

Help and support for families living with MSUD



CLIMB (Children Living with Inherited Metabolic Diseases) is the United Kingdom's foremost provider of free metabolic disease information to young people, adults, families, professionals and other interested groups. CLIMB's vision is to provide Metabolic Disease specific information, advice and support to children, young people, adults, families and professionals in the United Kingdom and to provide information and support to families worldwide.

Web: www.climb.org.uk

Written in collaboration with BIMDG dietitians.

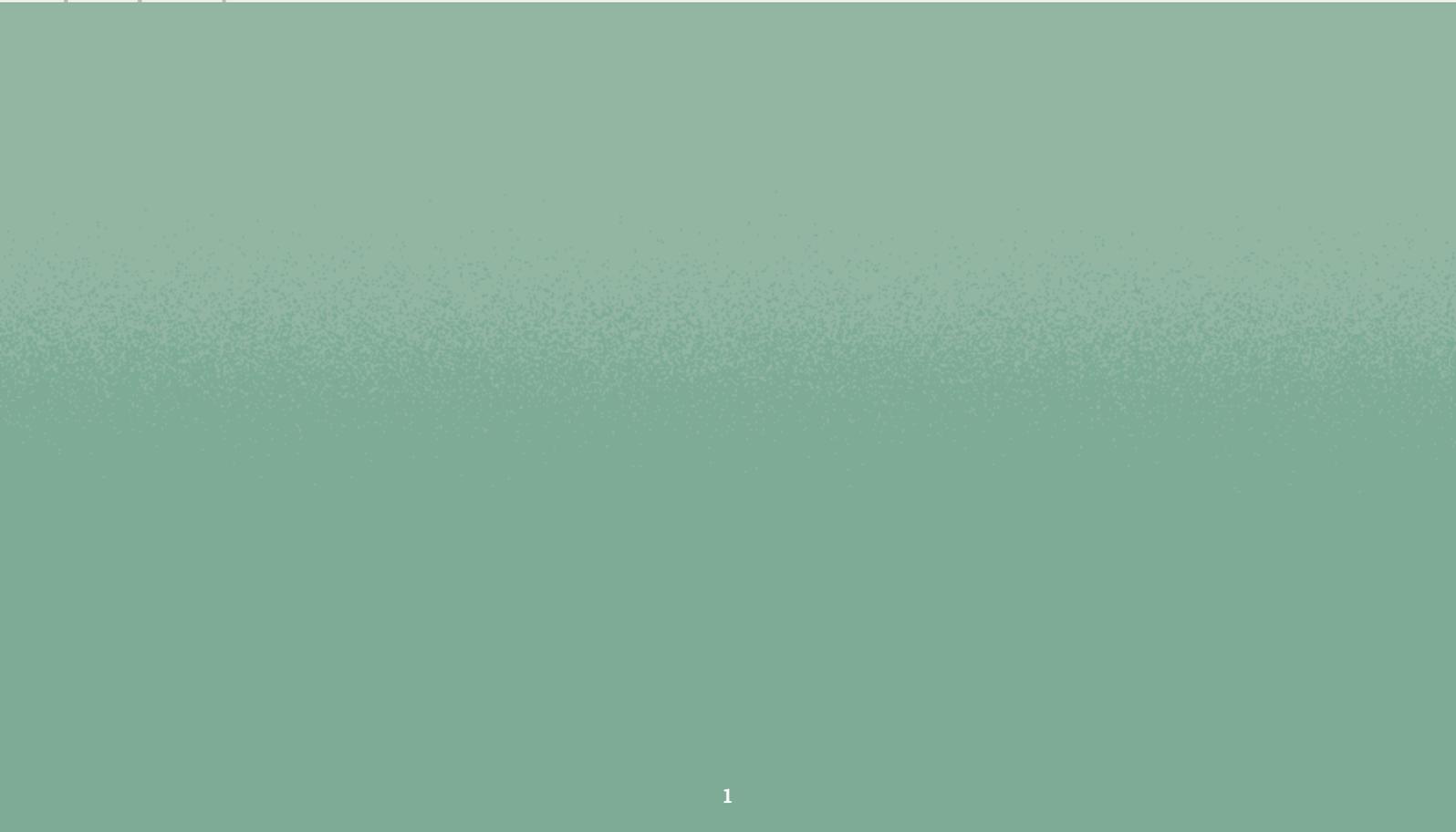
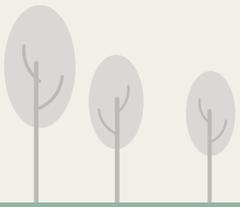
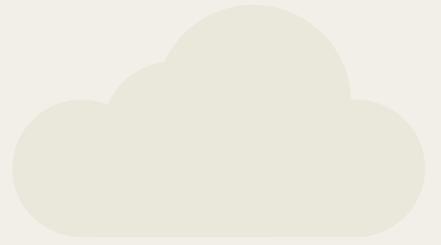
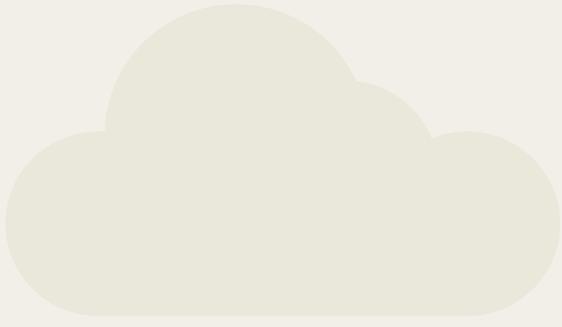
An introduction to Maple Syrup Urine Disease (MSUD)



Innovation in Nutrition

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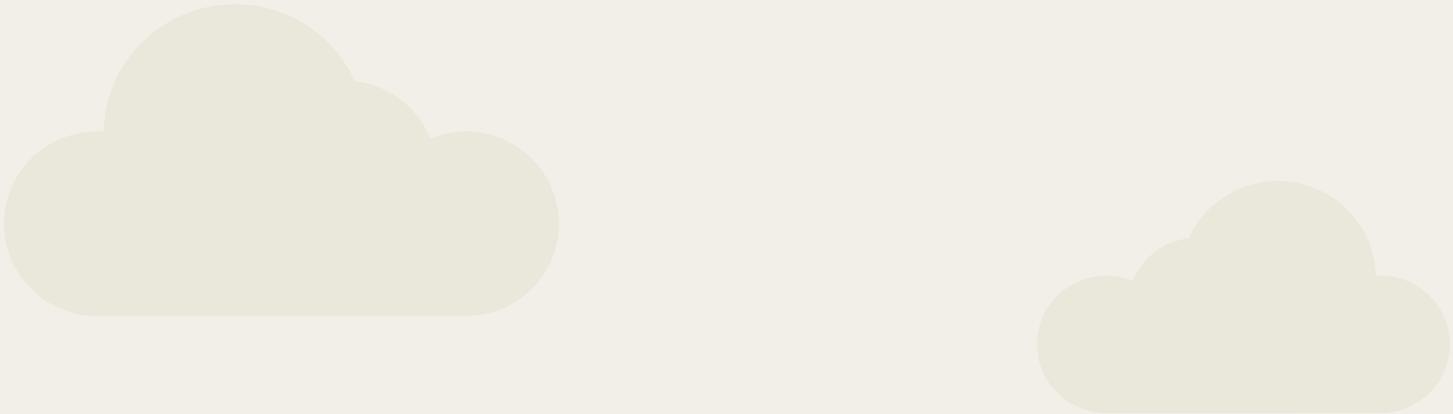
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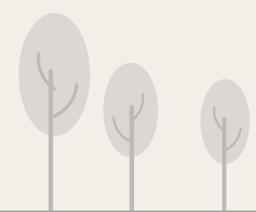
A cartoon illustration of a young girl with long brown hair, wearing a blue shirt and red boots, holding a large white sign. The sign has the text "What is MSUD?" written on it in a bold, dark green font. The background is a simple landscape with green grass, a few trees, and two clouds in a light sky.

