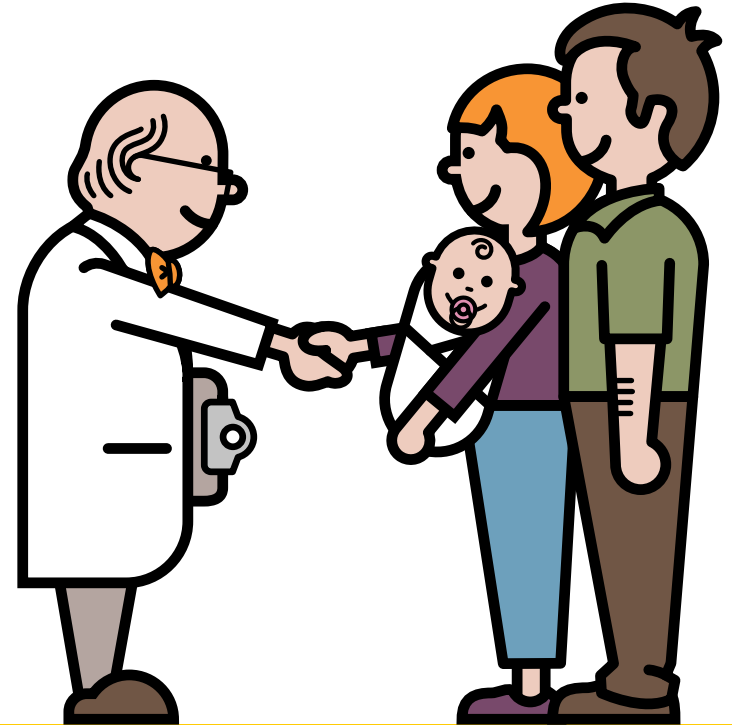
VERSION 2, FEBRUARY 2017

PA

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

# PA

Information for parents  
following a new diagnosis



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

**TEMPLE**



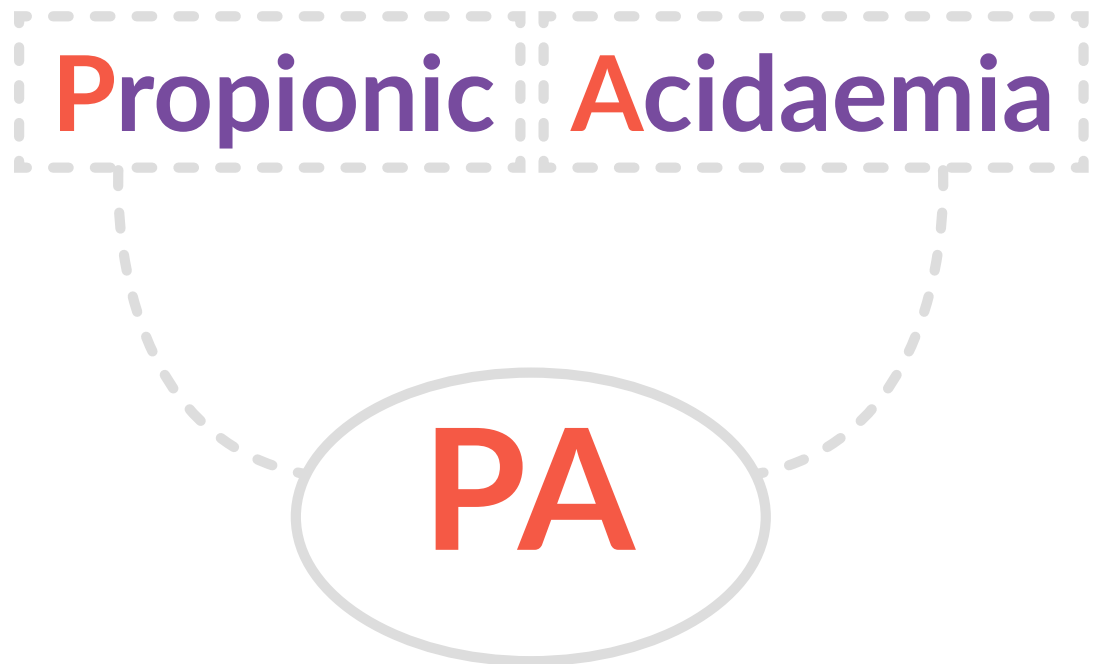
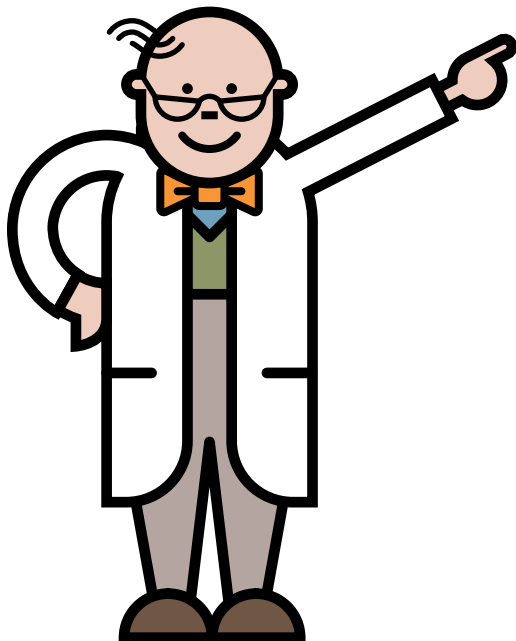
Tools Enabling Metabolic Parents LEarning

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

# What is PA?

PA stands for Propionic Acidaemia

It is an inherited metabolic condition

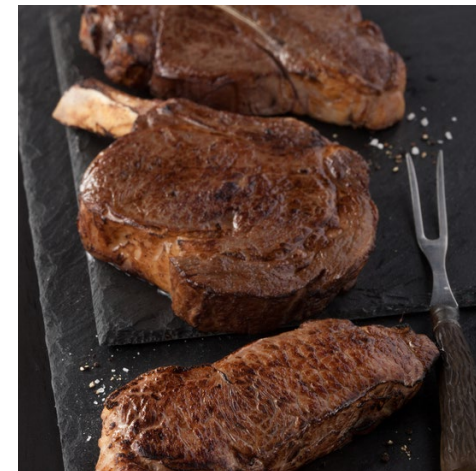
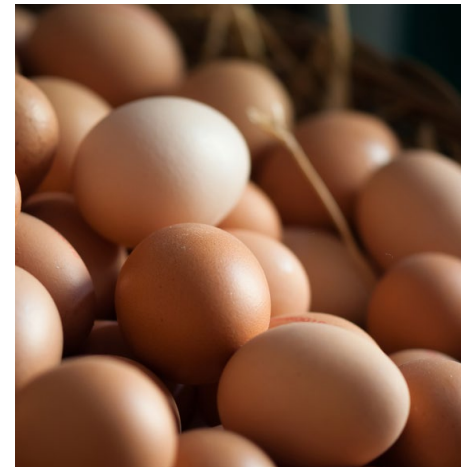


# PA and protein

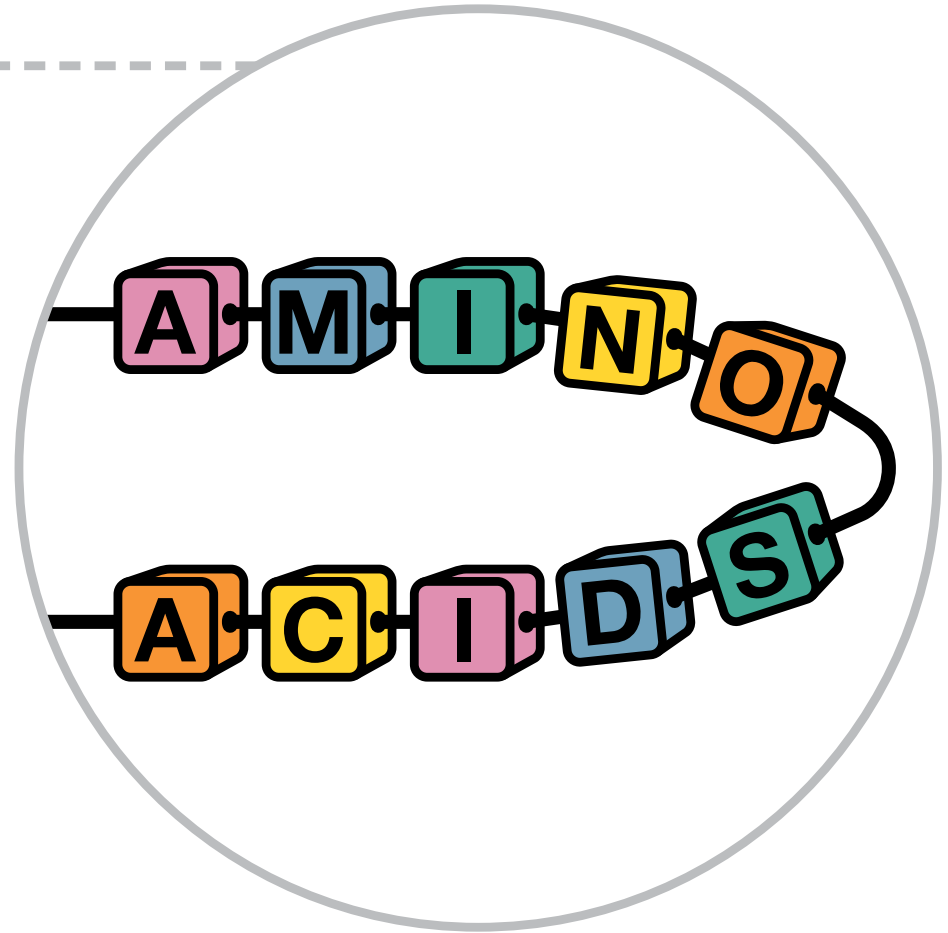
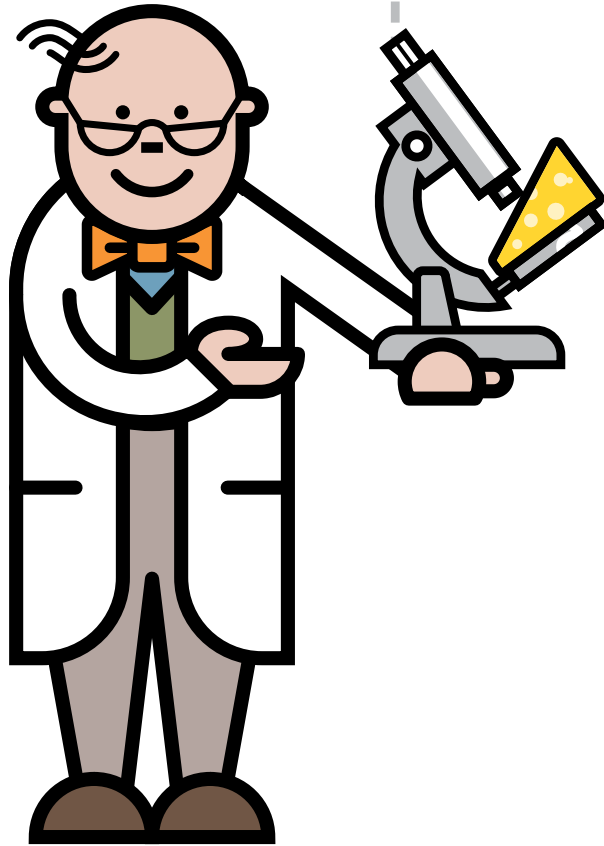
PA affects the way your baby breaks down protein

