

An introduction to the

Organic Acidaemia: Methylmalonic Acidaemia (MMA)

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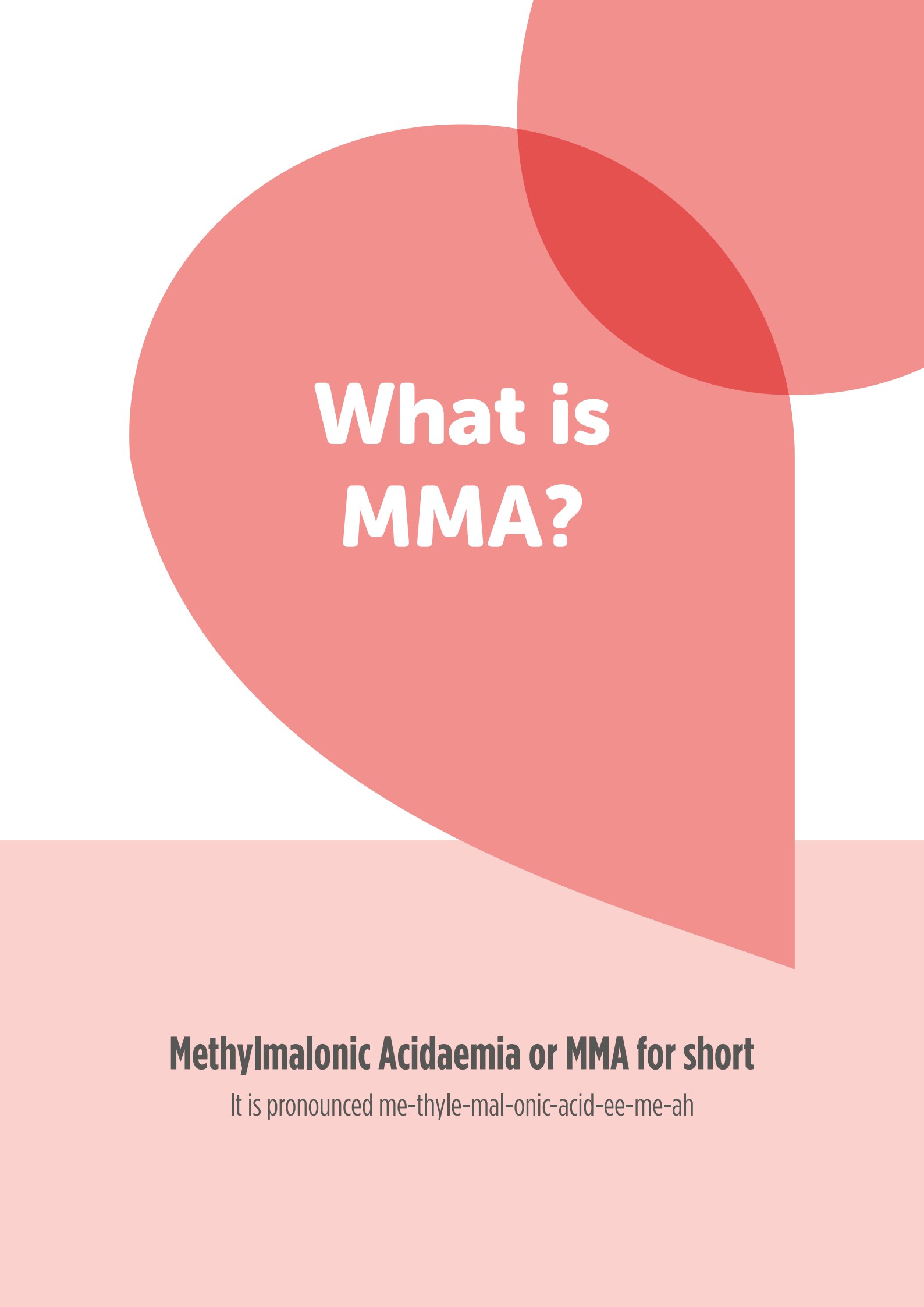
This is an introduction to the Organic Acidaemia; methylmalonic acidaemia (MMA) following the diagnosis of your child with this condition.

Consult your metabolic team for diagnosis and treatment for MMA.

This booklet is not designed to replace any advice given by the metabolic team at the hospital.

If you are unclear about anything written in this booklet, please ask your dietitian for further information.

This booklet was written with Prof Anita MacDonald who is a metabolic dietitian from Birmingham Children's Hospital, who has over 40 years of experience working with parents/carers and children with MMA.



What is MMA?

Methylmalonic Acidaemia or MMA for short

It is pronounced me-thyle-mal-onic-acid-ee-me-ah

MMA is an inherited metabolic condition which affects the way the body breaks down protein

If left untreated, chemicals in the blood build up to toxic levels.

This can cause several problems:

- It can damage the brain and kidneys.
- It may cause delays in normal development like walking and talking.

Other symptoms:

- Poor feeding
- Vomiting
- Dehydration
- Floppy infant
- Excessively sleepy
- Rapid breathing
- Seizures

MMA can be life threatening if left unmanaged.

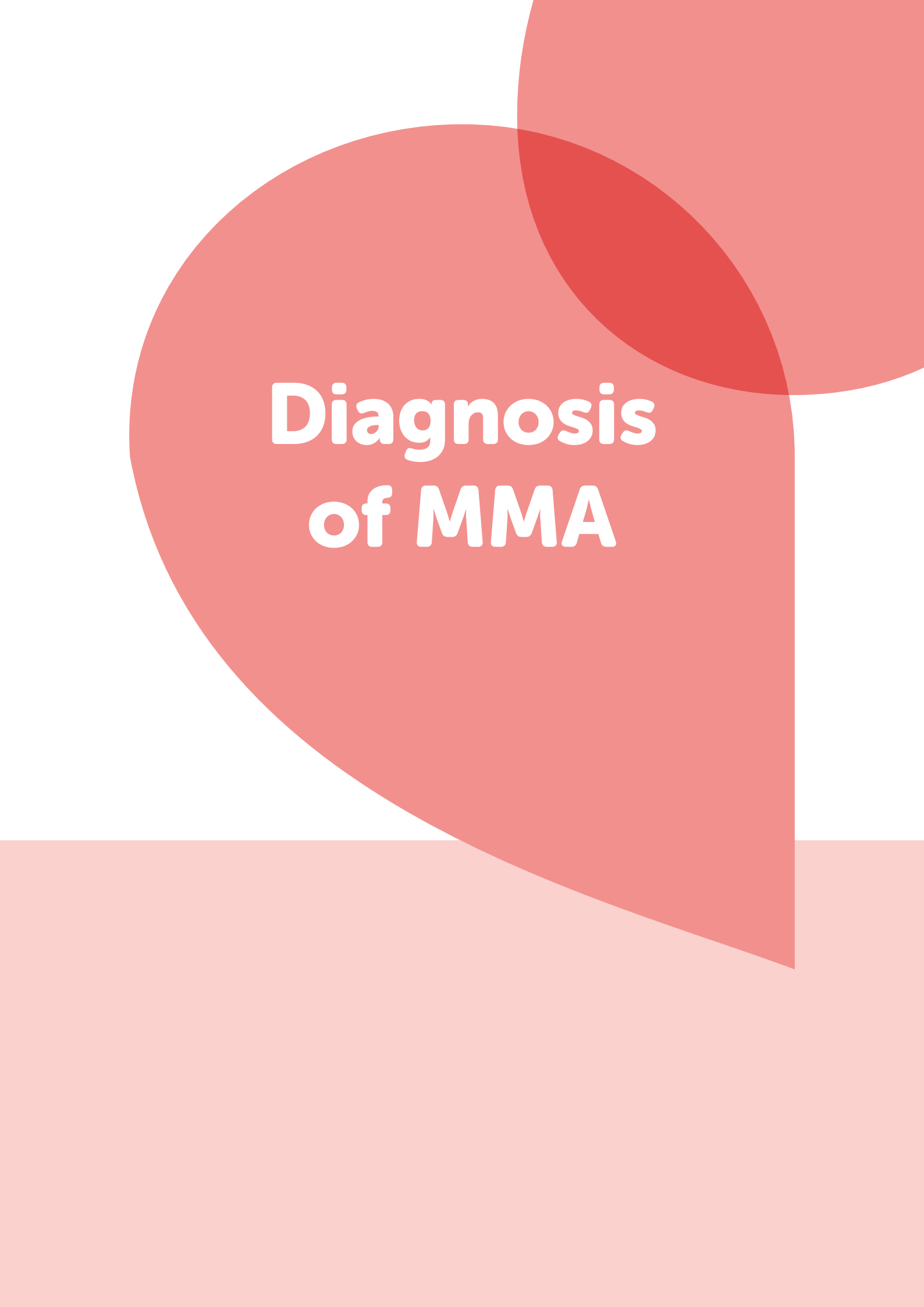
There are 2 main forms of MMA:

