

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

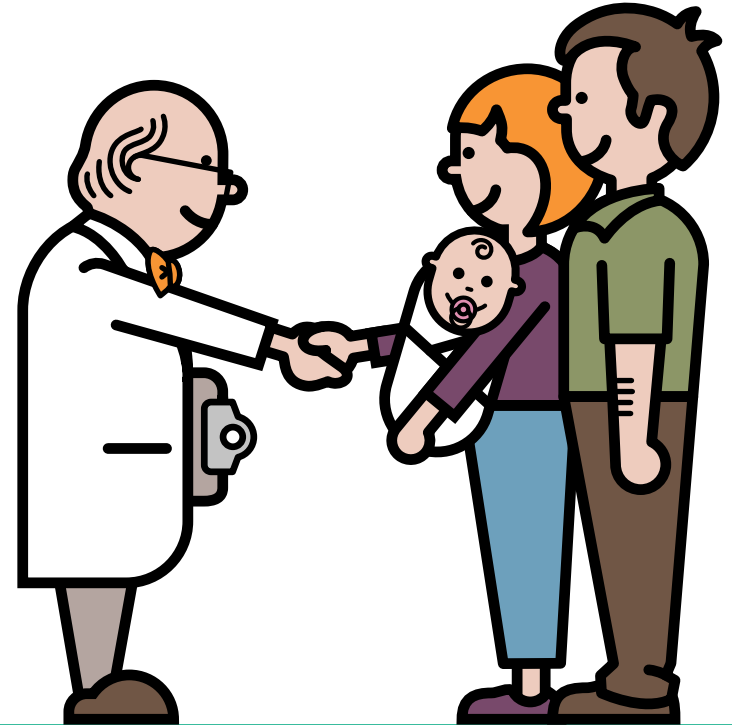
VERSION 2, FEBRUARY 2017

MSUD

Supported by 
as a service to metabolic medicine

MSUD

Information for families following
a positive newborn screening



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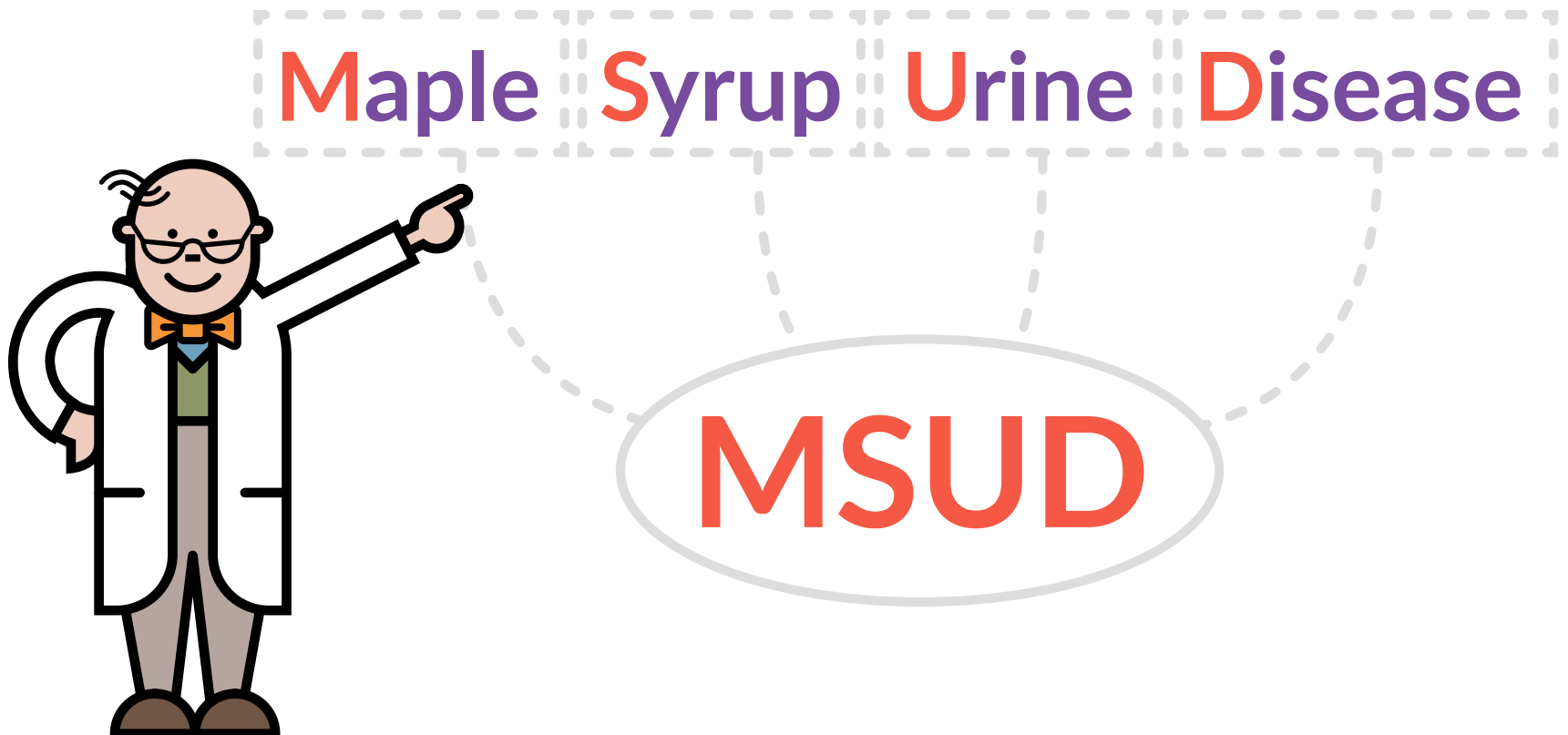
Tools Enabling Metabolic Parents LEarning

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

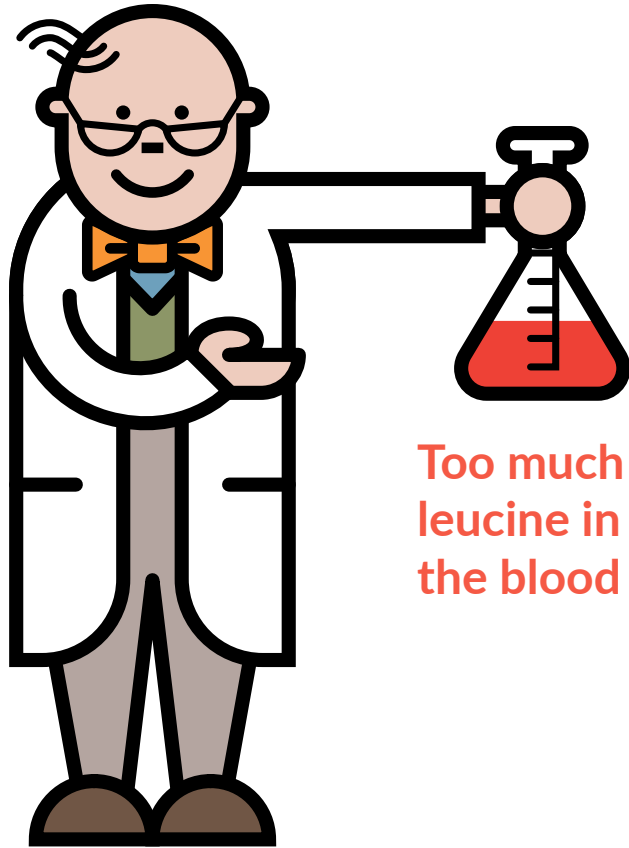
What is MSUD?

MSUD stands for Maple Syrup Urine Disease

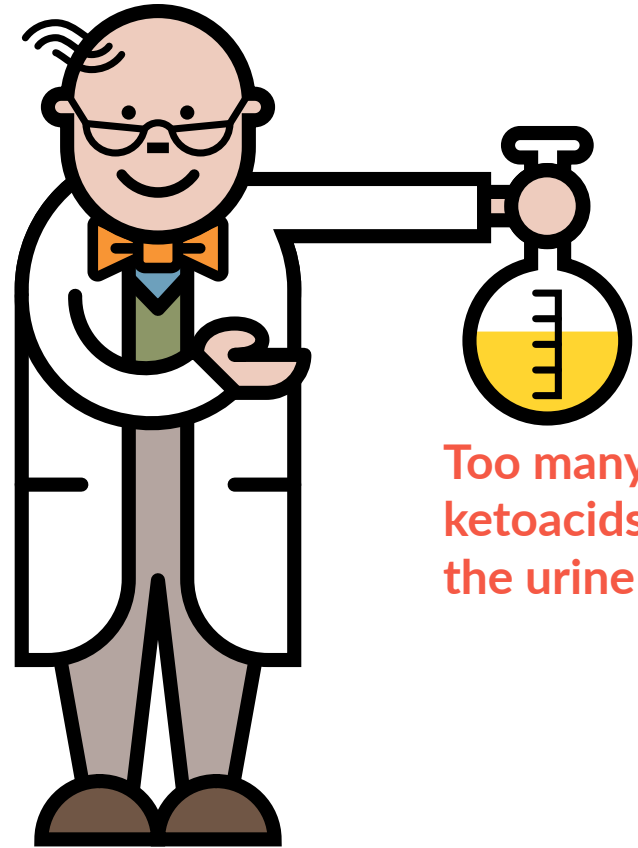
It is an inherited metabolic condition



What is MSUD?



