

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 3, APRIL 2020

MMA

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).

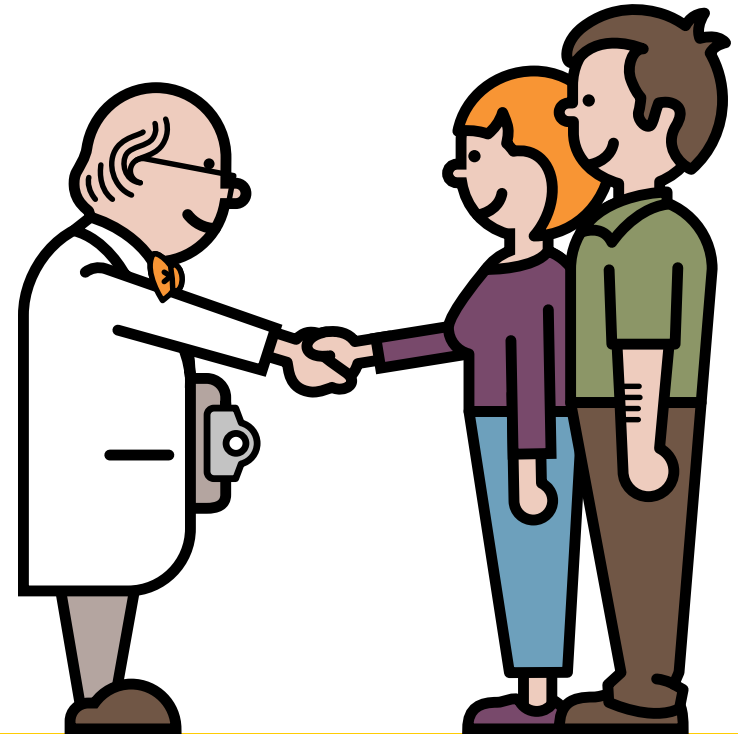
The team are Rachel Skeath, Karen van Wyk, Pat Portnoi and Anita MacDonald. The group is facilitated by Heidi Chan from Nutricia.

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.

MMA

Information for parents following
a new diagnosis



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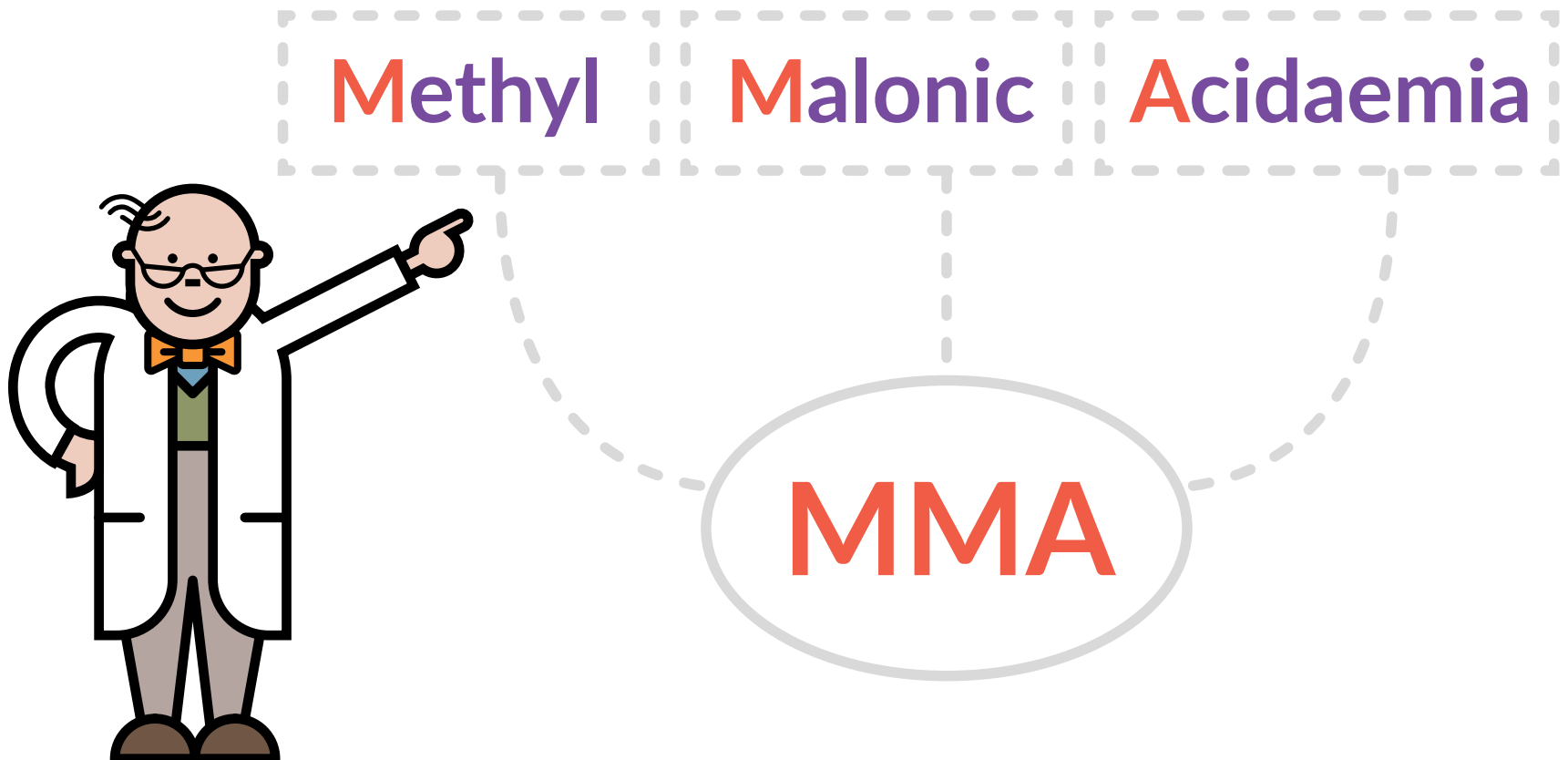
Tools Enabling Metabolic Parents LEarning

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What is MMA?

MMA stands for Methyl Malonic Acidaemia

It is an inherited metabolic condition

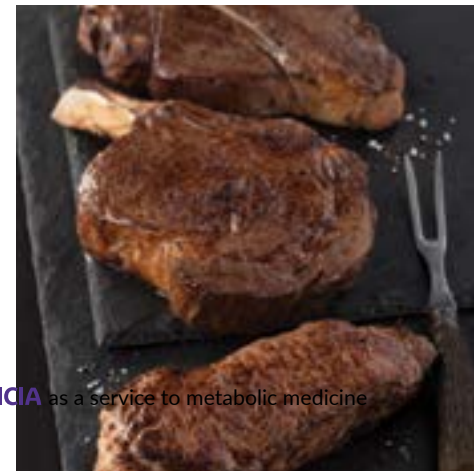


MMA and protein

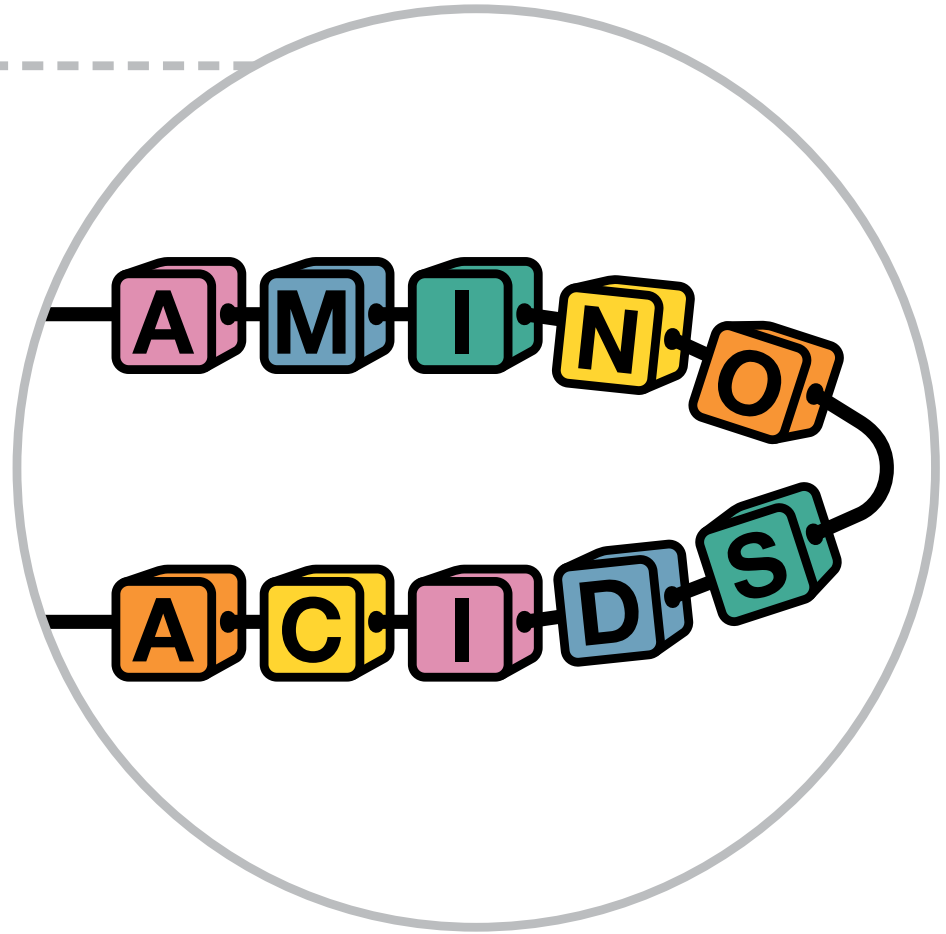
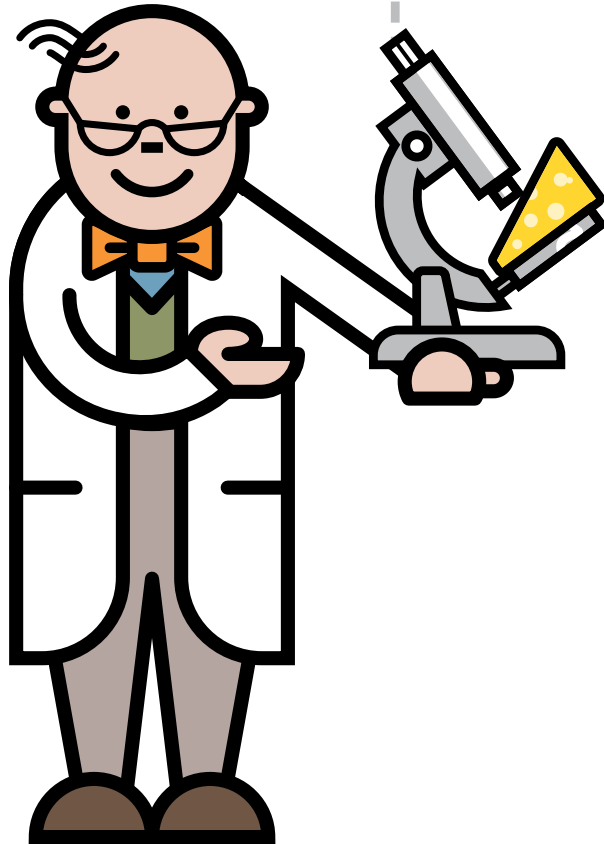
MMA affects the way your baby breaks down protein