B12 responsive MMA - This can be managed by injections of vitamin B12 and may not need to follow a special diet.

B12 non-responsive MMA - This form does not respond to large doses of B12 and is managed by following a protein restricted diet alongside medication.

Your child has been diagnosed with B12 non-responsive MMA .

This booklet will explain the management of B12 non-responsive MMA.



Diagnosis of MMA

MMA is identified by blood and urine tests

High levels of a chemical called methylmalonic acid and other chemicals are found in the blood and urine.

Once diagnosed a child will be referred to a specialist metabolic centre to ensure they receive the care they need.

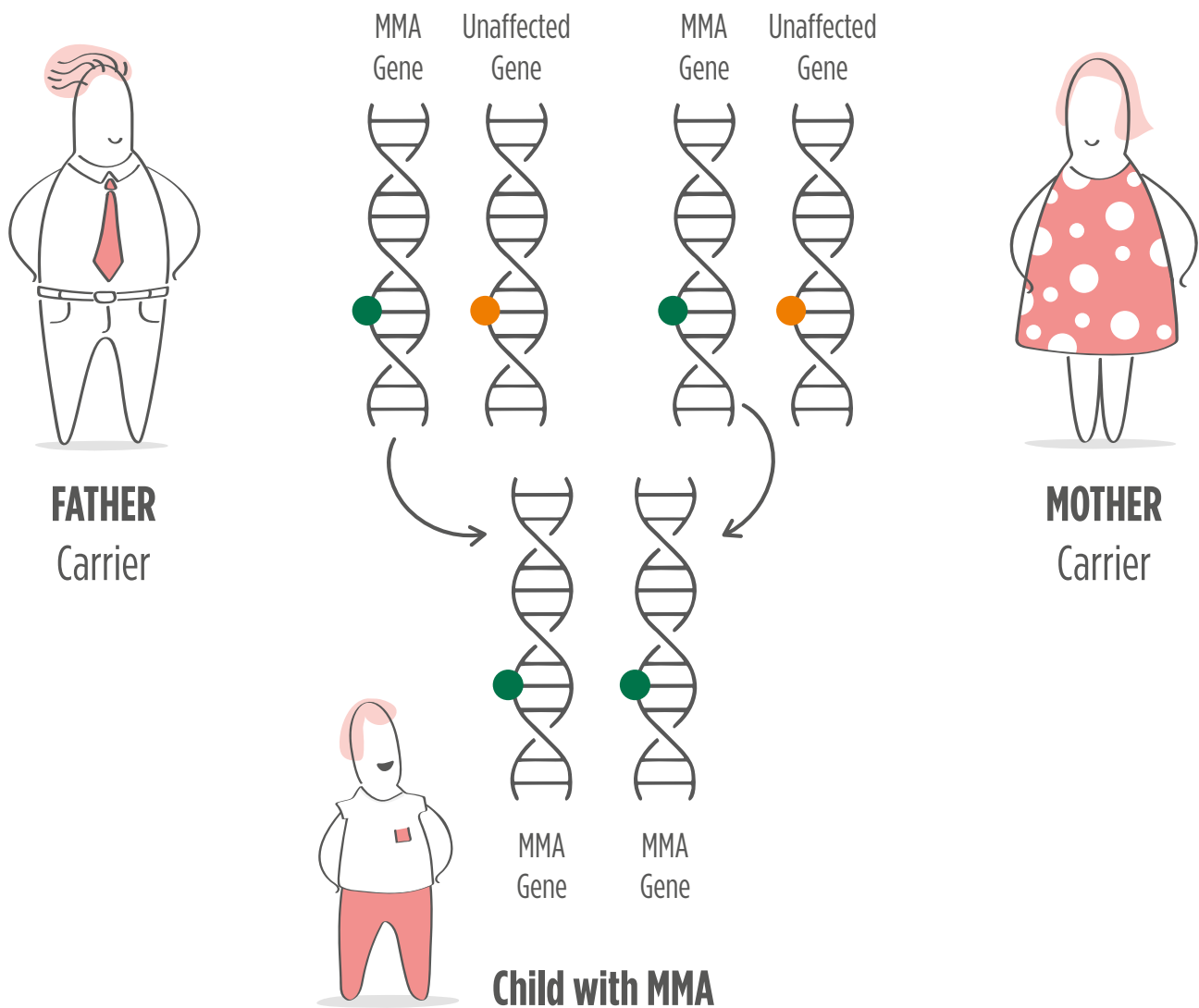
In newborns diagnosed with MMA, if managed early and effectively they can have a good quality of life



**Why does
my child
have MMA?**

MMA is an inherited genetic condition

It is nobody's fault and there is nothing you could have done to prevent it.