Too much
leucine in
the blood



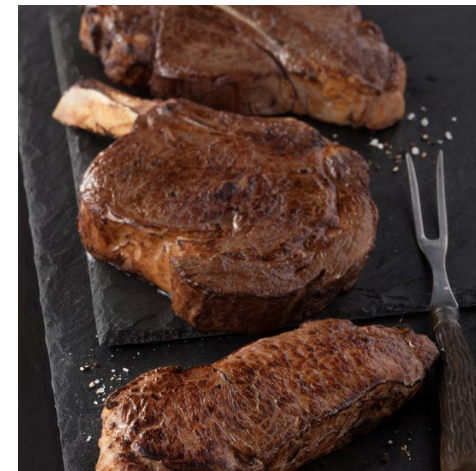
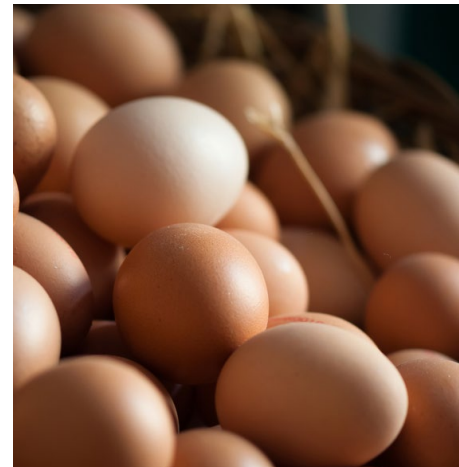
Too many
ketoacids in
the urine

MSUD and protein

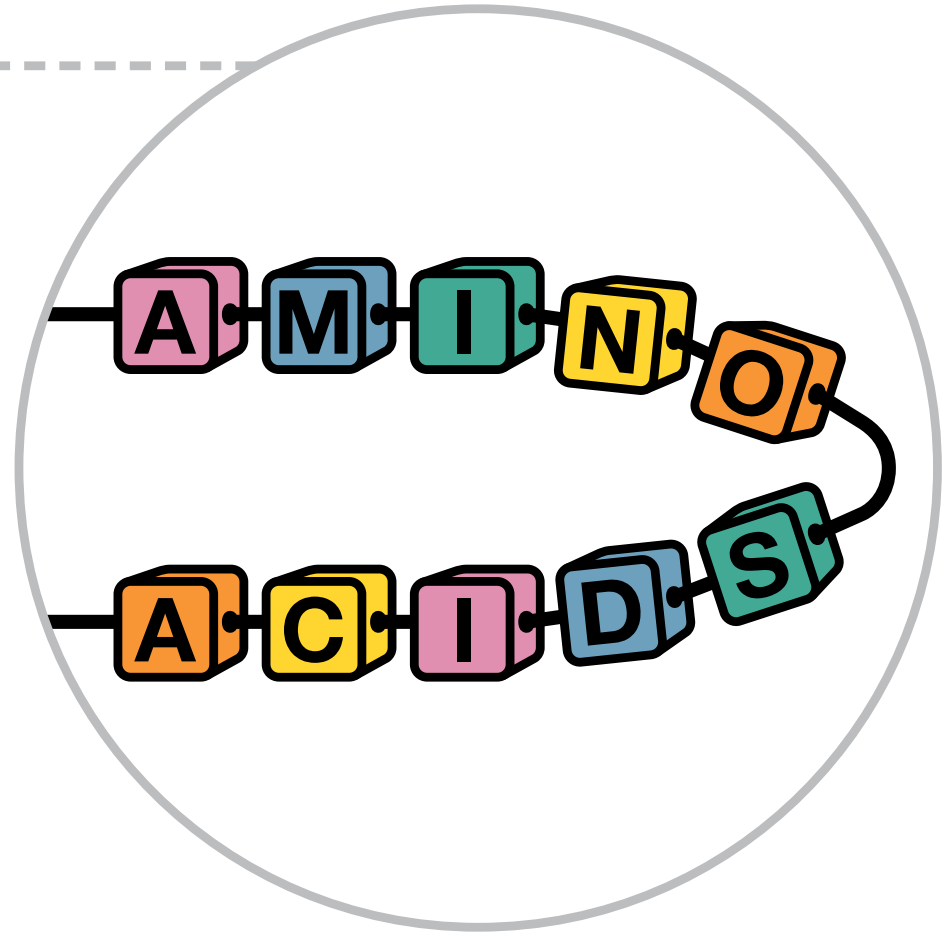
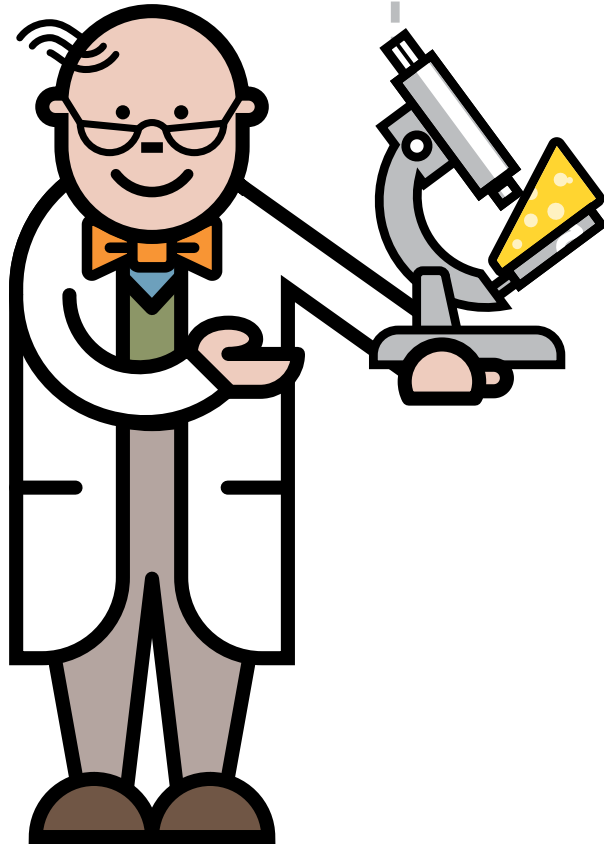
MSUD affects the way your baby breaks down protein

Many foods contain protein

The body needs protein for growth and repair



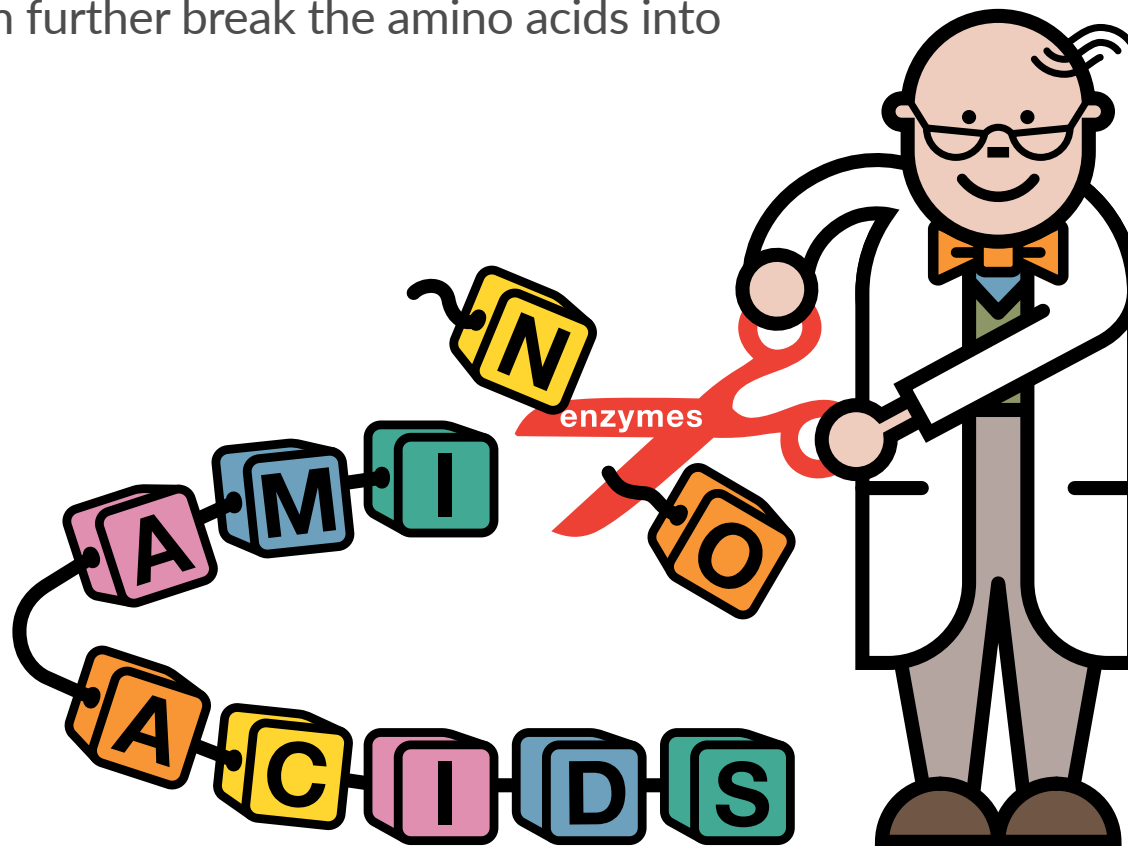
What is protein?



Protein and enzymes

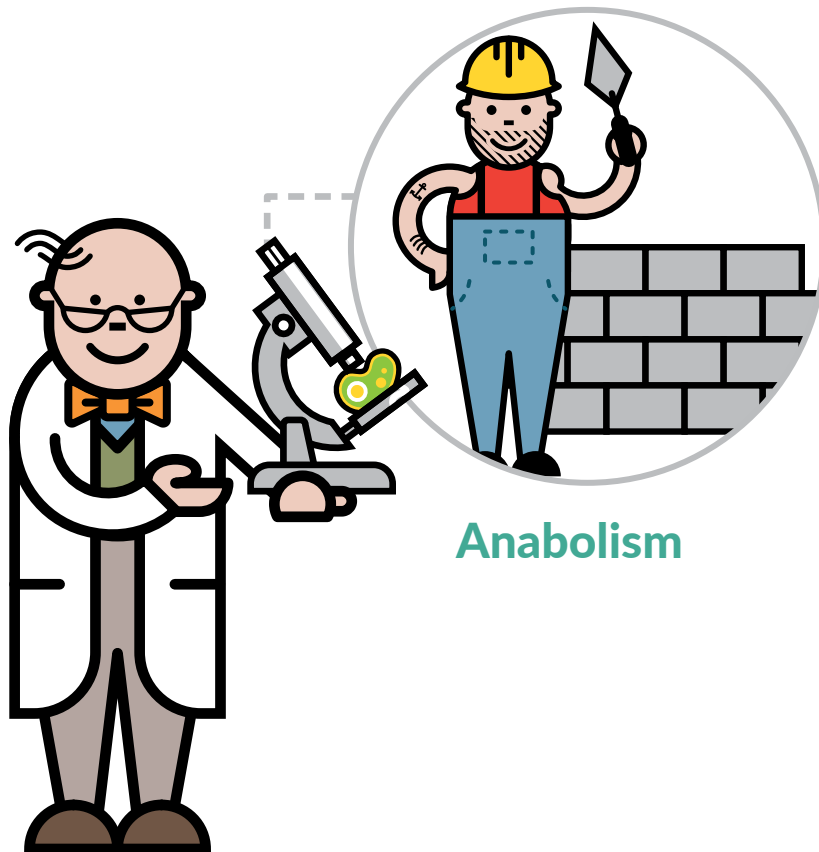
Protein is broken down into amino acids (building blocks of protein) by enzymes (which are like chemical scissors).

