

# 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, MAY 2021

IVA

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).

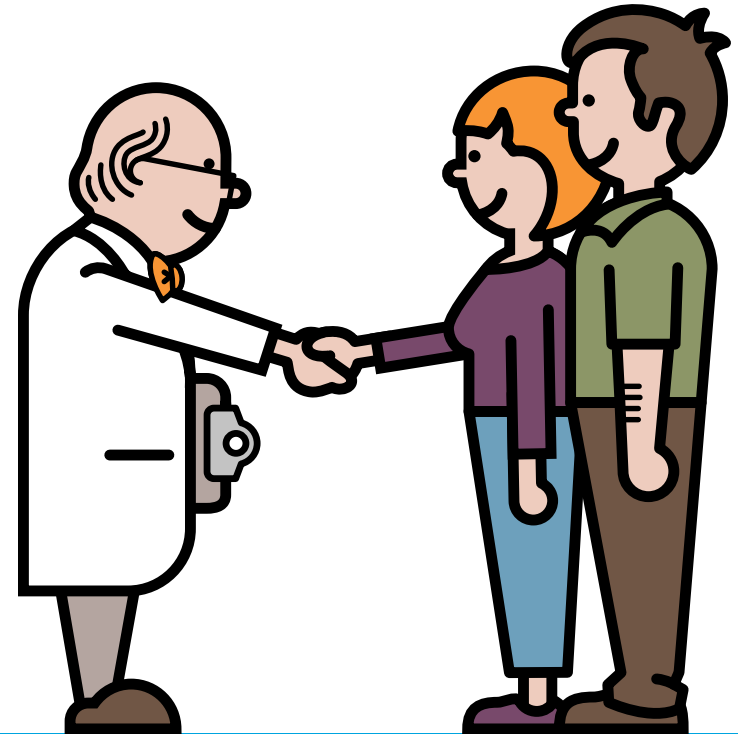
The team are Rachel Skeath, Karen van Wyk, Pat Portnoi and Anita MacDonald. The group is facilitated by Heidi Chan from Nutricia.

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.**

# IVA

Information for families following  
a positive newborn screening



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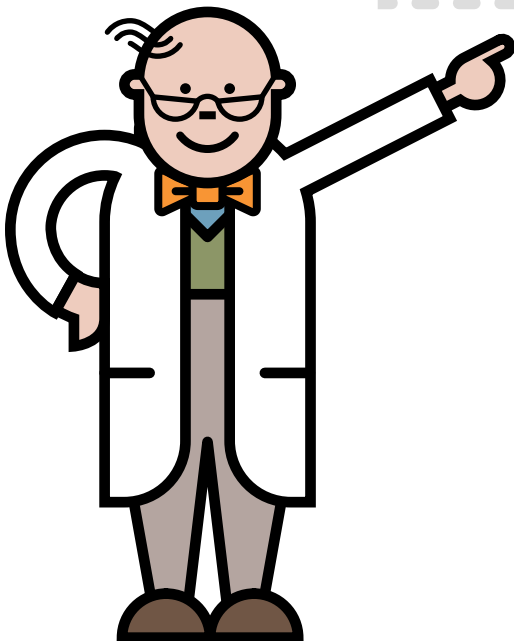
# What is IVA?

IVA stands for Isovaleric Acidaemia

It is an inherited metabolic condition

IsoValeric Acidaemia

IVA

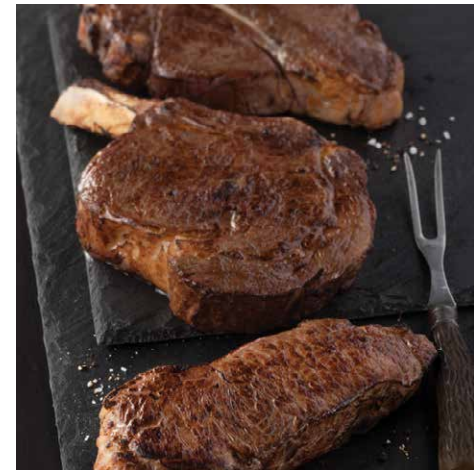


# IVA and protein

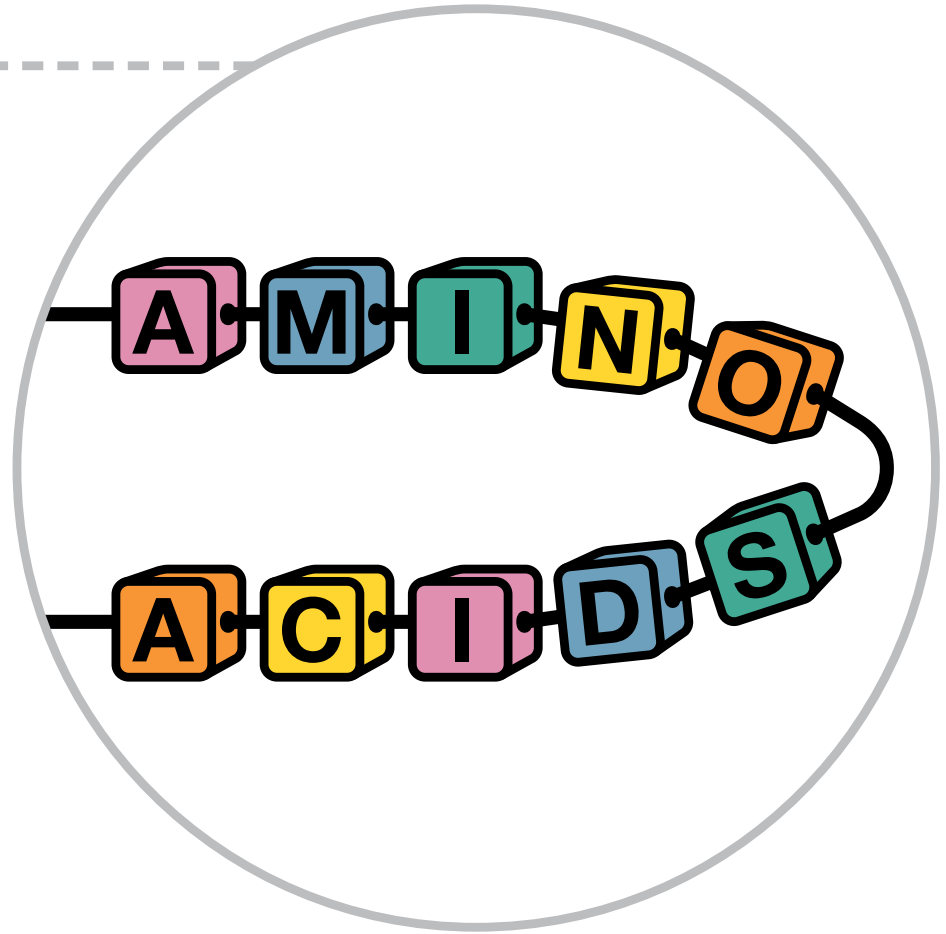
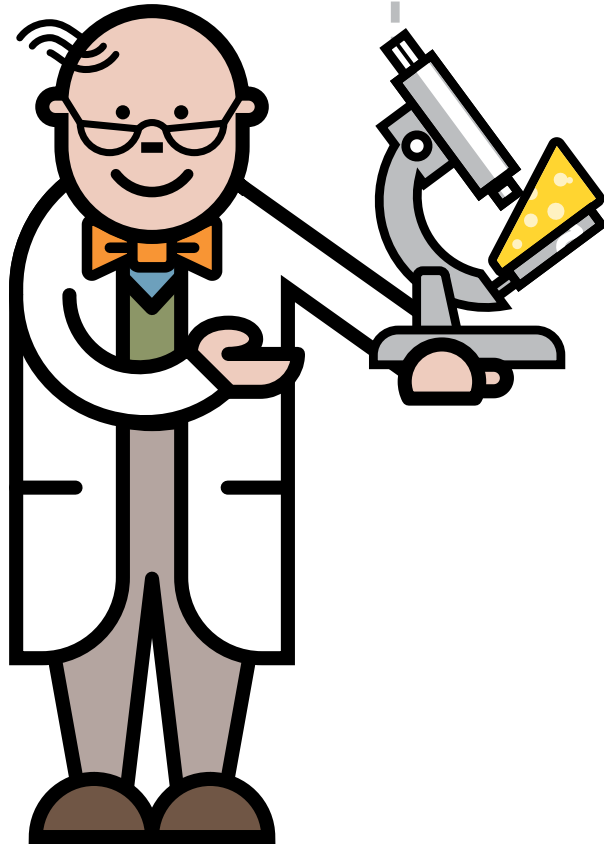
IVA affects the way your baby breaks down protein

