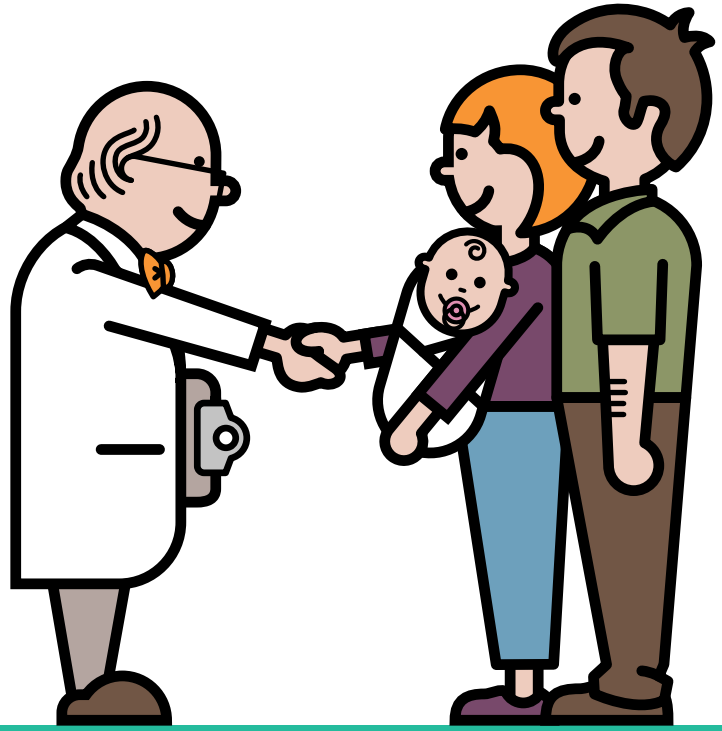
Maple Syrup Urine Disease

MSUD

Information for families after a positive newborn screening



Adapted by the Dietitians Group BIMDG

BIMDG
British Inherited Metabolic Diseases Group

BASED ON THE ORIGINAL TEMPLE WRITTEN BY BURGARD AND WENDEL

Reviewed & revised for North America by: A. Huber

This version of the TEMPLE tool, originally adapted by the Dietitians group of the BIMDG for use within the UK and Ireland, has been further adapted by Nutricia North America for use within United States and Canada. This version no longer represents clinical or dietetic practice in the UK or Ireland.