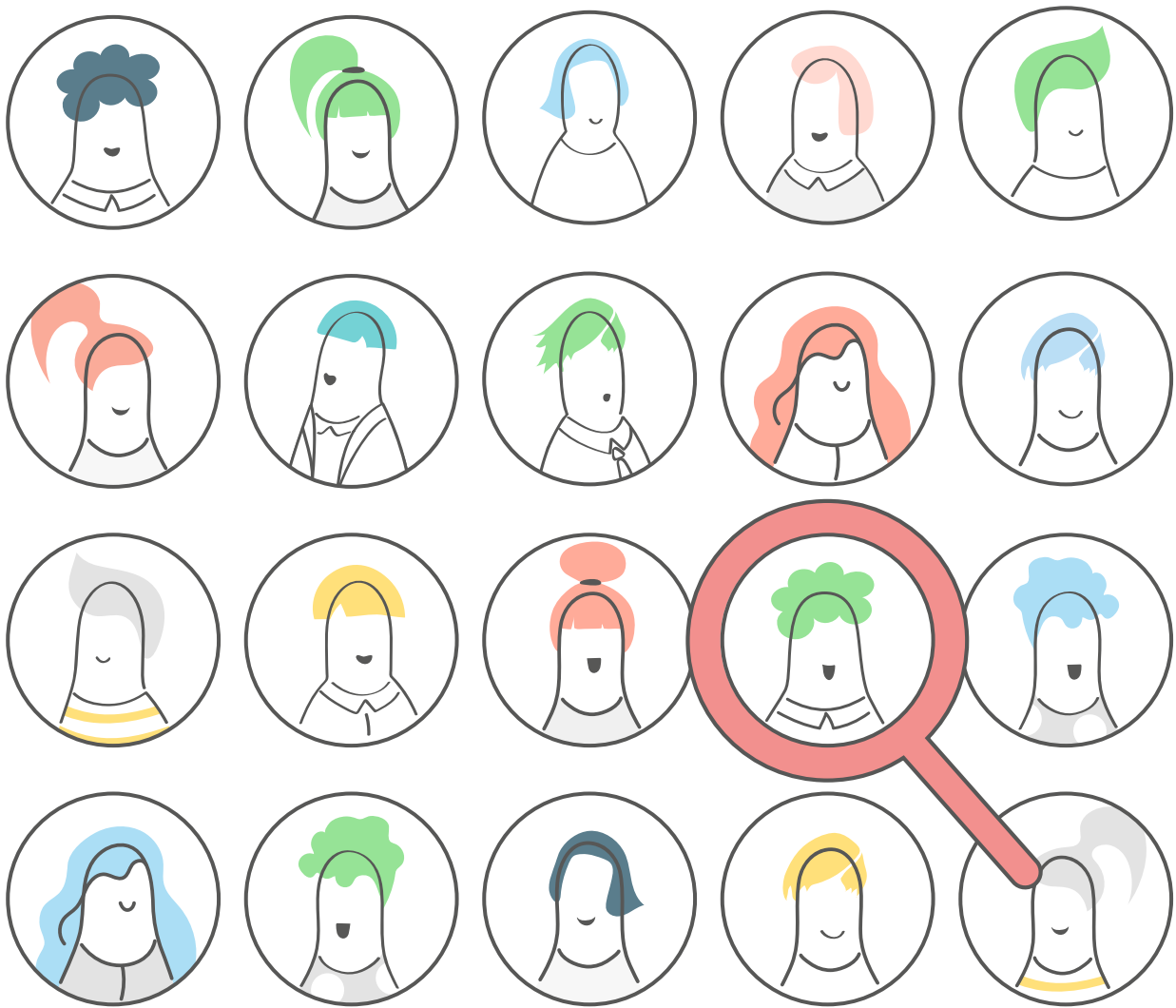


As a parent of a child with MMA, you have one MMA gene and one unaffected gene.
This is known as being a carrier.

For a child to inherit MMA, it requires both parents to be carriers of the MMA gene.

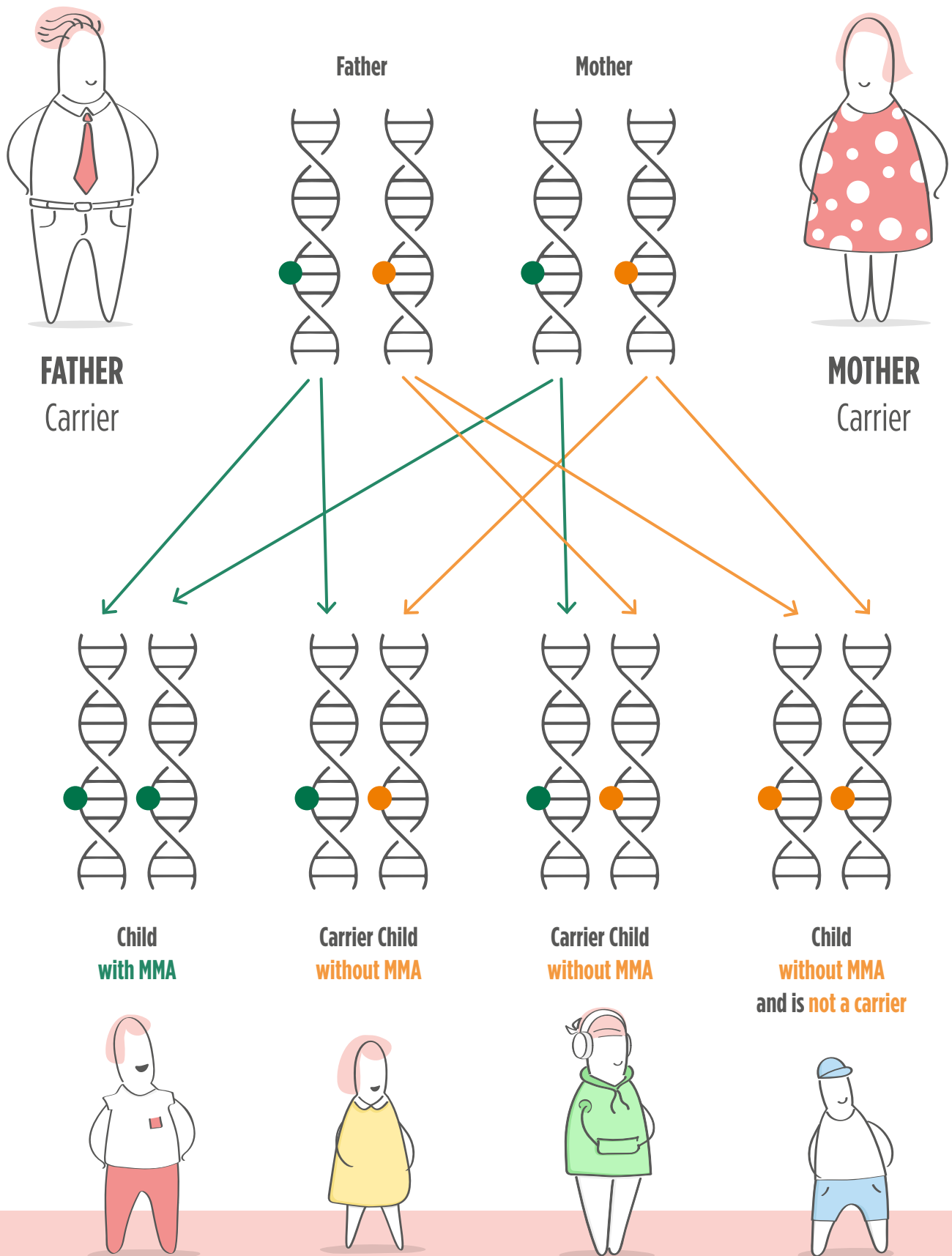
Your child has inherited 2 MMA genes, one from the father and one from the mother and so therefore has MMA.

There are a small number of people who are carriers of the MMA gene



People who are carriers for MMA do not have MMA themselves and in fact do not normally know they are carriers. The MMA gene does not cause a problem to them.

When 2 people who are carriers of the MMA gene conceive a child there is a 1 in 4 chance of that child having MMA.



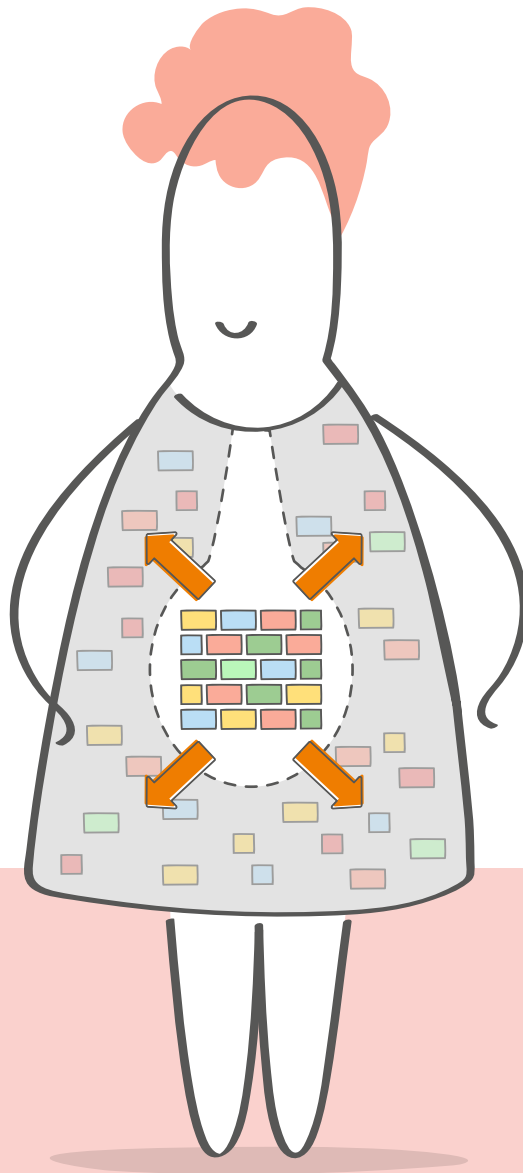


**The science
bit... protein
explained**

Why do we need protein?