Many foods contain protein

The body needs protein for growth and repair



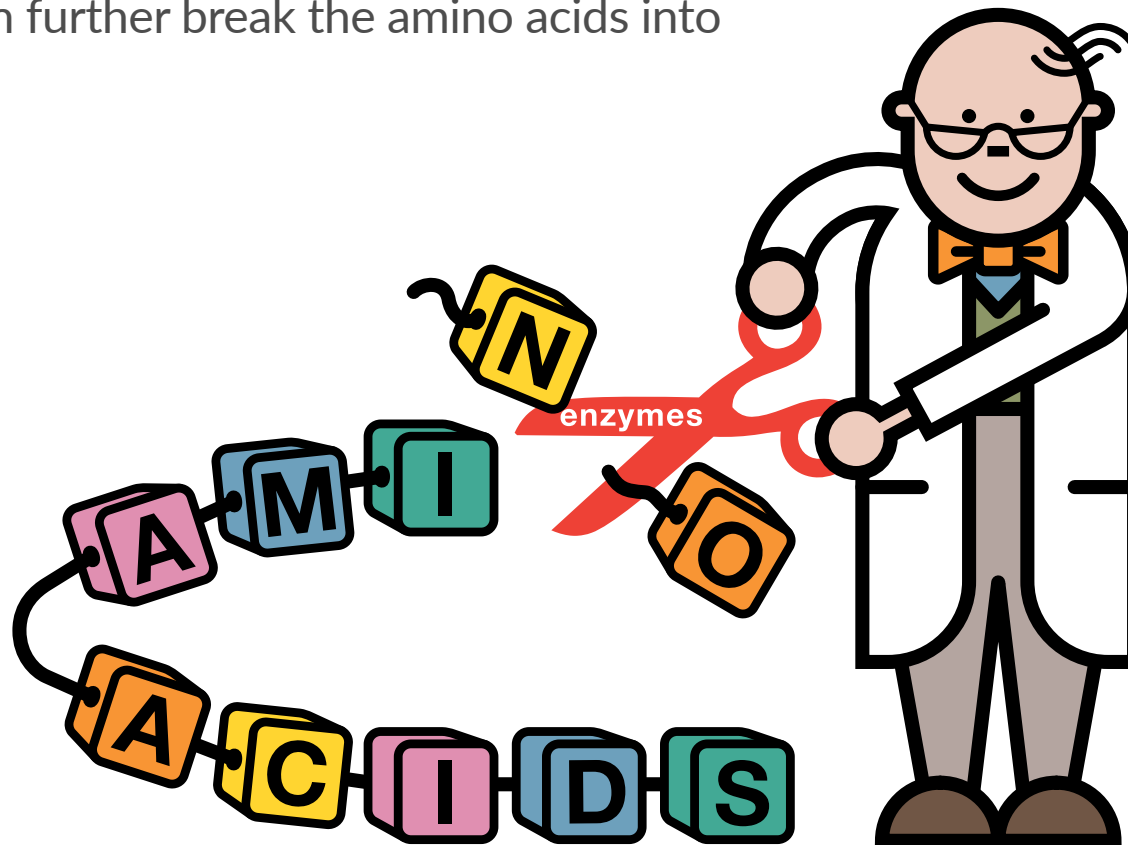
# What is protein?



# 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.

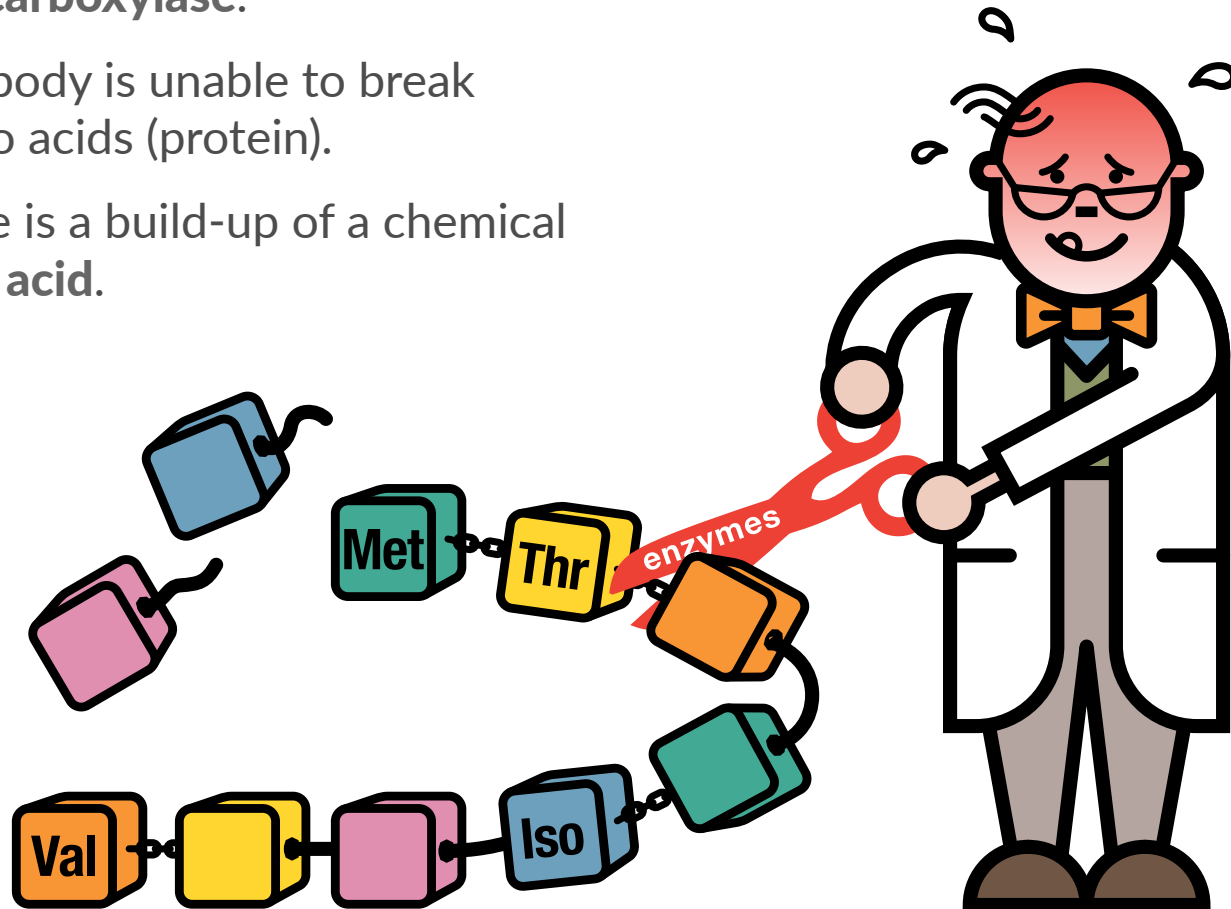


# What happens in PA?

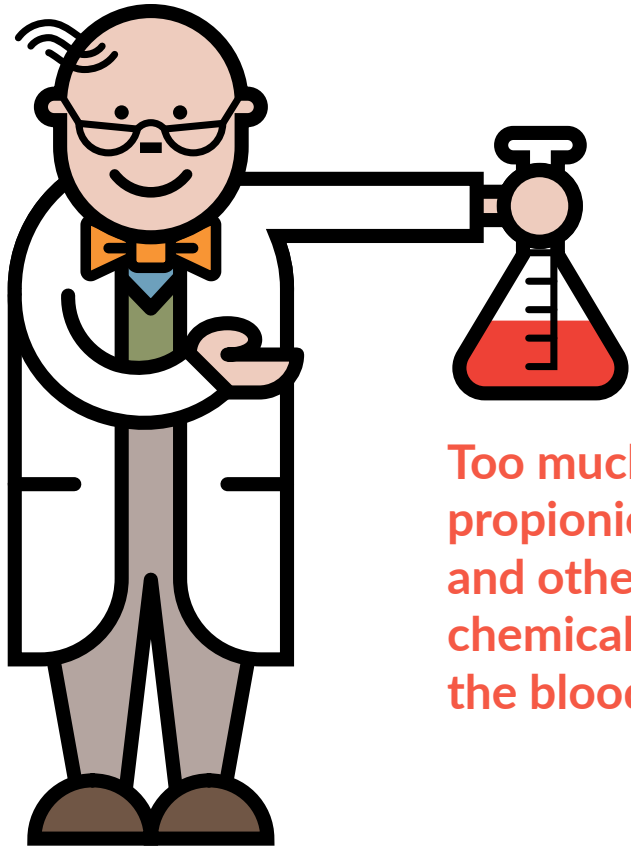
In PA, the body lacks an enzyme called **propionyl-CoA carboxylase**.

This means the body is unable to break down four amino acids (protein).

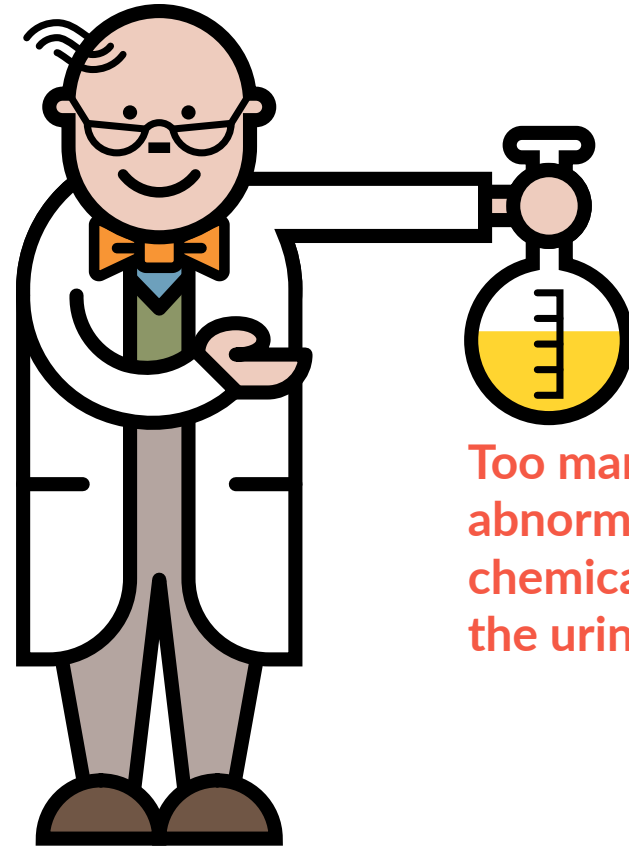
As a result, there is a build-up of a chemical called **propionic acid**.