Many foods contain protein

The body needs protein for growth and repair



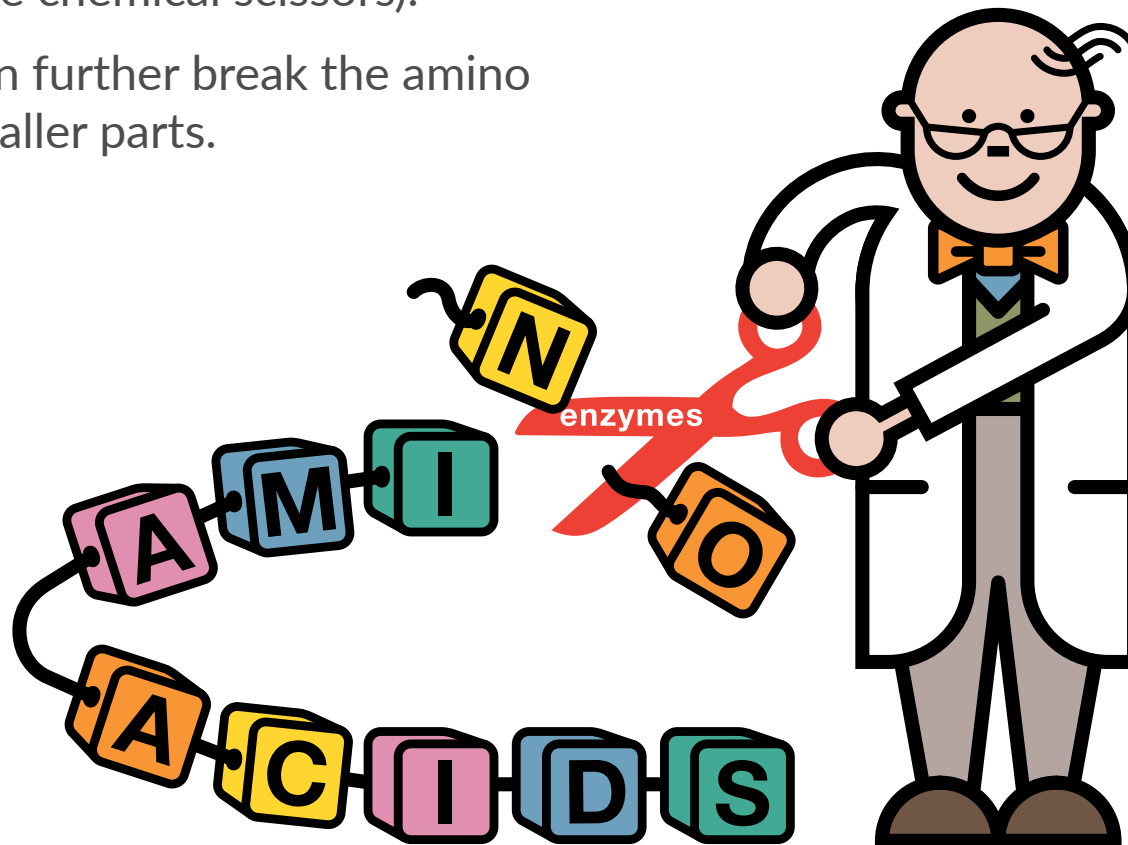
What is protein?



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.

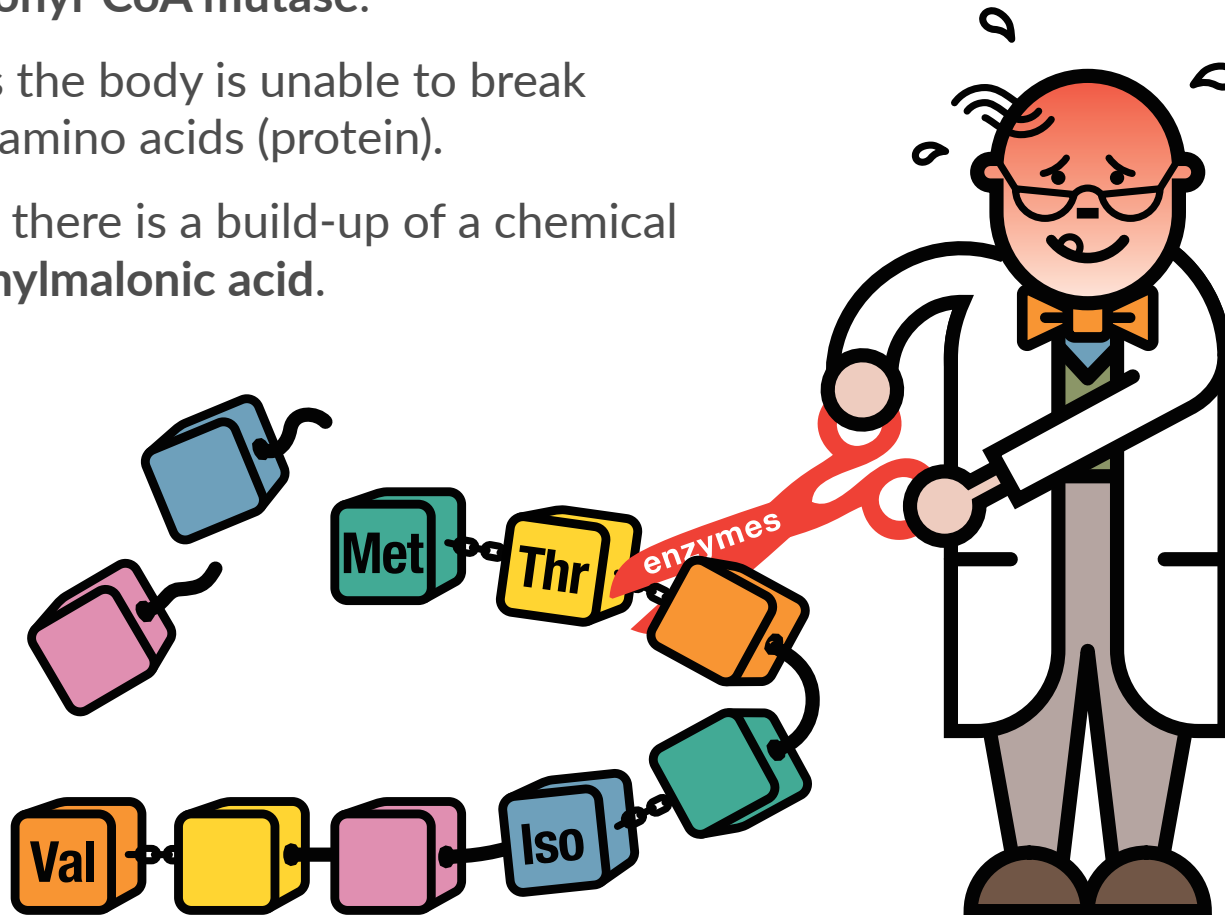


What happens in MMA?

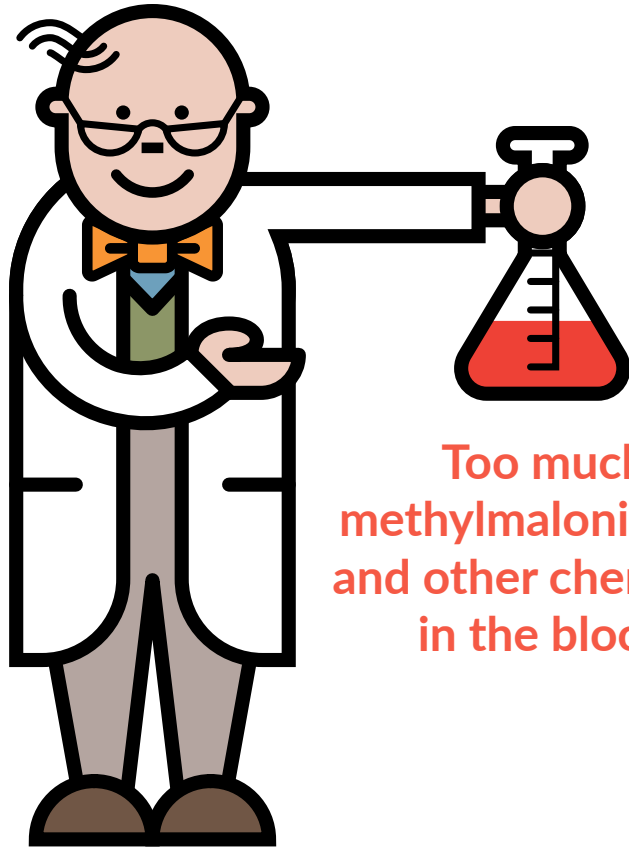
In MMA, the body lacks an enzyme called **methylmalonyl-CoA mutase**.