**What is
MSUD?**

**Maple Syrup Urine Disease
or MSUD for short.**

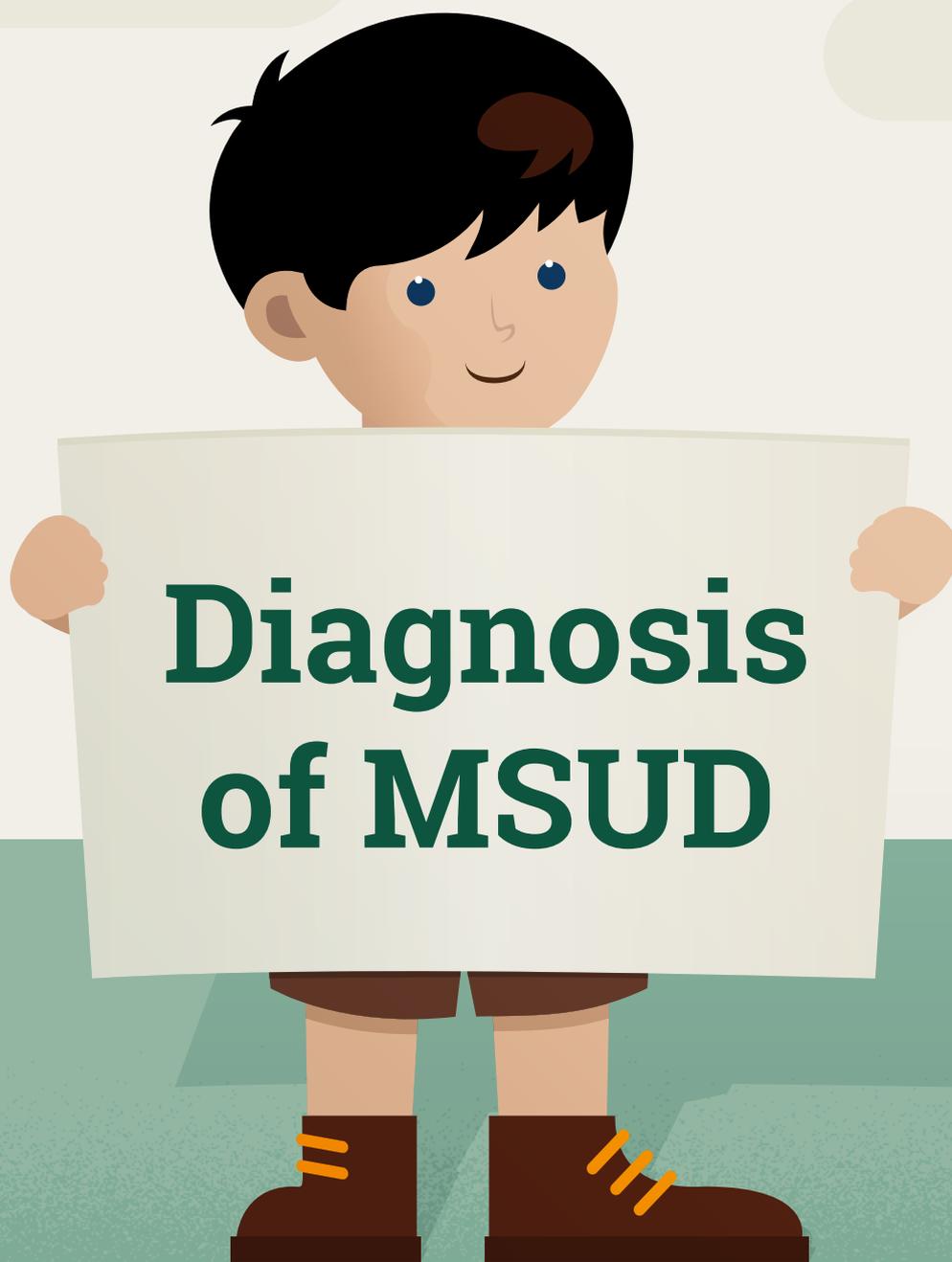


MSUD is a manageable condition which affects the way the body uses protein.

- 
- 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.



Babies 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.



MSUD is diagnosed by a blood test called “The heel prick test”



This blood test is carried out by the midwife around 5 days after birth. Under the national newborn screening programme every baby in the UK and Ireland is screened for this condition.

A cartoon illustration of a woman with long brown hair, wearing a blue top and red boots, holding a large white sign. The sign contains the text "Why does my baby have MSUD?". The background features a green field, a white sky with two clouds, and three stylized trees on the right.

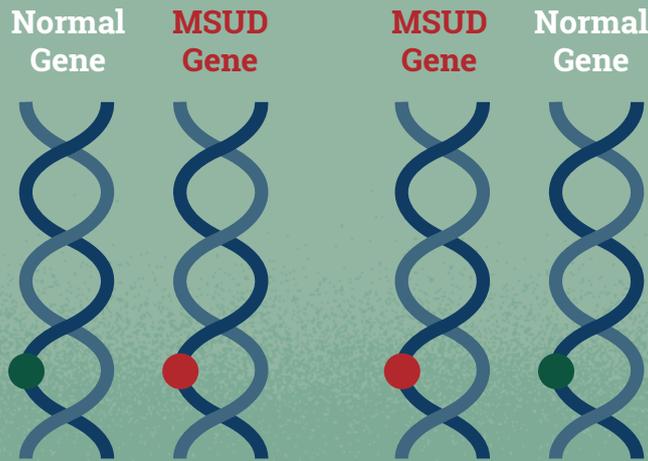
**Why does
my baby
have MSUD?**

MSUD is an inherited condition

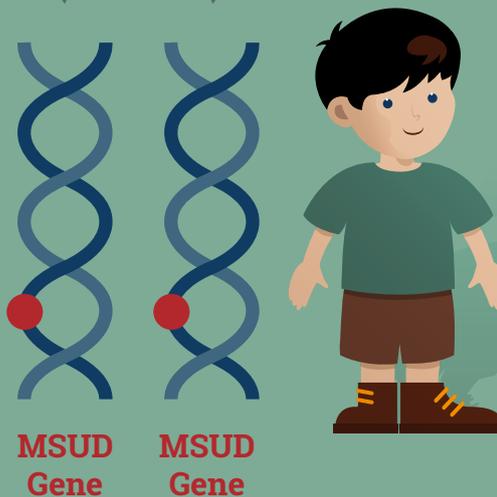
It's nobody's fault and there is nothing you could have done to prevent it.