Many foods contain protein

The body needs protein for growth and repair



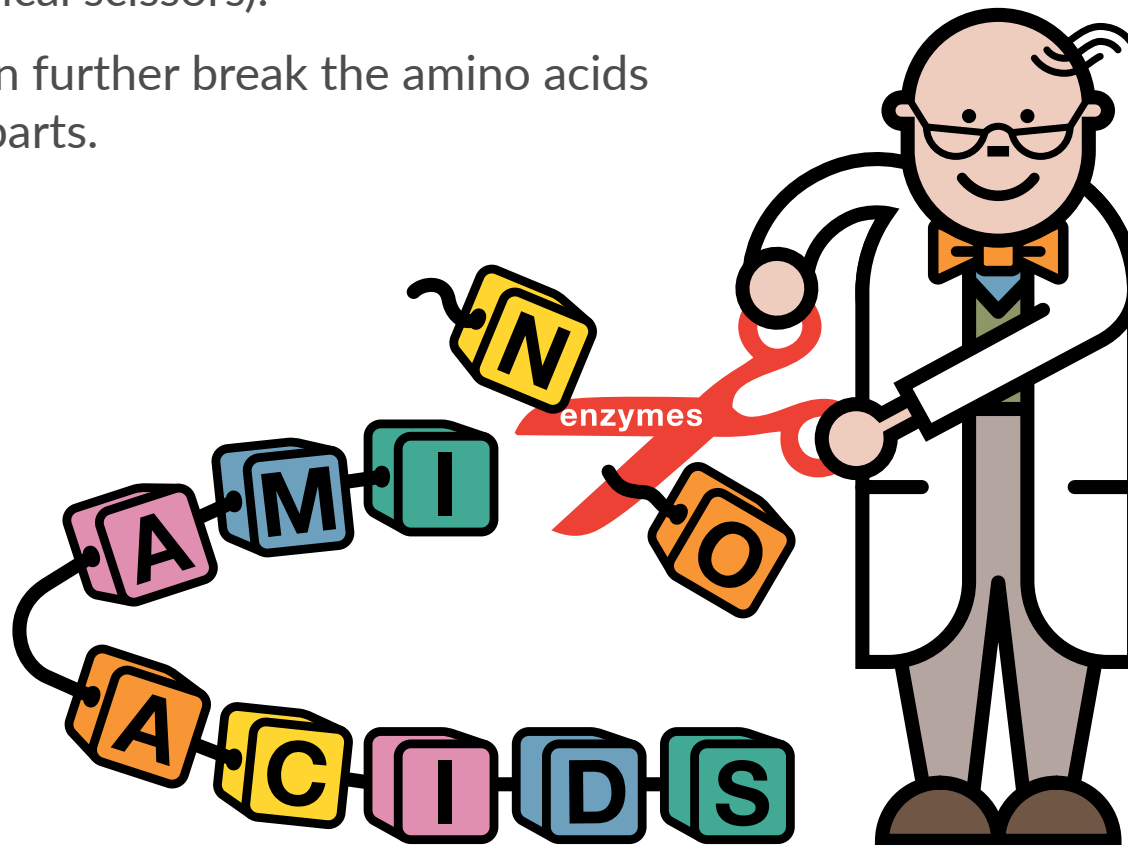
# What is protein?



# Protein and enzymes

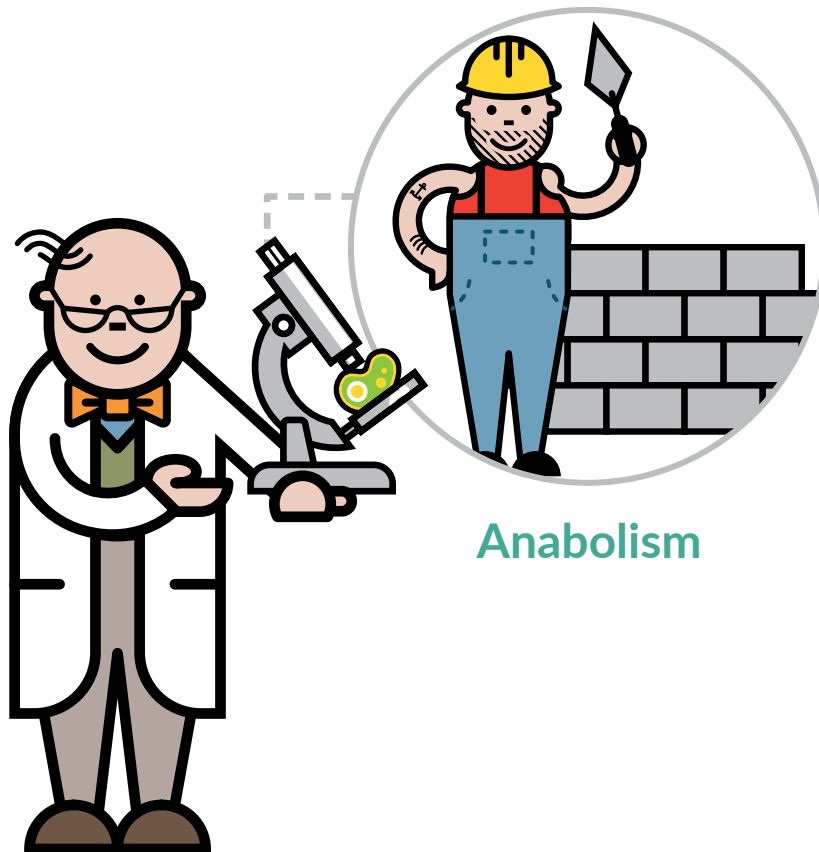
Protein is broken down into amino acids (building blocks of protein) by enzymes (which are like chemical scissors).

Enzymes then further break the amino acids into smaller parts.

