

TEMPLE



Tools **E**nabling **M**etabolic **P**arents **L**Earning

ADAPTED BY THE DIETITIANS GROUP

BIMDG

British Inherited Metabolic Diseases Group



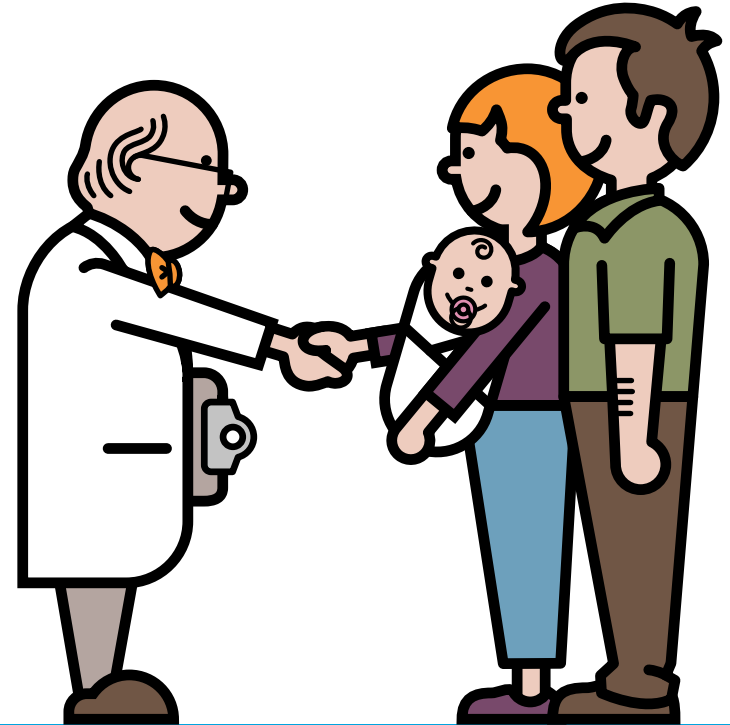
BASED ON THE ORIGINAL TEMPLE WRITTEN BY
BURGARD AND WENDEL
VERSION 2, FEBRUARY 2017

IVA

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

IVA

Information for families following
a positive newborn screening



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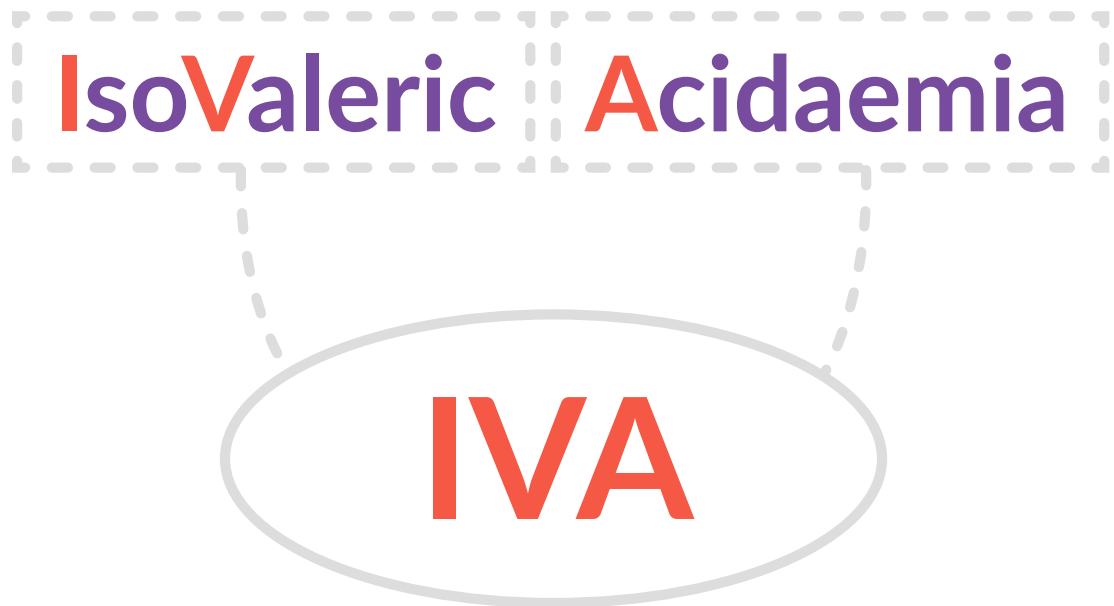
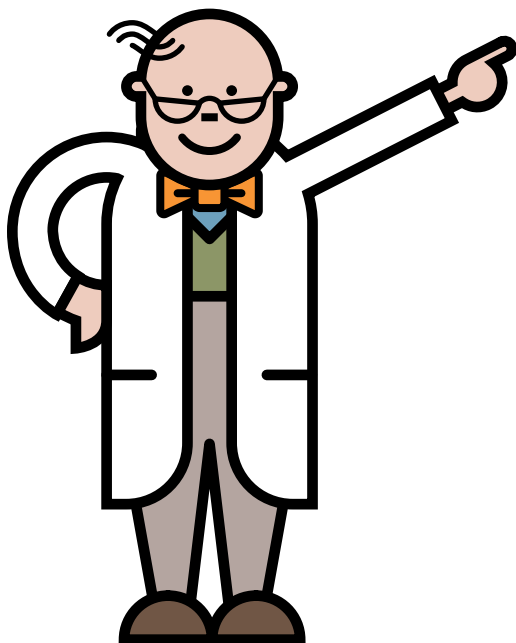
Tools Enabling Metabolic Parents LEarning

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

What is IVA?

IVA stands for Isovaleric Acidaemia

It is an inherited metabolic condition

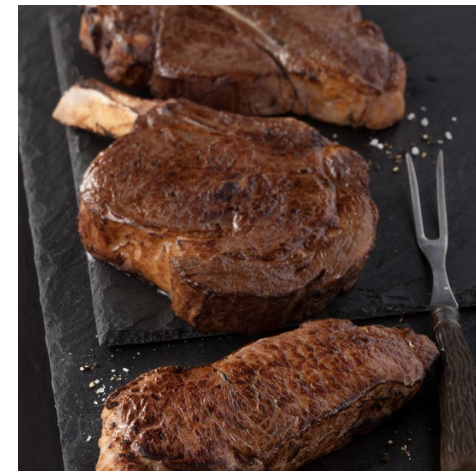
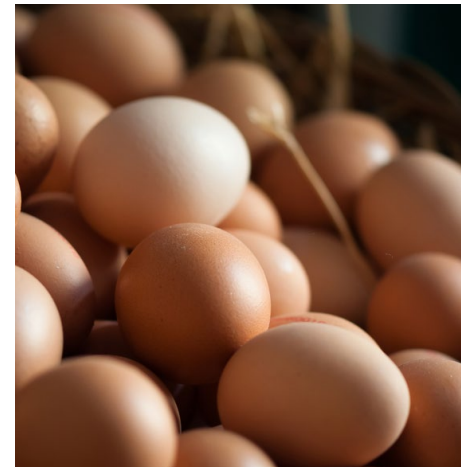


IVA and protein

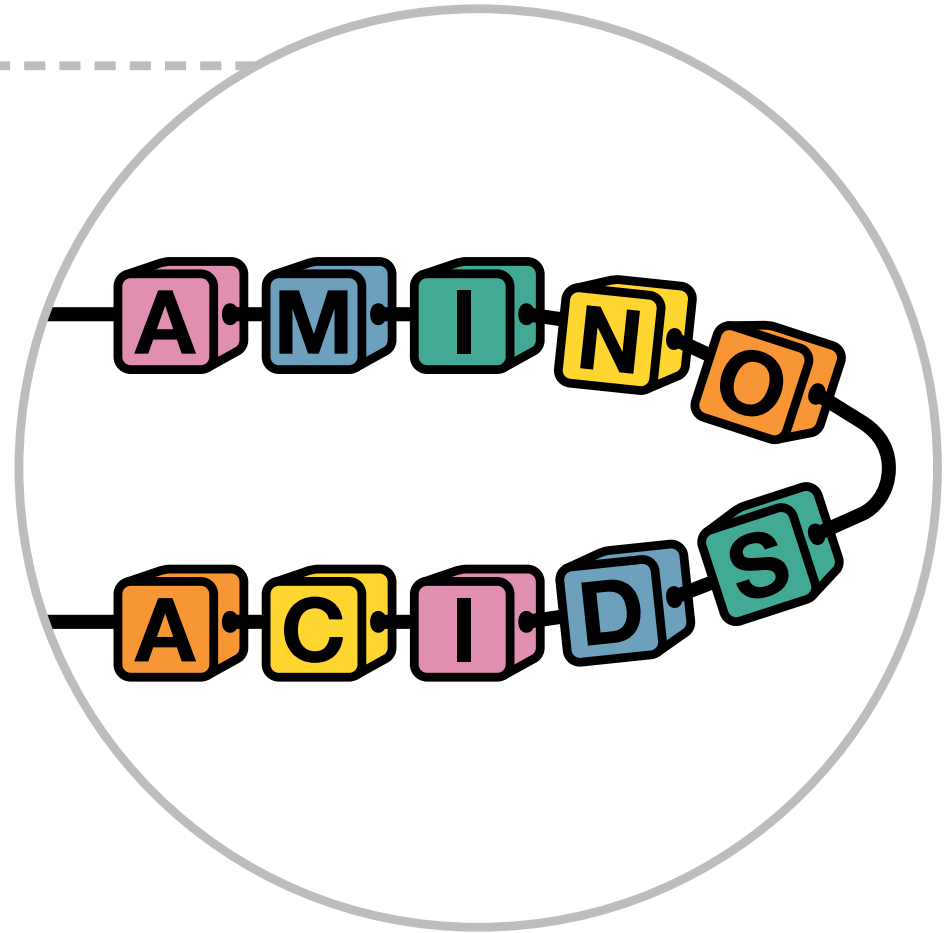
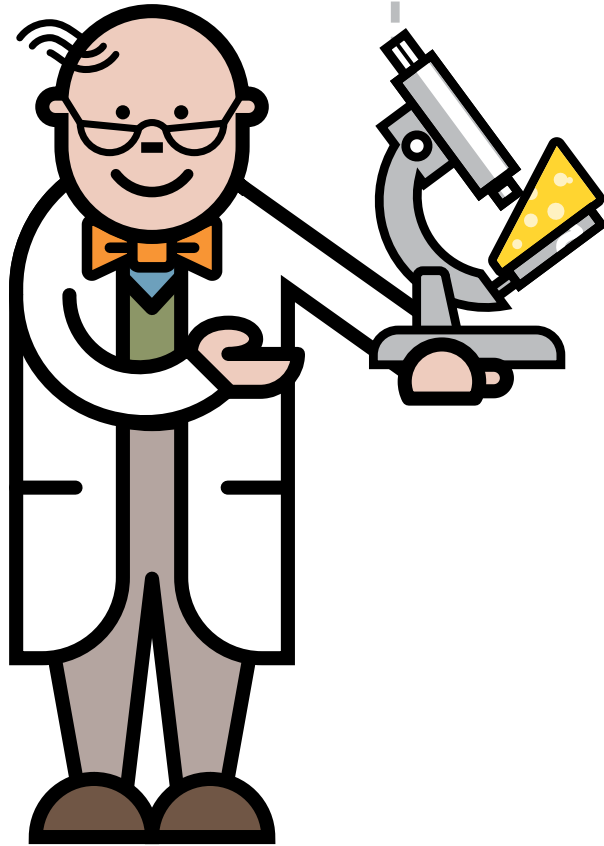
IVA affects the way your baby breaks down protein