FATHER
Carrier



MOTHER
Carrier



Child with MSUD

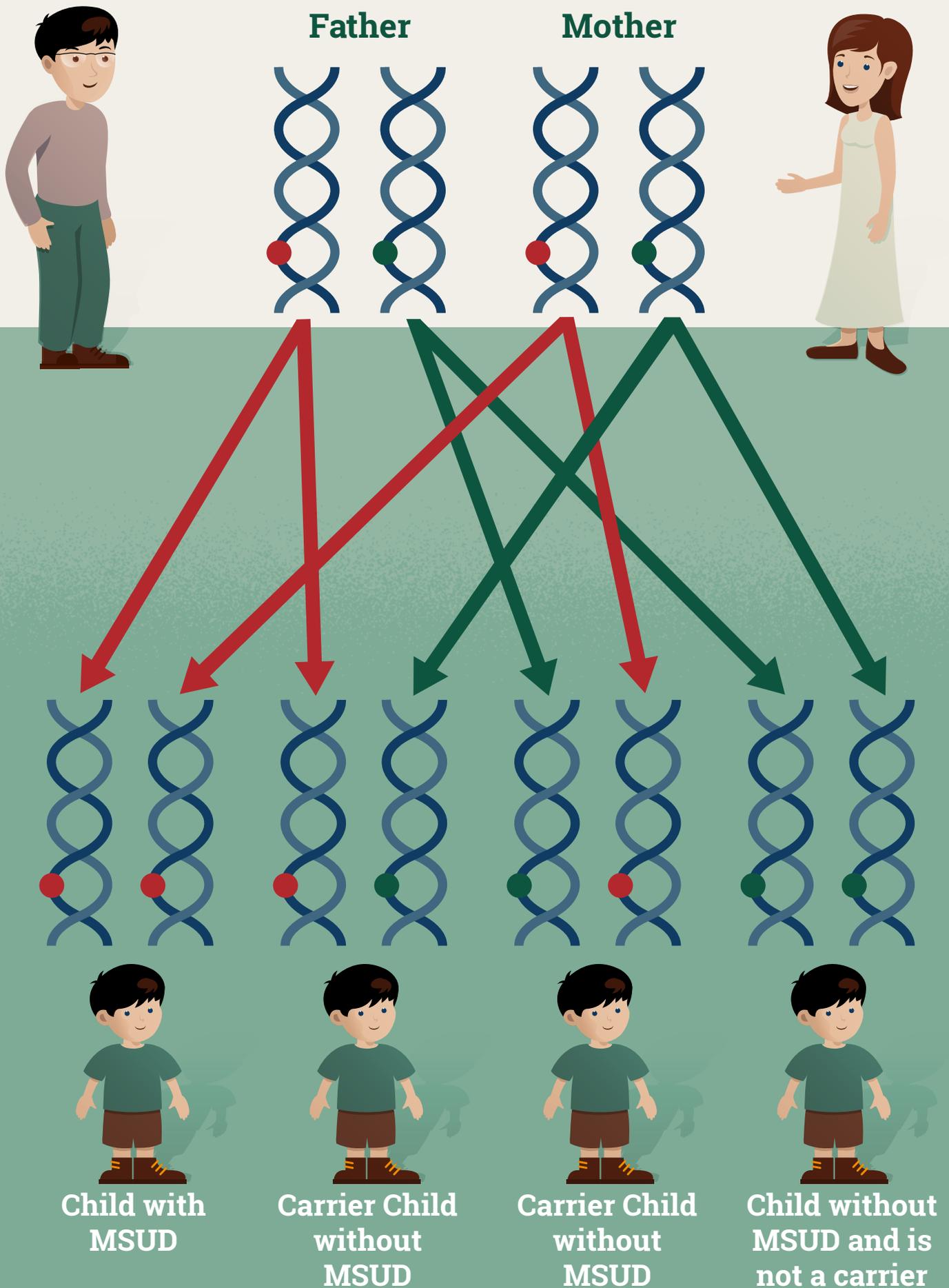
As a parent of a child with MSUD, you have one MSUD gene and one normal one. This is known as being a carrier.

Your child has inherited 2 MSUD genes, one from mum and one from dad.

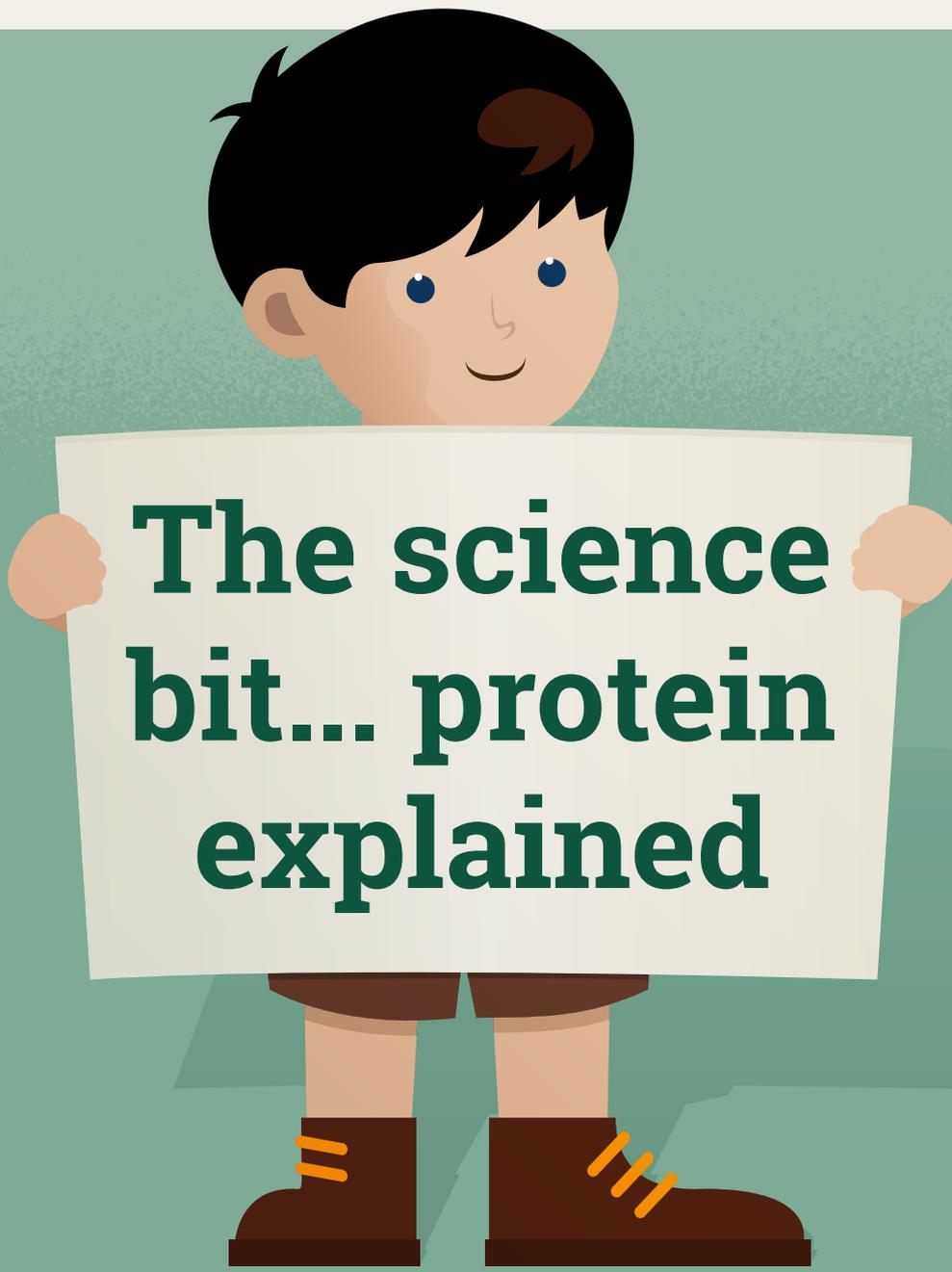
There are a small number of people who are carriers of the MSUD gene



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



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.