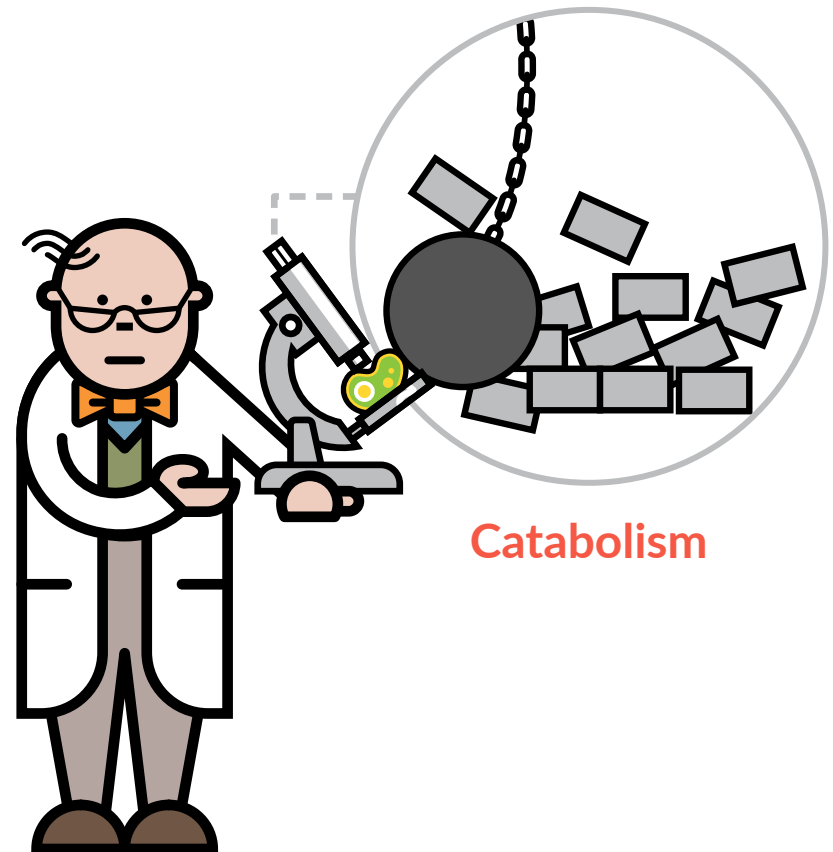


# Protein metabolism

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



Anabolism



Catabolism

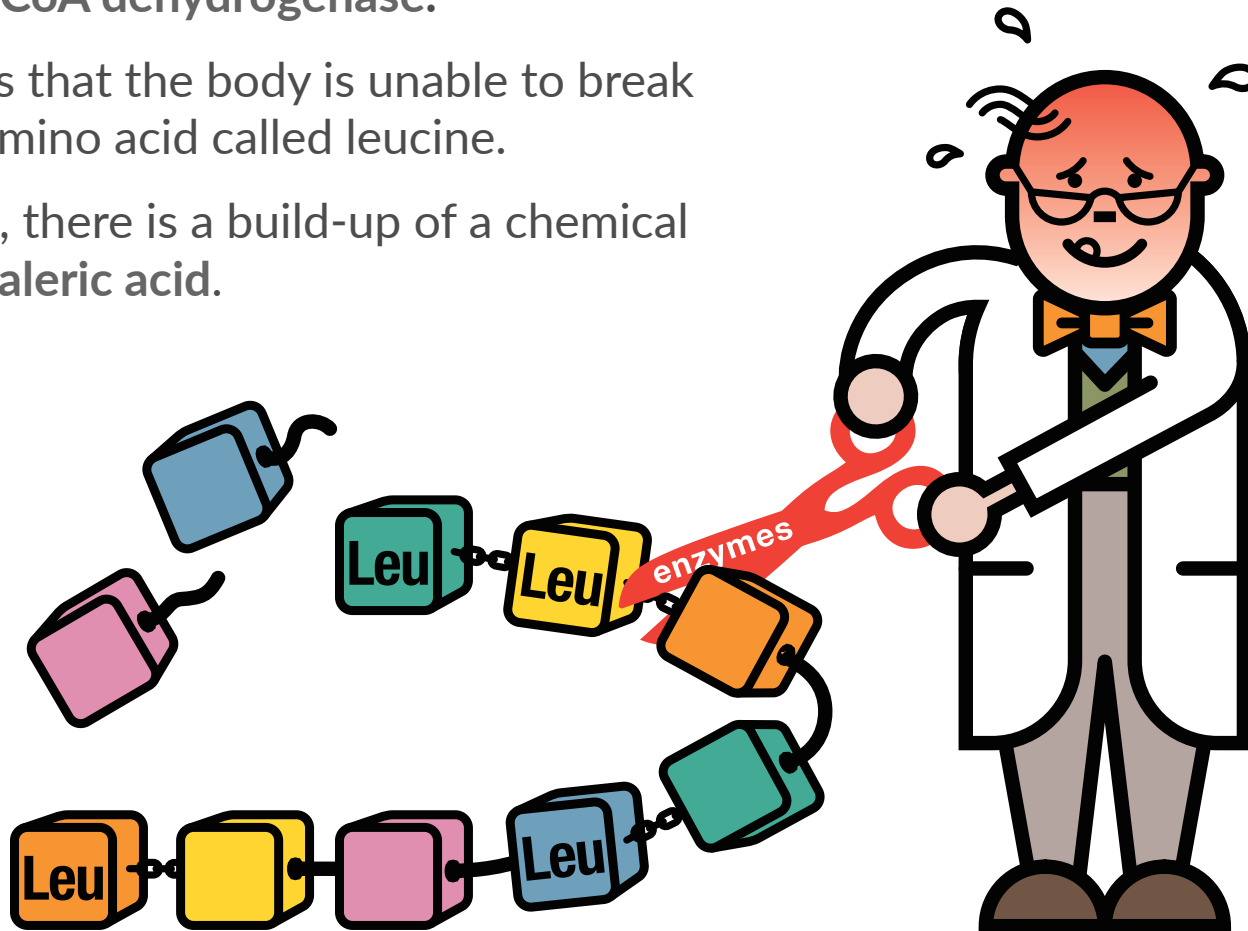


# What happens in IVA?

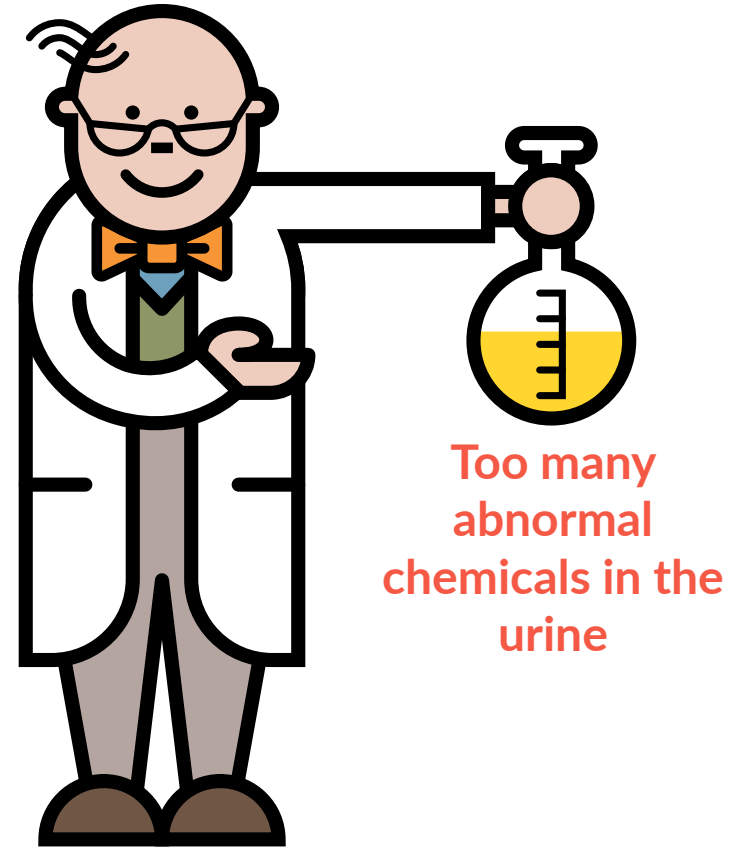
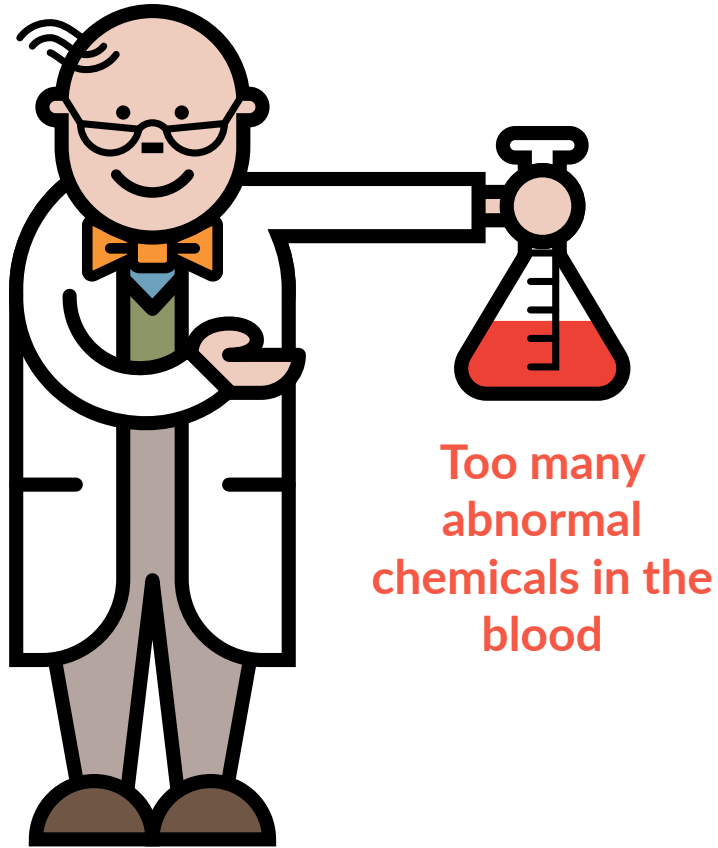
In IVA, the body lacks an enzyme called isovaleryl-CoA dehydrogenase.

This means that the body is unable to break down an amino acid called leucine.

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



# What does this cause?



# How is IVA diagnosed?

IVA is diagnosed by newborn screening. High levels of isovaleric acid are found in the blood.



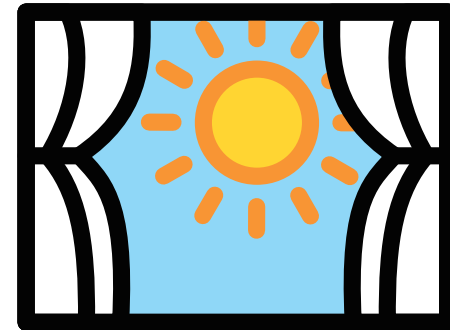
# What are the symptoms in IVA?

Some babies with IVA become ill in the first few days of life.

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

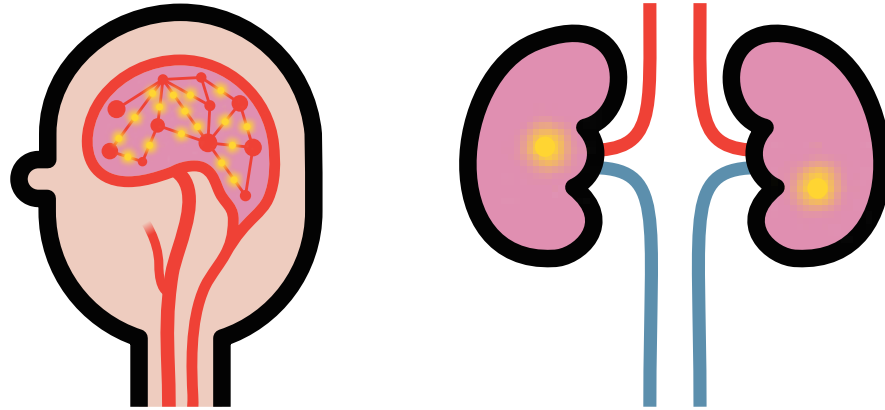
**The effects of IVA quickly become life-threatening if unmanaged**

Some children may not develop early symptoms but present when they are a little older with learning difficulties.

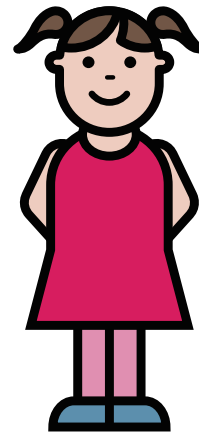


# What can go wrong in IVA?

The build up of harmful chemicals can damage the brain and other organs.



