

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

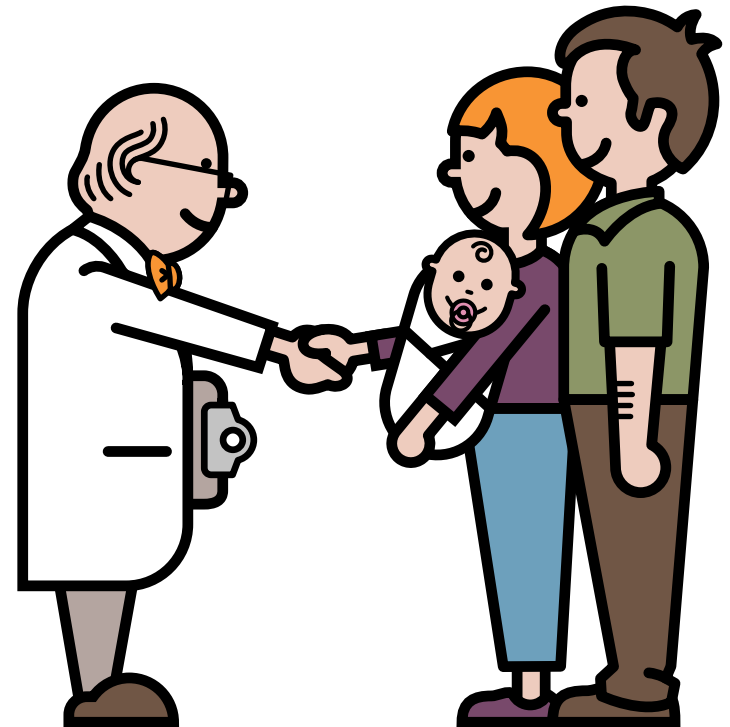
VERSION 2, FEBRUARY 2017

GA1

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

GA1

Information for families following
a positive newborn screening



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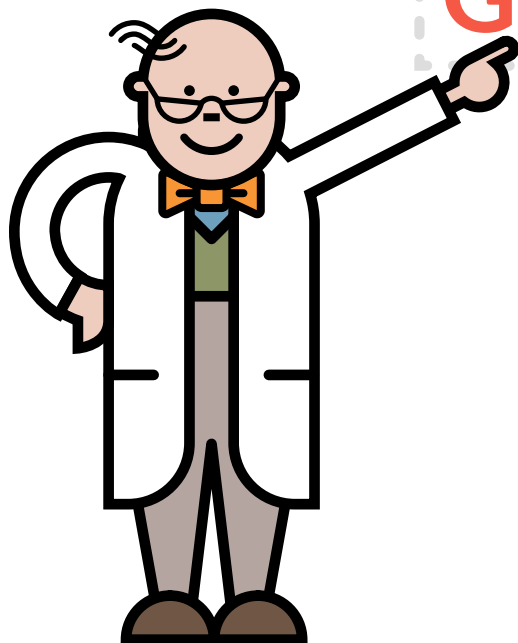
Tools Enabling Metabolic Parents LEarning

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

What is GA1?

GA1 stands for Glutaric Aciduria Type 1

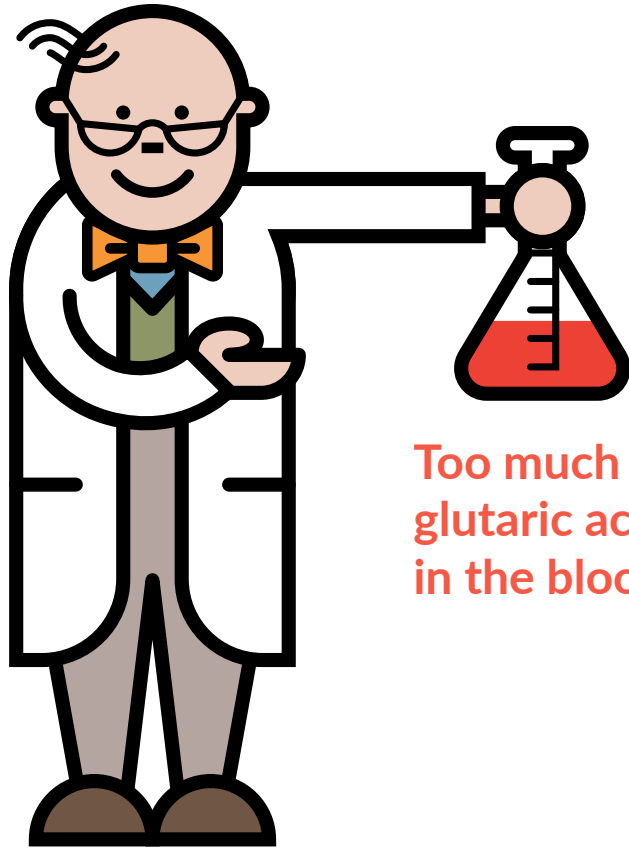
It is an inherited metabolic condition



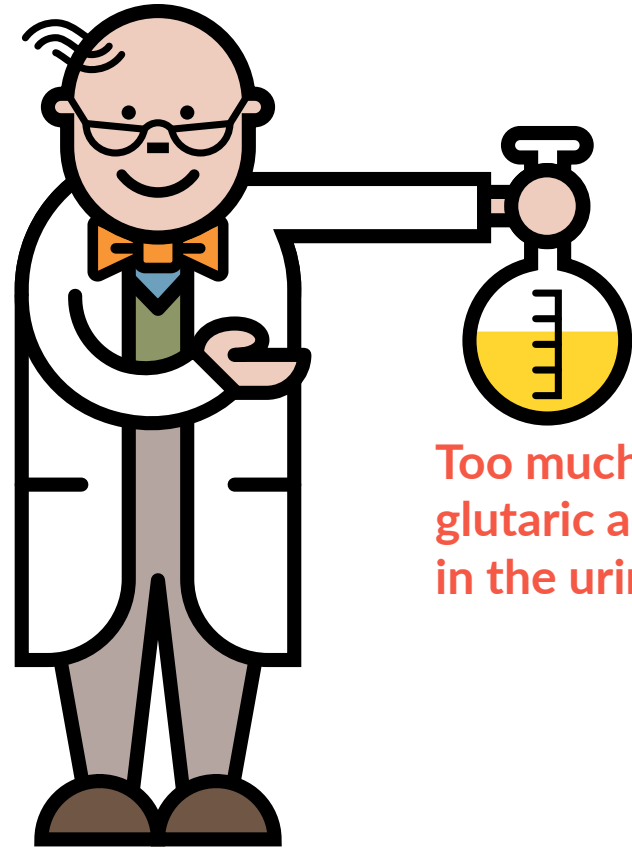
Glutaric Aciduria Type 1

GA1

What is GA1?



Too much
glutaric acid
in the blood



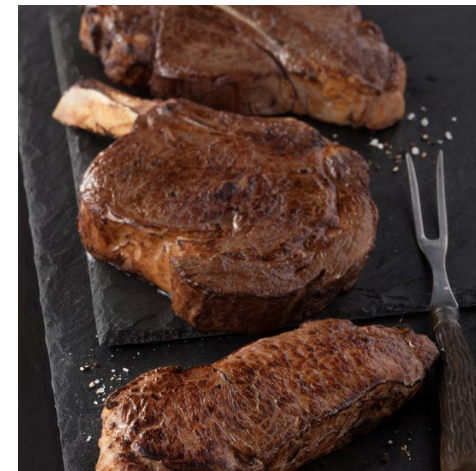
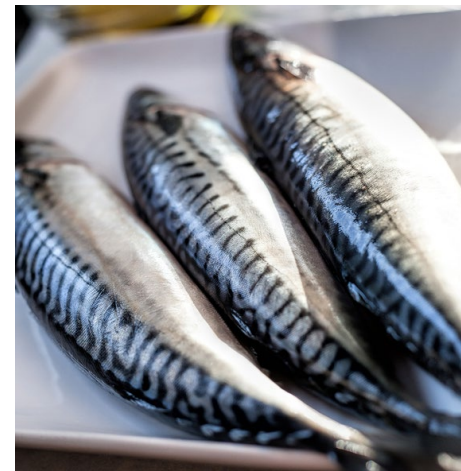
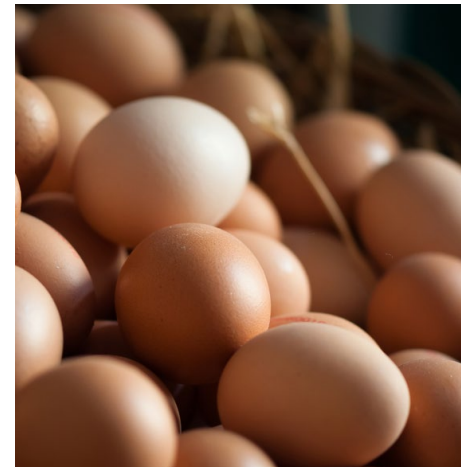
Too much
glutaric acid
in the urine

