

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 3, APRIL 2020

PA

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.

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).

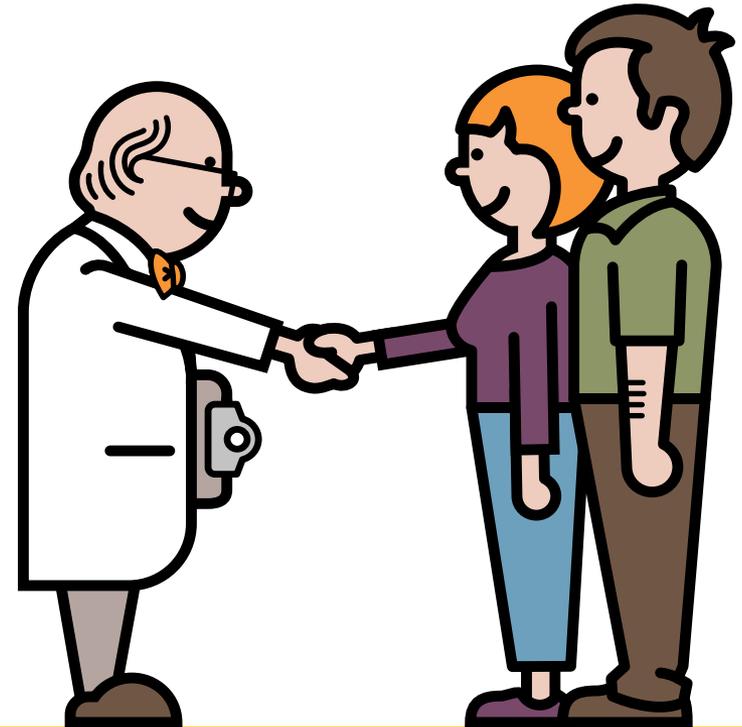
The team are Rachel Skeath, Karen van Wyk, Pat Portnoi and Anita MacDonald. The group is facilitated by Heidi Chan from Nutricia.

Each module produced is reviewed by a consultant clinician who is a member of the BIMDG.

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

PA

Information for parents following
a new diagnosis



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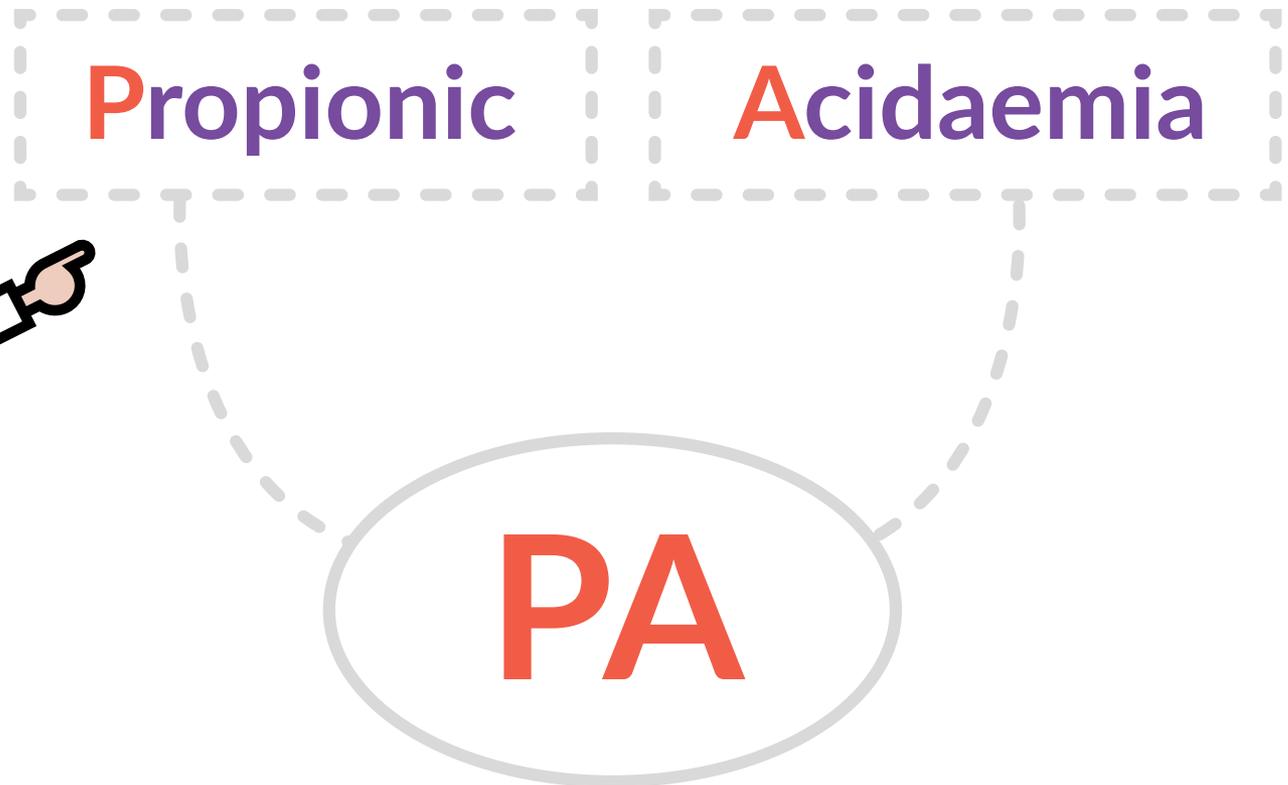
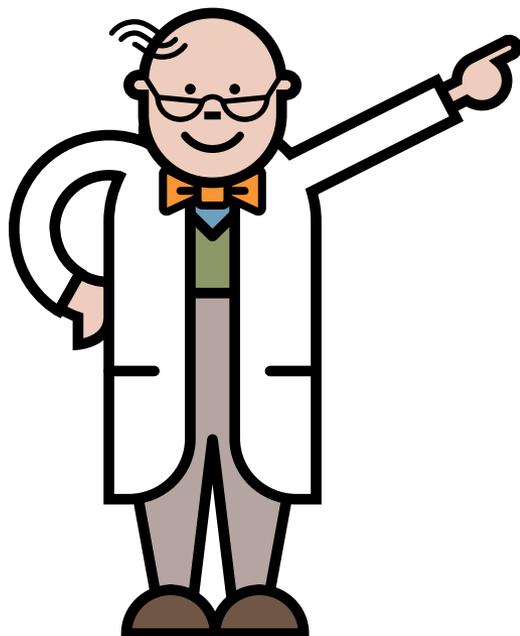
Tools Enabling Metabolic Parents LEarning

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

What is PA?

PA stands for Propionic Acidaemia

It is an inherited metabolic condition

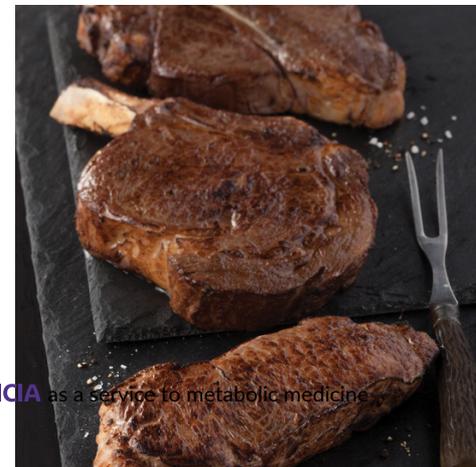
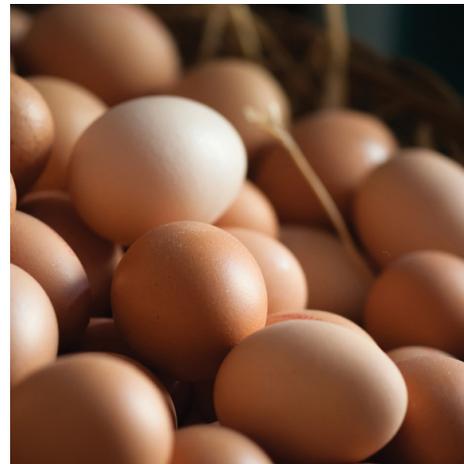