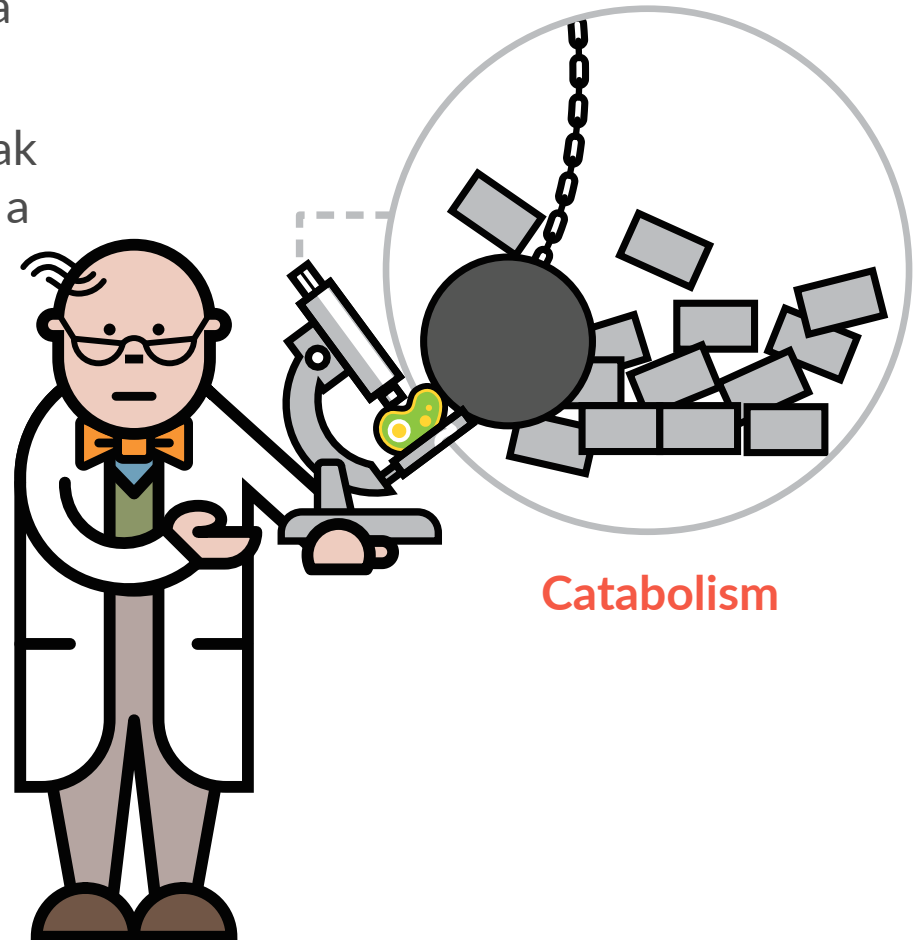
It may cause delays to normal development like walking and talking.



# What else happens in IVA?

If the body does not receive enough food e.g. during illness, 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.



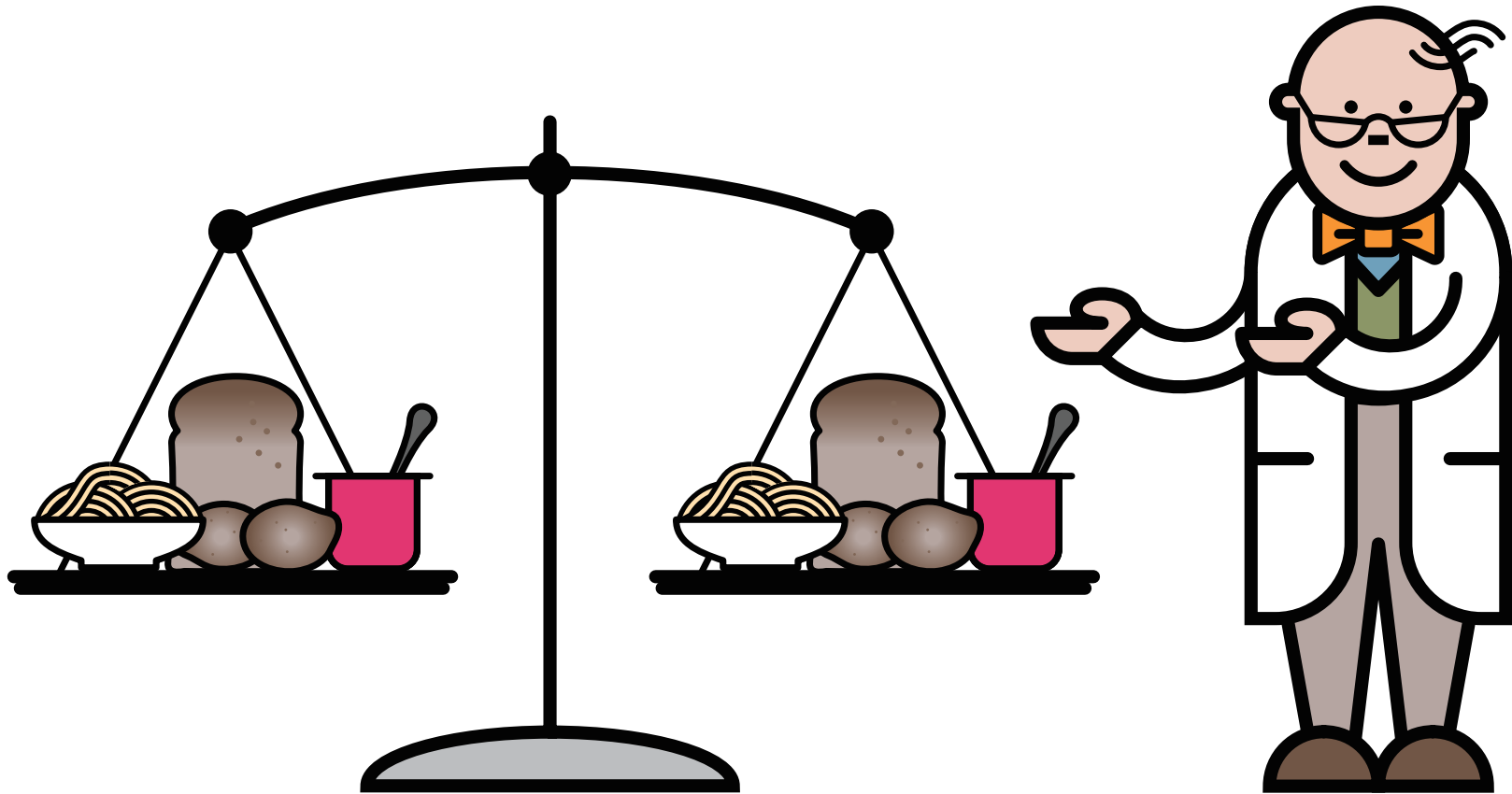
# Metabolic crisis

- In a **metabolic crisis** there is a build up of isovaleric acid and other toxic chemicals such as ammonia
- It is usually triggered by childhood illnesses e.g. vomiting and diarrhoea, fasting for too long or not having enough energy from food
- There should be no delay in management
- Avoidance of a metabolic crisis is essential



# Protein balance is needed in IVA

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





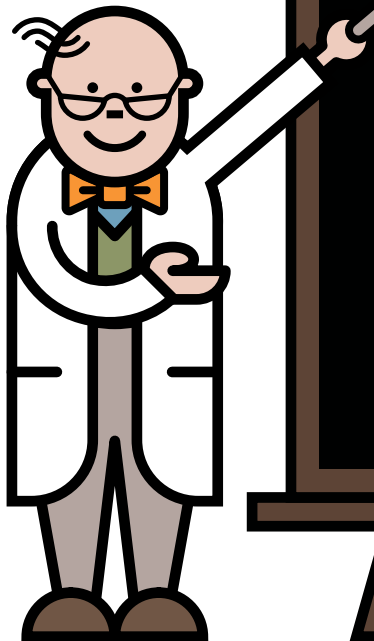
# How is IVA managed day to day?

IVA is managed with the following special diet and medications:

A protein restricted diet

Glycine

Carnitine medication



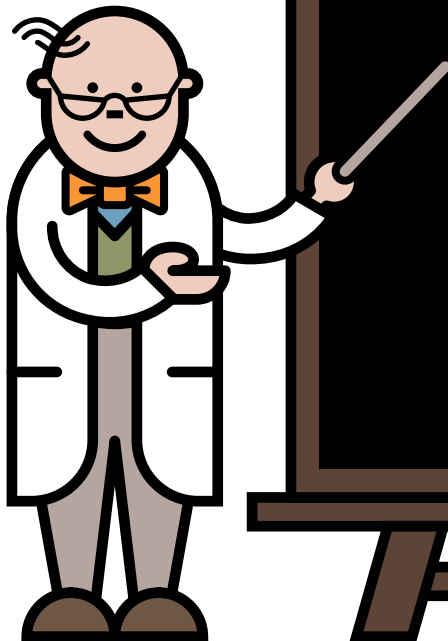
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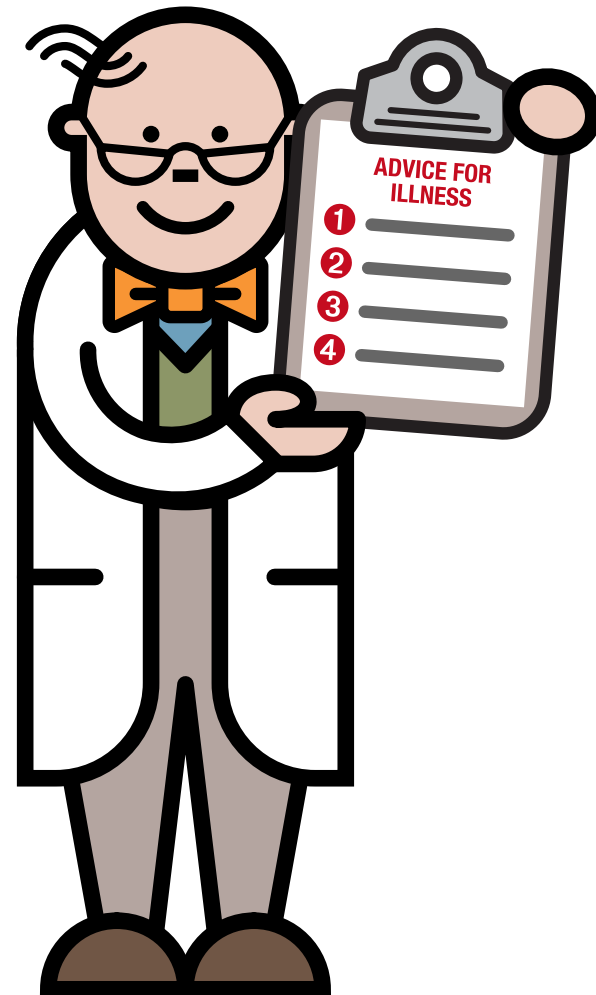
Glycine