TEMPLE
Tools Enabling Metabolic Parents LEarning

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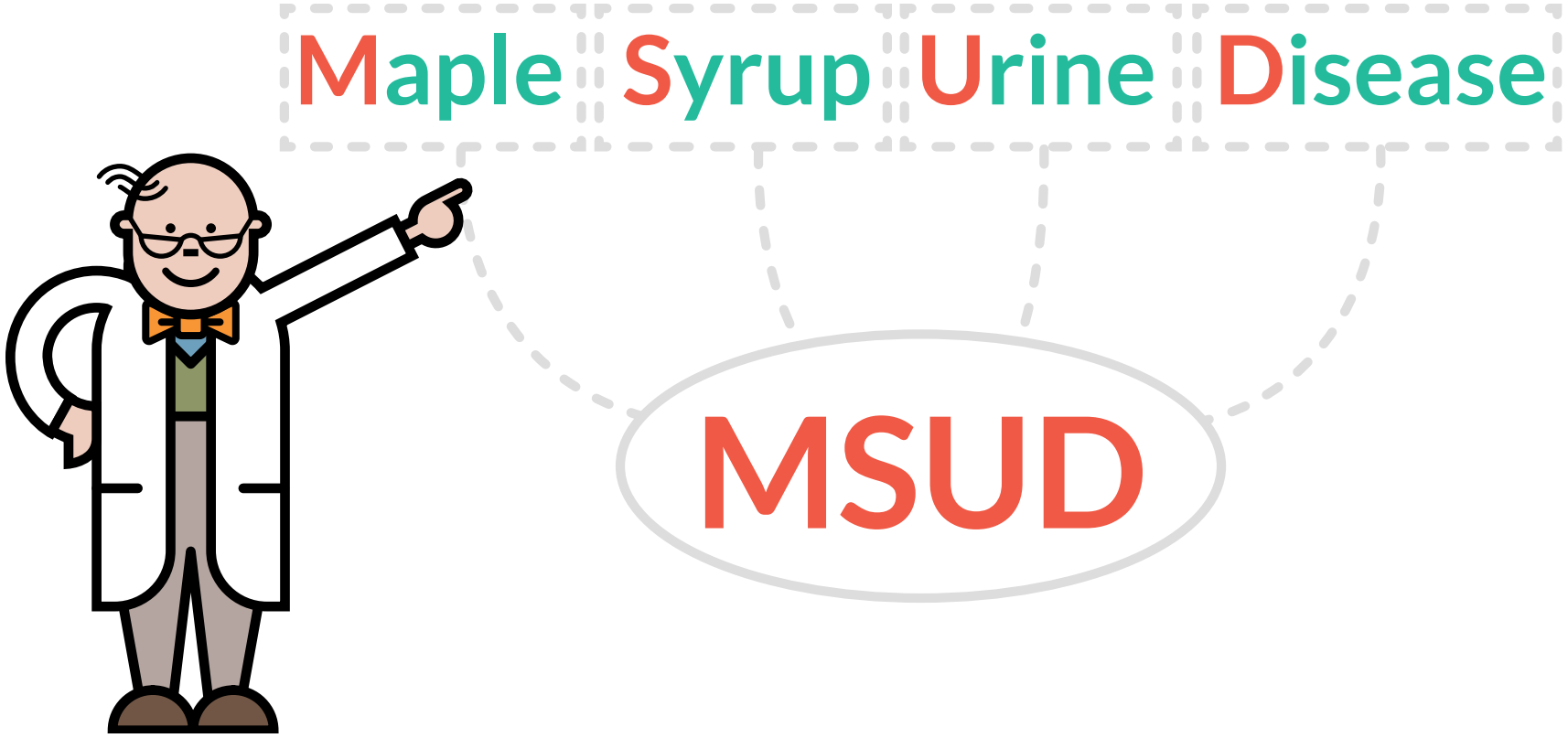
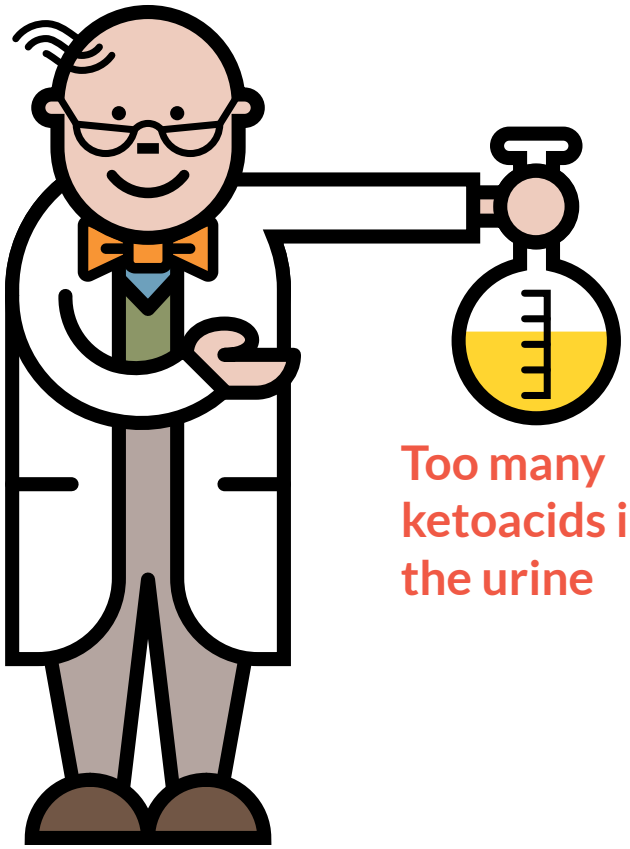
What is MSUD?

MSUD stands for maple syrup urine disease.

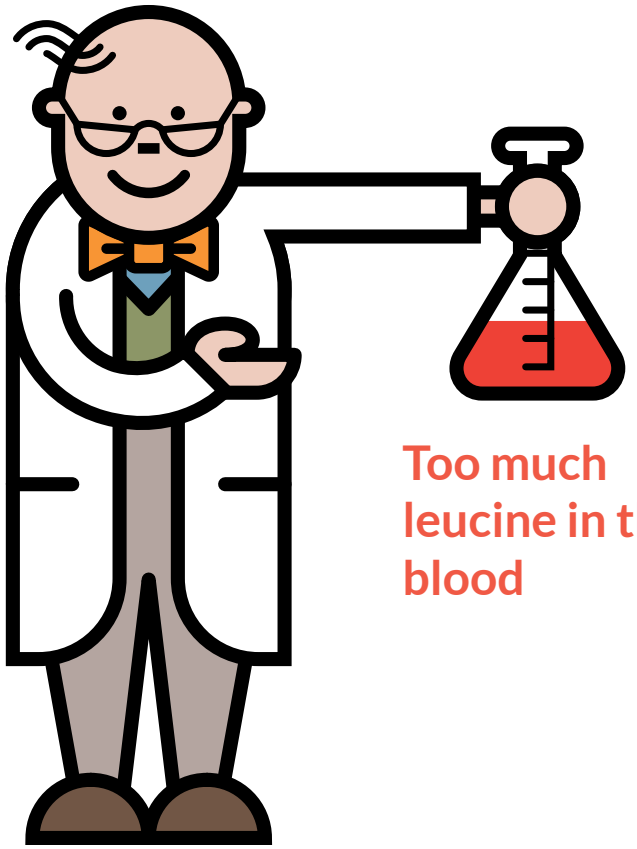
It is an inherited metabolic condition.