# What does this cause?



Too much propionic acid and other chemicals in the blood



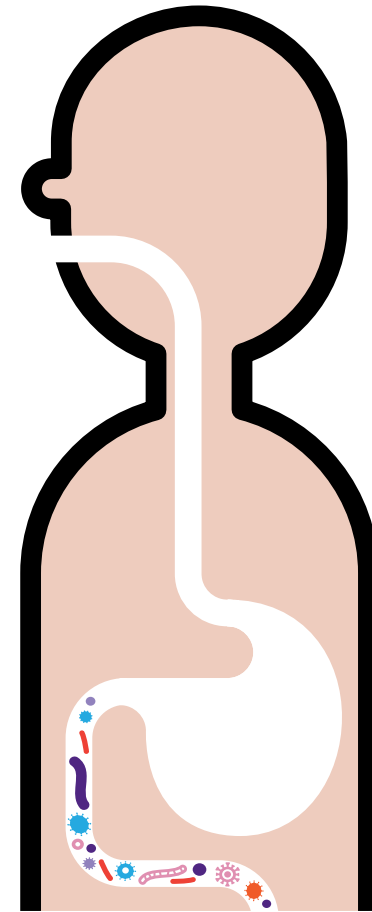
Too many abnormal chemicals in the urine



# Other sources of propionic acid

Propionic acid also comes from:

- The breakdown of fatty acids. The body will use these for energy when it has gone a long time without food
- Gut bacteria



# How is PA diagnosed?

PA is diagnosed by measuring chemicals containing propionic acid in the blood and urine.

It can also be diagnosed by looking at enzyme levels and at the body's genes.



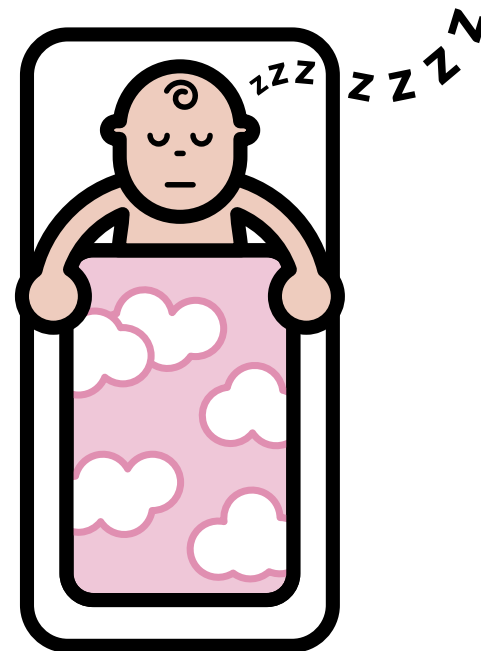
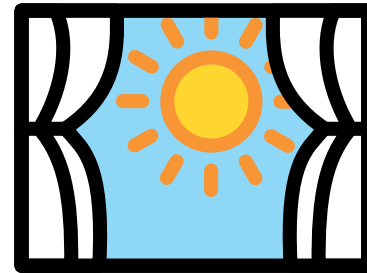
# What are the symptoms in PA?

Symptoms commonly start in the first few days of life.

Symptoms include:

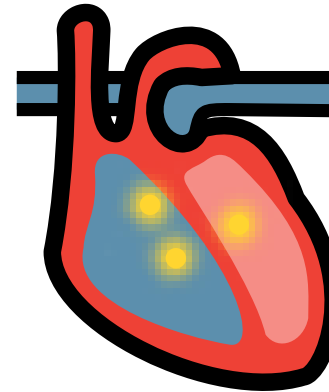
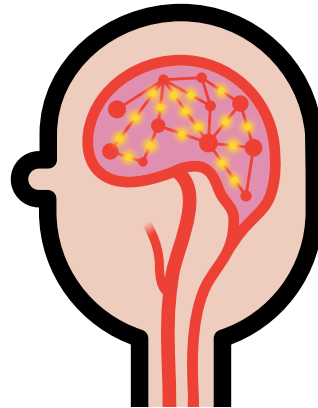
- poor feeding
- vomiting
- dehydration (lack of body fluids)
- floppy baby
- excessively sleepy
- rapid breathing
- seizures

**The effects of PA quickly become life-threatening if untreated**

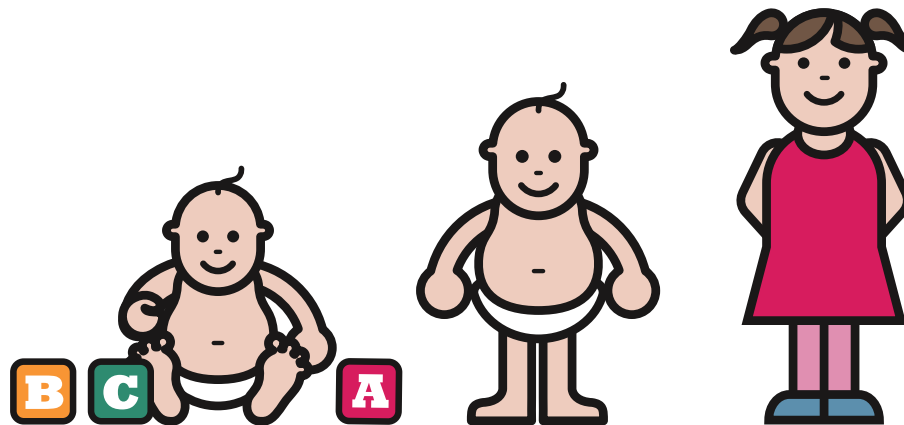


# What can go wrong in PA?

The build up of harmful chemicals can damage the brain and heart and cause problems with other organs.



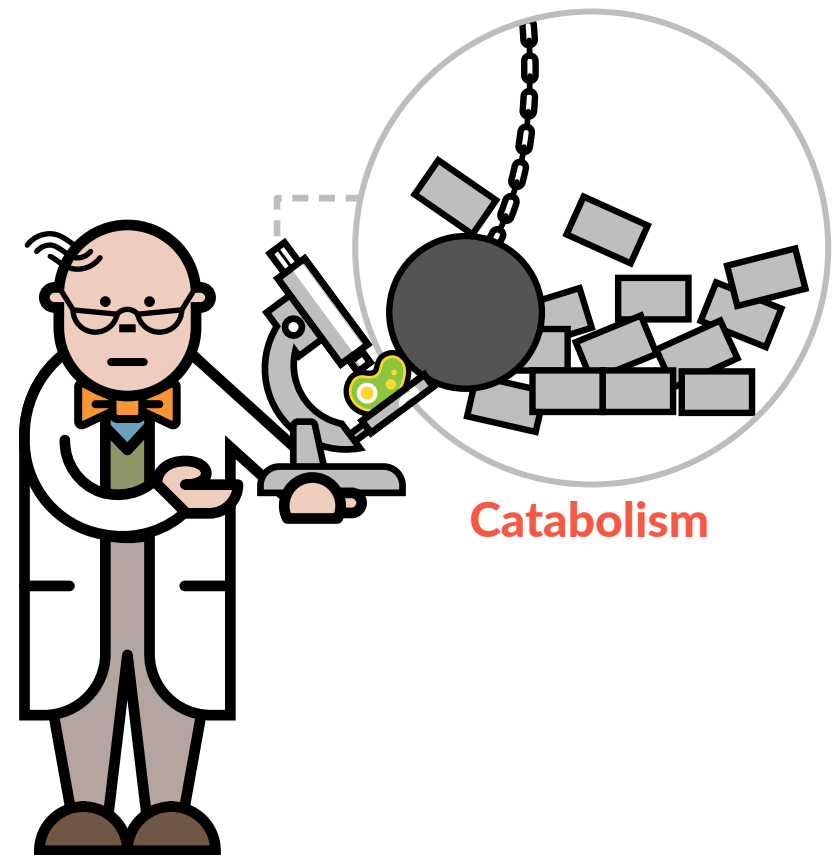
It may cause delays to normal development like walking and talking.



# What else happens in PA?

If the body does not receive enough food e.g. during illness or the body has gone without food for too long, there may be a shortage of energy supply.

This causes **catabolism** which is a break down of body protein and can lead to a metabolic crisis.



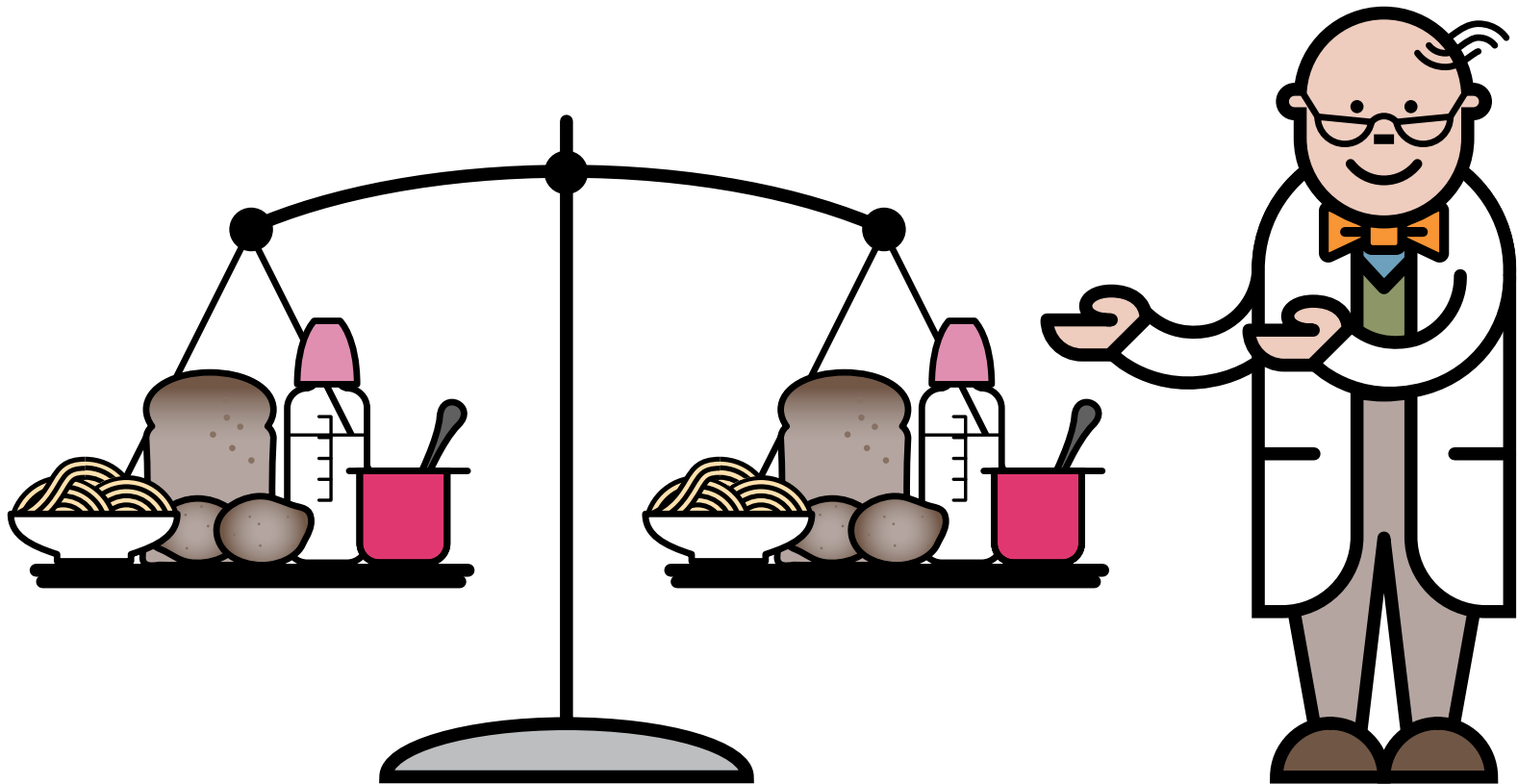
# Metabolic crisis

- In a **metabolic crisis** there is a build up of propionic acid and other toxic chemicals such as ammonia
- It is usually triggered by childhood illnesses e.g. vomiting and diarrhoea or fasting for too long
- There should be no delay in treatment
- Avoidance of a metabolic crisis is essential



# Protein balance is needed in PA

In PA, it is important that enough protein is given for growth  
...but not too much as toxic chemicals will be made.