Carnitine medication



# How is IVA 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



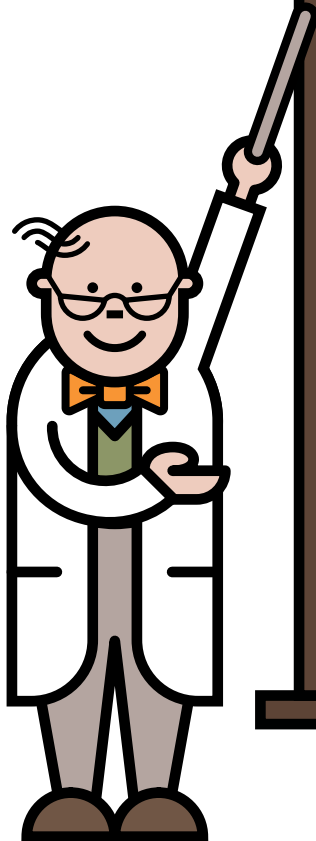
# How is IVA managed during illness?

Stop all protein in food & drink

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

Glycine medication

Carnitine medication



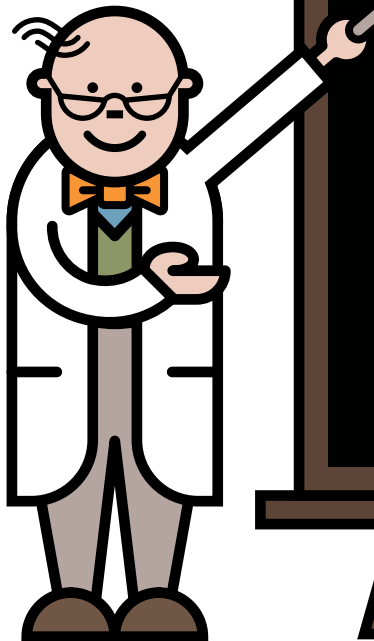
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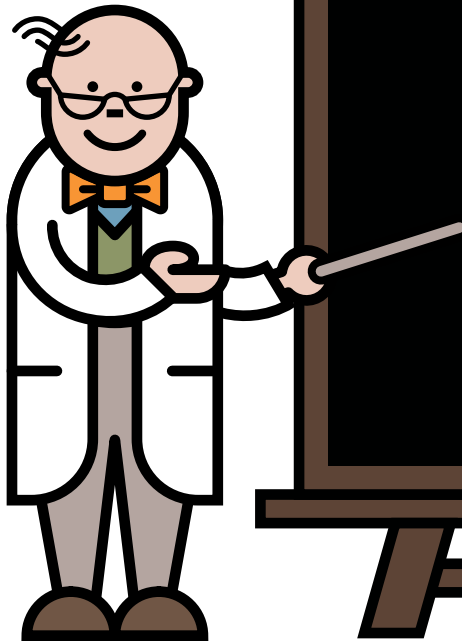
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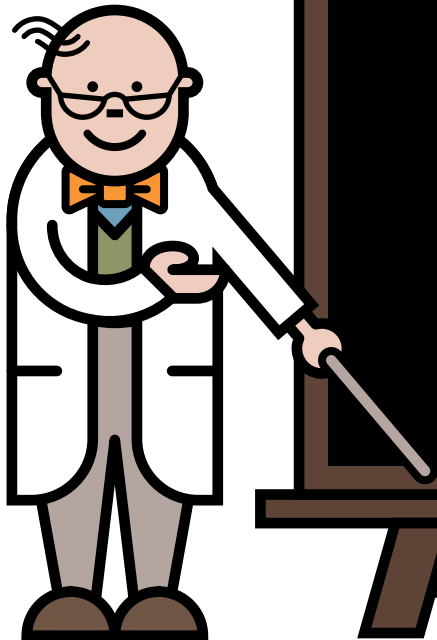
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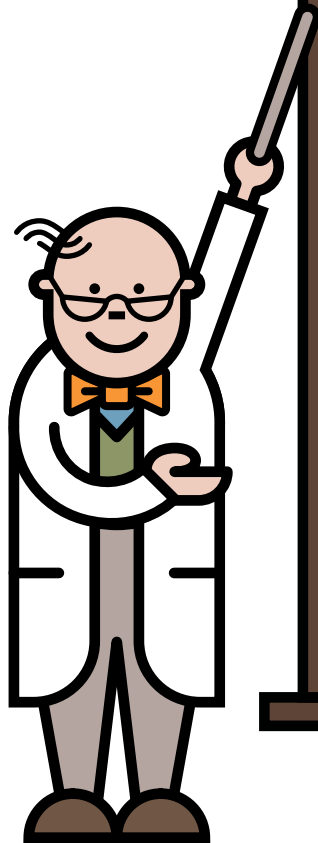
Glycine medication

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# 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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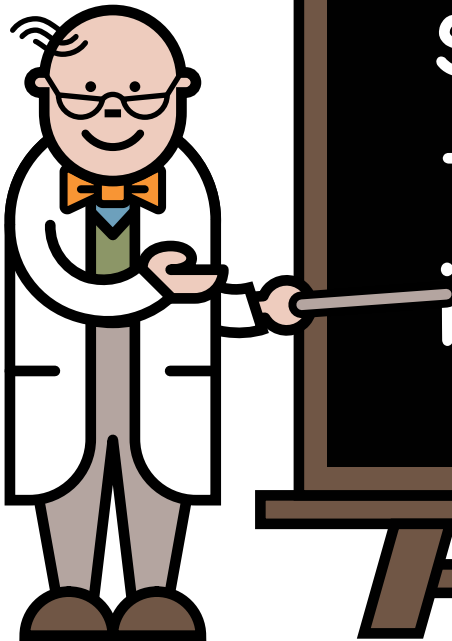


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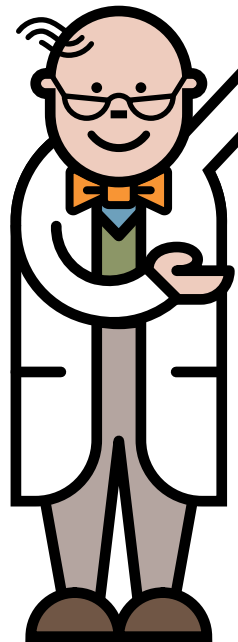


# Key message

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



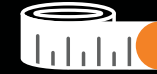
# How is IVA monitored?



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



Height and weight

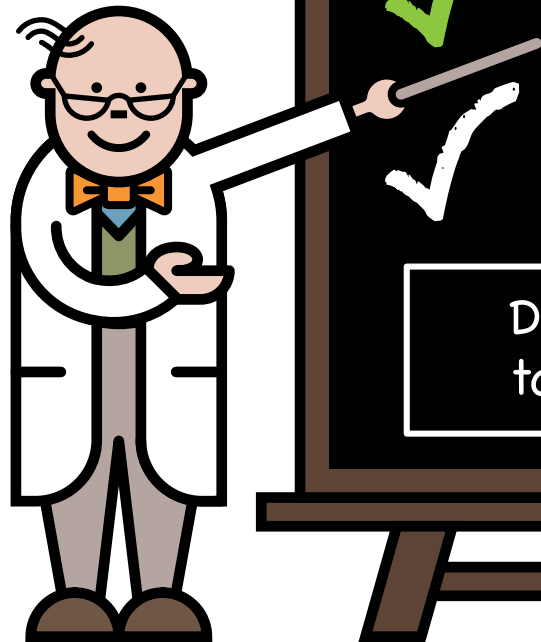


Developmental checks



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

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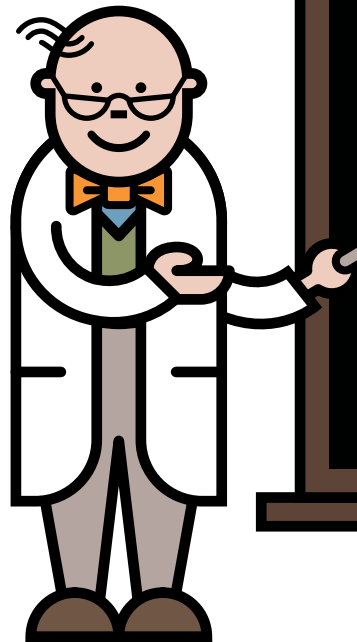