What is MSUD?



Too many ketoacids in the urine



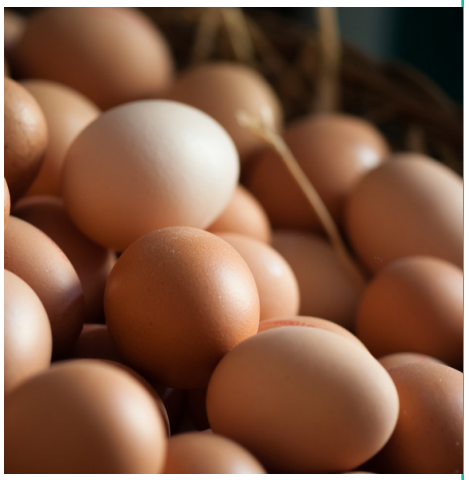
Too much leucine in the blood

How does MSUD affect the body?

MSUD affects the way the body breaks down protein.

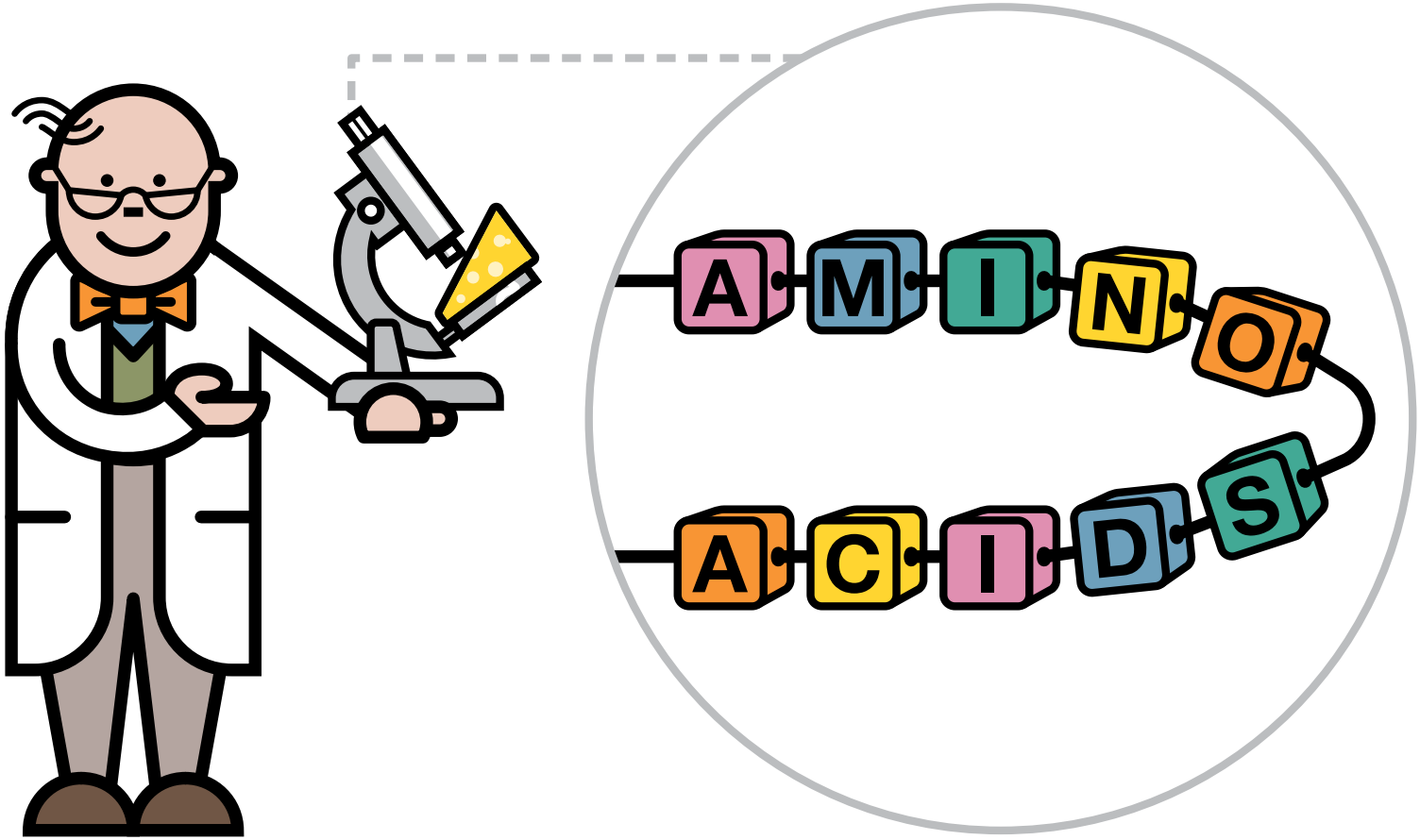
Protein is found in our bodies and in many foods. The body needs protein for growth and repair.



