An introduction to

Maple Syrup Urine Disease (MSUD)

comidamed™

Contents

Important information

This is an introduction to Maple Syrup Urine Disease (MSUD), following diagnosis of your child with this condition. Consult your metabolic team for diagnosis and treatment for MSUD.

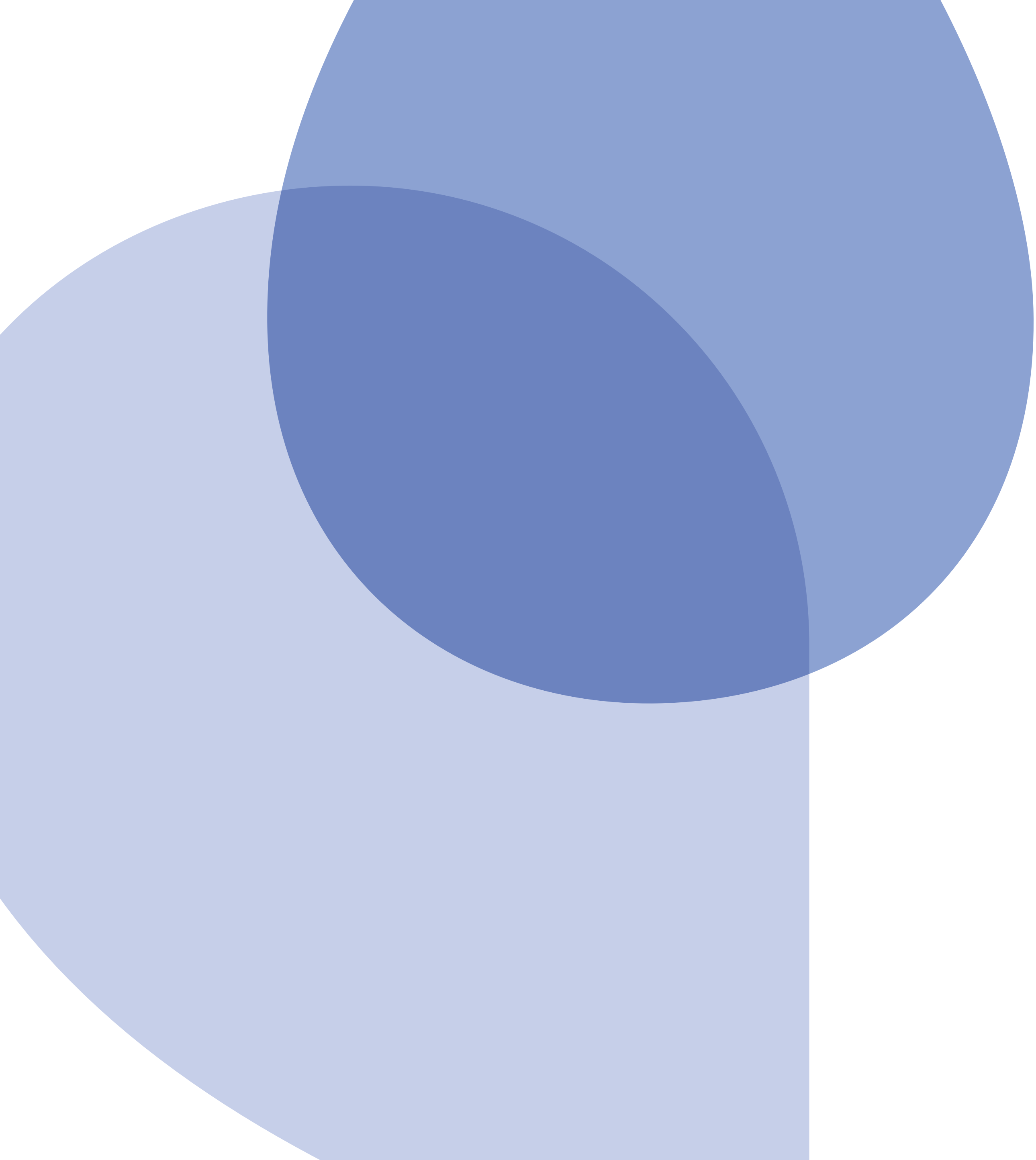
This booklet is not designed to replace any advice given by the metabolic team at the hospital. If you are unclear about anything written in this booklet, please ask your dietitian for further information.

What is MSUD?

Maple Syrup Urine Disease
or MSUD for short.

MSUD is a manageable condition which affects the way the body uses protein.

- Children born with MSUD cannot use parts of protein called the branch chain amino acids (BCAAs).
- There are 3 branch chain amino acids and they are called Leucine, Isoleucine and Valine.
- If left untreated, these amino acids build up in the body and cause brain damage. This can cause an unusual sweet smell in both the urine and sweat, which is where the name comes from.
- The management for MSUD is a strict lifelong special diet low in Leucine, Isoleucine and Valine and emergency management during illness.



Infants with MSUD are identified by newborn screening.

Special dietary management and careful monitoring can improve long term outcome. If treated effectively and early, children with MSUD can do well.

Diagnosis of MSUD

MSUD is diagnosed by a blood test called "The heel prick test"

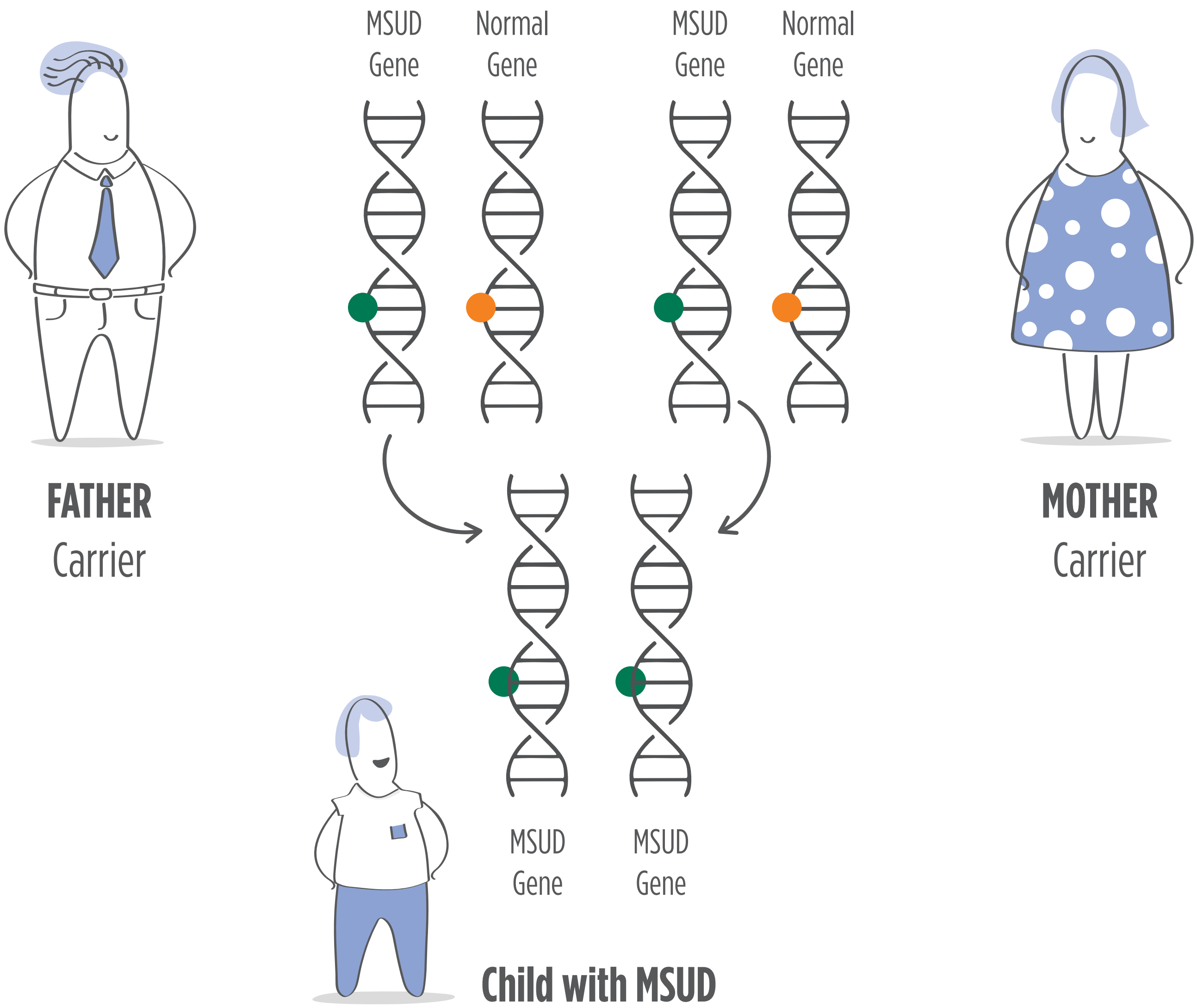
This blood test (heel prick test) is carried out by a healthcare professional after birth.

