

# 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 4, JANUARY 2025

GAI

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).

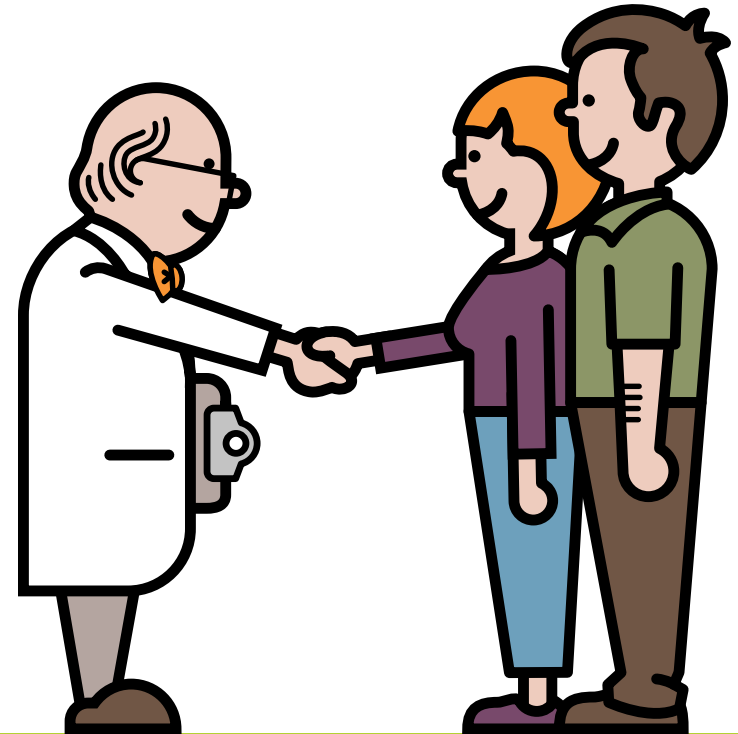
The team are Rachel Skeath, Karen van Wyk, Pat Portnoi and Anita MacDonald. The group is facilitated by Heidi Chan from Nutricia.

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.**

# GAI

Information for families following  
a positive newborn screening



ADAPTED BY THE DIETITIANS GROUP

**BIMDG**

British Inherited Metabolic Diseases Group



BASED ON THE ORIGINAL TEMPLE WRITTEN BY  
BURGARD AND WENDEL

VERSION 3, OCTOBER 2020

**TEMPLE**

Tools Enabling Metabolic Parents LEarning

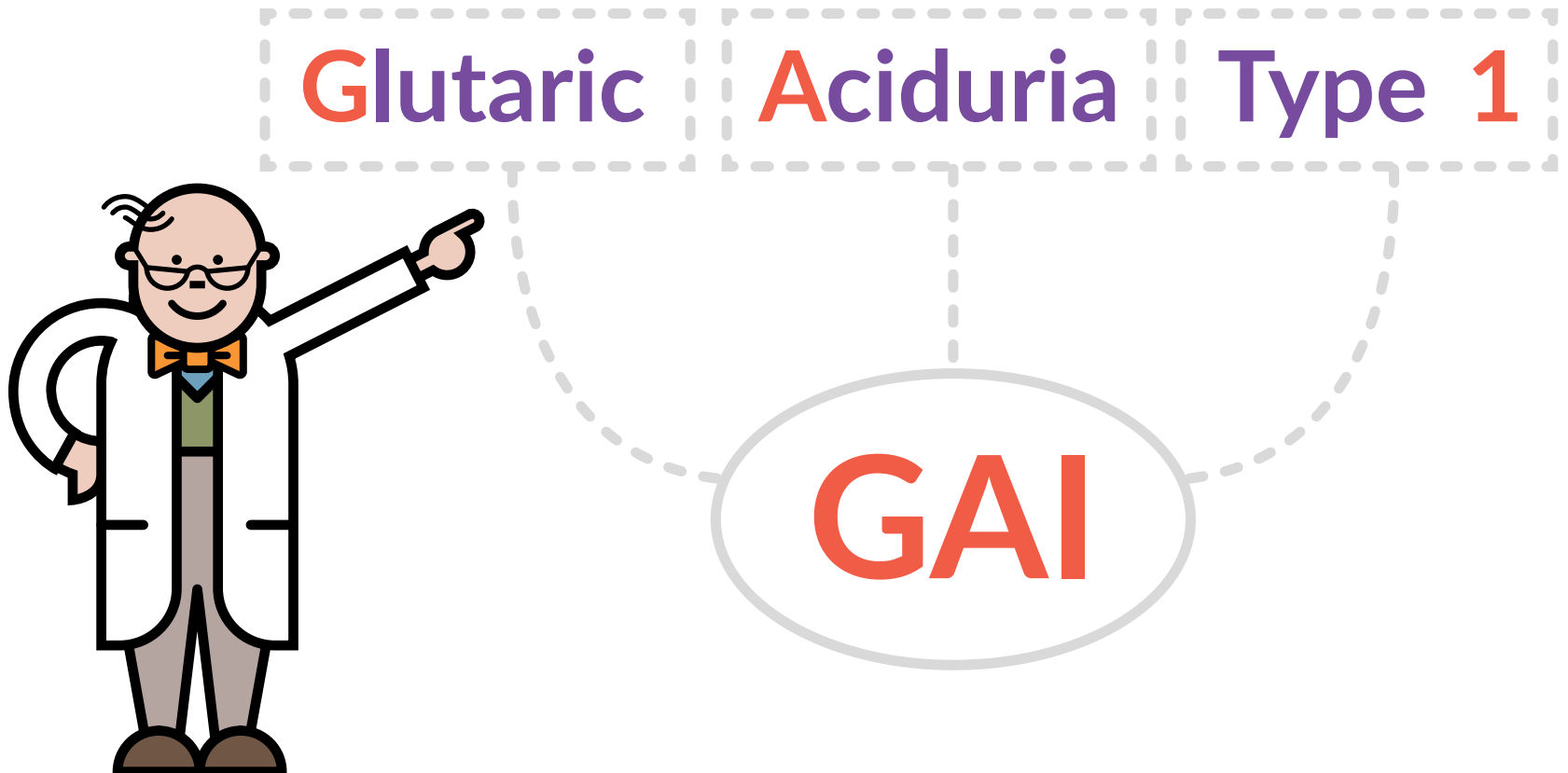


Supported by **NUTRICIA**  
as a service to metabolic medicine

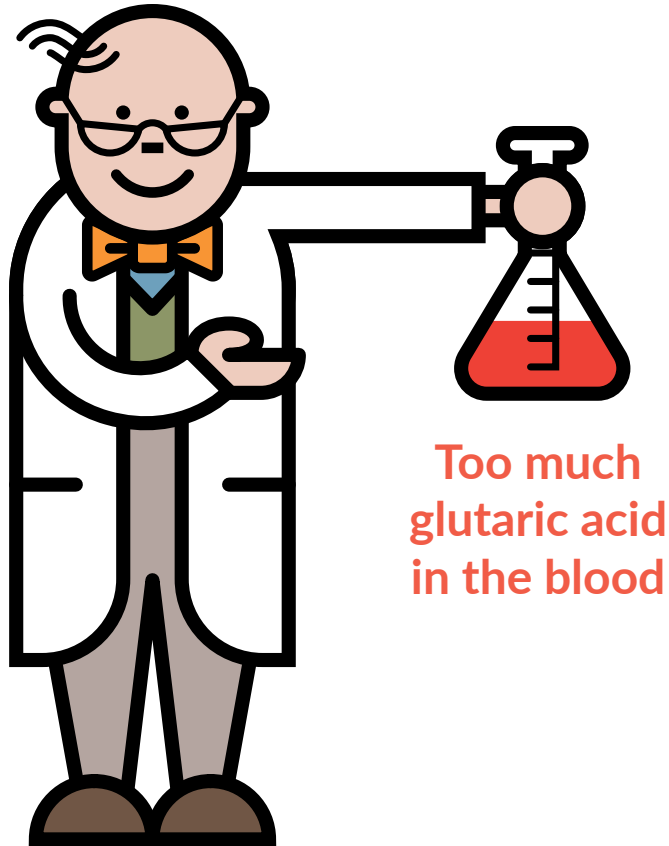
# What is GAI?

GAI stands for Glutaric Aciduria Type 1

It is an inherited metabolic condition



# What is GAI?

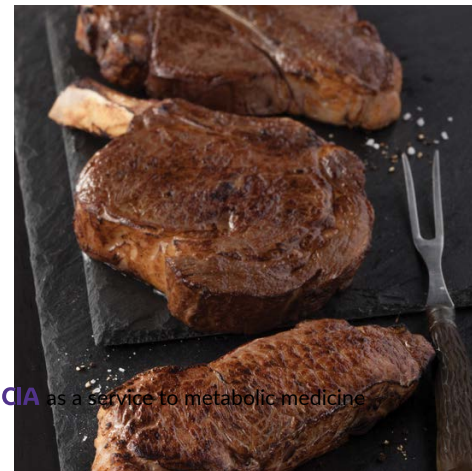
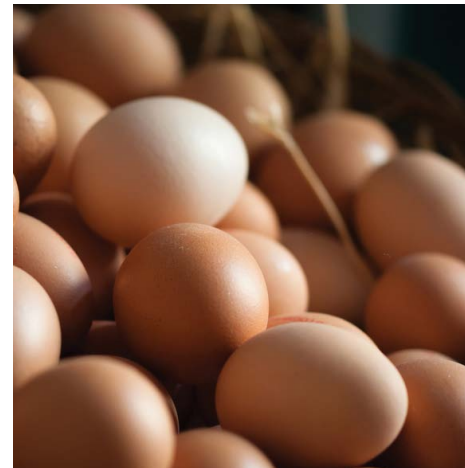


# GAI and protein

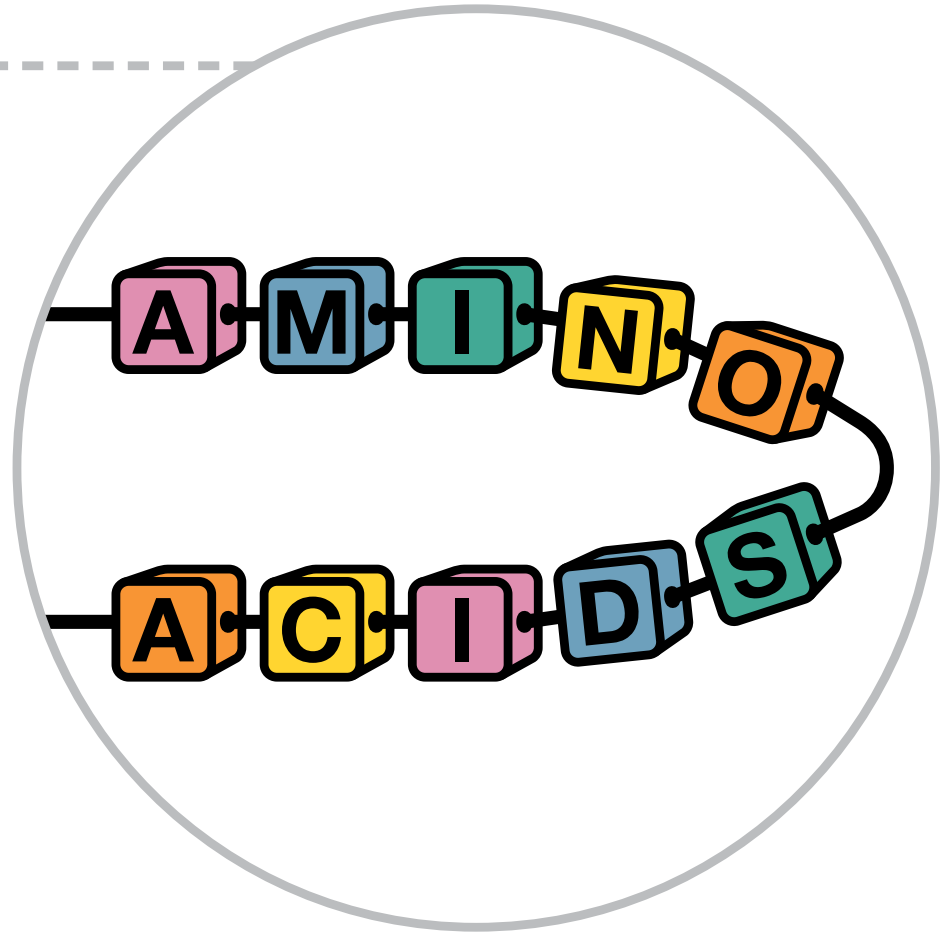
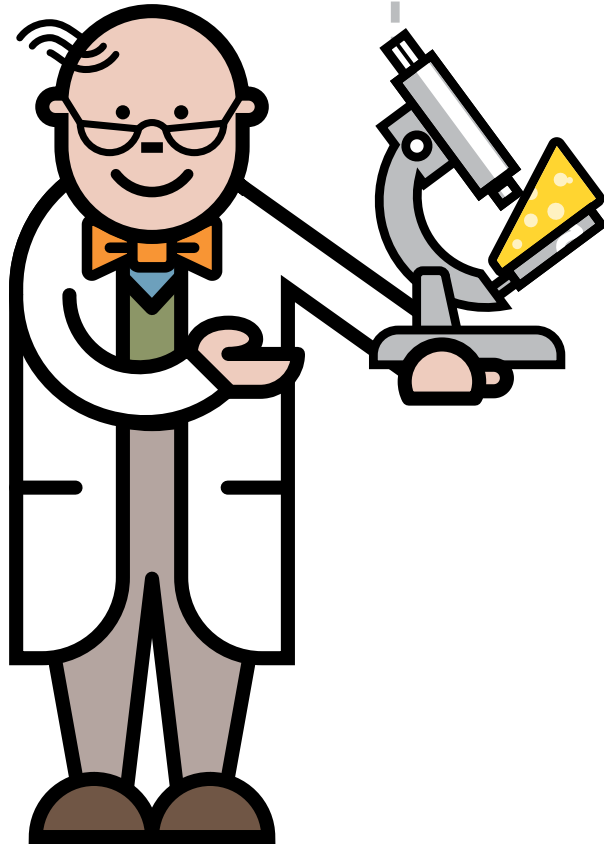
GAI affects the way your baby breaks down protein