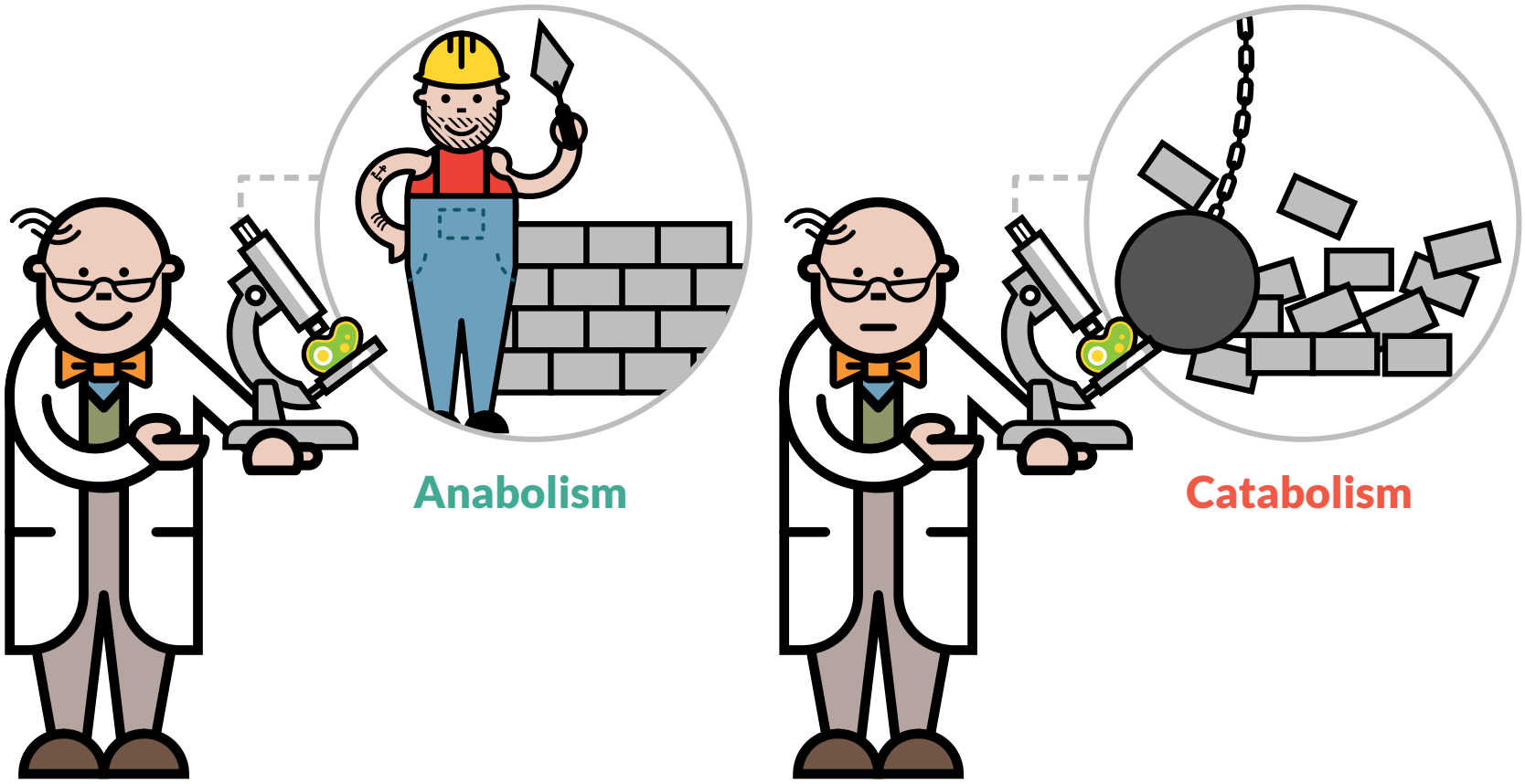
What is protein?

Protein consists of chains of many smaller units called amino acids.