A cartoon illustration of a young boy with black hair, blue eyes, and a slight smile. He is wearing brown shorts and brown boots with yellow laces. He is holding a large white sign in front of his chest. The background is a simple landscape with a green field, a white sky, and some stylized clouds and trees.

**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.

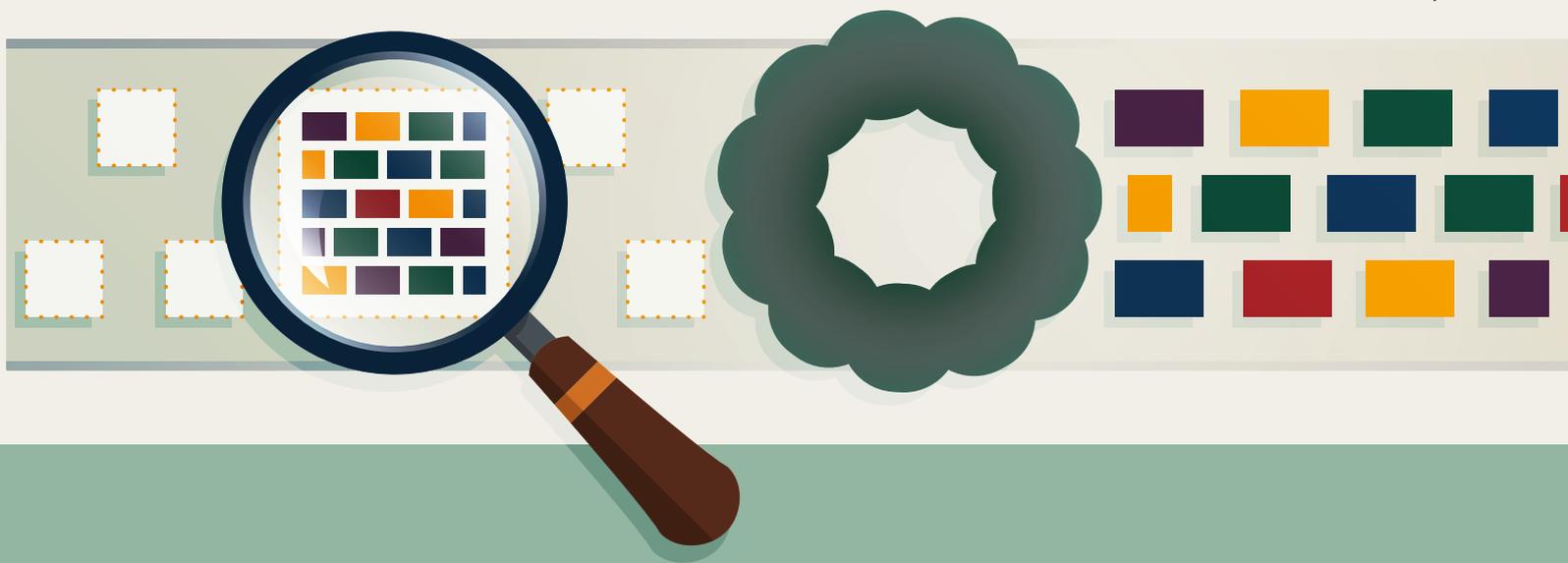
Leucine, Isoleucine and Valine are essential amino acids that a person with MSUD cannot process correctly.

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

What normally happens

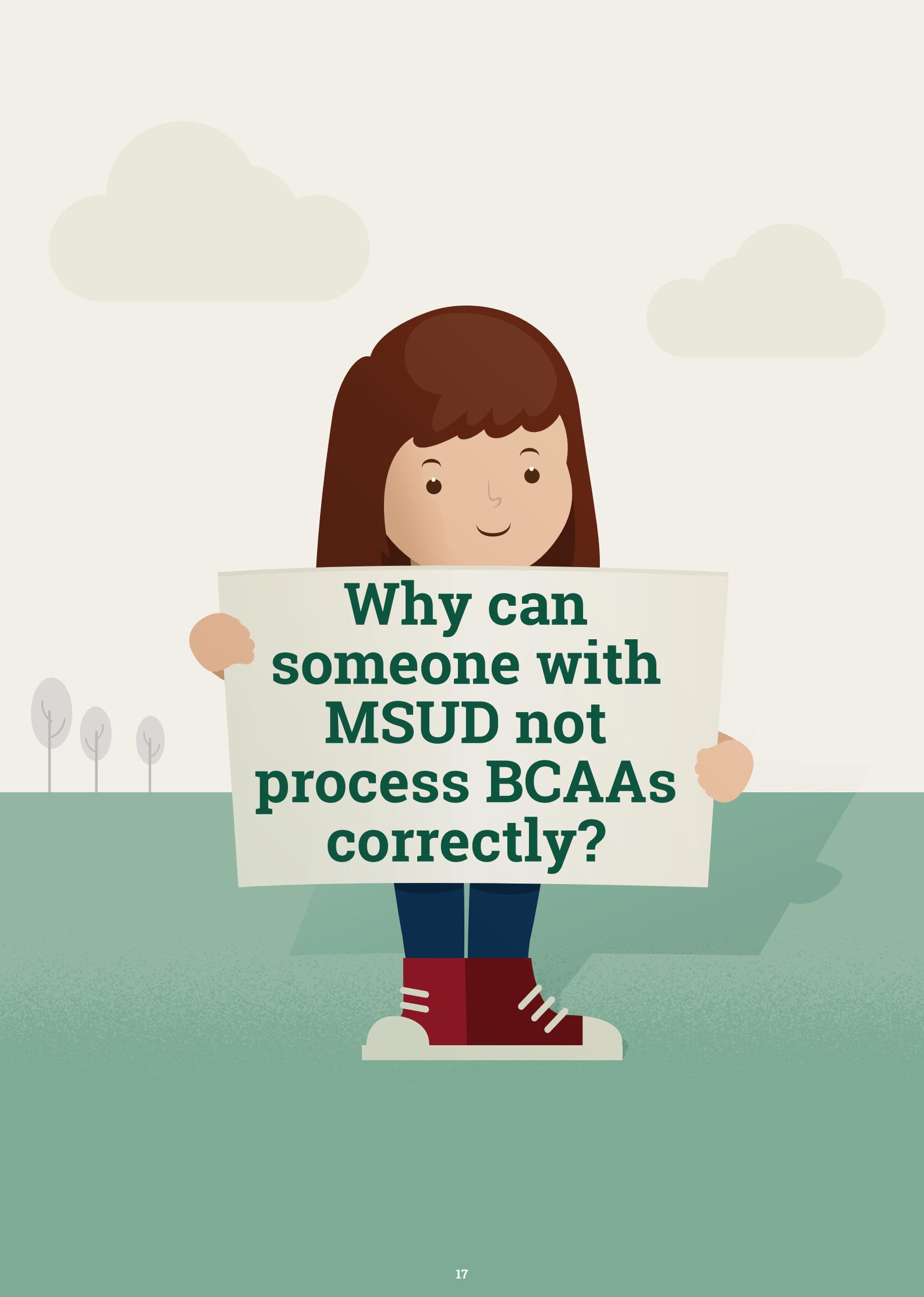
The conversion of protein into amino acids