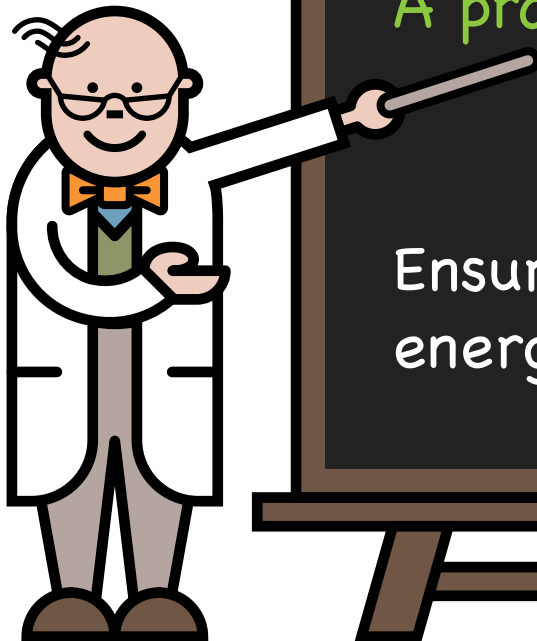
# How is PA managed day to day?

PA is managed with the following:

A protein restricted diet



Ensuring a sufficient energy supply



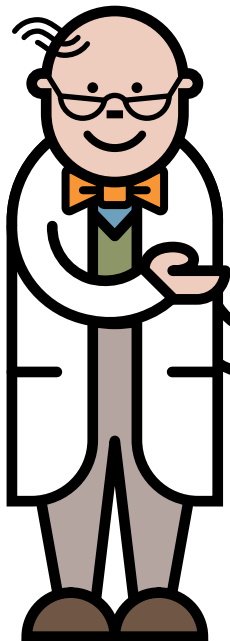


# How is PA managed day to day?

PA is managed with the following:

A protein restricted diet

Ensuring a sufficient  
energy supply



# How is PA managed day to day?

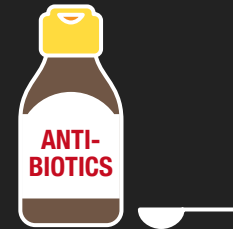
Regular feeding



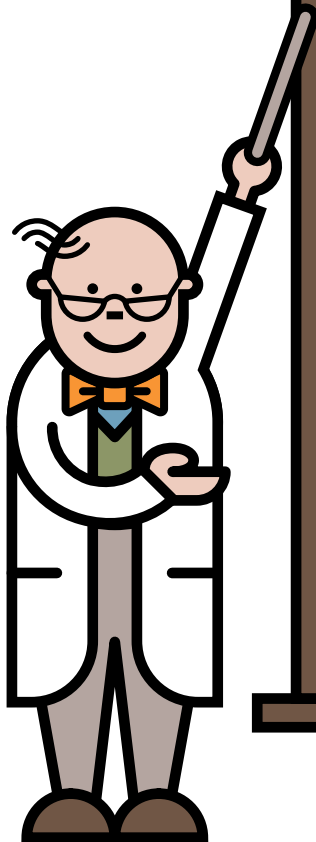
Carnitine medication



Antibiotics to control gut bacteria



Other medications may be necessary



# How is PA managed day to day?

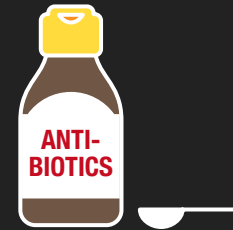
Regular feeding



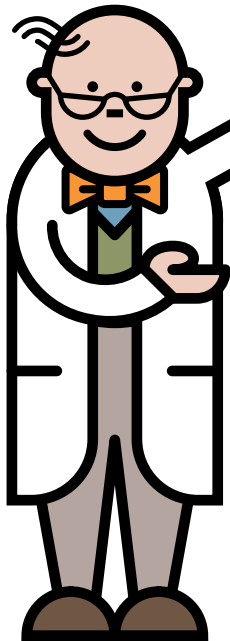
Carnitine medication



Antibiotics to control gut bacteria



Other medications may be necessary



# How is PA managed day to day?

Regular feeding



Carnitine medication



Antibiotics to control  
gut bacteria



Other medications may be necessary



# How is PA managed day to day?

Regular feeding



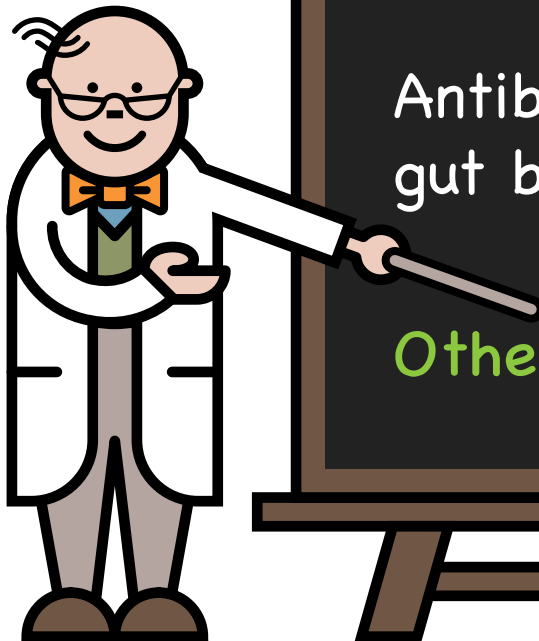
Carnitine medication



Antibiotics to control gut bacteria

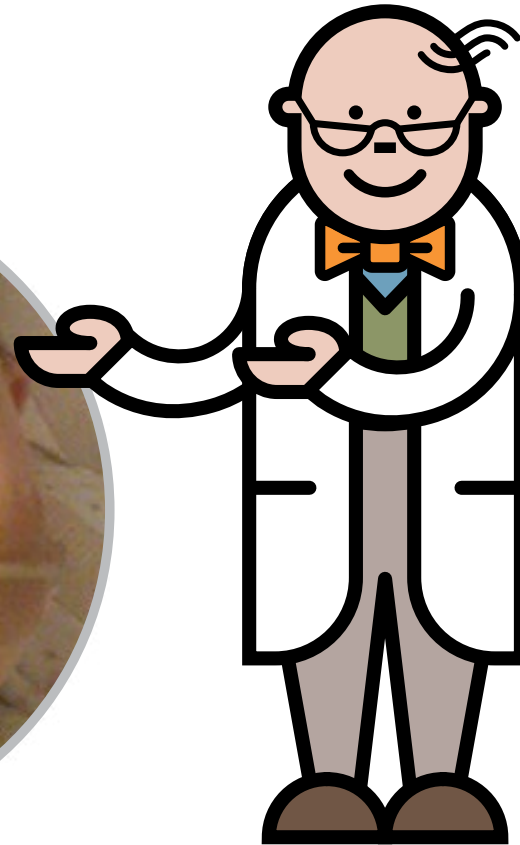


Other medications may be necessary



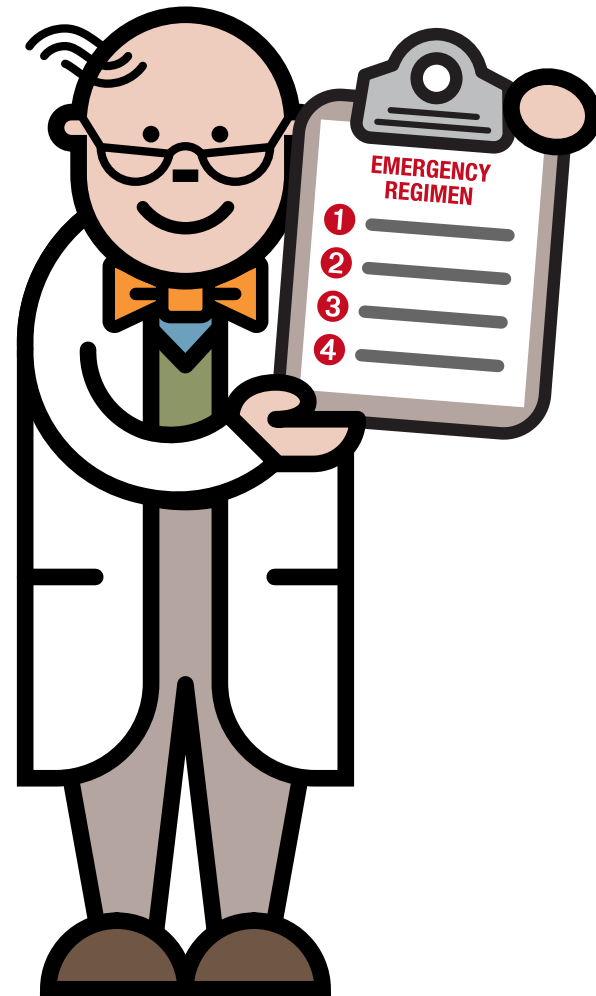
# Is tube feeding needed?

Tube feeding may be necessary to give regular feeds. This will ensure energy, nutrient and fluid needs are met and can help to reduce the production of abnormal chemicals.