GA1 and protein

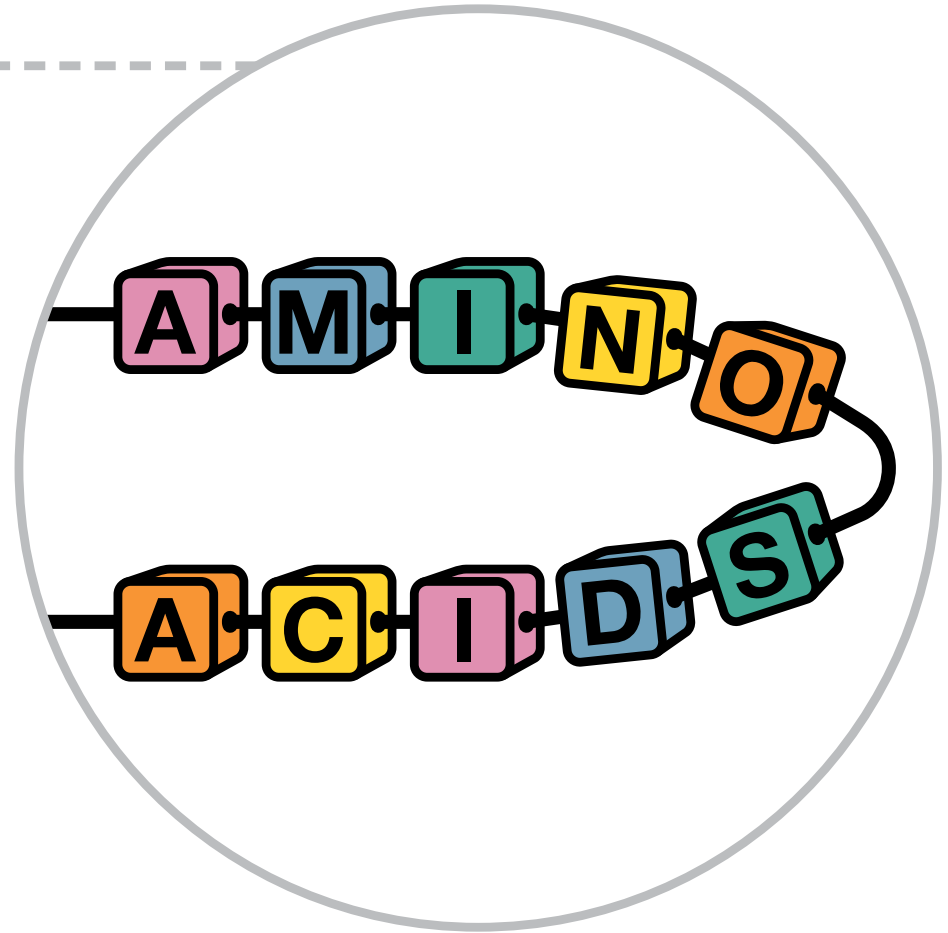
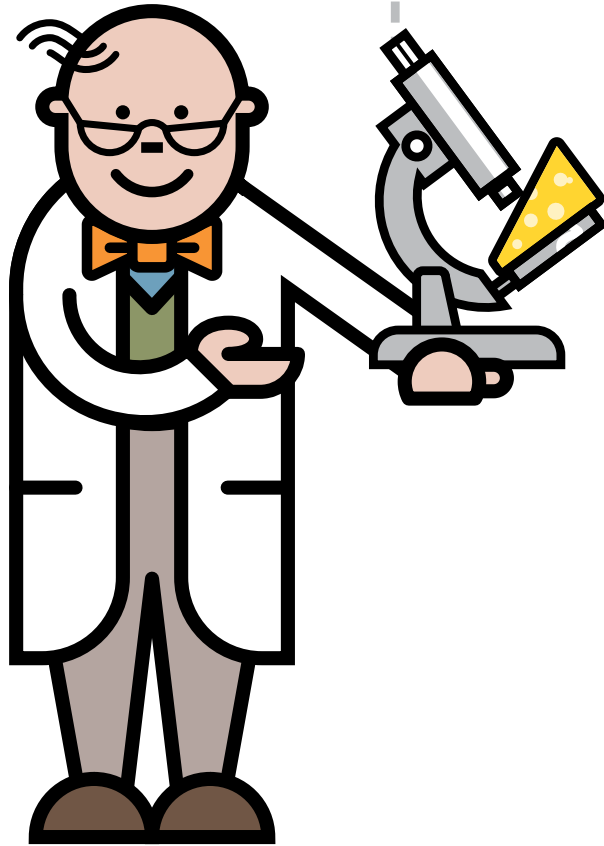
GA1 affects the way your baby breaks down protein

Many foods contain protein

The body needs protein for growth and repair



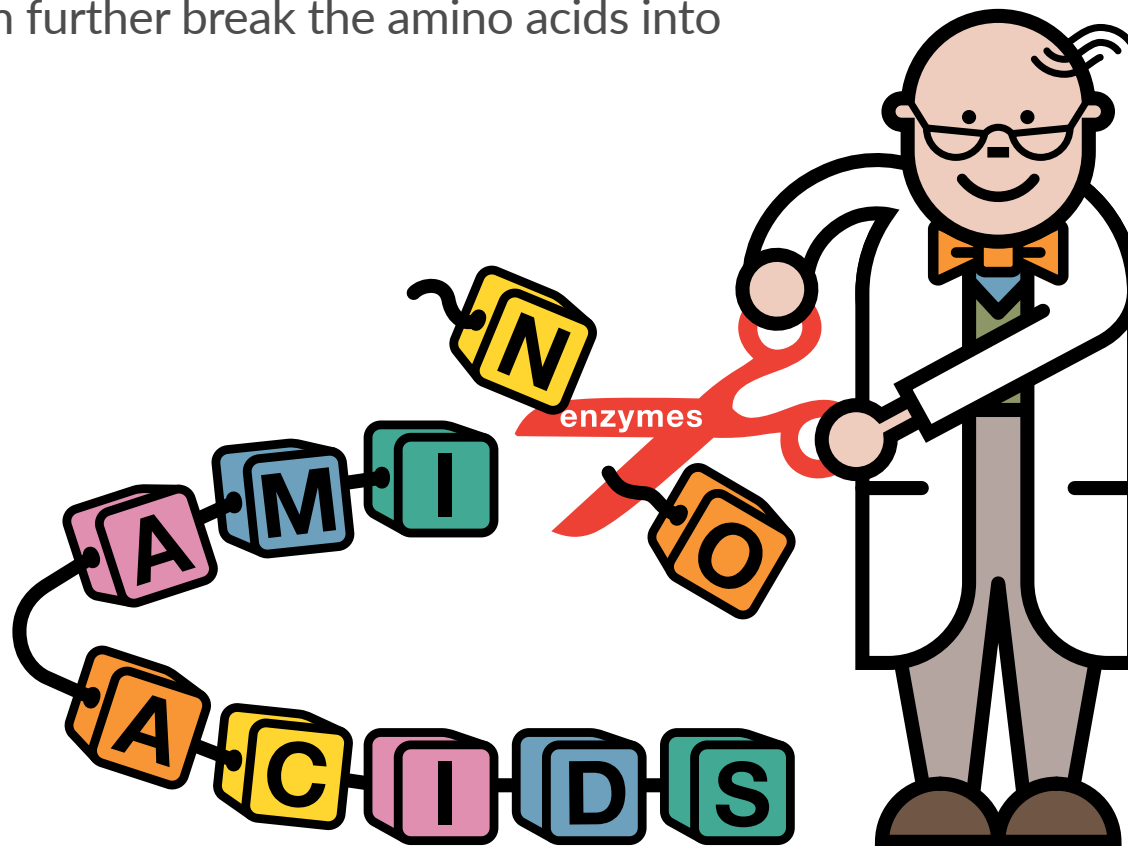
What is protein?



Protein and enzymes

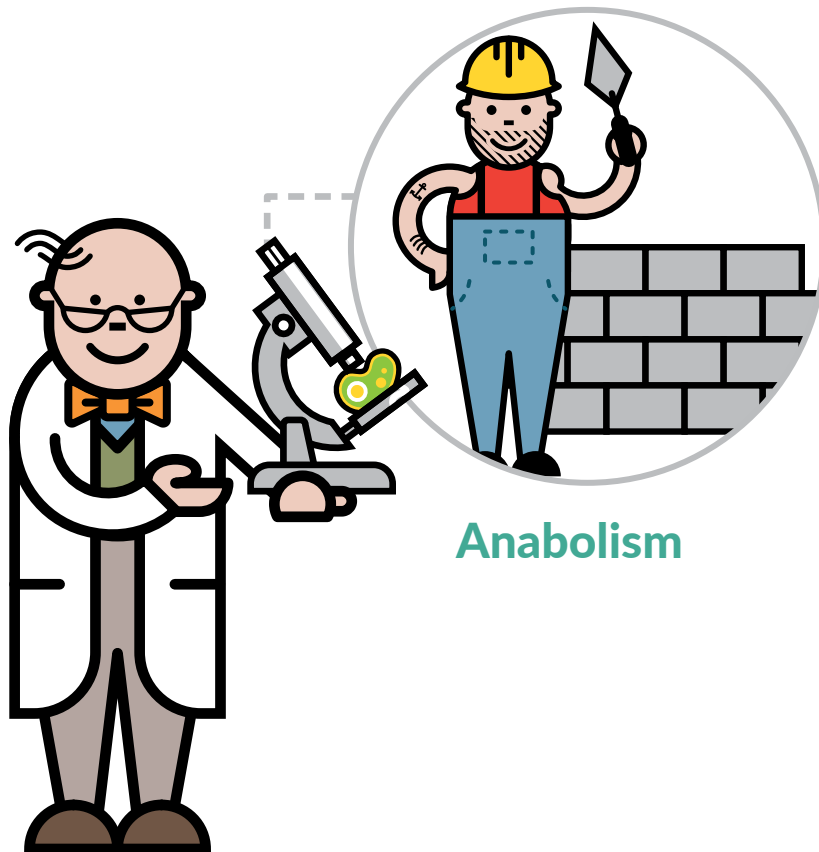
Protein is broken down into amino acids (building blocks of protein) by enzymes (which are like chemical scissors).

Enzymes then further break the amino acids into smaller parts.