This means the body is unable to break down four amino acids (protein).

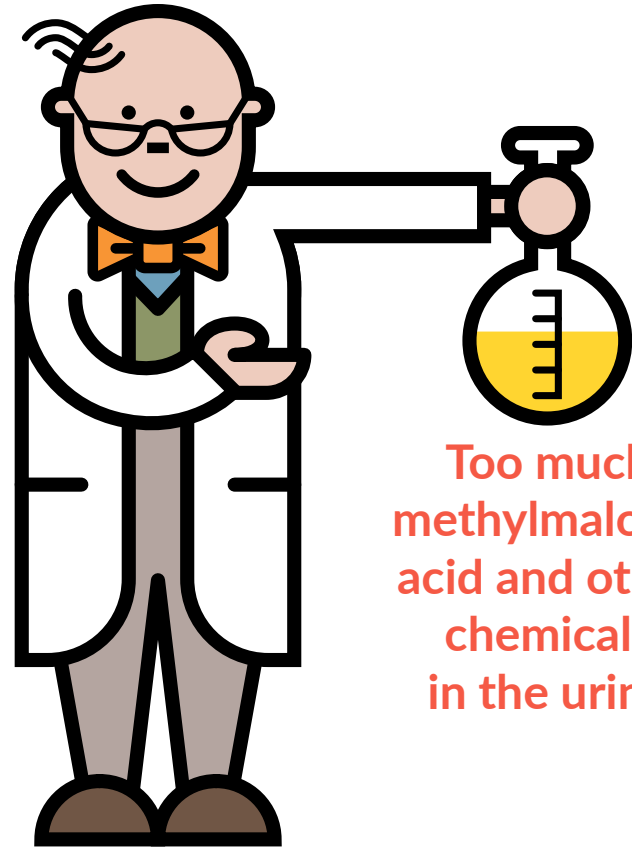
As a result, there is a build-up of a chemical called **methylmalonic acid**.



What does this cause?



Too much
methylmalonic acid
and other chemicals
in the blood

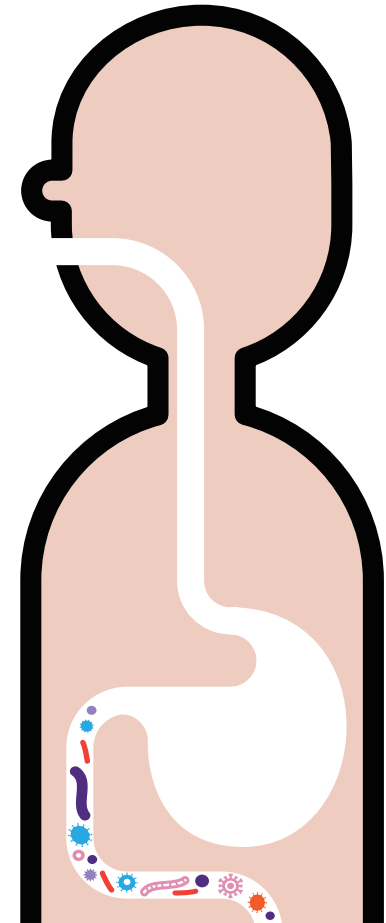


Too much
methylmalonic acid
and other chemicals
in the urine

Other sources of methylmalonic acid

Methylmalonic acid also comes from:

- The breakdown of fatty acids. The body will use these for energy when it has gone a long time without food
- Gut bacteria



How is MMA diagnosed?

MMA is diagnosed by measuring high levels of methylmalonic acid and other chemicals in the blood and urine. It can also be diagnosed by looking at enzyme levels and at the body's genes.



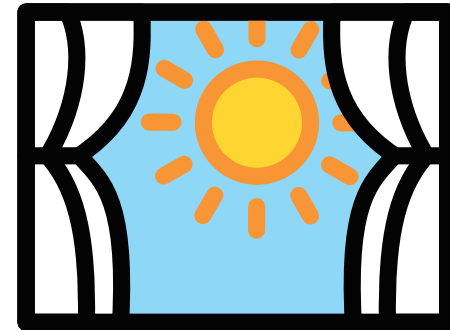
What are the symptoms in MMA?

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

Symptoms include:

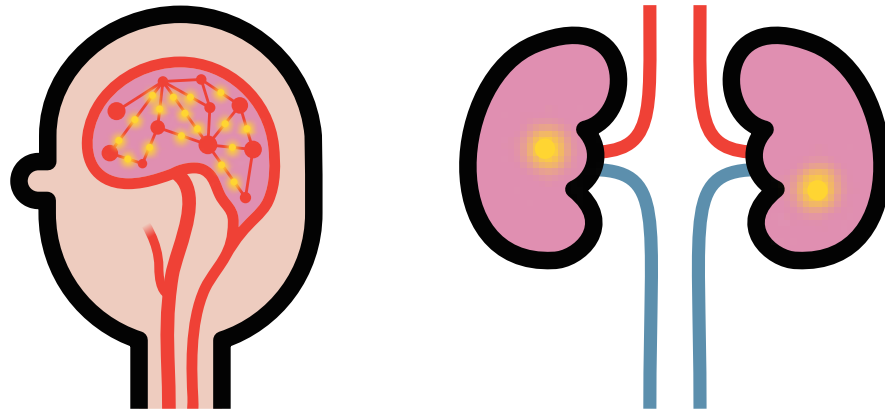
- poor feeding
- vomiting
- dehydration (lack of body fluids)
- floppy baby
- excessively sleepy
- rapid breathing
- seizures

The effects of MMA quickly become life-threatening if unmanaged

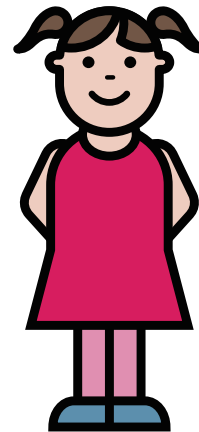


What can go wrong in MMA?

The build up of harmful chemicals can damage the brain and kidneys and cause problems with other organs.



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

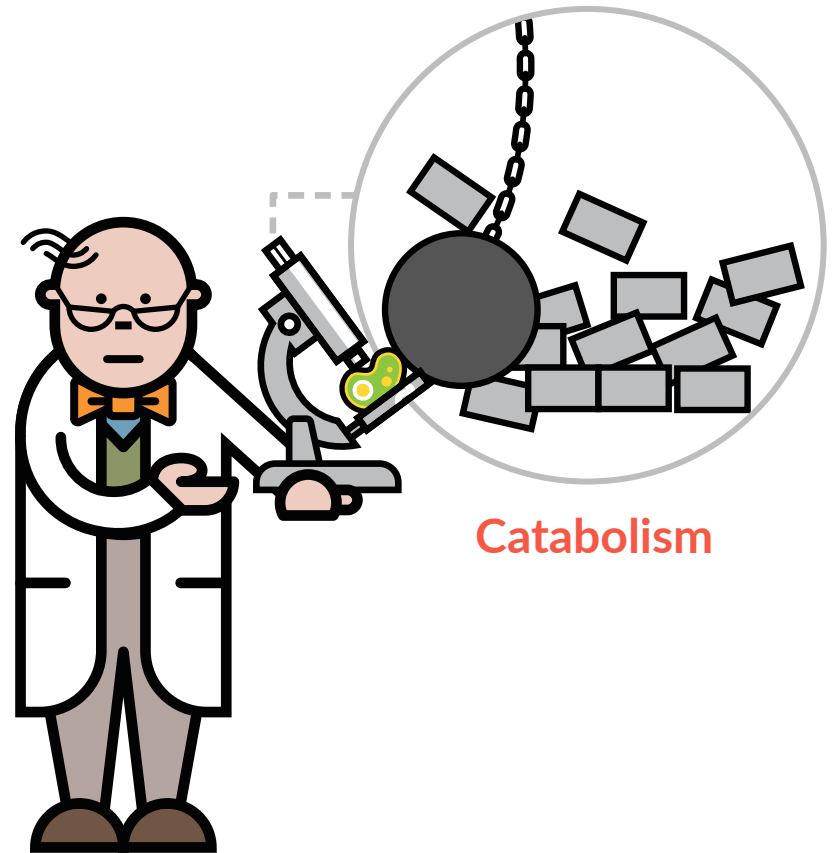


What else happens in MMA?

If the body does not receive enough food

e.g. during illness or the body has gone without food for too long, 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.