PA and protein

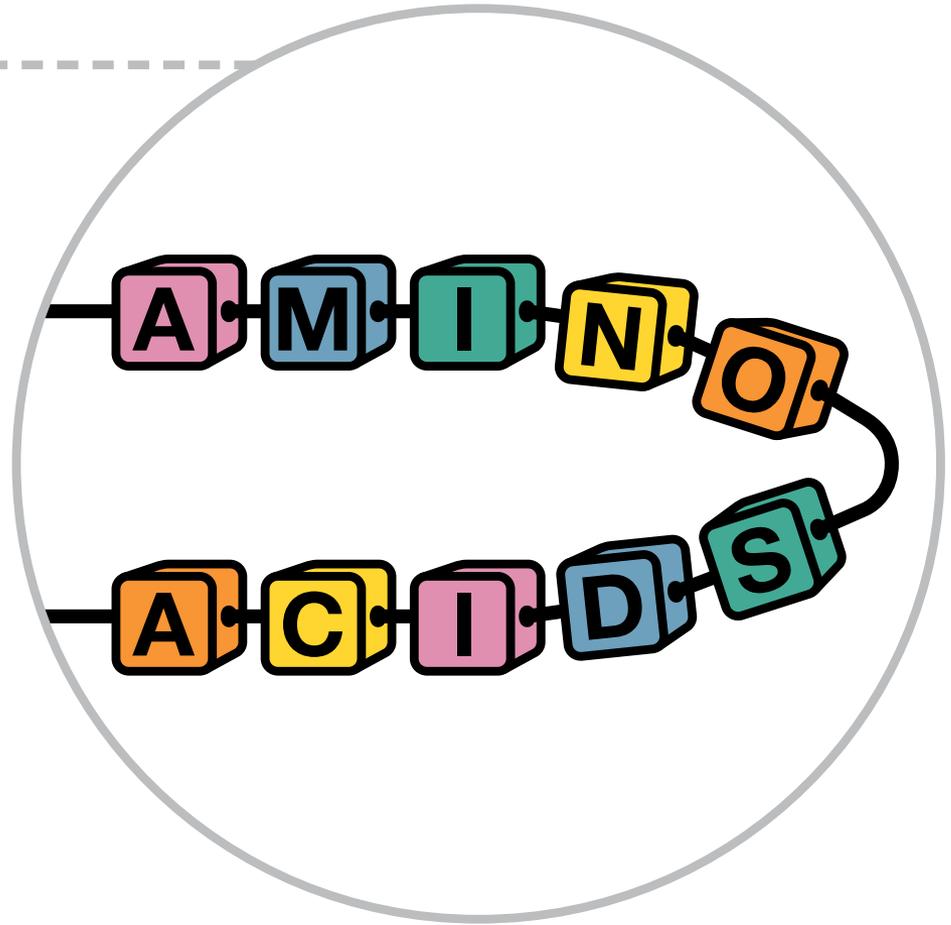
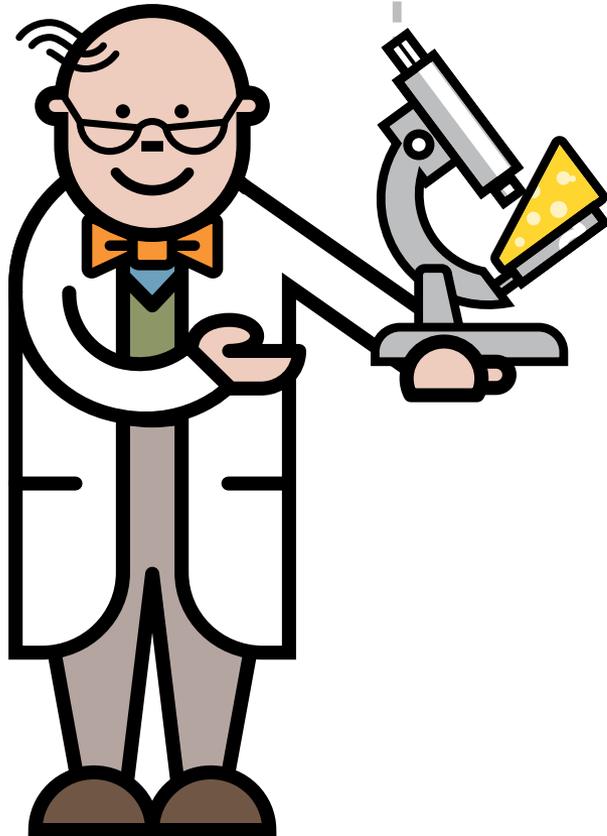
PA affects the way your baby breaks down protein

Many foods contain protein

The body needs protein for growth and repair



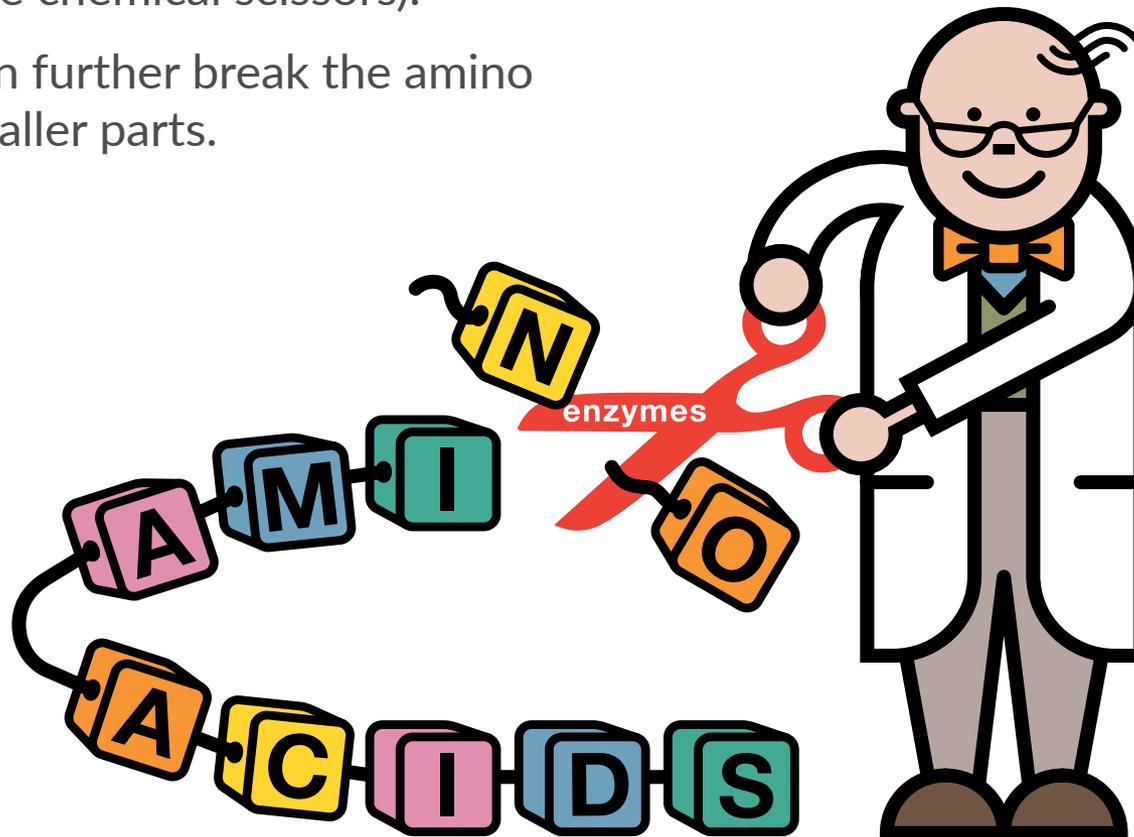
What is protein?



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.

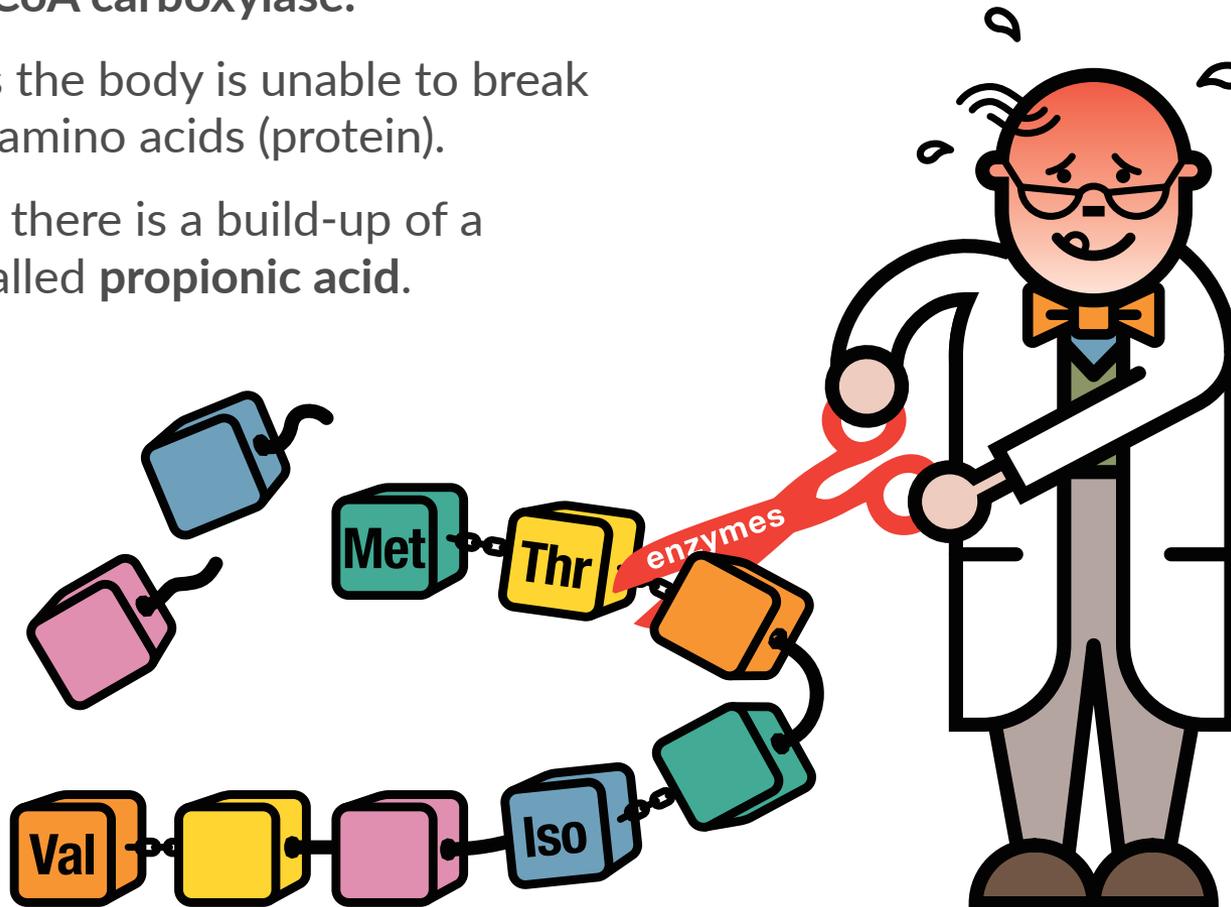


What happens in PA?