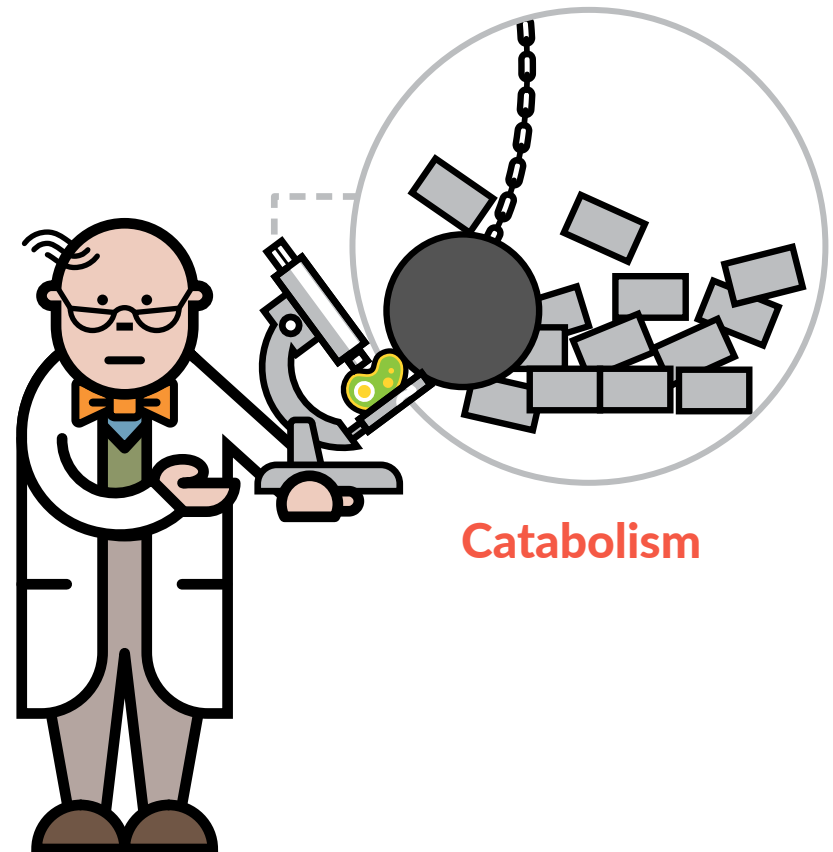


Protein metabolism

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



Anabolism

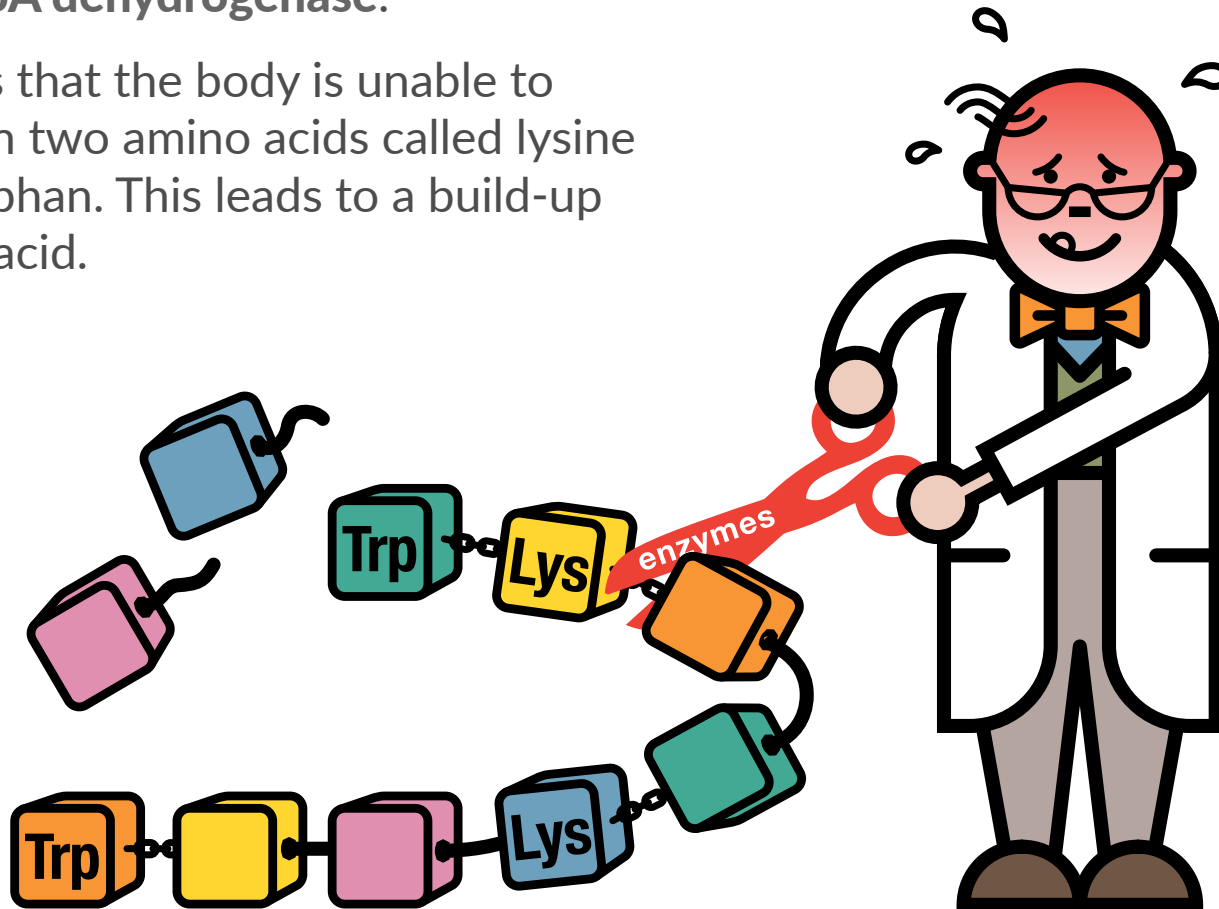


Catabolism

What happens in GA1?

In GA1, the body lacks an enzyme called **glutaryl-CoA dehydrogenase**.

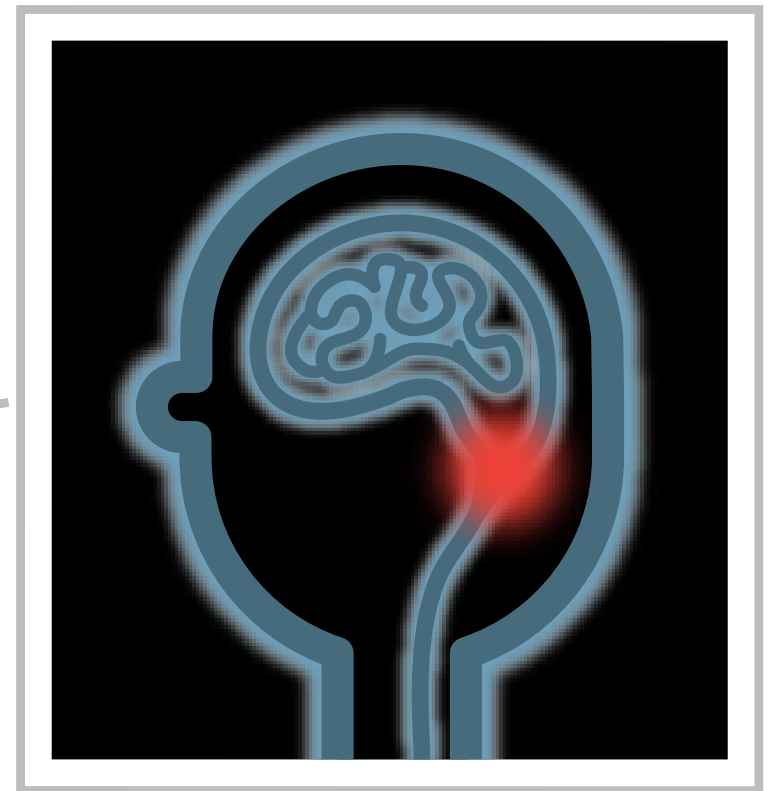
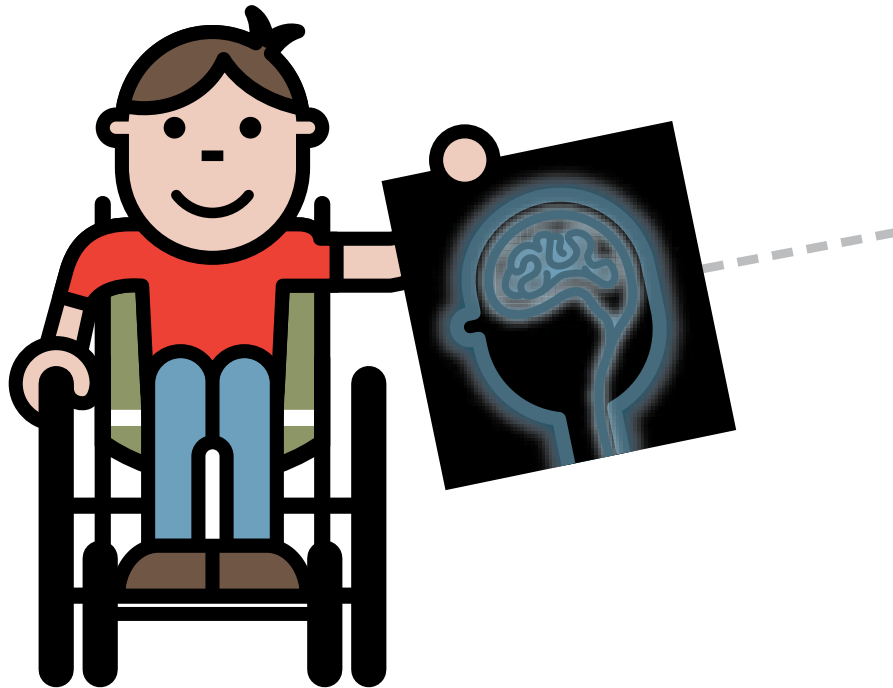
This means that the body is unable to break down two amino acids called lysine and tryptophan. This leads to a build-up of glutaric acid.



What can go wrong in GA1?

The basal ganglia in the brain controls movement.

The build up of glutaric acid damages the basal ganglia and causes movement problems.



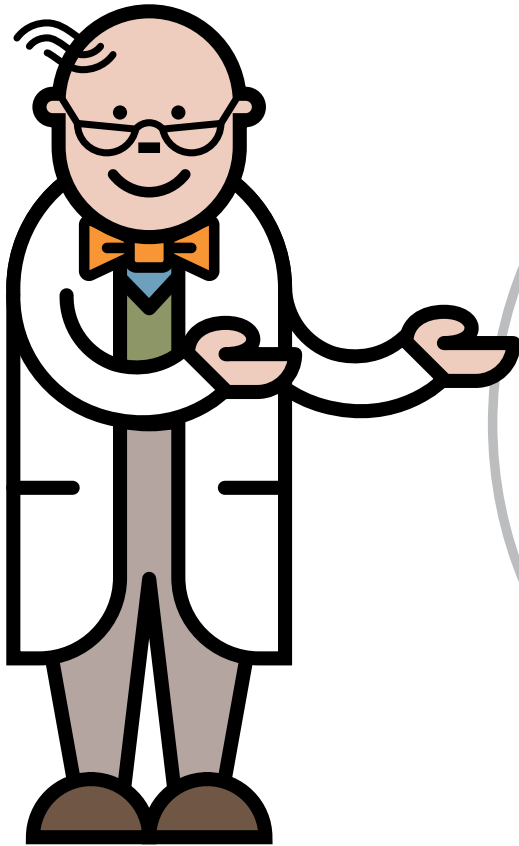
Metabolic crisis

- A **metabolic crisis** can trigger the **movement problems**. This is because there is a build-up of glutaric acid and other toxic chemicals
- It is usually caused by childhood infections or viruses causing high temperatures, vomiting and diarrhoea
- Avoidance of a metabolic crisis is essential



What about other symptoms in GA1?