Protein → Enzymes → Amino acids
(Building Blocks)



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

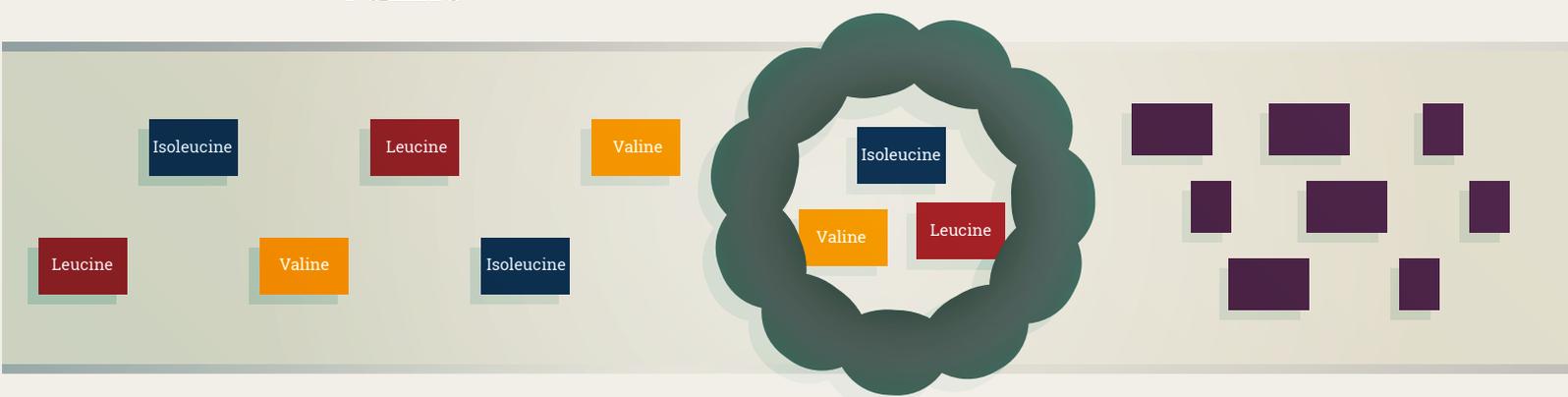
The body uses these amino acids for growing, building up muscle and helping the body stay healthy.

A cartoon illustration of a young girl with long brown hair, wearing a blue shirt and red boots, holding a large white sign. The sign contains the text: "Why can someone with MSUD not process BCAAs correctly?". The background features a green field, three small trees, and two light-colored clouds.

**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.

Leucine, Isoleucine, Valine → Enzymes → 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.



**Management
of MSUD...
the diet
explained**

A Regular Diet

Fruit and Vegetables

Carbohydrate

Very low protein

Moderately high in protein

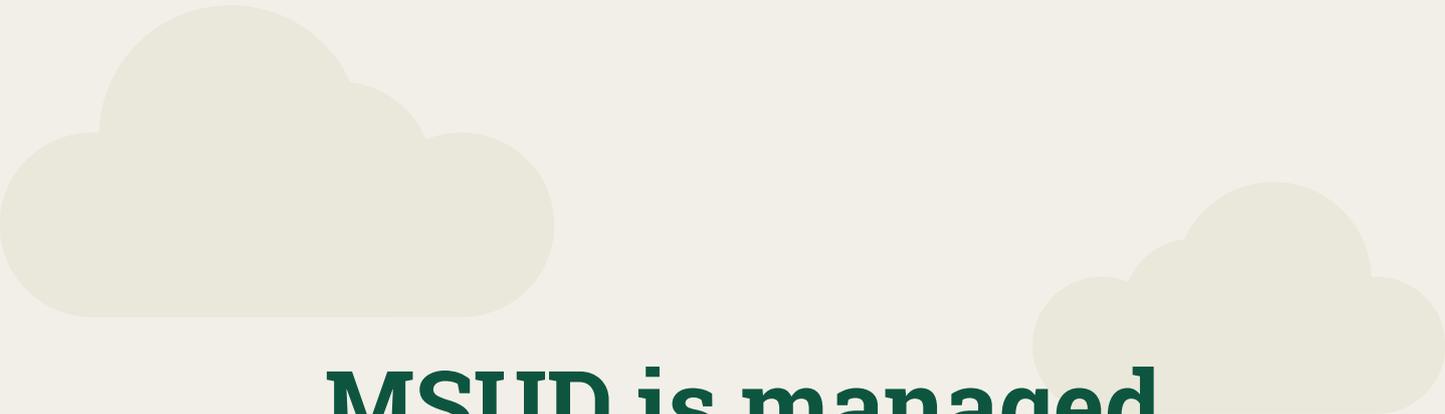
Protein free

High in protein

Fat and sugar

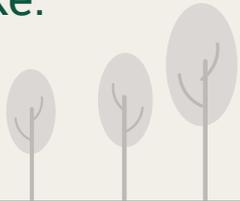
Protein

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

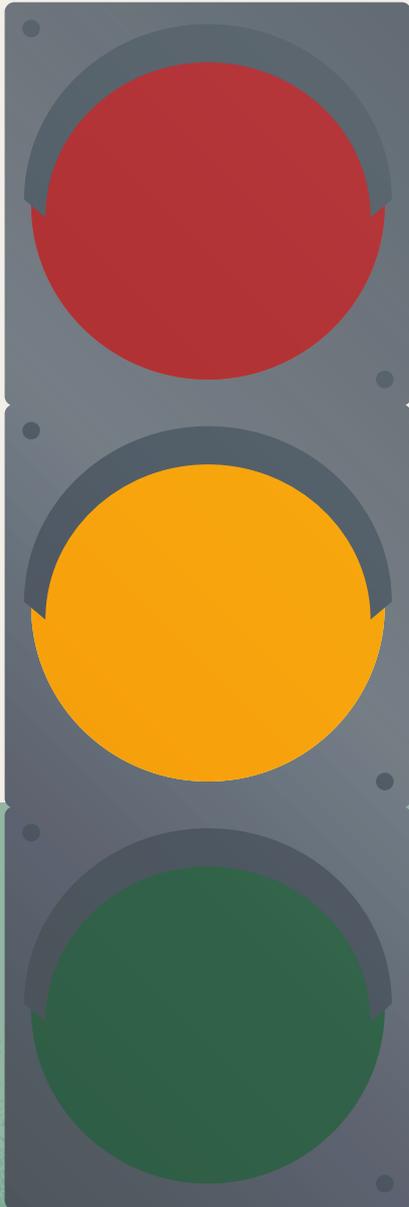
This is because Leucine in the blood is generally higher than Isoleucine and Valine in people with MSUD, so the diet is therefore managed by a controlled Leucine intake.



- 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!

Do not eat these foods

Too high in protein/Leucine

AMBER Go Cautiously!

These foods can be eaten in measured amounts (exchanges)

Contains some protein/Leucine

GREEN Go!