Enzymes then further break the amino acids into smaller parts.

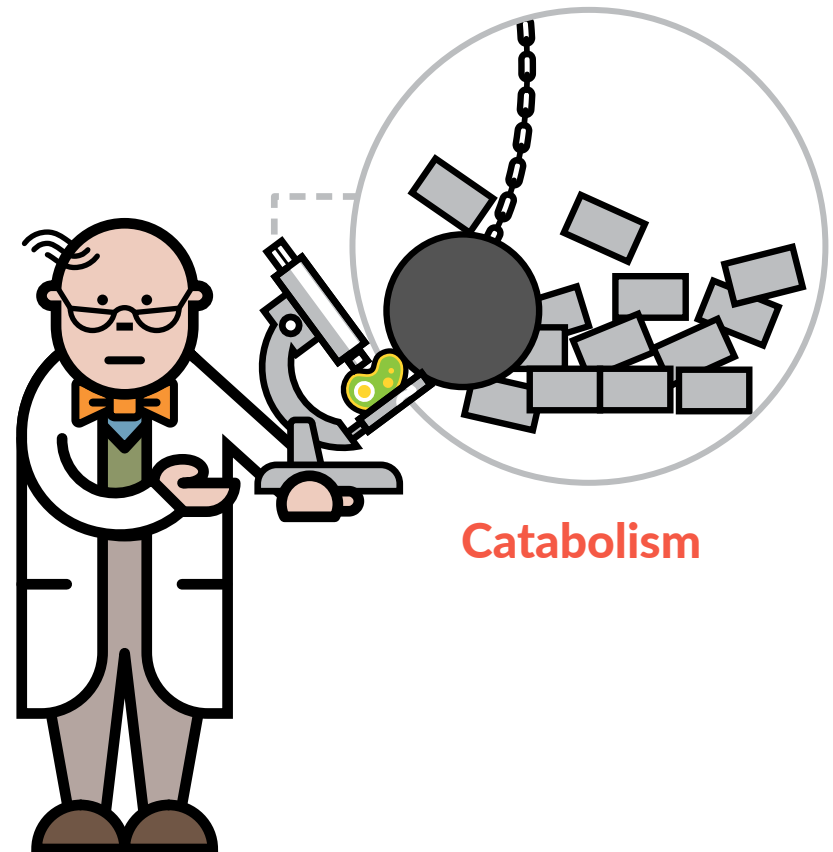


Protein metabolism

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



Anabolism

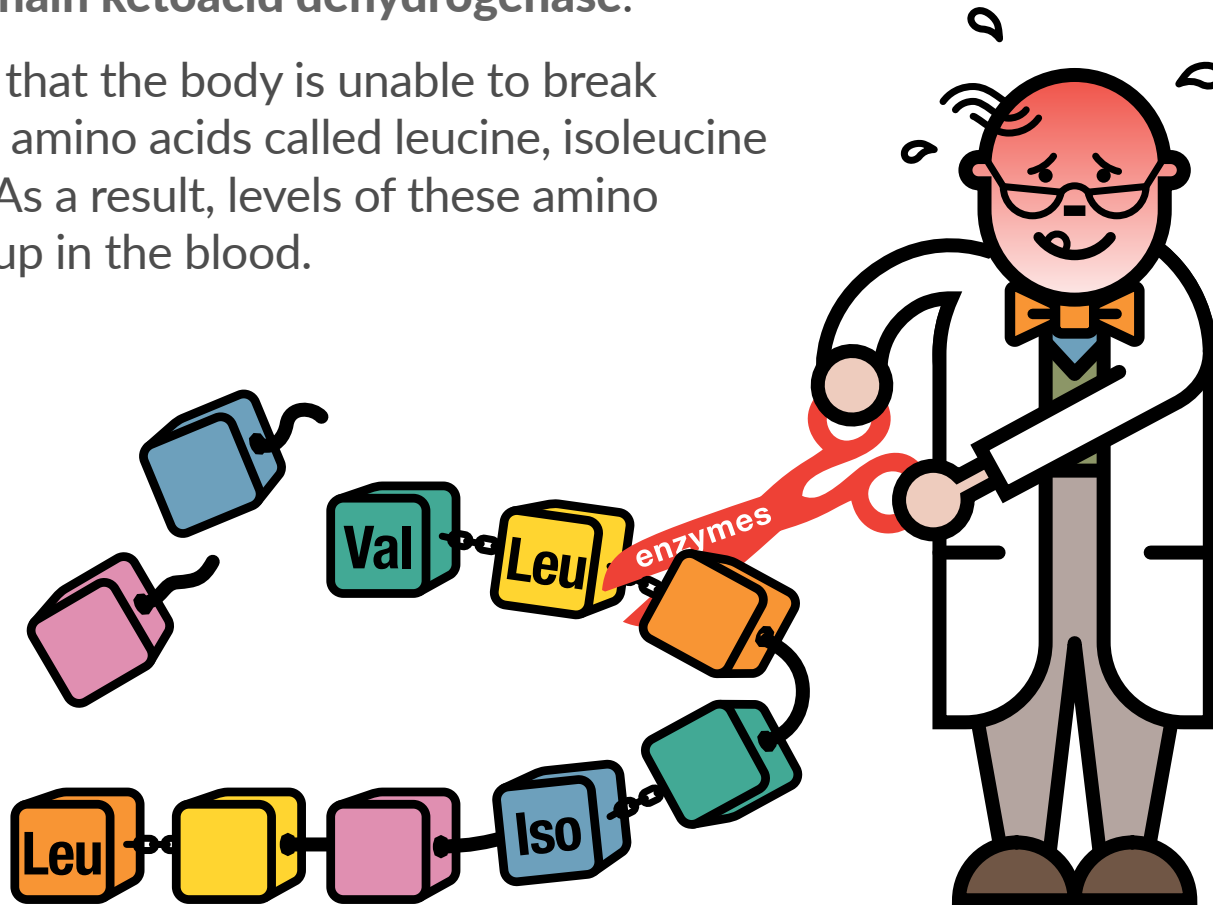


Catabolism

What happens in MSUD?

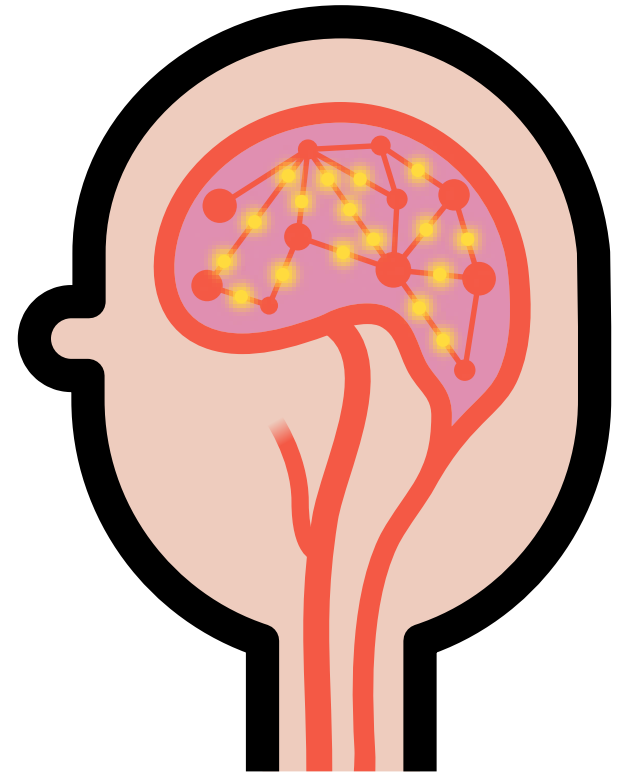
In MSUD, the body lacks an enzyme called **branched chain ketoacid dehydrogenase**.

This means that the body is unable to break down three amino acids called leucine, isoleucine and valine. As a result, levels of these amino acids build up in the blood.



What can go wrong in MSUD?

The build-up of harmful chemicals can cause damage to the brain. It may cause delays to normal development like walking and talking.



Early management can prevent brain damage and learning difficulties

What are the symptoms in MSUD?

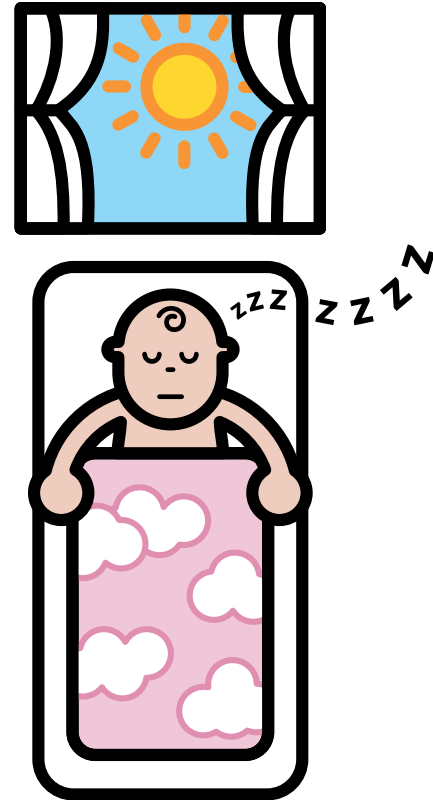
Some babies with MSUD become ill in the first few days of life before the newborn screening result is available.

Symptoms include:

- poor feeding
- vomiting
- dehydration (lack of body fluids)
- floppy baby
- excessively sleepy
- seizures
- rapid breathing
- sweet-smelling urine (like maple syrup)

The effects of MSUD quickly become life-threatening if left untreated.

Some children may develop symptoms at a later stage. They may present with developmental delay or with an acute childhood illness such as vomiting or diarrhoea.



How is MSUD diagnosed?

MSUD is usually diagnosed by newborn screening. High levels of leucine are found in the blood.



How is MSUD managed?