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

Why does my child have MSUD?

MSUD is an inherited condition

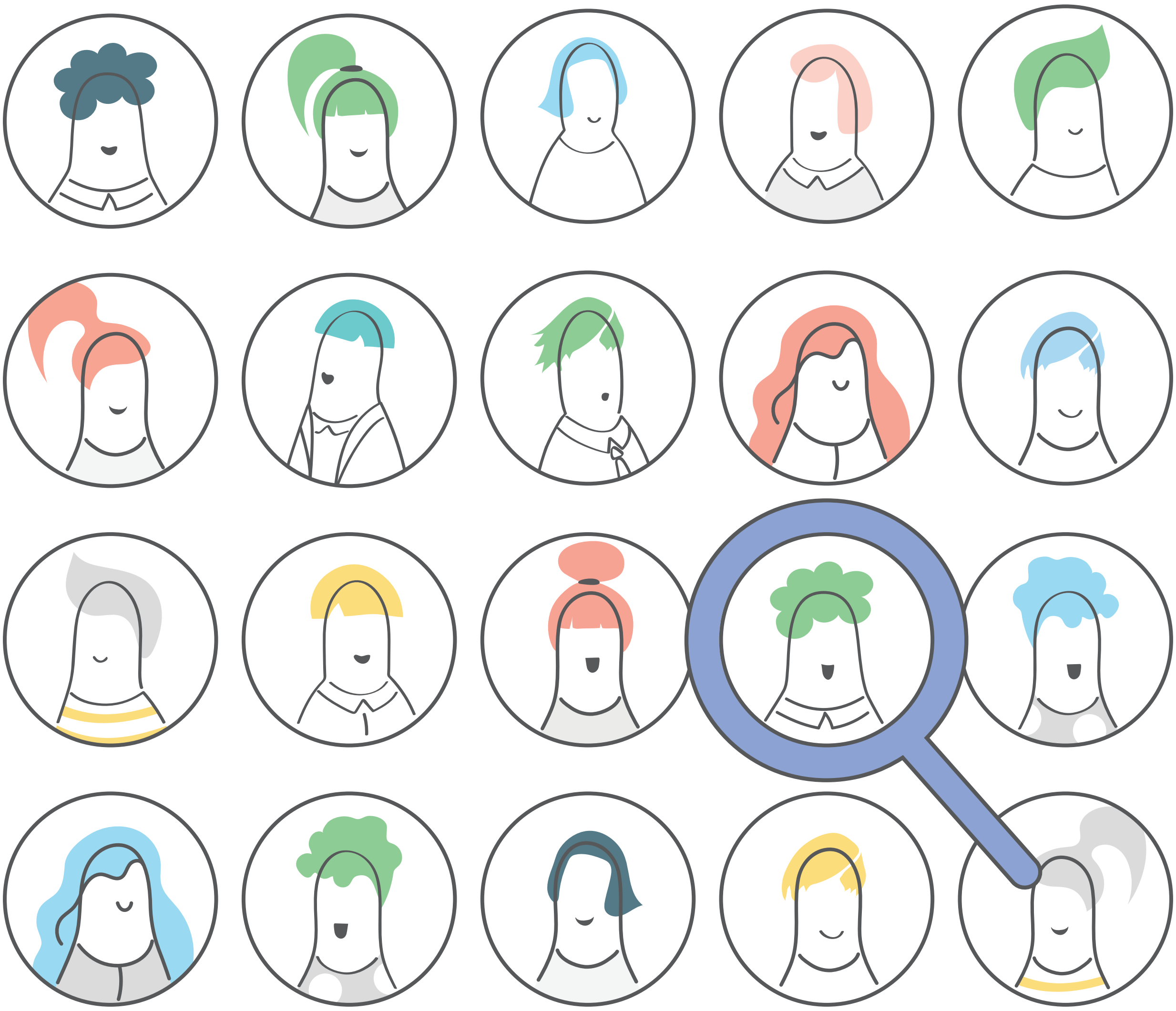
It's nobody's fault and there is nothing you could have done to prevent it.



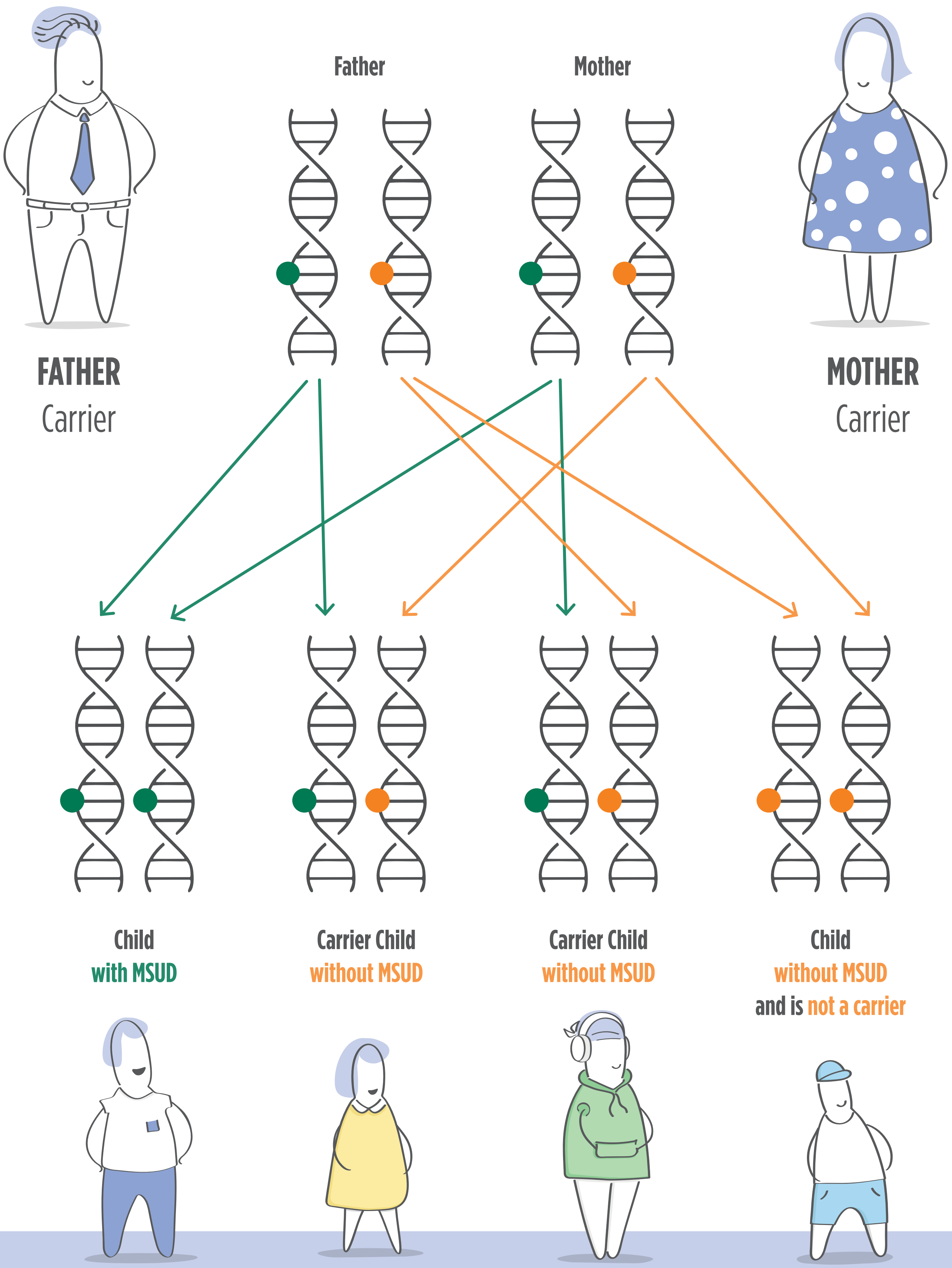
As a parent of a child with MSUD, you have one MSUD gene and one normal one. This is known as being a carrier.