Babies with GA1 are usually healthy at birth although many are born with a larger than average head size.



How is GA1 diagnosed?

GA1 is diagnosed by newborn screening. High levels of glutaric acid are found in the blood.



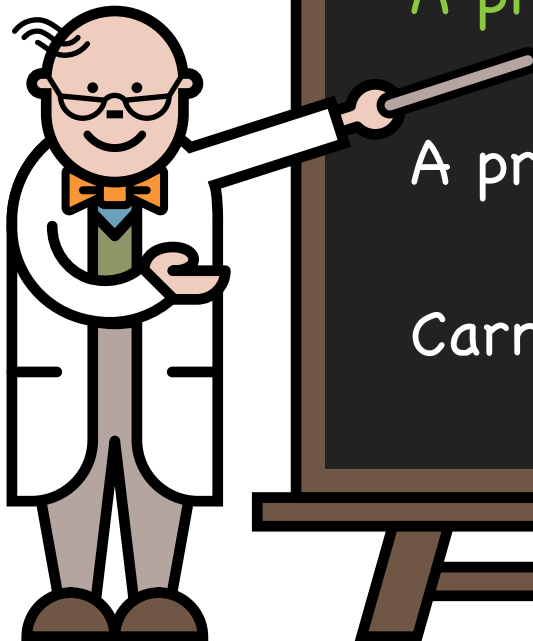
How is GA1 managed day to day?

GA1 is managed with the following special diet and medication:

A protein restricted diet

A protein substitute

Carnitine medication



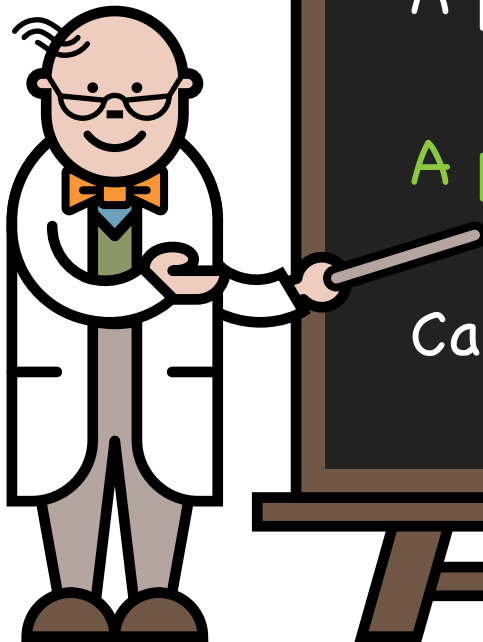
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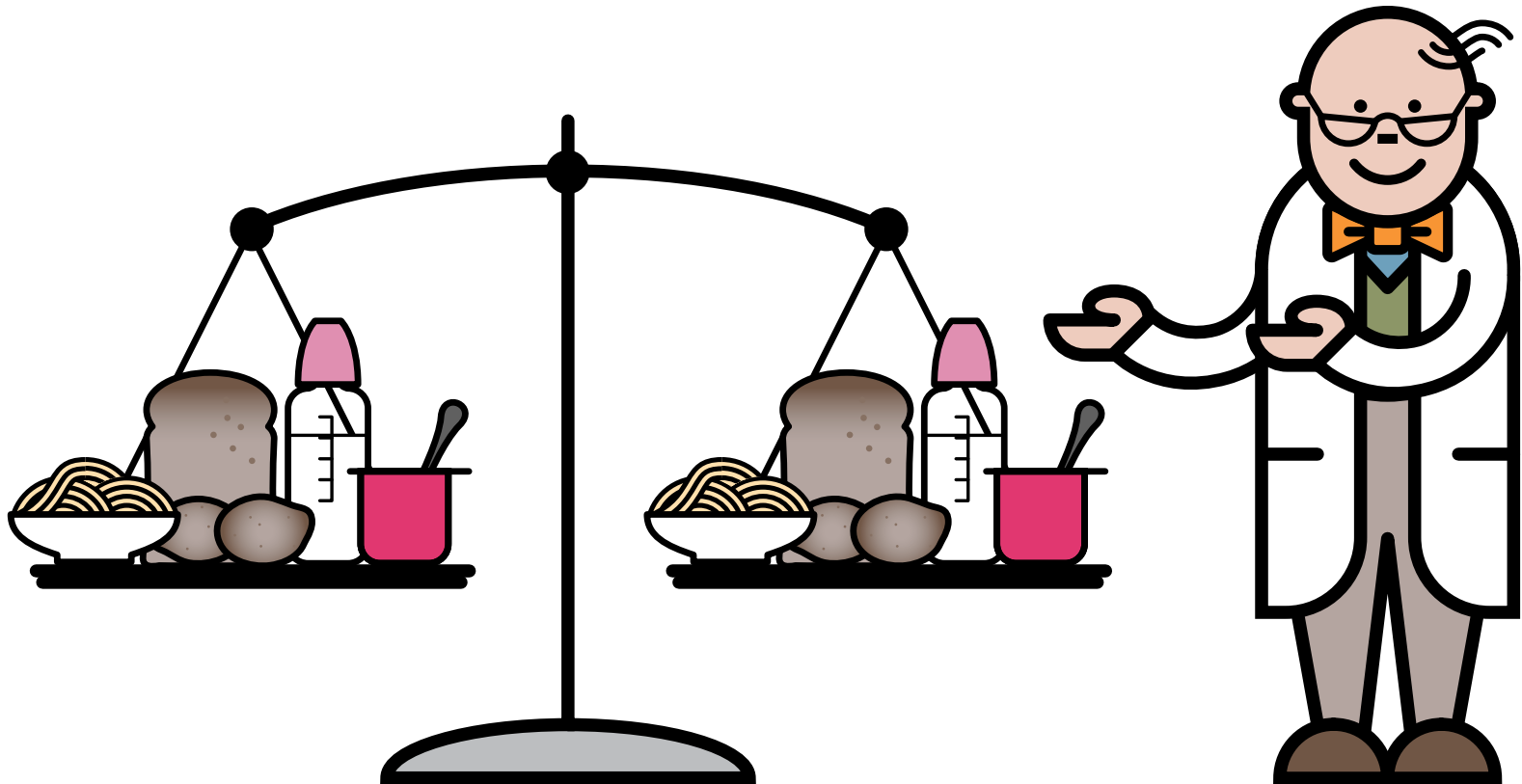
A protein substitute

Carnitine medication



Protein balance is needed in GA1

In GA1, it is important that enough protein is given for growth
...but not too much as toxic chemicals will be made.



Measured protein intake

In babies, a restricted amount of protein is given from breast milk or measured amounts of infant formula.

The amount given will be monitored regularly by your specialist metabolic dietitian.

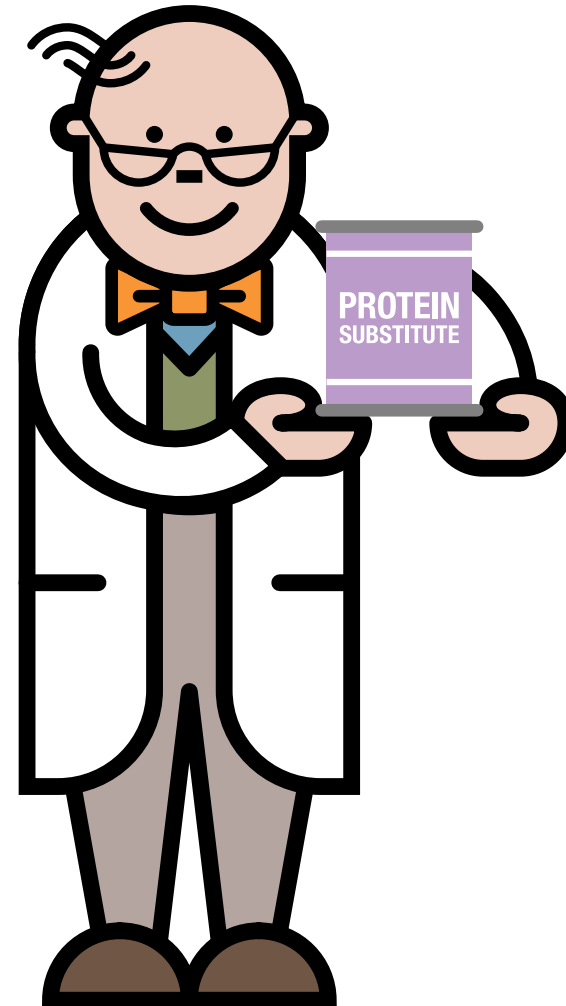


Protein substitute

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.



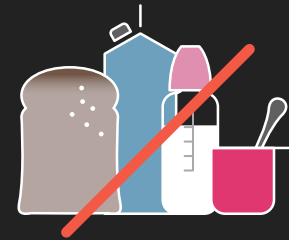
How is GA1 managed during illness?

- During any childhood illness, an emergency regimen is given
- Illnesses can cause catabolism or protein breakdown
- This will lead to a build up of glutaric acid and cause a metabolic crisis



How is GA1 managed during illness?