Many foods contain protein

The body needs protein for growth and repair



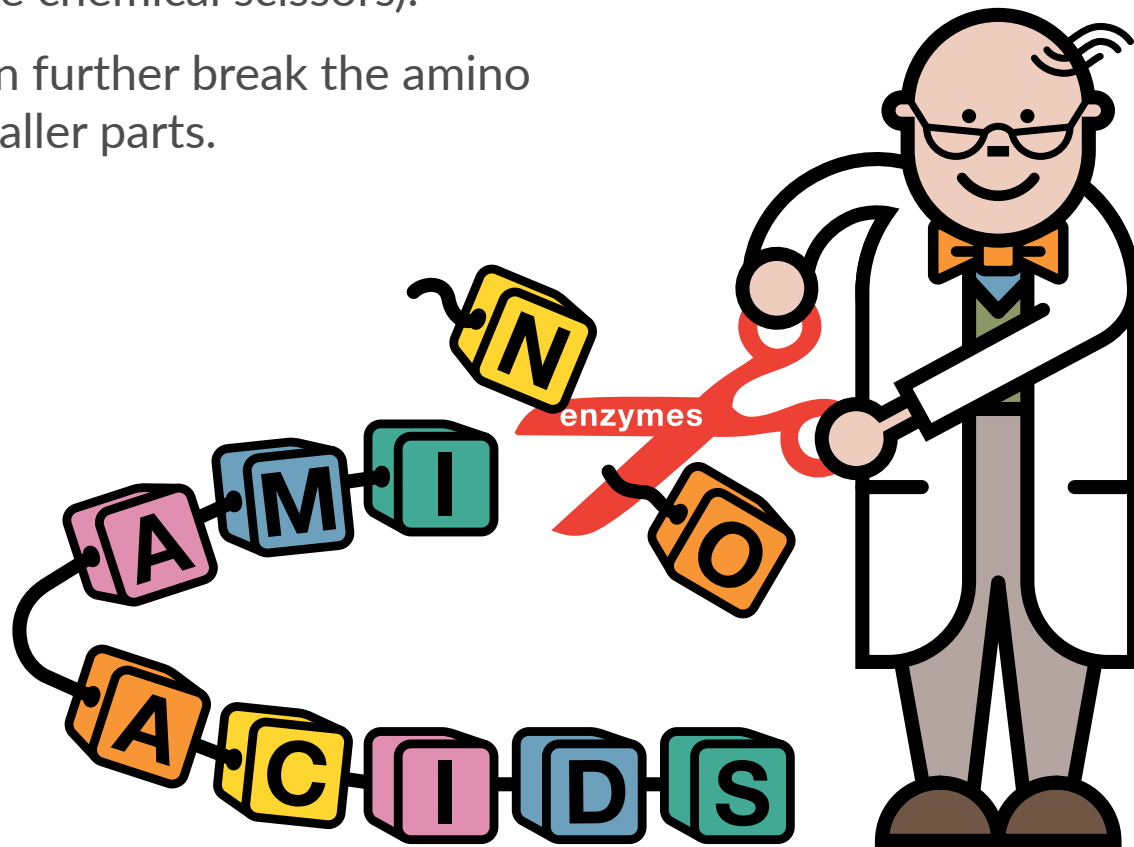
# What is protein?



# Protein and enzymes

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

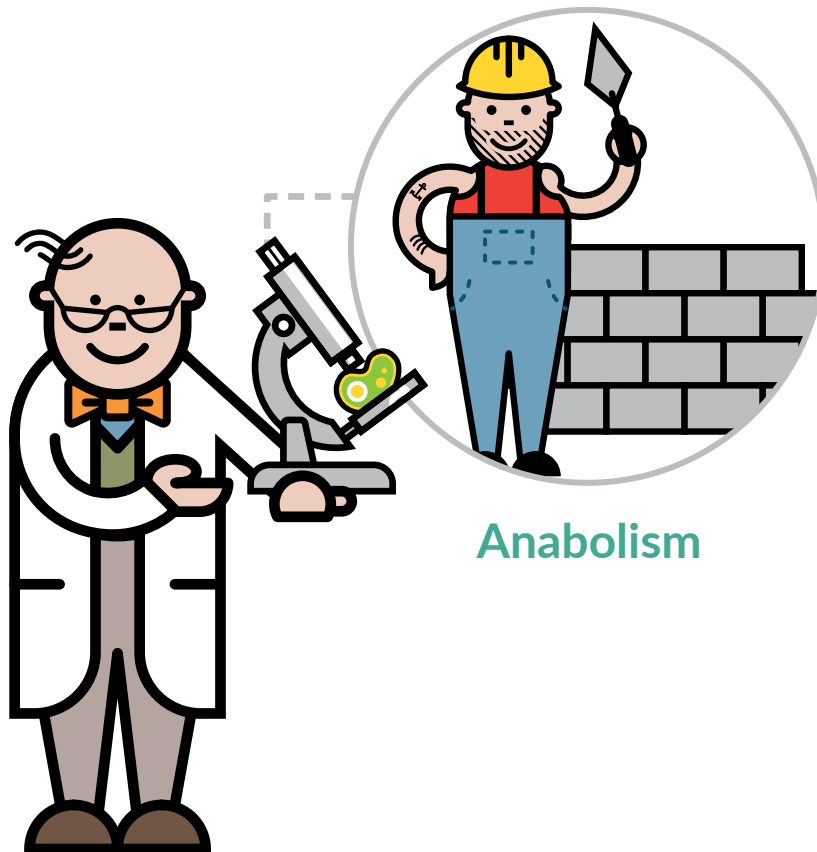
Enzymes then further break the amino acids into smaller parts.



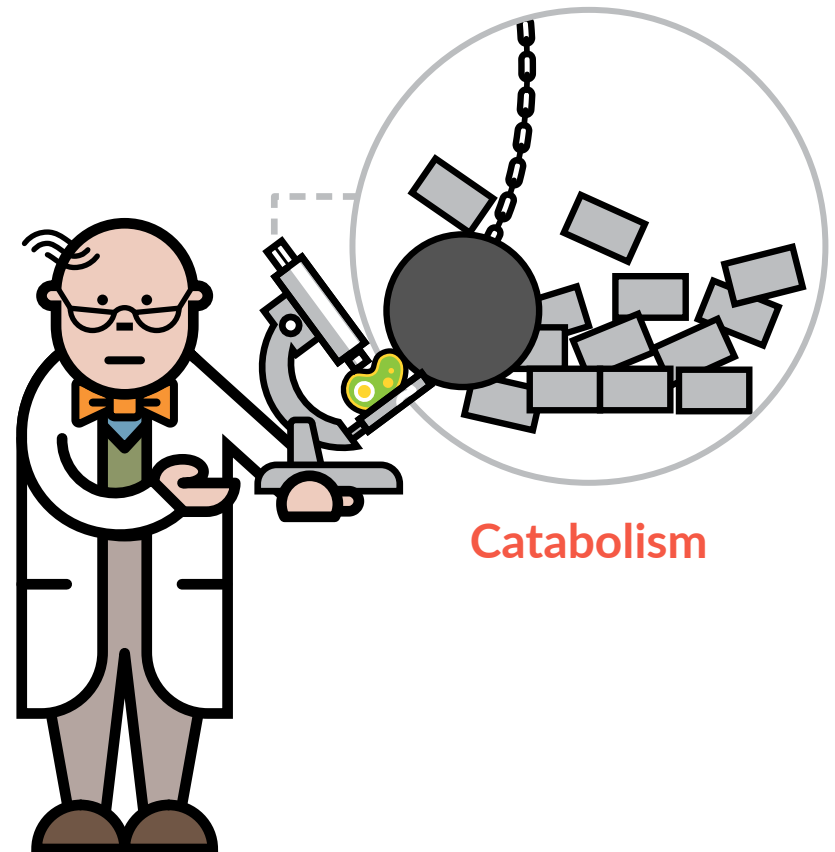


# Protein metabolism

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



Anabolism

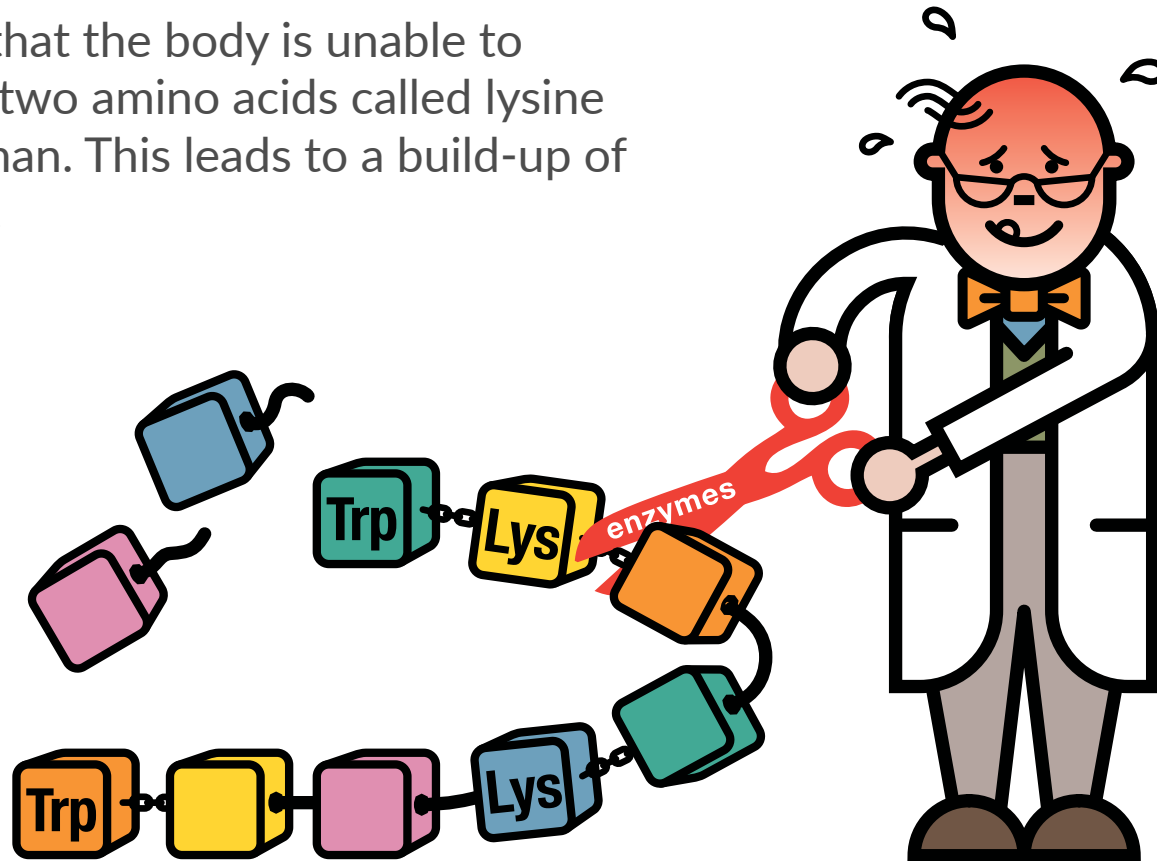


Catabolism

# What happens in GAI?

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

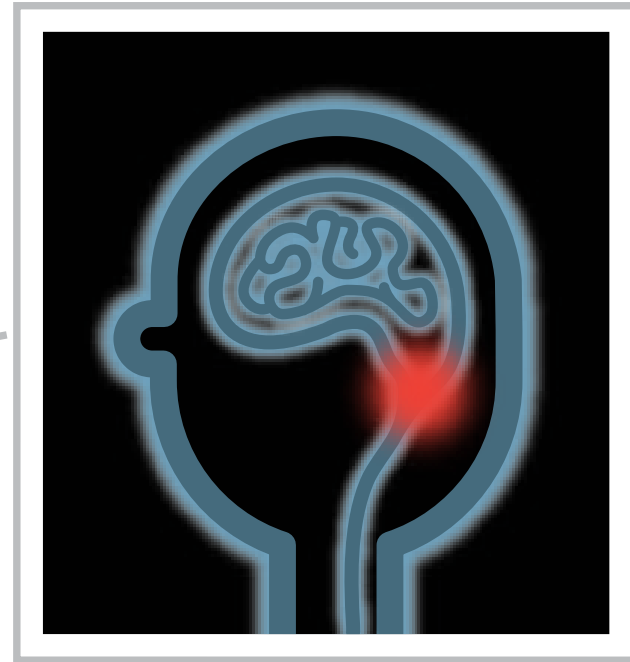
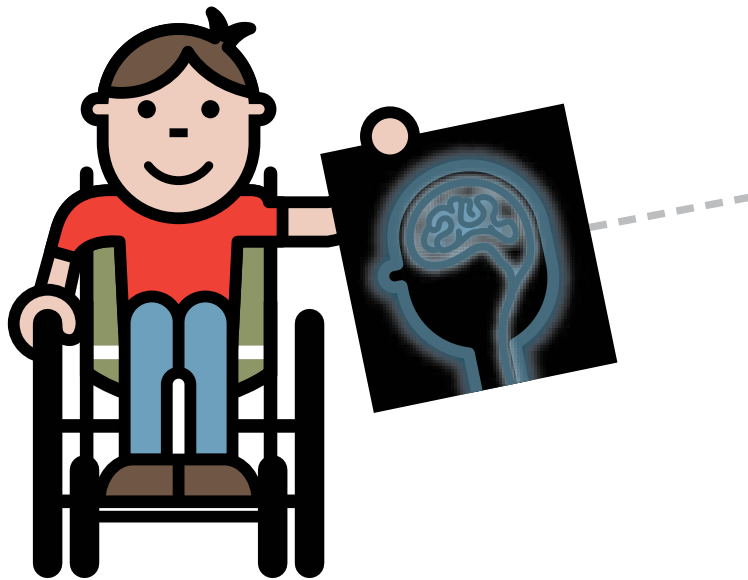
This means that the body is unable to break down two amino acids called lysine and tryptophan. This leads to a build-up of glutaric acid.