Protein is one of the nutrients needed by the body.

It helps to build, repair and maintain body cells and tissues, like your skin, muscles, organs, blood and even bones.

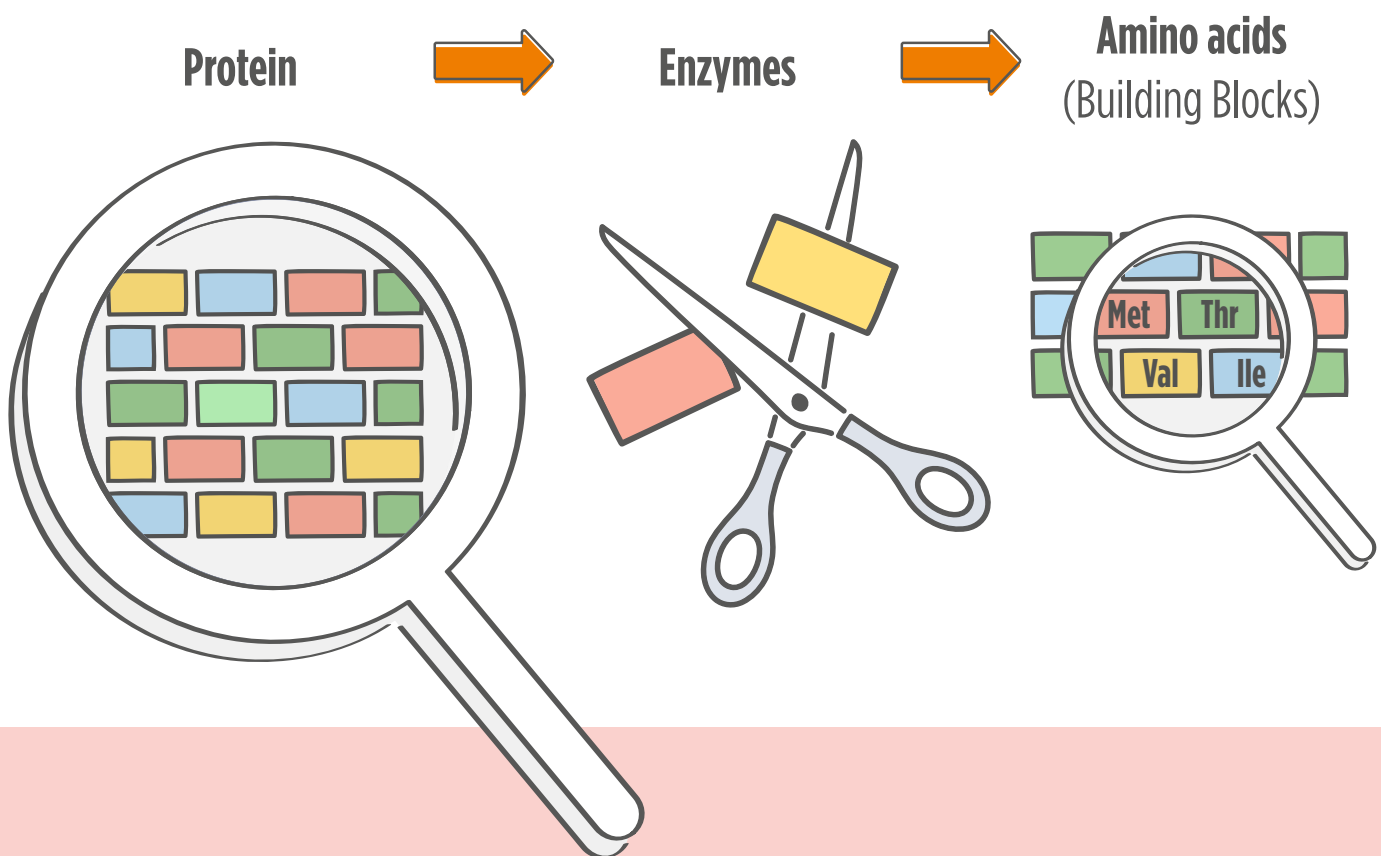


Protein is made up of different building blocks called amino acids.

When protein is eaten it is broken down into these amino acids so they can be absorbed into the blood stream and travel to where they are needed.

The conversion of protein into amino acids

Enzymes are special proteins which act like chemical scissors needed for the breakdown of protein into amino acids.
There are 20 amino acids (building blocks) that make up protein.

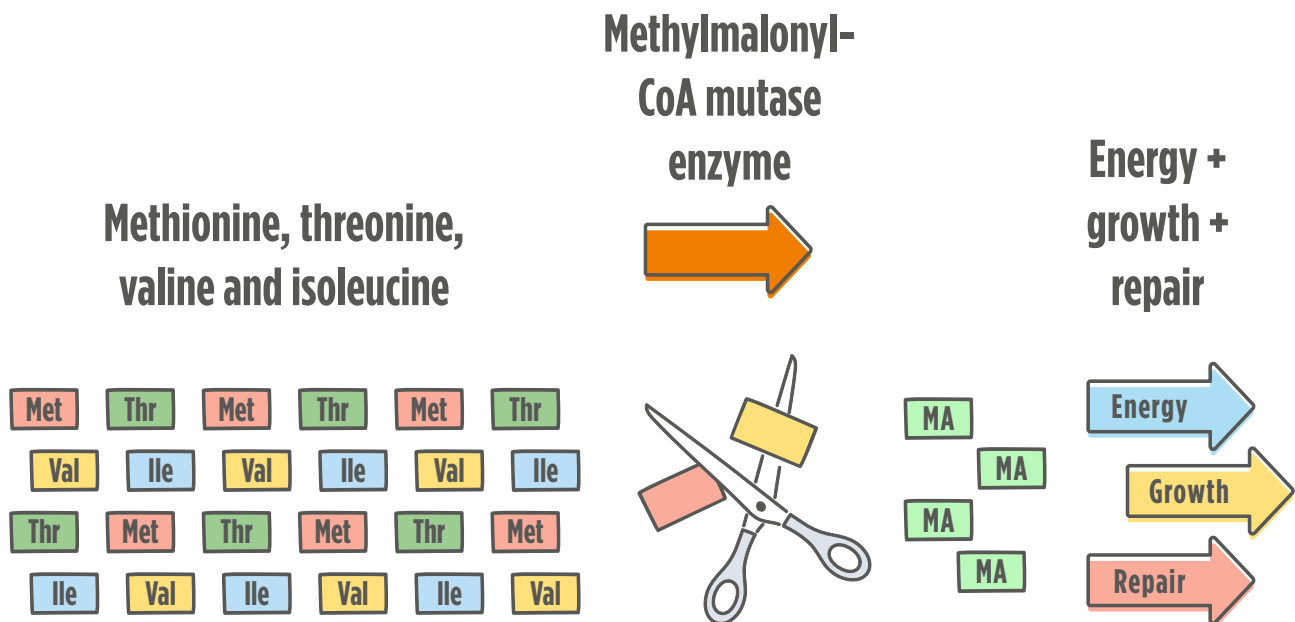


Methionine, threonine, valine and isoleucine are 4 of these amino acids and are used by the body to make new chemicals.

It is these four amino acids that a person with MMA cannot process correctly.