# How is PA managed during illness?

- During any childhood illness, an emergency regimen is given
- This is to ensure sufficient energy supply to prevent the build up of harmful chemicals that cause a metabolic crisis



# How is PA managed during illness?

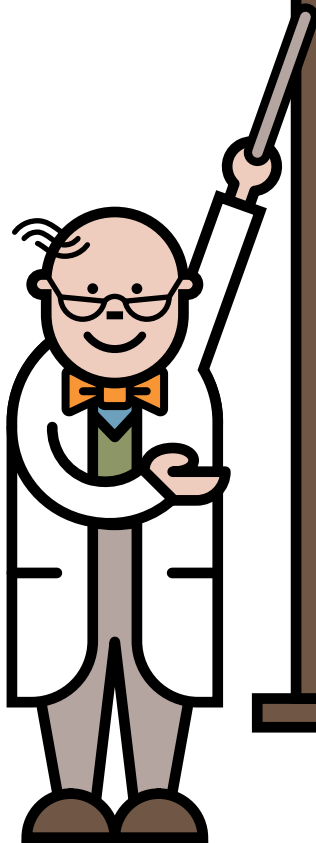
Stop all protein in food & drink



Start the emergency regimen. This is made up of glucose polymer



Continue medication as prescribed



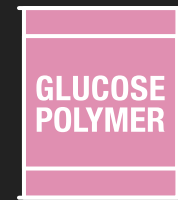


# How is PA managed during illness?

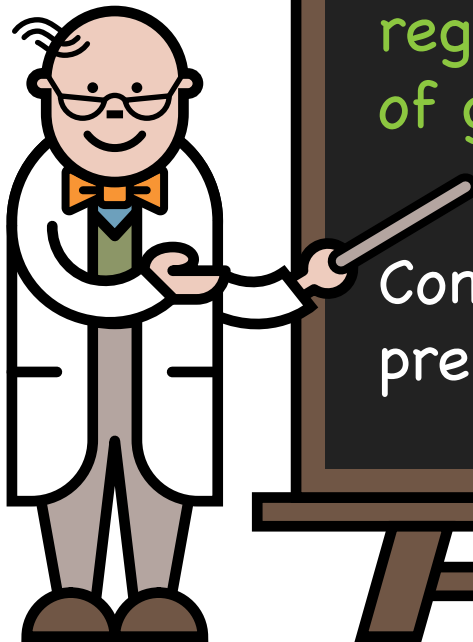
Stop all protein in food & drink



Start the emergency regimen. This is made up of glucose polymer



Continue medication as prescribed

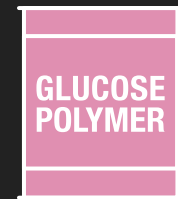


# How is PA managed during illness?

Stop all protein in food & drink



Start the emergency regimen. This is made up of glucose polymer

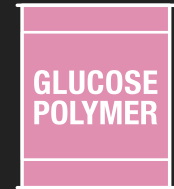


Continue medication as prescribed



# Checklist for illness

Always take full amounts of  
emergency feeds as prescribed



If symptoms continue and/or you are  
worried, go immediately to the hospital

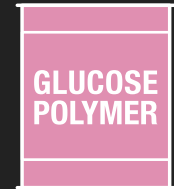


Regularly update your metabolic team



# Checklist for illness

Always take full amounts of emergency feeds as prescribed



If symptoms continue and/or you are worried, go immediately to the hospital

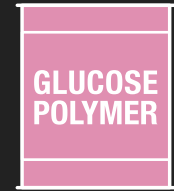


Regularly update your metabolic team



# Checklist for illness

Always take full amounts of  
emergency feeds as prescribed



If symptoms continue and/or you are  
worried, go immediately to the hospital



Regularly update your metabolic team

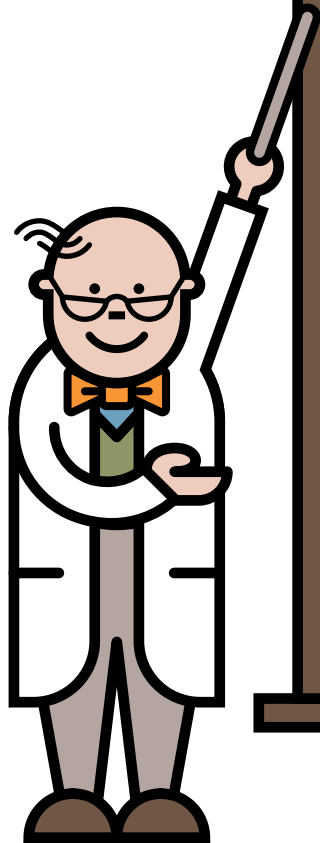


## Key message

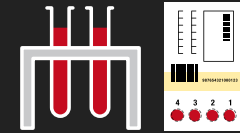
It is imperative that emergency feeds are started **promptly** and there are **no delays** in management.



# How is PA monitored?



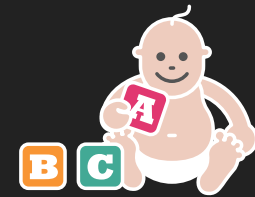
Frequent blood tests to check amino acids, nutrient and chemical levels



Height and weight



Developmental checks



Diet and medications are adjusted according to age, weight and blood chemical levels

# How is PA monitored?

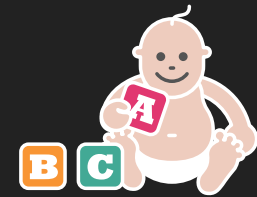
Frequent blood tests to check amino acids, nutrient and chemical levels



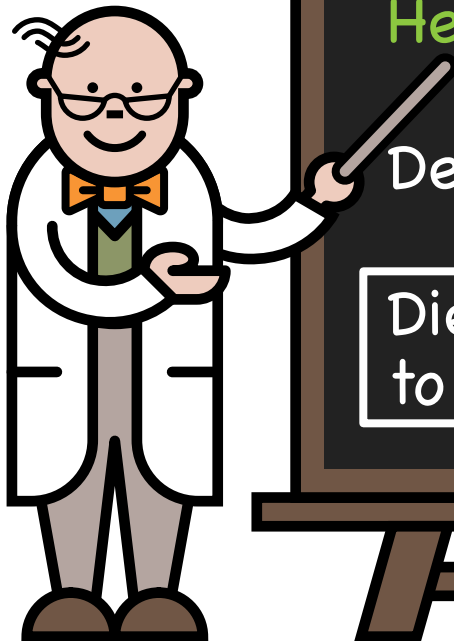
Height and weight



Developmental checks



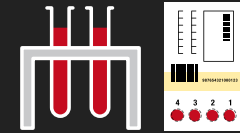
Diet and medications are adjusted according to age, weight and blood chemical levels





# How is PA monitored?

Frequent blood tests to check amino acids, nutrient and chemical levels



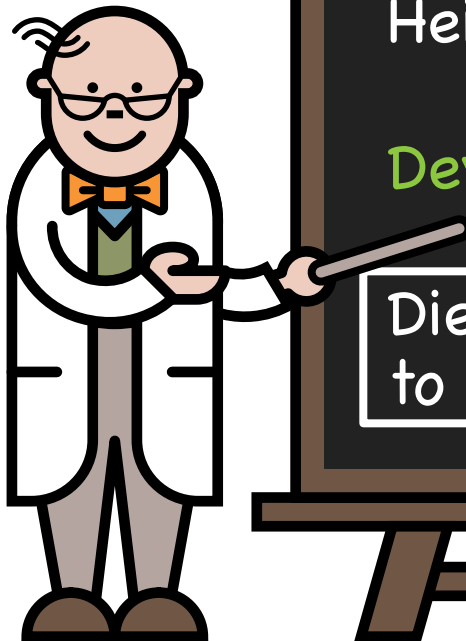
Height and weight



Developmental checks

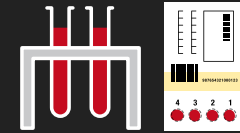


Diet and medications are adjusted according to age, weight and blood chemical levels



# How is PA monitored?

Frequent blood tests to check amino acids, nutrient and chemical levels



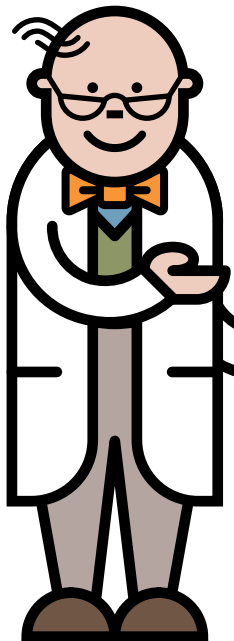
Height and weight



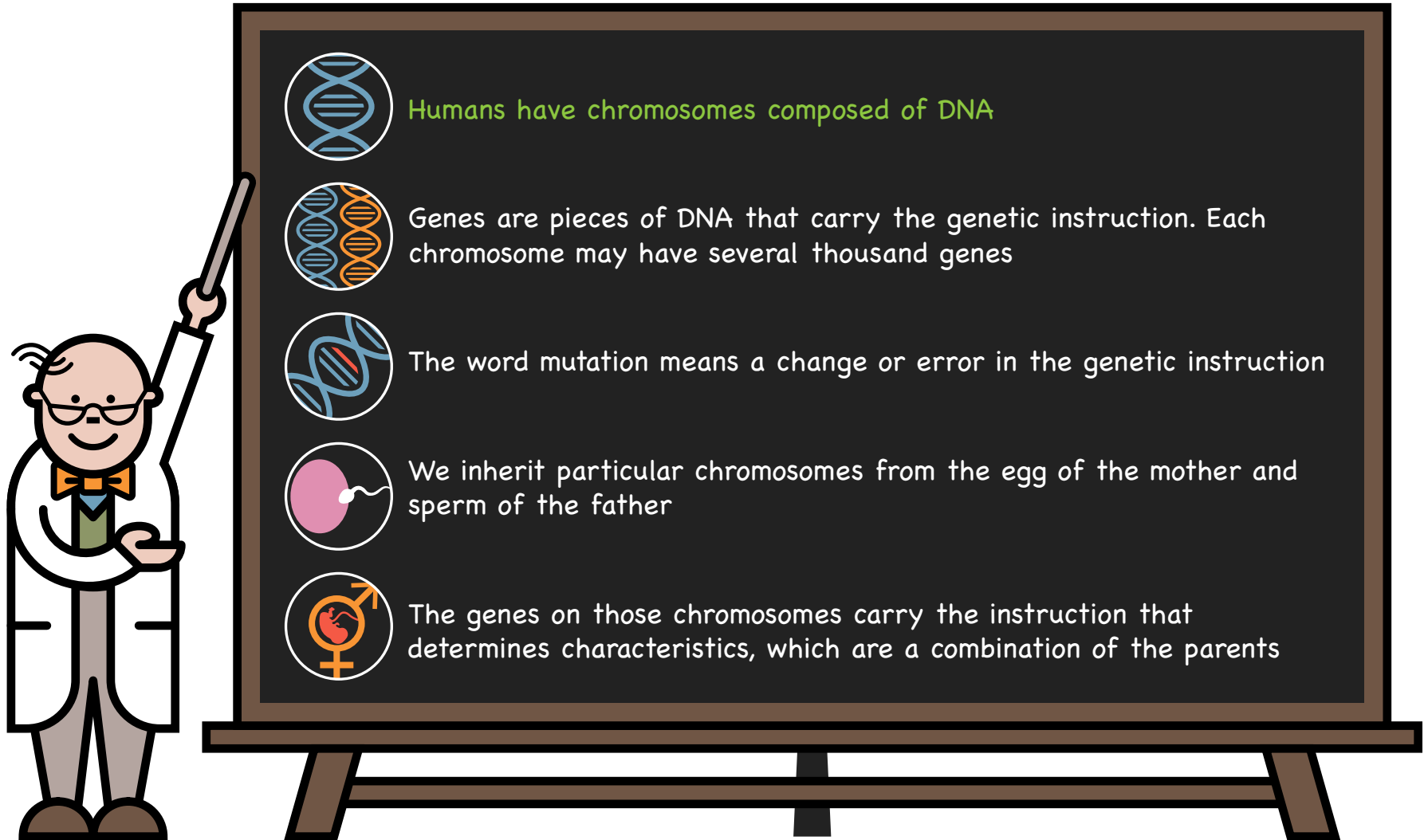
Developmental checks





Diet and medications are adjusted according to age, weight and blood chemical levels