# What can go wrong in GAI?

The basal ganglia in the brain controls movement.

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



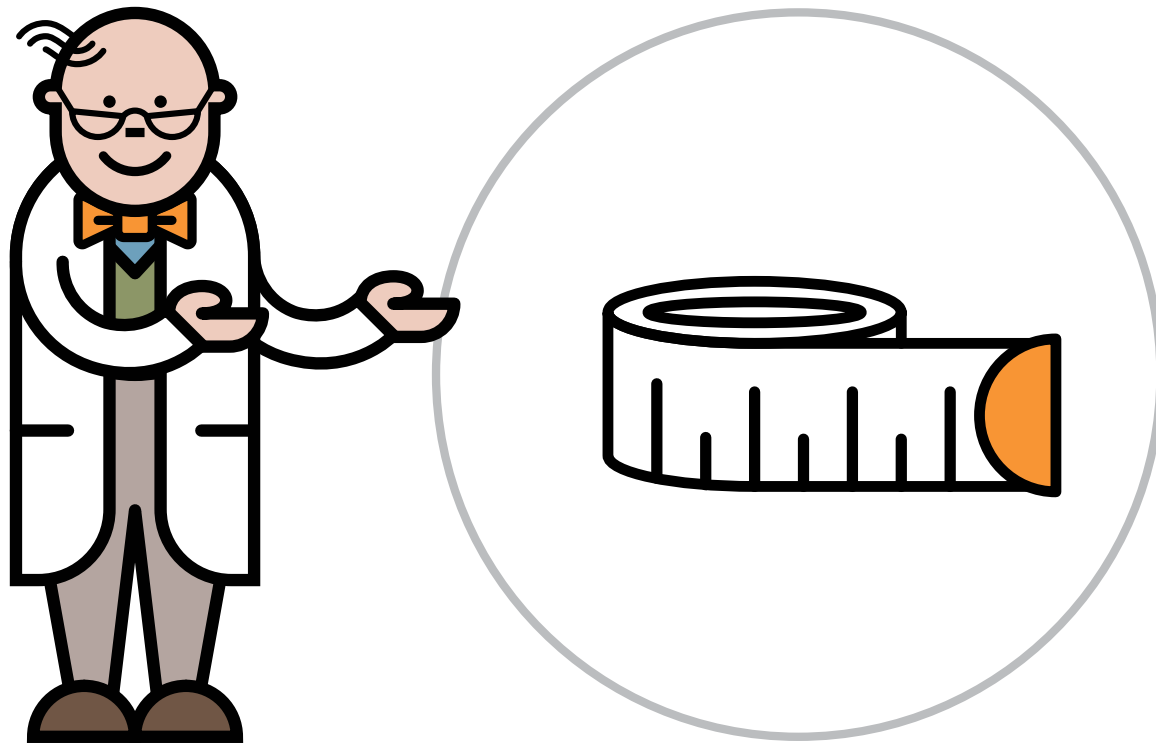
# Metabolic crisis

- 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 GAI?

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



# How is GAI diagnosed?

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



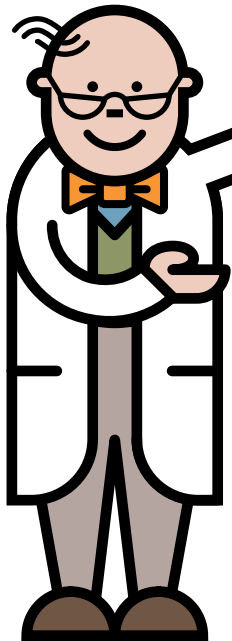
# How is GAI managed day to day?

GAI is managed with the following special diet and medication:

A protein restricted diet

A protein substitute

Carnitine medication



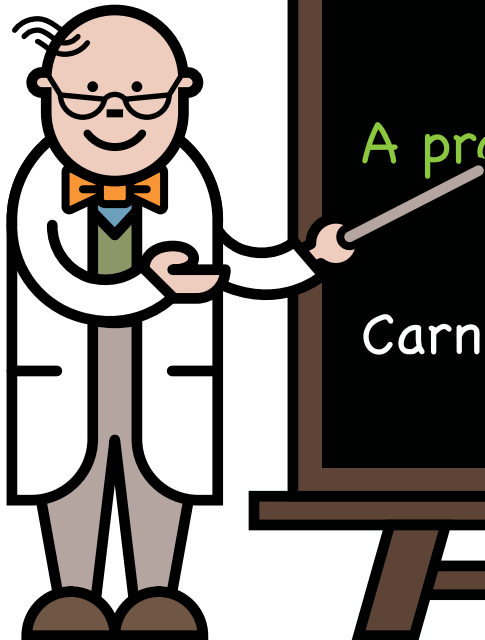
# How is GAI managed day to day?

GAI is managed with the following special diet and medication:

A protein restricted diet

A protein substitute

Carnitine medication





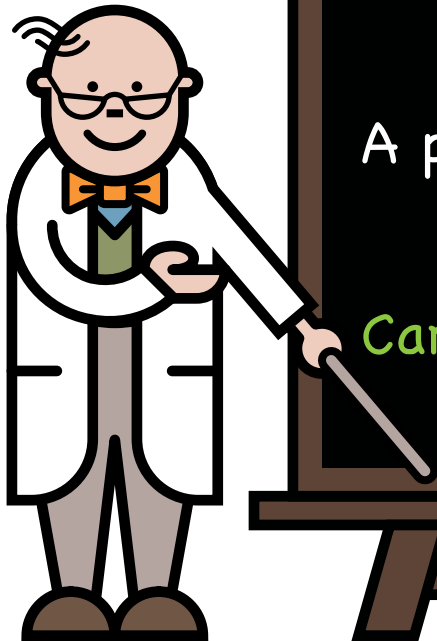
# How is GAI managed day to day?

GAI is managed with the following special diet and medication:

A protein restricted diet

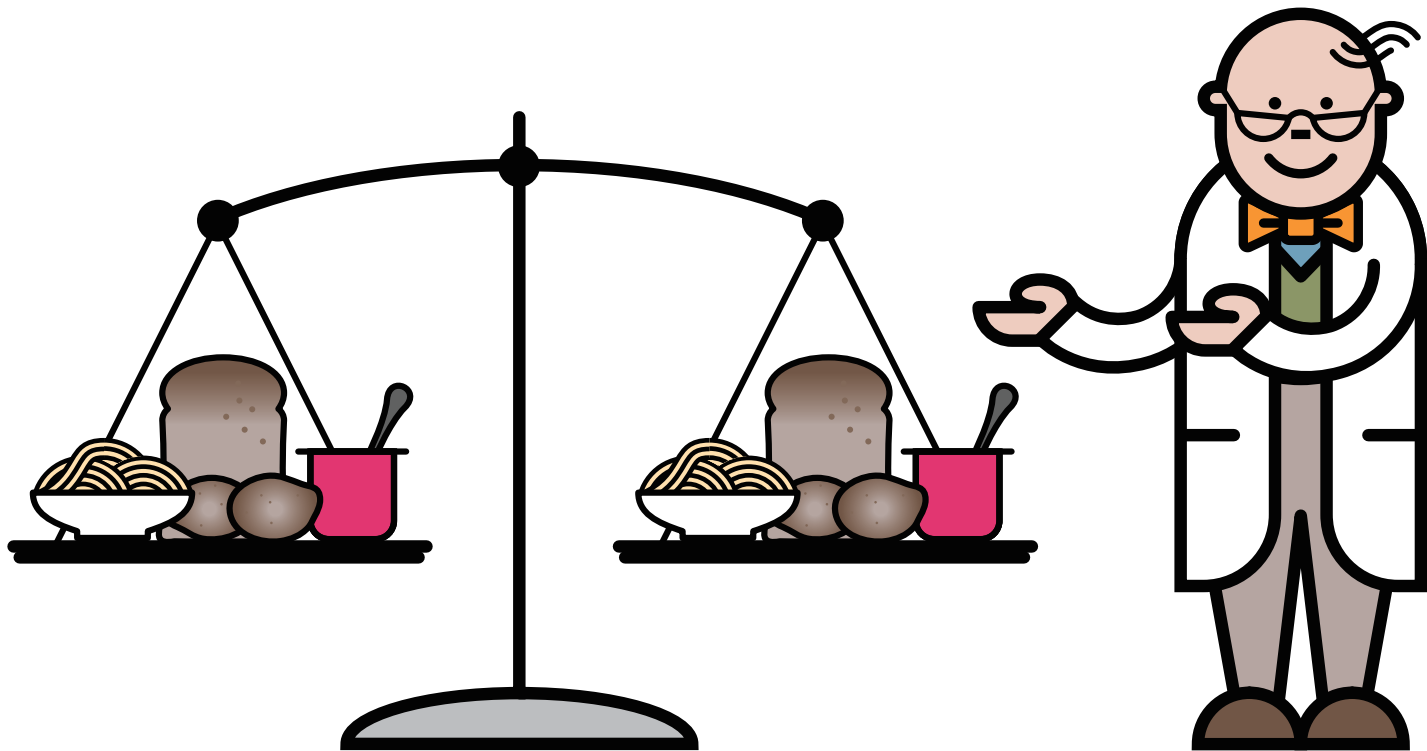
A protein substitute

Carnitine medication



# Protein balance is needed in GAI

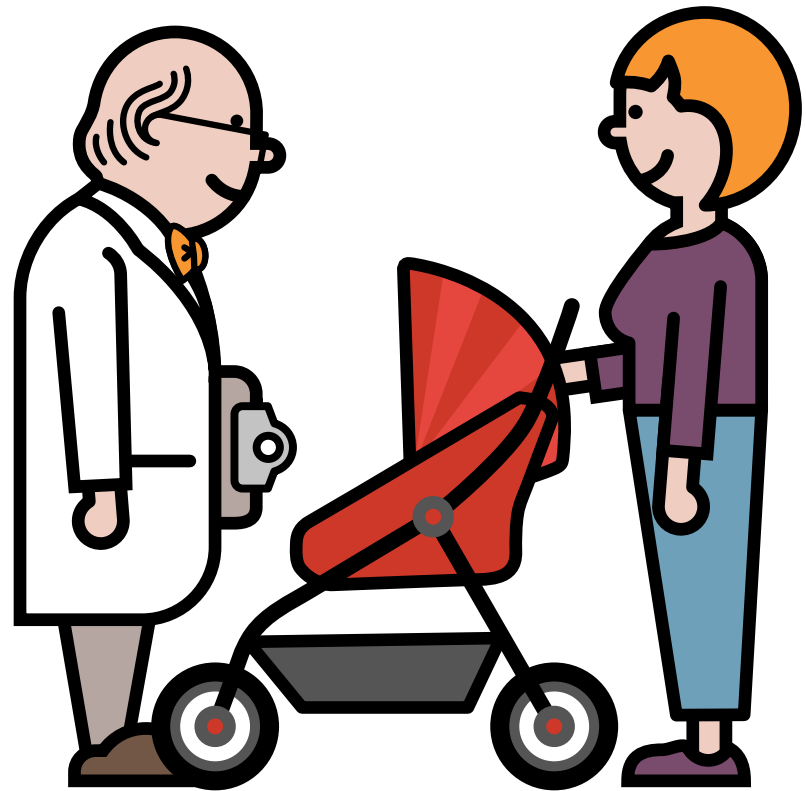
In GAI, it is important that enough protein is given for growth ... but not too much as toxic chemicals will be made.