Why can someone with MMA not process methionine, threonine, valine and isoleucine

A person without MMA



Some of the methionine, threonine, valine and isoleucine are broken down further to make methylmalonic acid and other chemicals.

Normally the liver produces an enzyme called **methylmalonyl-CoA mutase** which is used to convert methylmalonic acid to energy, which is used for growth and repair.

A person with MMA

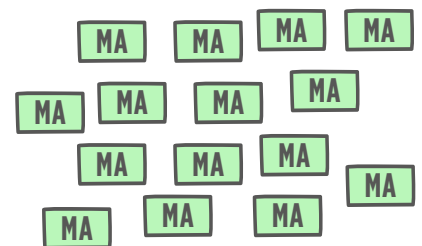
Faulty/no
methylmalonyl-
CoA mutase enzyme



Methionine, threonine,
valine and isoleucine



Buildup of
methylmalonic acid
and other harmful
chemicals



In MMA the methylmalonyl-CoA mutase enzyme
is either not made by the liver or it does not work properly.

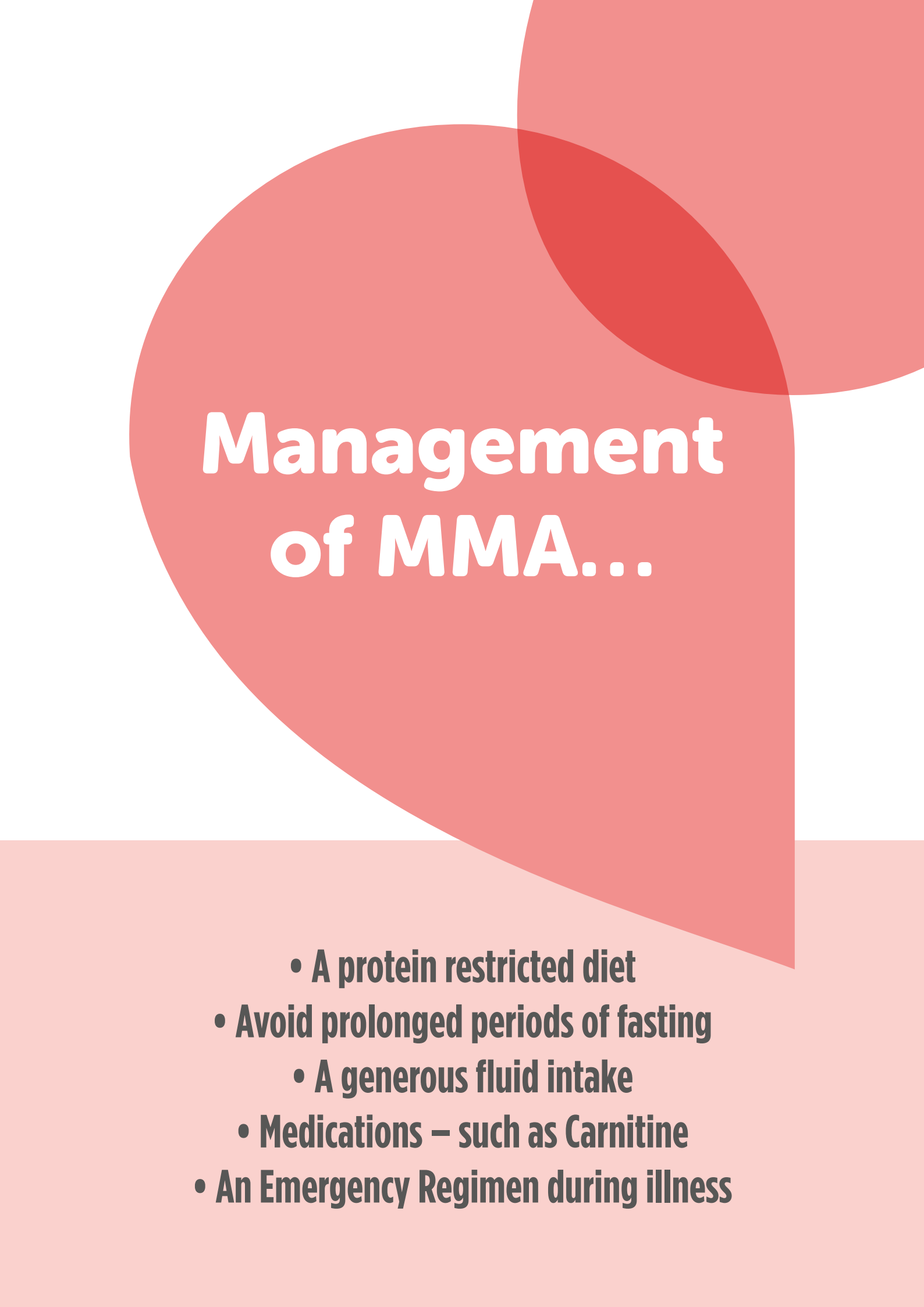
This causes methylmalonic acid and other harmful chemicals to build up
and can damage the brain, kidneys and other organs.

Other sources of methylmalonic acid

The management of MMA aims to reduce the build up of methylmalonic acid and other harmful chemicals from all sources.

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



Management of MMA...

- **A protein restricted diet**
- **Avoid prolonged periods of fasting**
 - **A generous fluid intake**
 - **Medications – such as Carnitine**
- **An Emergency Regimen during illness**