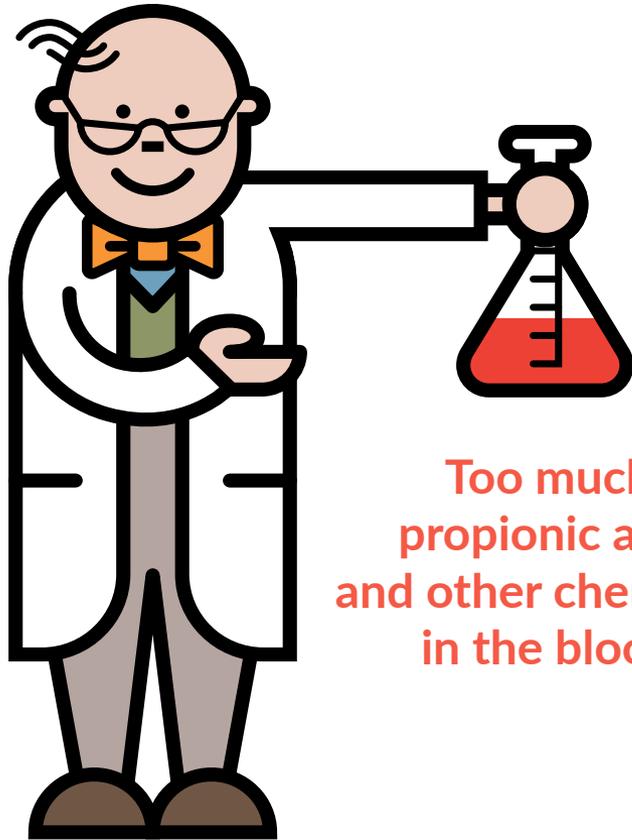
In PA, the body lacks an enzyme called **propionyl-CoA carboxylase**.

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

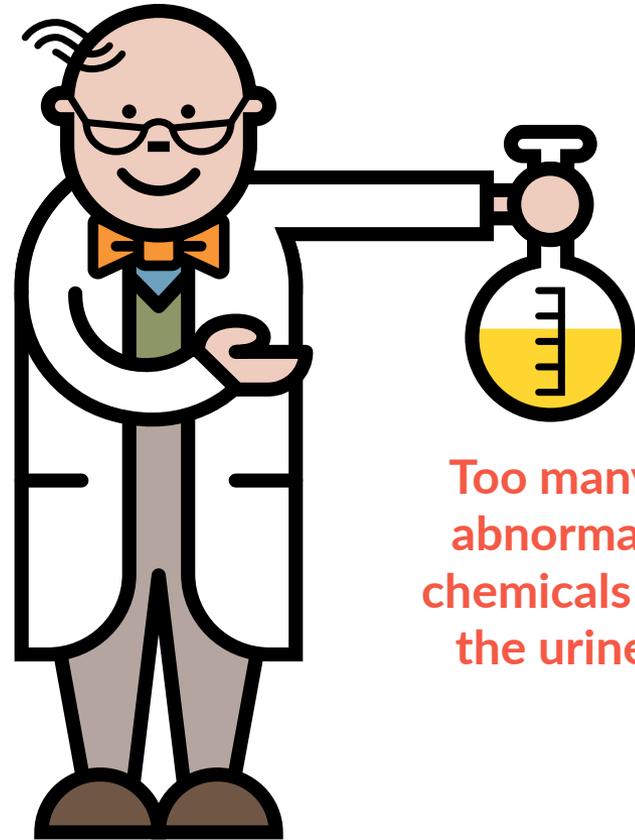
As a result, there is a build-up of a chemical called **propionic acid**.



What does this cause?



Too much propionic acid and other chemicals in the blood

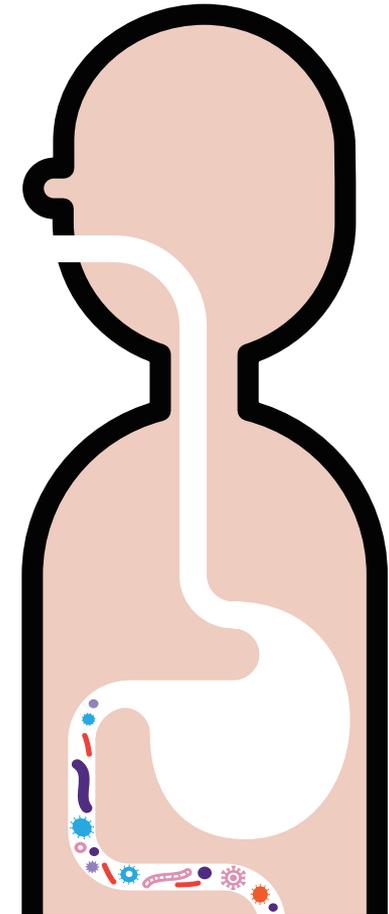


Too many abnormal chemicals in the urine

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.



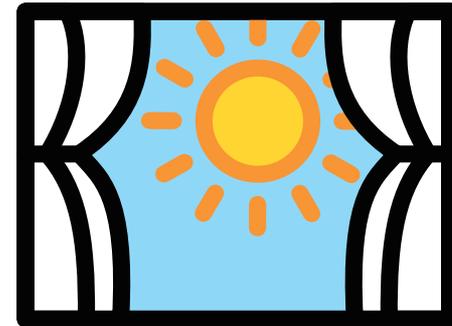
What are the symptoms in PA?

Symptoms commonly start in the first few days of life.

Symptoms include:

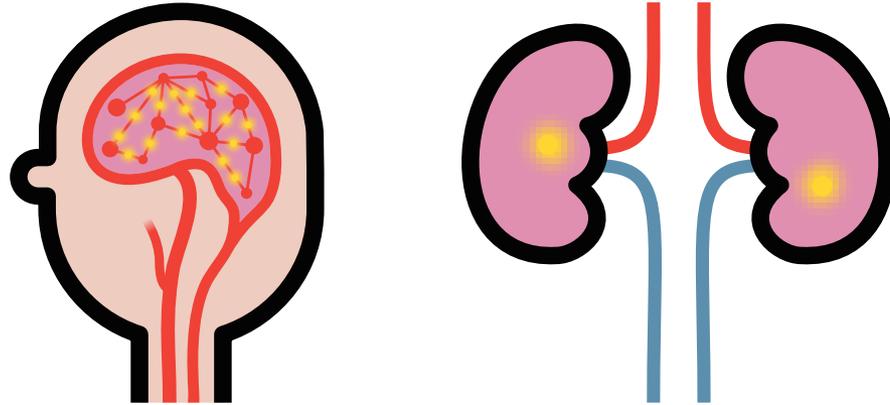
- poor feeding
- vomiting
- dehydration (lack of body fluids)
- floppy baby
- excessively sleepy
- rapid breathing
- seizures

The effects of PA quickly become life-threatening if unmanaged

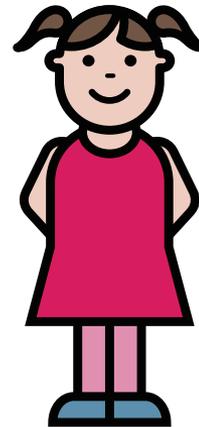


What can go wrong in PA?

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



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

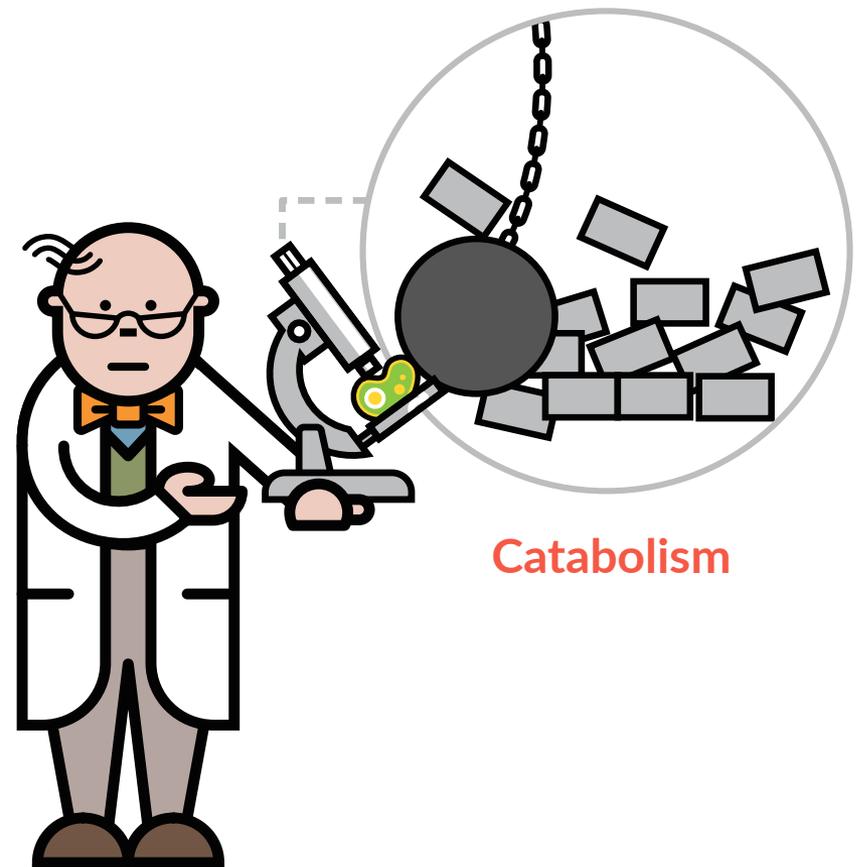


What else happens in PA?