Many foods contain protein

The body needs protein for growth and repair



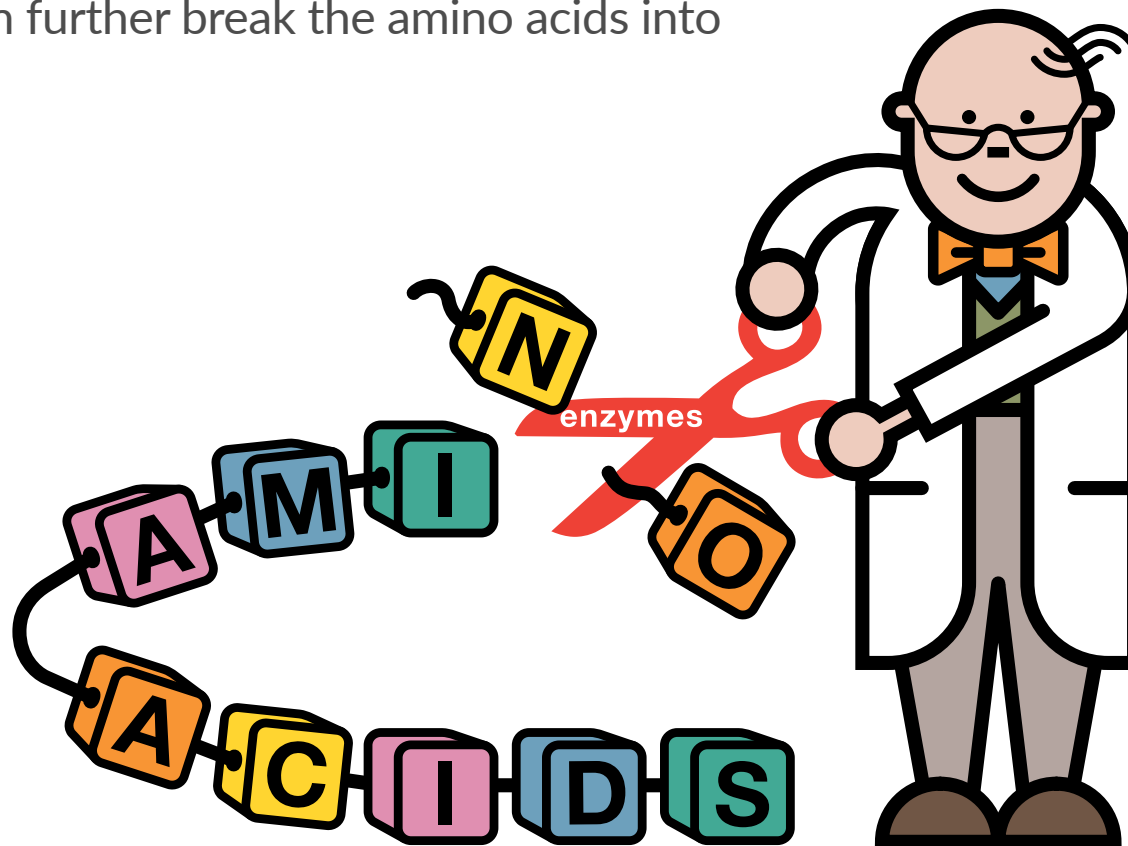
What is protein?



Protein and enzymes

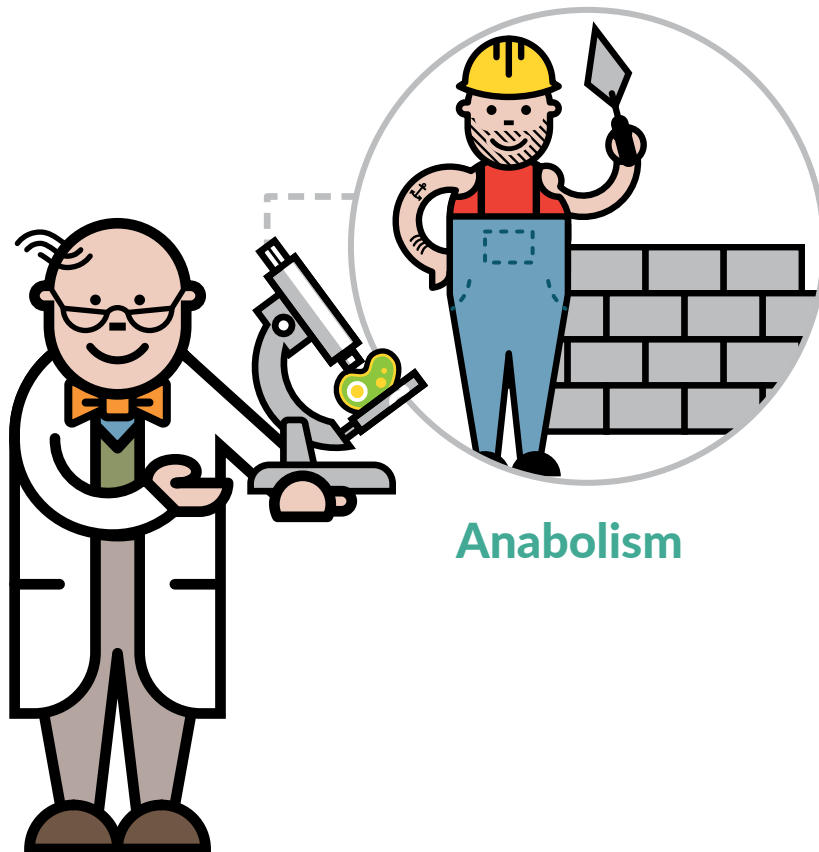
Protein is broken down into amino acids (building blocks of protein) by enzymes (which are like chemical scissors).

Enzymes then further break the amino acids into smaller parts.