# Measured protein intake

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

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

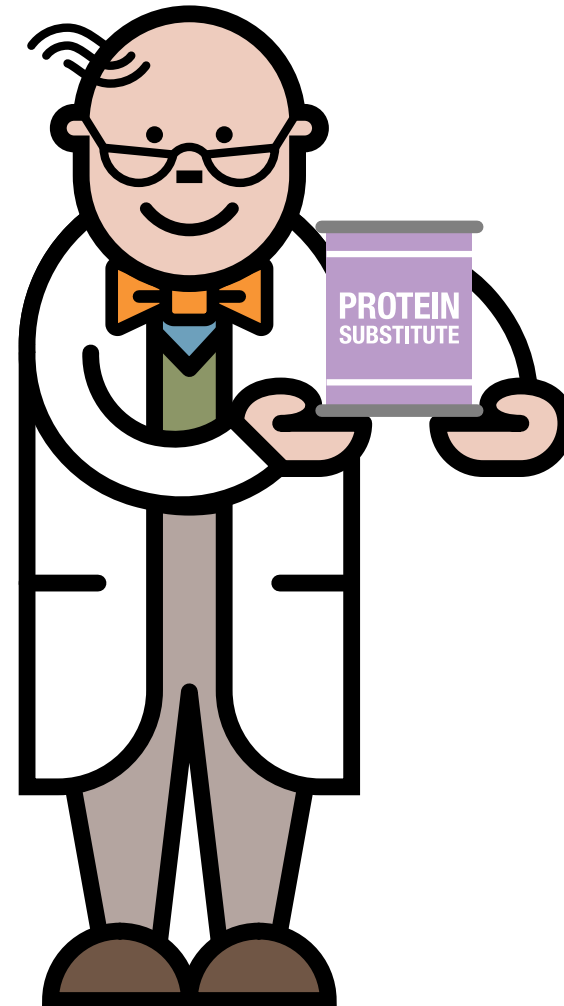


# Protein substitute

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.

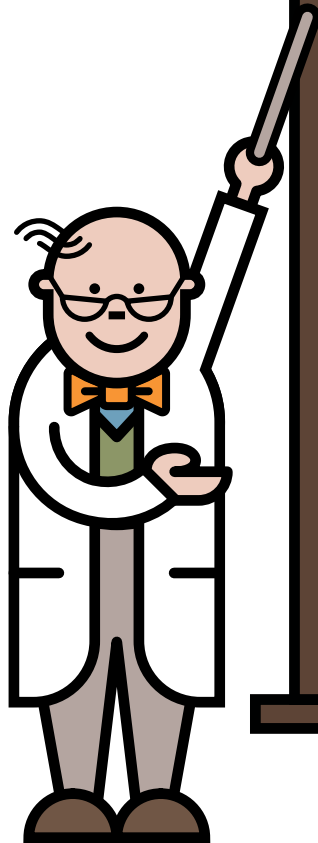


# How is GAI managed during illness?

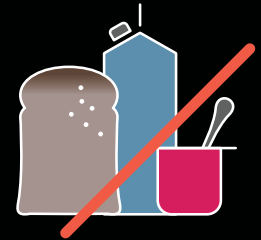
- During any childhood illness, an emergency regimen is given
- Illnesses can cause catabolism or protein breakdown
- This will lead to a build up of glutaric acid and cause a metabolic crisis



# How is GAI 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



# How is GAI 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



# How is GAI managed during illness?



Stop all protein in food & drink

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

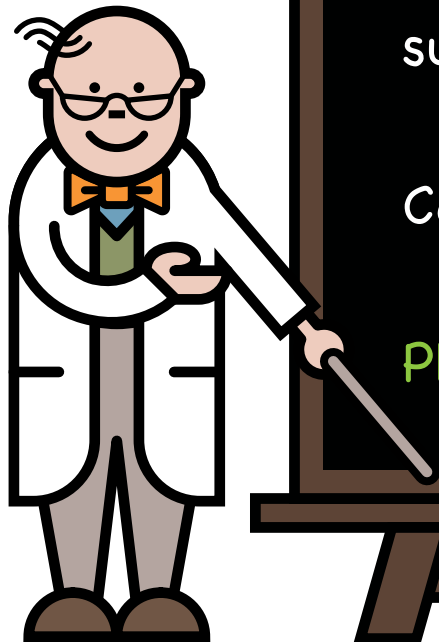
Carnitine medication as prescribed

Phone your metabolic team

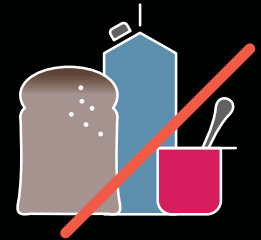




# How is GAI managed during illness?



Stop all protein in food & drink



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



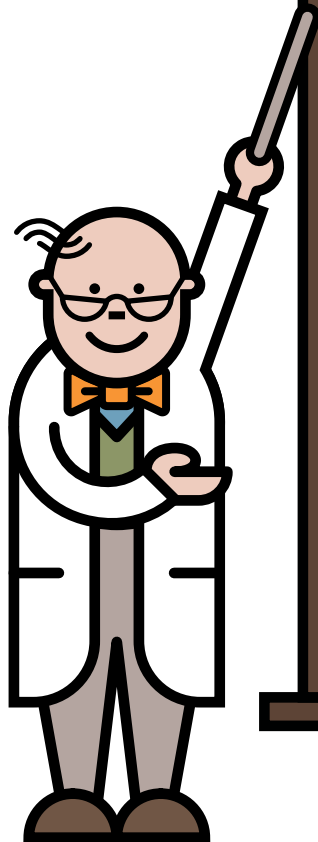
Carnitine medication as prescribed



Phone your metabolic team



# 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



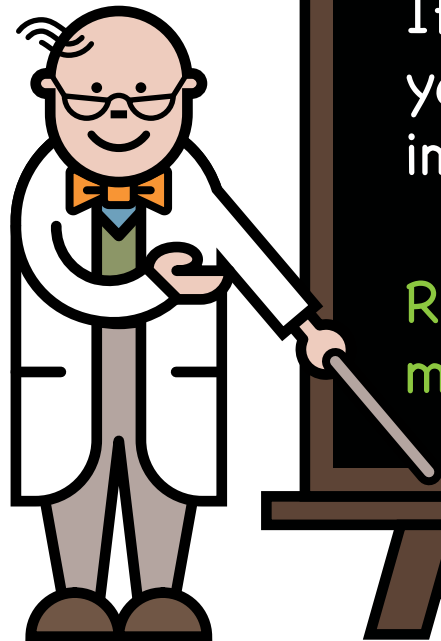
If symptoms continue and/or  
you are worried, go  
immediately to the hospital



Regularly update your  
metabolic team



# Checklist for illness



Always take full amounts  
of emergency feeds as  
prescribed



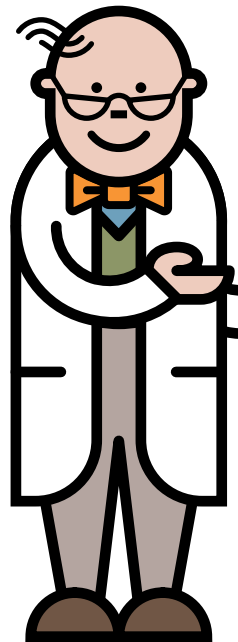
If symptoms continue and/or  
you are worried, go  
immediately to the hospital



Regularly update your  
metabolic team

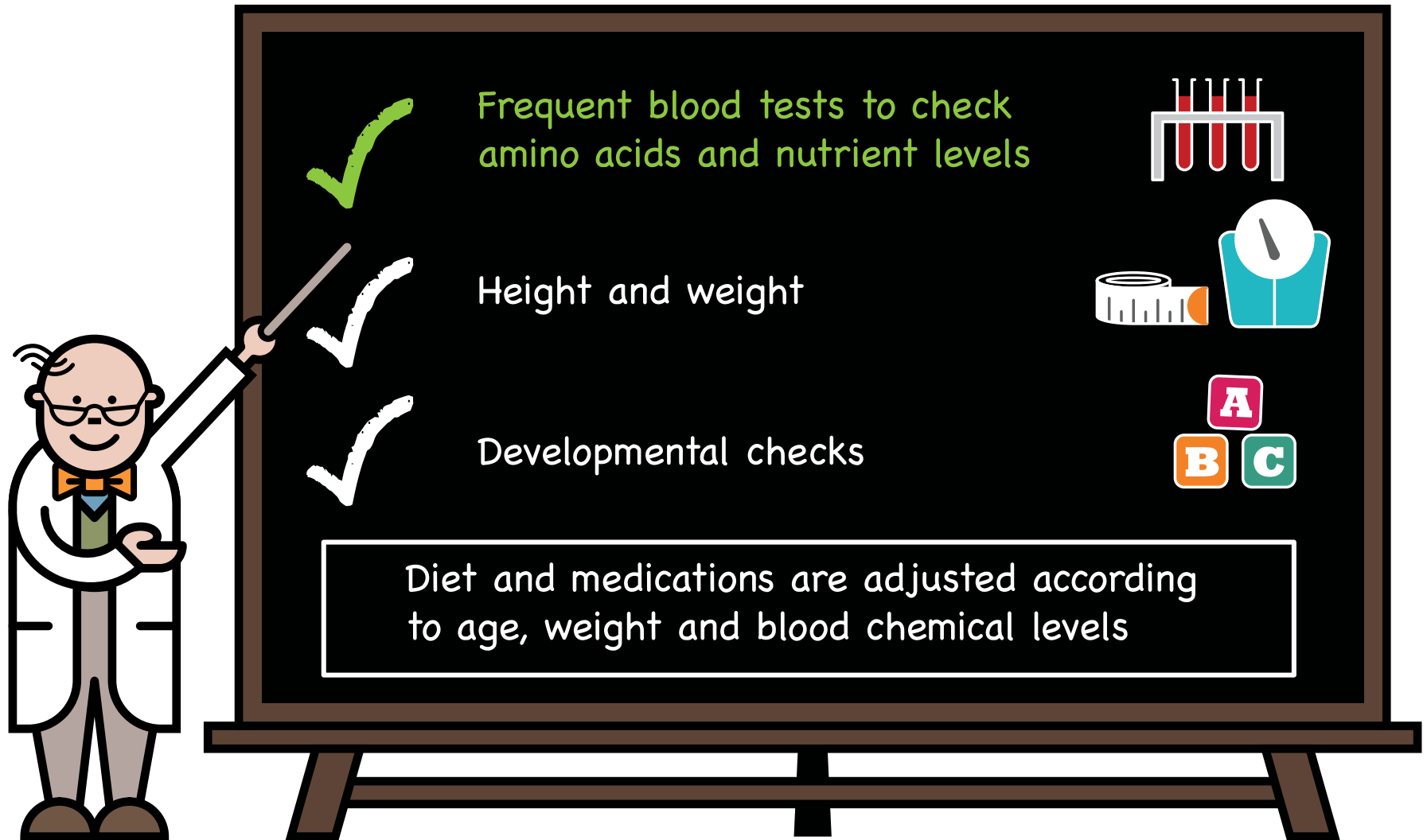


## Key message

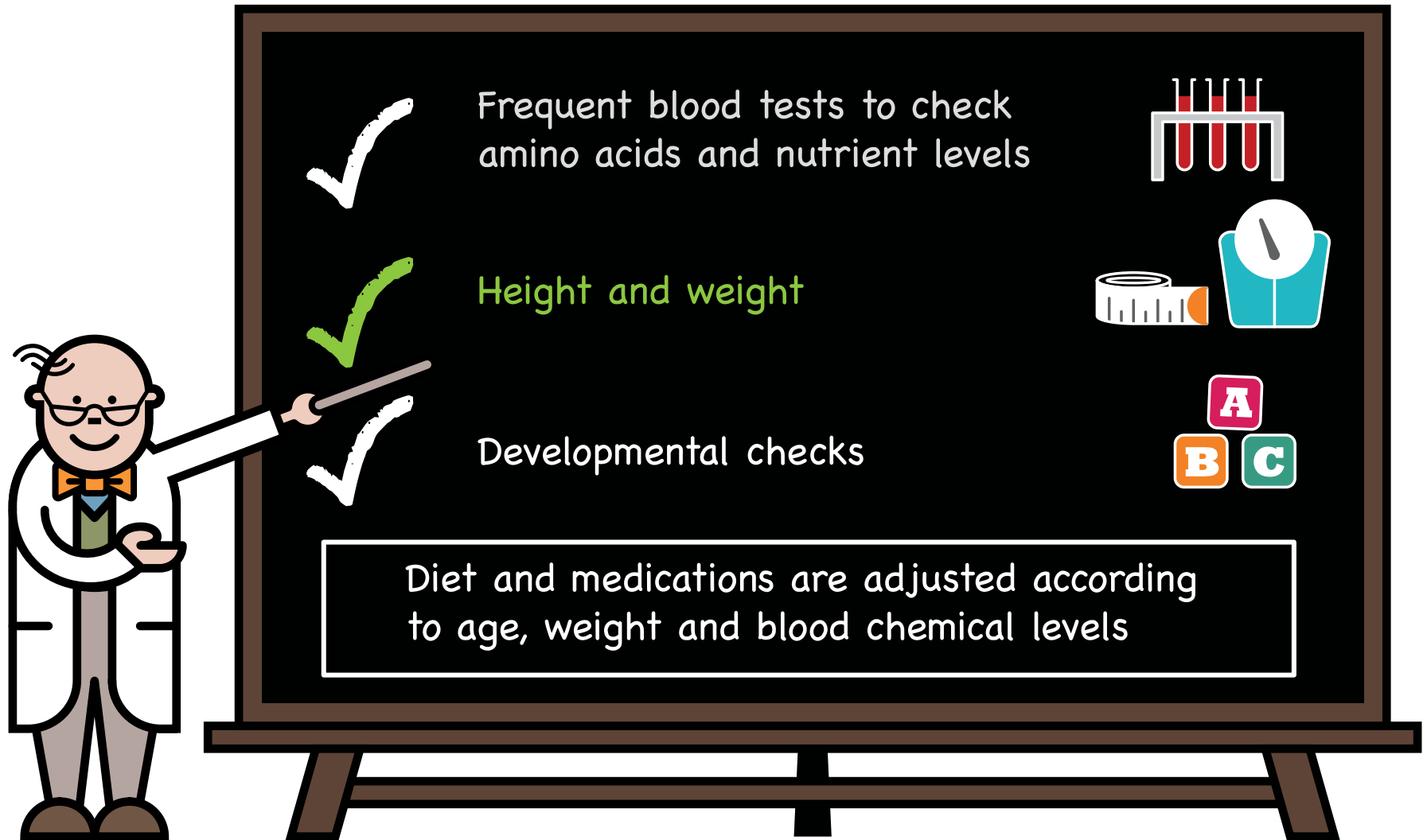


It is imperative that  
emergency feeds are  
started **promptly** and  
there are **no delays**  
in management.