Your child has inherited 2 MSUD genes, one from the mother and one from the father.

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



People who are carriers for MSUD do not have MSUD themselves and the faulty gene does not cause a problem to them.



Father

Mother

FATHER
Carrier

MOTHER
Carrier

Child
with MSUD

Carrier Child
without MSUD

Carrier Child
without MSUD

Child
without MSUD
and is not a carrier

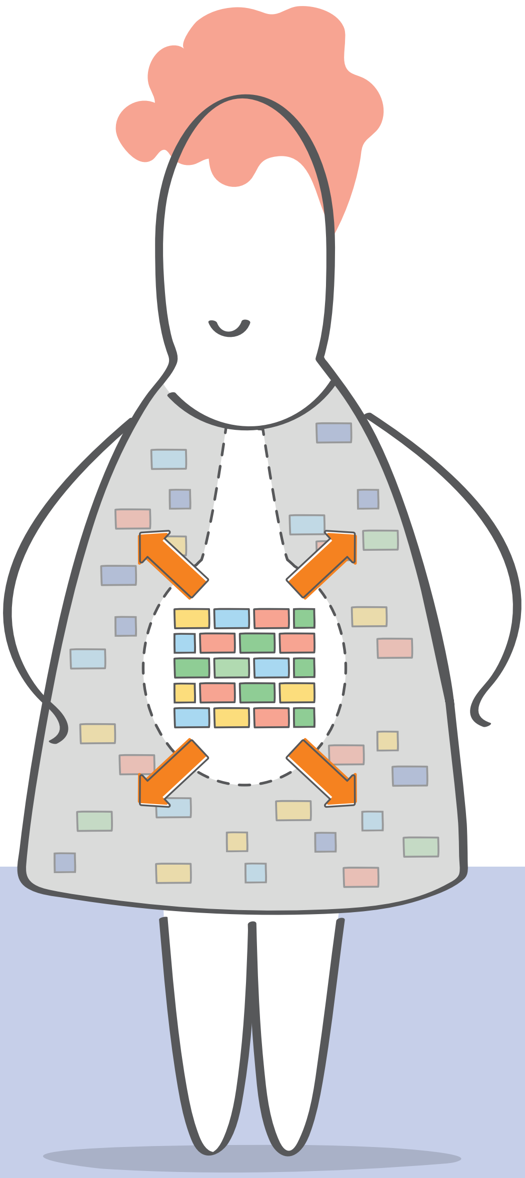
When 2 people who are carriers of the MSUD gene conceive a child there is a 1 in 4 chance of that child having MSUD.

The science bit… protein explained

Why do we need protein?

Protein is a nutrient needed by the body.

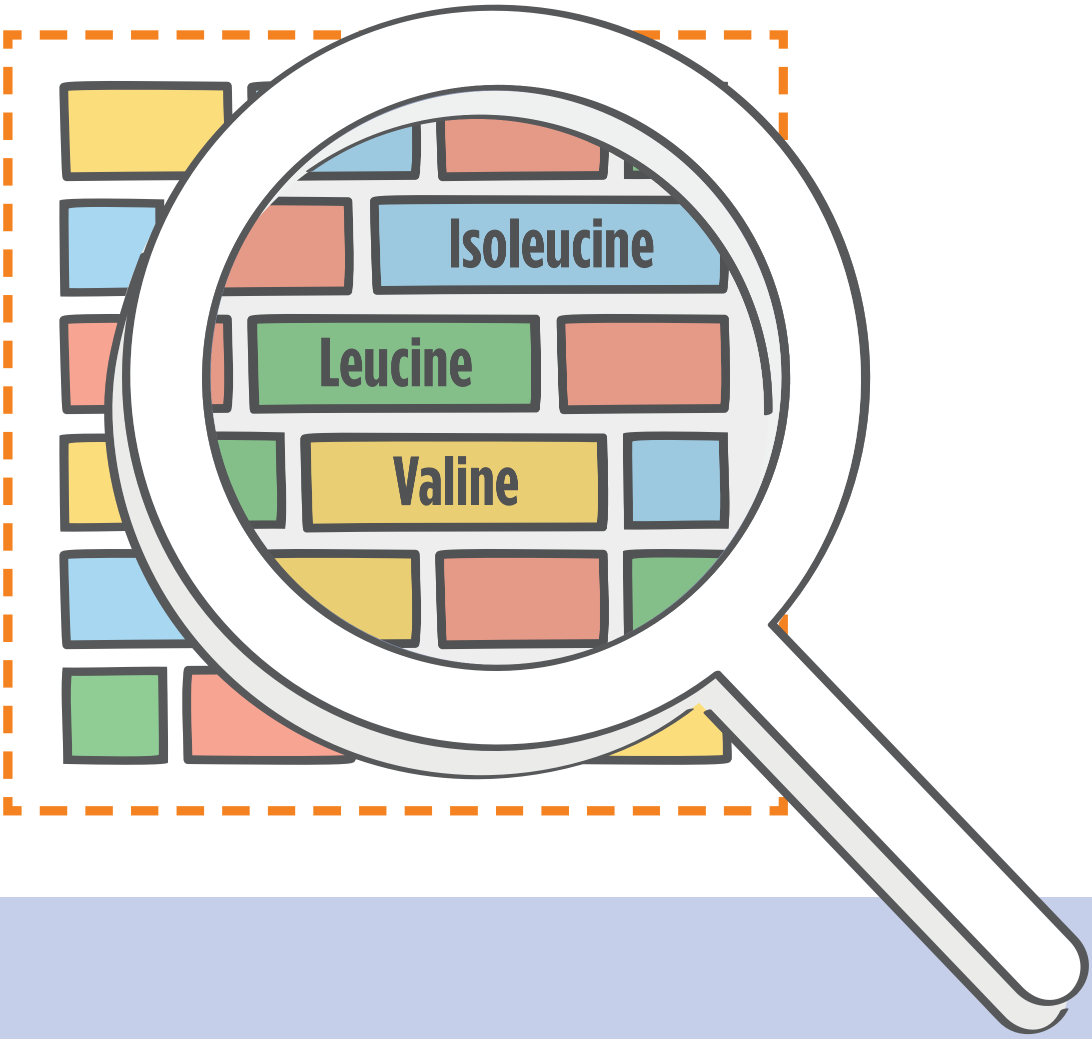
It helps to build, repair and maintain body cells and tissues, like your skin, muscles, organs, blood and even bones.



When protein is eaten, it is broken down in the body (during digestion) into smaller pieces (like building blocks).

These smaller pieces are called amino acids.

Protein is made up of many building blocks called amino acids



There are 20 amino acids (building blocks) that make up protein.