Stop all protein in food & drink



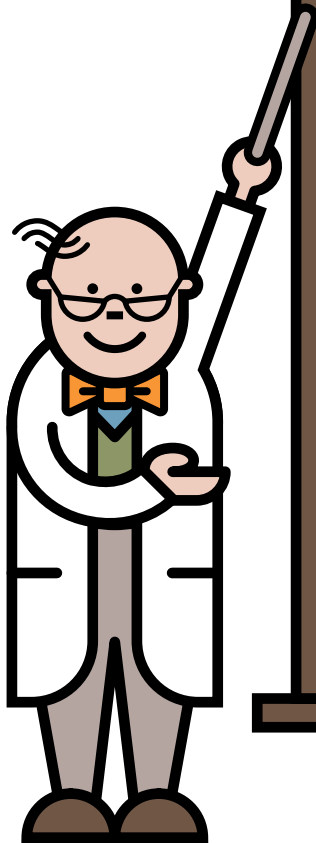
Start the emergency regimen.
This is made up of protein
substitute and glucose polymer



Carnitine medication as prescribed



Phone your metabolic team



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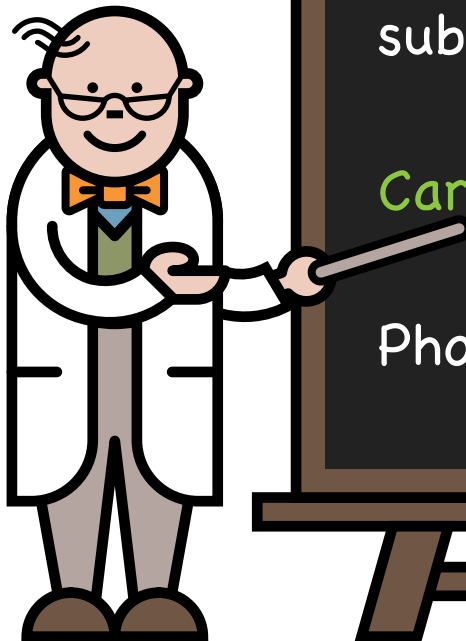
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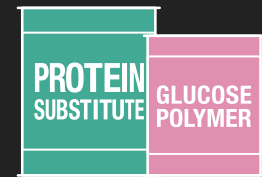


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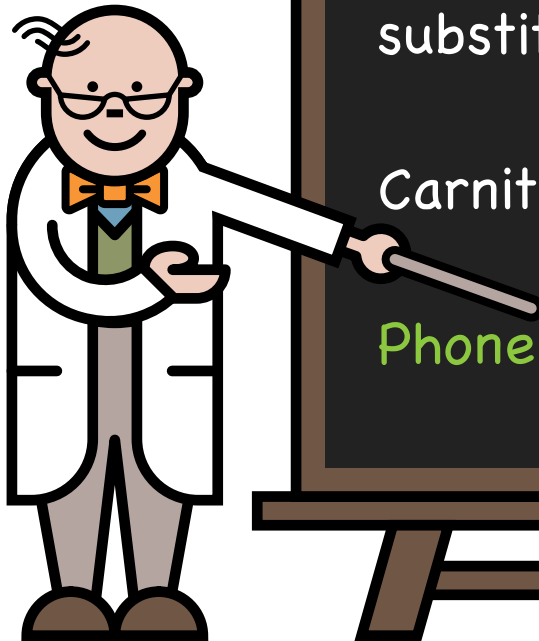
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Carnitine medication as prescribed



Phone 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



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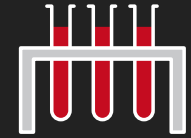
Key message

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



How is GA1 monitored?

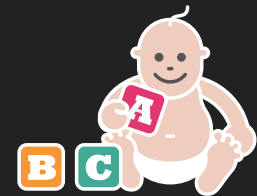
Frequent blood tests to check amino acid and nutrient levels ✓



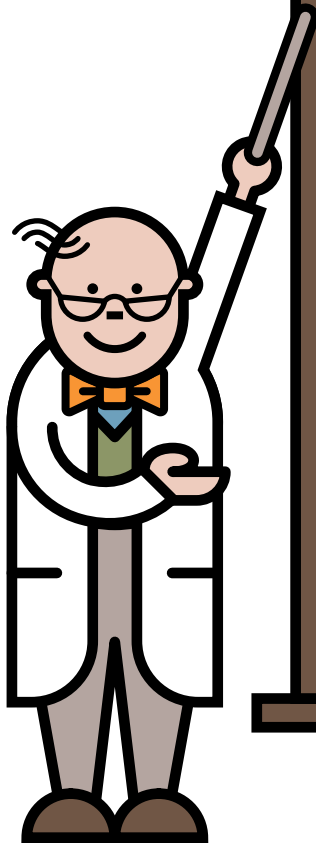
Height and weight



Developmental checks

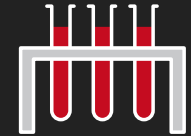


Diet and medications are adjusted according to age, weight and blood chemical levels



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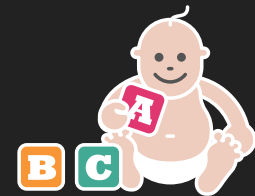
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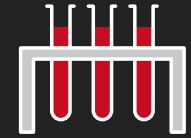


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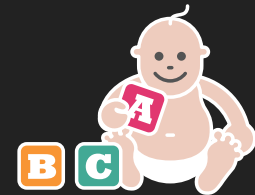
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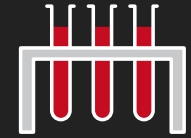


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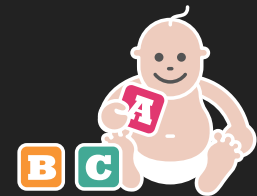
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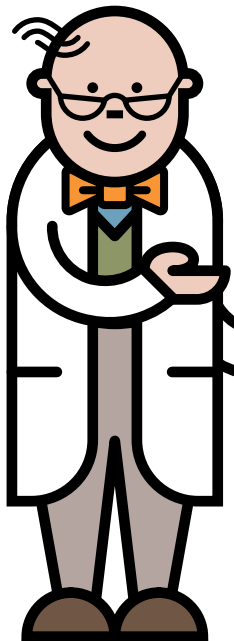
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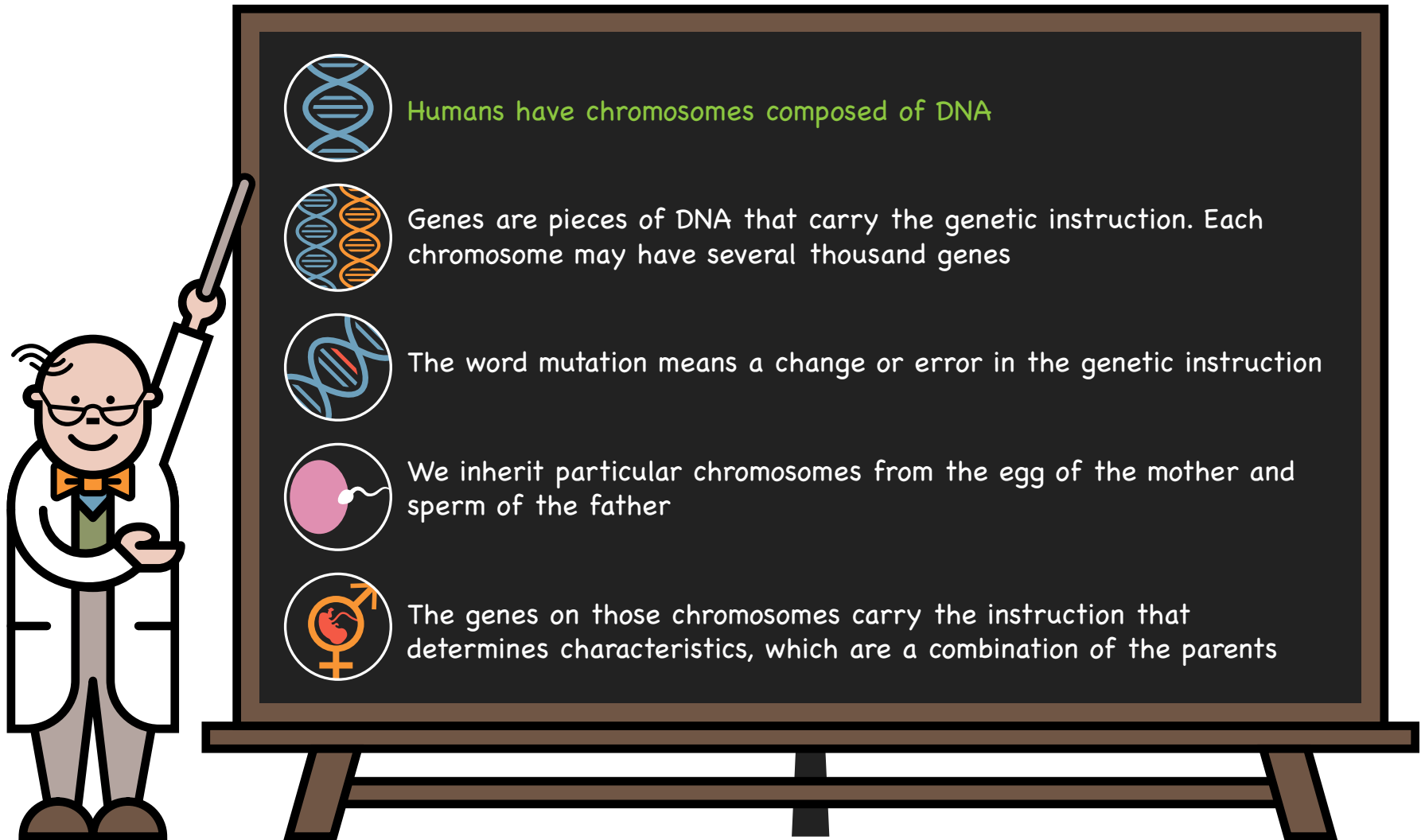
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Chromosomes, genes, mutations