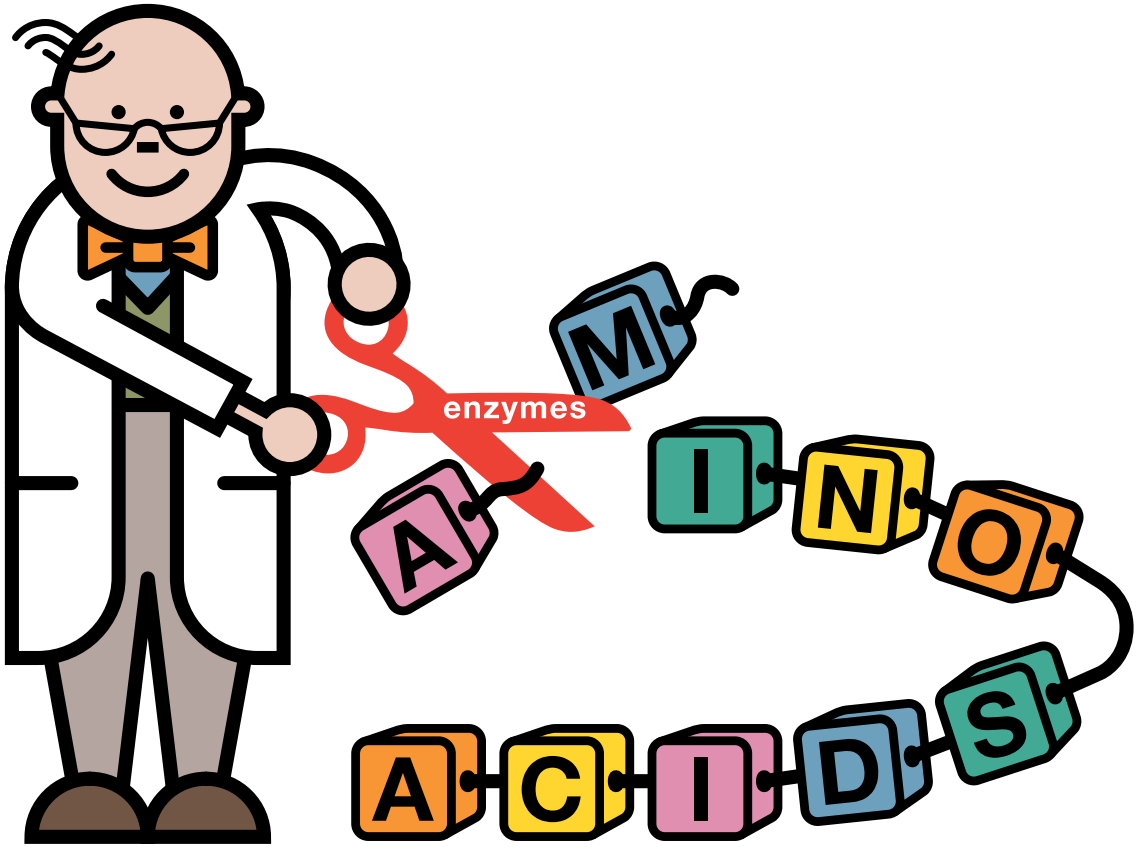
Protein metabolism

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



What do enzymes do?

Enzymes help with metabolism by functioning like scissors.
They break down proteins into smaller parts, including amino acids.