These foods can be eaten freely

Very low in/ free from protein/Leucine

RED STOP!

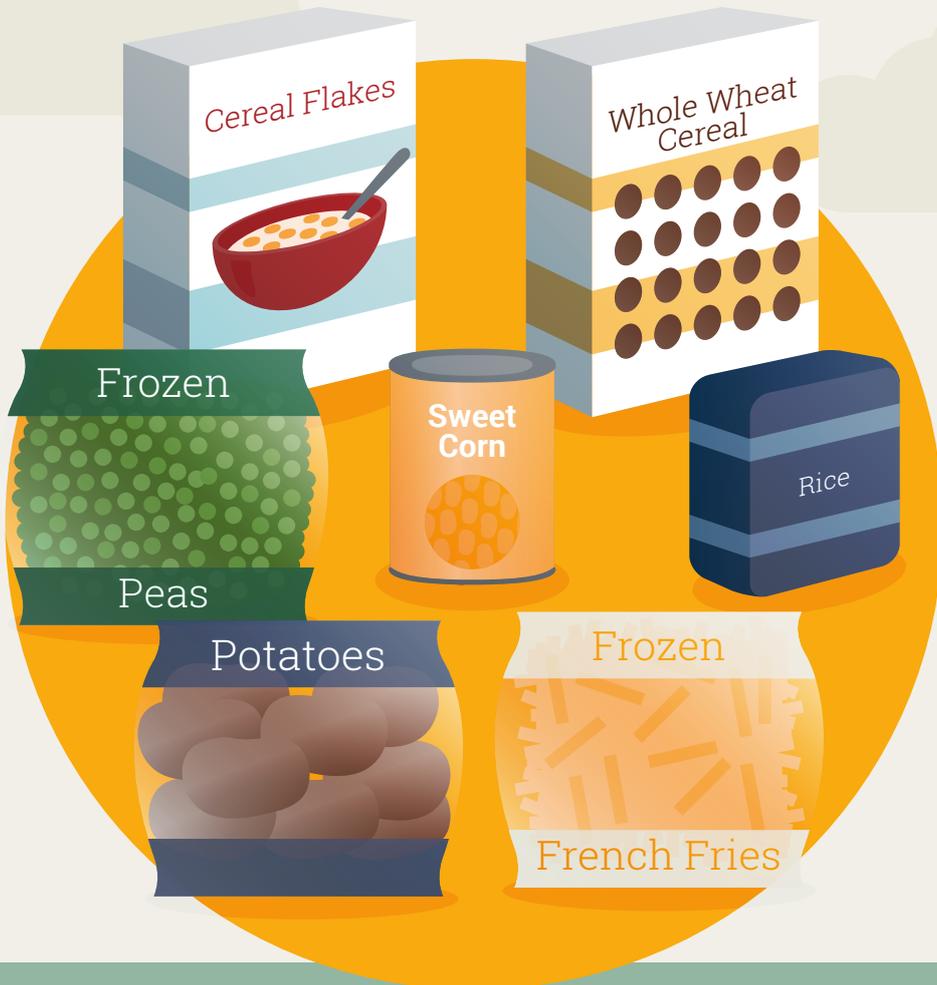
Do not eat these foods



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

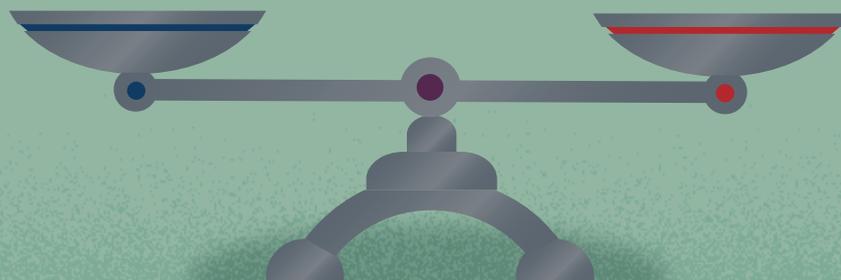
AMBER Go Cautiously!