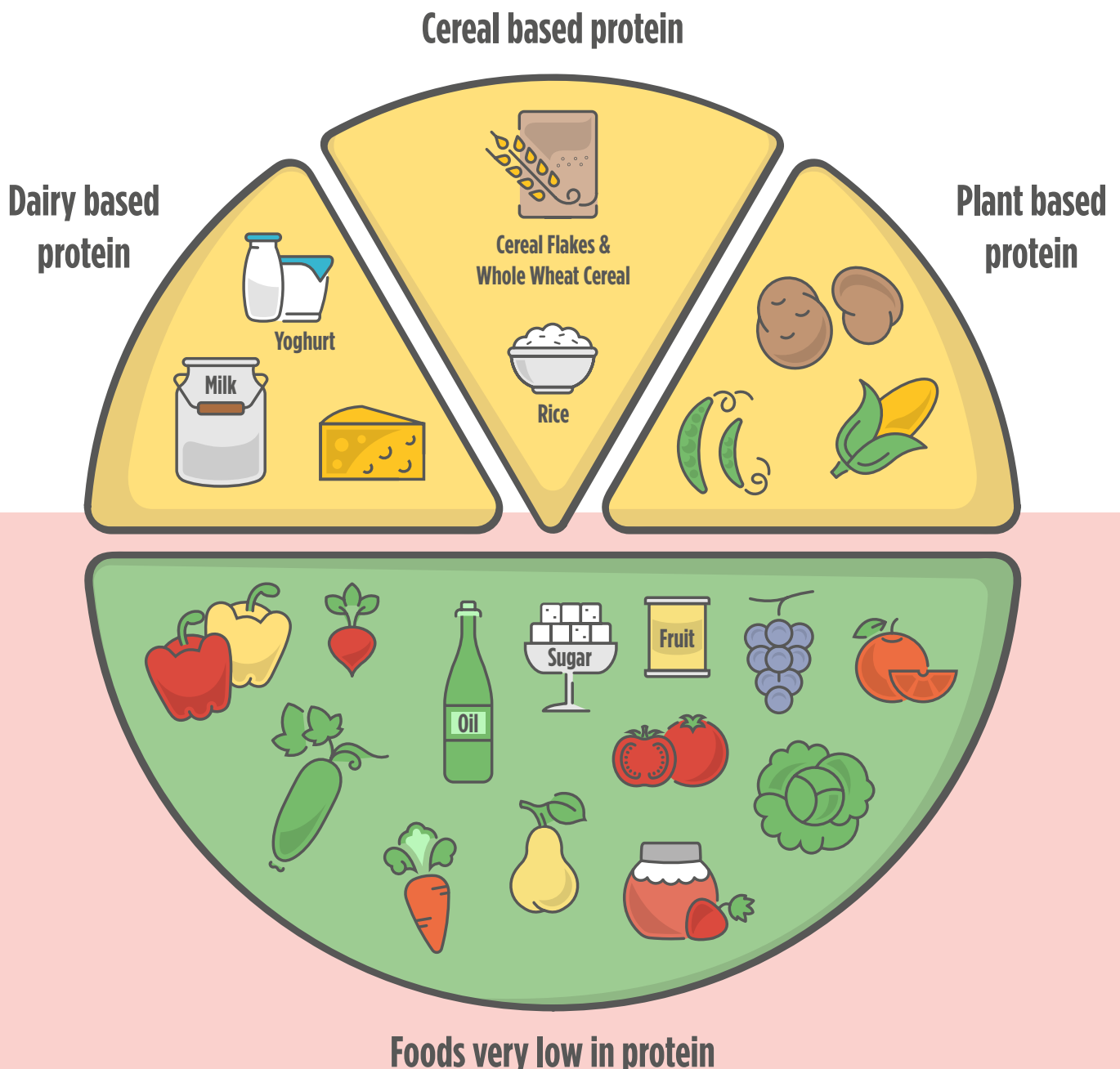
Diet Management

A typical diet has too much protein and therefore methionine, threonine, valine and isoleucine for a person with MMA.

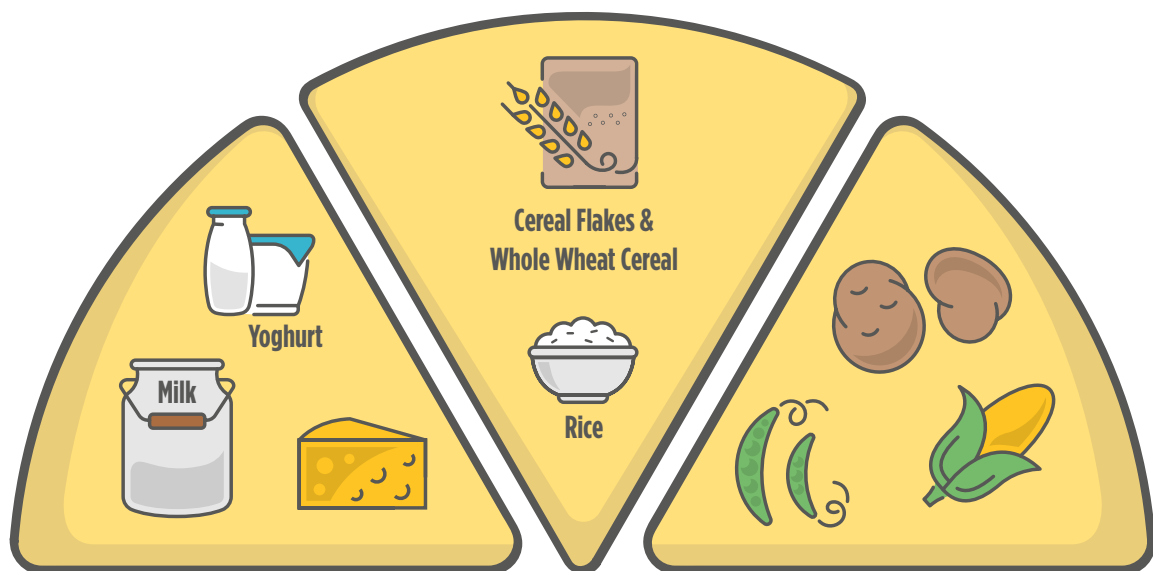
In MMA, it is important that enough protein is given for growth and development, but not too much as harmful chemicals will be made.

Therefore they must follow a protein restricted diet.

The diet will typically be made up as follows:



Measured amounts of protein



Your dietitian will advise you how much protein your child will tolerate.

This will depend on the severity of the condition.

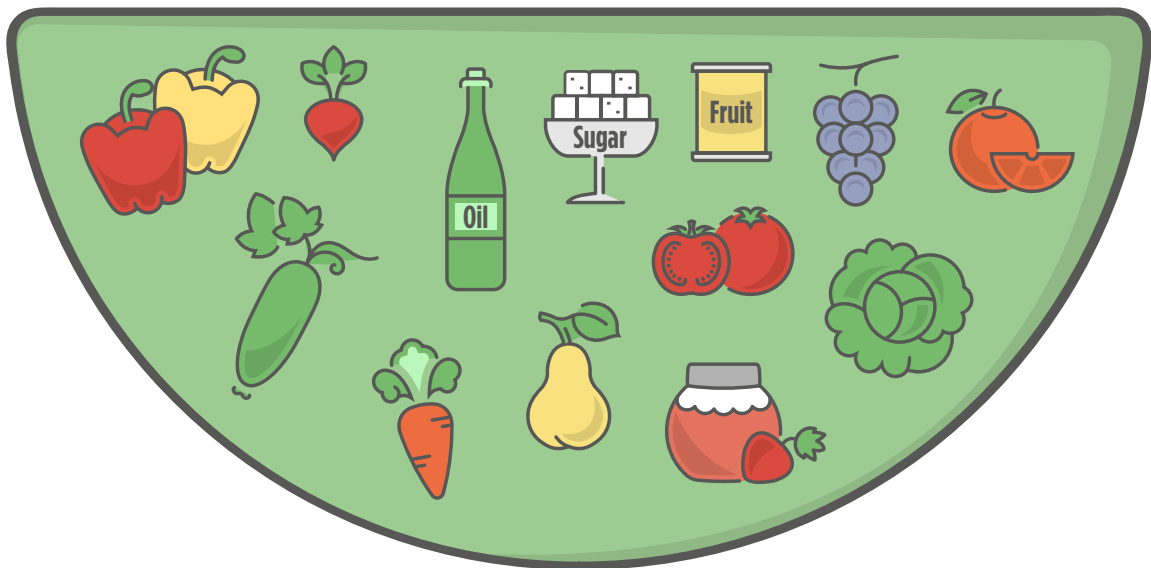
These foods need to be measured and taken everyday.

They will mainly come from dairy, cereal and plant based foods.

Your dietitian will teach you how to measure protein.

They will provide you with a list of foods suitable for your child.

Low protein foods



These foods are very low in protein and can be eaten without being measured.

These foods should make up the majority of your child's diet.

They provide an important source of energy and variety in the diet.

Your dietitian will provide you with a list of foods suitable for your child.

Ensure a sufficient energy supply

This is essential because if the body goes without food for too long there may be a shortage of energy supply.