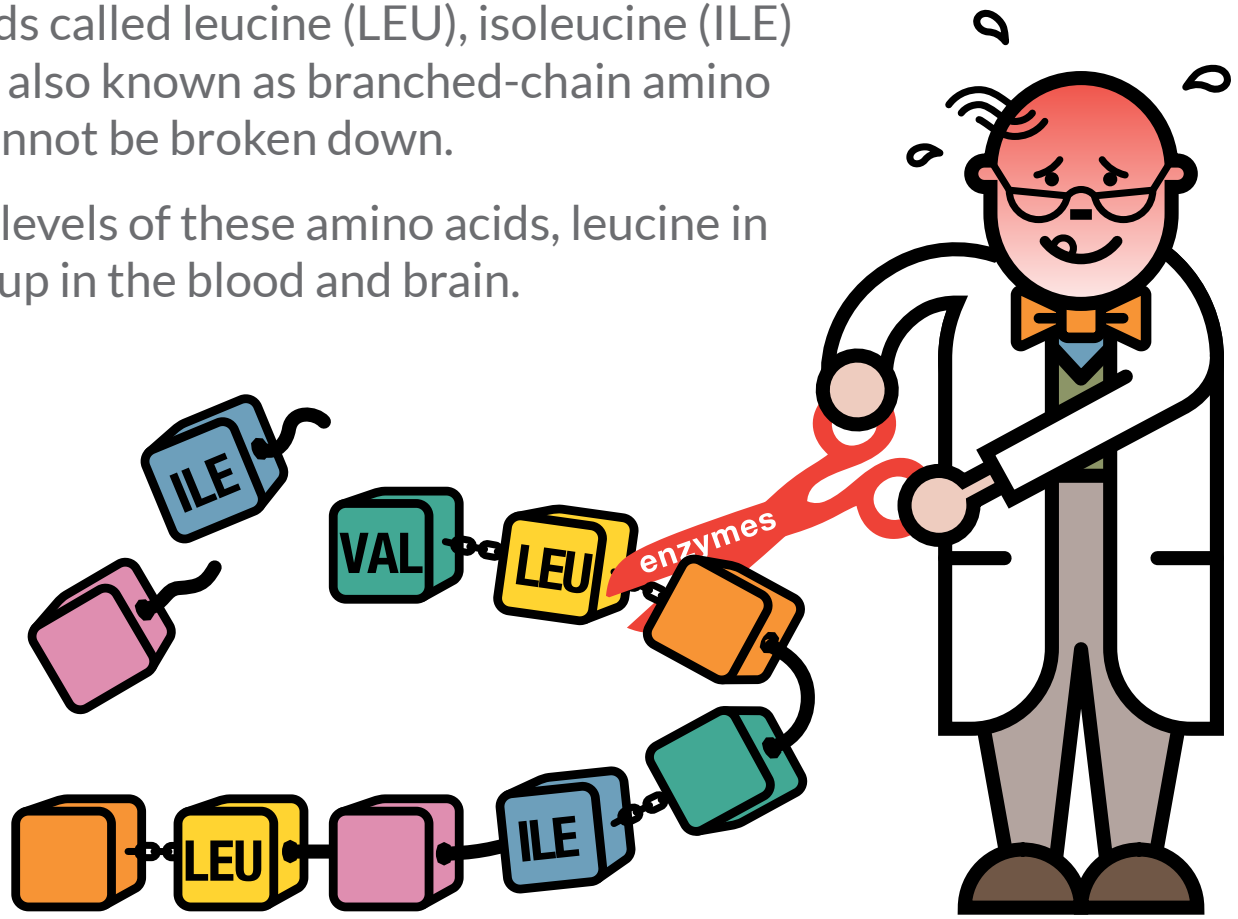


What happens in MSUD?

MSUD is due to deficiency of an enzyme complex called **branched-chain ketoacid dehydrogenase (BCKAD)**.

Three amino acids called leucine (LEU), isoleucine (ILE) and valine (VAL) also known as branched-chain amino acids (BCAA), cannot be broken down.

This means that levels of these amino acids, leucine in particular, build up in the blood and brain.



What can go wrong in MSUD?

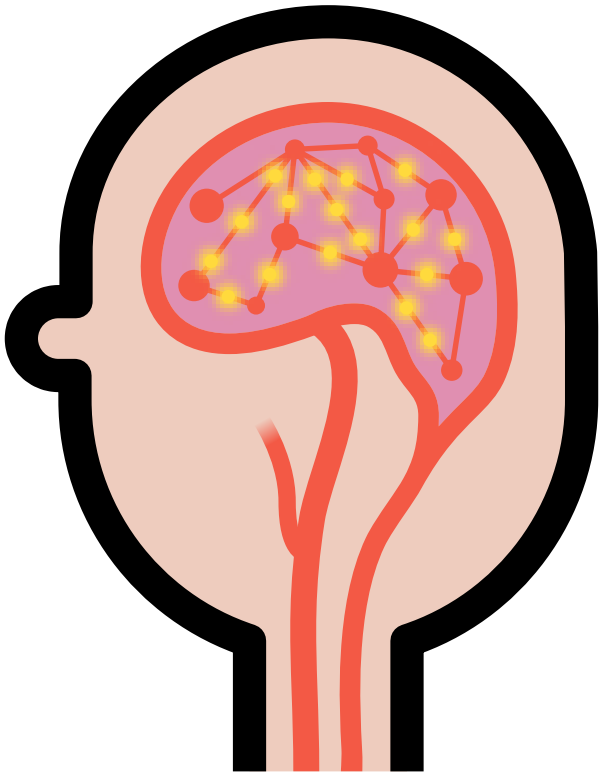
Leucine and other substances build up to high toxic levels in the blood and brain.

It may affect babies and children in different ways.

Left unmanaged, some babies develop symptoms such as sleepiness and irritability in the first few days. Babies may deteriorate and even go into a coma which can lead to brain damage.