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

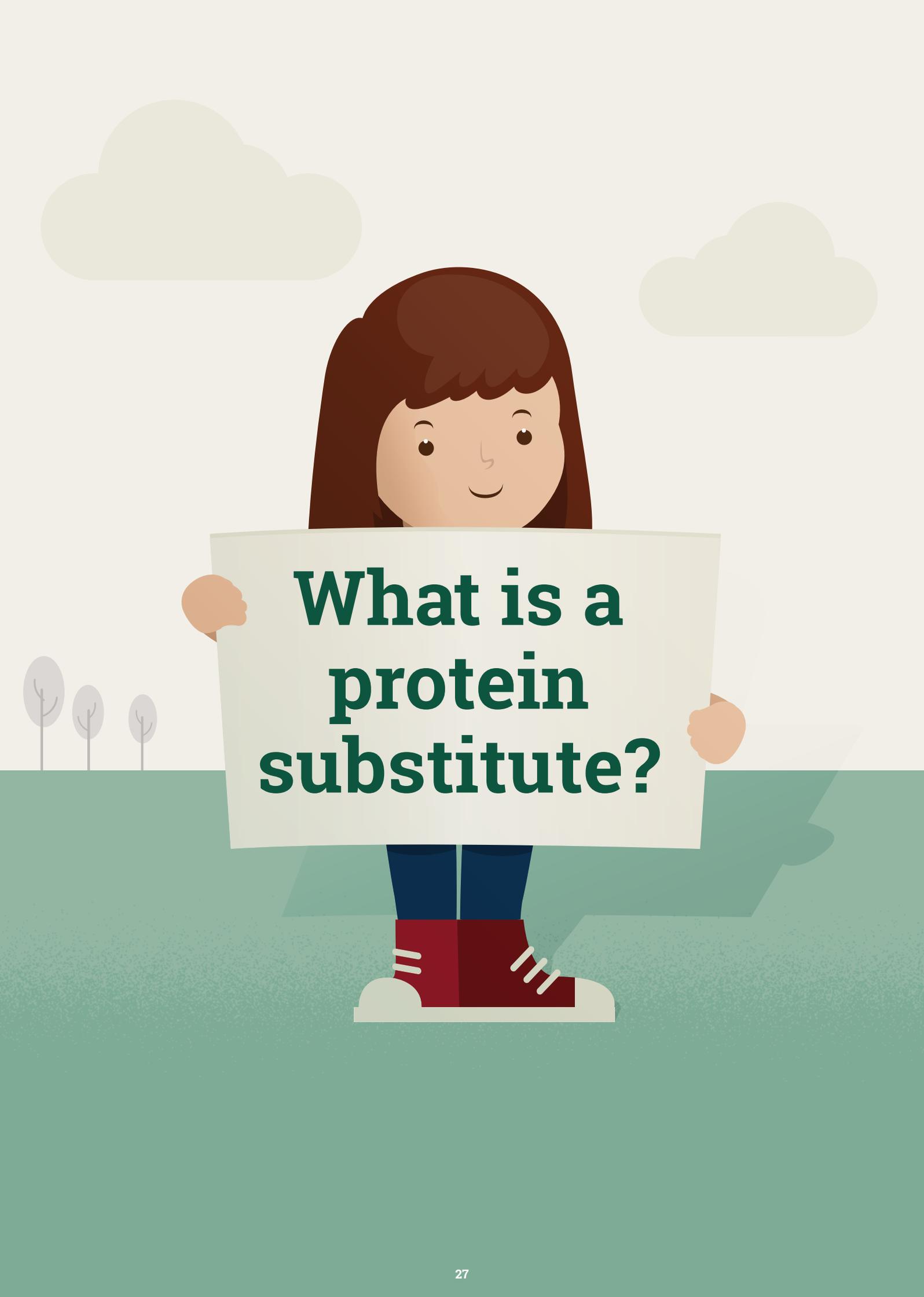
**Special
manufactured
foods**



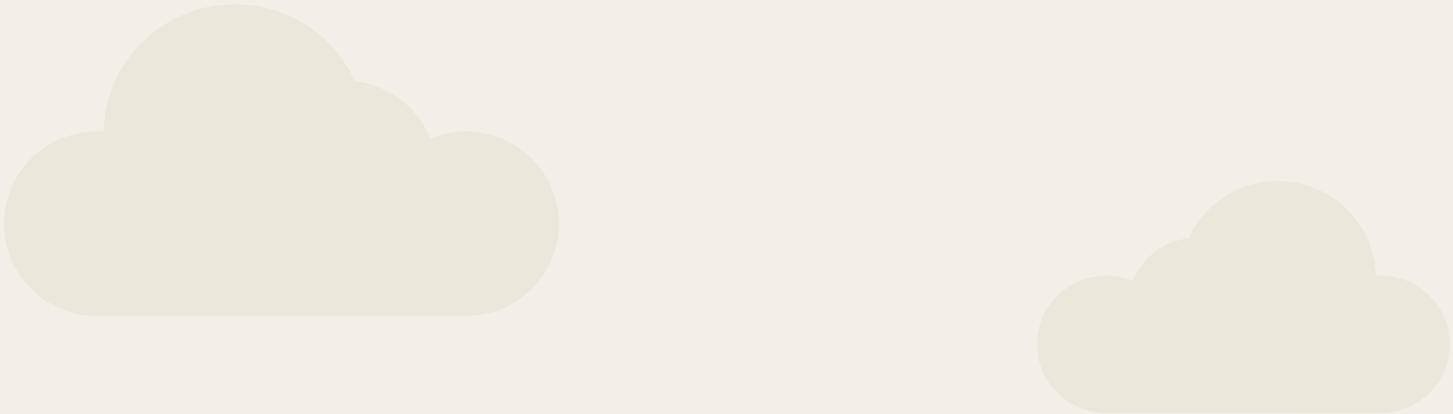
Free foods

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.

A cartoon illustration of a young girl with long brown hair, wearing a blue shirt and red boots, holding a large white sign. The sign contains the text "What is a protein substitute?". The background features a green field, a few trees, and two clouds.

**What is a
protein
substitute?**



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

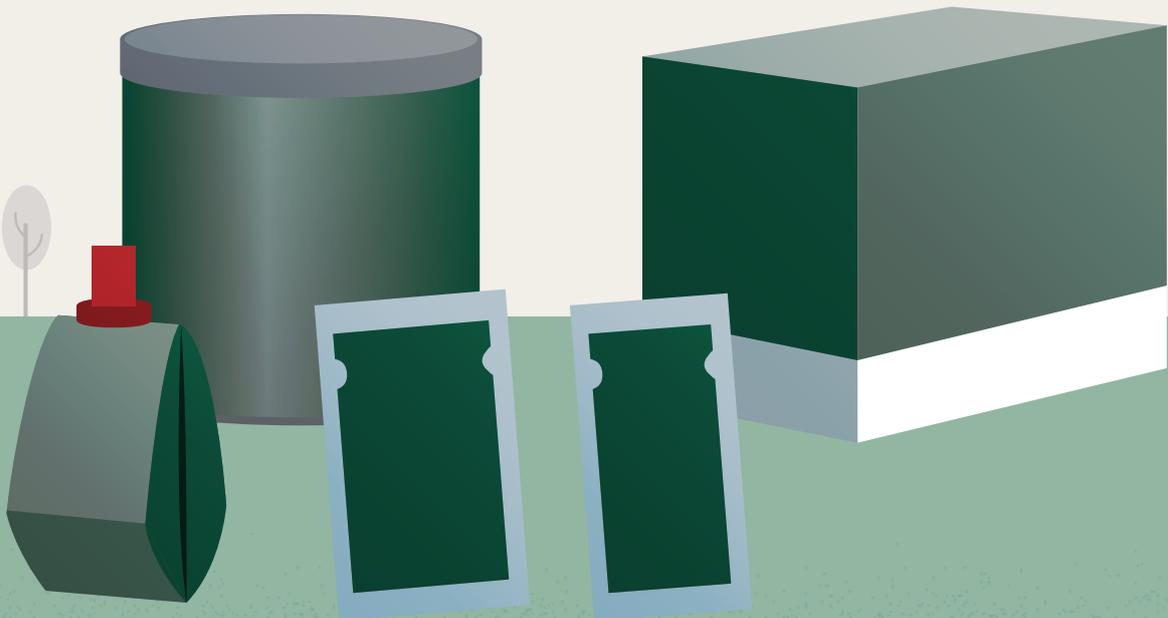
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- A protein substitute is a specially made medical food, which can come in various formats like liquids or powders that can be made up to various consistencies.
 - A protein substitute contains NO Leucine, Isoleucine or Valine. But it contains all 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.

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.



A cartoon illustration of a young boy with black hair, blue eyes, and a slight smile. He is wearing brown shorts and brown boots with yellow laces. He is holding a large white sign in front of his chest. The background is a simple landscape with a green field, a few grey trees, and two light green clouds in the sky.

**How will I
feed my new
born baby?**

Infants: breastfeeding



Step 1: Once your child is diagnosed, you will be seen by a dietitian. Your baby will be given a special 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 breastmilk taken by the baby. Breastmilk provides limited but essential BCAAs for normal growth.

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

Infants: non-breastfeeding (bottle feeding)



Step 1: Once your child is diagnosed, you will be seen by a dietitian. Your baby will be given a special 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 baby formula will be given along with the BCAA-free formula to provide the limited BCAAs essential for normal growth.

A cartoon illustration of a young girl with long brown hair, wearing a blue top and red boots with white laces. She is holding a large white sign in front of her chest. The sign has the word "Weaning" written on it in a bold, dark green font. The background is a simple landscape with a green field, three small trees on the left, and two light-colored clouds in the sky.

Weaning

When to start weaning*

Your dietitian will advise you when to start solid foods,* but it will be at the same time as babies 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.