MSUD is managed with the following special diet:

Limited high protein foods



Measured amounts of leucine (protein) containing foods

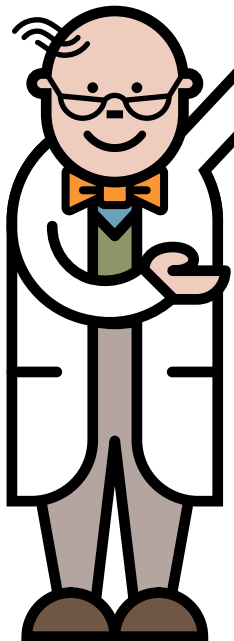
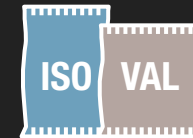


A protein substitute



Low protein foods

Isoleucine and valine supplements



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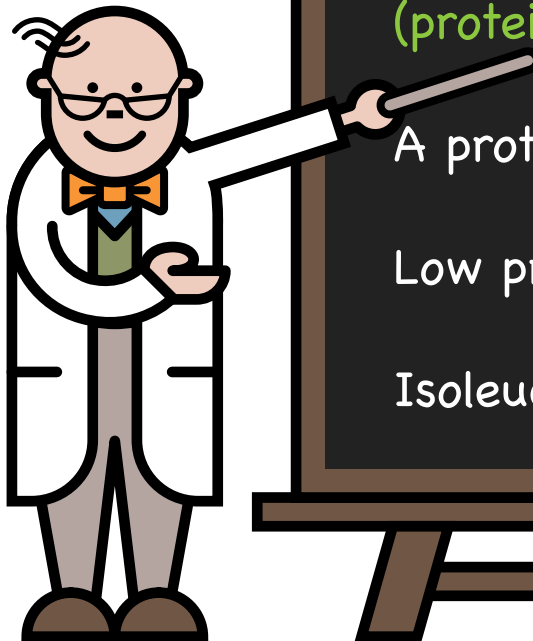
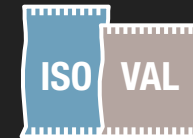


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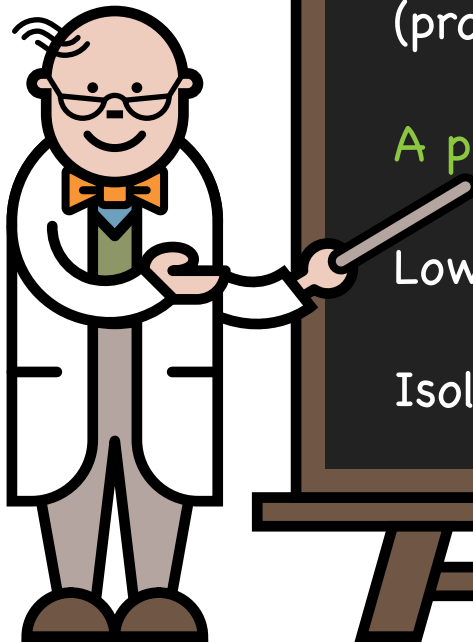
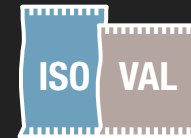


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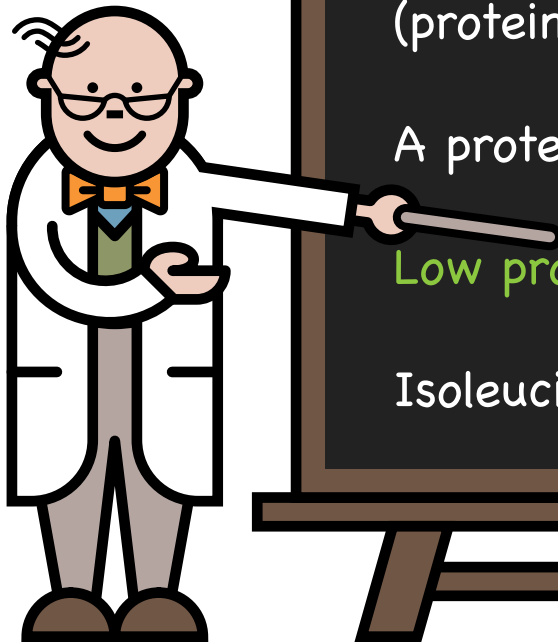
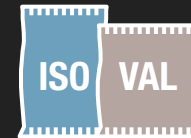


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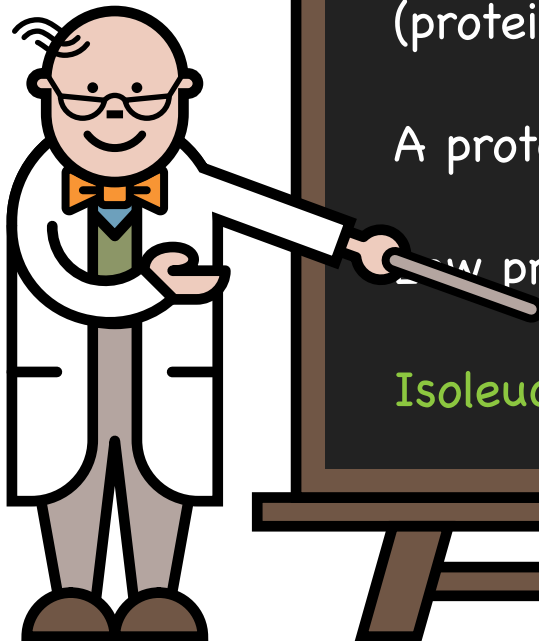
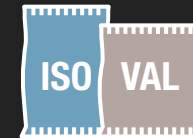


A protein substitute



Low protein foods

Isoleucine and valine supplements



High protein foods

These foods are high in leucine (protein) and must be avoided: **meat, fish, eggs, cheese, bread, pasta, nuts, seeds, soya, Quorn and tofu.**



Measured leucine intake

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

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

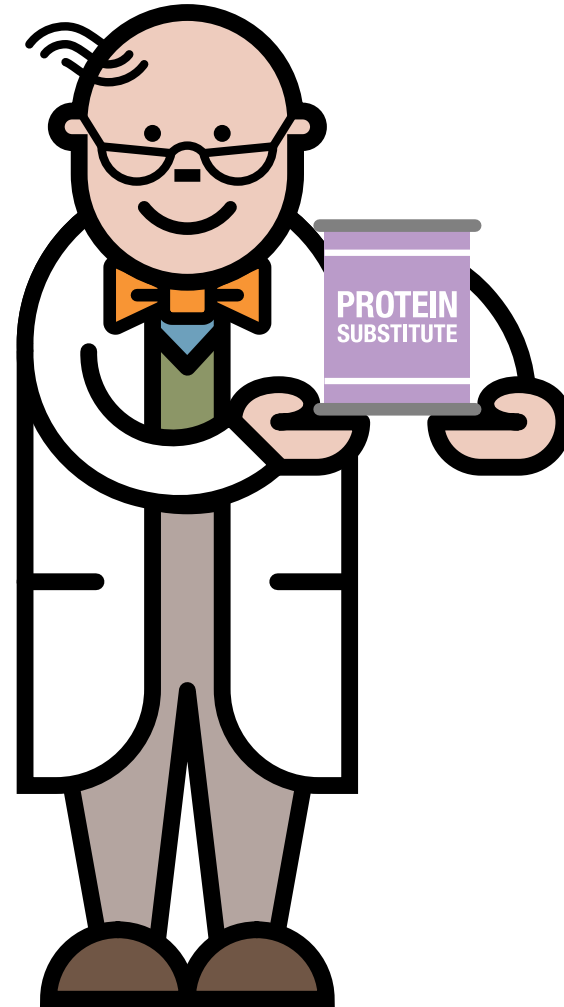


Protein substitute

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.

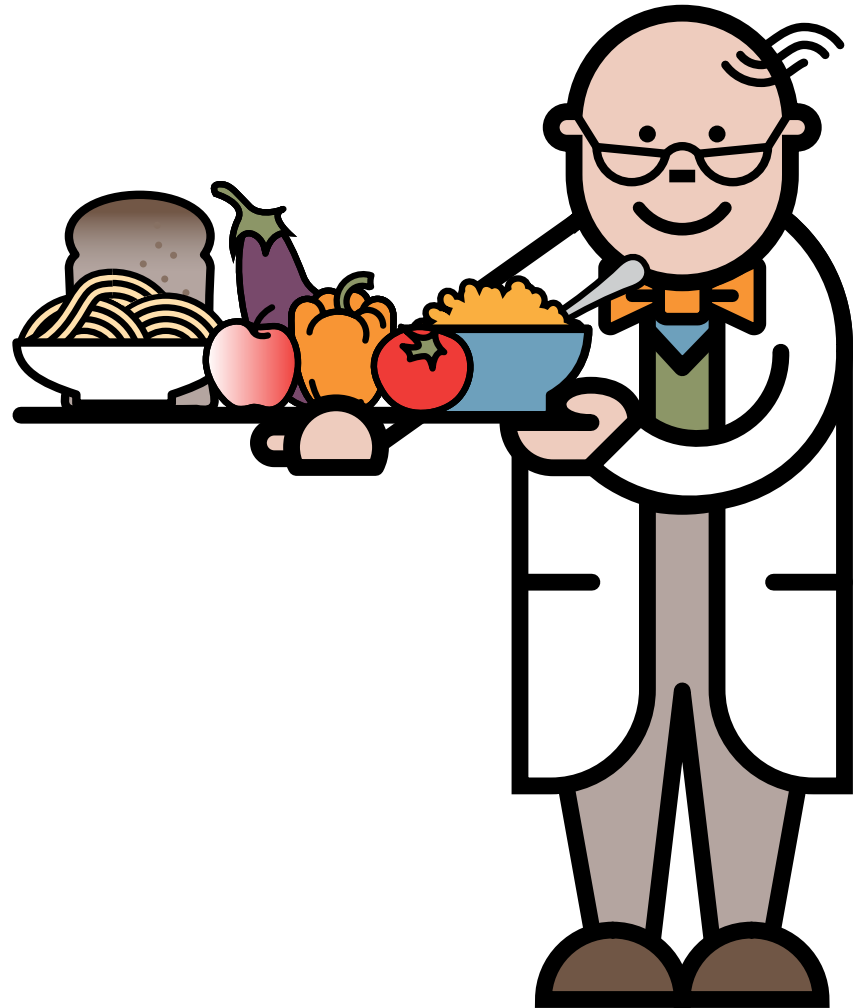


Low protein foods

There are many low protein foods. These include fruit, many vegetables and prescribable low protein foods such as bread and pasta.