 Humans have chromosomes composed of DNA

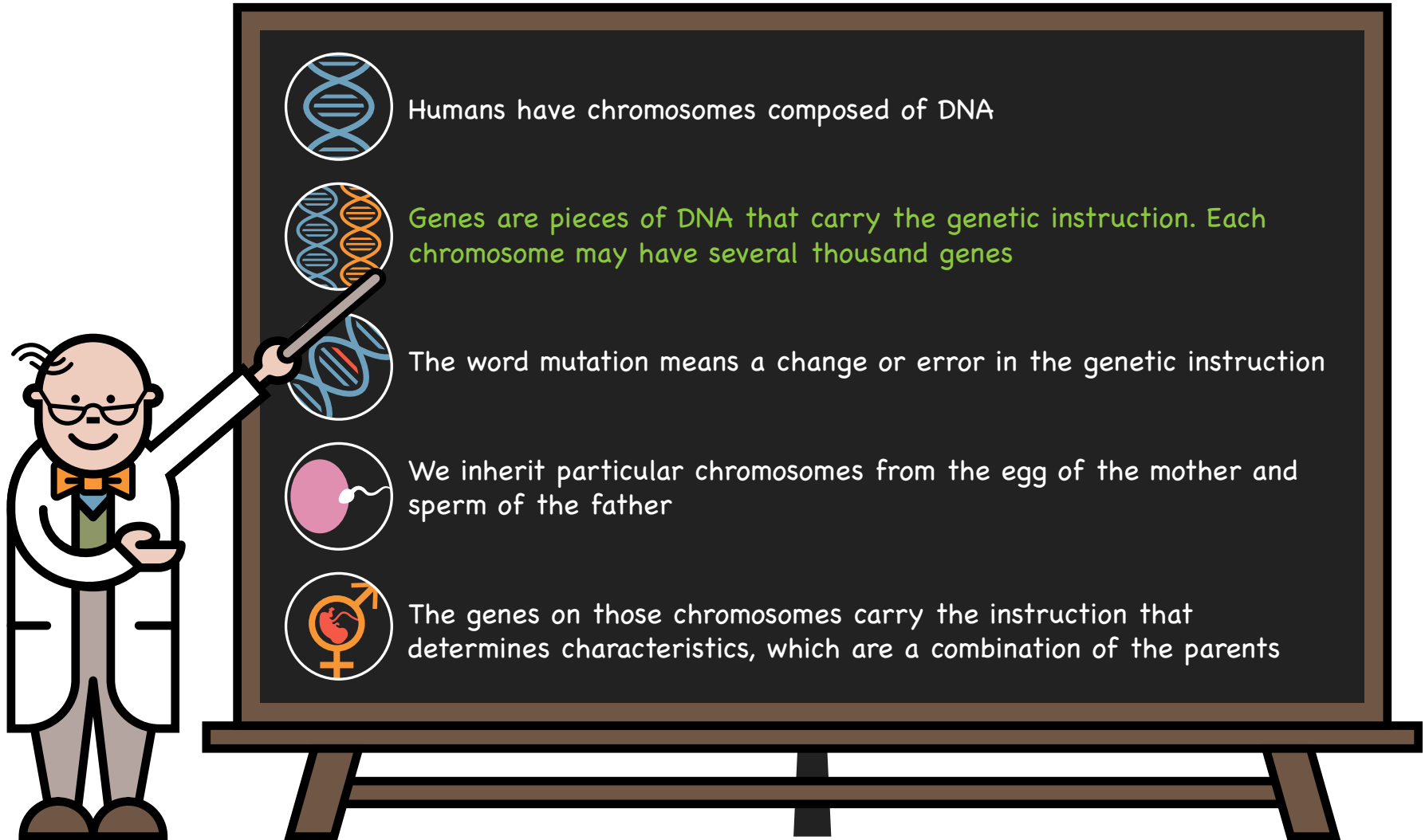
 Genes are pieces of DNA that carry the genetic instruction. Each chromosome may have several thousand genes


 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




 Humans have chromosomes composed of DNA

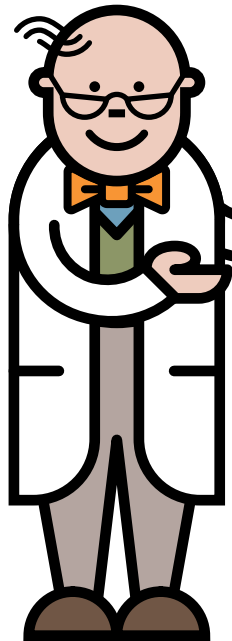
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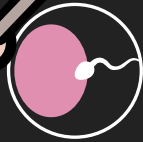
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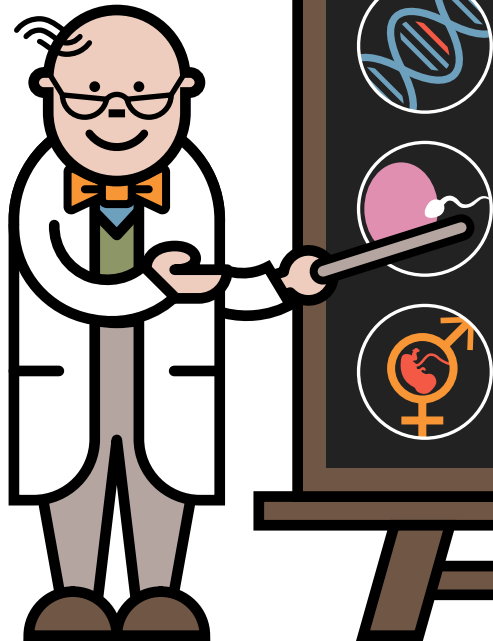


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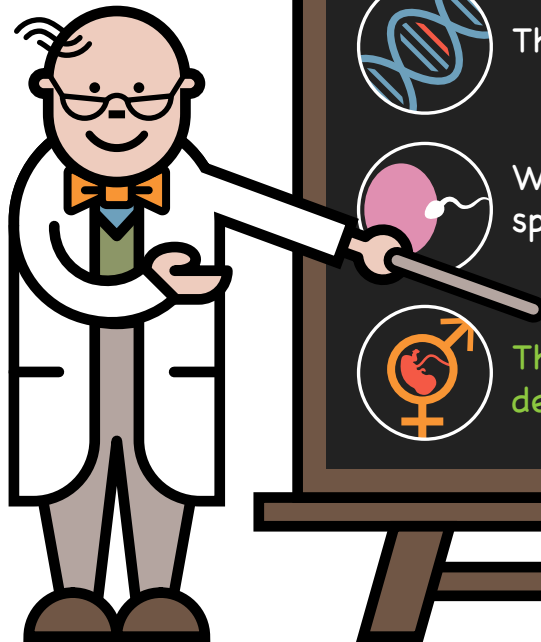


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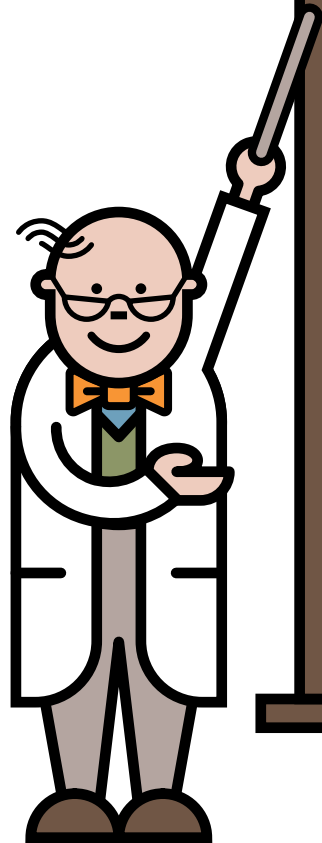


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Inheritance



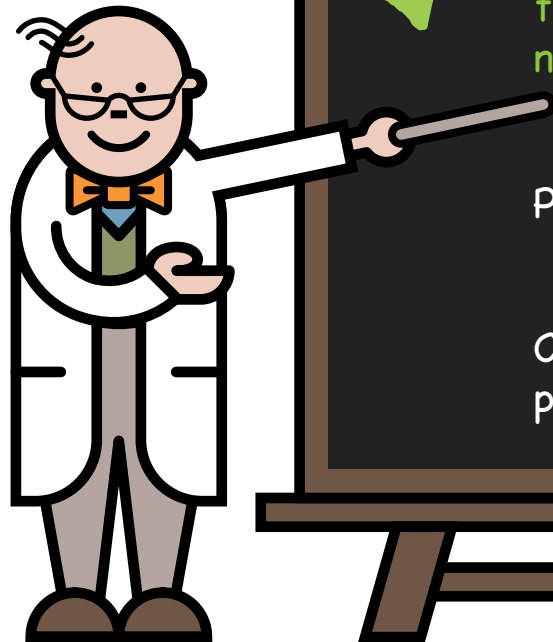
GA1 is an inherited condition. There is nothing that could have been done to prevent your baby from having GA1

Everyone has a pair of genes that make the glutaryl-CoA dehydrogenase enzyme. In children with GA1, neither of these genes works correctly. These children inherit one non-working GA1 gene from each parent

Parents of children with GA1 are carriers of the condition

Carriers do not have GA1 because the other gene of this pair is working correctly

Inheritance



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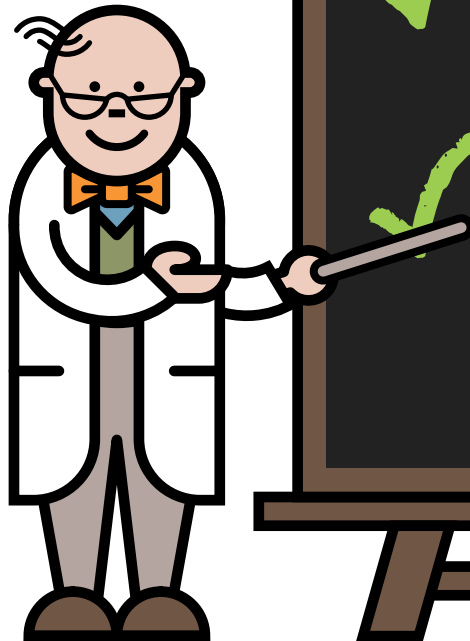