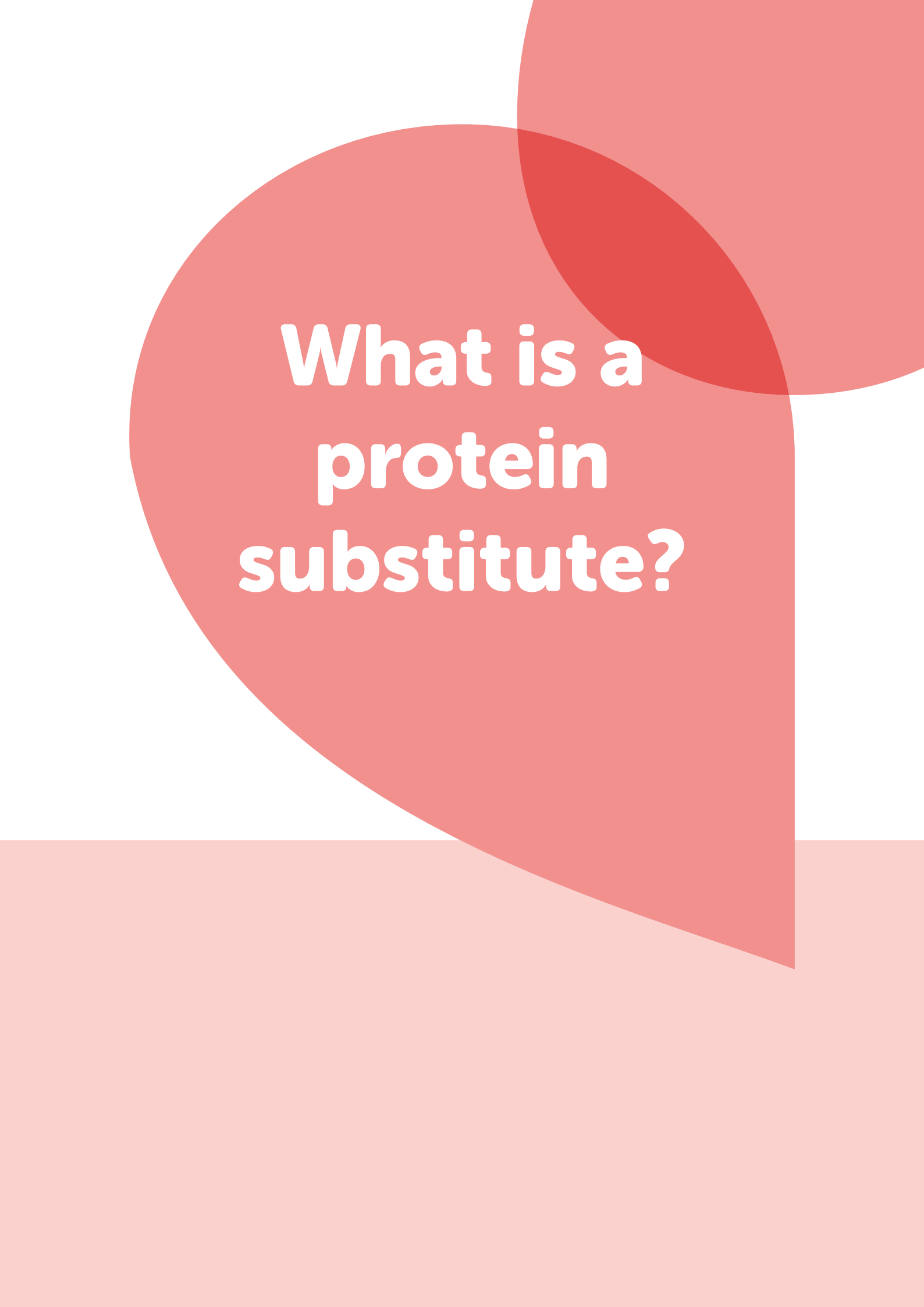
This causes the body to breakdown protein and will cause the build up of methylmalonic acid and other harmful chemicals – this must be avoided.

Avoid prolonged periods of fasting.

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 harmful chemicals.

This will be discussed in more detail with your dietitian.



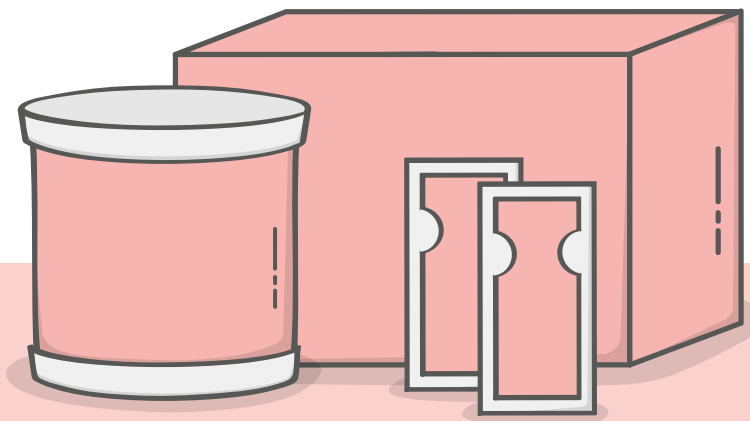
**What is a
protein
substitute?**

Why does a person with MMA need to take a protein substitute?

Due to the protein restricted diet, some individuals may be unable to get all the nutrients they need by food alone. They may need to take a protein substitute*.

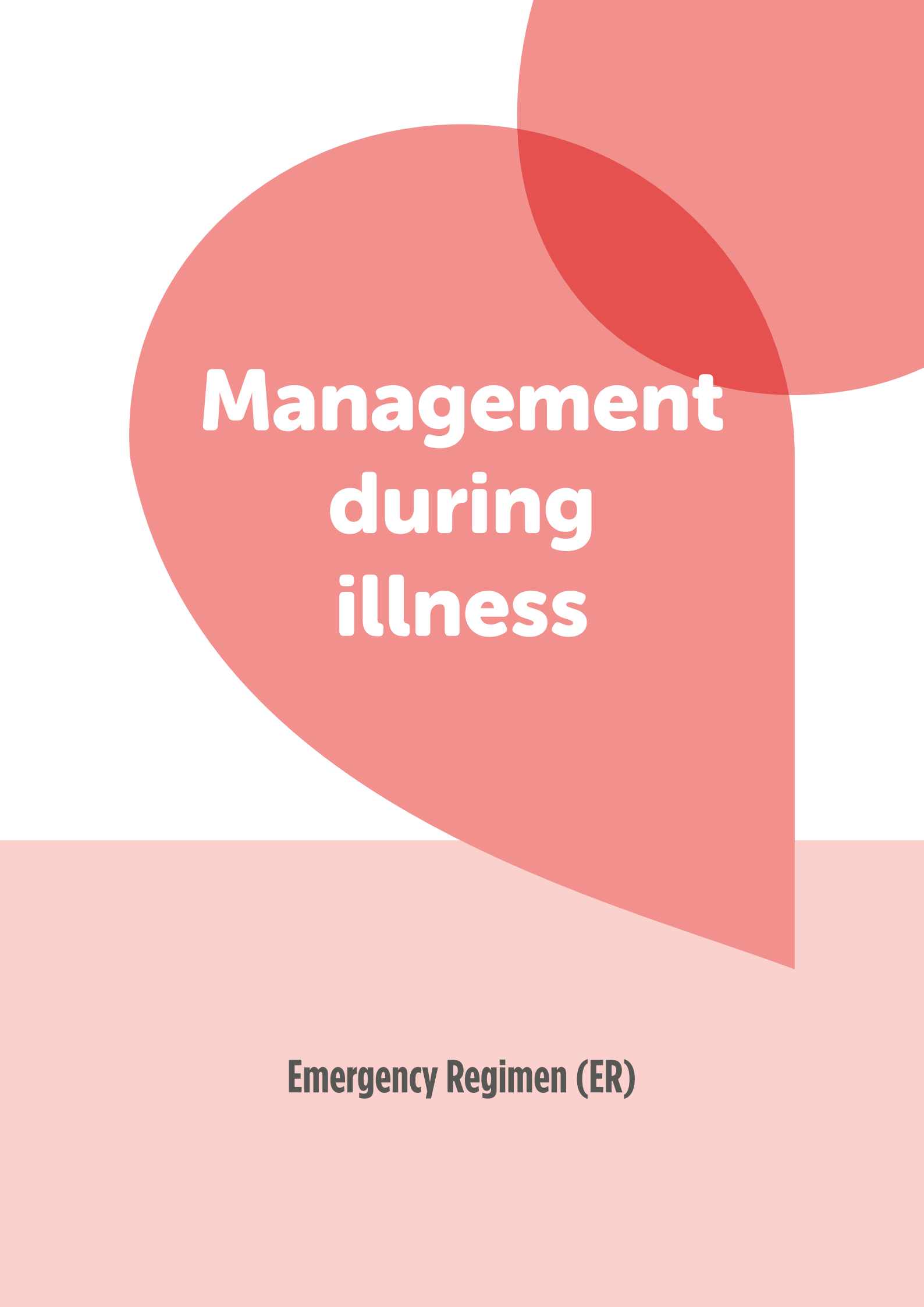
A protein substitute is a specially made medical food. It is methionine, threonine and valine free and has low levels of isoleucine, but it contains all the other amino acids in protein that your child needs to grow and develop.