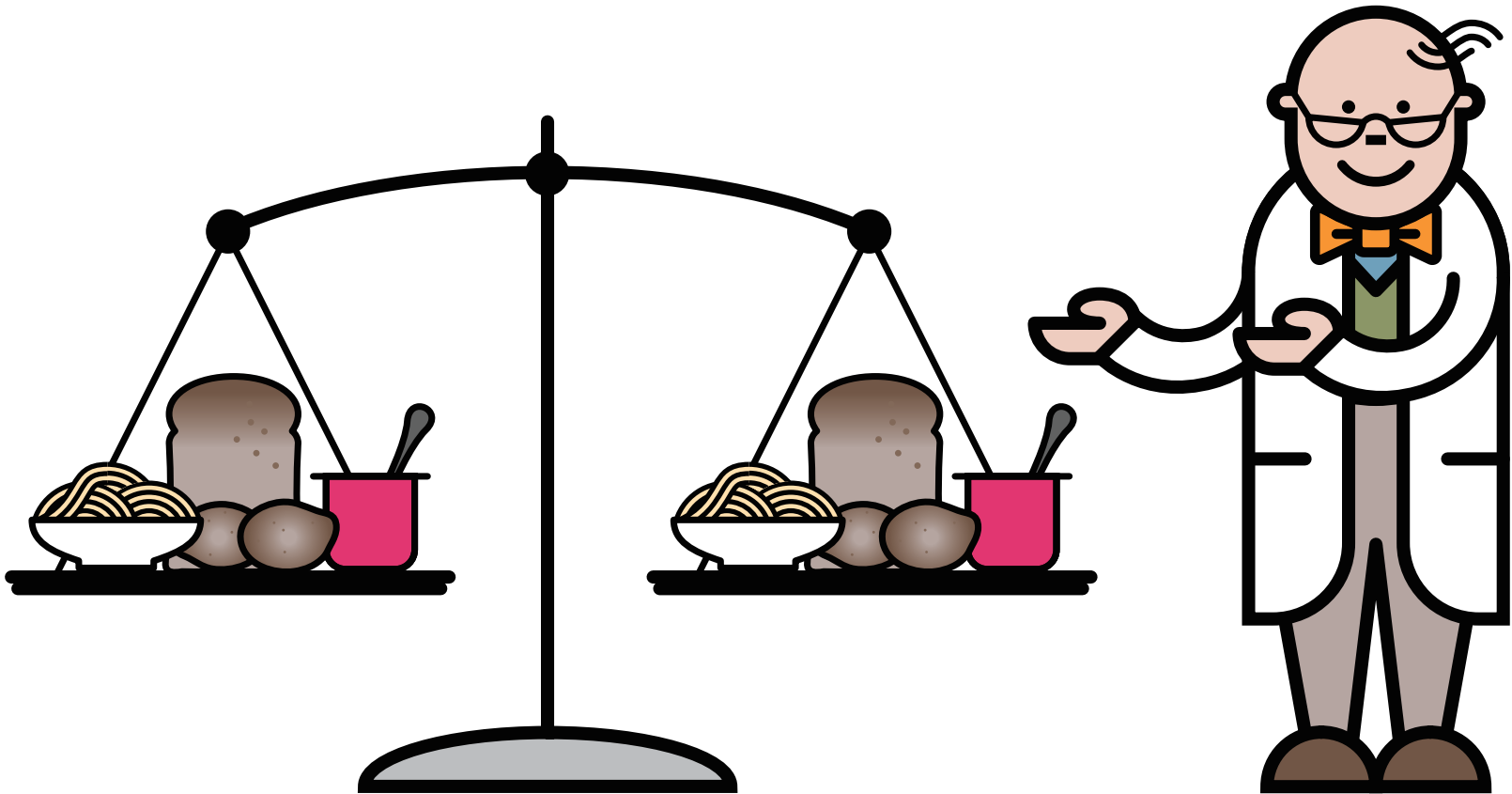
Metabolic crisis

- 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 management
- Avoidance of a metabolic crisis is essential



Protein balance is needed in MMA

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

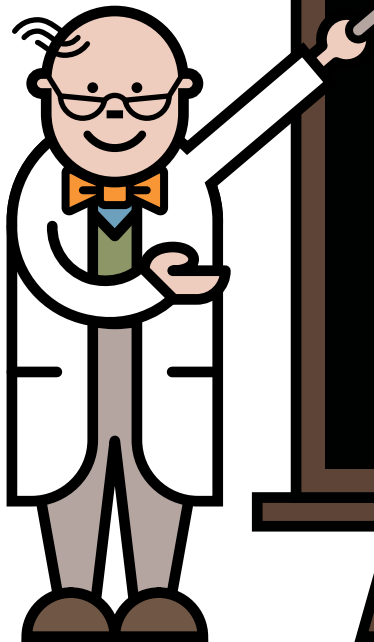


How is MMA managed day to day?

MMA is managed with the following:

A protein restricted diet

Ensuring a sufficient energy supply

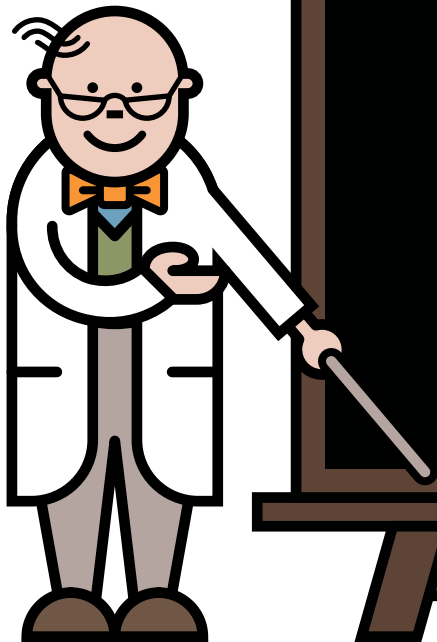


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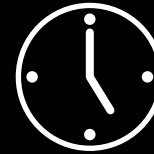


How is MMA managed day to day?

A generous fluid intake



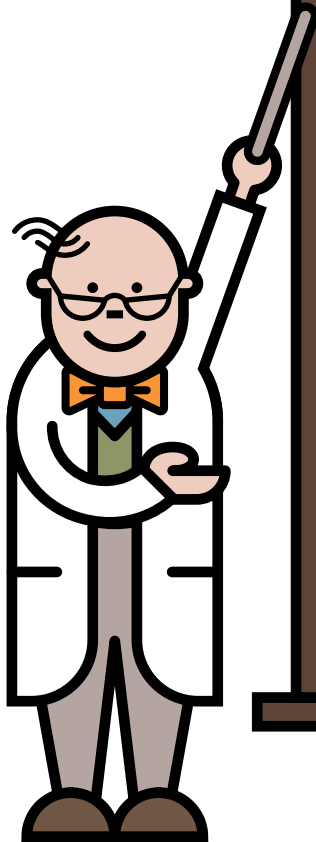
Regular feeding



Carnitine medication



Other medications may be necessary



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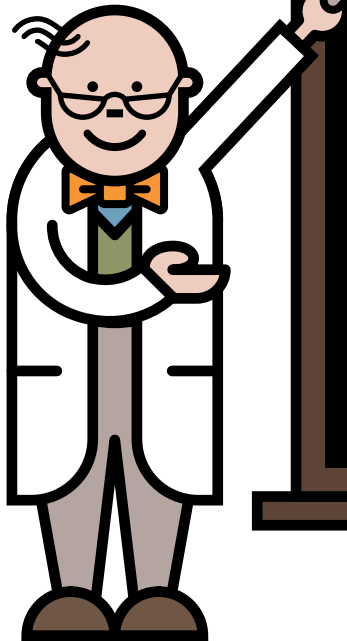
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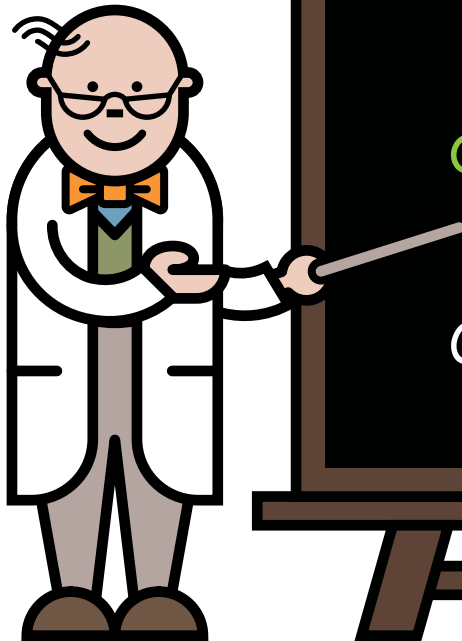
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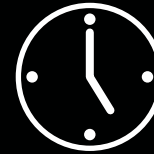