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.



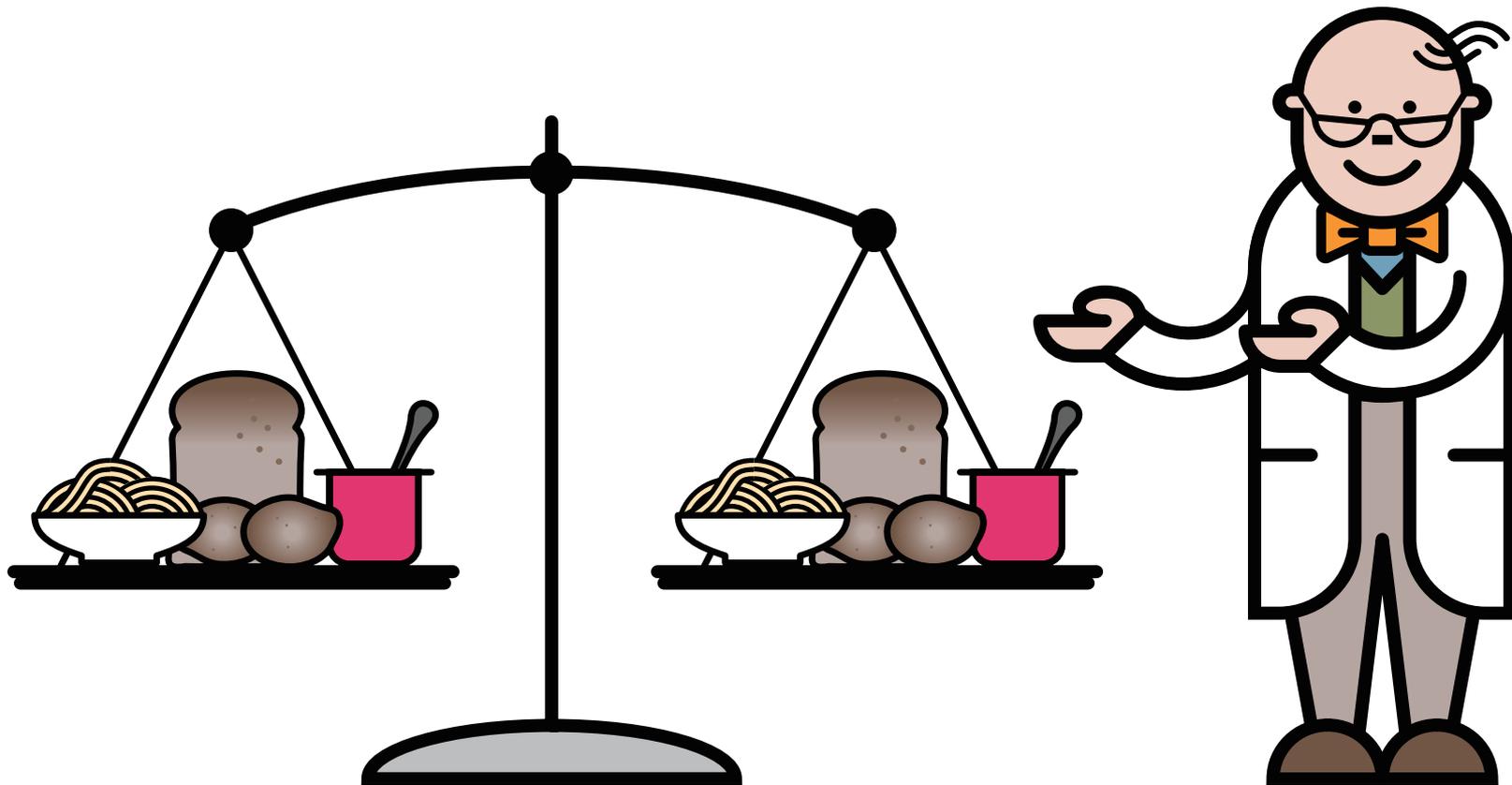
Metabolic crisis

- 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 management
- 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.

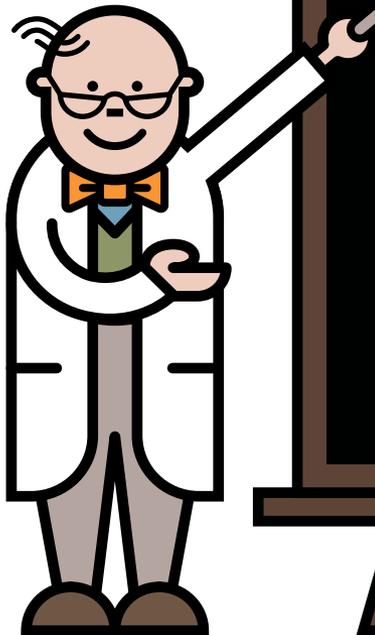


How is PA managed day to day?

PA is managed with the following:

A protein restricted diet

Ensuring a sufficient energy supply

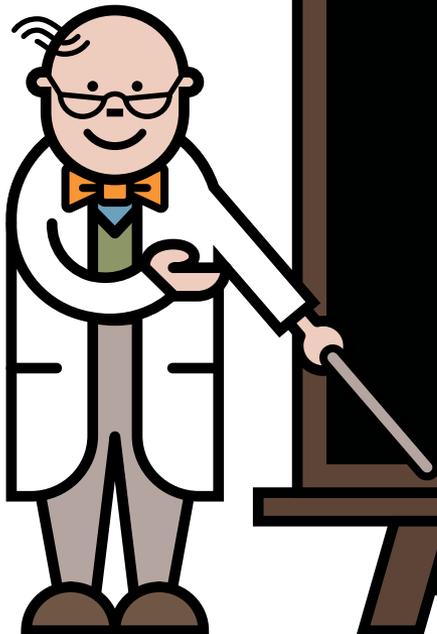


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How is PA managed day to day?

Regular feeding



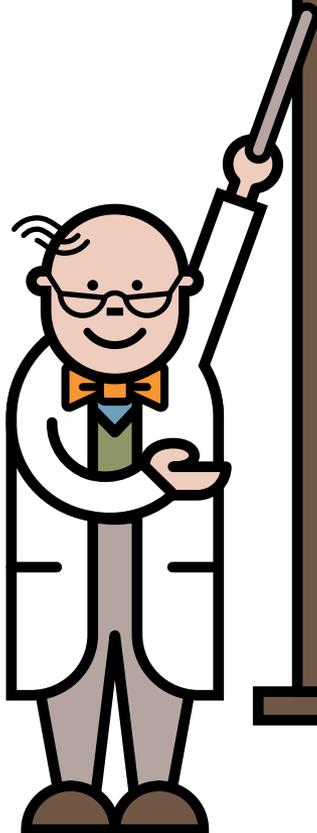
Carnitine medication



Antibiotics to control gut bacteria



Other medications may be necessary



How is PA managed day to day?