They provide:

- a source of energy
- variety in the diet



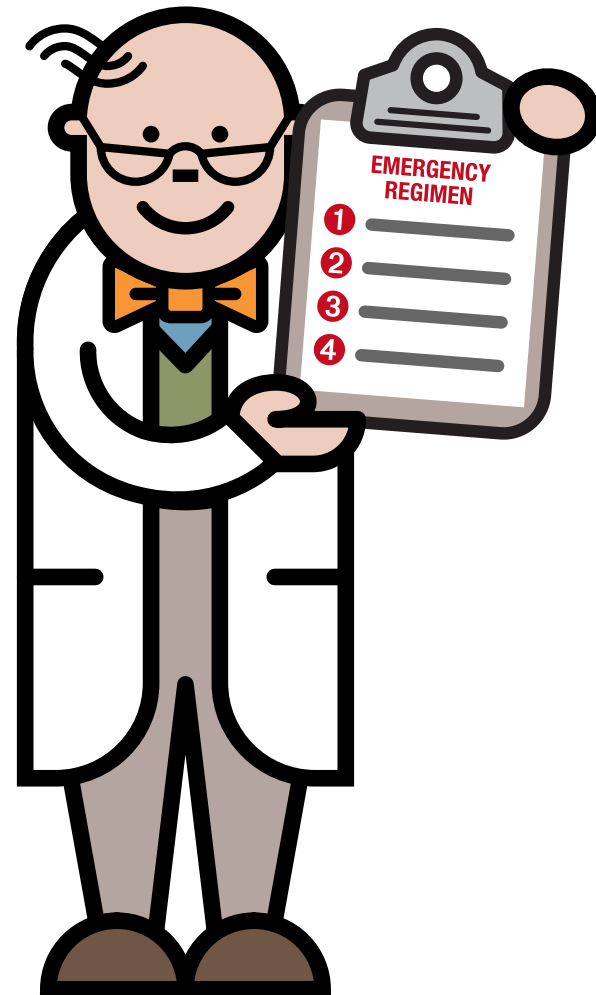
Metabolic crisis

- A '**metabolic crisis**' causes a build-up of leucine and other toxic chemicals
- It is usually triggered by childhood illnesses e.g. vomiting and diarrhoea
- It is important to manage a metabolic crisis quickly and properly



How is MSUD managed during illness?

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



How is MSUD managed during illness?

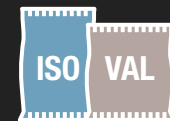
Stop all protein in food & drink



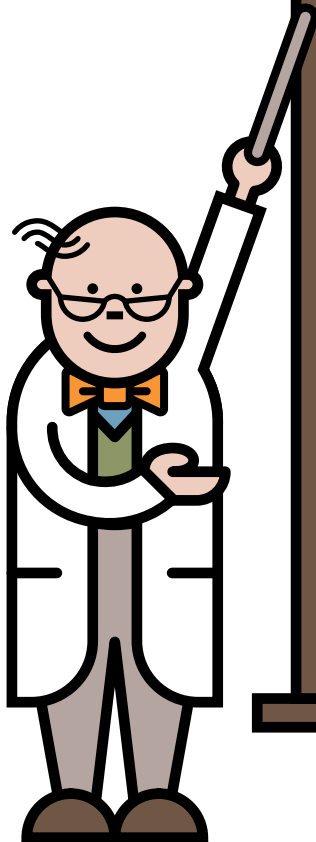
Start the emergency regimen.
This is made up of protein
substitute and glucose polymer



Continue isoleucine and valine
supplements



Do a blood test and phone
your metabolic team

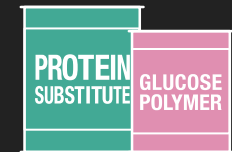


How is MSUD managed during illness?

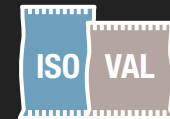
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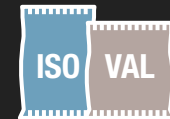
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Continue isoleucine and valine supplements



Do a blood test and 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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Regularly update your metabolic team

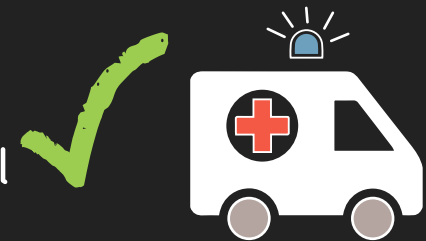


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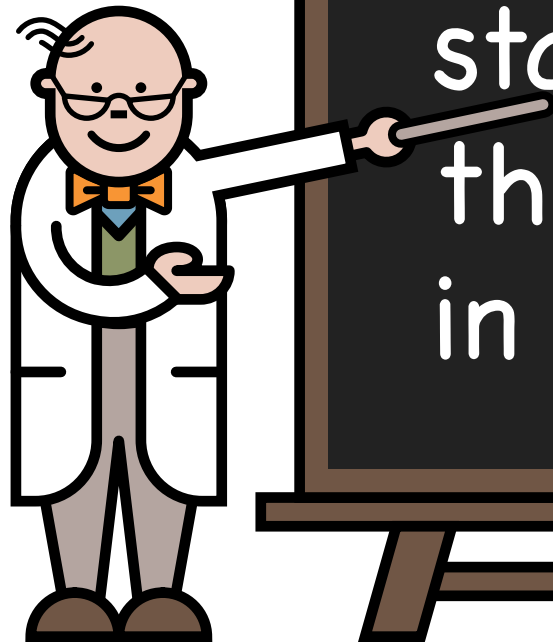
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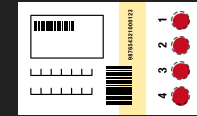
Key message



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

How is MSUD monitored?

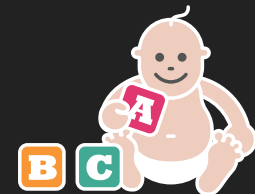
Frequent blood tests to check leucine, isoleucine and valine levels



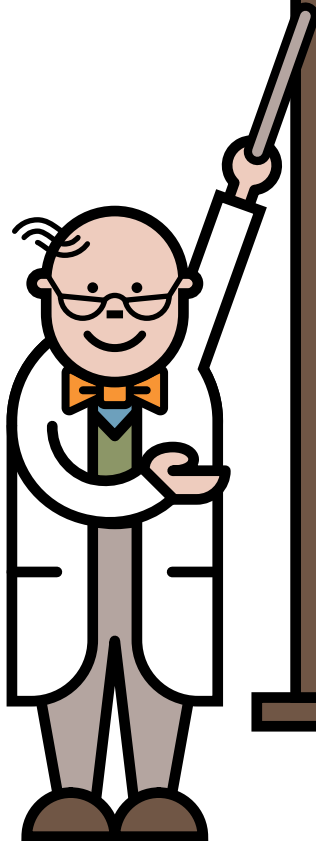
Height and weight



Developmental checks

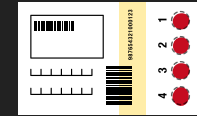


Diet is adjusted according to age, weight and blood tests



How is MSUD monitored?

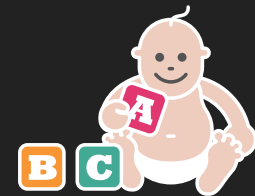
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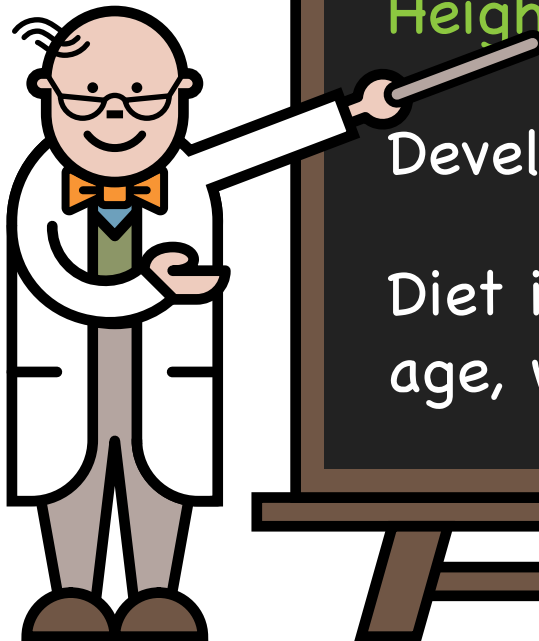
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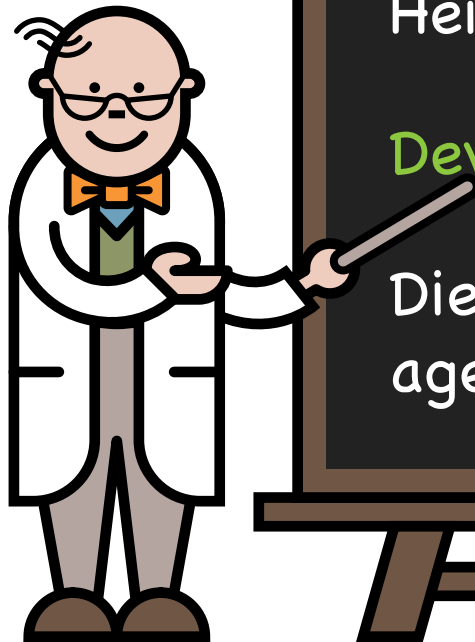
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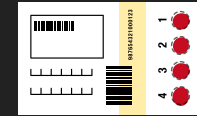
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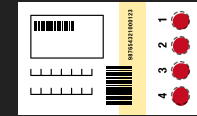


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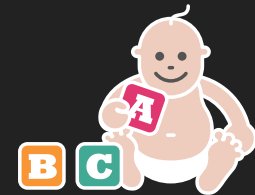
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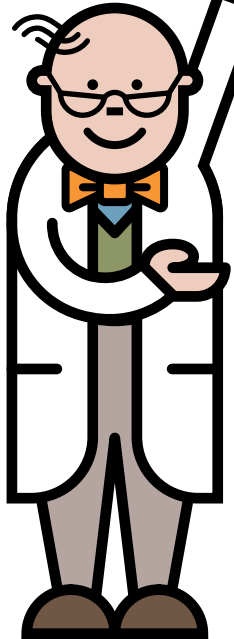
Developmental checks



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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