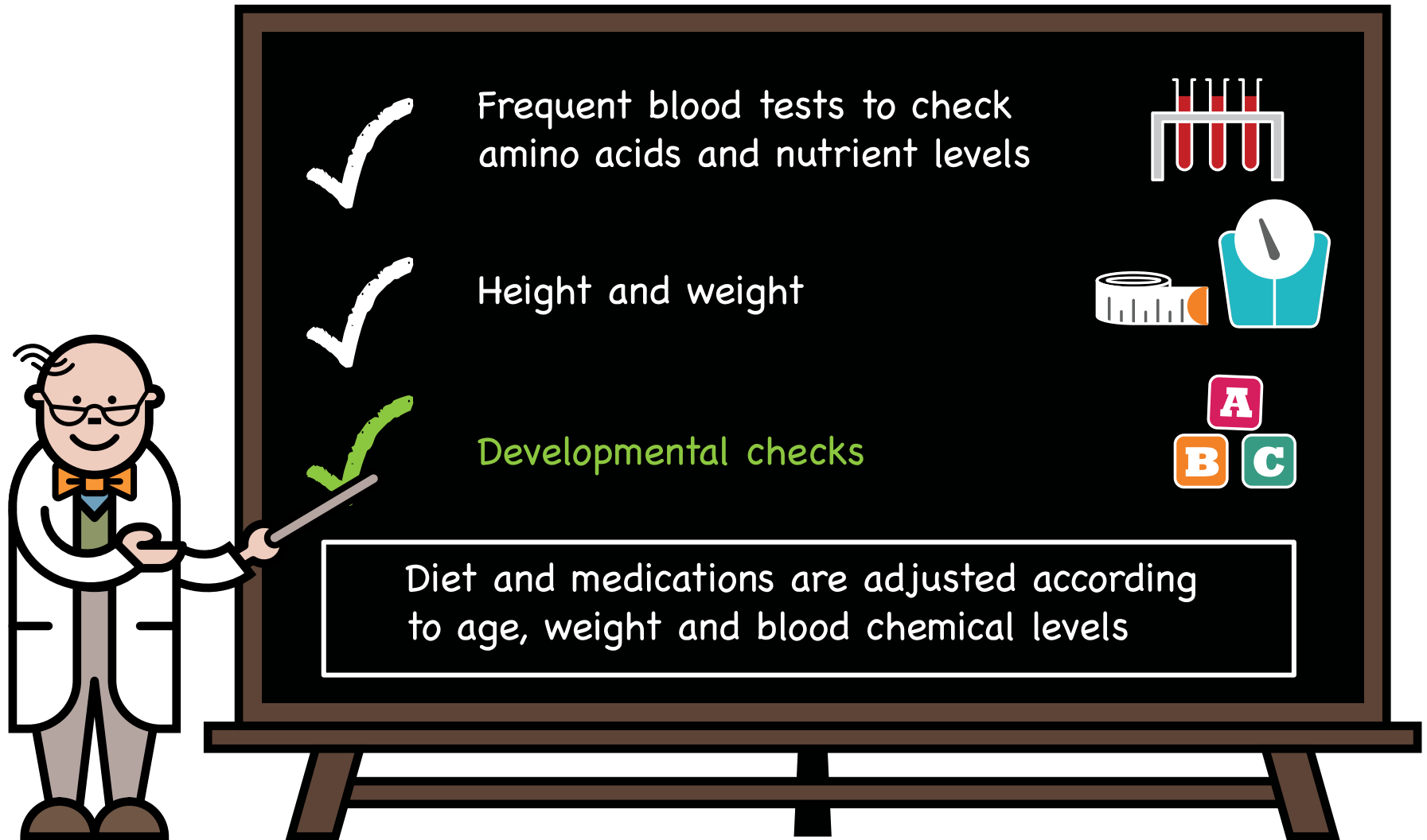
# How is GAI monitored?