Regular feeding



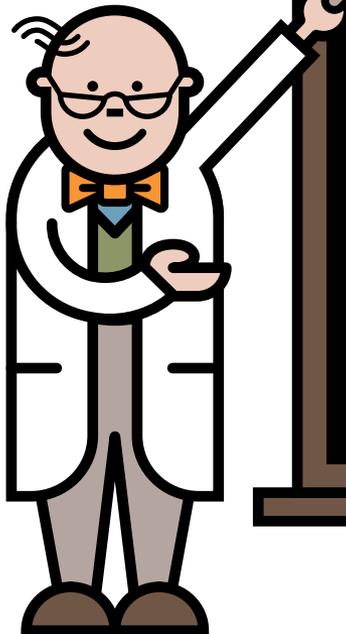
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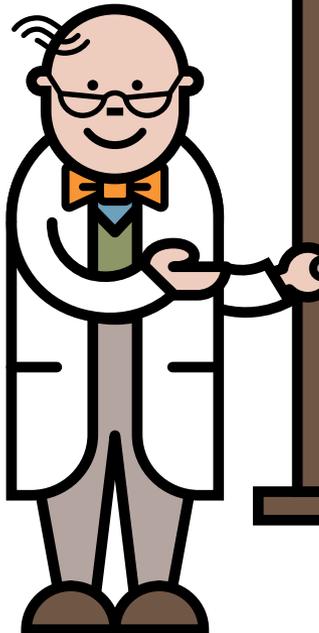
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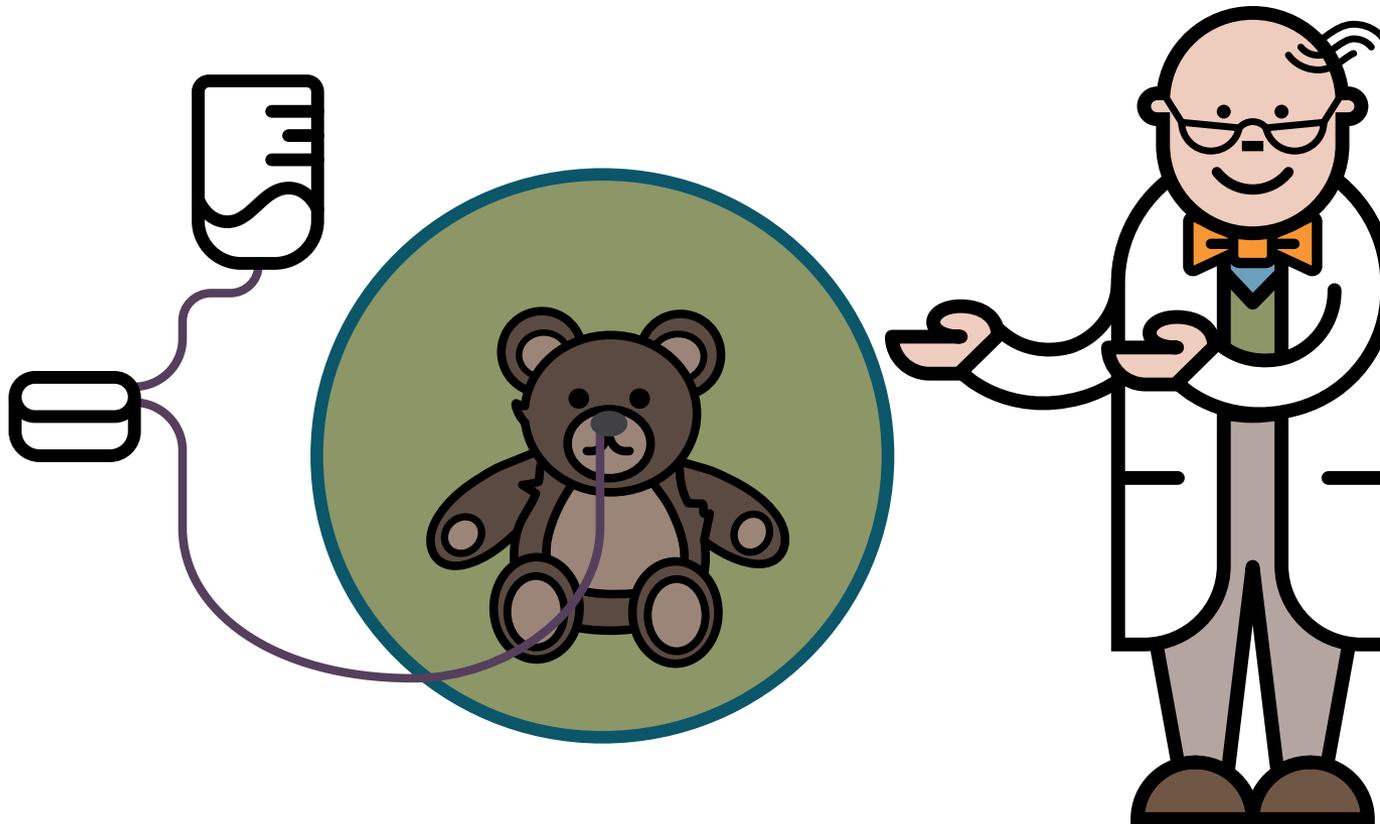


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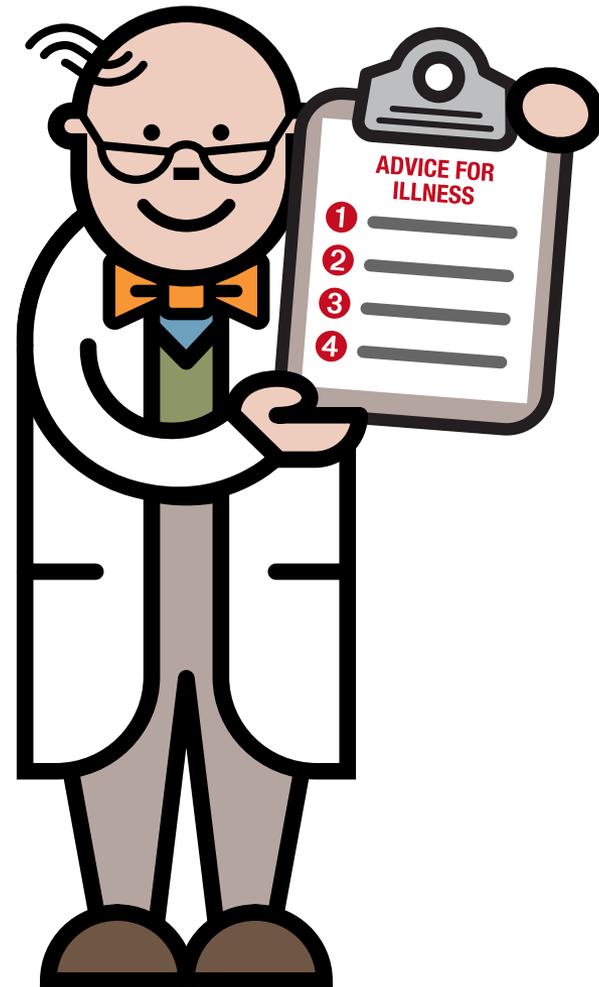
Is tube feeding needed?

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 avoid a lack of energy supply and build-up of harmful chemicals that cause a metabolic crisis



How is PA managed during illness?

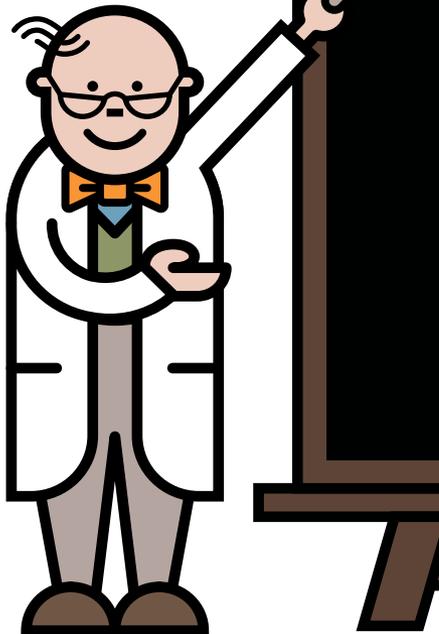
Stop all protein in food & drink



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



Continue medication as prescribed



How is PA managed during illness?

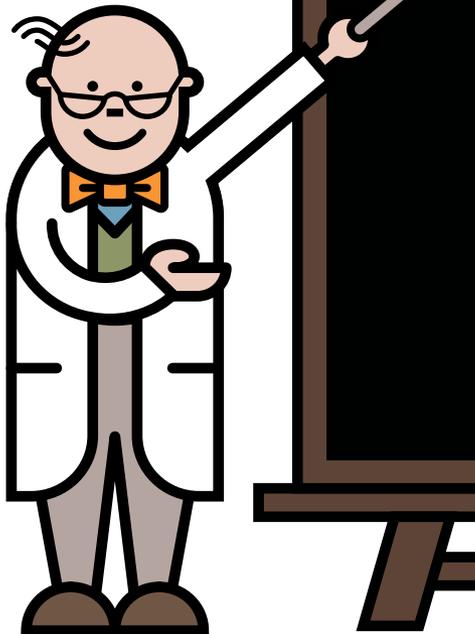
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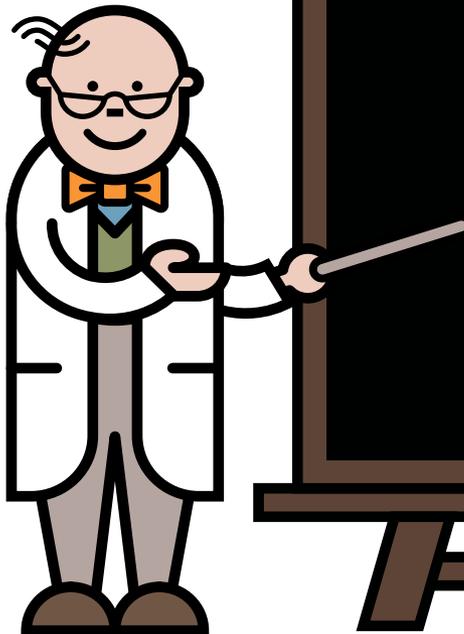
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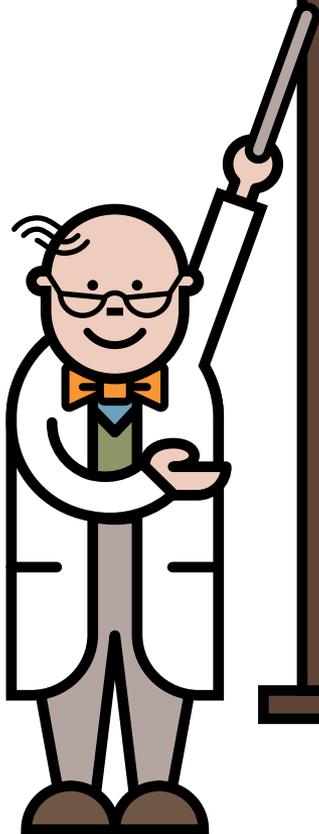
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Continue medication as prescribed



Checklist for illness



Always take full amounts
of emergency feeds as
prescribed



If symptoms continue and/or
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immediately to the hospital