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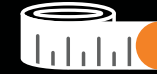
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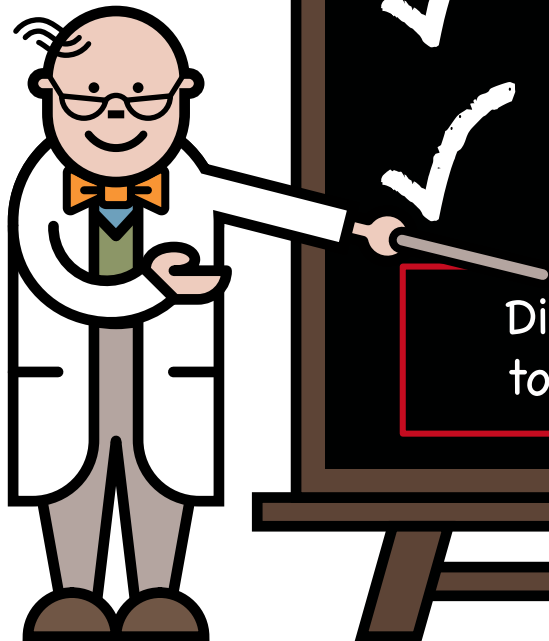
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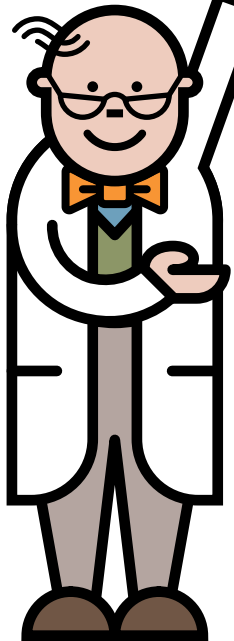


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# Chromosomes, genes, mutations



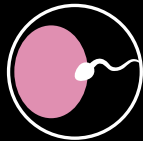
Humans have chromosomes composed of DNA



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

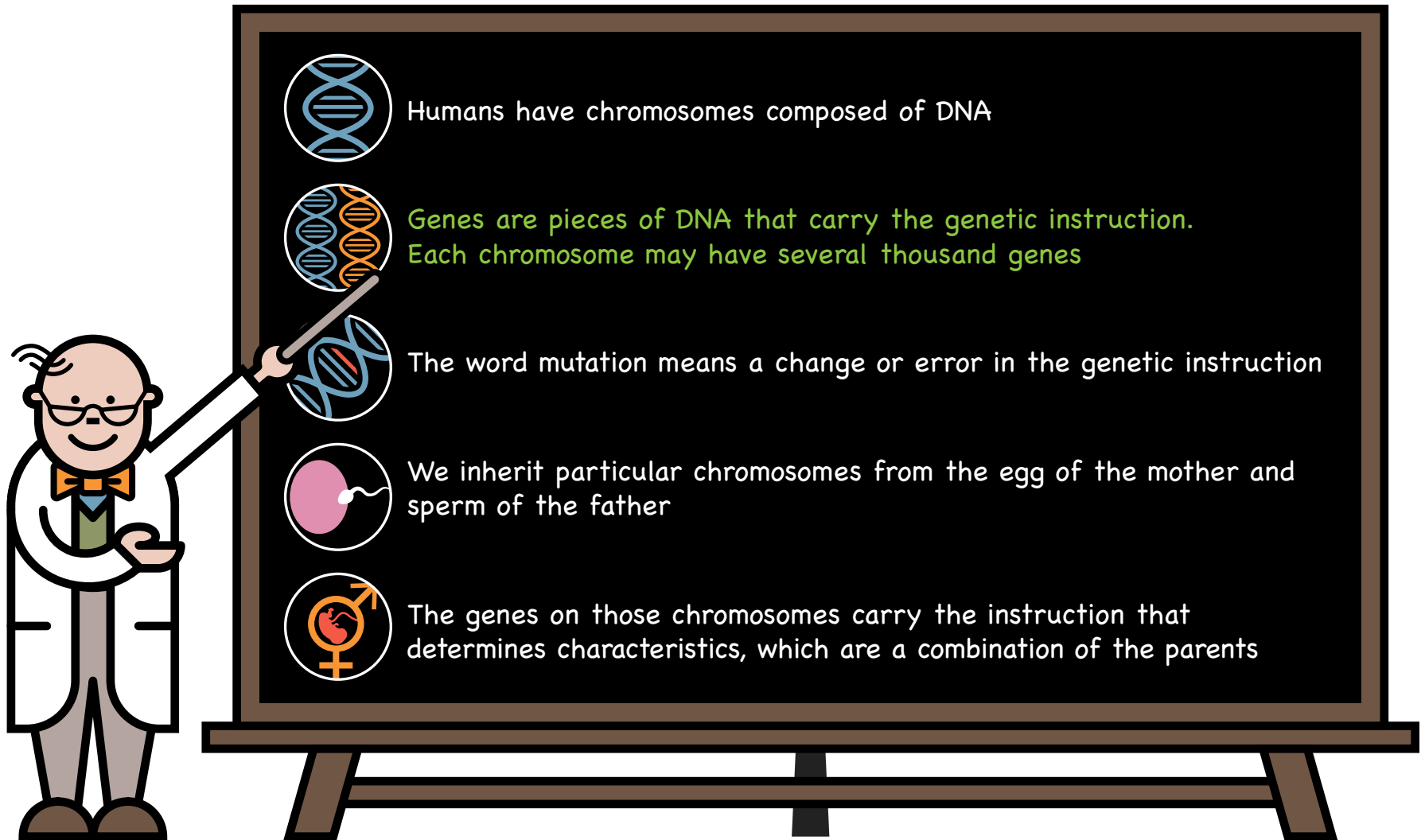



We inherit particular chromosomes from the egg of the mother and sperm of the father





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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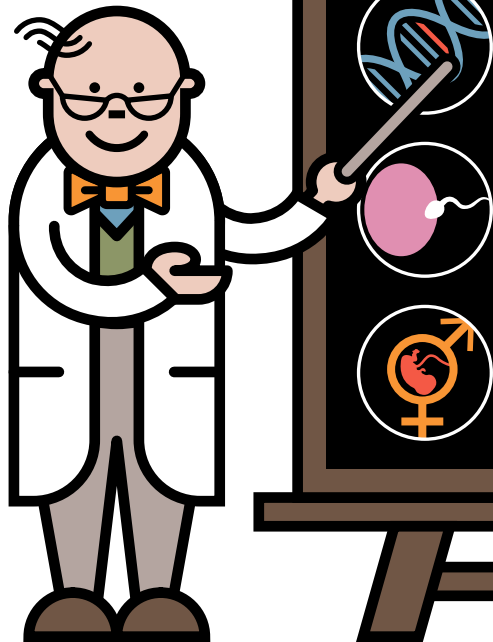
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# Chromosomes, genes, mutations



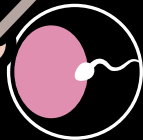
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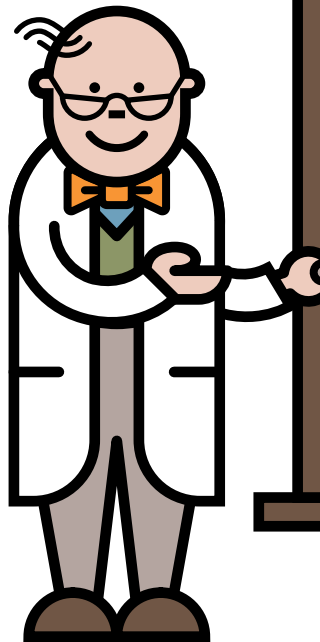


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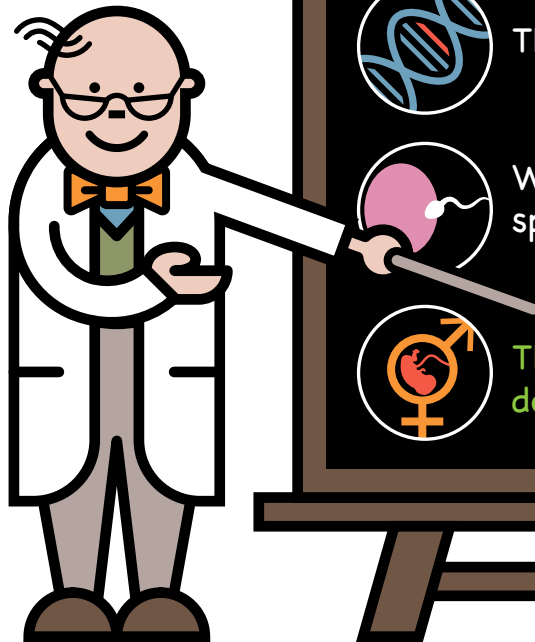


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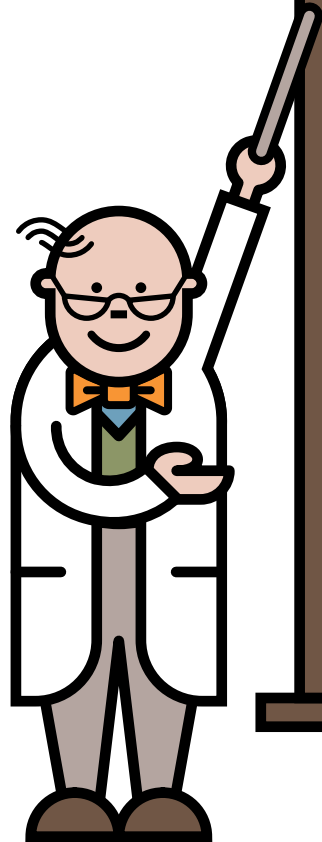


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# Inheritance



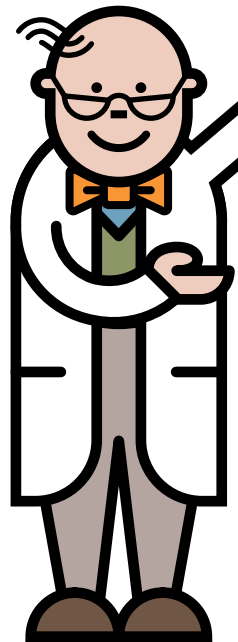
IVA is an inherited condition. There is nothing that could have been done to prevent your baby from having IVA