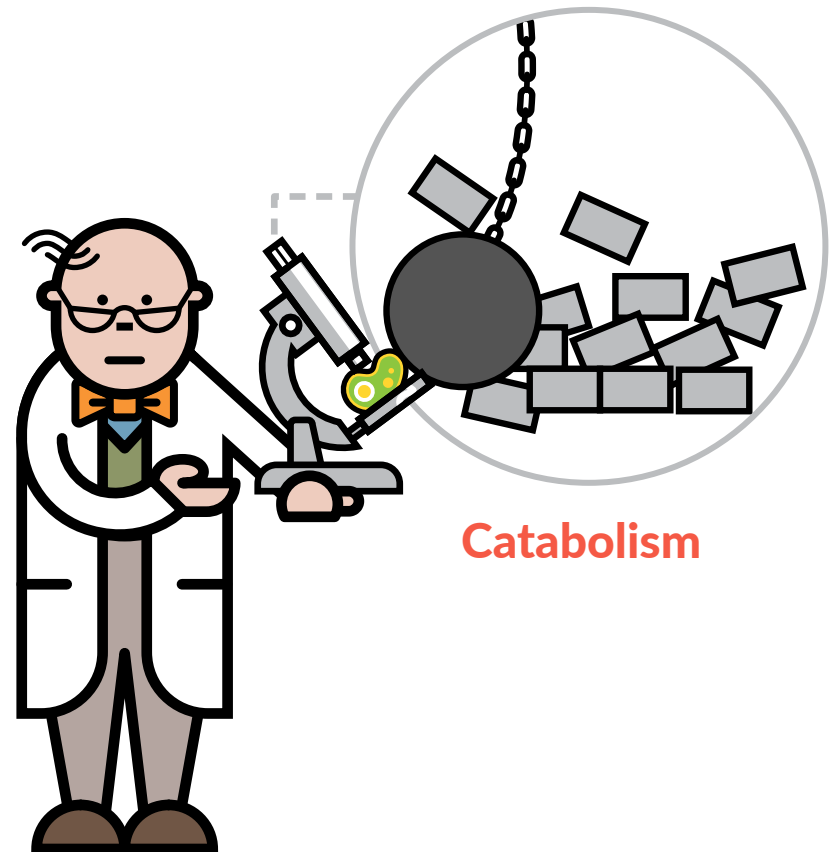


Protein metabolism

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



Anabolism



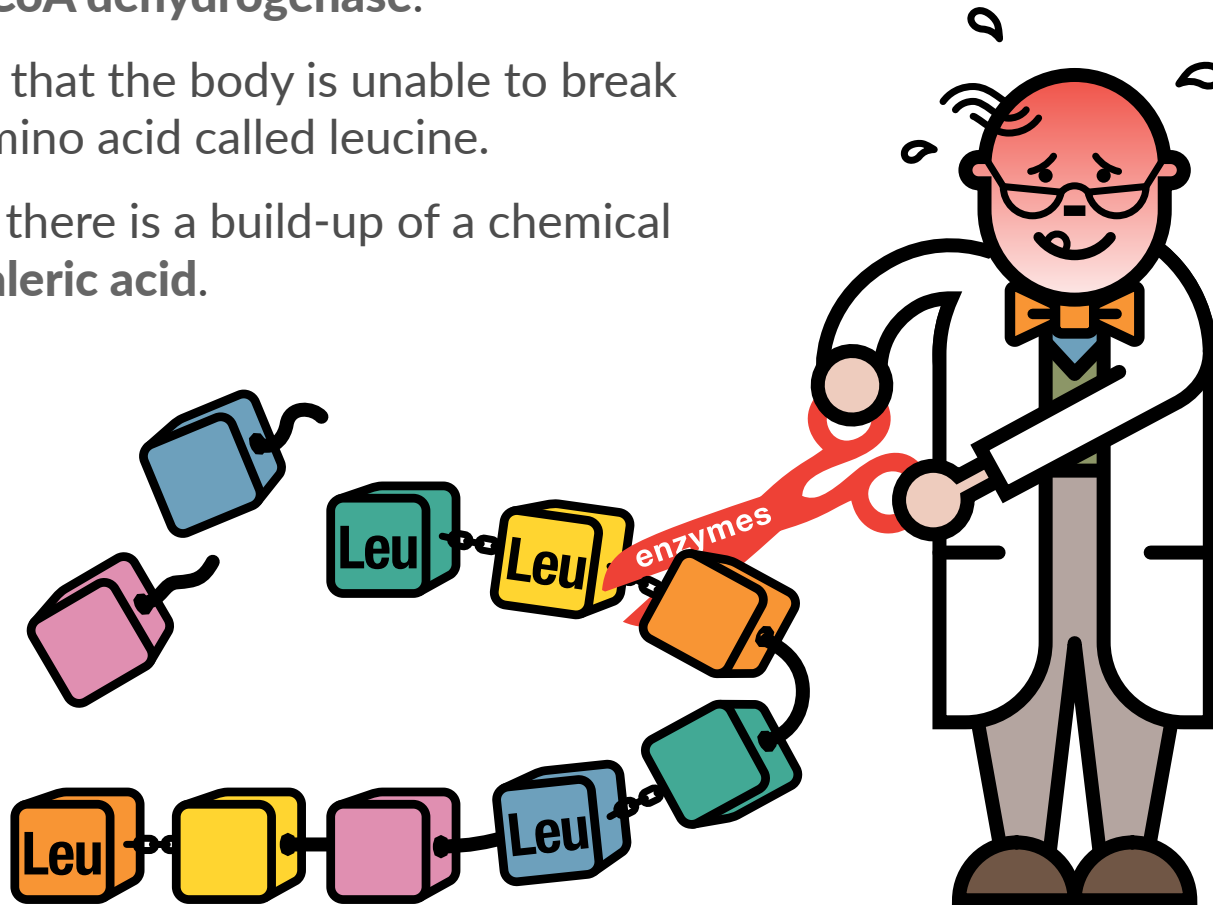
Catabolism

What happens in IVA?

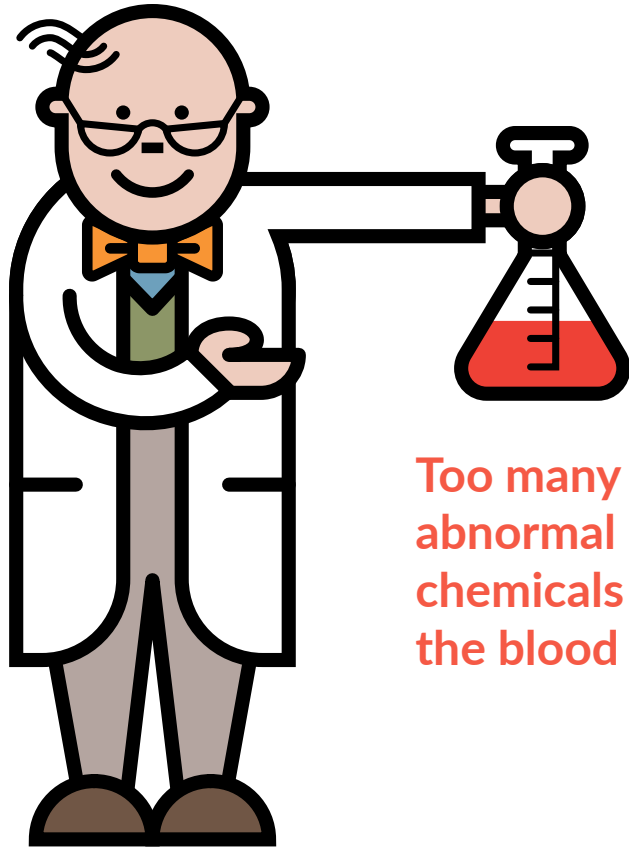
In IVA, the body lacks an enzyme called **isovaleryl-CoA dehydrogenase**.

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

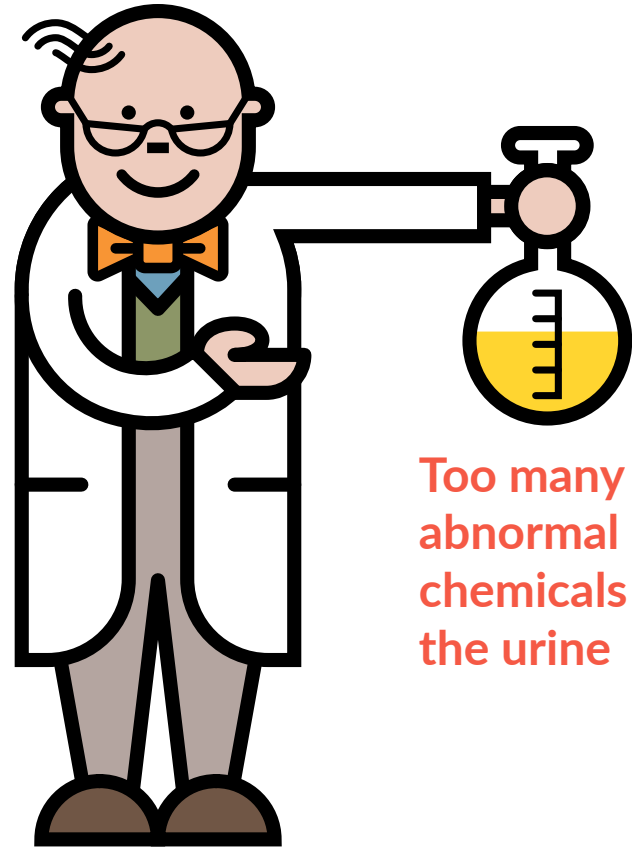
As a result, there is a build-up of a chemical called **isovaleric acid**.



What does this cause?



Too many
abnormal
chemicals in
the blood



Too many
abnormal
chemicals in
the urine

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?

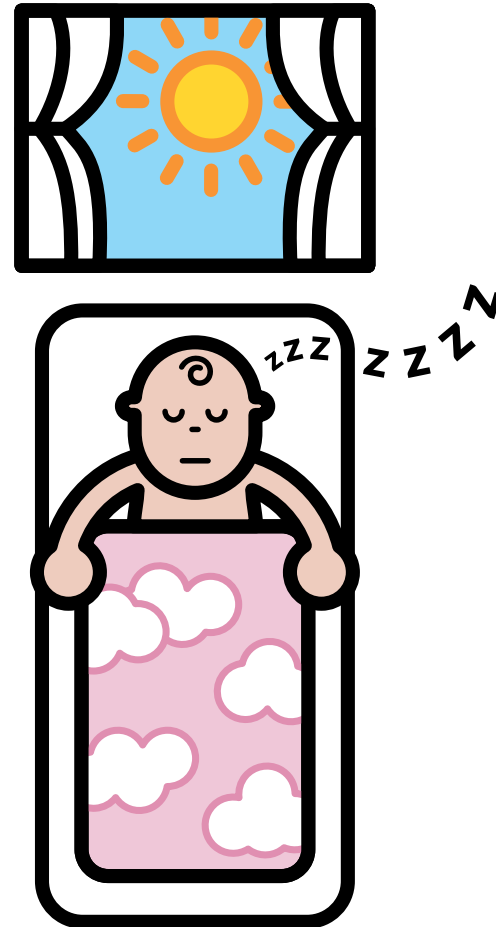
Some babies with IVA become ill in the first few days of life.