# Chromosomes, genes, mutations




 Humans have chromosomes composed of DNA

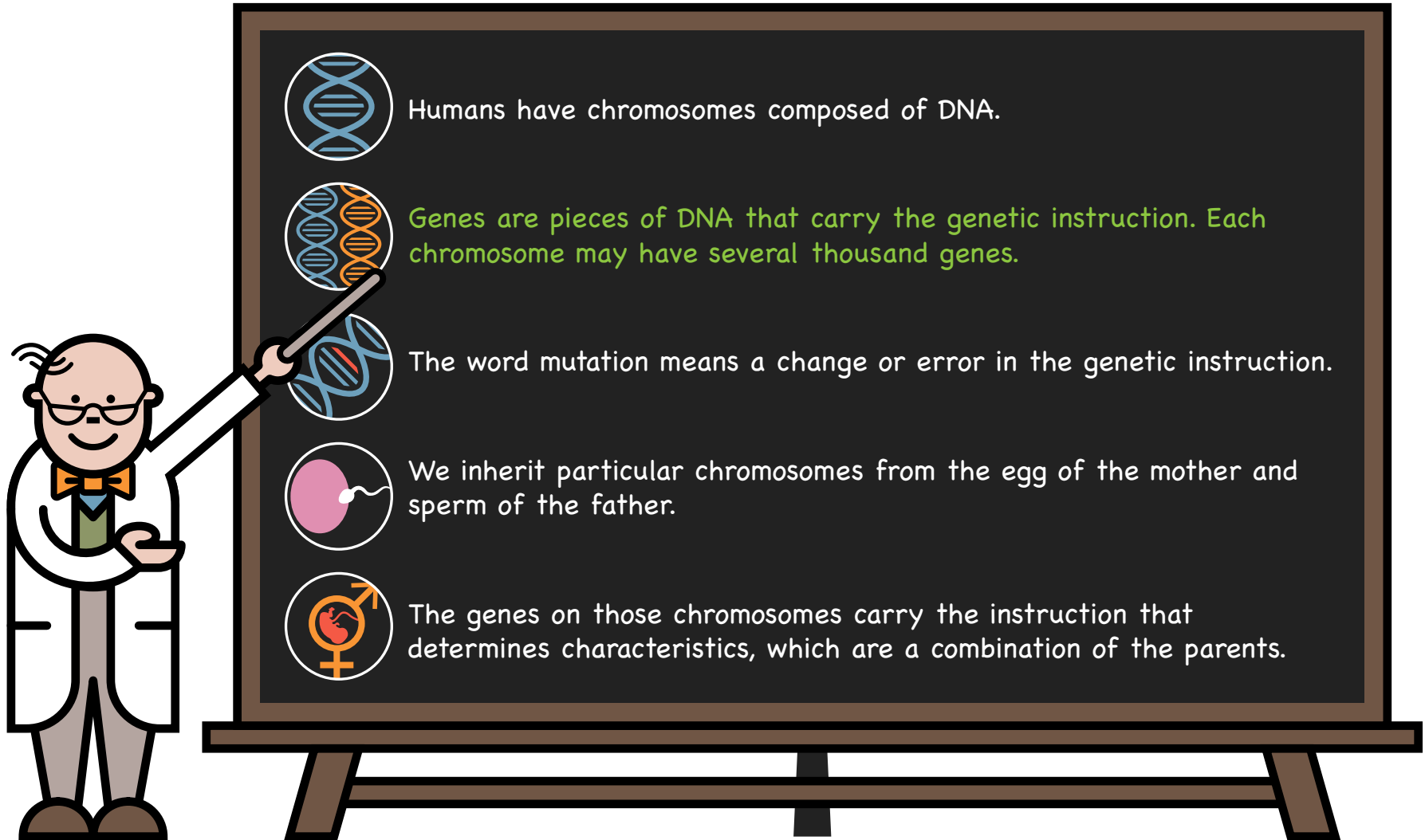
 Genes are pieces of DNA that carry the genetic instruction. Each chromosome may have several thousand genes

 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

# Chromosomes, genes, mutations



Humans have chromosomes composed of DNA.

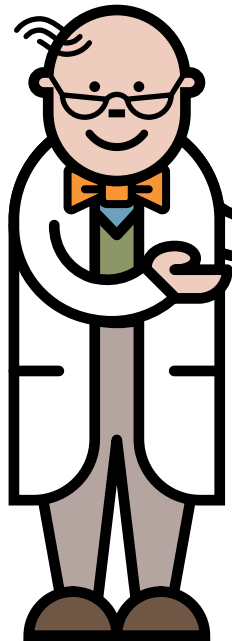
Genes are pieces of DNA that carry the genetic instruction. Each chromosome may have several thousand genes.

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.



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.

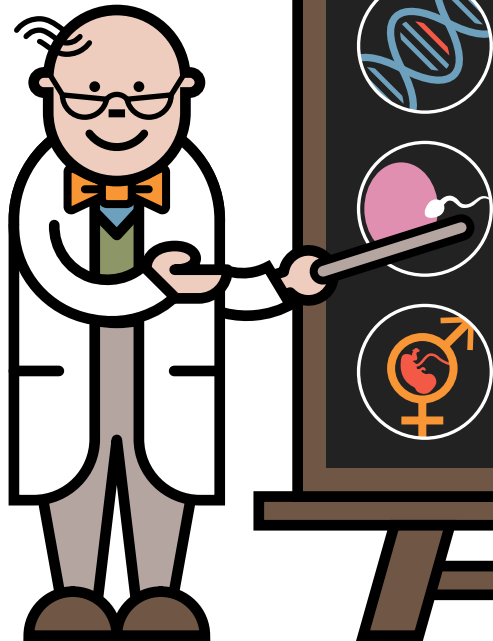


We inherit particular chromosomes from the egg of the mother and sperm of the father.



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.



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.

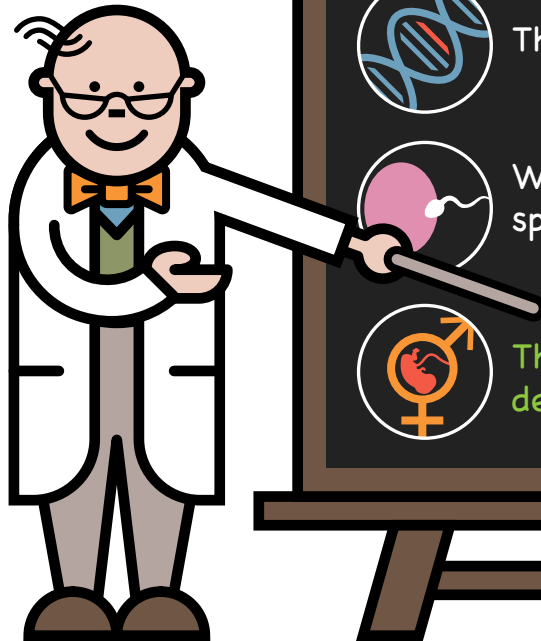


We inherit particular chromosomes from the egg of the mother and sperm of the father.



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.



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.

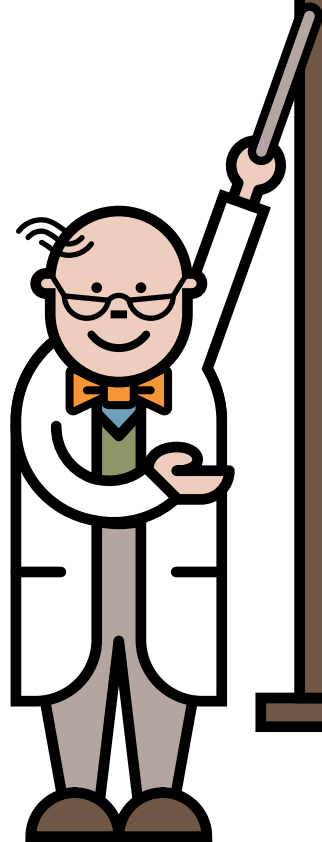


We inherit particular chromosomes from the egg of the mother and sperm of the father.



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

# Inheritance



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

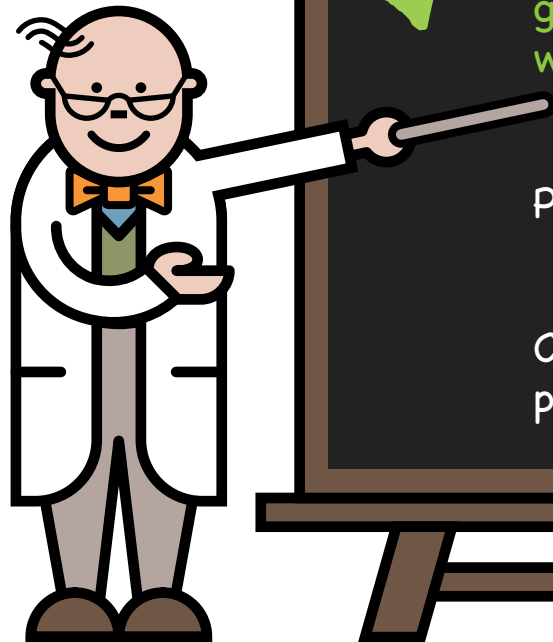
Everyone has a pair of genes that make the propionyl-CoA carboxylase enzyme. In children with PA, neither of these genes works correctly. These children inherit one non-working PA gene from each parent