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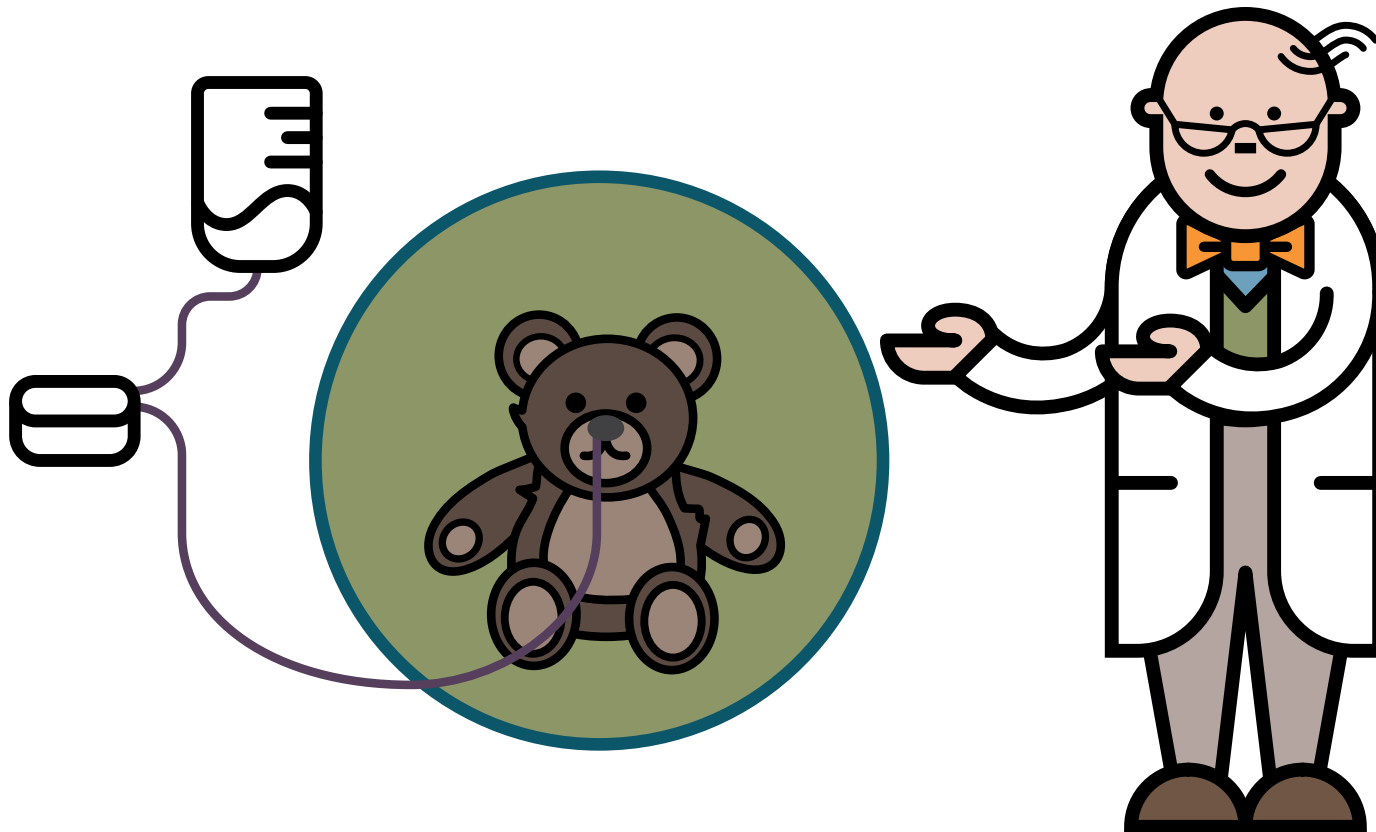


Other medications may be necessary



Is tube feeding needed?

Tube feeding may be necessary to give regular feeds. This will ensure energy, nutrient and fluid needs are met and can help to reduce the production of abnormal chemicals.



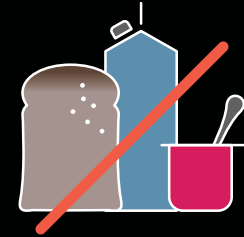
How is MMA managed during illness?

- 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 MMA managed during illness?

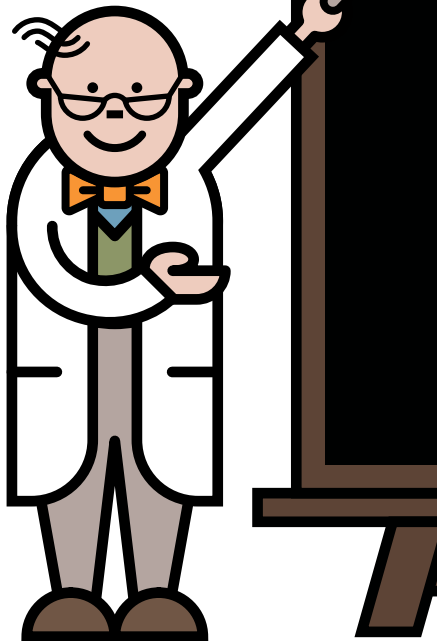
Stop all protein in food & drink



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

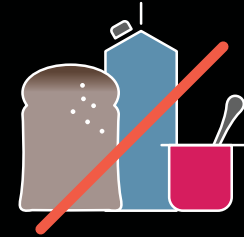


Continue medication as prescribed



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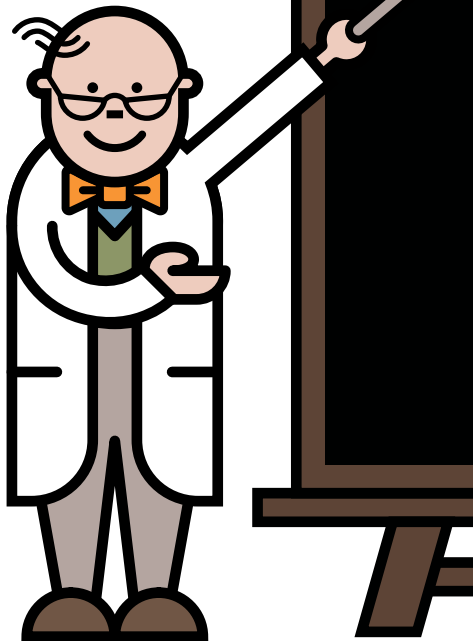
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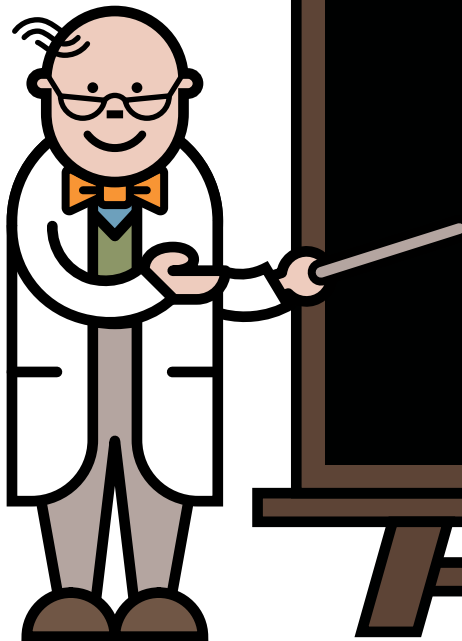
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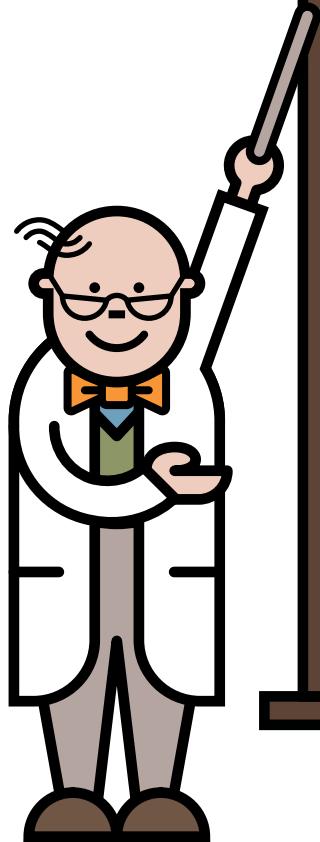
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Continue medication as prescribed



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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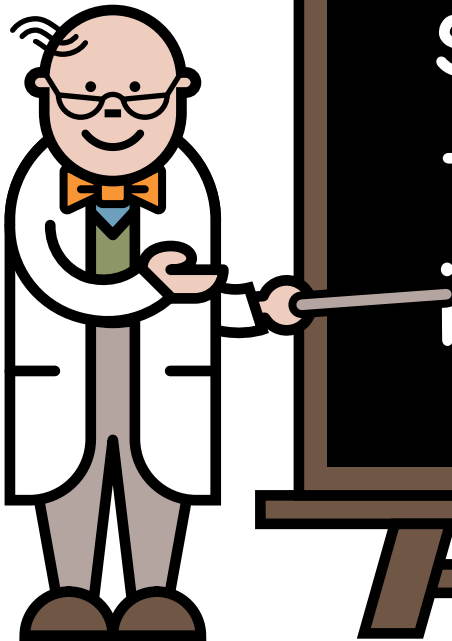


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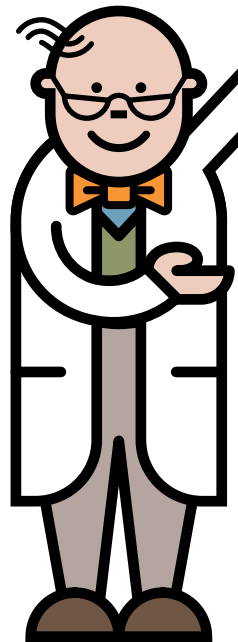


Key message

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



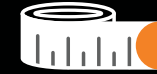
How is MMA monitored?