# How is GAI monitored?

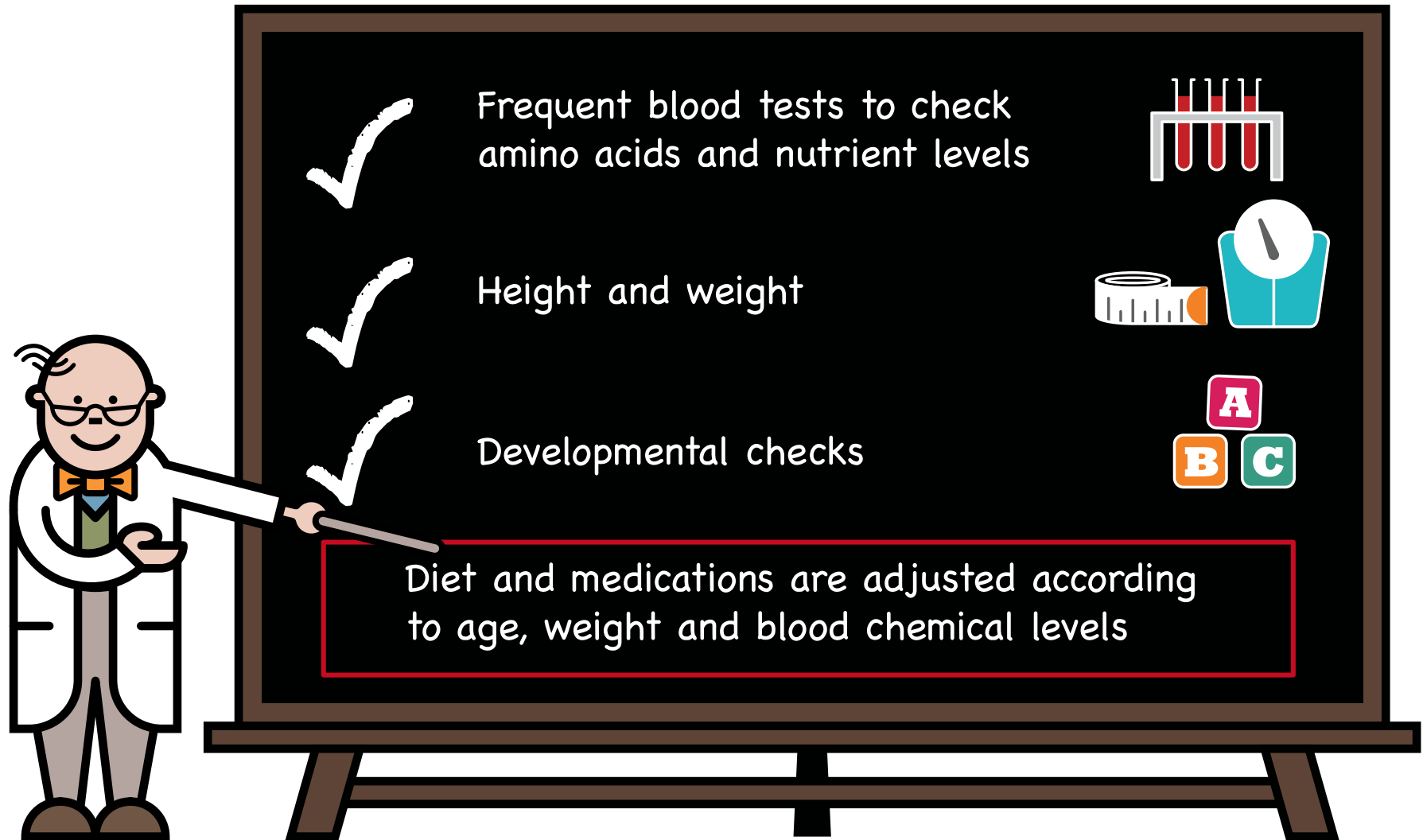


# How is GAI monitored?

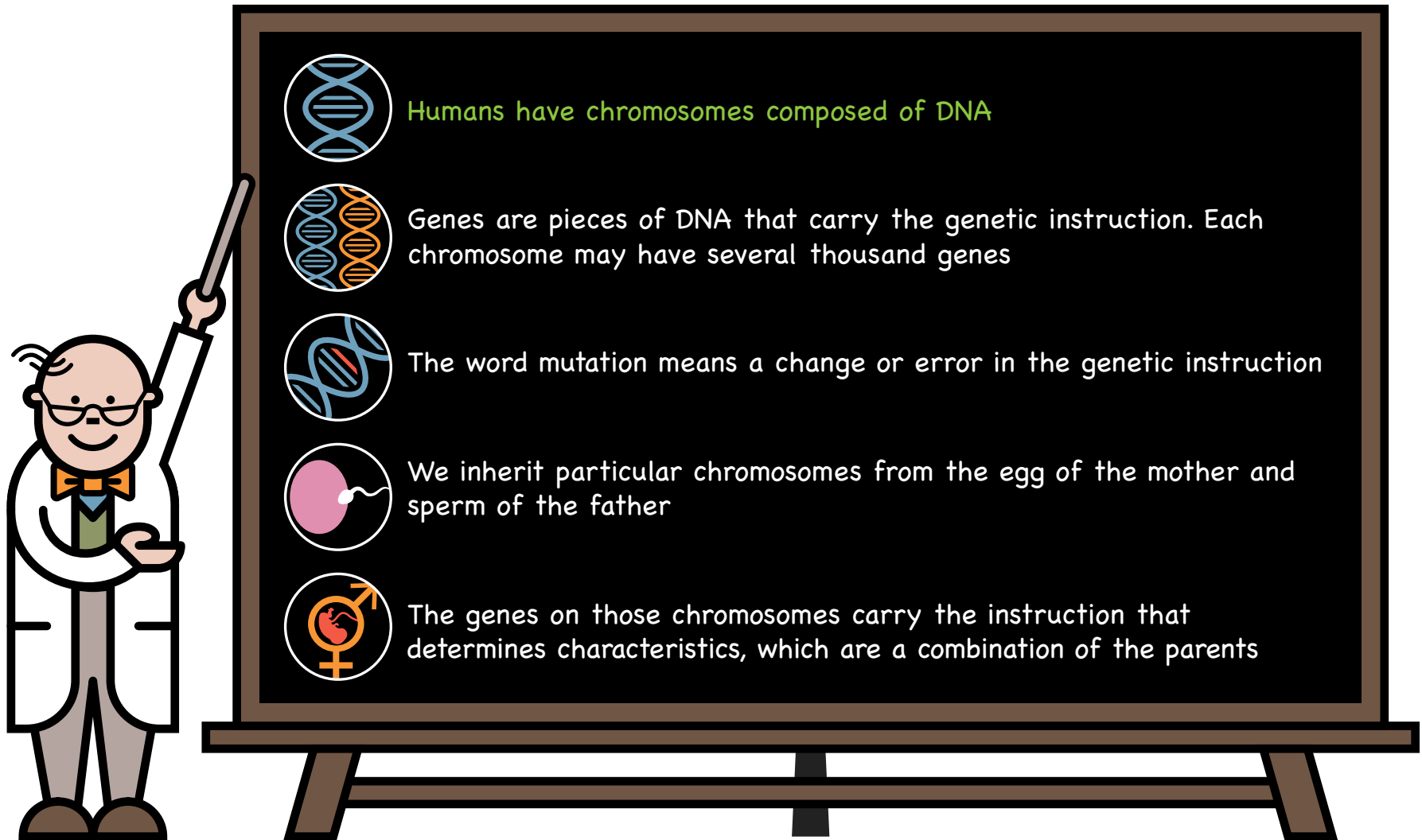




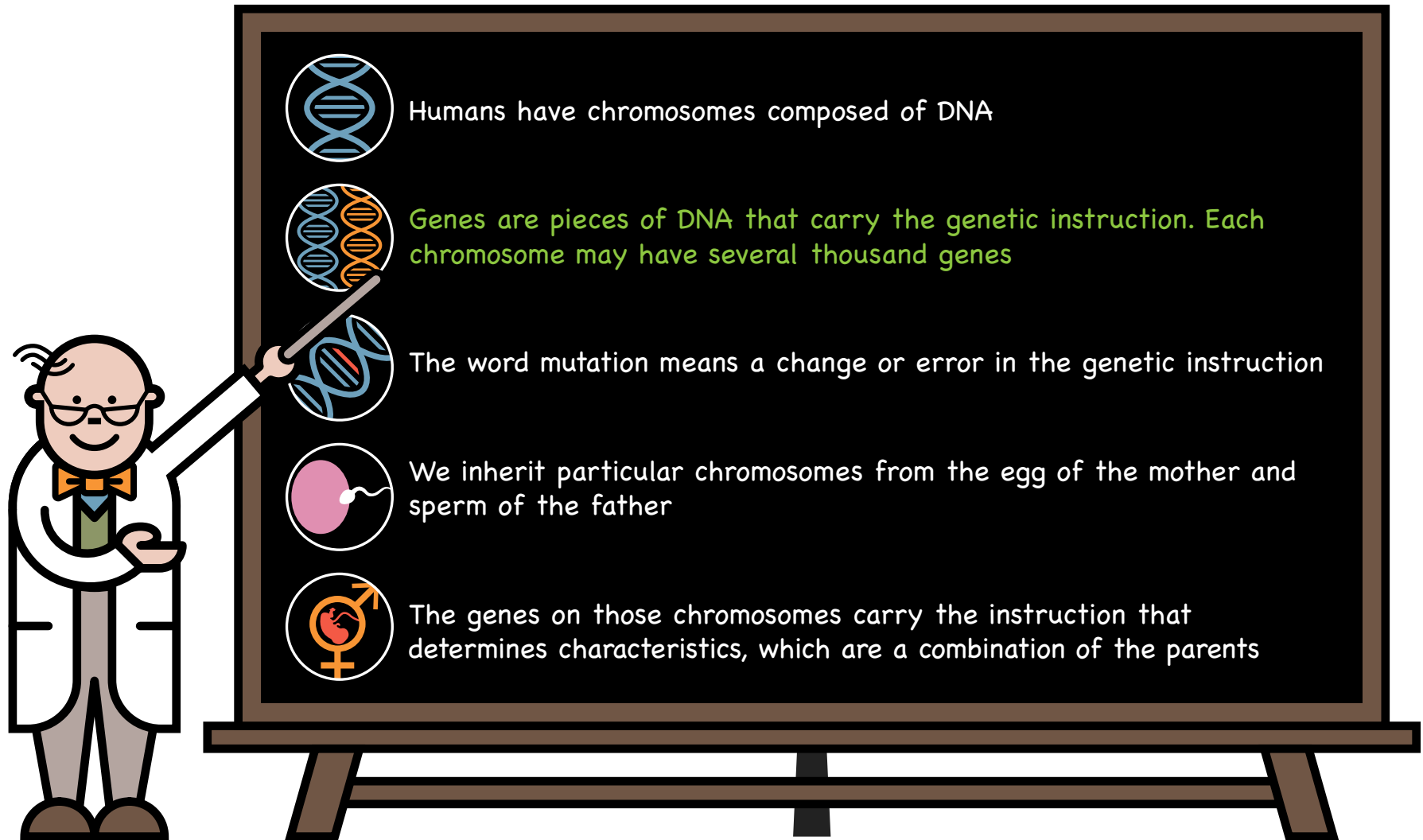
# How is GAI monitored?



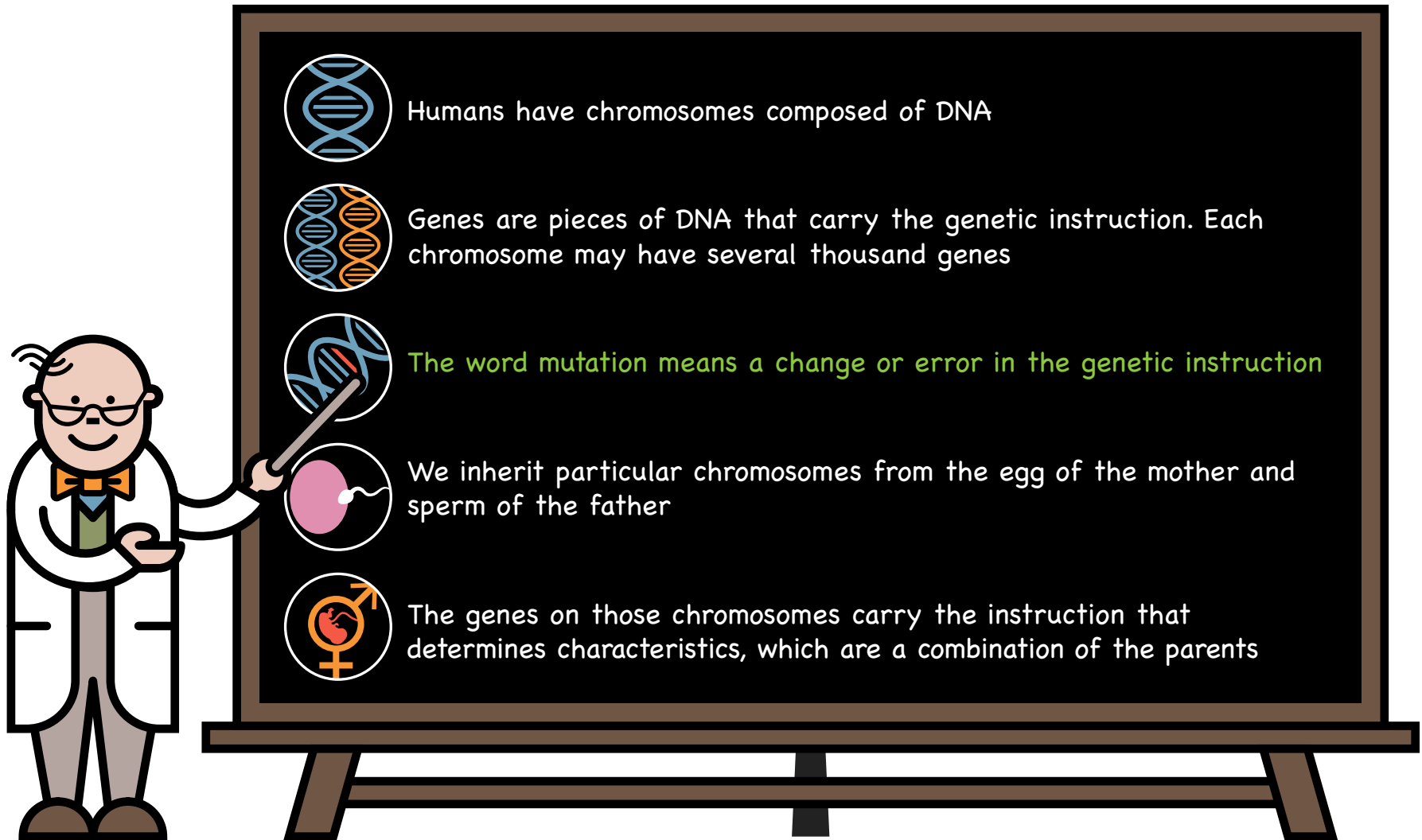
# Chromosomes, genes, mutations



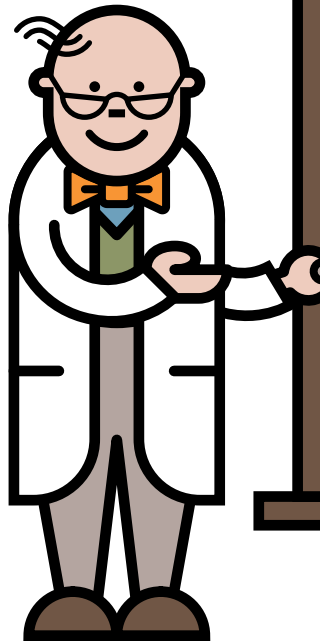
# Chromosomes, genes, mutations



# Chromosomes, genes, mutations



# 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

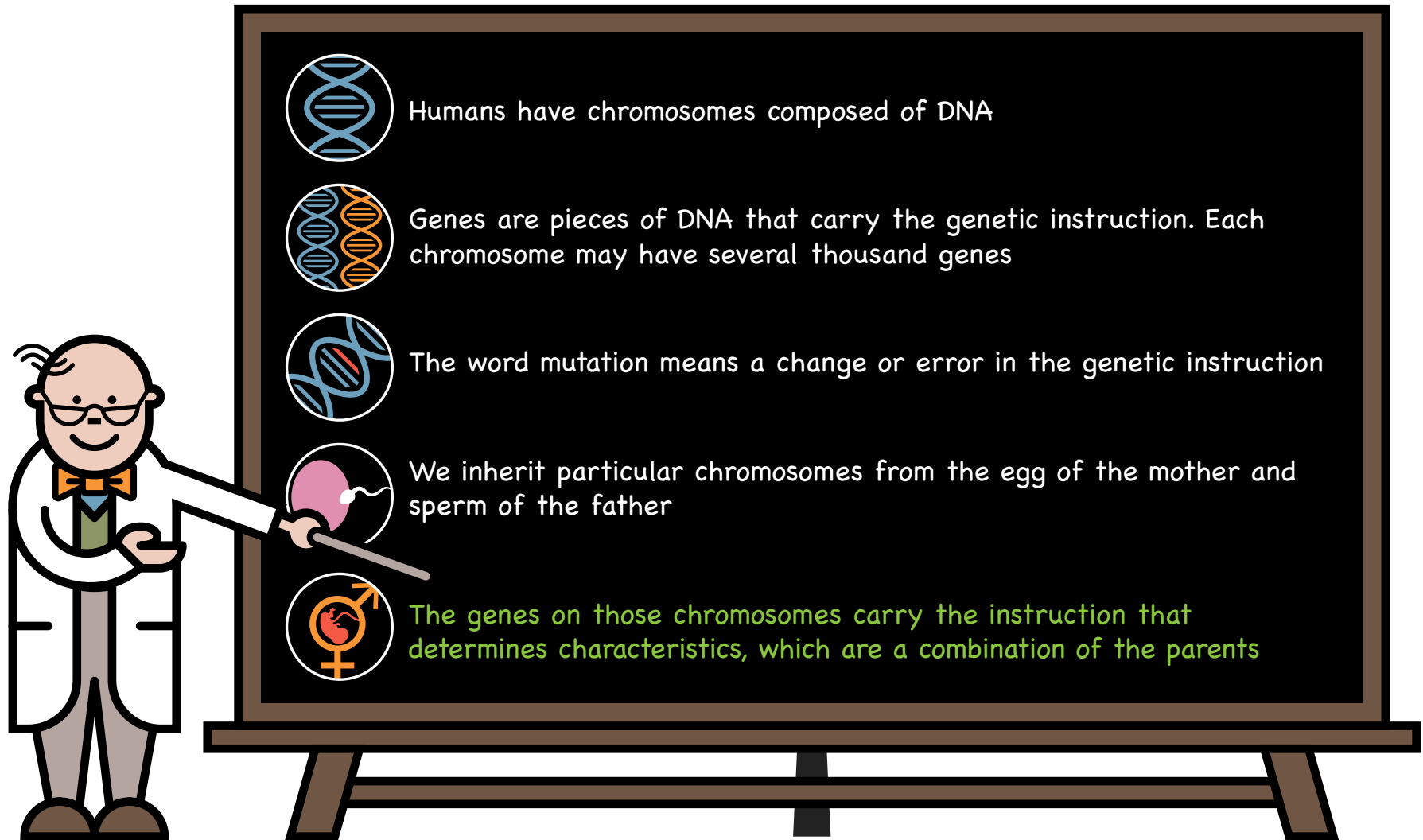


We inherit particular chromosomes from the egg of the mother and sperm of the father

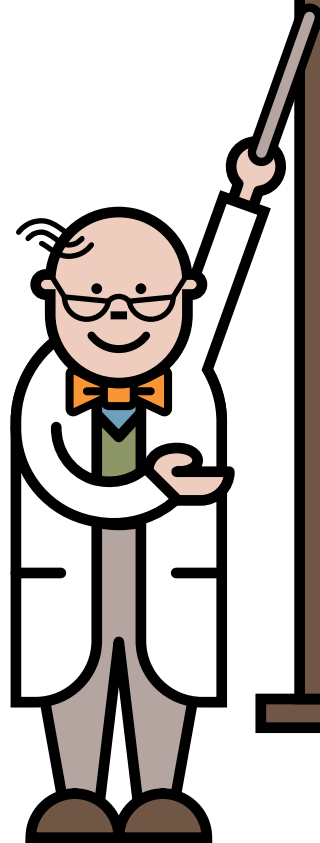


The genes on those chromosomes carry the instruction that determines characteristics, which are a combination of the parents

# Chromosomes, genes, mutations



# Inheritance



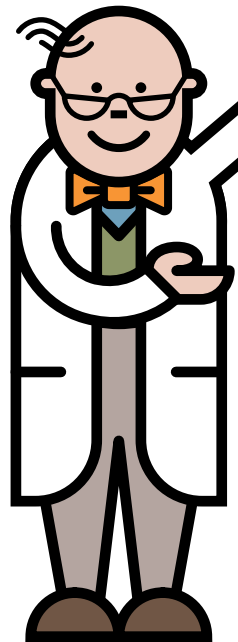
GAI is an inherited condition. There is nothing that could have been done to prevent your baby from having GAI