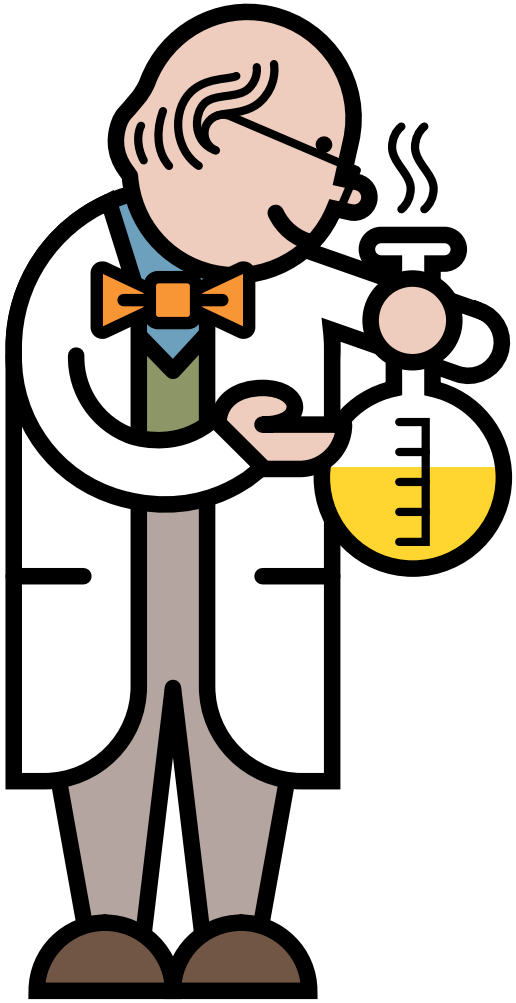
Other children may develop symptoms at a later stage, and this may be caused by an infection such as the flu or vomiting illness.

Early management can prevent brain damage and learning difficulties.



What about other symptoms?

The urine and sweat may smell sweet (like maple syrup) before diagnosis or when unwell.



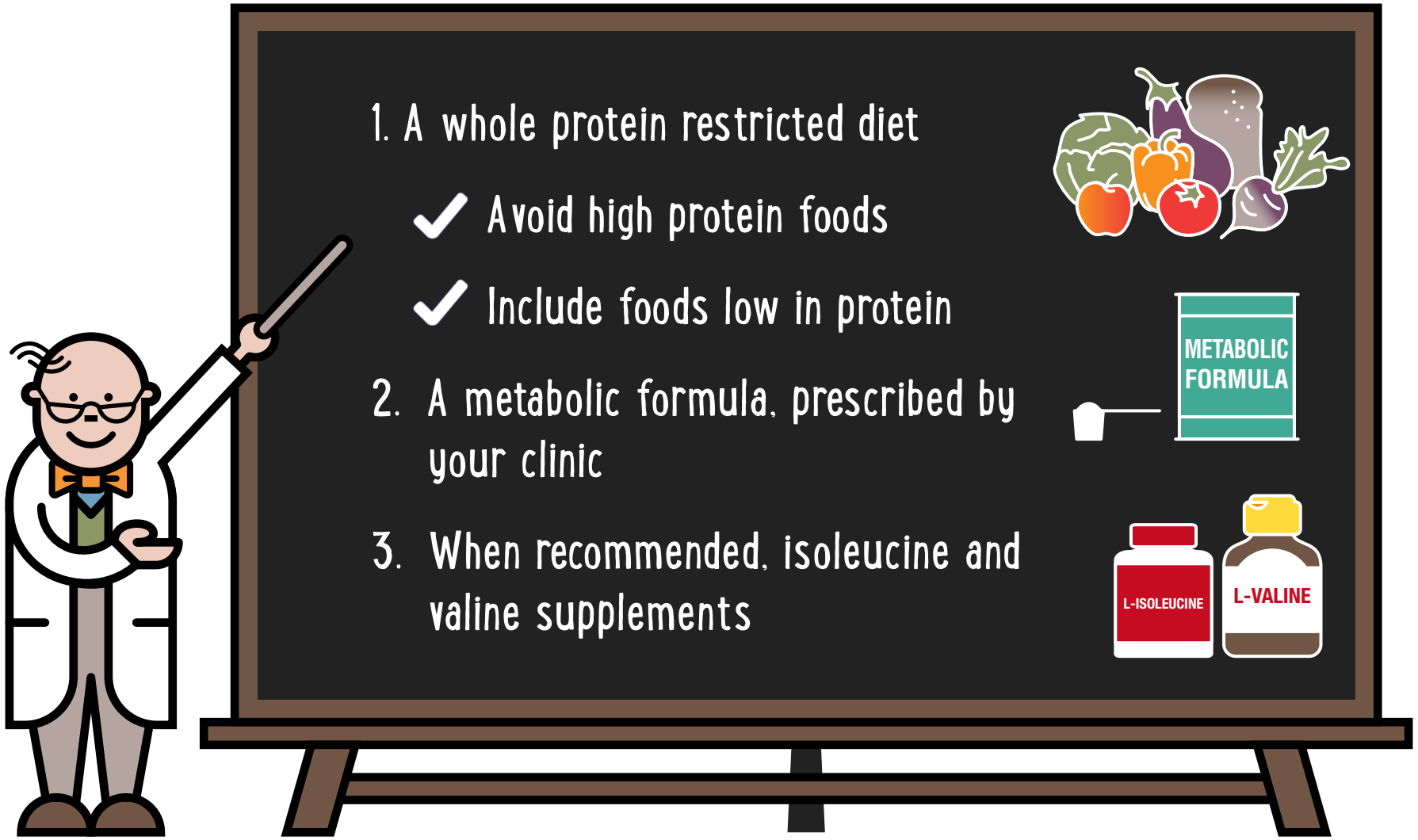
How is MSUD diagnosed?