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



Height and weight

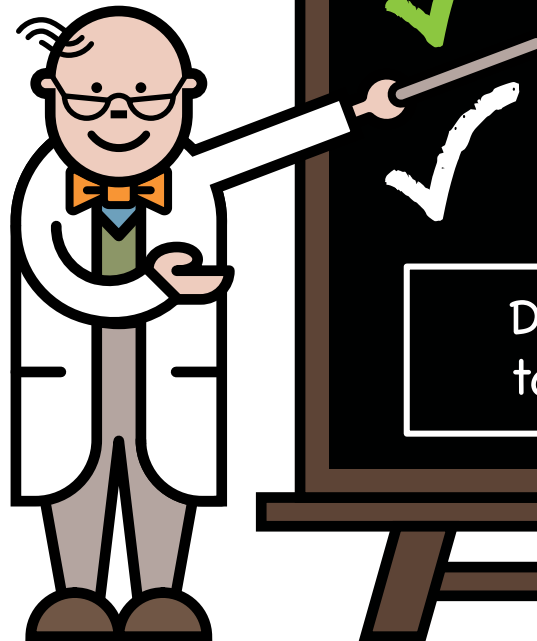


Developmental checks



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

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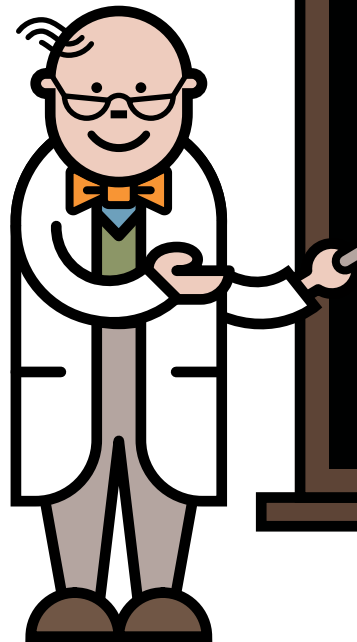


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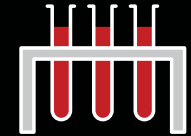


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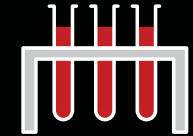
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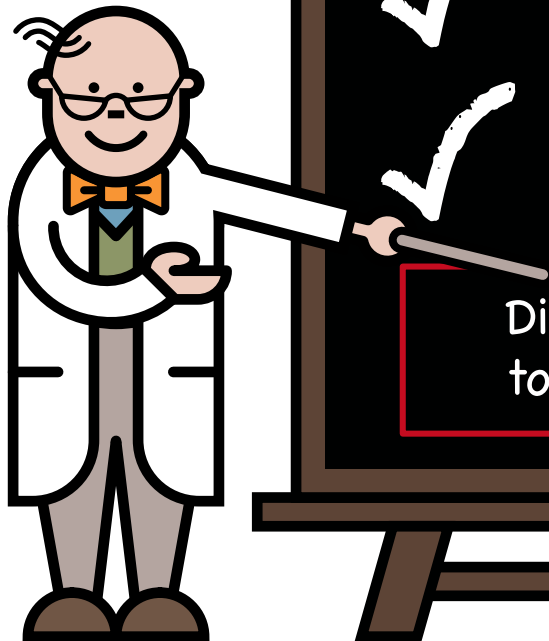
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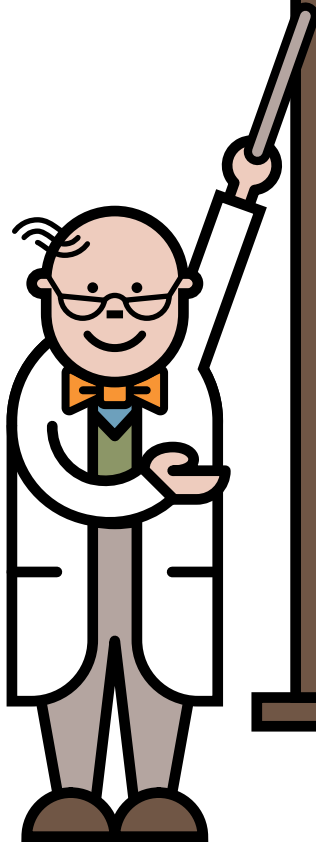
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Chromosomes, genes, mutations



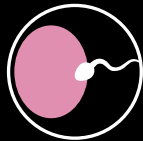
Humans have chromosomes composed of DNA



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

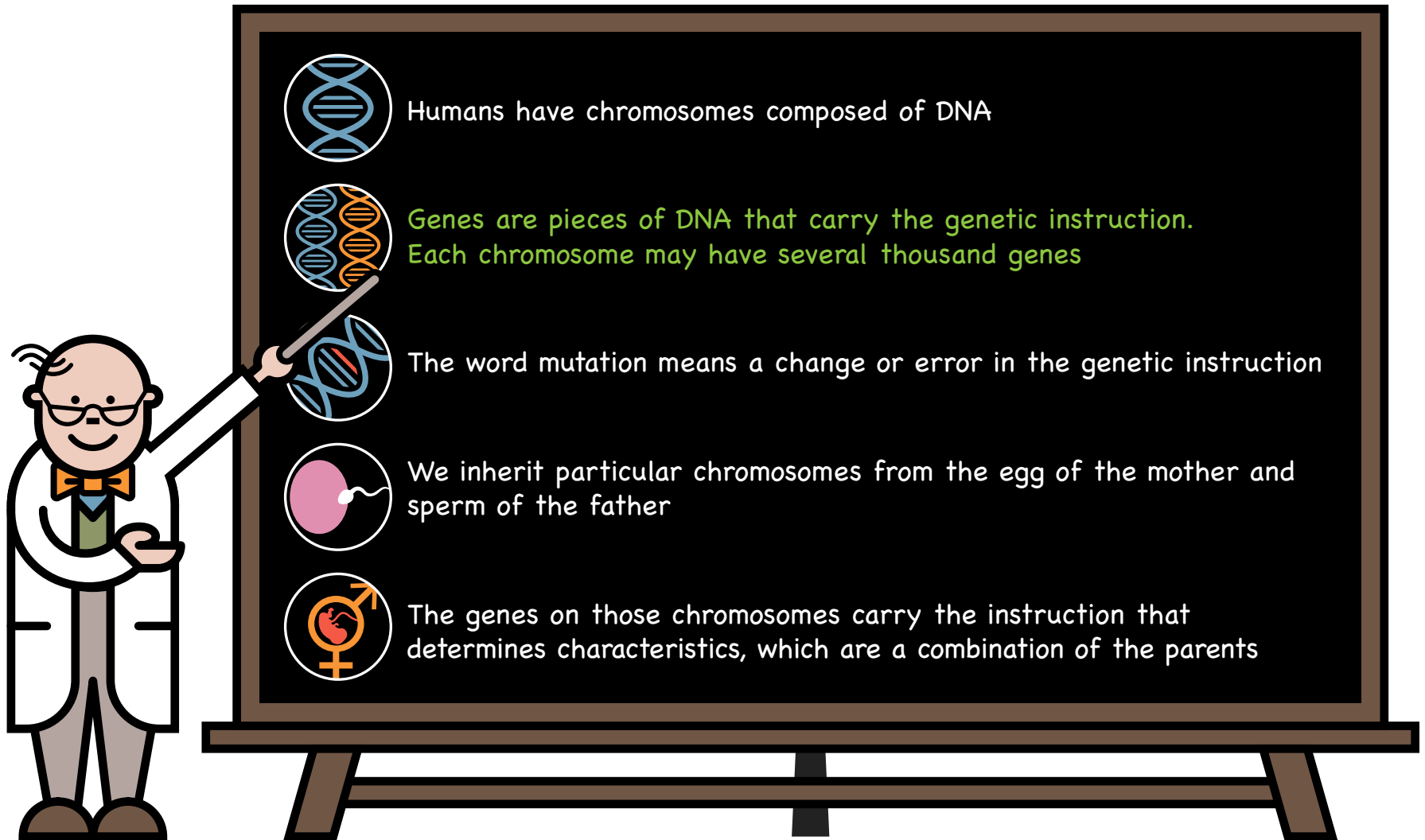



We inherit particular chromosomes from the egg of the mother and sperm of the father





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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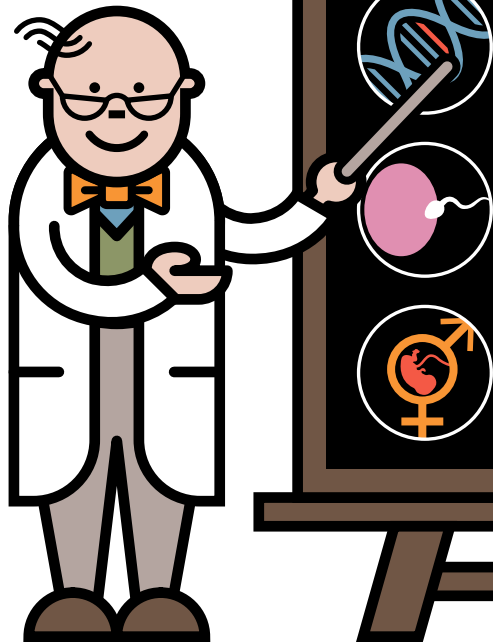
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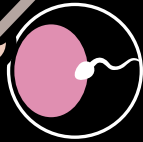
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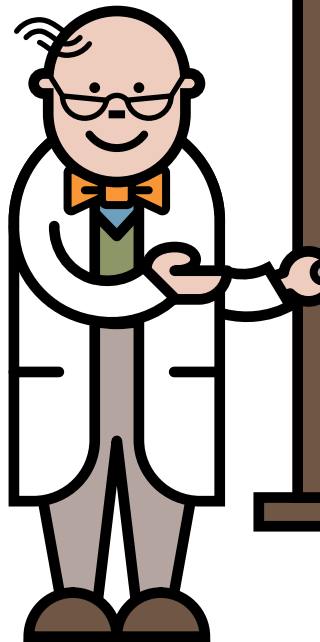


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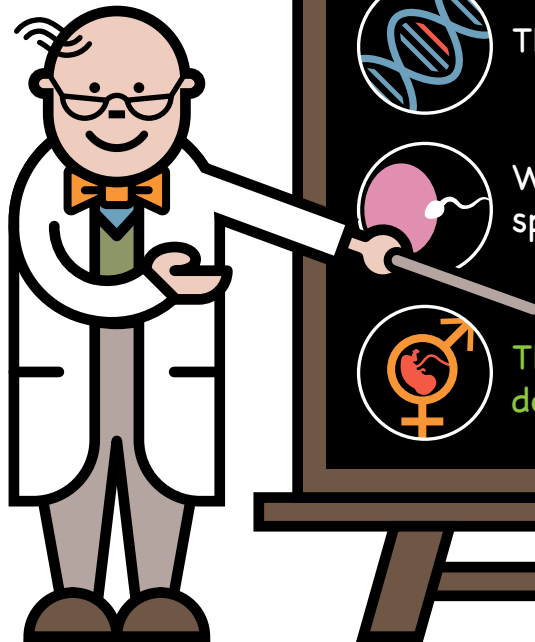