Regularly update your
metabolic team



Checklist for illness



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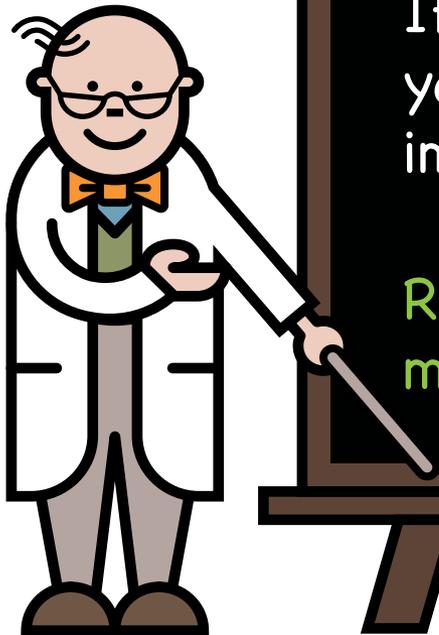
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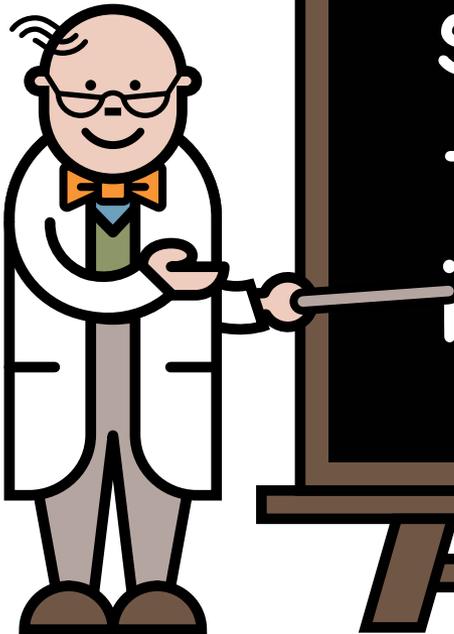


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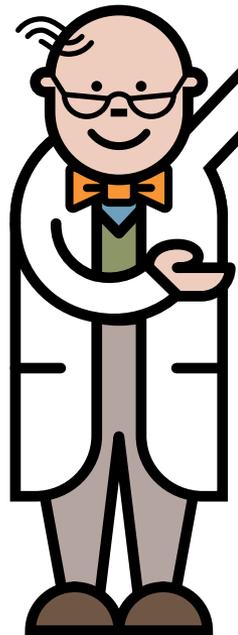


Key message

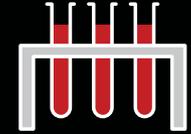
It is imperative that emergency feeds are started **promptly** and there are **no delays** in management.



How is PA monitored?



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



Height and weight

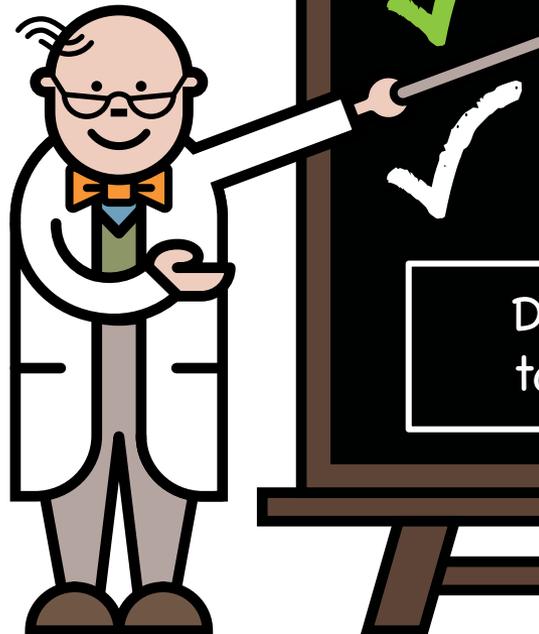


Developmental checks



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

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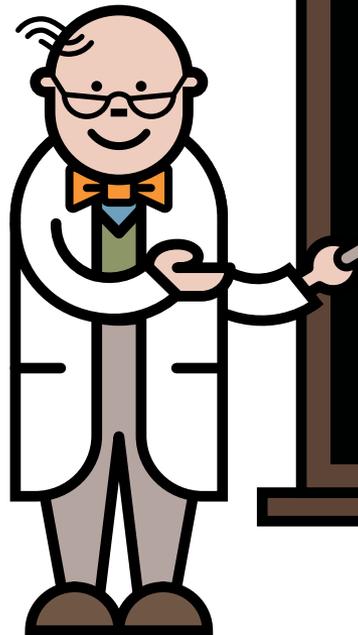


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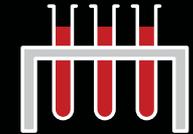
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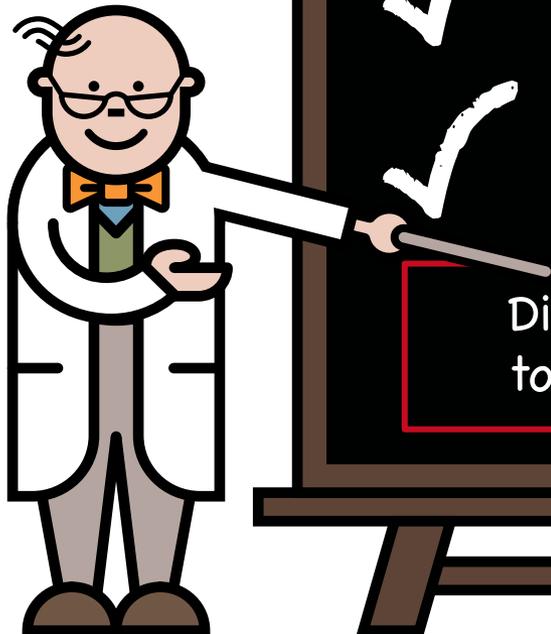
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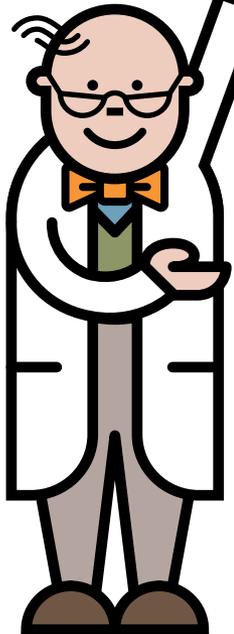
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Chromosomes, genes, mutations



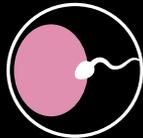
Humans have chromosomes composed of DNA



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

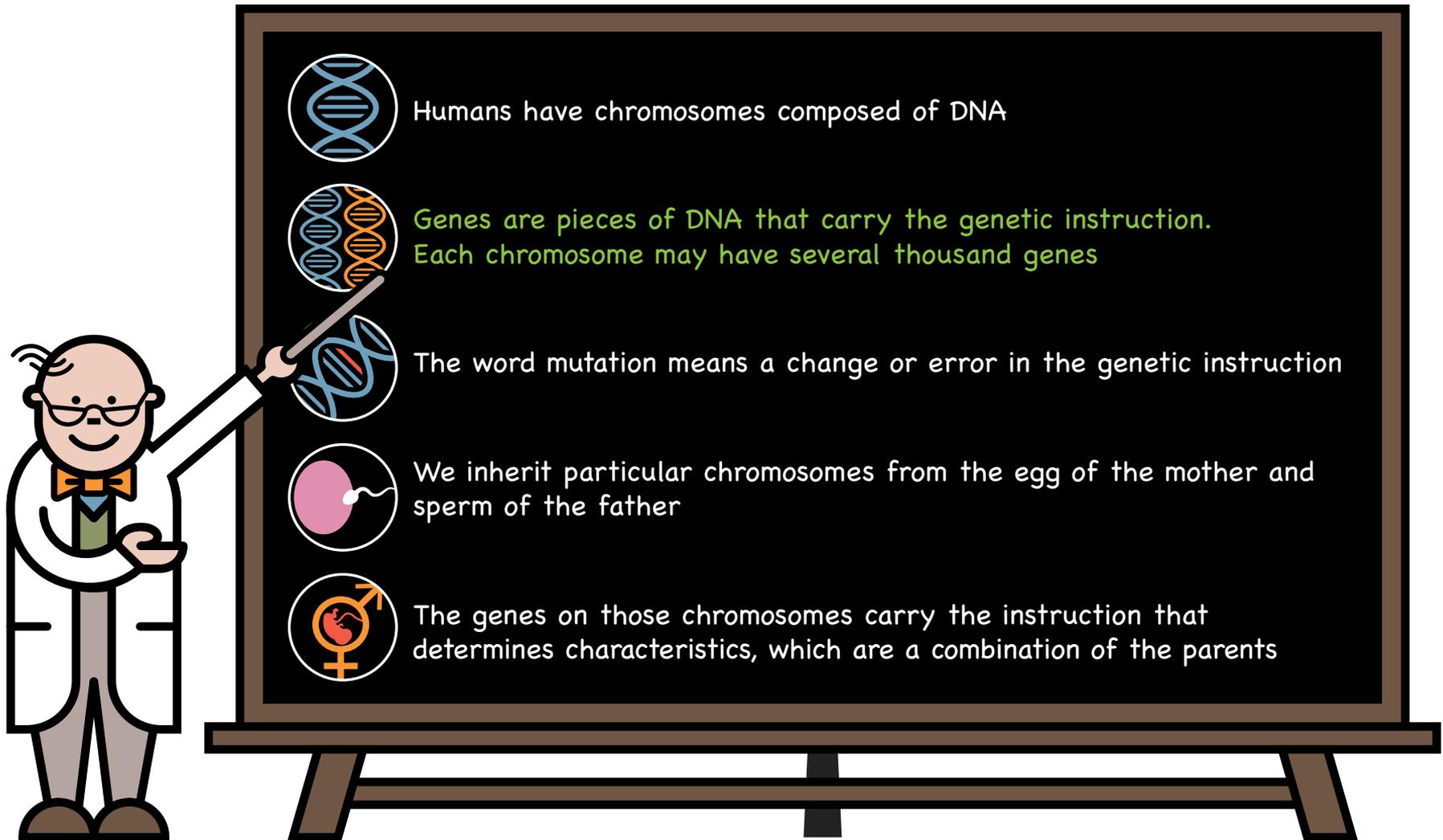


We inherit particular chromosomes from the egg of the mother and sperm of the father