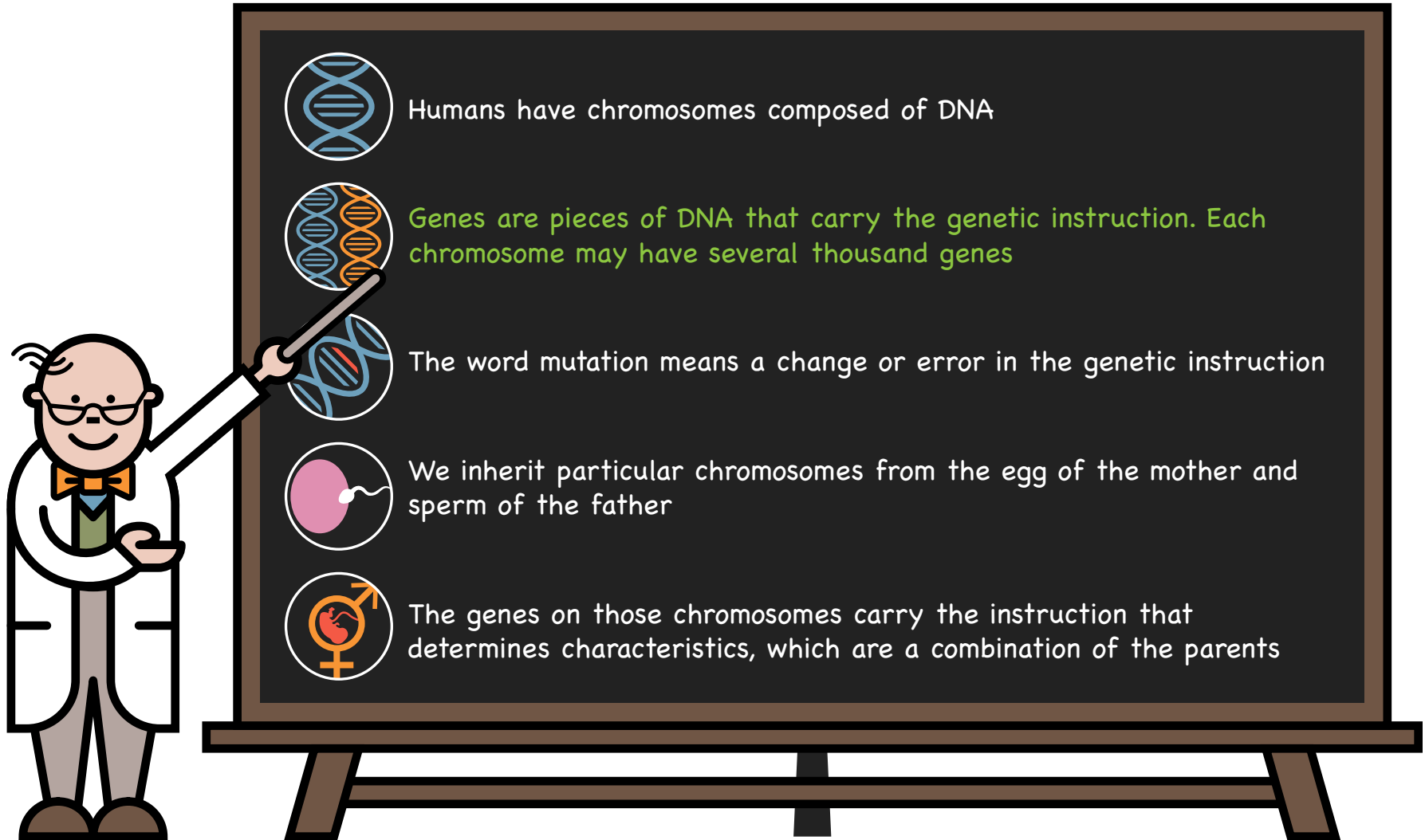



We inherit particular chromosomes from the egg of the mother and sperm of the father





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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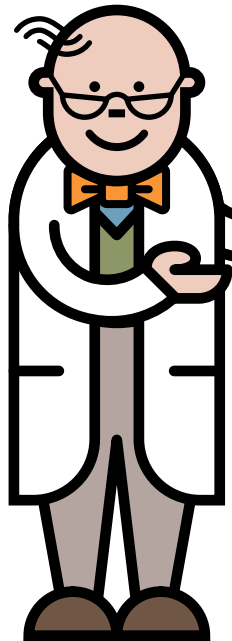
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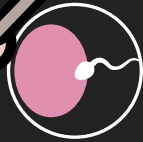
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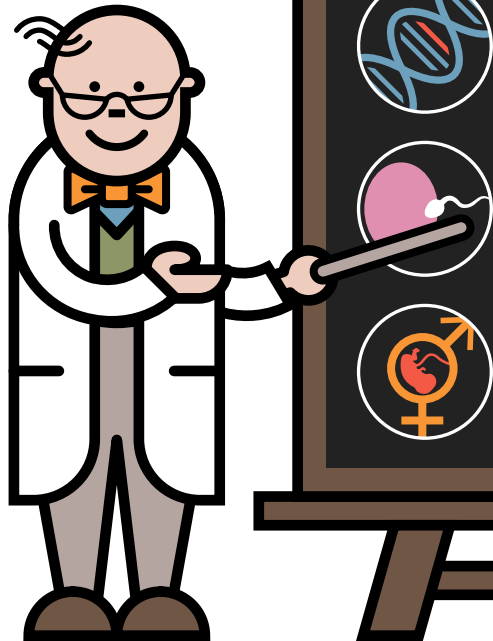


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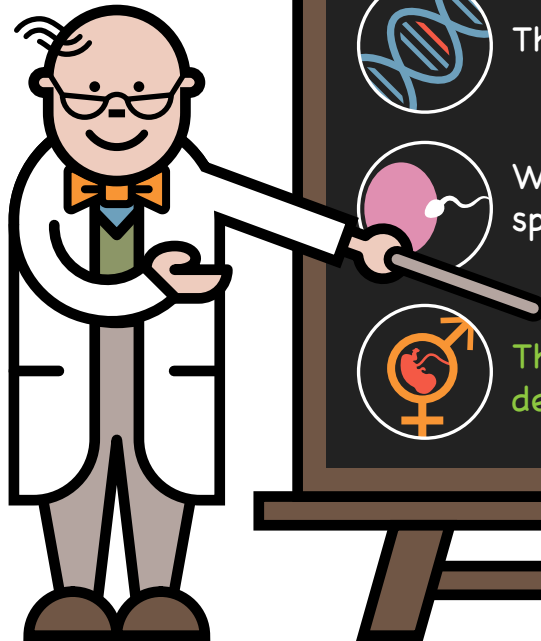


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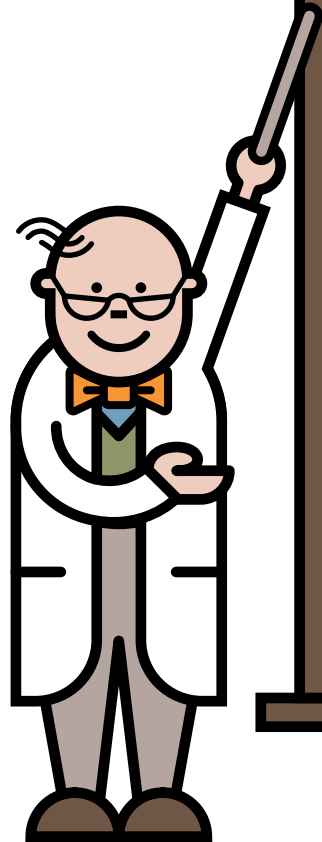


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Inheritance



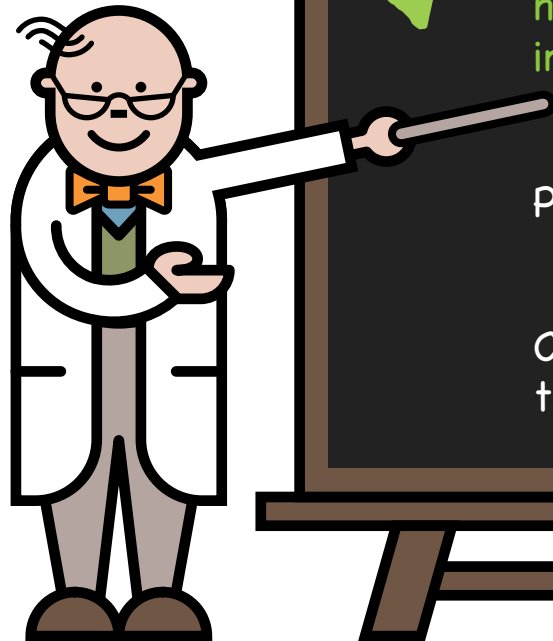
MSUD is an inherited condition. There is nothing that could have been done to prevent your baby from having MSUD

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

Parents of children with MSUD are carriers of the condition

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

Inheritance



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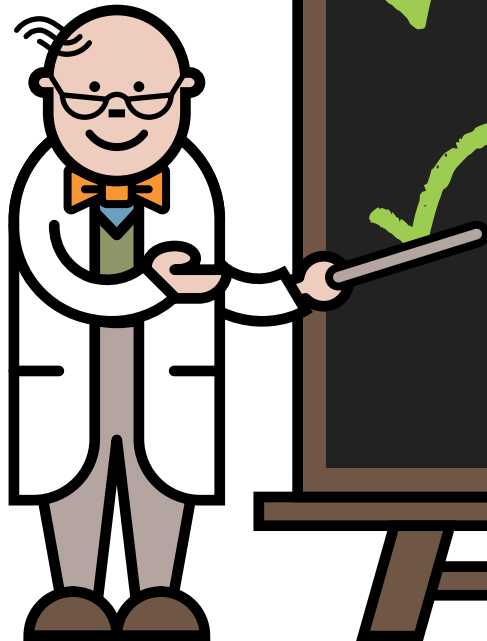