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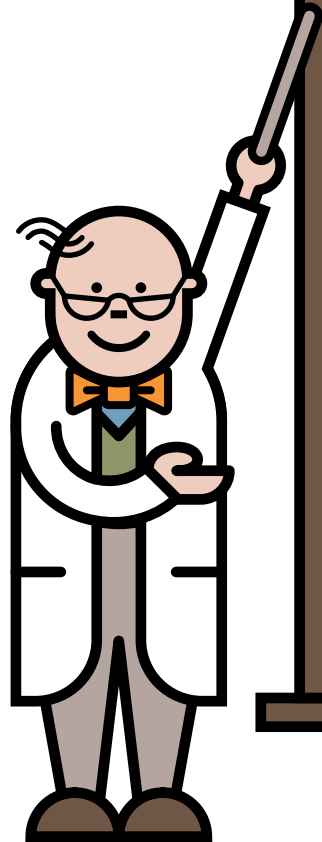


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Inheritance



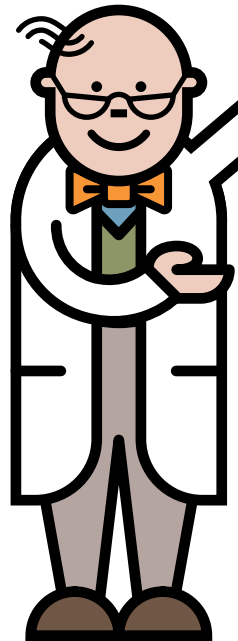
MMA is an inherited condition. There is nothing that could have been done to prevent your baby from having MMA

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

Parents of children with MMA are carriers of the condition

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

Inheritance



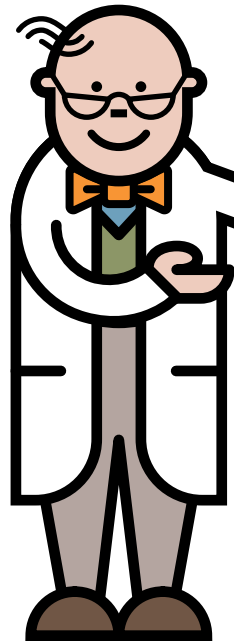
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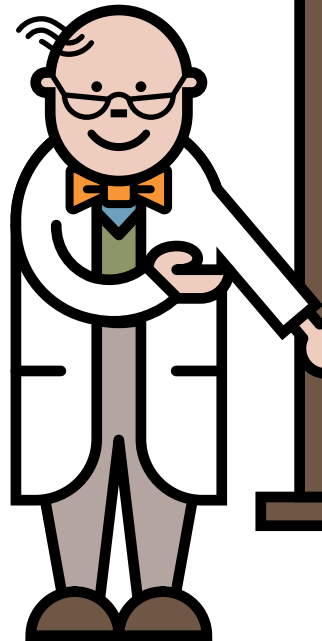
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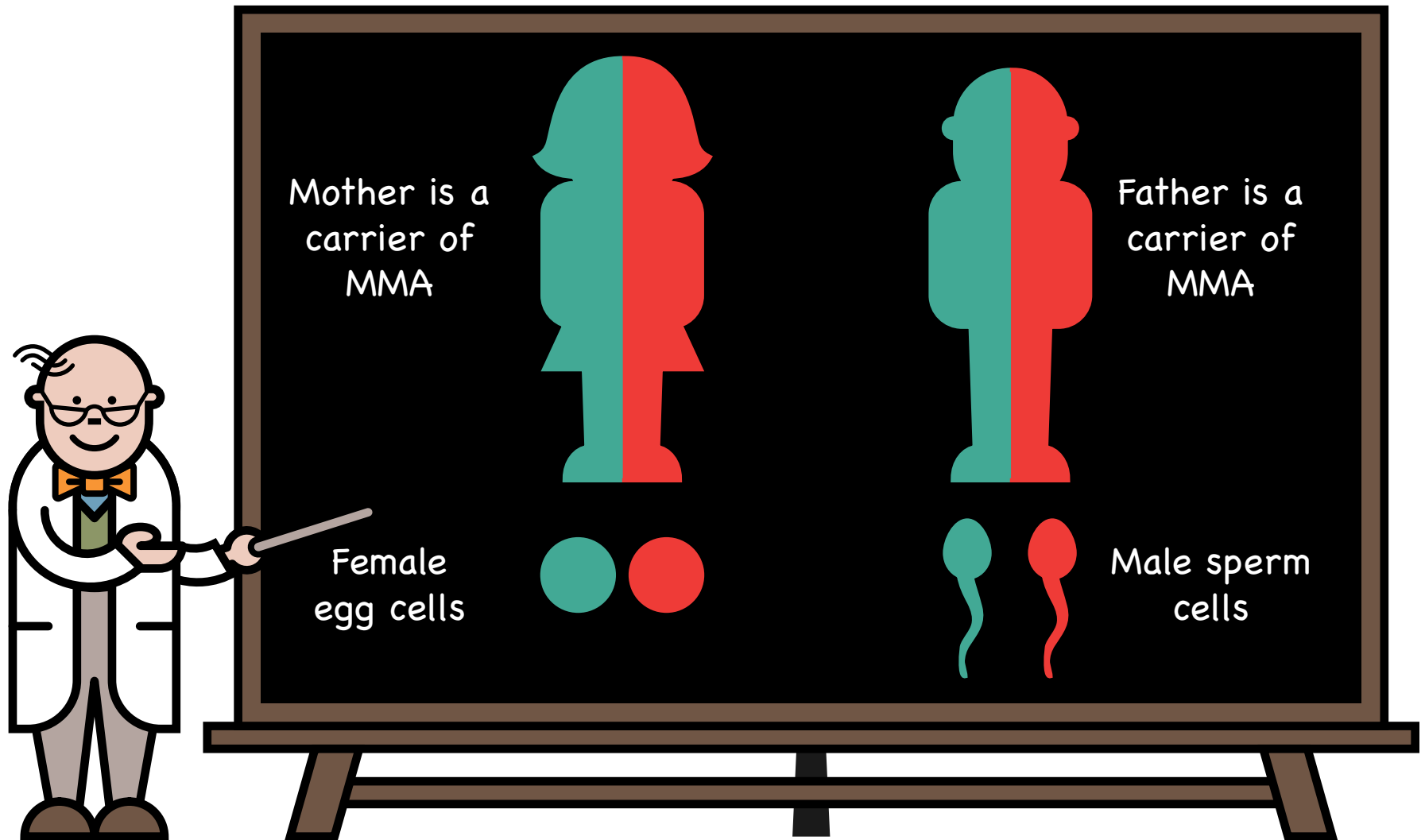
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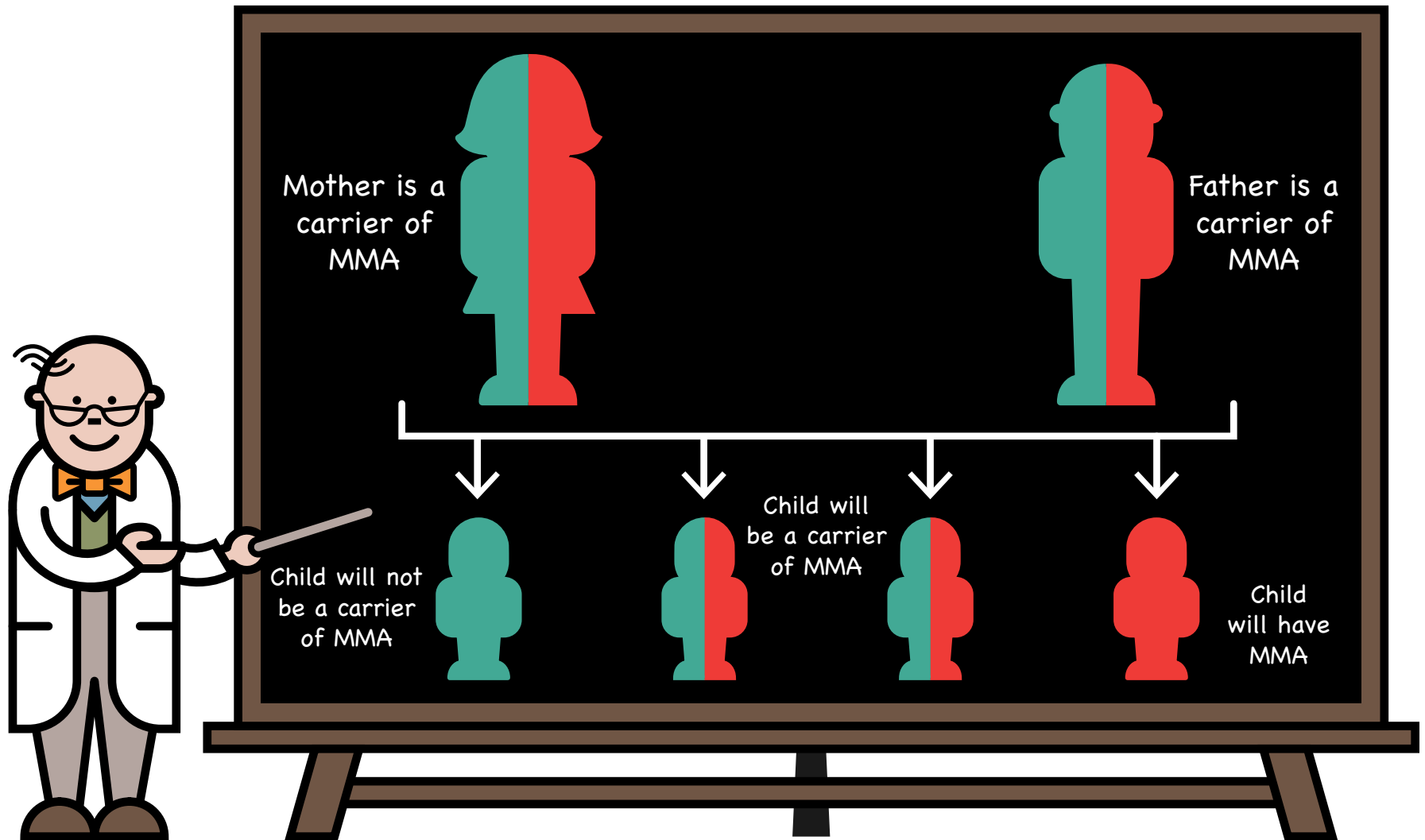
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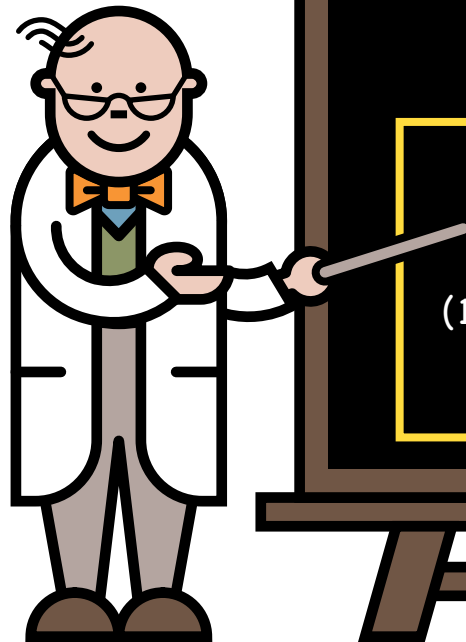
Autosomal recessive (carriers of MMA)