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

Chromosomes, genes, mutations



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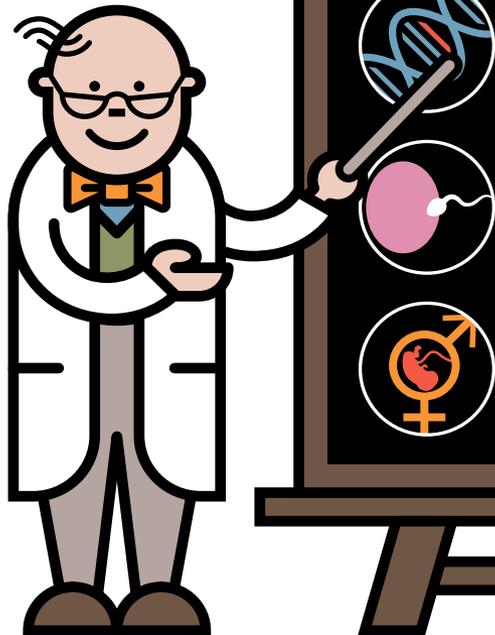
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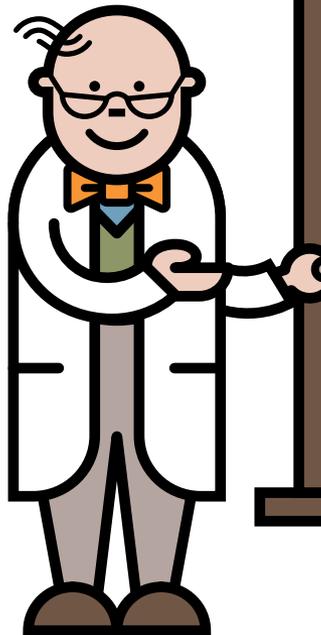


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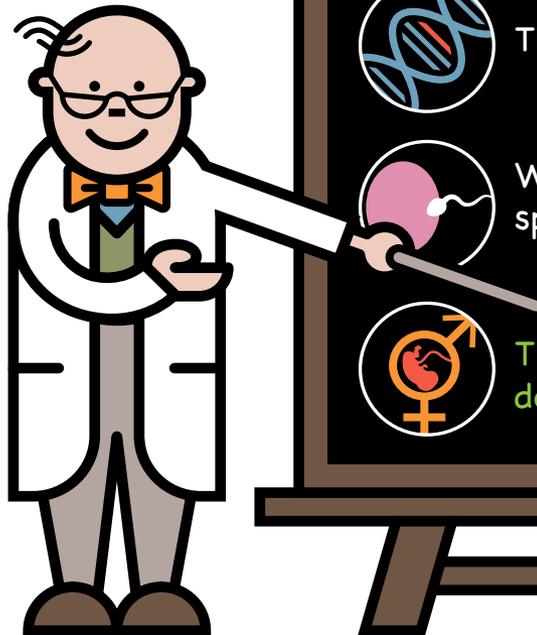


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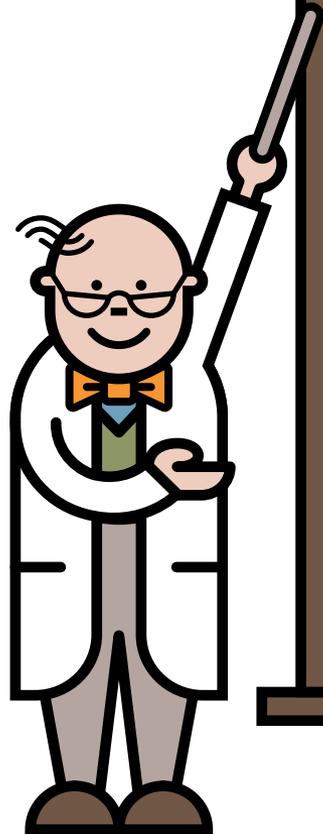


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Inheritance



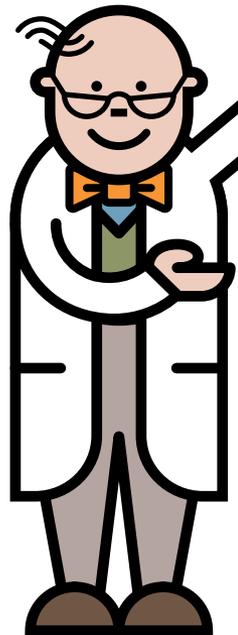
PA is an inherited condition. There is nothing that could have been done to prevent your baby from having PA