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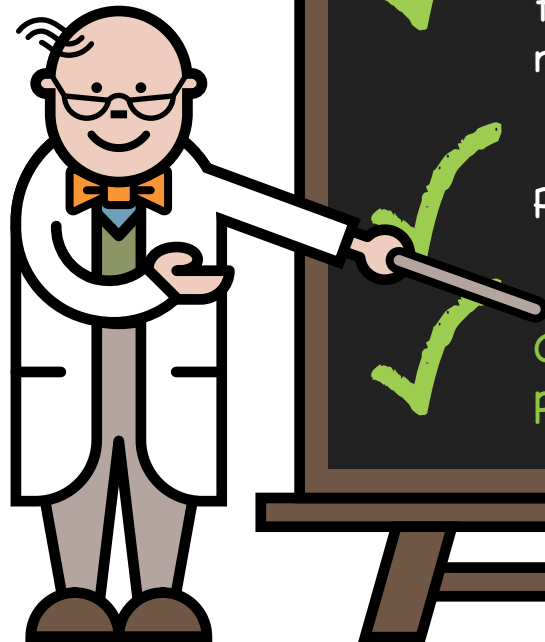
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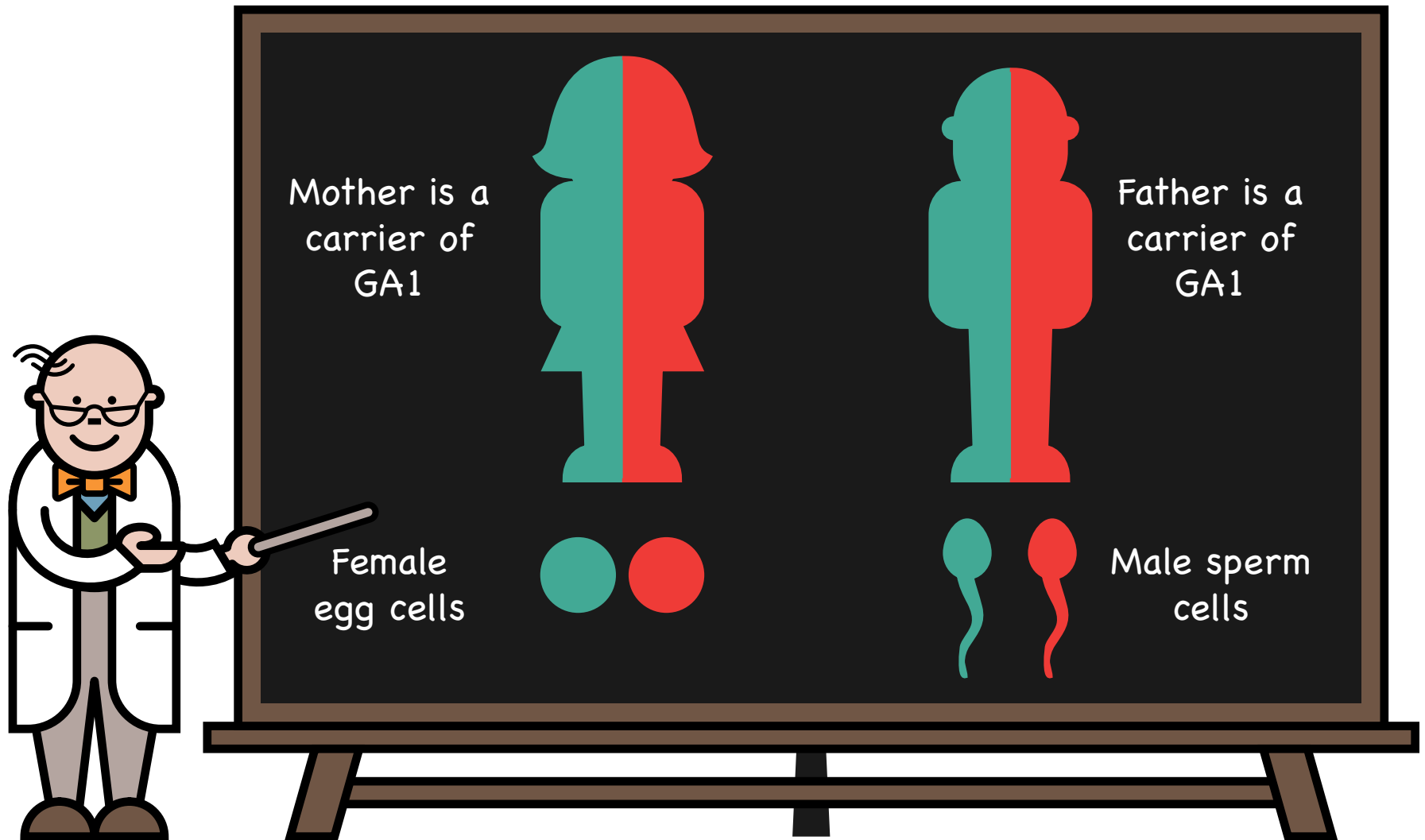


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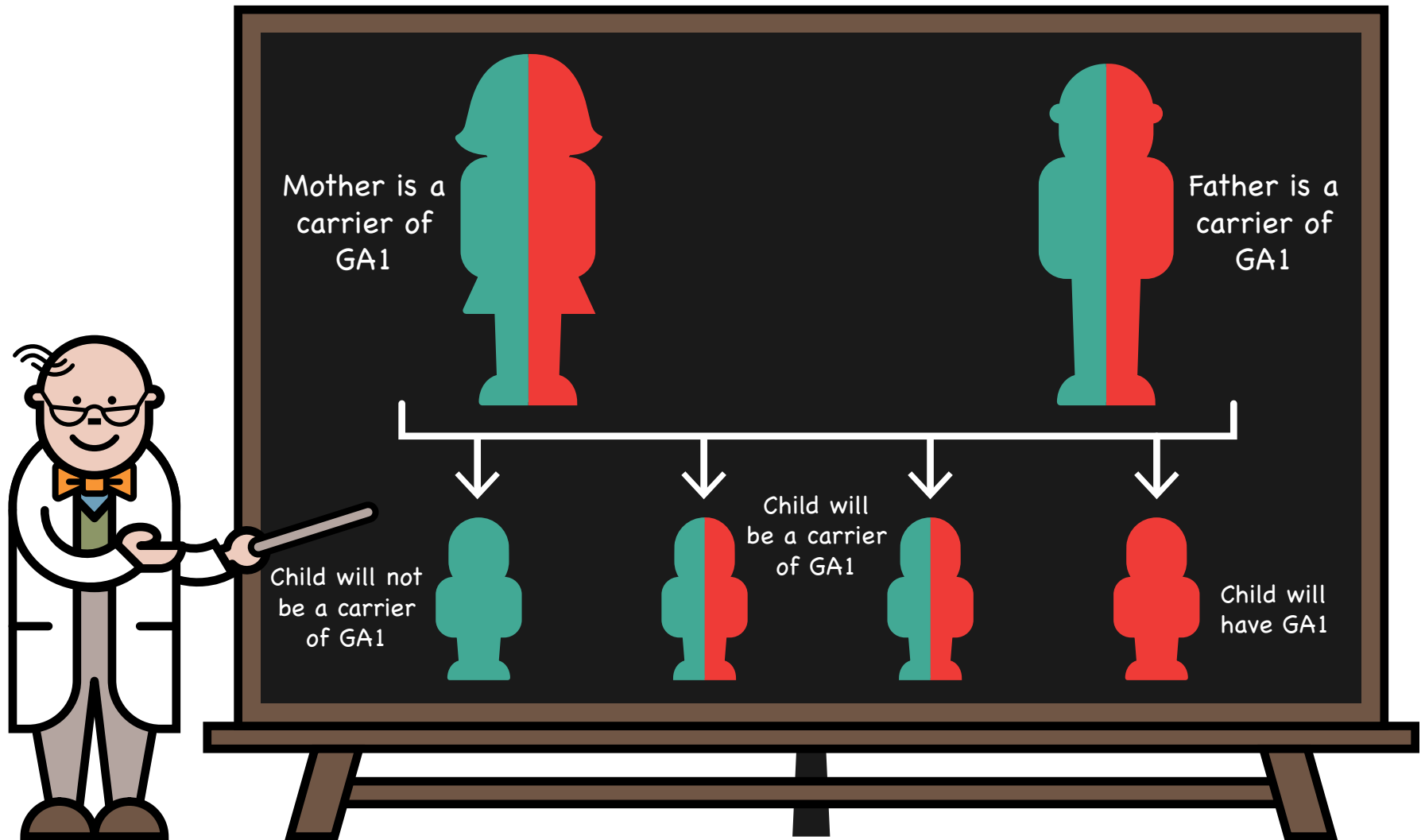


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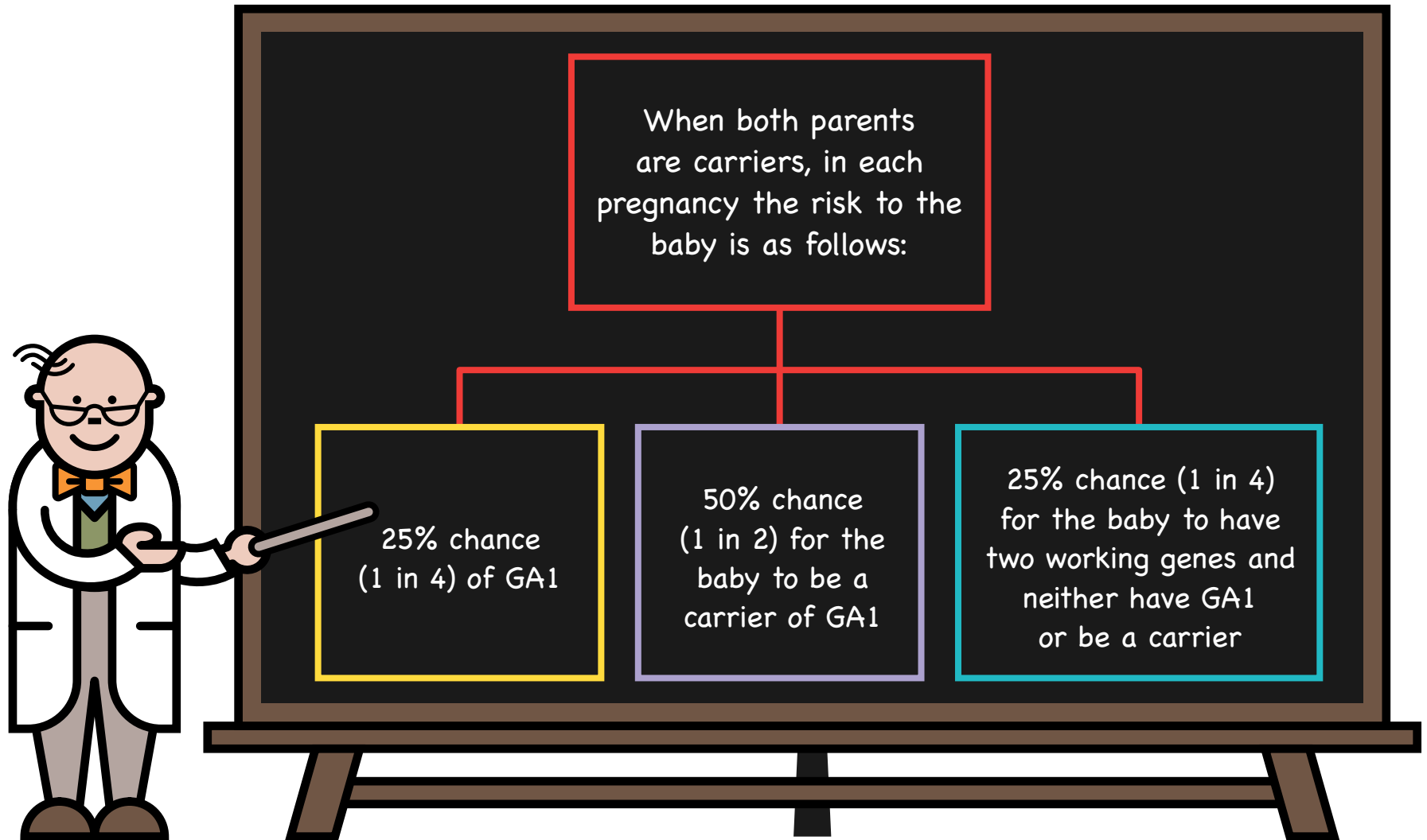
Inheritance – Autosomal recessive (carriers of GA1)