Symptoms include:

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

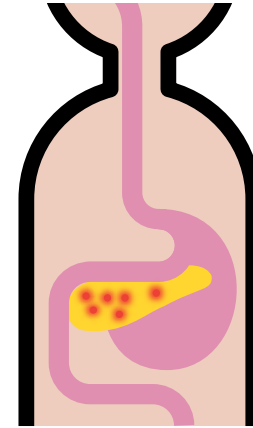
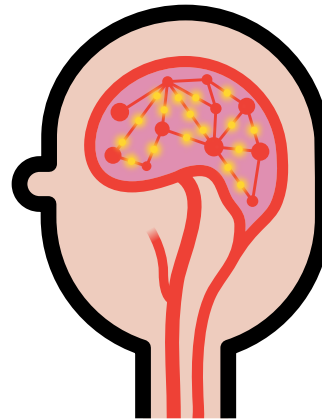
The effects of IVA quickly become life-threatening if untreated

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

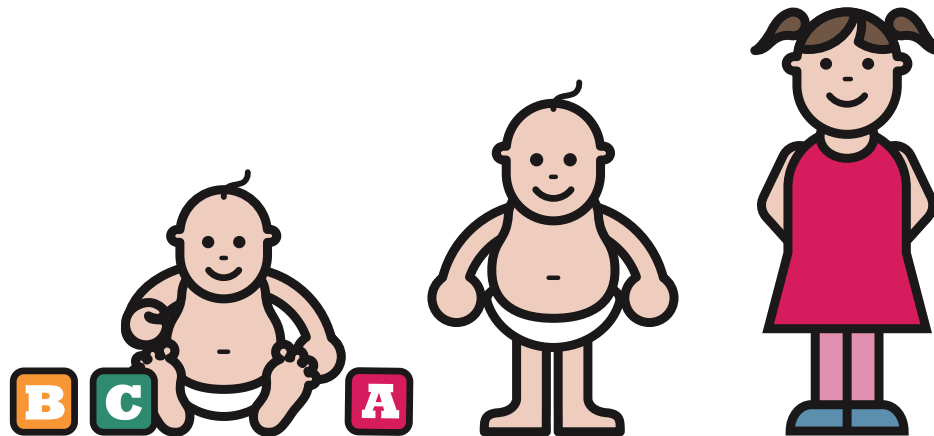


What can go wrong in IVA?

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



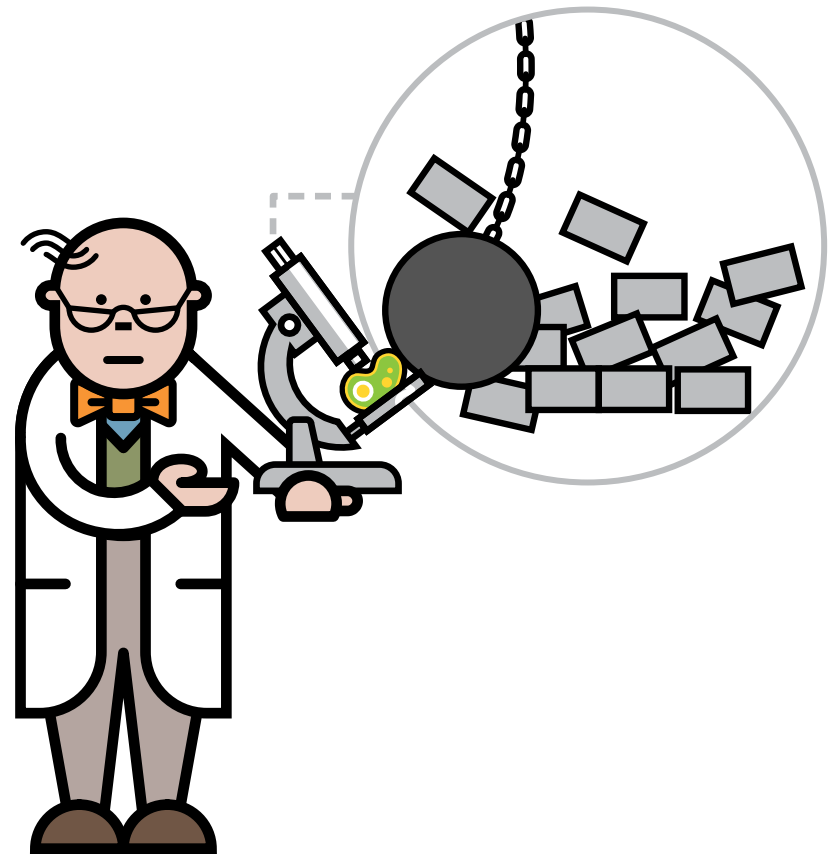
It may cause delays to normal development like walking and talking.



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.



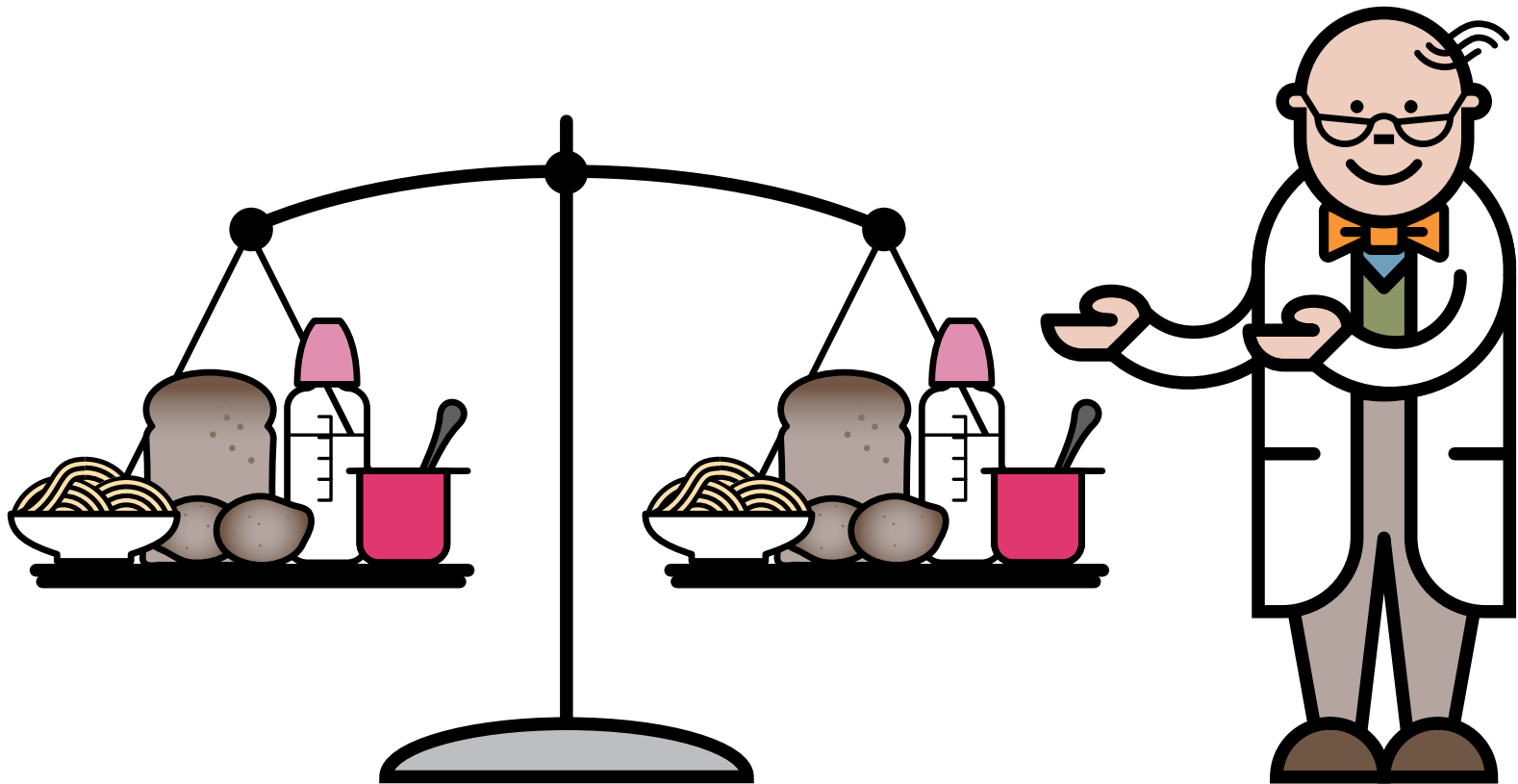
Metabolic crisis

- In a **metabolic crisis** there is a build up of methylmalonic 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 treatment
- 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.



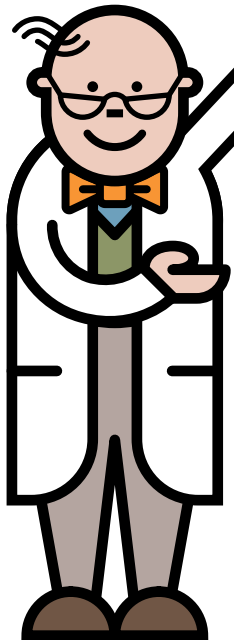
How is IVA managed day to day?

IVA is managed with the following special diet and medications:

A protein restricted diet

Glycine

Carnitine medication



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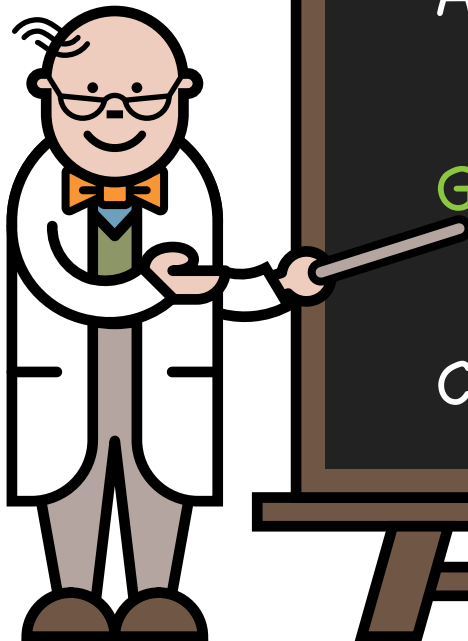
A protein restricted diet



Glycine



Carnitine medication



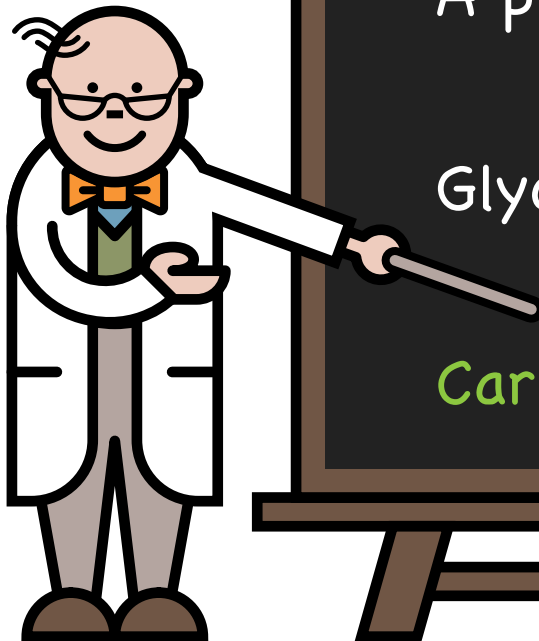
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How is IVA managed during illness?