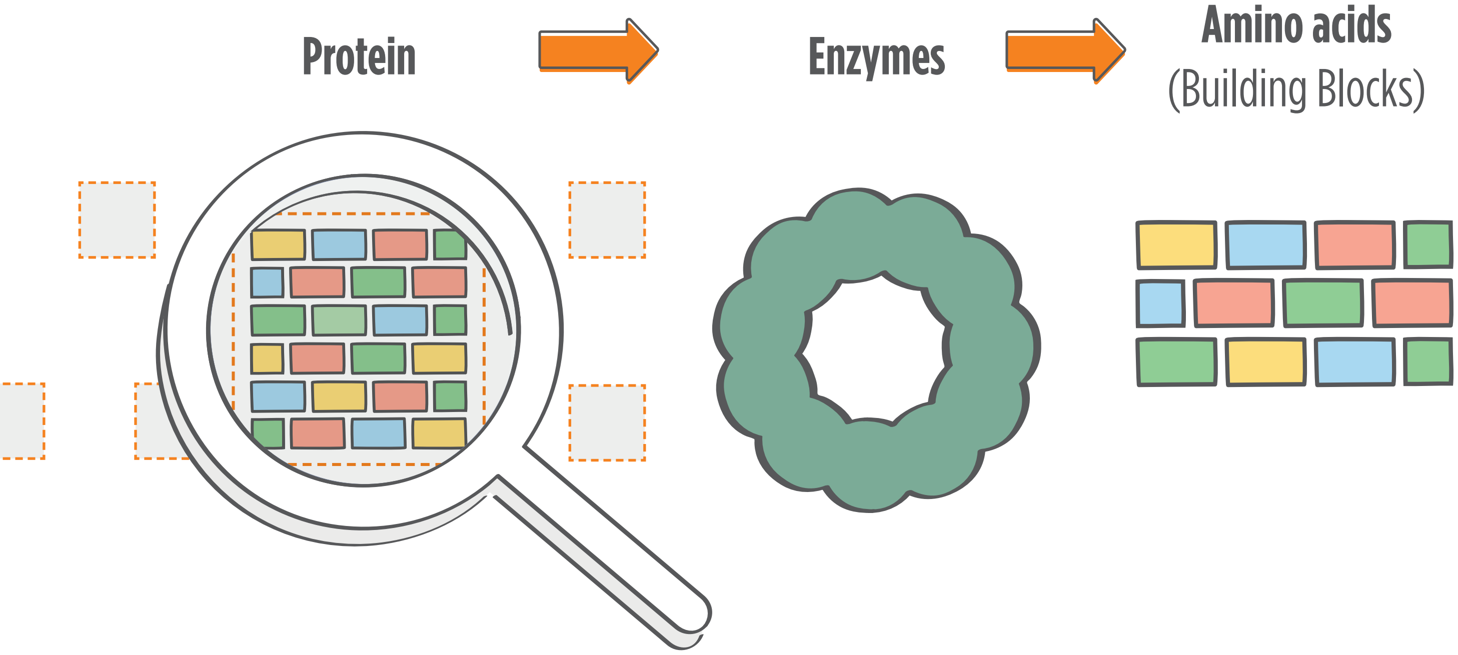
Some of these are ESSENTIAL and cannot be made in the body – so they must come from the food we eat.

Leucine, Isoleucine and Valine are essential amino acids that a person with MSUD cannot process correctly.

These are also referred to as branch chain amino acids (BCAAs).

What normally happens

The conversion of protein into amino acids



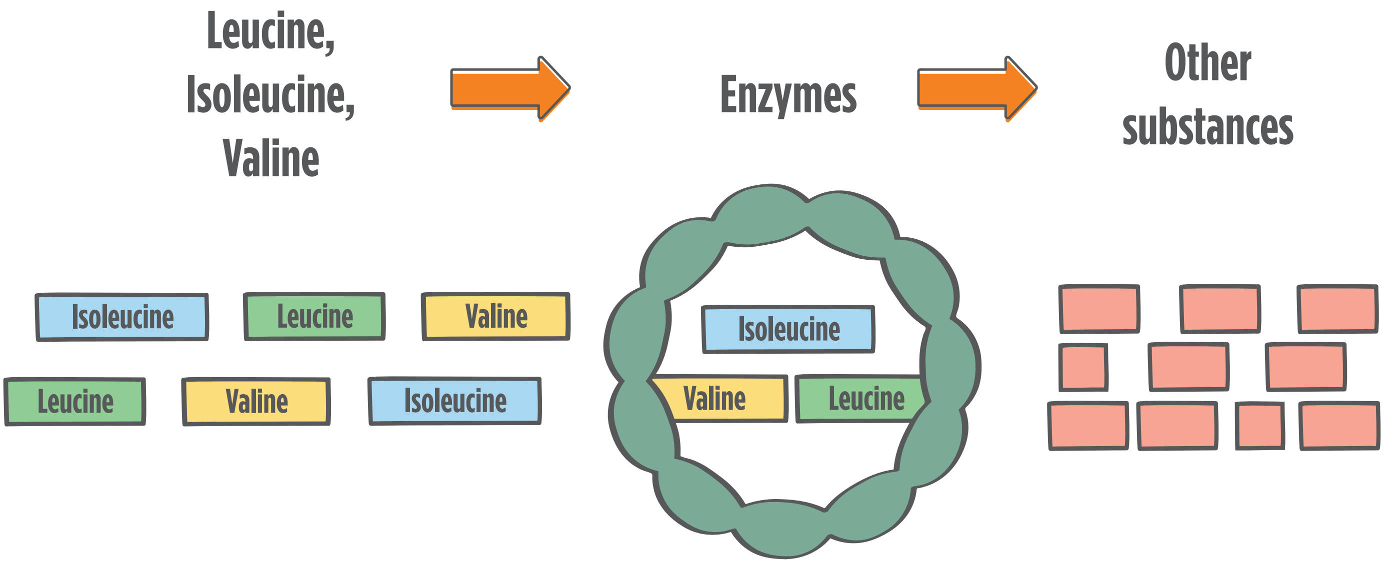
Protein → **Enzymes** → **Amino acids** (Building Blocks)

Enzymes are like chemical scissors needed for the breakdown of protein into amino acids.

The body uses these amino acids for growing, building up muscle and helping the body stay healthy.

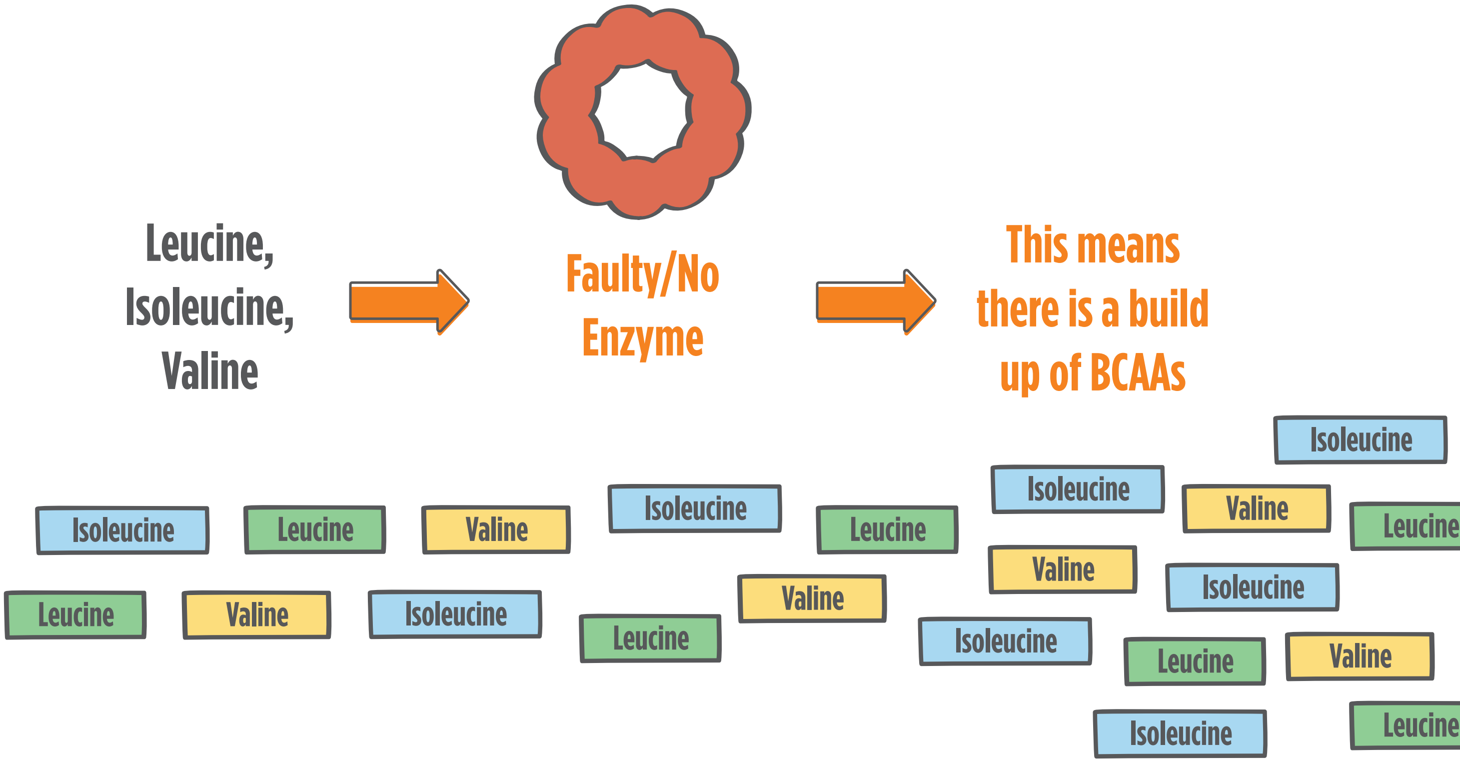
Why can someone with MSUD not process BCAAs correctly?