Parents of children with PA are carriers of the condition

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



# Inheritance



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

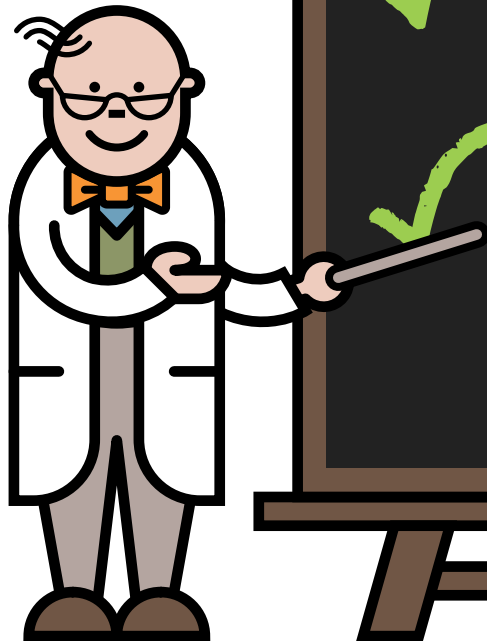


Everyone has a pair of genes that make the propionyl-CoA carboxylase enzyme. In children with PA, neither of these genes works correctly. These children inherit one non-working PA gene from each parent

Parents of children with PA are carriers of the condition

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

# Inheritance



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



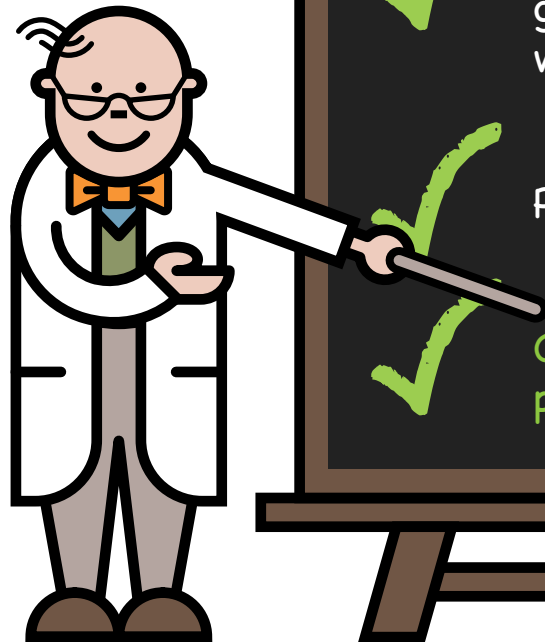
Everyone has a pair of genes that make the propionyl-CoA carboxylase enzyme. In children with PA, neither of these genes works correctly. These children inherit one non-working PA gene from each parent



Parents of children with PA are carriers of the condition

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

# Inheritance



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



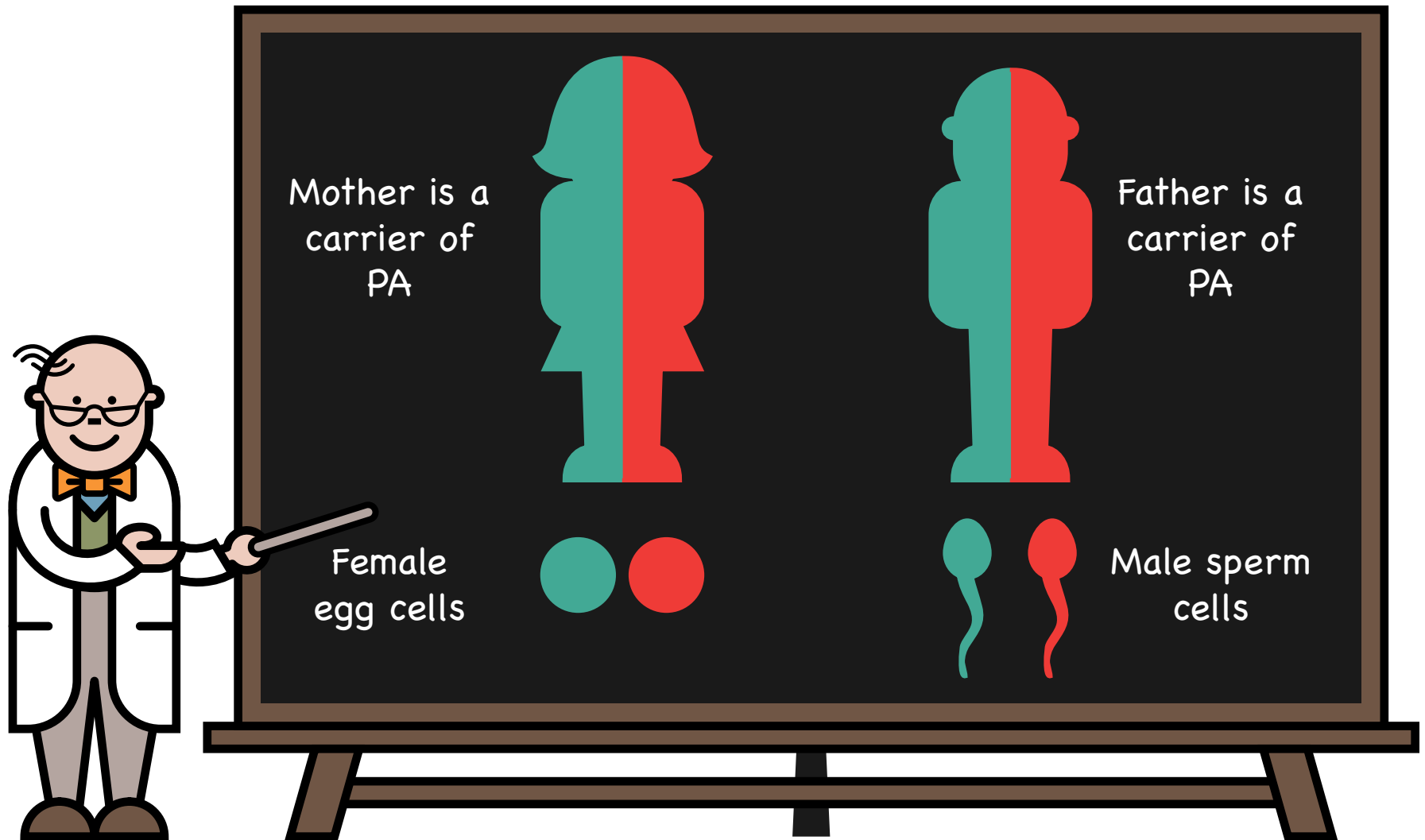
Everyone has a pair of genes that make the propionyl-CoA carboxylase enzyme. In children with PA, neither of these genes works correctly. These children inherit one non-working PA gene from each parent



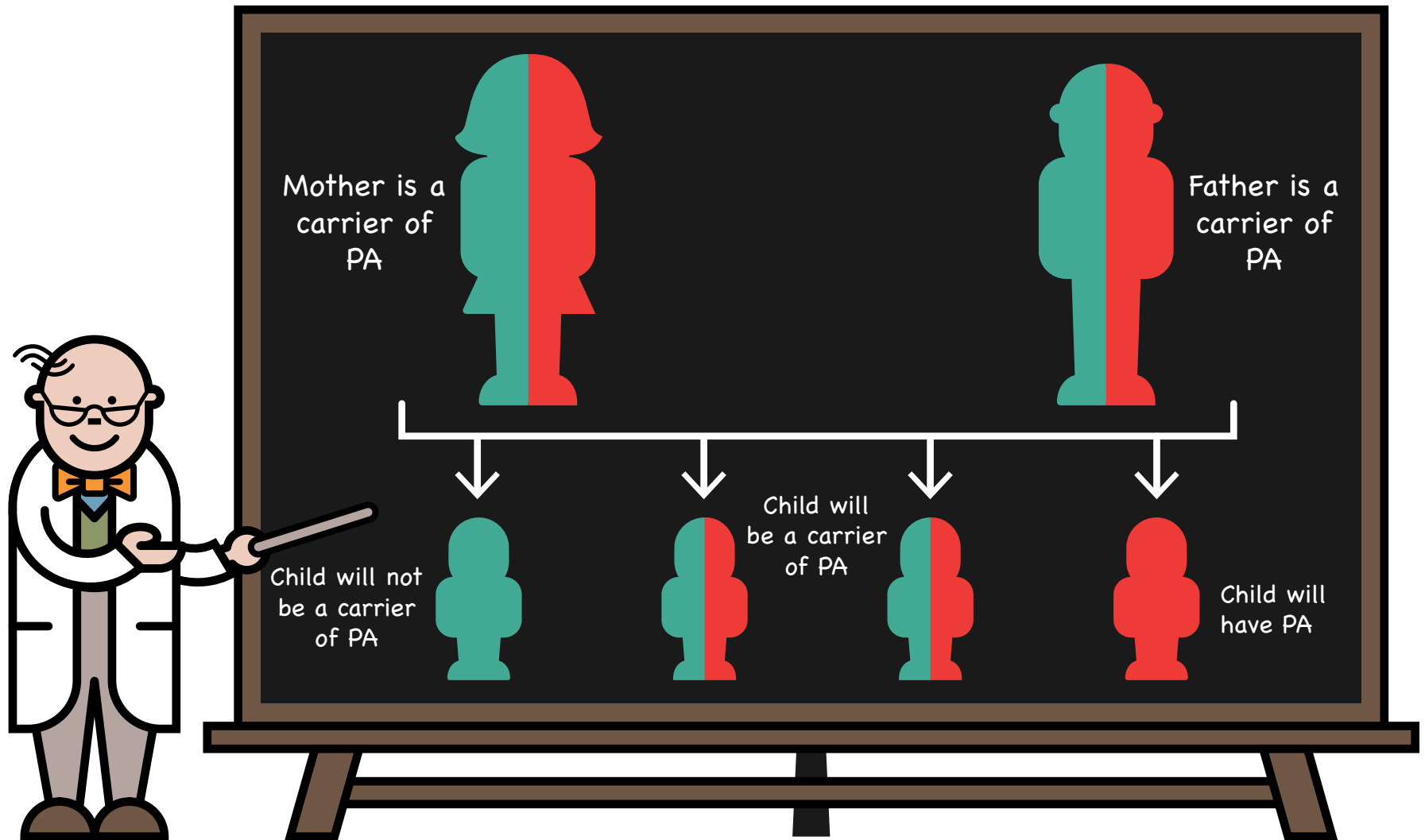
Parents of children with PA are carriers of the condition