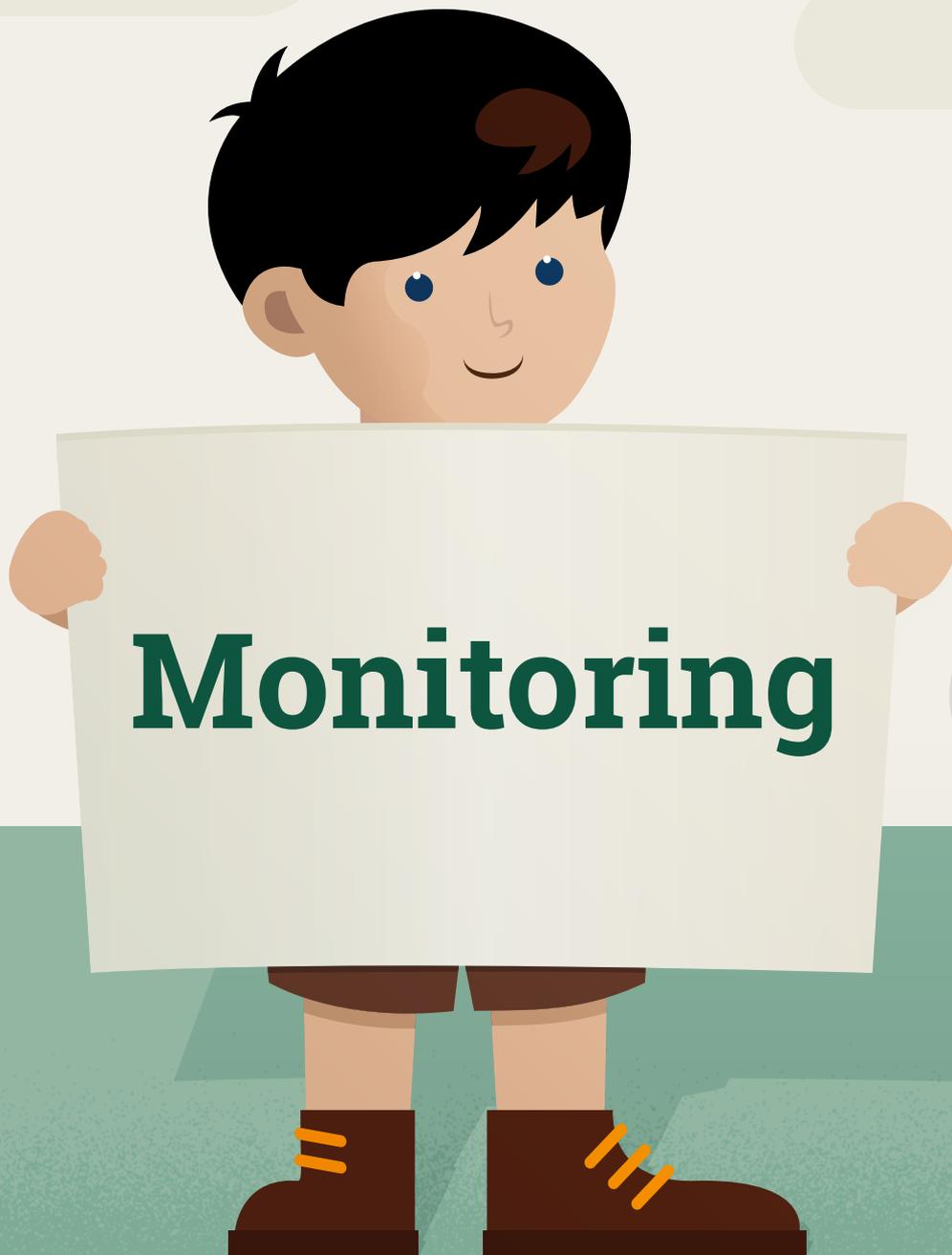
* Weaning may occur a little earlier than 6 months, depending on your baby, but it should never be started before 17 weeks.

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.

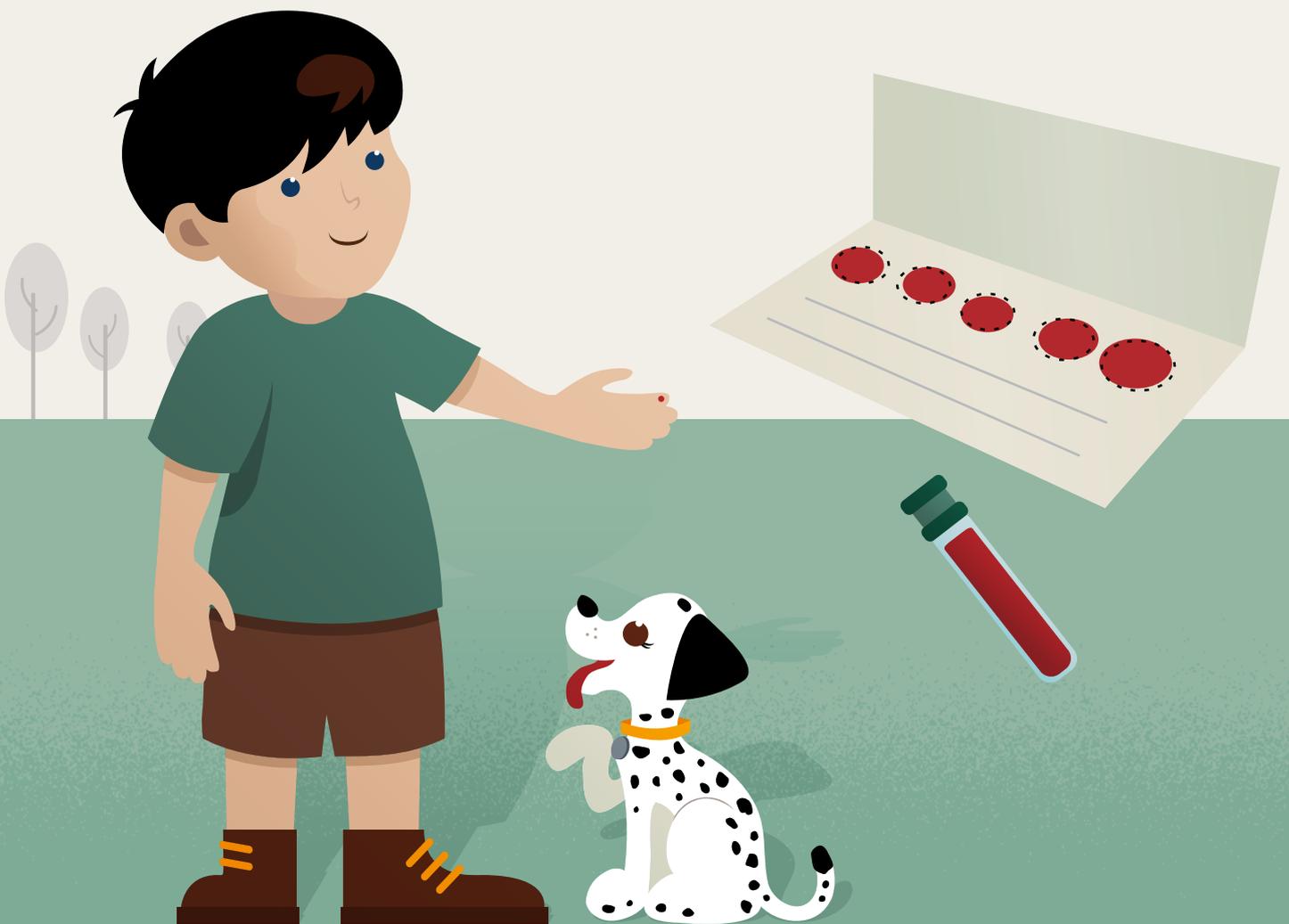


Leucine free foods should still be included in the diet as well. Your child will also continue their gradual switch from their BCAA-free formula to second stage protein substitute.



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 babies 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.

An illustration of a woman with long brown hair, wearing a blue top and red boots, holding a large white sign. The sign contains the text 'As your child grows up'. The background features a green field, a white sky with two light green clouds, and three stylized trees on the right side.

**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.

Notes