In a person **without** MSUD the Leucine, Isoleucine and Valine (the BCAAs) are converted to other substances.



Normally, the liver produces a group of enzymes which break down the BCAAs into other substances which have important functions in the body.

In a person **with** MSUD, the conversion of BCAAs does not happen as it should.



Leucine,
Isoleucine,
Valine

Faulty/No
Enzyme

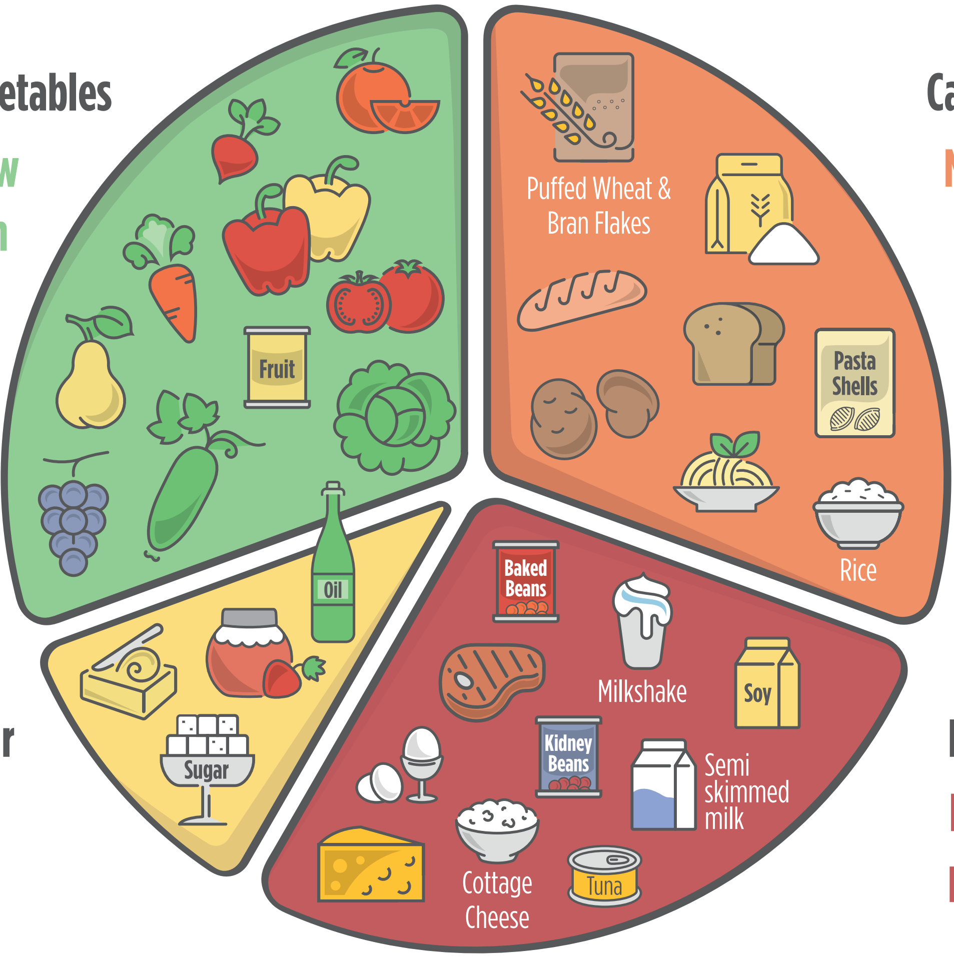
This means
there is a build
up of BCAAs

In MSUD the group of enzymes that break down Leucine, Isoleucine and Valine do not work properly. This means that BCAA's build up in the blood. High levels of Leucine or BCAA's are toxic in the blood and this results in damage to tissues in the body, including the brain.

This is the reason a special diet, low in protein, needs to be followed.

Management of MSUD… The diet explained

A Regular Diet



Fruit and Vegetables

Very low protein

Carbohydrate

Moderately high in protein

Fat and sugar

Protein free

Protein

High in protein

A regular diet has too much protein/BCAAs for a person with MSUD. Therefore they must follow a special diet.

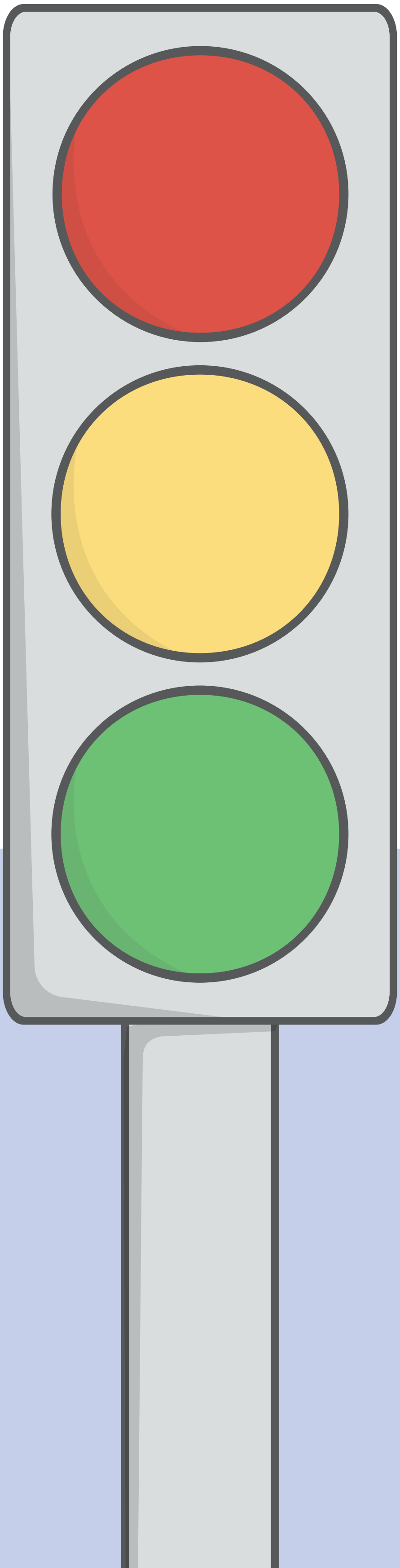
MSUD is managed by following a strict low protein/ low Leucine diet

This is because Leucine in the blood is generally higher than Isoleucine and Valine in people with MSUD, so the diet is therefore managed by a controlled Leucine intake.