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

Parents of children with GAI are carriers of the condition

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

# Inheritance



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



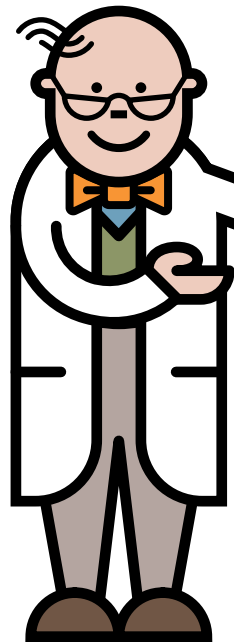
Everyone has a pair of genes that make the glutaryl-CoA dehydrogenase enzyme. In children with GAI, neither of these genes works correctly. These children inherit one non-working GAI gene from each parent

Parents of children with GAI are carriers of the condition

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



# Inheritance



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



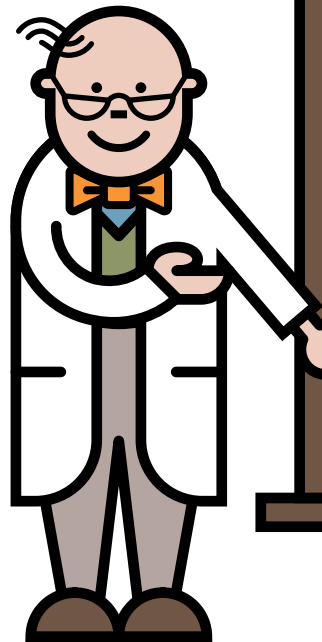
Everyone has a pair of genes that make the glutaryl-CoA dehydrogenase enzyme. In children with GAI, neither of these genes works correctly. These children inherit one non-working GAI gene from each parent



Parents of children with GAI are carriers of the condition

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

# Inheritance



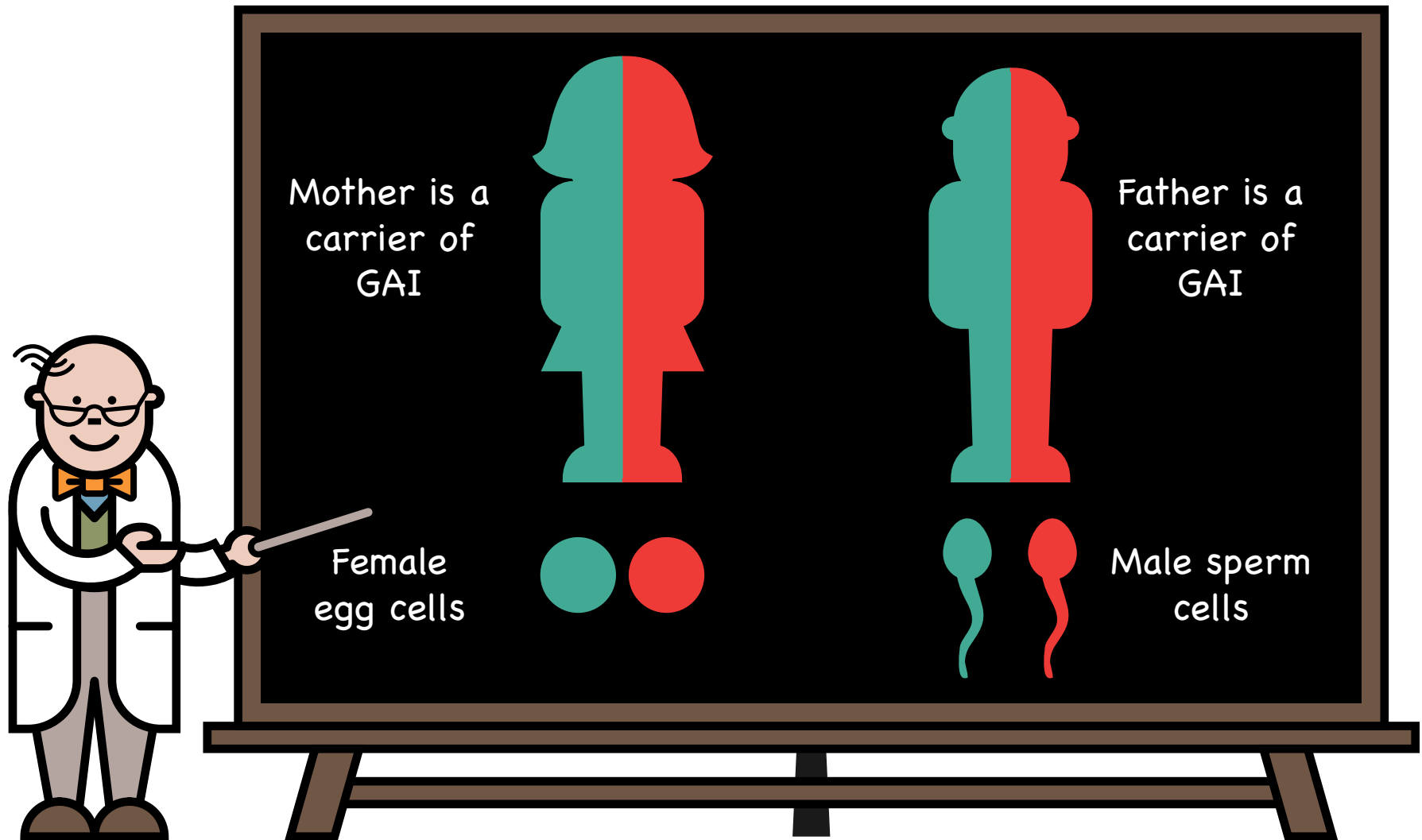
✓ GAI is an inherited condition. There is nothing that could have been done to prevent your baby from having GAI

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

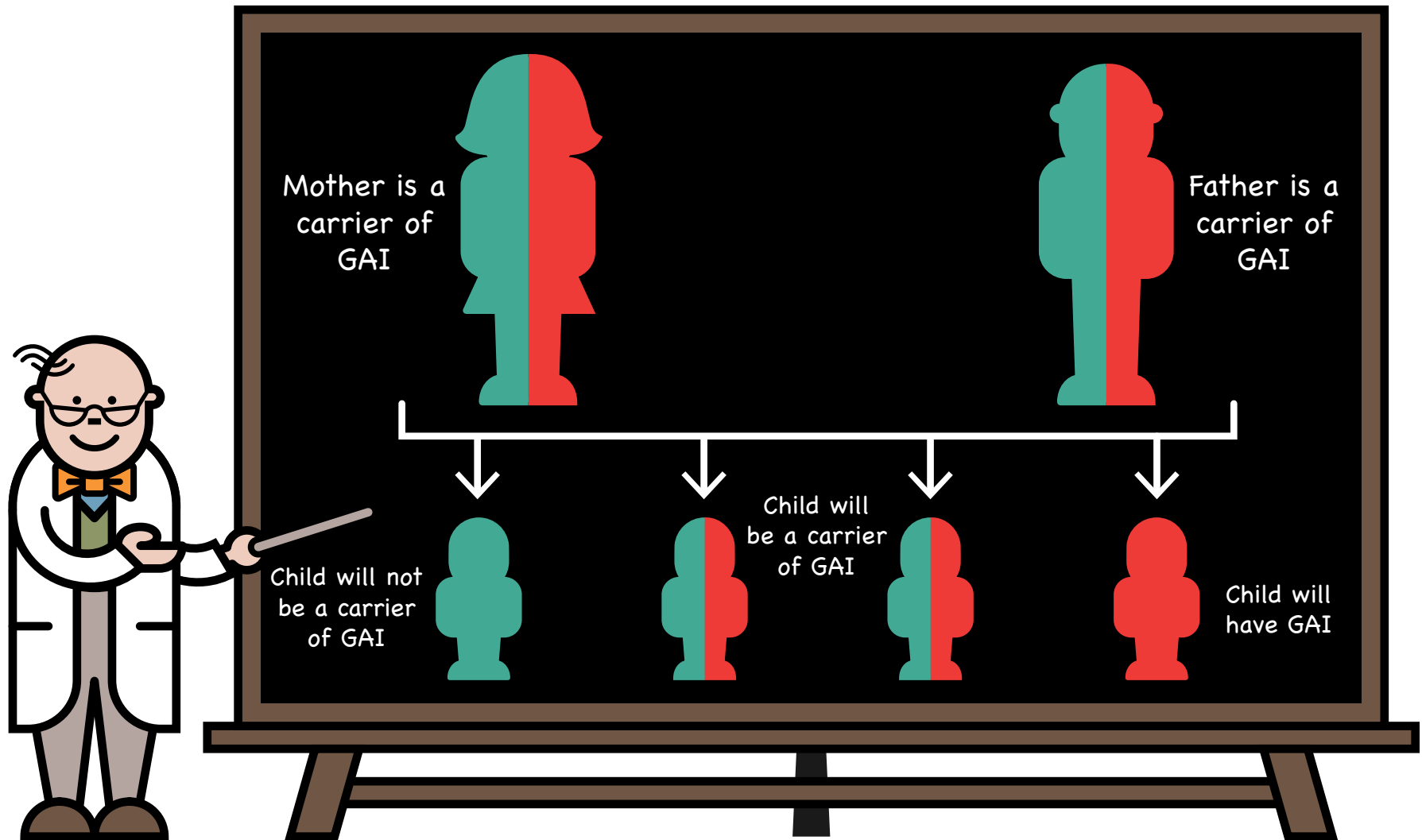
✓ Parents of children with GAI are carriers of the condition

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

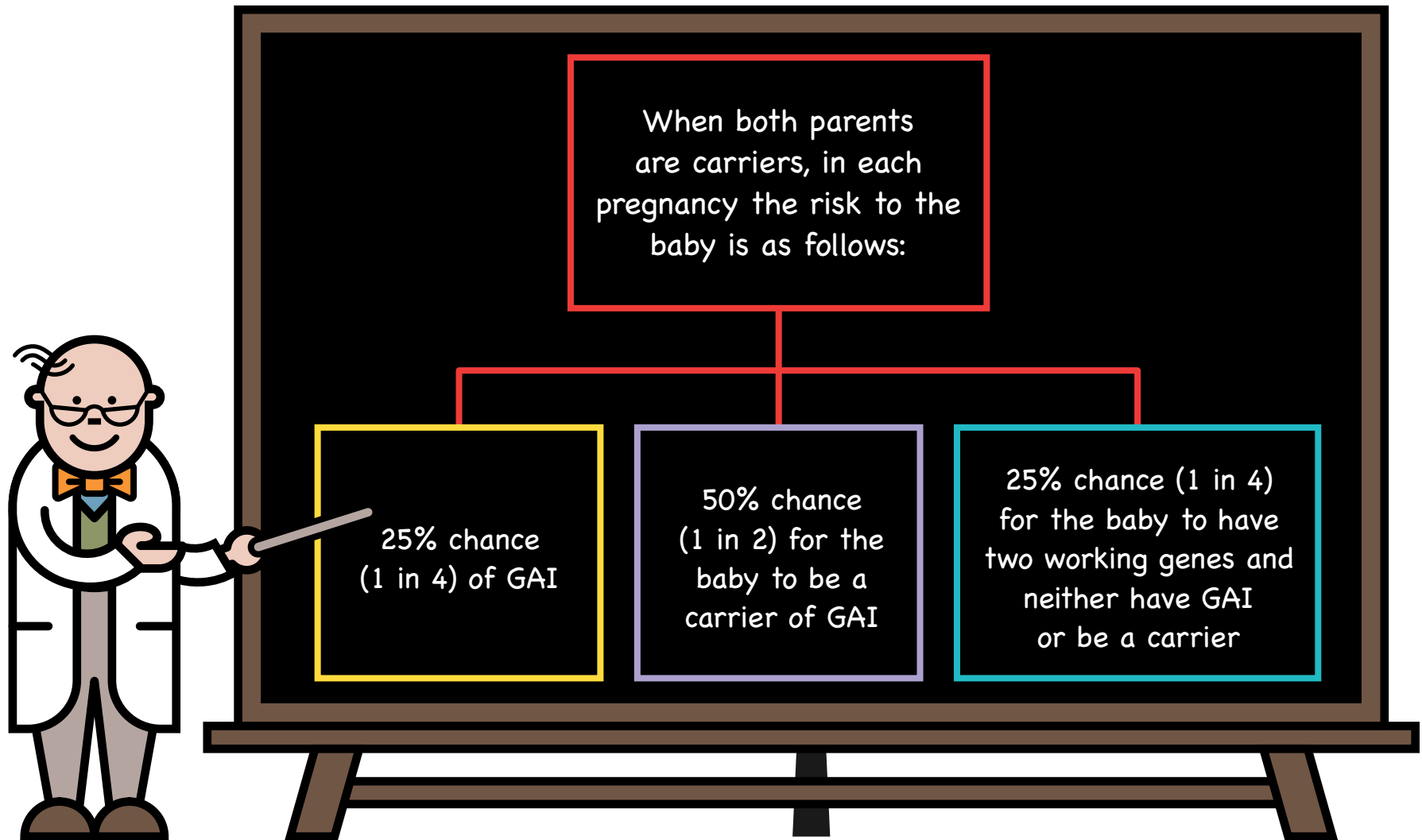
# Inheritance – Autosomal recessive (carriers of GAI)