- 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



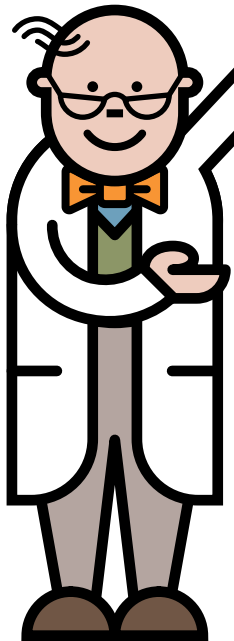
Start the emergency regimen. This is made up of glucose polymer



Glycine medication



Carnitine medication



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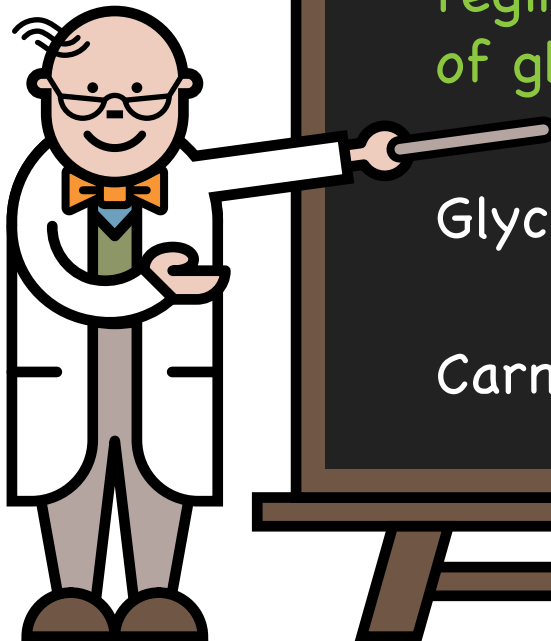
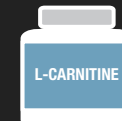
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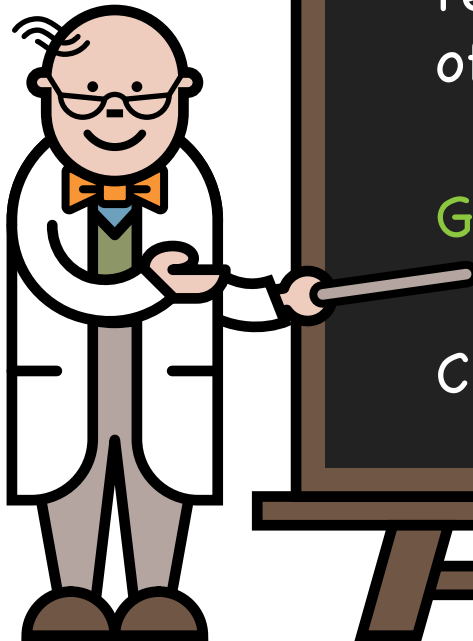
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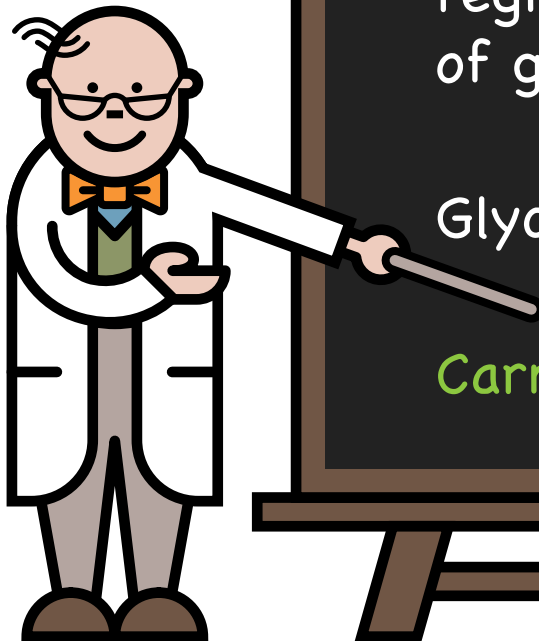
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Glycine medication



Carnitine medication



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



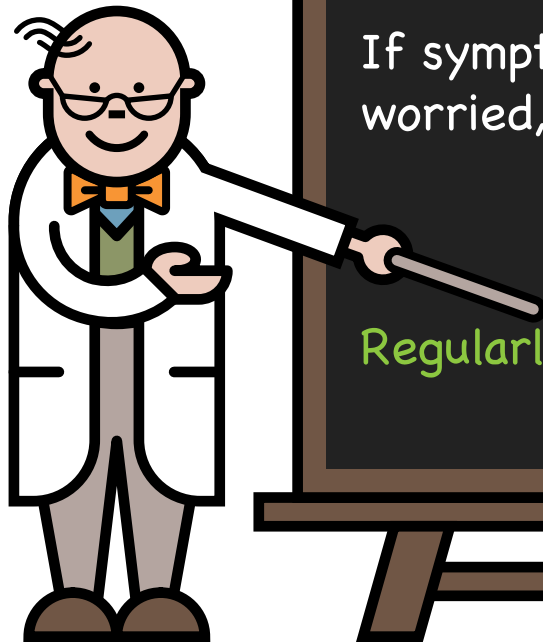
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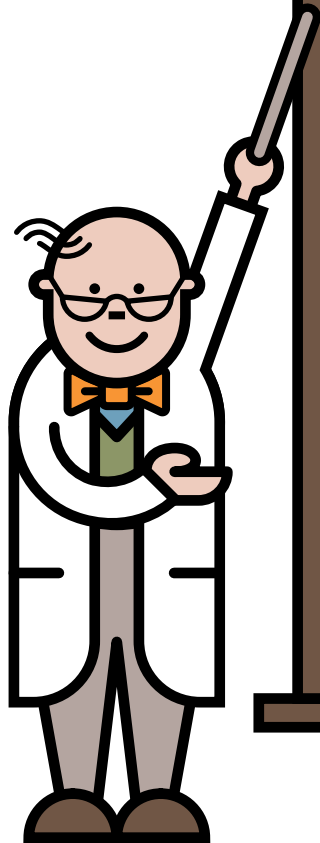


Key message

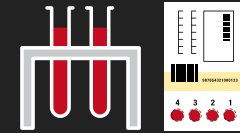


It is imperative that emergency feeds are started **promptly** and there are **no delays** in management.

How is IVA monitored?



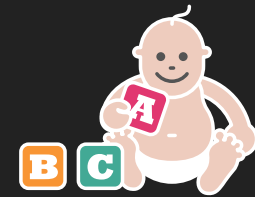
Frequent blood tests to check amino acids, nutrients and chemicals



Height and weight



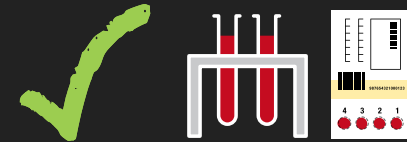
Developmental checks



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

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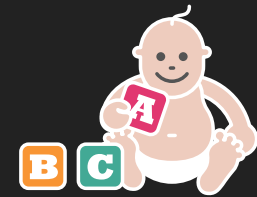
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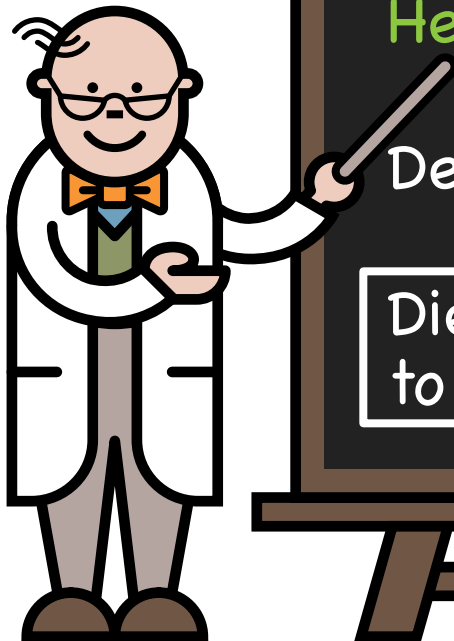
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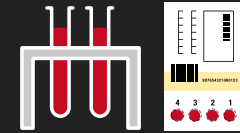


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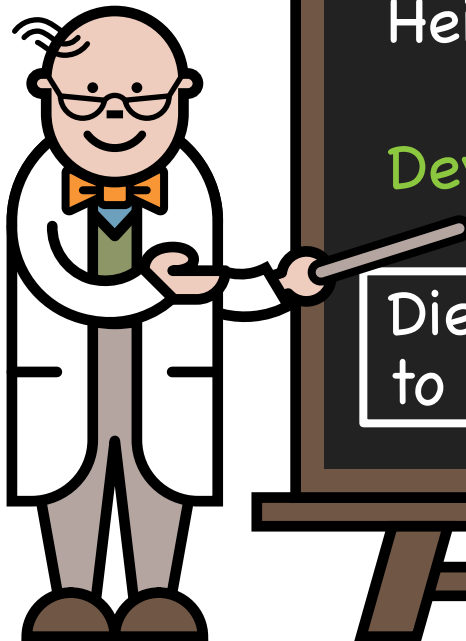
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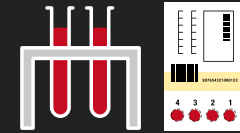