- Leucine is found in all protein containing foods, e.g. meat, fish, eggs, cheese, chicken, quorn, nuts, soya, pulses and in foods such as normal bread, biscuits and chocolate.
- In order to manage MSUD, **all protein foods must be restricted** – and a low protein / low Leucine diet must be followed.
- All amino acids have a role or function in the body. BCAAs help us with our growth, development and tissue repair.
- It is important that someone with MSUD gets some Leucine to grow normally, but not so much that it becomes harmful.

Traffic Light System

The low protein/low Leucine diet a person with MSUD must follow is best described by using the traffic light system:



RED: STOP!

Too high in protein / Leucine

Do not eat these foods.

AMBER: Go Cautiously!

Contains some protein / Leucine

These foods can be eaten in measured amounts (exchanges).

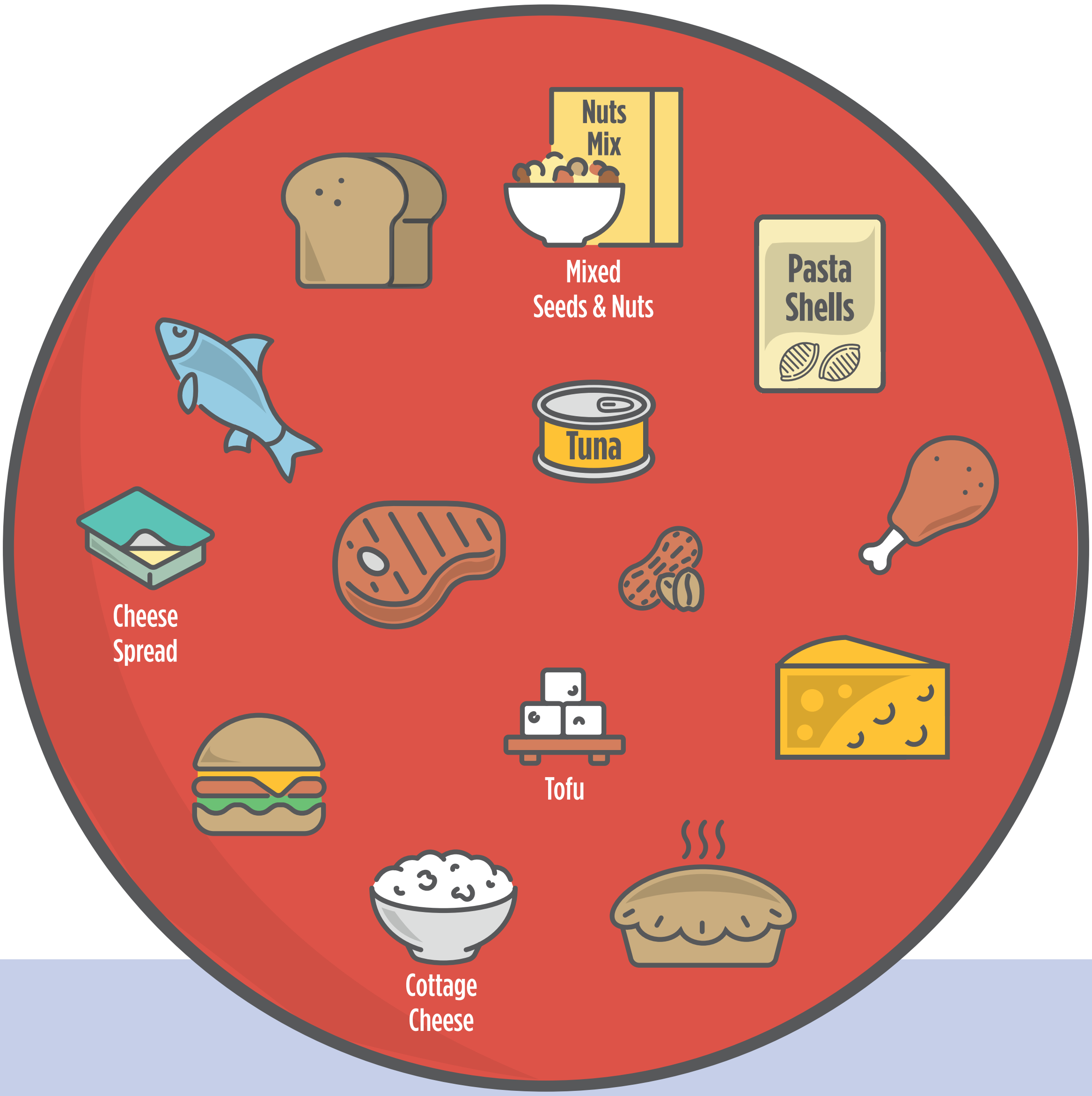
GREEN: Go!

Very low in / free from protein / Leucine

These foods can be eaten freely.

RED: STOP!

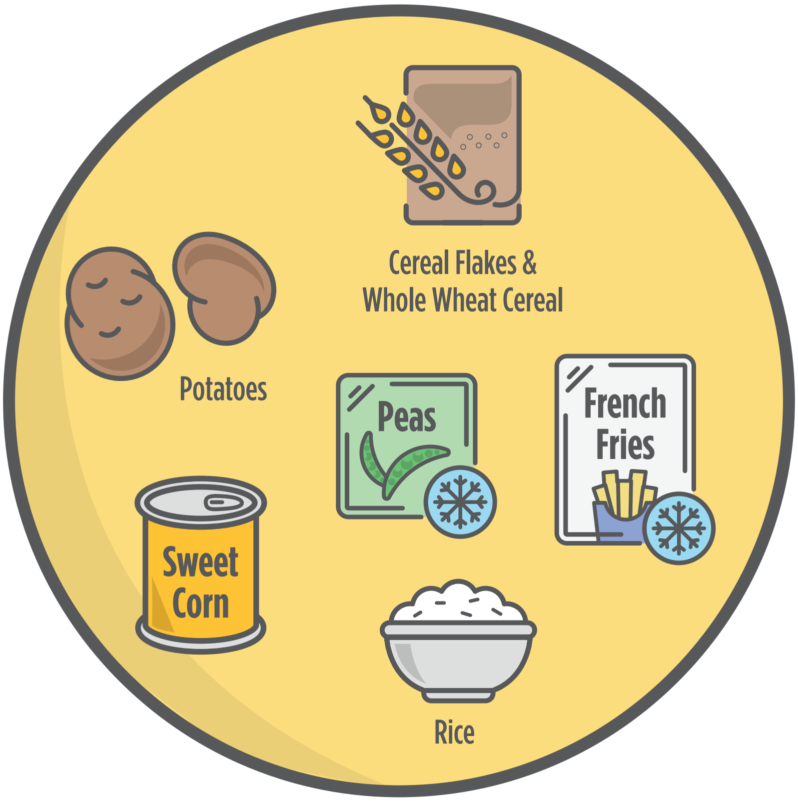
Do not eat these foods.

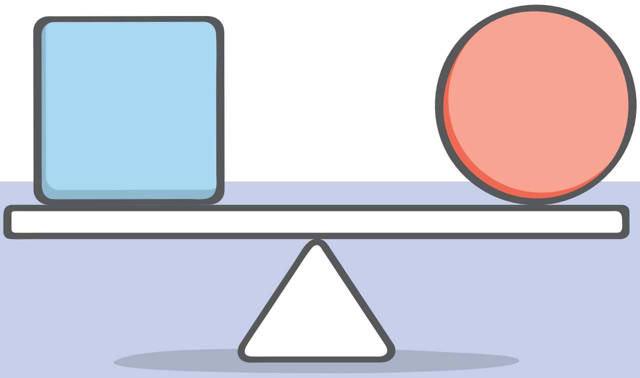


These foods are all rich in protein and therefore high in Leucine so they are not allowed.

AMBER: Go Cautiously!

These foods can be eaten in measured amounts and are known as exchanges.