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 MMA

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

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

Take home messages



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

The condition is managed with a protein restricted diet, regular feeding, a generous fluid intake and medications

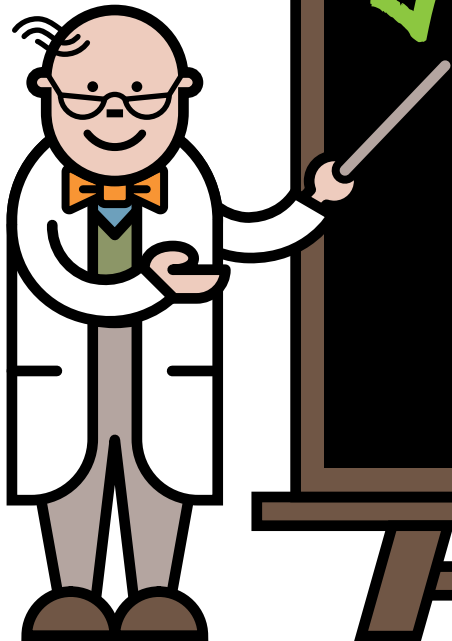
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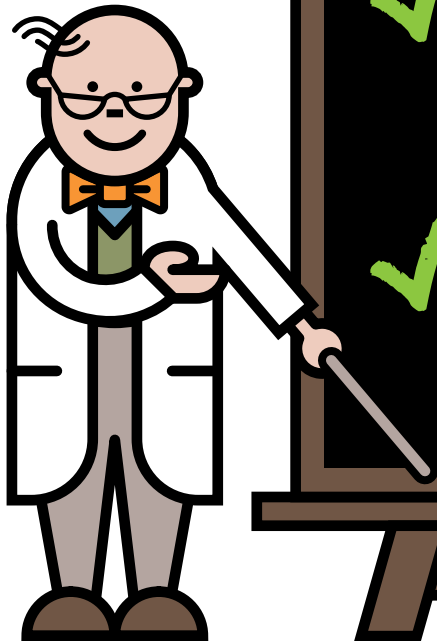


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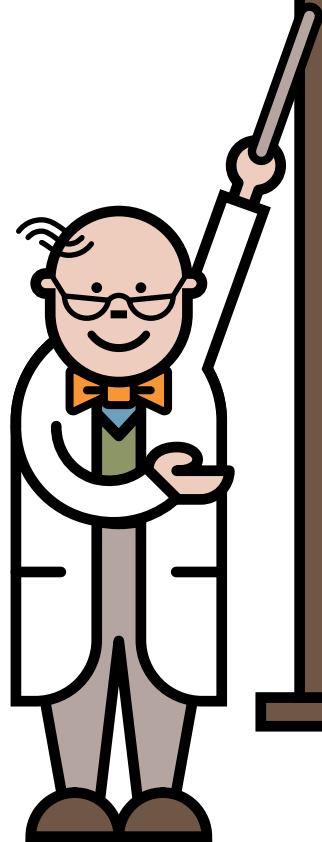
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Helpful hints



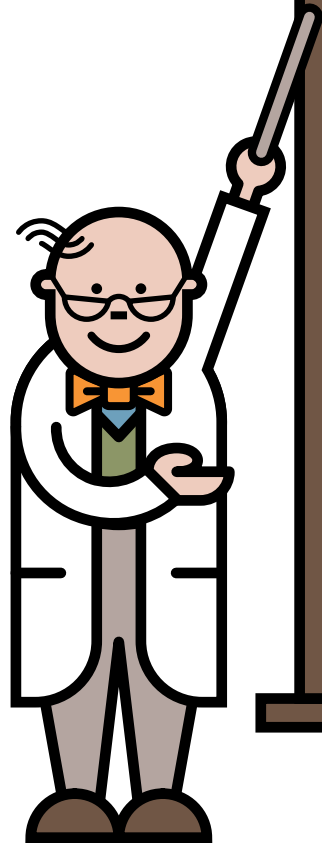
Always ensure you have a good supply of your dietary products and medicines and that they are in date

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

Helpful hints



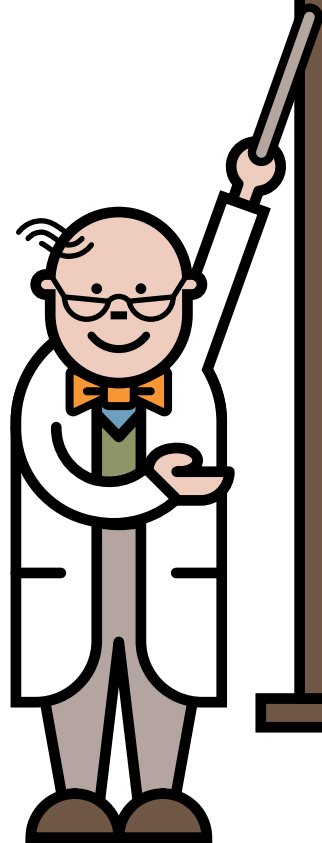
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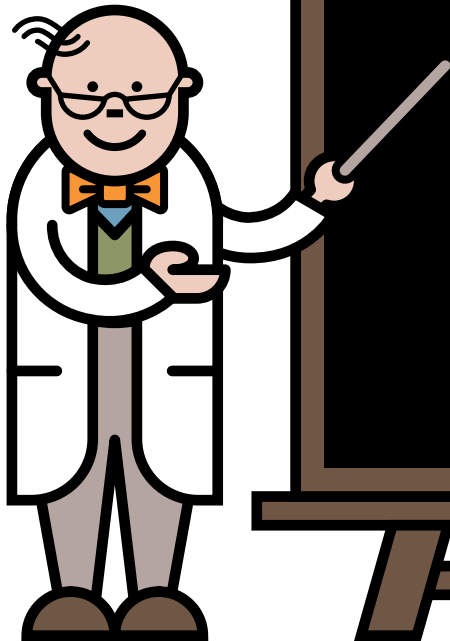
Always ensure you have a good supply of your dietary products and medicines and that they are in date



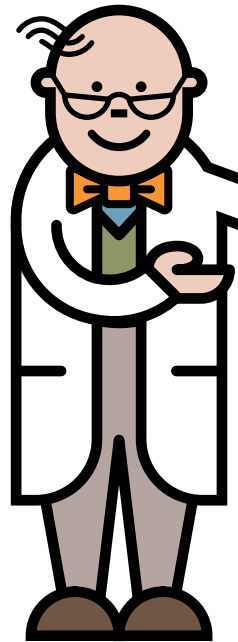
Your dietary products and medications are prescribed. These are obtained via a pharmacy or home delivery

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

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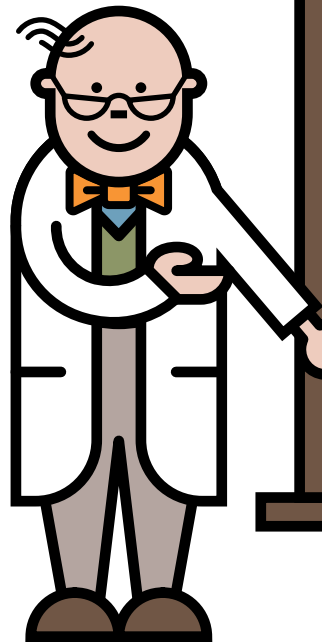
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Who's who

- 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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