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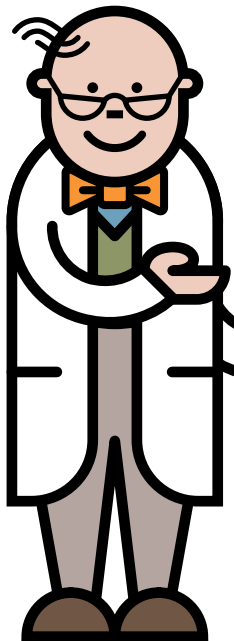
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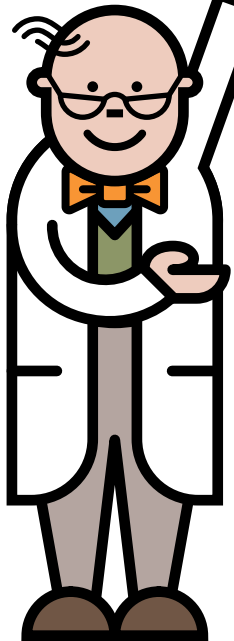
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Chromosomes, genes, mutations



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

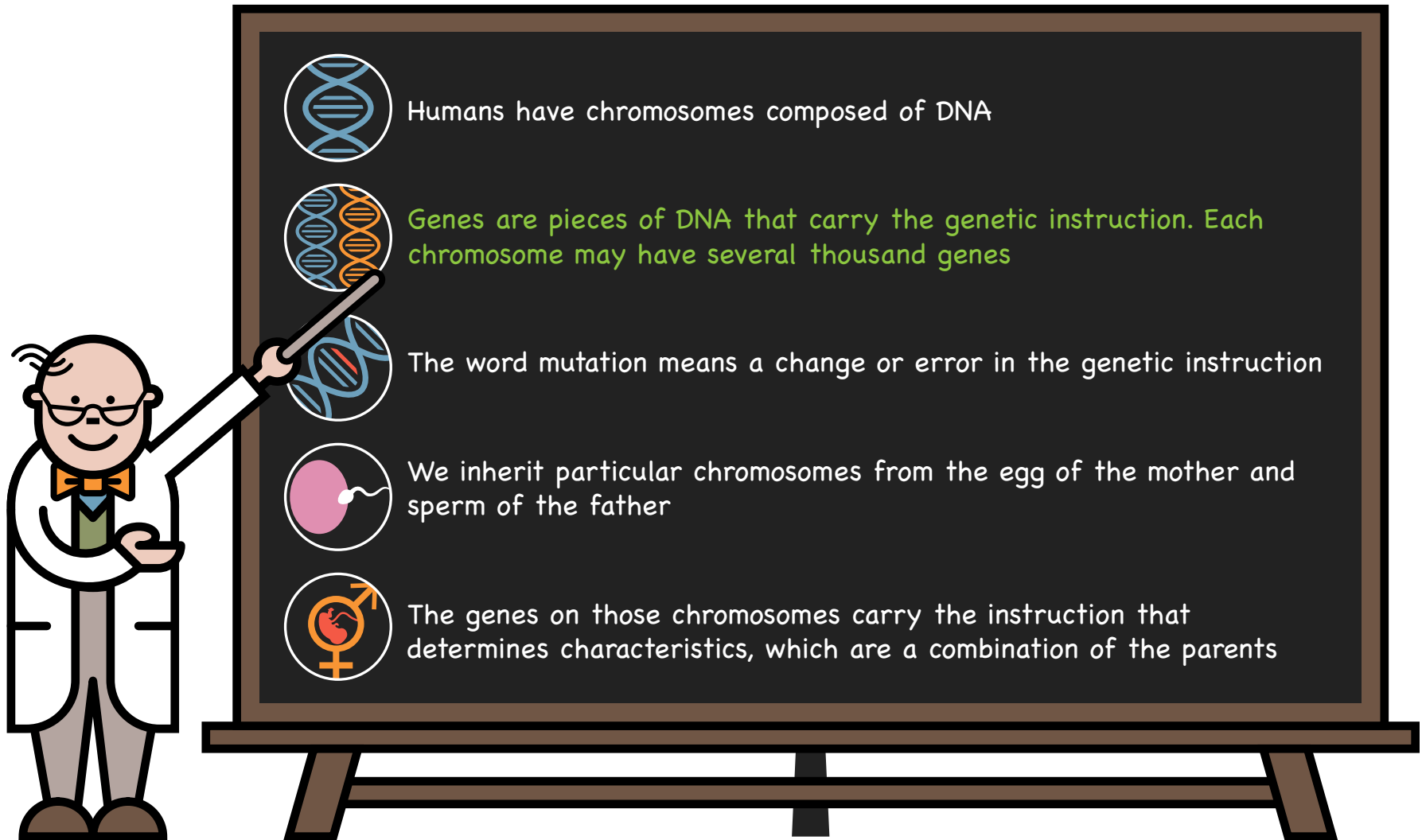



We inherit particular chromosomes from the egg of the mother and sperm of the father





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

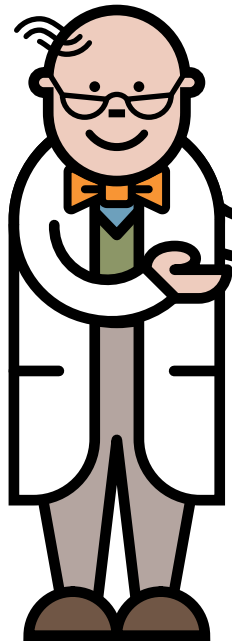
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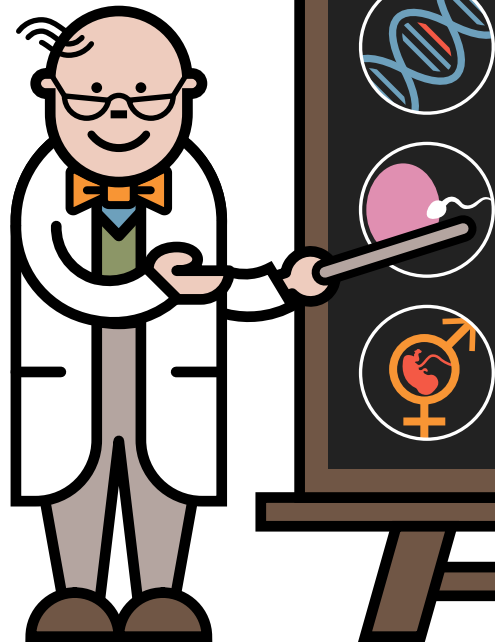


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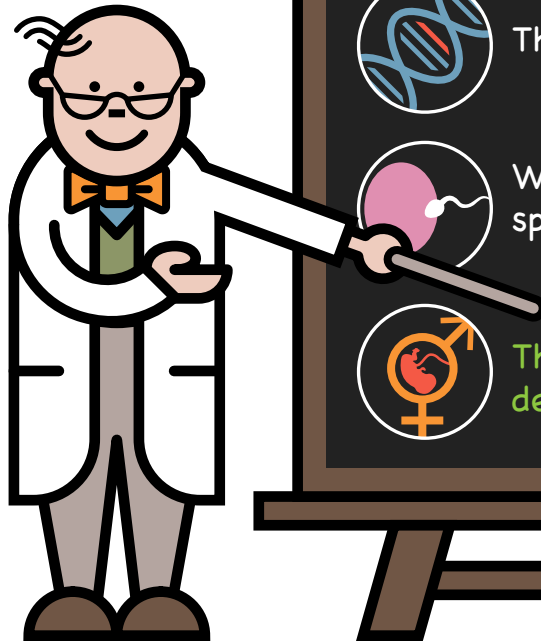


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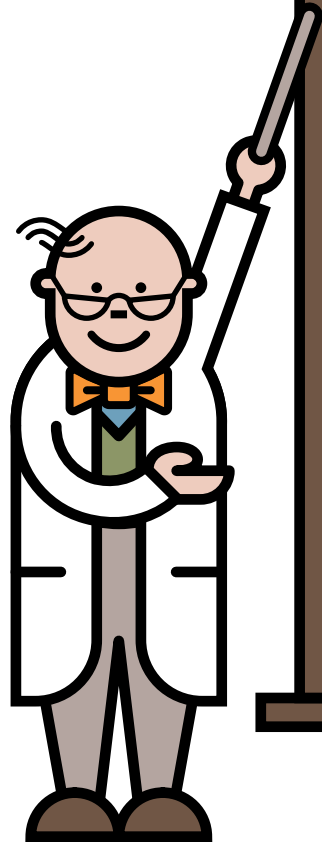


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Inheritance



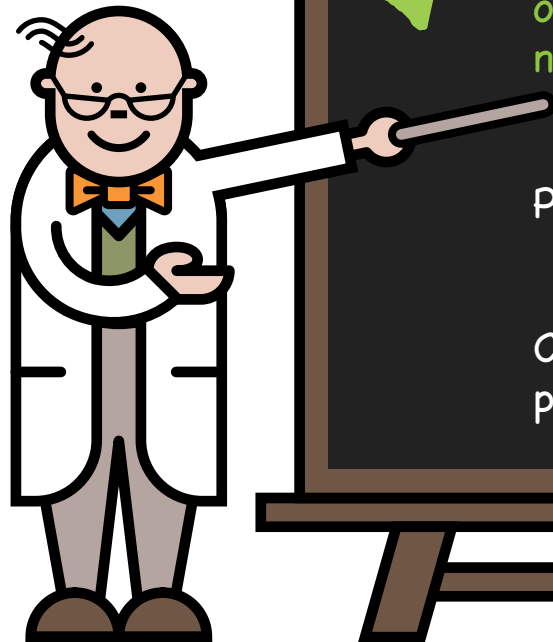
IVA is an inherited condition. There is nothing that could have been done to prevent your baby from having IVA