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

Parents of children with IVA are carriers of the condition

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

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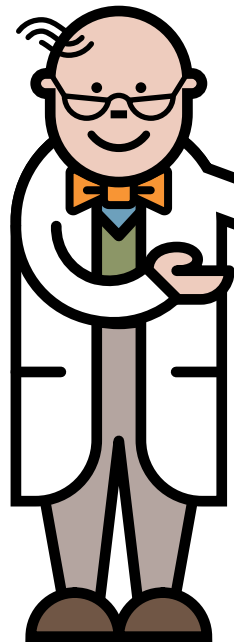


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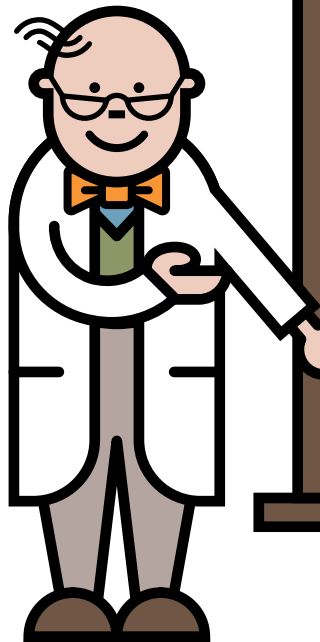


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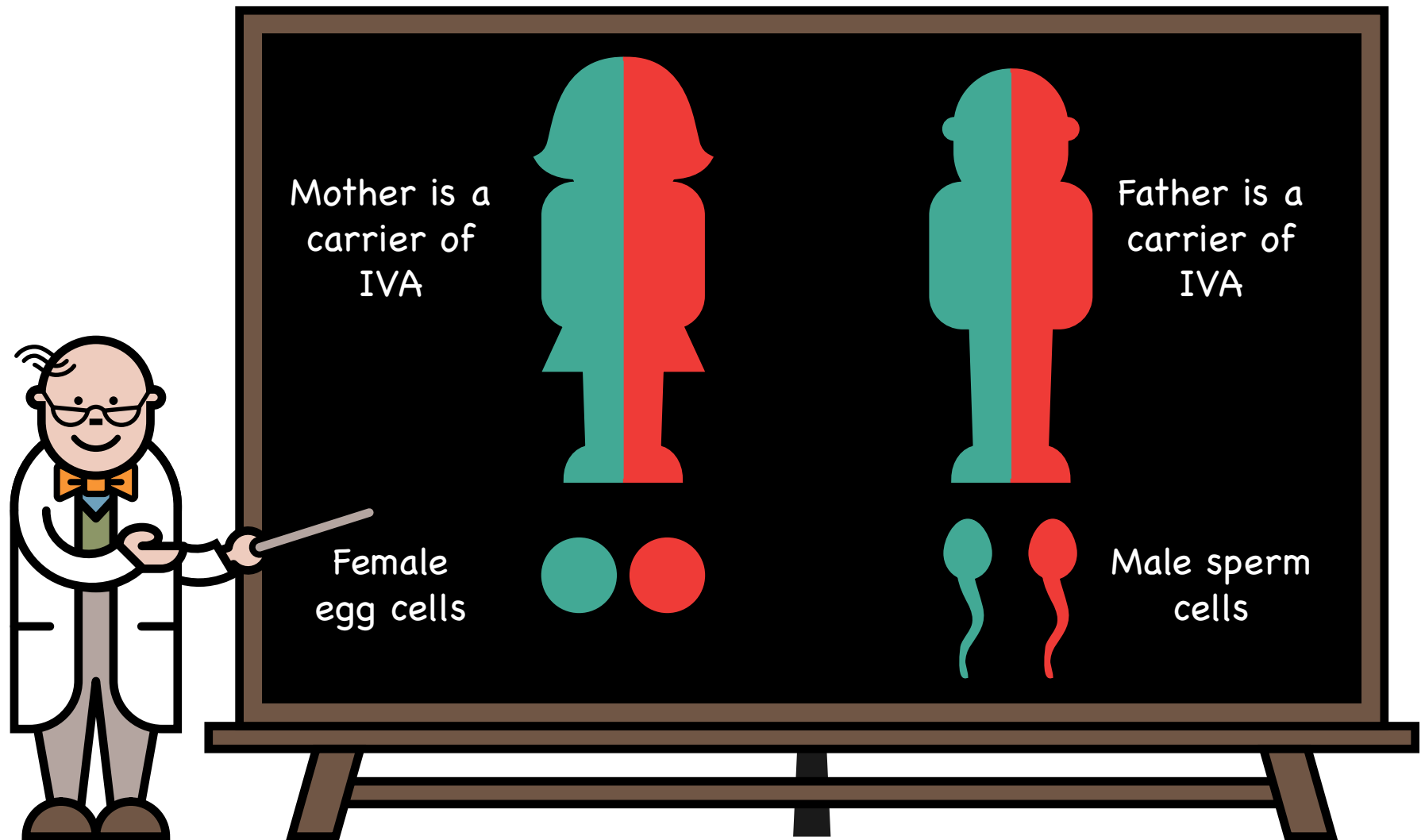
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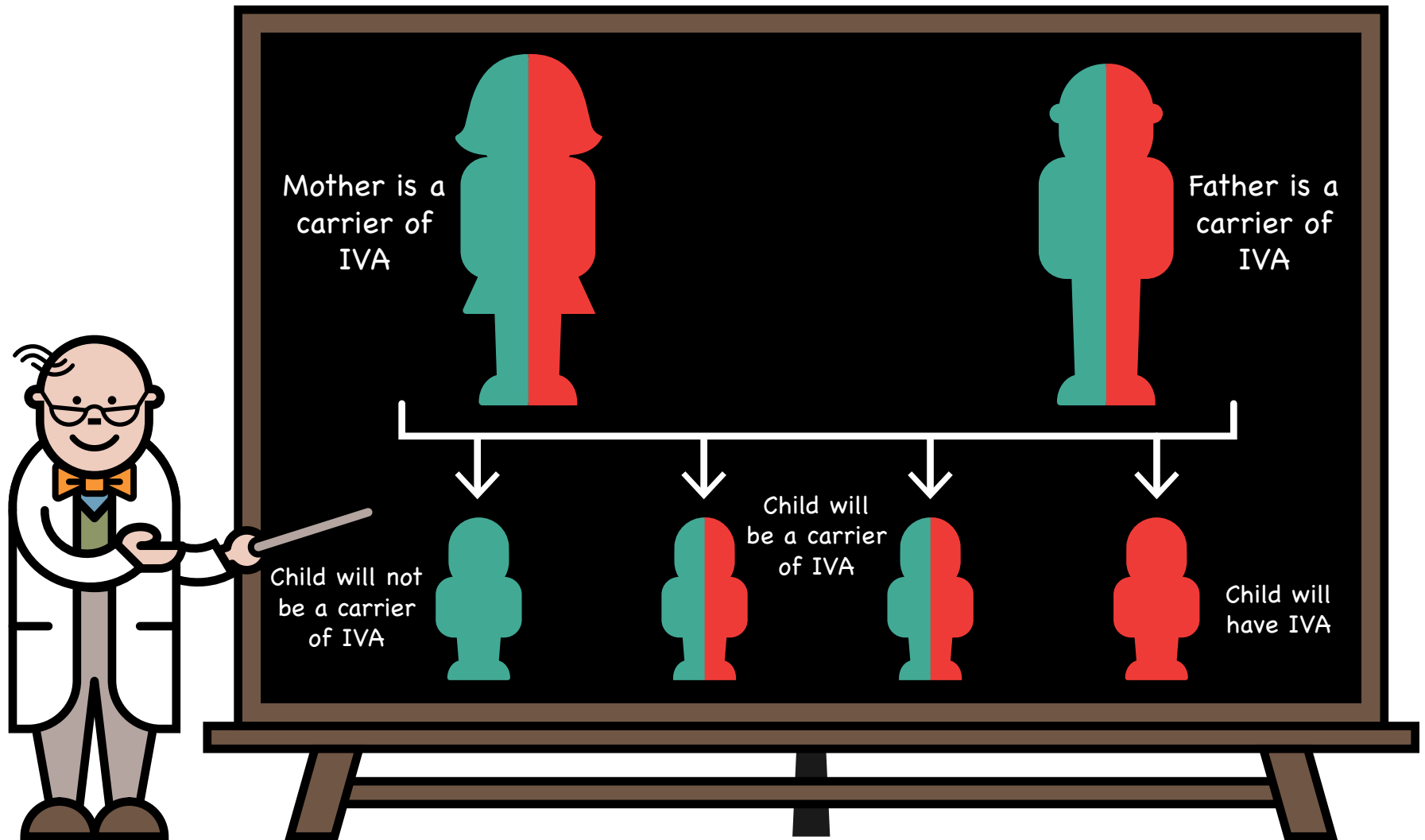
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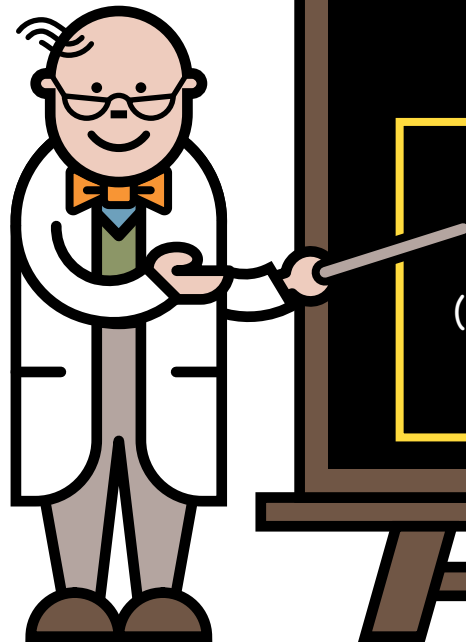
# Inheritance – Autosomal recessive (carriers of IVA)