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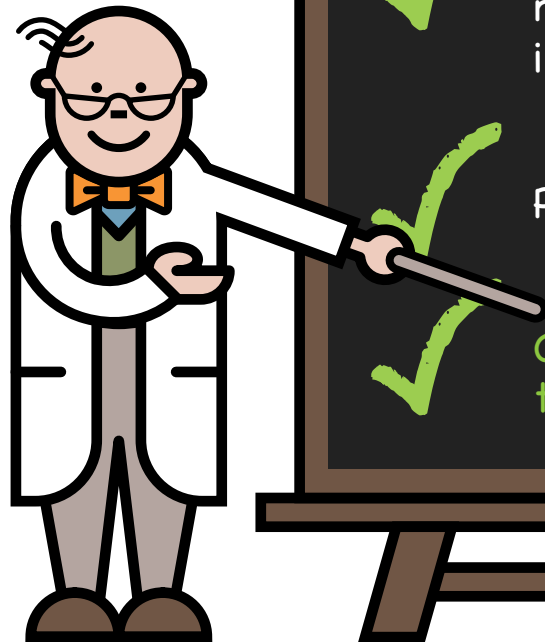
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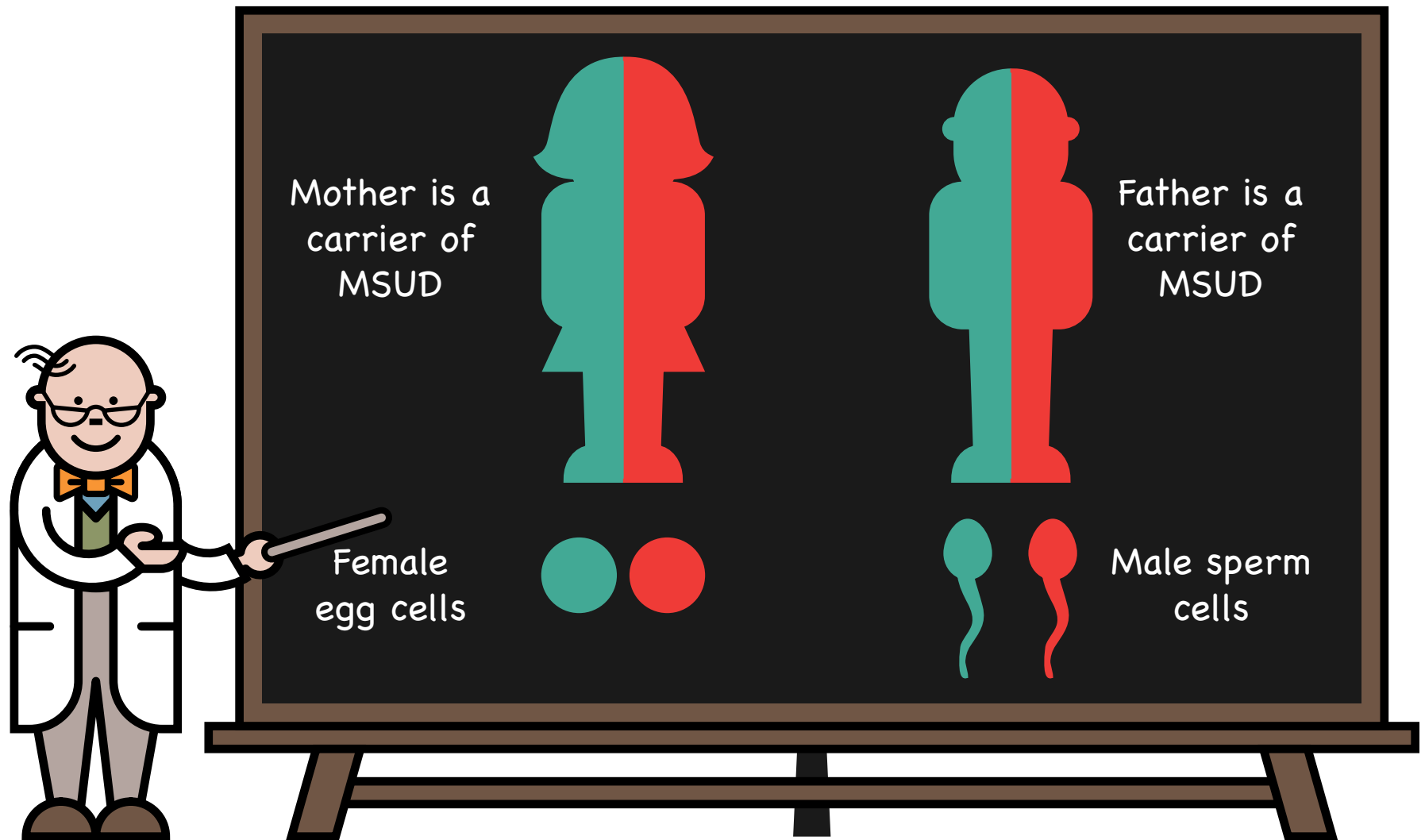


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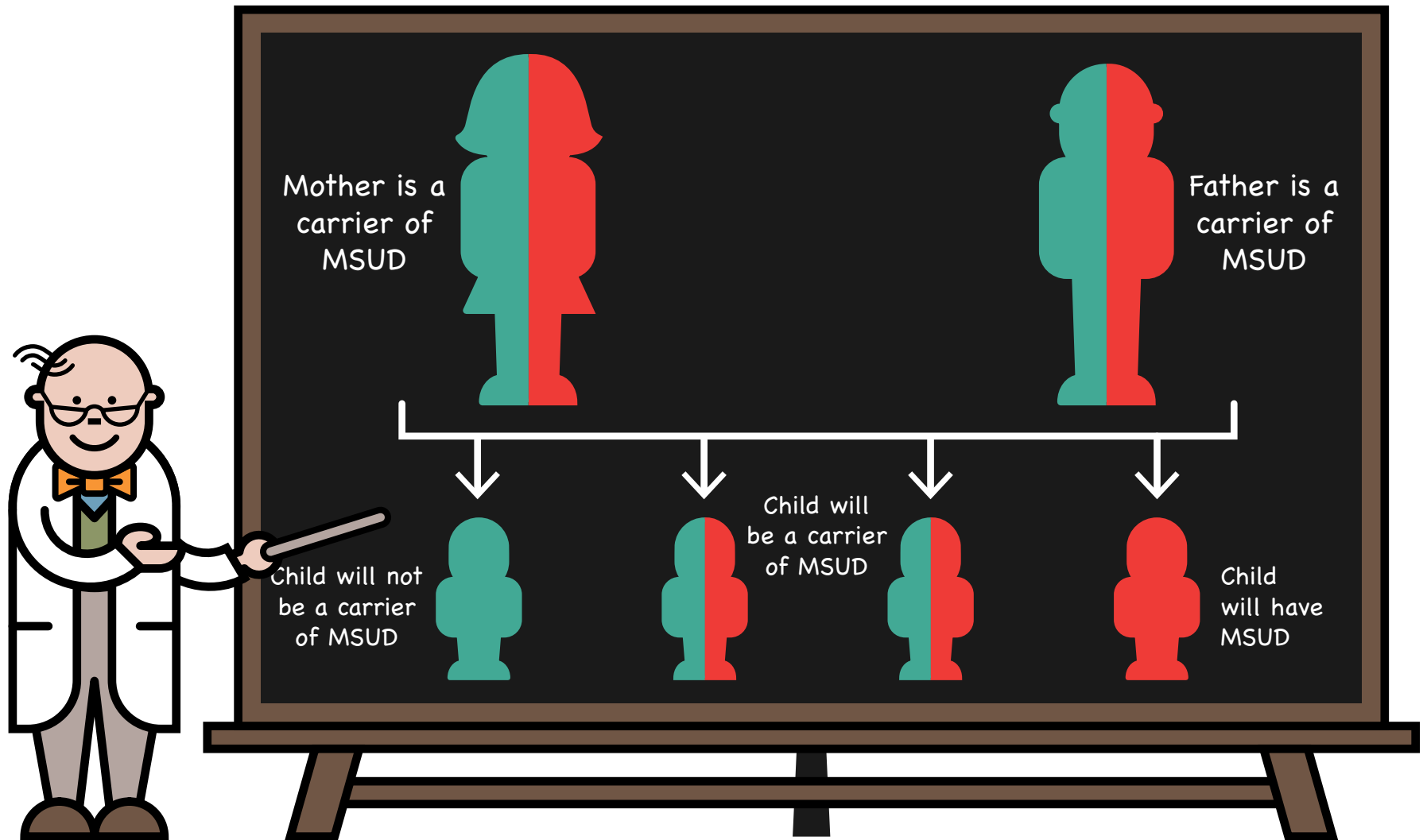


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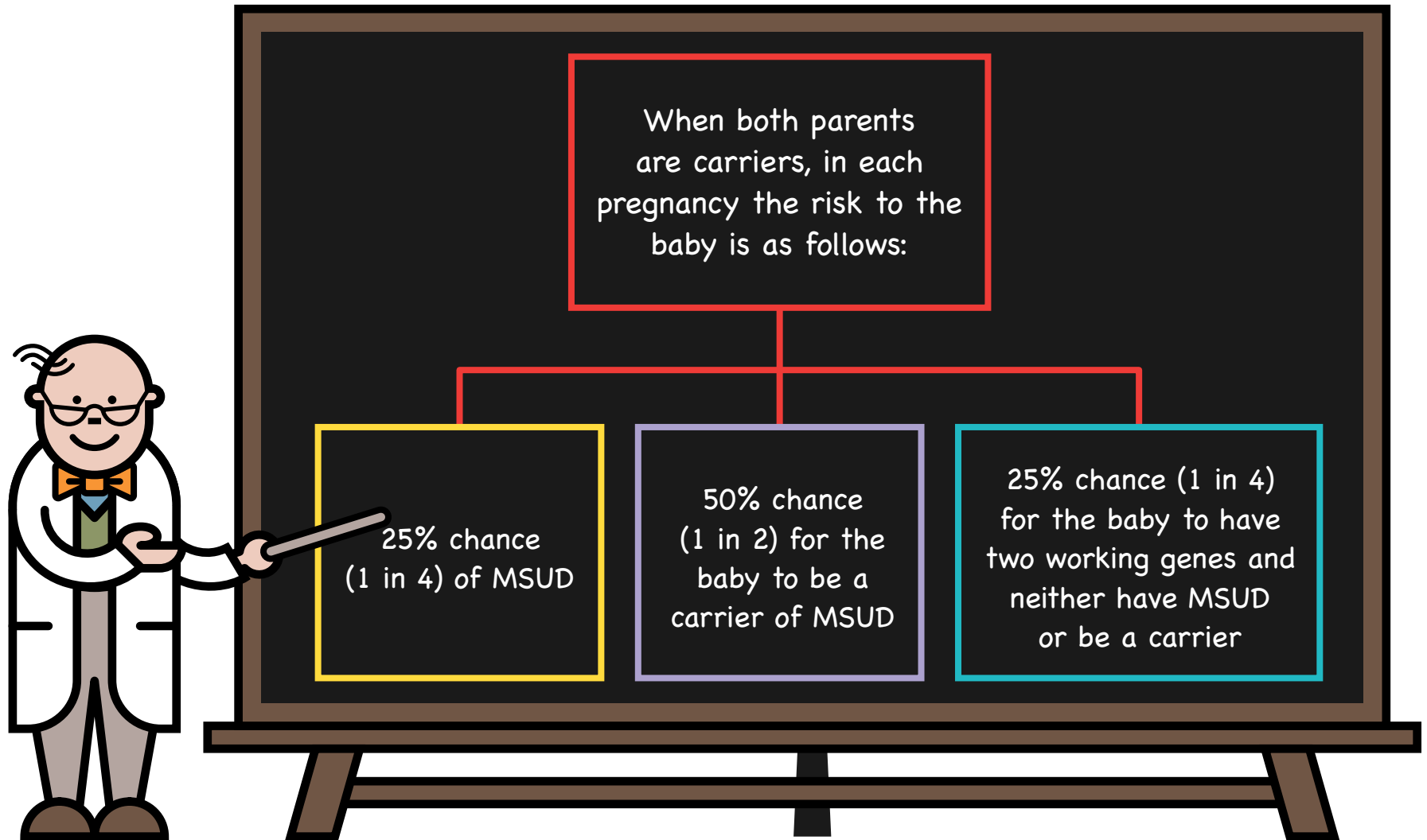
Inheritance – Autosomal recessive (carriers of MSUD)