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



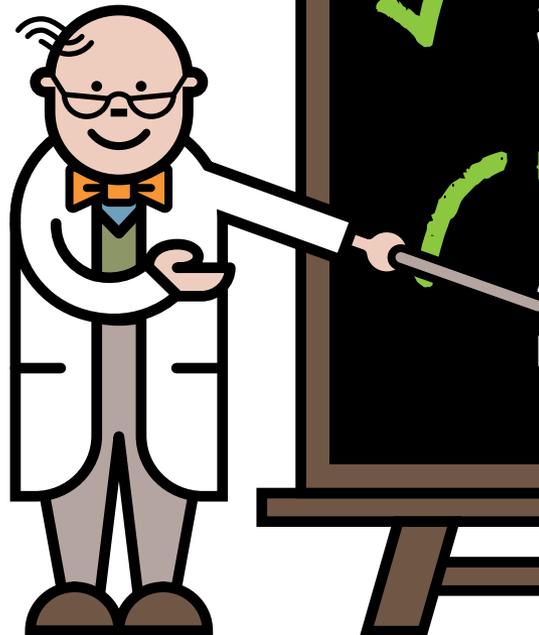
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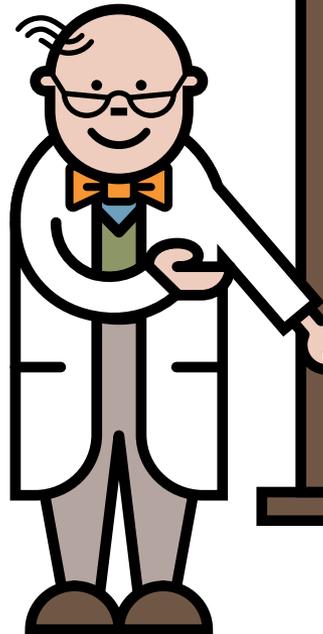
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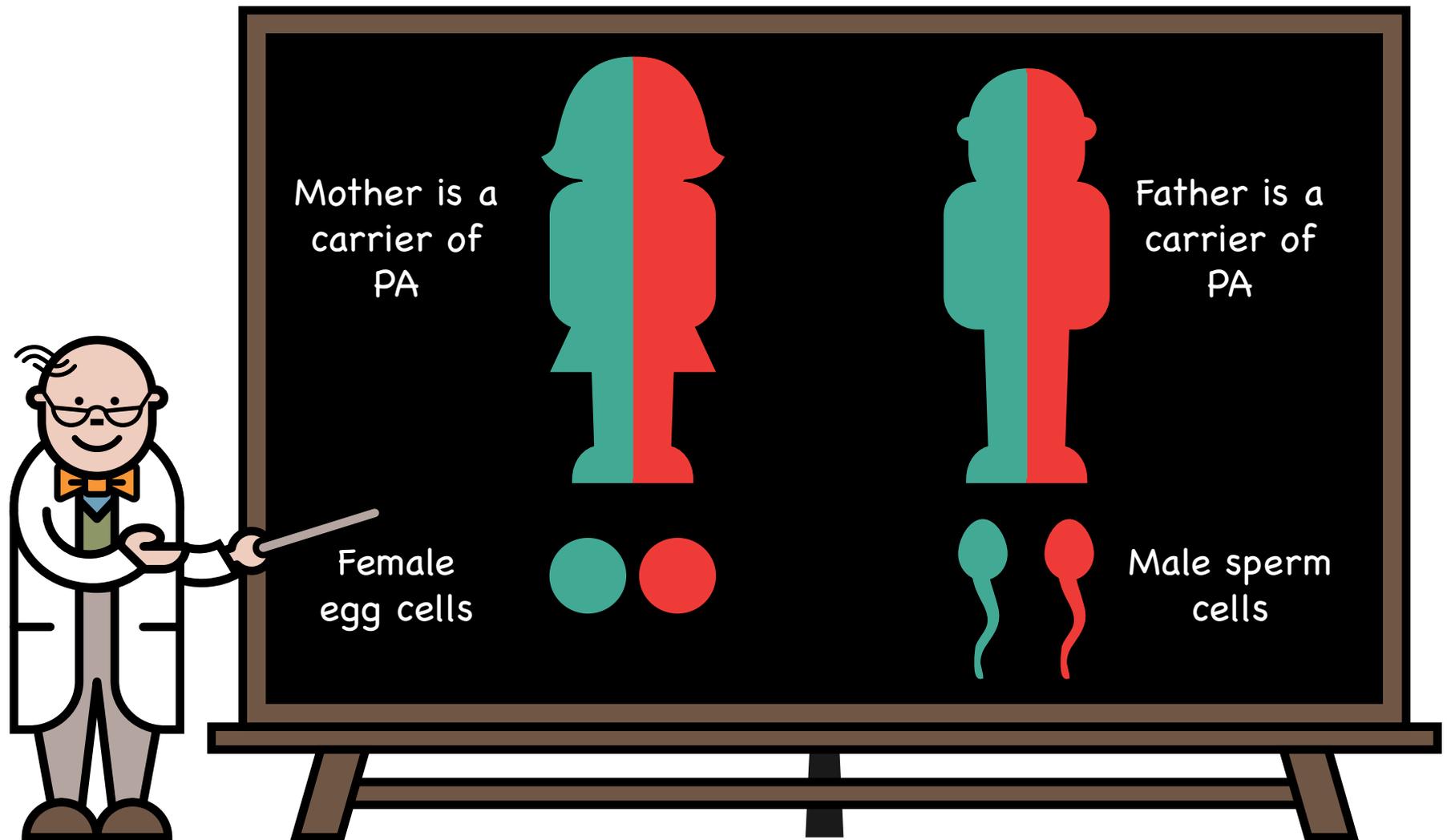
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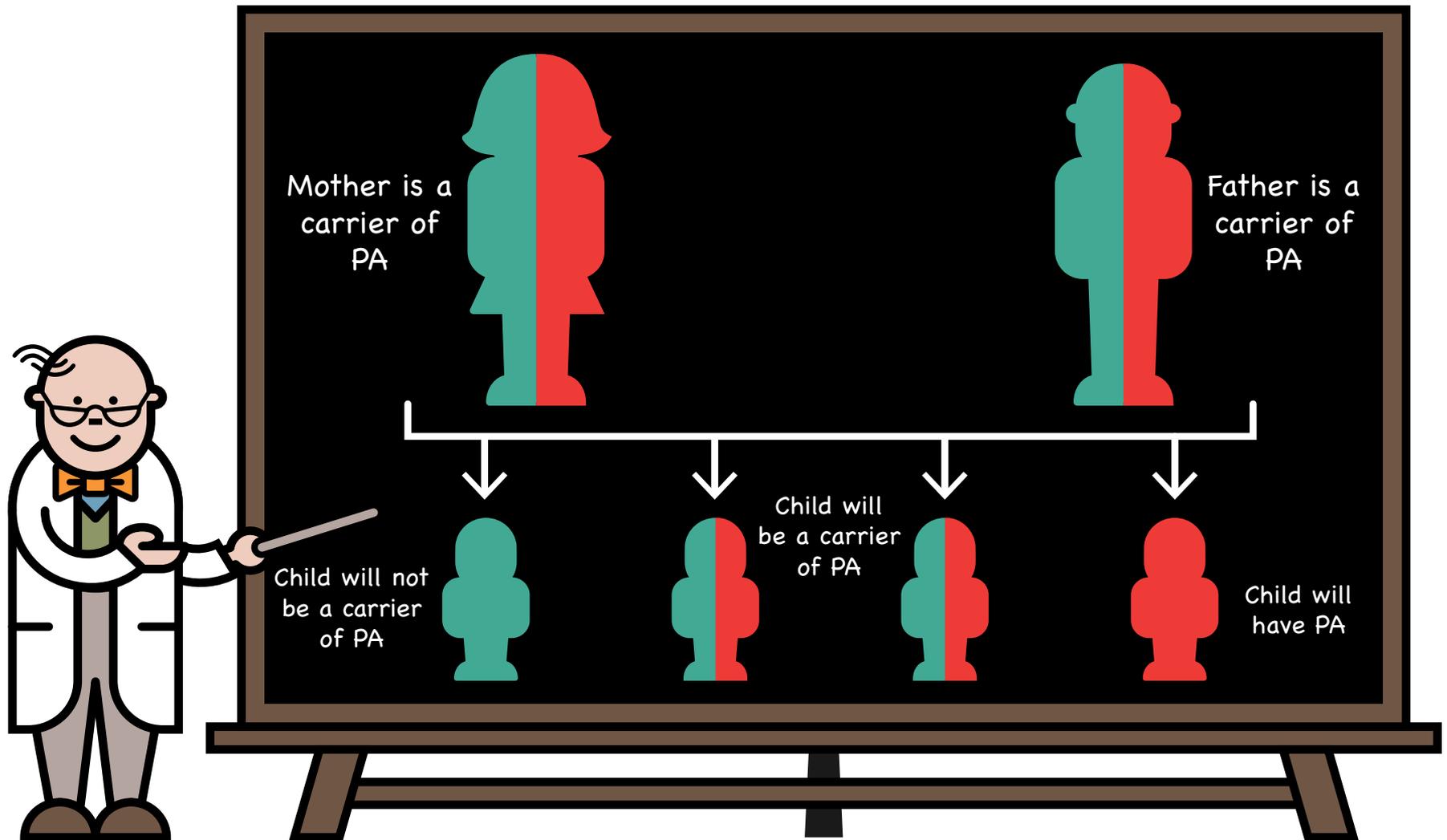
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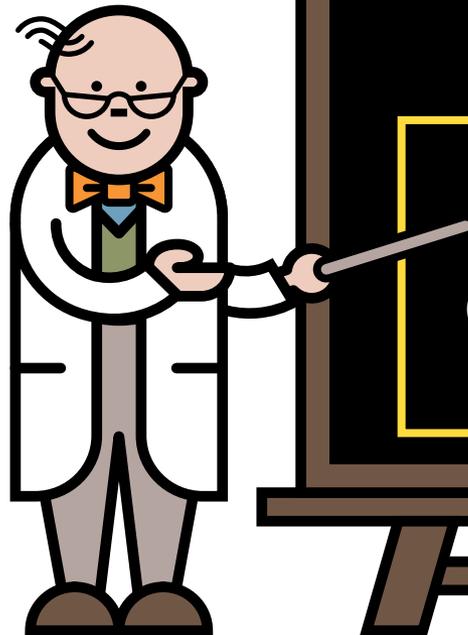
Inheritance – Autosomal recessive (carriers of PA)



Inheritance – Autosomal recessive – possible combinations



Future pregnancies



When both parents are carriers, in each pregnancy the risk to the baby is as follows:

25% chance
(1 in 4) of PA

50% chance
(1 in 2) for the baby to be a carrier of PA

25% chance (1 in 4) for the baby to have two working genes and neither have PA or be a carrier

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 management

Take home messages

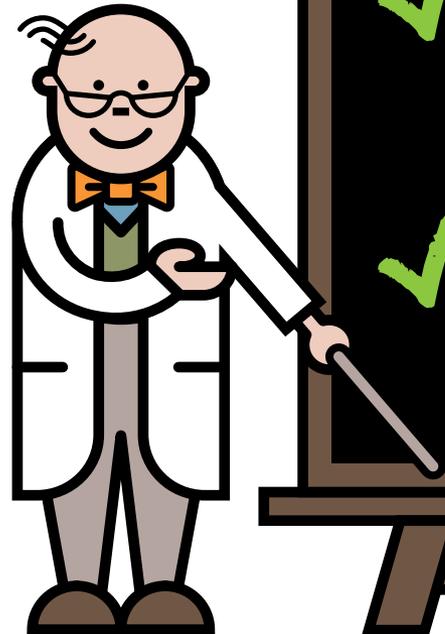
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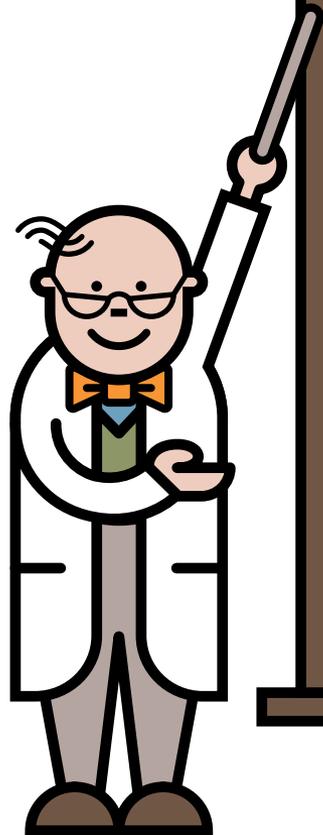


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Helpful hints



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



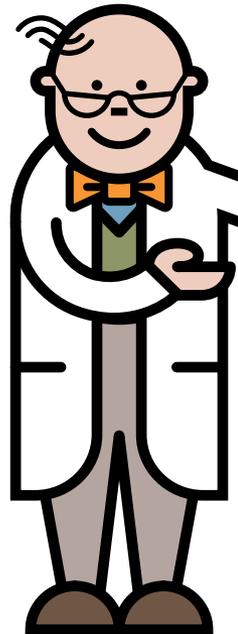
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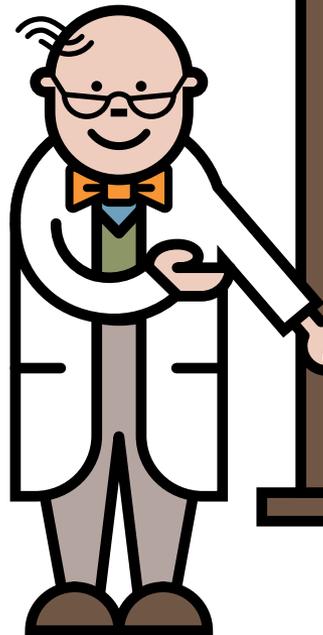
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Who's who

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

Visit www.lowproteinconnect.com 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.



www.bimdg.org.uk

www.nutricia.co.uk

www.metabolicsupportuk.org