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

Inheritance



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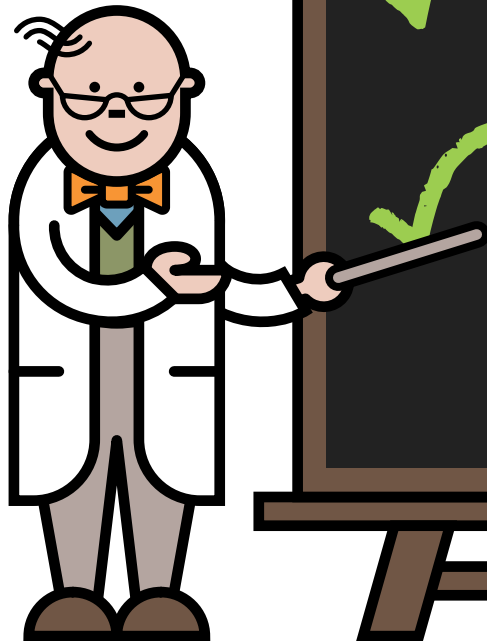


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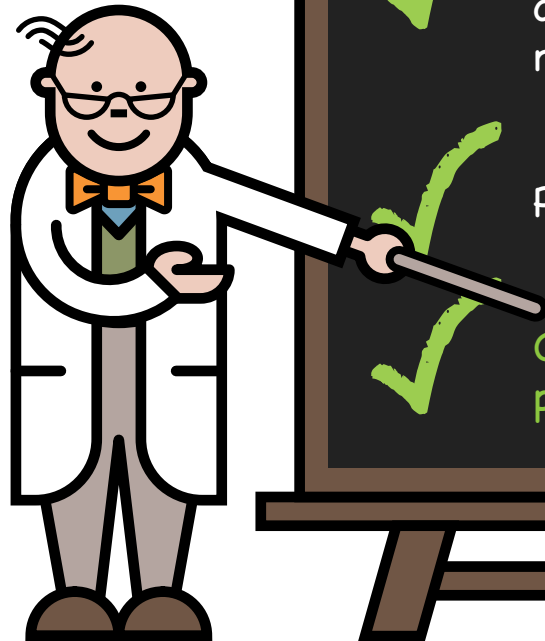
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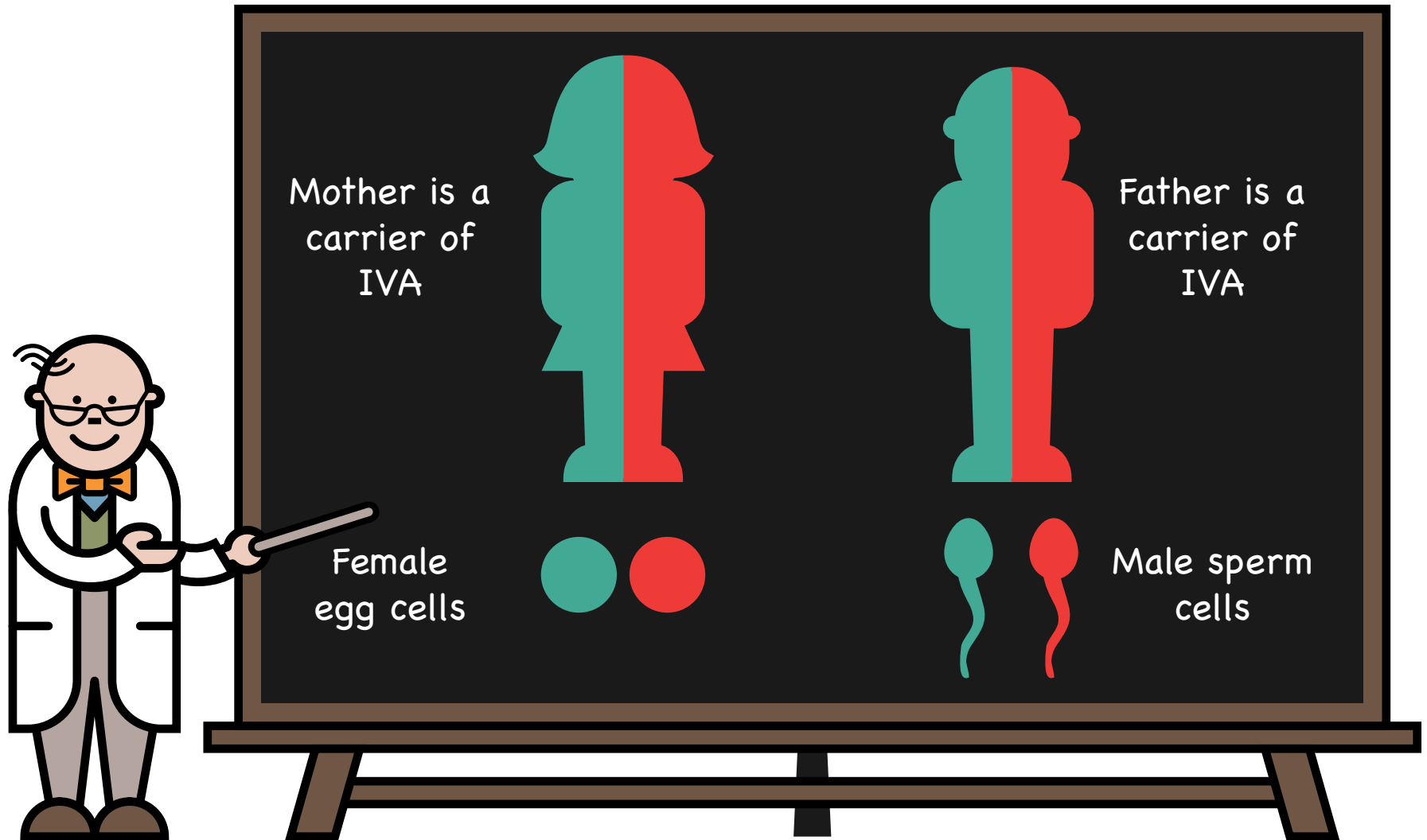


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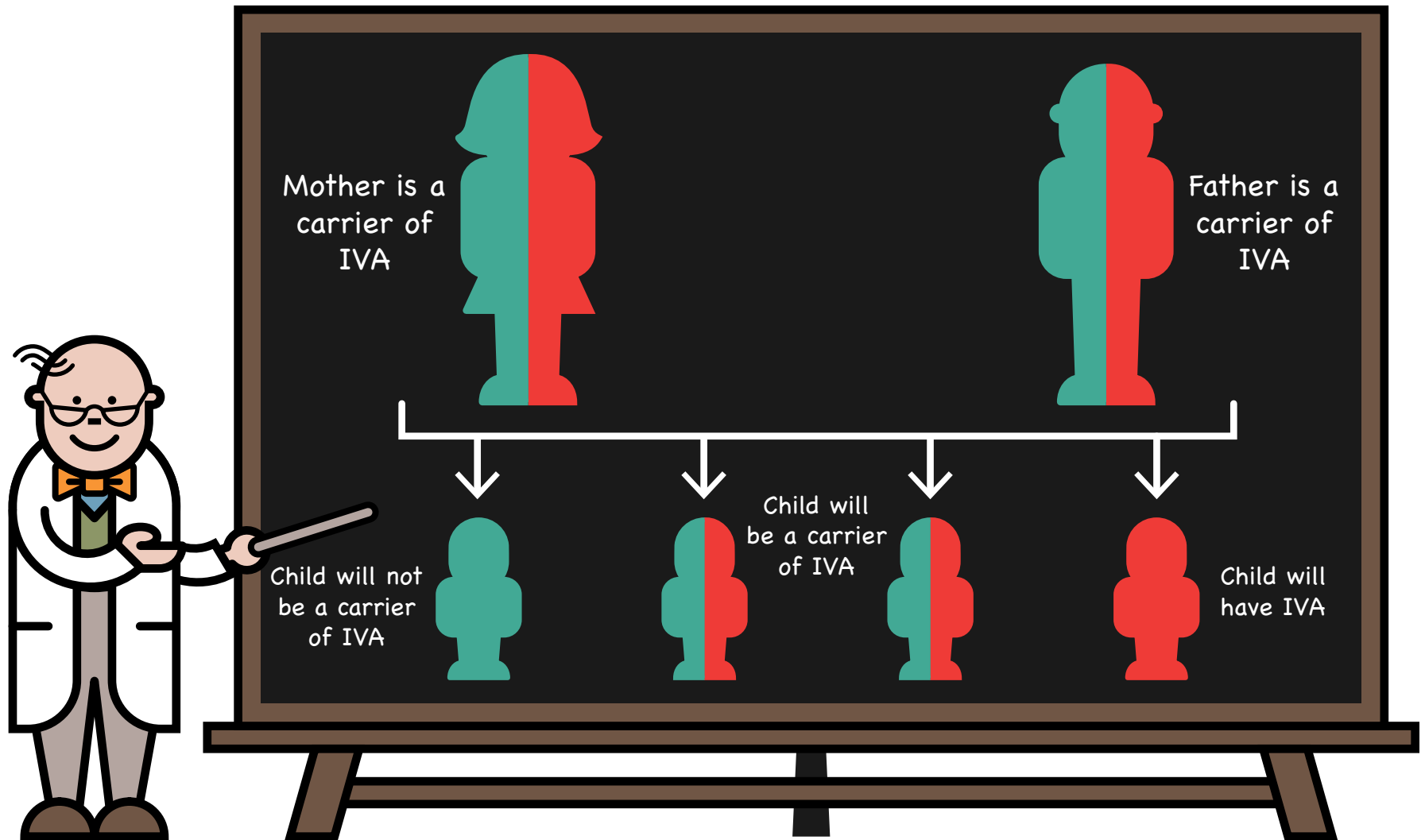