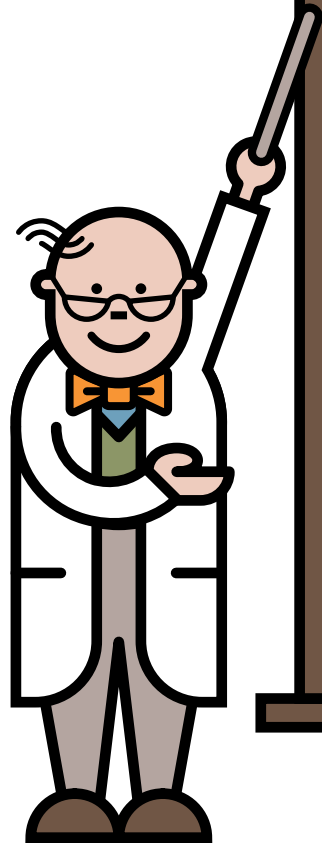
Inheritance – Autosomal recessive – possible combinations



Future pregnancies



Take home messages



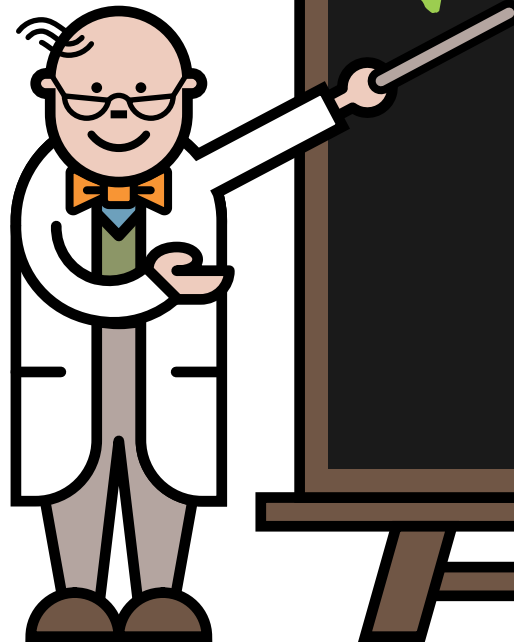
MSUD is a serious inherited metabolic disorder that can lead to severe brain damage

Damage can be prevented with a protein restricted diet, a protein substitute and appropriate illness management

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

Regular blood tests are essential to monitor the management of MSUD

Take home messages



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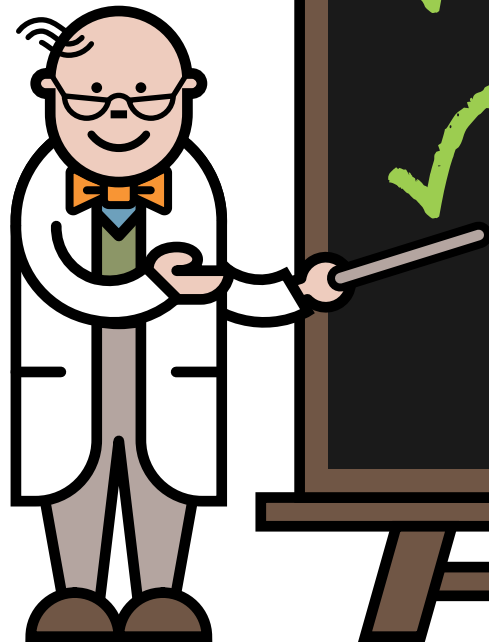


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Take home messages



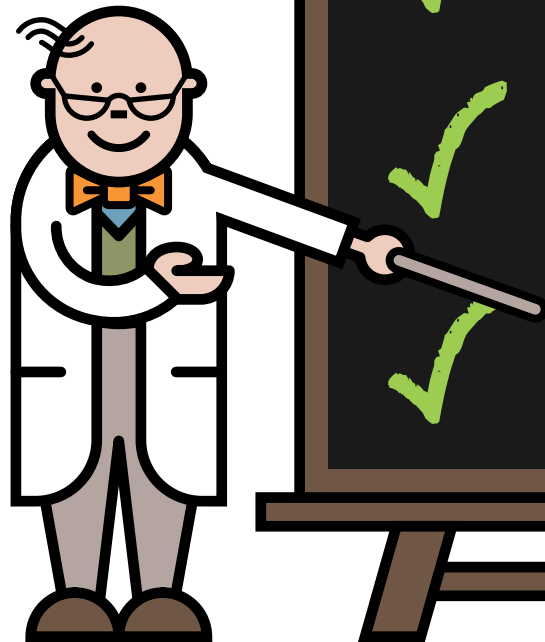
✓ MSUD is a serious inherited metabolic disorder that can lead to severe brain damage

✓ Damage can be prevented with a protein restricted diet, a protein substitute and appropriate illness management

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

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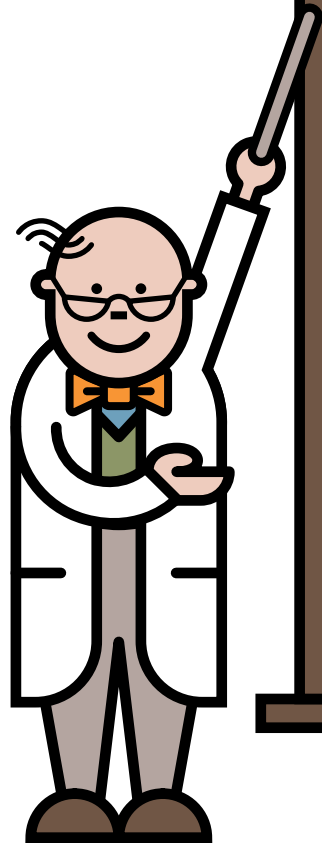
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Helpful hints



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

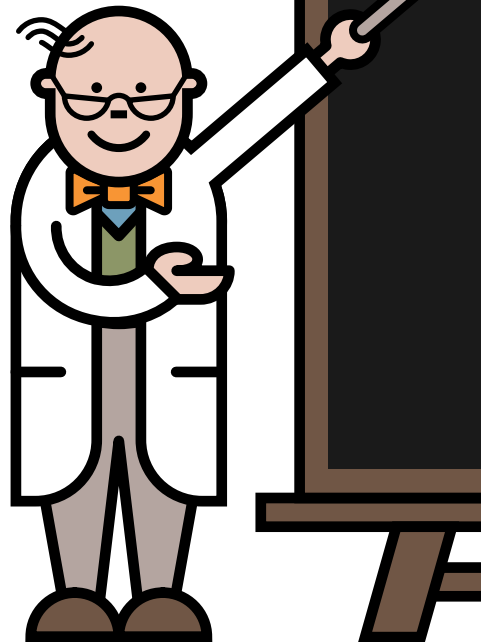
Your dietary products are prescribed by your GP. These are obtained via a pharmacy or home delivery

Always ensure you have your emergency feed products and a written emergency plan

Always ensure you have sufficient blood testing equipment and send samples on a regular basis

Medications to control fever should be given as normally recommended – always keep supplies available

Helpful hints



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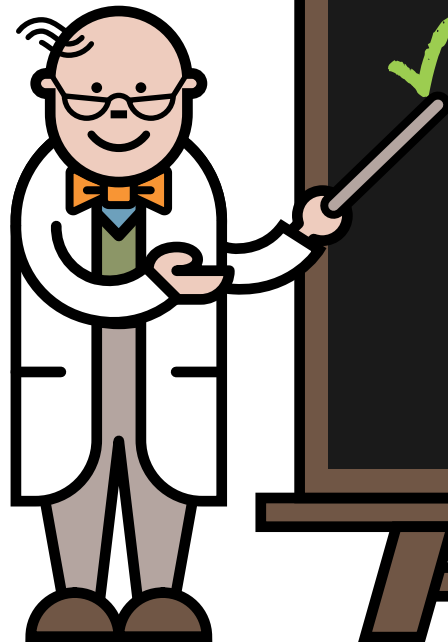
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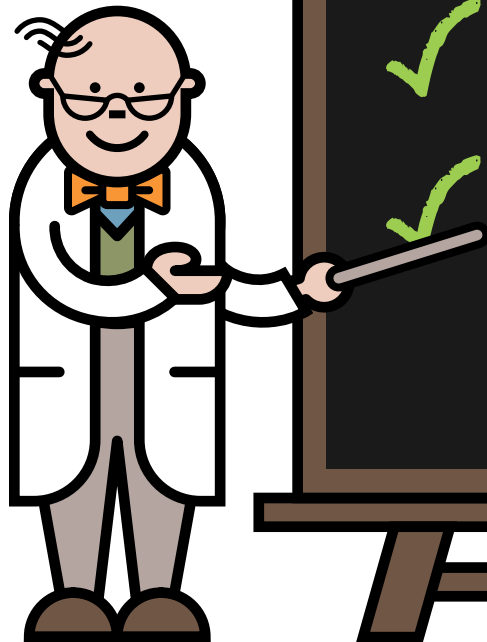
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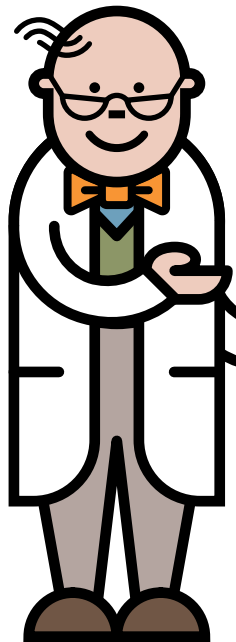
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Who's who

- 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
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ClimBTM

Children Living with
Inherited Metabolic Diseases

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