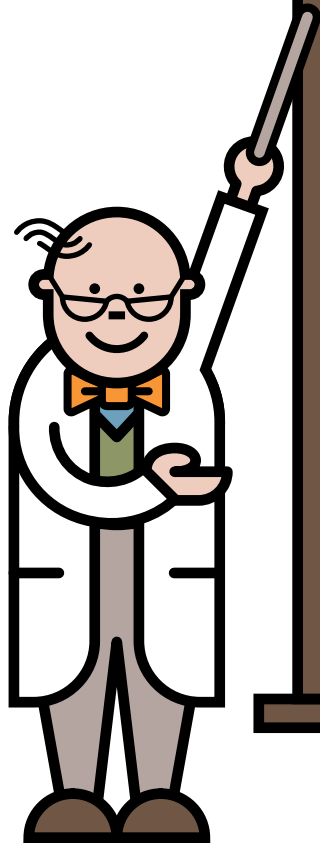
Inheritance – Autosomal recessive – possible combinations



Future pregnancies



Take home messages



✓ GA1 is a serious inherited metabolic disorder that can lead to severe movement problems

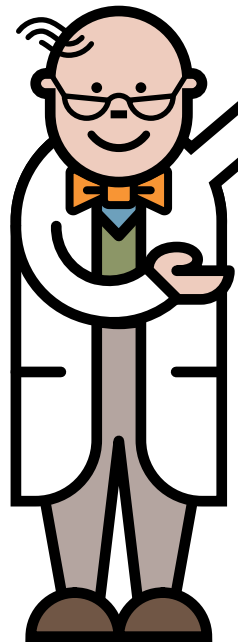
Children are very vulnerable in the first 6 years of life

Damage can be prevented with a protein restricted diet, a protein substitute and carnitine

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

With good management, severe movement problems can be prevented in the majority of children

Take home messages



GA1 is a serious inherited metabolic disorder that can lead to severe movement problems



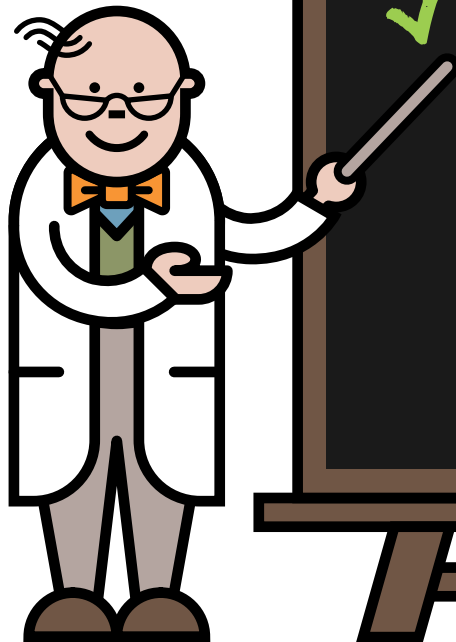
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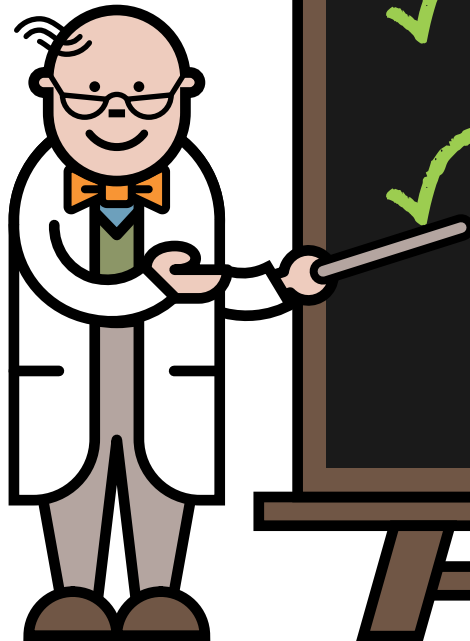


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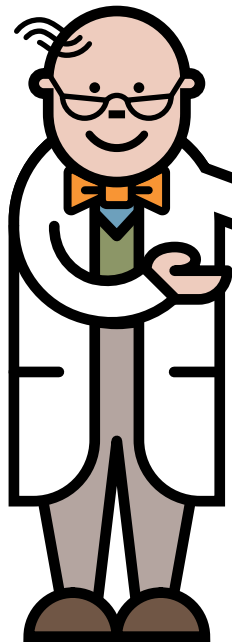
Take home messages



- ✓ GA1 is a serious inherited metabolic disorder that can lead to severe movement problems
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Helpful hints

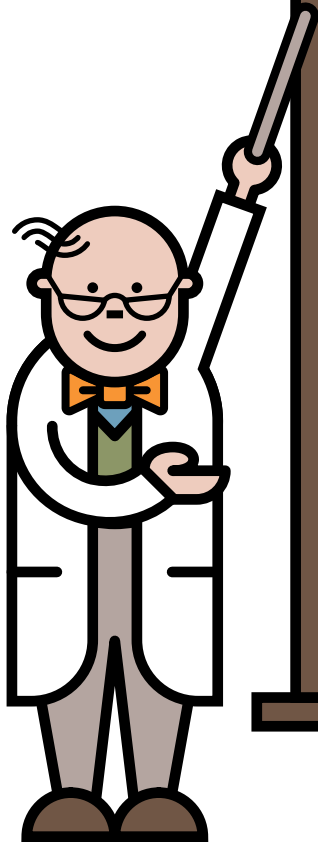


Always ensure you have a good supply of your dietary products and medicines and that they are in date

Your dietary products and medications are prescribed by your GP. 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



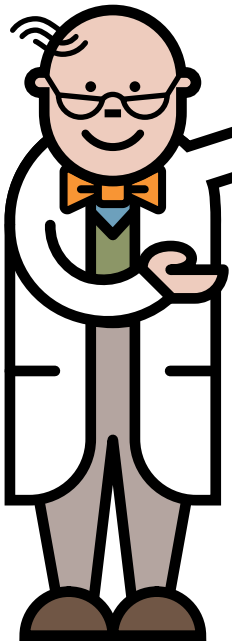
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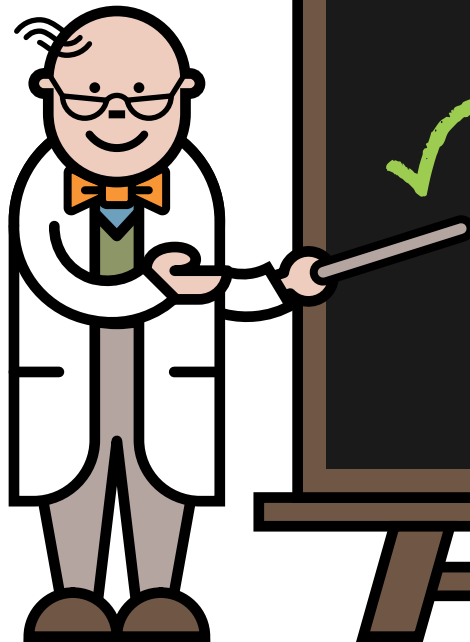
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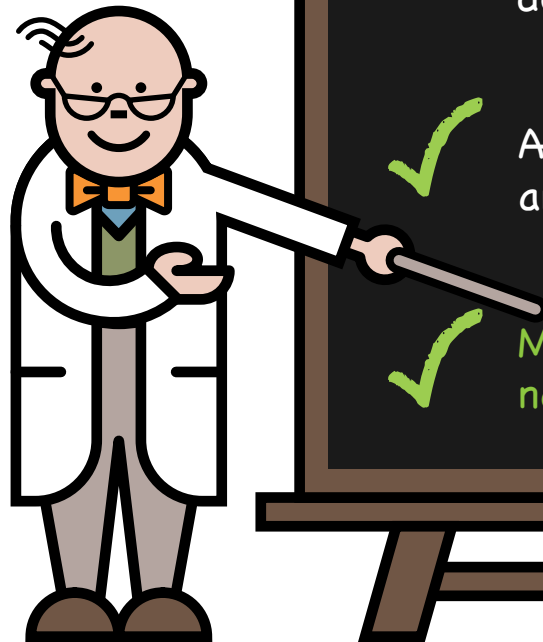
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Who's who

- 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.



BIMDG

British Inherited Metabolic Diseases Group



www.bimdg.org.uk

NUTRICIA
Advanced Medical Nutrition

www.nutricia.co.uk



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Children Living with
Inherited Metabolic Diseases

www.climb.org.uk