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

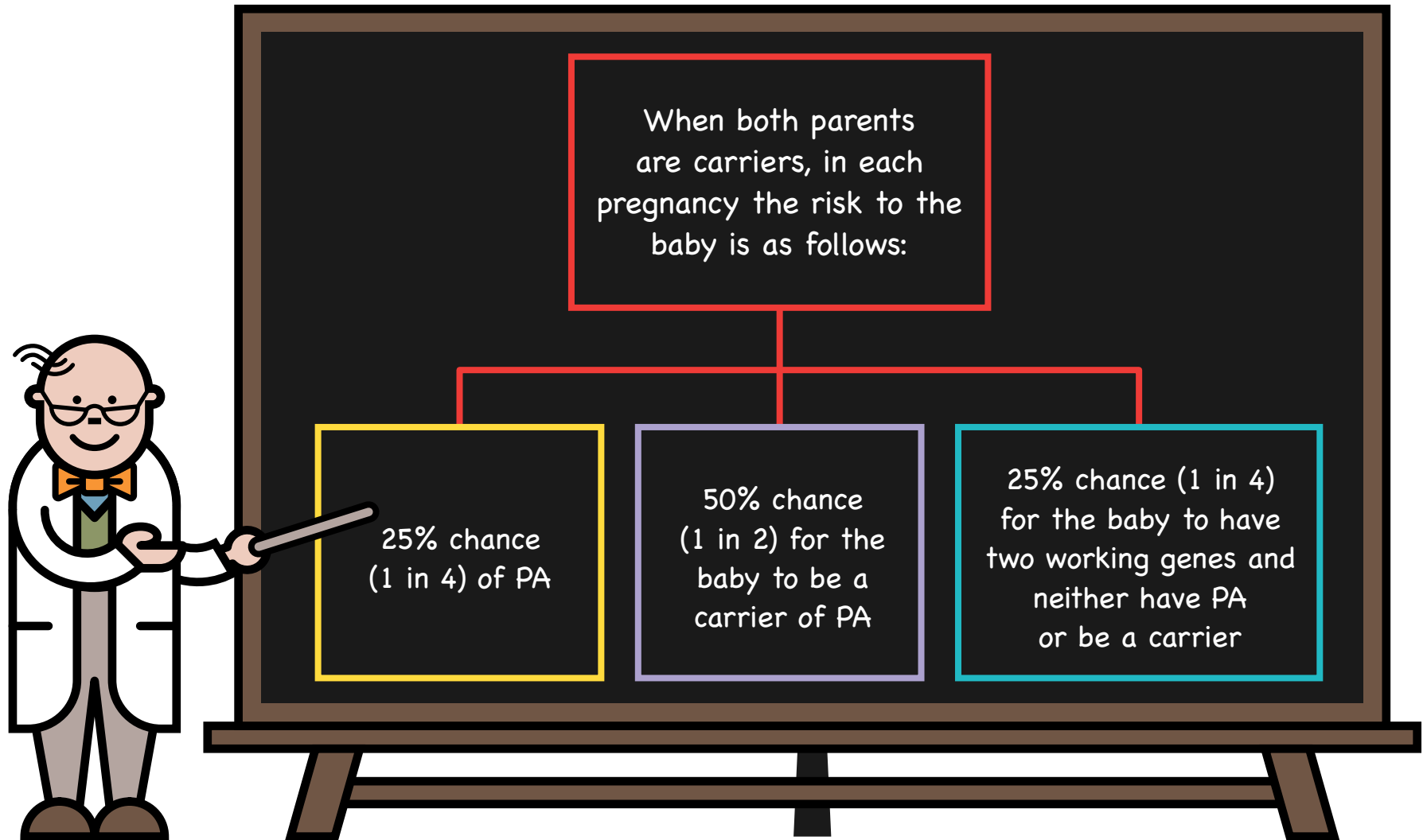
# Inheritance – Autosomal recessive (carriers of PA)



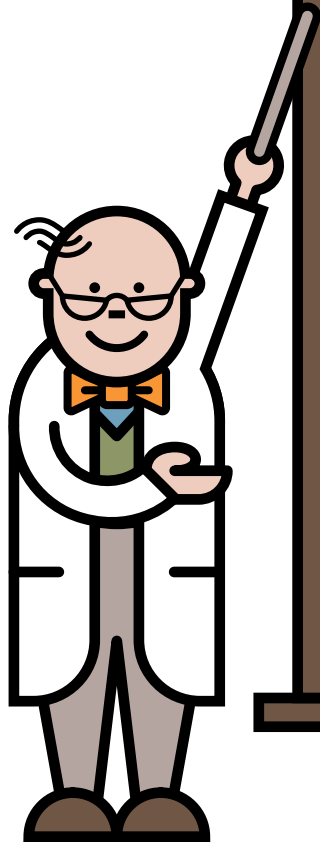
# Inheritance – Autosomal recessive – possible combinations



# Future pregnancies



# Take home messages



✓ PA is a serious inherited metabolic disorder that can lead to severe problems

The condition is managed with a protein restricted diet, regular feeding, and medications

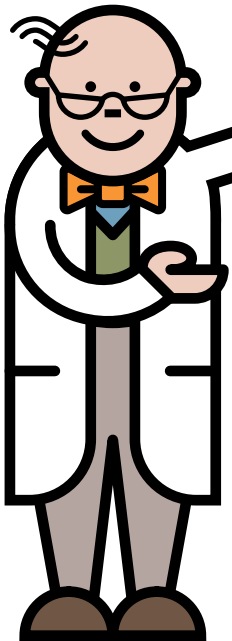
Remember, during illness, it is imperative that emergency feeds are started promptly, followed strictly and there are no delays in treatment

# Take home messages

✓ PA is a serious inherited metabolic disorder that can lead to severe problems

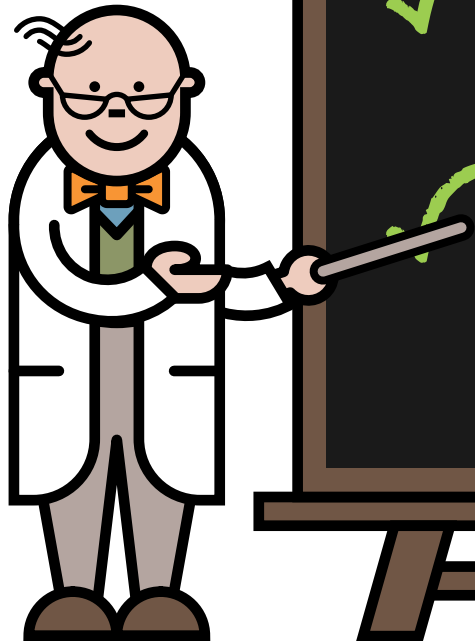
✓ The condition is managed with a protein restricted diet, regular feeding, and medications

Remember, during illness, it is imperative that emergency feeds are started promptly, followed strictly and there are no delays in treatment





# Take home messages

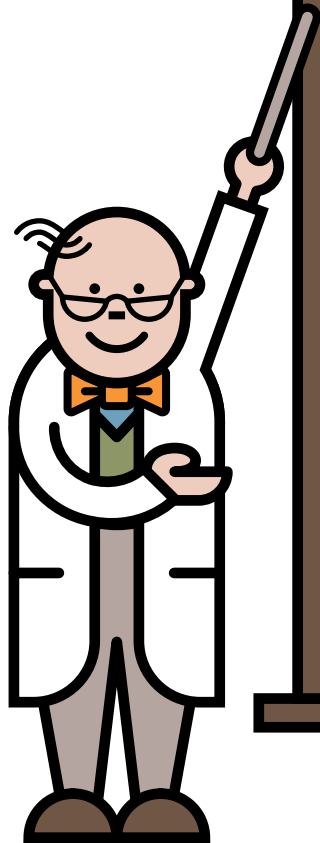


✓ PA is a serious inherited metabolic disorder that can lead to severe problems

✓ The condition is managed with a protein restricted diet, regular feeding, and medications

✓ Remember, during illness, it is imperative that emergency feeds are started promptly, followed strictly and there are no delays in treatment

# Helpful hints



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

Your dietary products and medications are prescribed. These are obtained via a pharmacy or home delivery

Always ensure you have your emergency feed products and a written emergency plan

Medications to control fever should be given as normally recommended – always keep supplies available

# Helpful hints



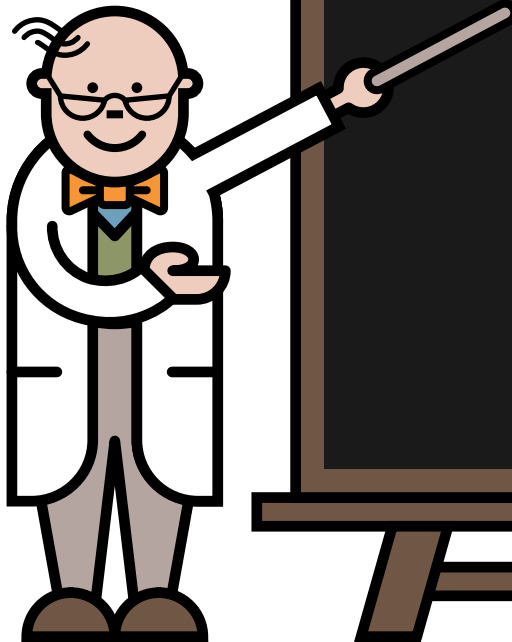
Always ensure you have a good supply of your dietary products and medicines and that they are in date



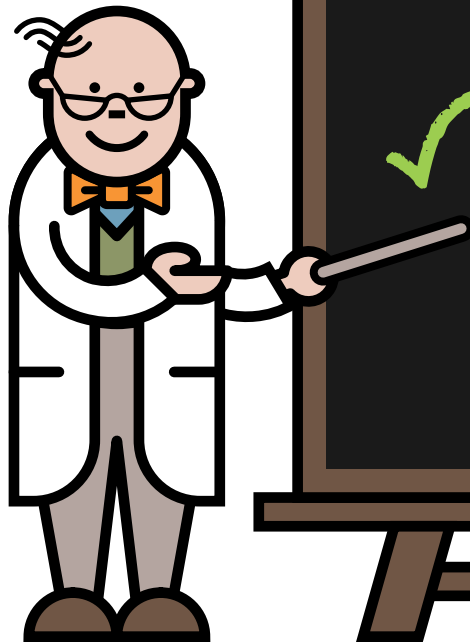
Your dietary products and medications are prescribed. These are obtained via a pharmacy or home delivery

Always ensure you have your emergency feed products and a written emergency plan

Medications to control fever should be given as normally recommended – always keep supplies available



# Helpful hints



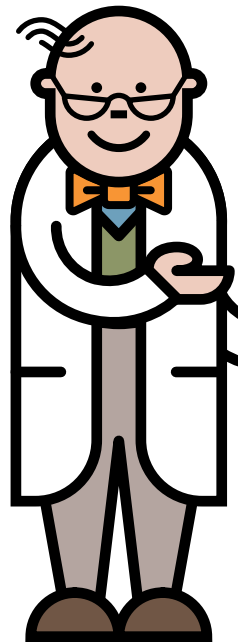
✓ Always ensure you have a good supply of your dietary products and medicines and that they are in date

✓ Your dietary products and medications are prescribed. These are obtained via a pharmacy or home delivery

✓ Always ensure you have your emergency feed products and a written emergency plan

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 medicines and that they are in date

✓ Your dietary products and medications are prescribed. These are obtained via a pharmacy or home delivery

✓ Always ensure you have your emergency feed products and a written emergency plan

✓ Medications to control fever should be given as normally recommended – always keep supplies available

# 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.



**BIMDG**  
British Inherited Metabolic Diseases Group



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

**NUTRICIA**  
Advanced Medical Nutrition

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



**Climb**<sup>TM</sup>

Children Living with  
Inherited Metabolic Diseases

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