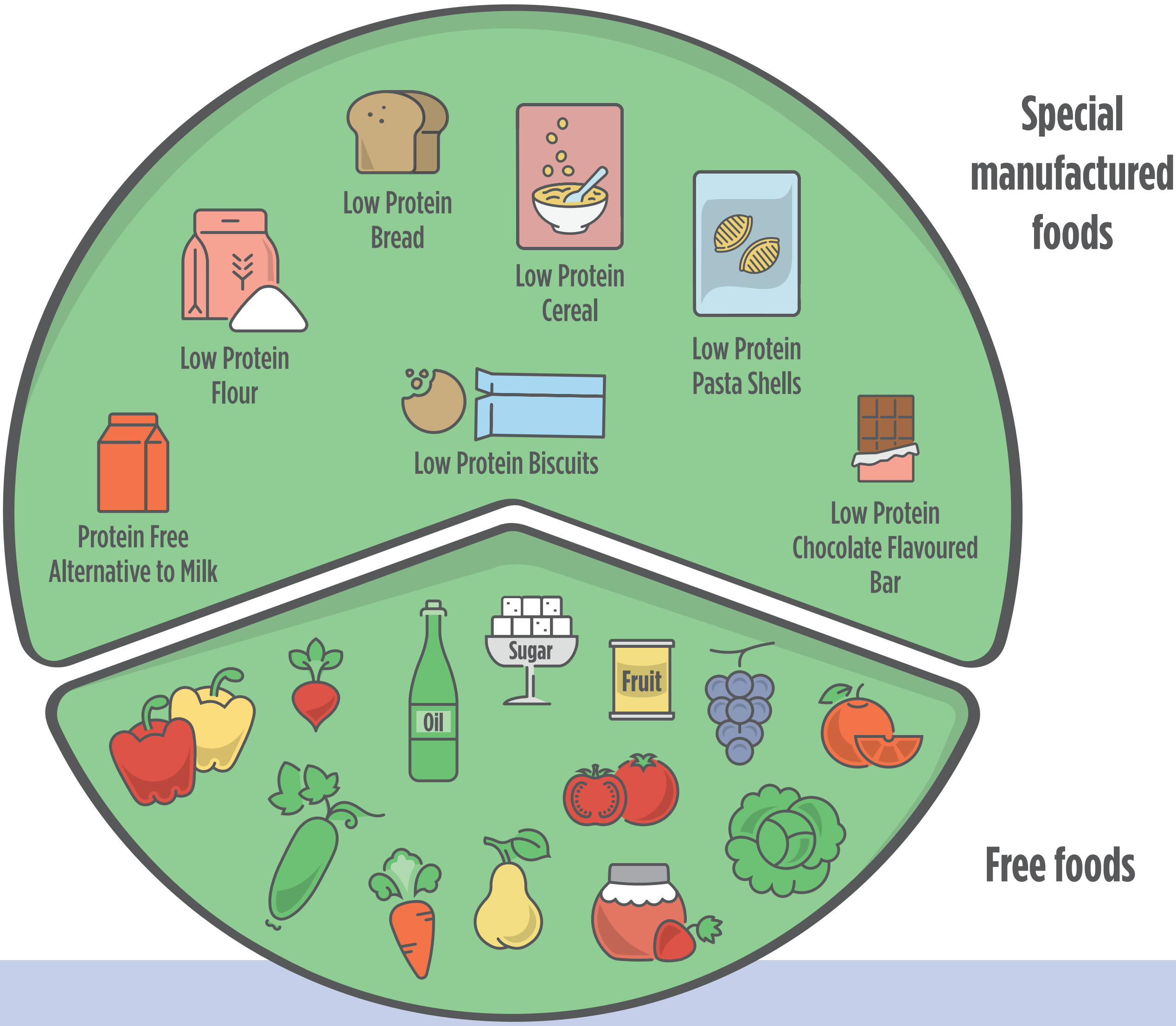
1 Exchange

50mg Leucine
or 0.5g Protein

Other foods like these contain small amounts of protein. These foods can be eaten in measured amounts. The measure is called an exchange. These foods are spread out between the day's meals to provide small amounts of essential Leucine. The quantities allowed will vary from person to person and from time to time in the same person.

GREEN: Go!

These foods can be eaten freely.



These foods are either naturally low in Leucine or have been specially made to be low in protein and so are “FREE from exchanges” or do not have to be measured*.

*Please note certain low protein manufactured foods will need to be counted as part of Leucine exchanges. Please check with your dietitian.

What is a protein substitute?

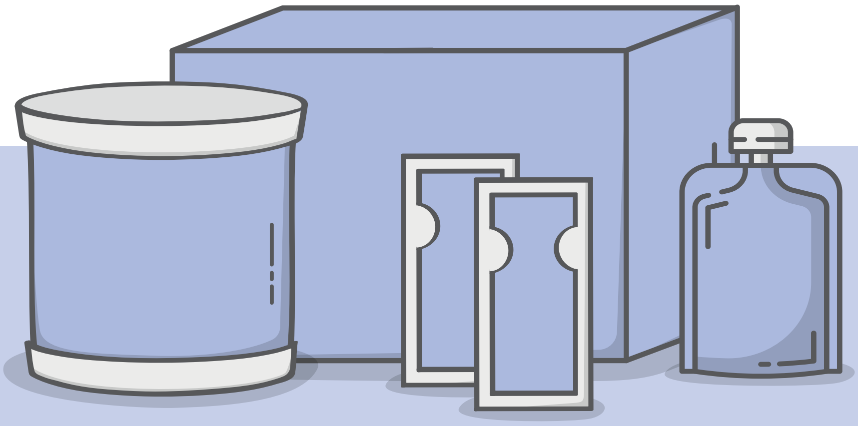
Why does a person with MSUD need to take a protein substitute?

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

A protein substitute is a specially made medical supplement. It contains NO added Leucine, Isoleucine or Valine. But it contains all of the other amino acids in protein that your child needs to grow.

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

The protein substitute is an extremely important part of the diet for a person with MSUD and it must be taken regularly and evenly spread over the day. This helps to keep the BCAA levels steady throughout the day.



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

---

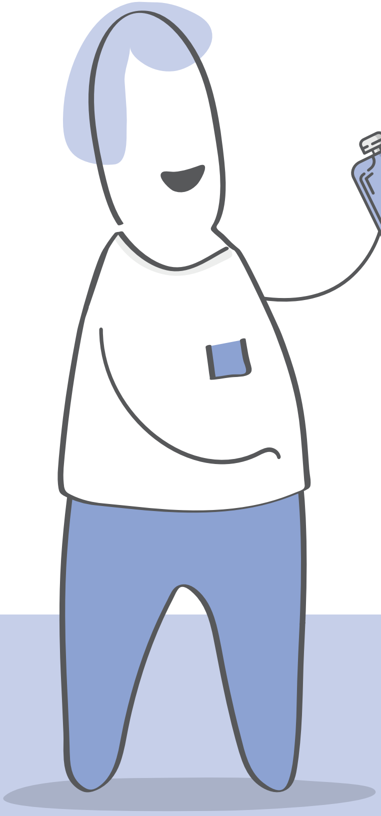
*Protein substitutes designed for the dietary management of MSUD are Foods for Special Medical Purposes which must be used under medical supervision.

Protein Substitutes

Many protein substitutes are designed to fit easily into everyday life.

Your dietitian will advise you on which protein substitute is best for your child at each stage of their development.

Your child may require additional Isoleucine and Valine supplements if levels of these fall below the desired range.



How will I feed my newborn?

Infants: breastfeeding

Step 1: Once your child is diagnosed, you will be seen by a dietitian. Your infant will be given a BCAA free formula until the levels of BCAAs fall back to normal levels (usually within a few days). Some children may need additional medical treatment to achieve normal BCAA levels.

Step 2: Once the levels of BCAAs are under control a small amount of BCAA free formula is given before a breastfeed to restrict the amount of breast milk taken by the infant. Breast milk provides limited but essential BCAAs for normal growth.

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

Infants: non-breastfeeding (Bottle feeding)

Step 1: Once your child is diagnosed, you will be seen by a dietitian. Your infant will be given a BCAA free formula until the levels of BCAAs fall back to normal levels (usually within a few days). Some children may need additional medical treatment to achieve normal BCAA levels.

Step 2: Once the levels of BCAAs are under control a small restricted amount of standard infant formula will be given along with the BCAA free formula to provide the limited BCAAs essential for normal growth.