Most protein substitutes also contain vitamins, minerals and other important nutrients to ensure your child is getting what they need.



If your child requires a protein substitute, your dietitian will discuss this with you in more detail.

* Protein substitutes designed for the dietary management of MMA are Foods for Special Medical Purposes which must be used under medical supervision.



Management during illness

Emergency Regimen (ER)

As with all infants and children, illness will occur from time to time. However, those with MMA will need to start a special feed called the **emergency regimen (ER)** as soon as illness starts. **DO NOT DELAY.**

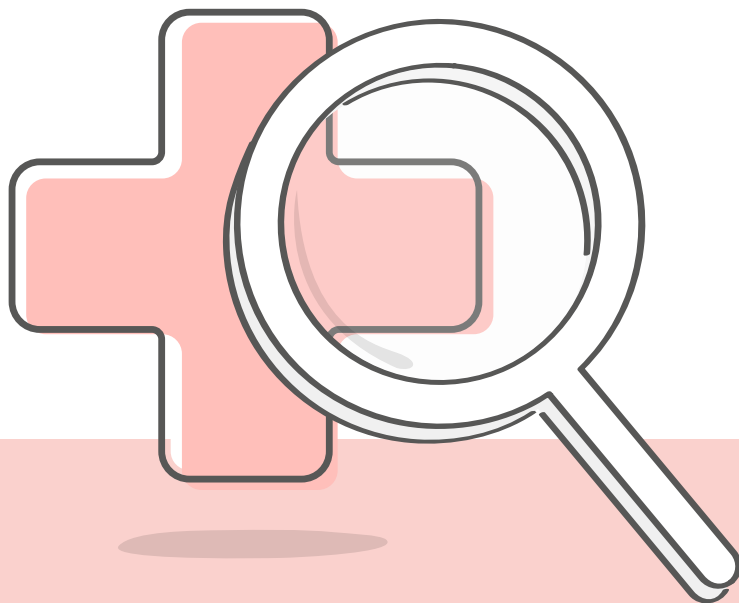
The ER is made up of a glucose polymer.

Your child's metabolic team will provide an emergency regimen specific for your child and will teach you in advance about what to do in times of illness.

This is an extremely important aspect of dietary management.

When your child gets ill, contact your child's metabolic team immediately and keep in regular contact with them.

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



An ER can help minimise the impact of illness on your child.

Any common childhood illness or infection can cause methylmalonic acid levels to rise - avoidance of this is essential.

If left untreated, it can become life threatening.

The ER supplies energy to prevent a buildup of these harmful chemicals.

**Always make sure you have your ER products and a written emergency plan -
check that the products are in date.**

**During illness –
start the ER promptly –
do not delay**

- Stop all protein in food and drink temporarily
- Always complete the full amount of the emergency feed
 - Continue on medication as directed by your doctor
- Maintaining your infant's fluid intake during illness is important and extra fluid may be necessary
- Keep in regular contact with your metabolic team.





**How will I
feed my
newborn?**

Feeding your infant



Your infant may be given a special methionine, threonine, valine and isoleucine free formula and a protein free formula alongside breast milk / standard infant formula.

You will be advised by the dietitian on how much of each to give.

Your dietitian will go through this with you in more detail

Breastfeeding offers many benefits for infant and mother, it is recommended you discuss this with your health care professional.



Introducing complementary feeding to your infant



Monitoring

You and your child will need to attend your specialist metabolic centre to meet with the metabolic team for ongoing monitoring and check-ups.

At your child's monitoring visits, the team will spend time with you to ensure your child's condition is being well managed and tailored to best suit the needs of your child and family.

To do this they look at many different factors such as:

- Your child's **overall health**, growth and well-being.
- **Blood tests** - blood results give a good indication on how well your child is doing.
- Your child's **nutritional intake** - it is helpful to bring diet diaries to your clinic visits.

Your dietitian and doctor will discuss any changes that may be needed to your child's diet or medication.

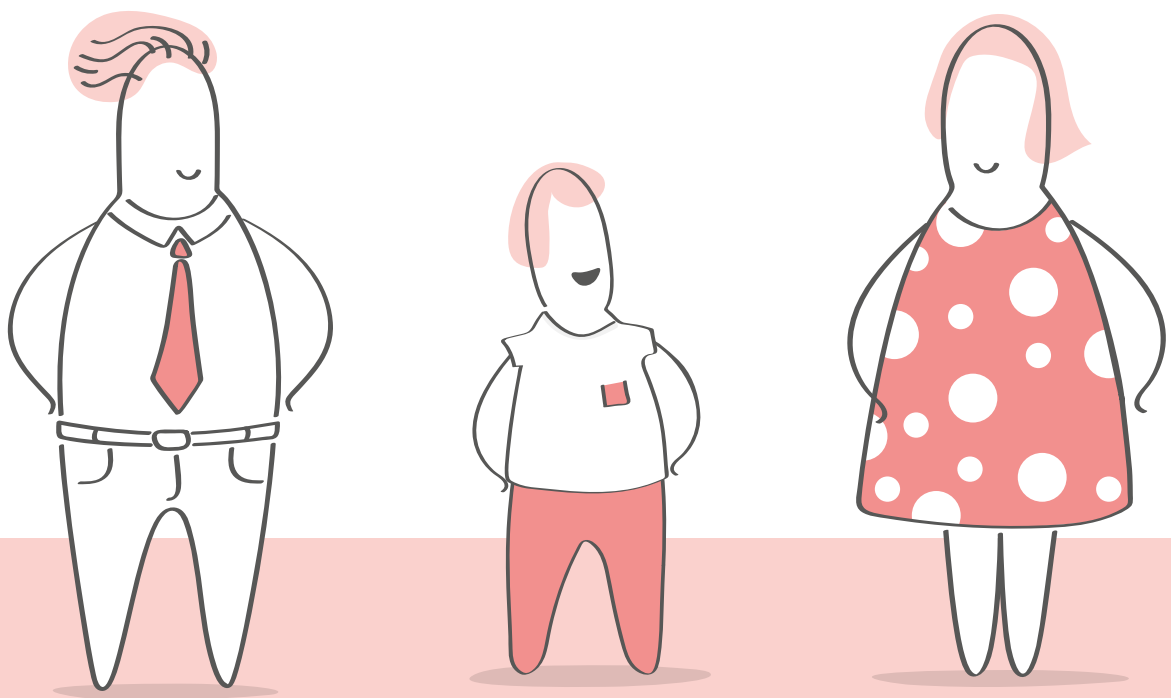
It is very important to attend these monitoring visits. They will also give you opportunities to discuss concerns or questions you might have.



**As your
child
grows up**

Children / Teenagers / Adults

Following the prescribed diet, medication and acting quickly at times of illness ensures the best chance of improving long term outcome.



Closing comments

Before today, you may not have heard of MMA. It may now seem that there is a lot of new information to take on board and that your child will have a very different diet, and perhaps a different life, to what you might have imagined.

Your child's specialist metabolic centre will be there to support you and your child every step of the way. They will use their experience to answer any questions you might have and help overcome any challenges that might arise. In many cases they have encountered the same challenges you are facing with other families, and their recommendations have resulted in successful outcomes.