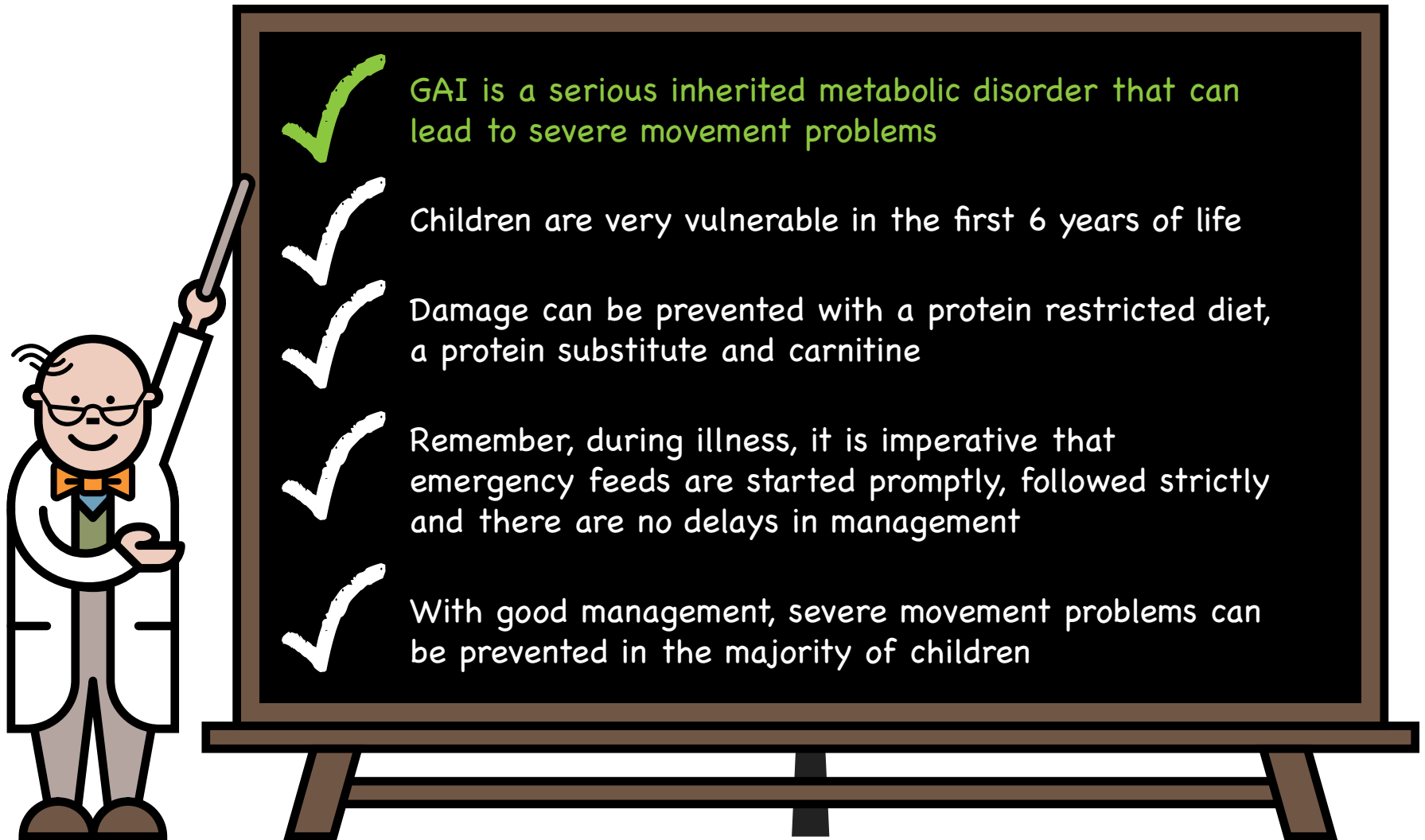
# Inheritance – Autosomal recessive – possible combinations



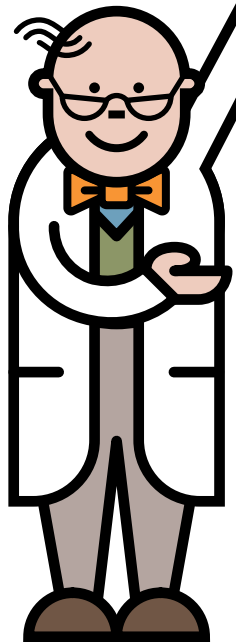
# Future pregnancies



# Take home messages



# Take home messages



GAI 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

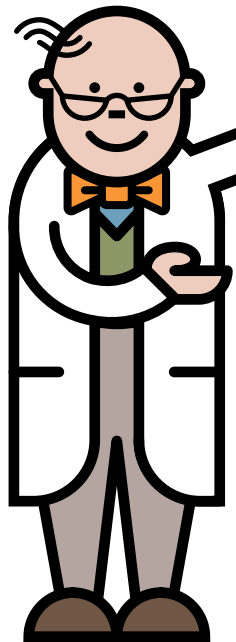


Remember, during illness, it is imperative that emergency feeds are started promptly, followed strictly and there are no delays in management



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

# Take home messages



GAI 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



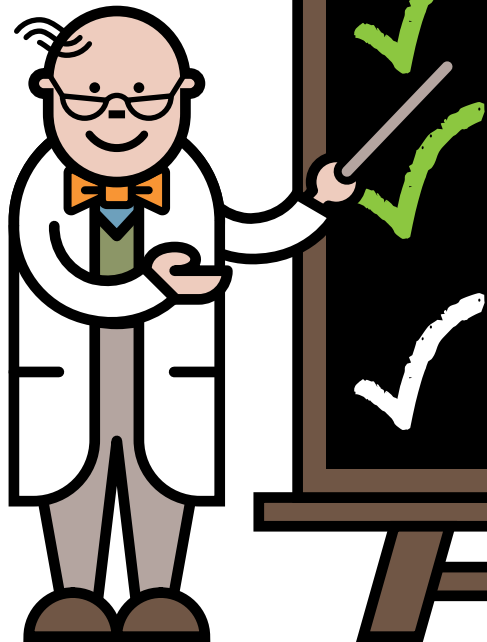
Remember, during illness, it is imperative that emergency feeds are started promptly, followed strictly and there are no delays in management



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



# Take home messages



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

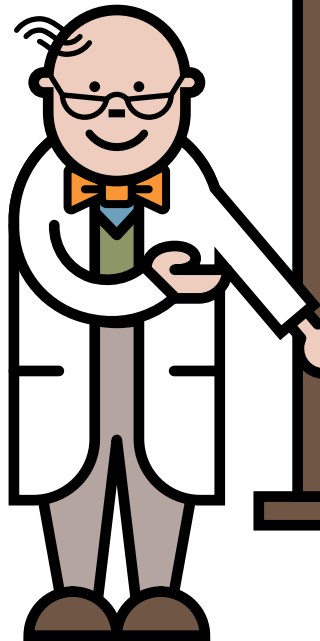
✓ Children are very vulnerable in the first 6 years of life

✓ 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



GAI 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

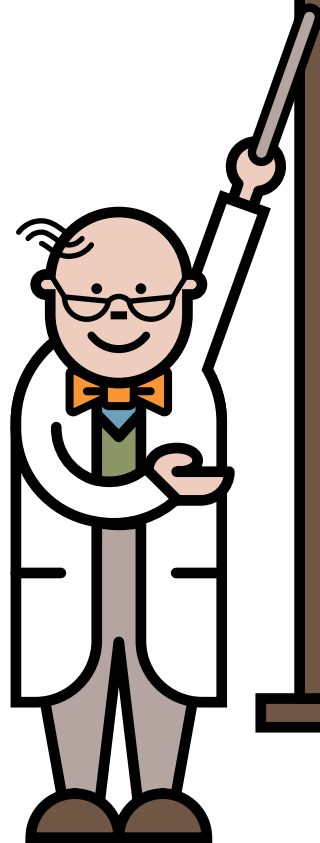


Remember, during illness, it is imperative that emergency feeds are started promptly, followed strictly and there are no delays in management



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

# Helpful hints



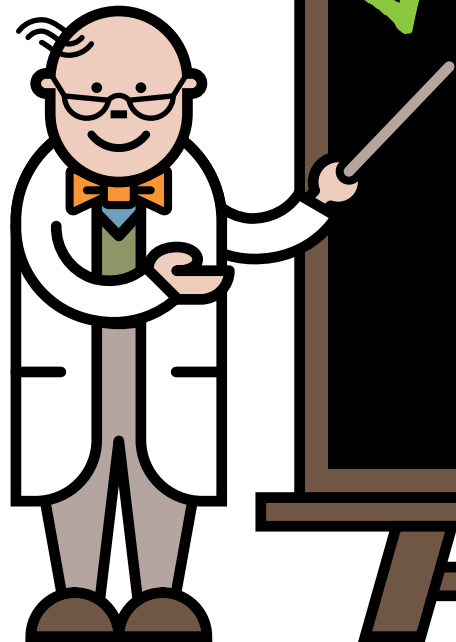
✓ Always ensure you have a good supply of your dietary products and medicines and that they are in date

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

# Helpful hints



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

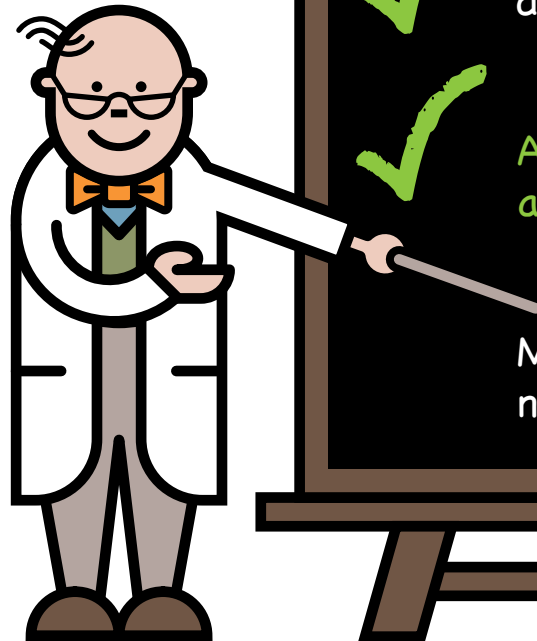


Your dietary products and medications are prescribed by your GP. These are obtained via a pharmacy or home delivery

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

# Helpful hints



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



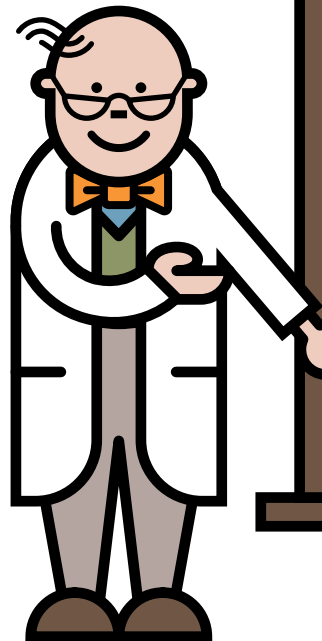
Your dietary products and medications are prescribed by your GP. These are obtained via a pharmacy or home delivery



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

# 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

# Who's who

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

Visit [www.nutricia.co.uk/patients-carers/living-with/low-protein-diet.html](http://www.nutricia.co.uk/patients-carers/living-with/low-protein-diet.html) 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.



@LowProConnect



LowProConnect



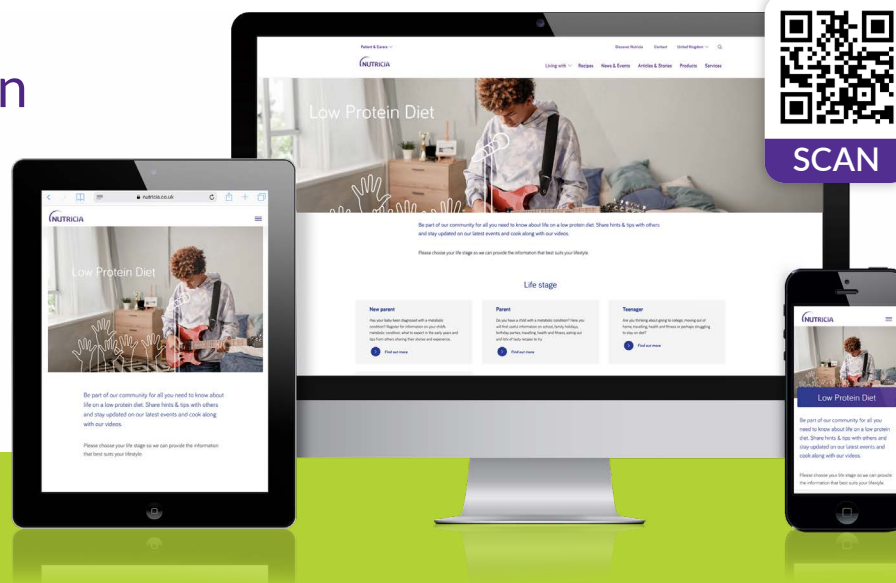
LowProteinConnect



LowProteinConnect



SCAN



**BIMDG**

British Inherited Metabolic Diseases Group



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

**NUTRICIA**  
LIFE-TRANSFORMING NUTRITION

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

**METABOLIC  
SUPPORT UK**

Your rare condition.  
Our common fight.

[www.metabolicsupportuk.org](http://www.metabolicsupportuk.org)