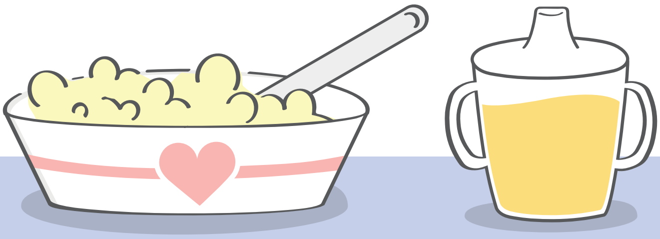
Weaning

When to start weaning*

Your dietitian will advise you when to start solid foods, but it will be at the same time as infants without MSUD.

These will initially be BCAA free foods or foods that have a very low protein (low Leucine) content.

This will help your child to develop a healthy eating pattern while eating a variety of fresh foods.

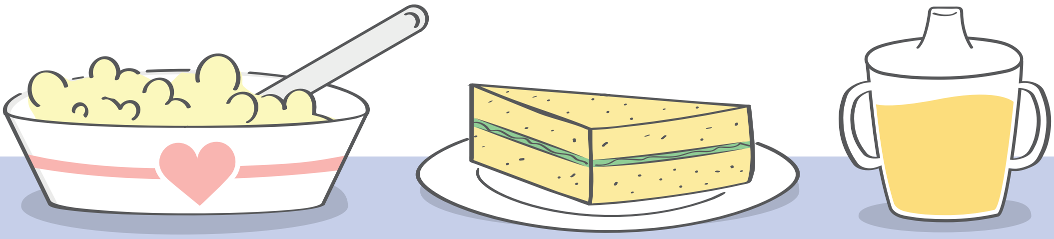


This is also the time that a second stage protein substitute can be introduced, your dietitian will advise you about this this.

* Weaning may occur a little earlier than 6 months, depending on your infant, but it should never be started before 17 weeks.

7 to 12 months

Your child will then be encouraged to move to more textured food and finger foods. Over time, they will get more of their Leucine exchanges from food (e.g. potato or permitted cereal).
Less exchanges will come from breast milk or formula milk.



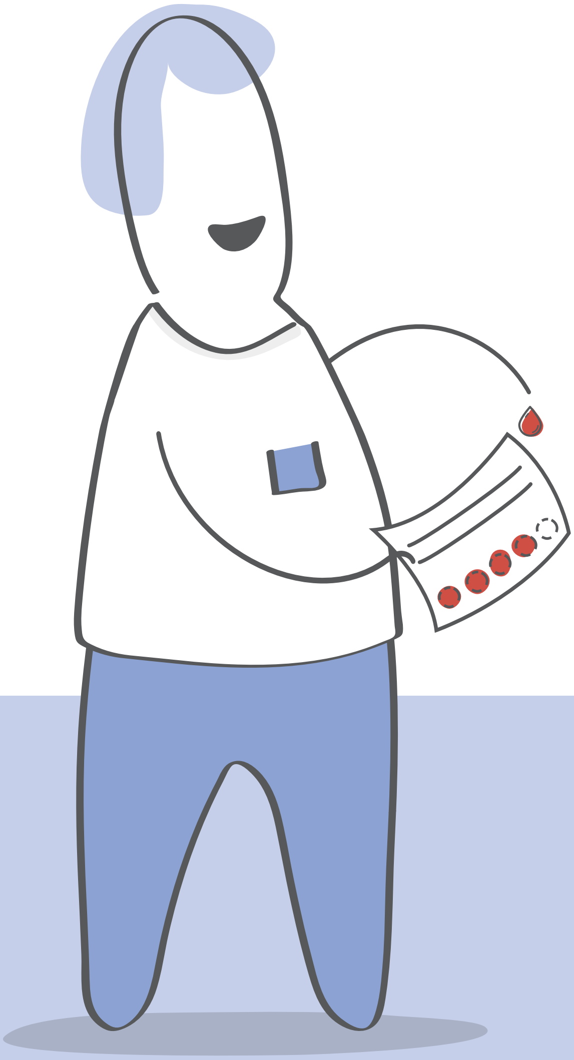
Leucine free foods should still be included in the diet as well.

Your child will also continue their gradual switch from their BCAA free formula to second stage protein substitute.

Monitoring

Your child will have regular blood tests and monitoring to make sure their BCAA levels are under control. This is very important to ensure that they do not have too little or too much BCAA in their diet.

Your child may require additional Isoleucine and Valine supplements if levels of these fall below the desired range.



Your dietitian will contact you once the results are processed and discuss any changes that may be needed to your child's diet.

Emergency Management

As with all infants and children illness will occur from time to time.

However, those with MSUD may need to start a special feed regimen called the **emergency regimen (ER)** as soon as illness starts.

Any common childhood illness or infection can cause Leucine levels to rise which may lead to vomiting, irritability, sleepiness and unusual breathing.

The ER is designed to supply plenty of energy. Your child will also continue with their usual protein substitute. In some cases, the amino acids Isoleucine and Valine are also added to the ER.

The ER helps to reduce the Leucine level to non-toxic levels.

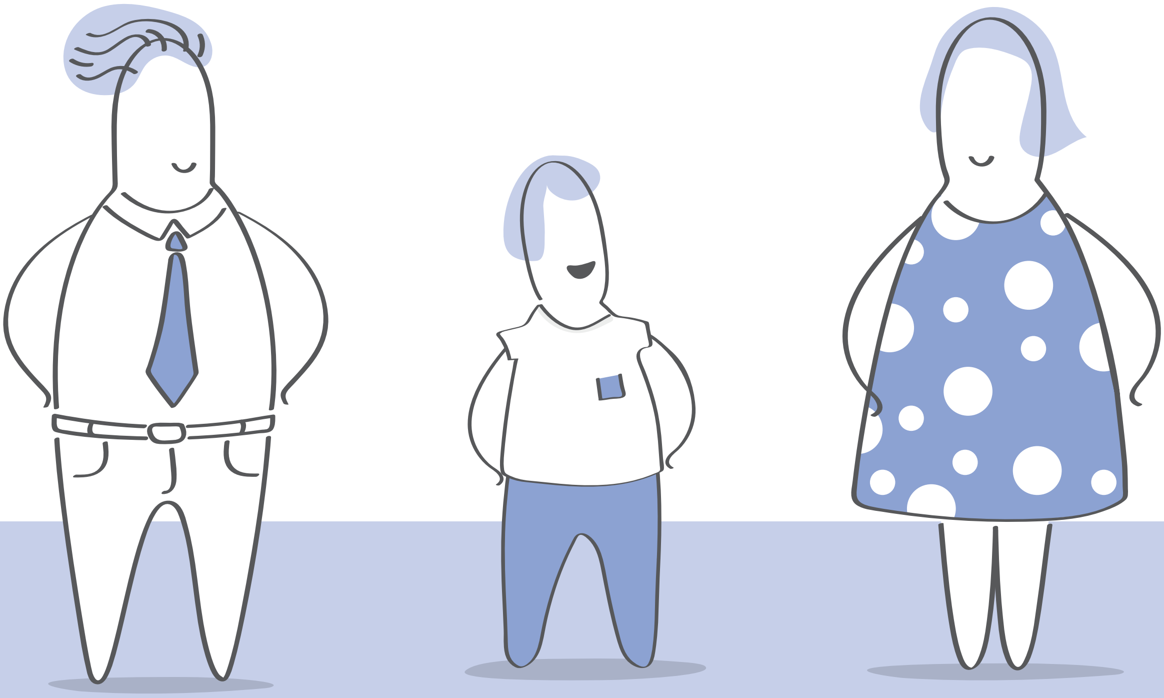
Your dietitian will prepare you in advance about what to do in times of illness.

As your child grows up

Children / Teenagers / Adults

Your child will continue to follow a low protein / low Leucine diet as they grow up into adulthood.

You, your child and dietitian will help choose the protein substitute that best suits them.



All information is correct at time of publication July 2025