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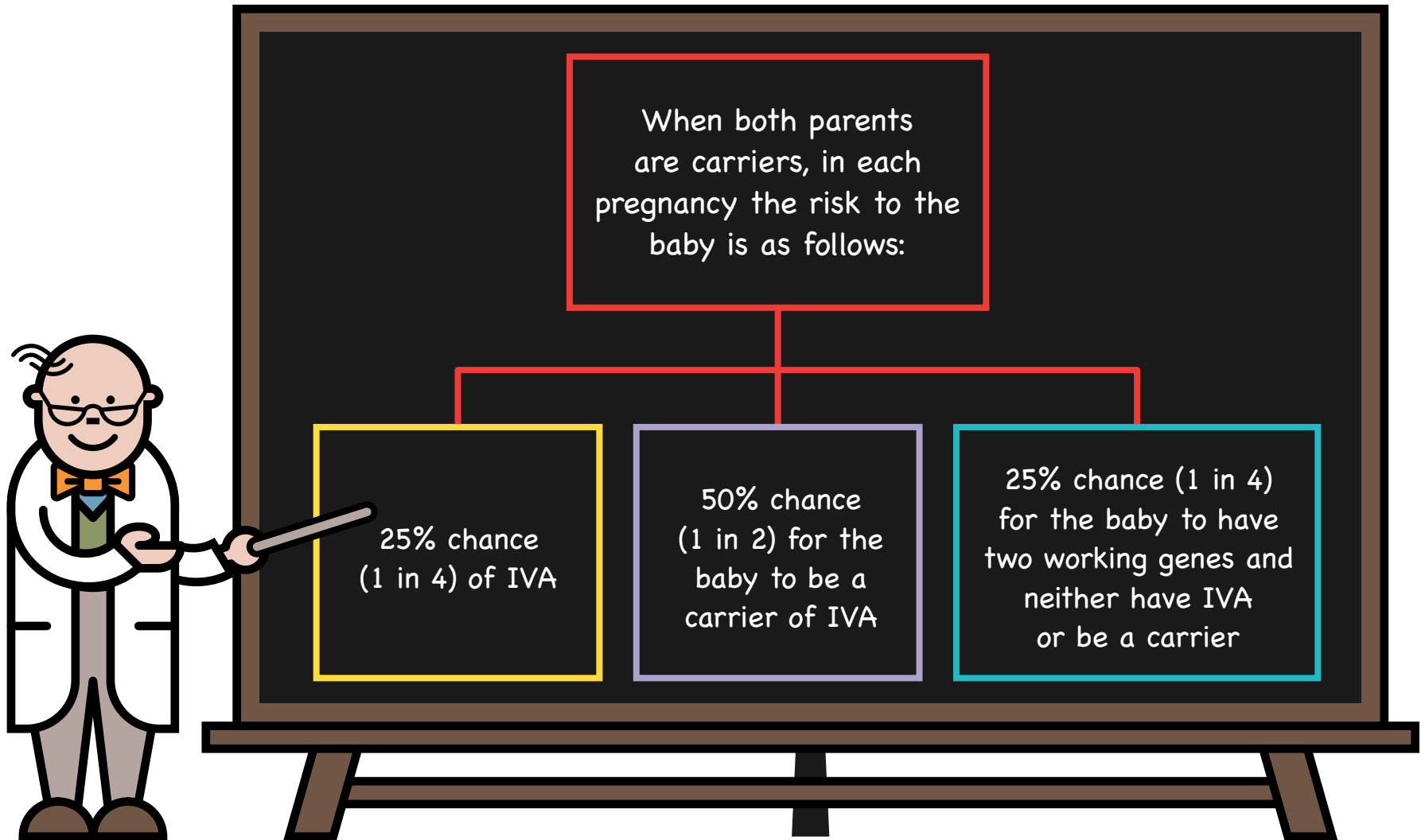
Inheritance – Autosomal recessive (carriers of IVA)



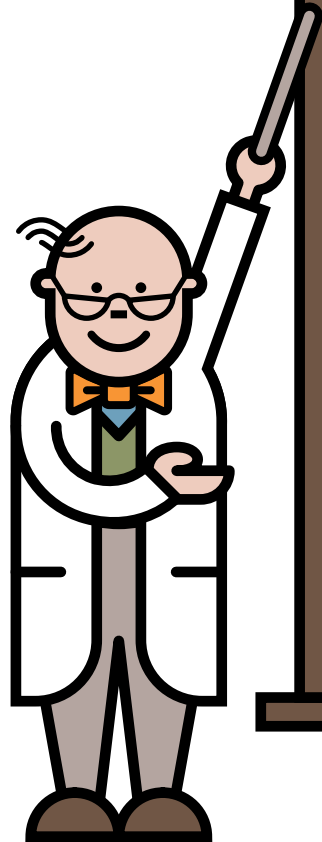
Inheritance – Autosomal recessive – possible combinations



Future pregnancies



Take home messages

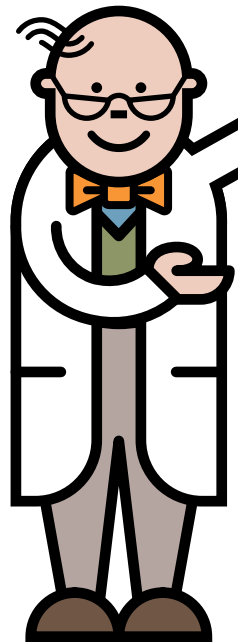


IVA is a serious inherited metabolic disorder that can lead to severe brain damage

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

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

Take home messages



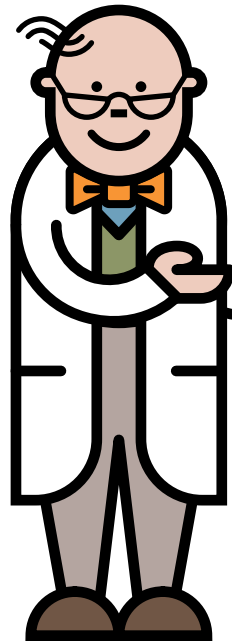
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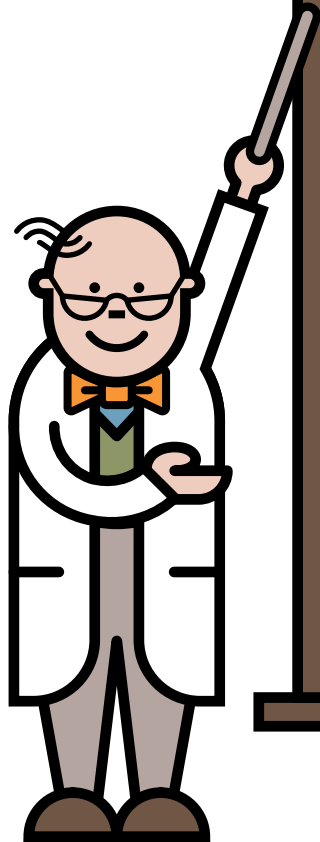


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Helpful hints



✓ Always ensure you have a good supply of your dietary products and 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

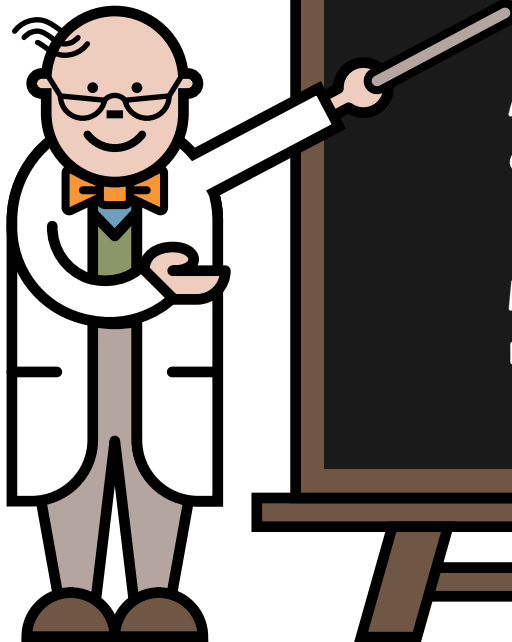
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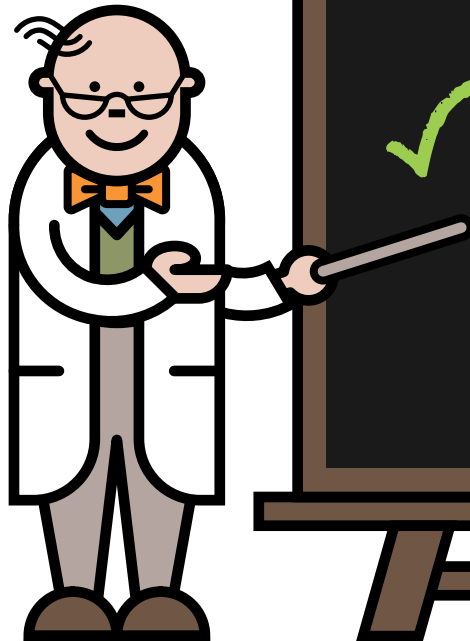
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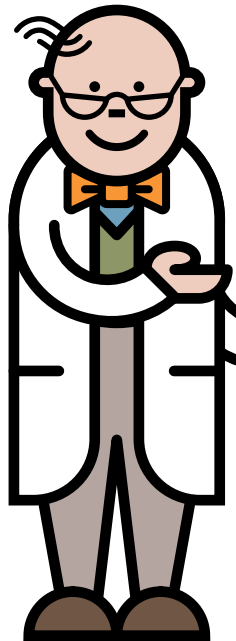
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Who's who

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

Visit www.lowproteinconnect.com
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.



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Inherited Metabolic Diseases

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