# Inheritance – Autosomal recessive – possible combinations



# Future pregnancies



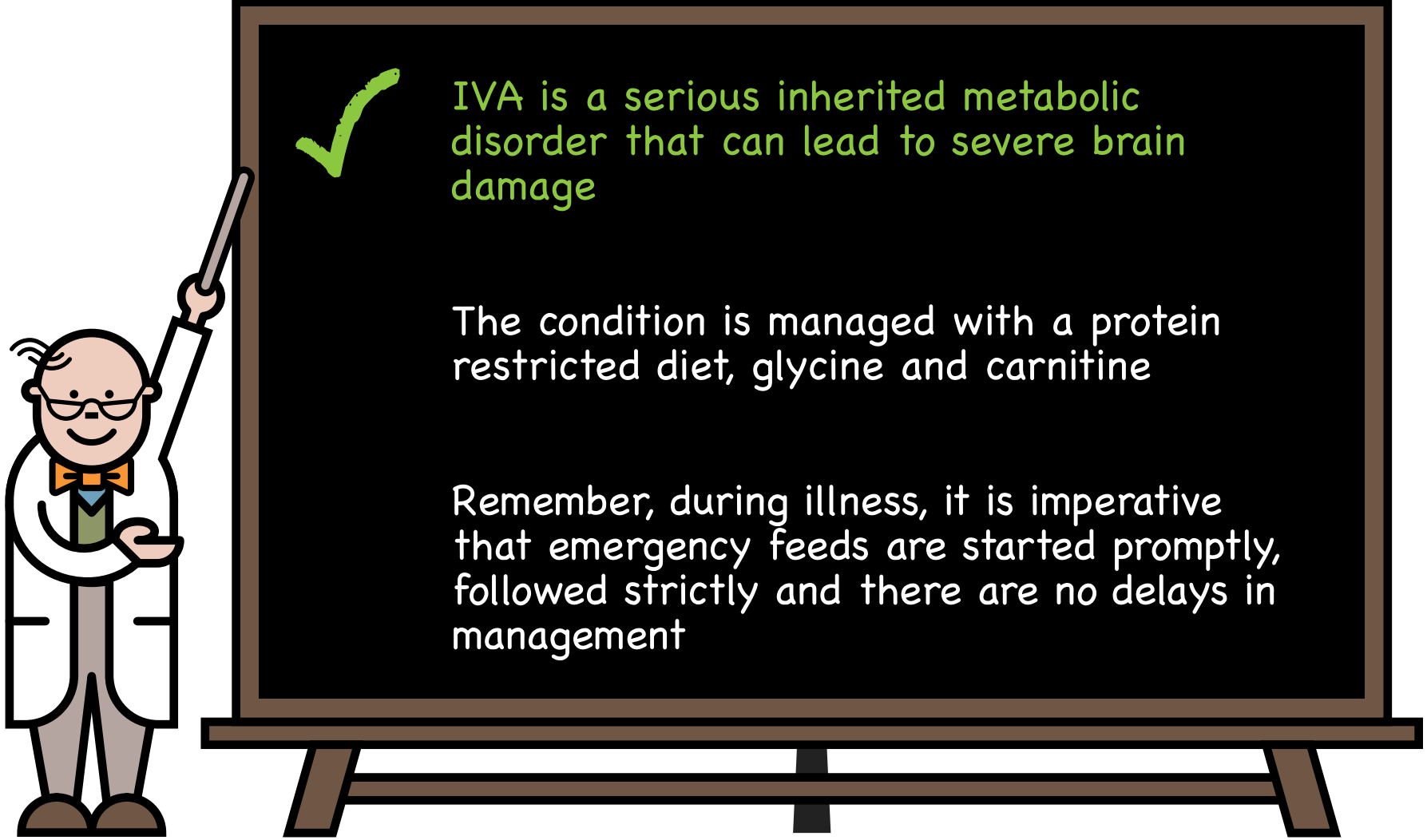
When both parents are carriers, in each pregnancy the risk to the baby is as follows:

25% chance  
(1 in 4) of IVA

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

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

# Take home messages



✓ IVA is a serious inherited metabolic disorder that can lead to severe brain damage

The condition is managed with a protein restricted diet, glycine and carnitine

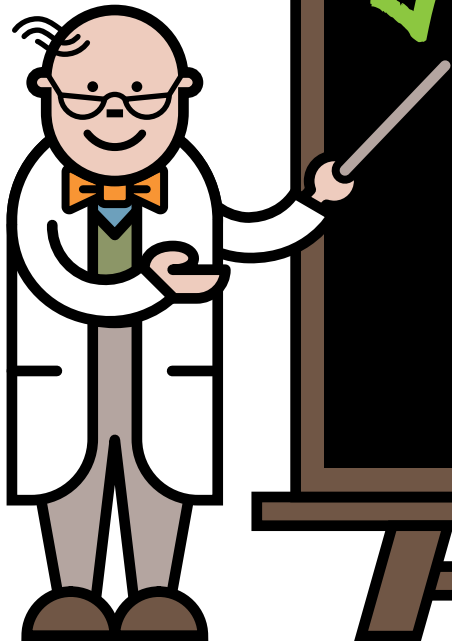
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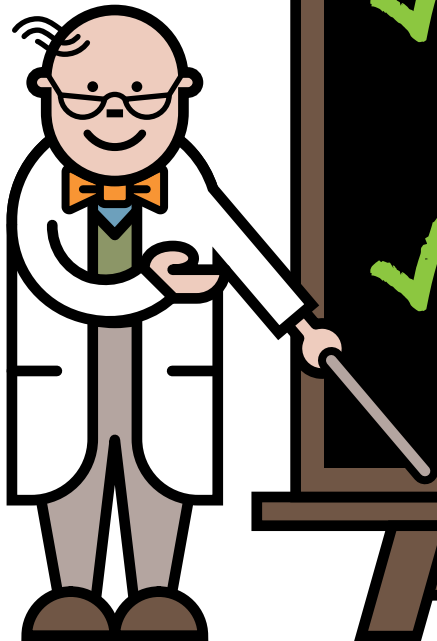


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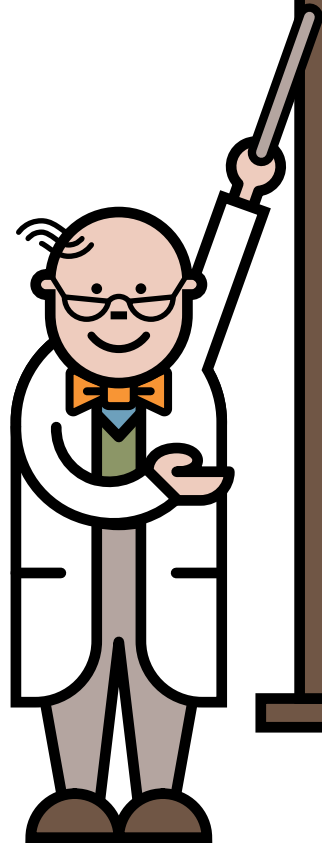
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# Helpful hints



Always ensure you have a good supply of your dietary products 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



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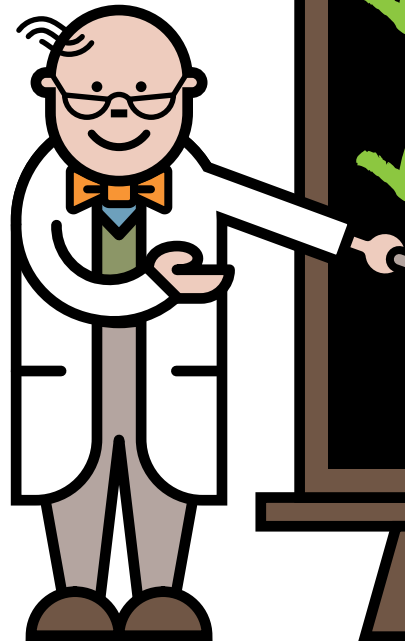
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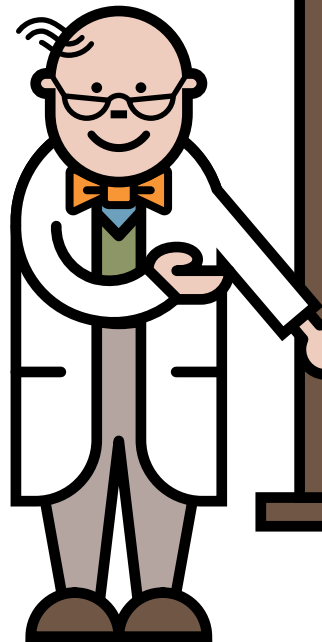
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# Who's who

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

Visit [www.lowproteinconnect.com](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



**NUTRICIA**  
LIFE-TRANSFORMING NUTRITION

**METABOLIC SUPPORT UK**  
Your rare condition.  
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[www.bimdg.org.uk](http://www.bimdg.org.uk)

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

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