As part of newborn screening, a few drops of blood are collected.

The blood sample is then analyzed and leucine, isoleucine and valine levels are measured.

A high level of these amino acids could mean your child has MSUD, which will prompt your clinician to do further testing to confirm the diagnosis.

How is MSUD managed day-to-day?



Avoid high protein foods

Foods rich in protein, and therefore BCAA*, should be avoided. This includes **meat, fish, eggs, cheese, milk, bread, pasta, nuts, soy and tofu**.

*Remember BCAA are the amino acids leucine (LEU), isoleucine (ILE) and valine (VAL)

Supported by NUTRICIA as a service to metabolic medicine
Dietary management of the condition should only be done under medical supervision.

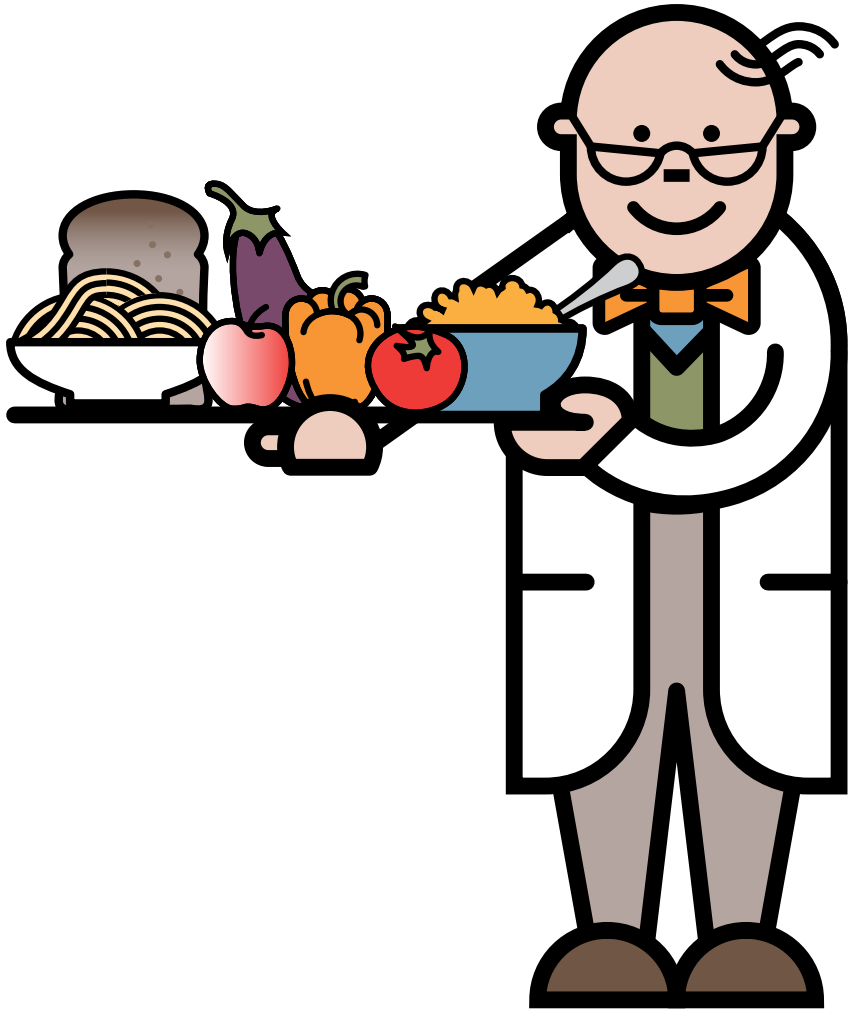
Include foods low in protein

These are foods that contain small amounts of BCAA* which can be used in typical quantities.

They include many fruits and vegetables, and special low protein foods.

They provide:

- An important source of energy
- Variety in the diet



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Low protein cooking

Cooking low-protein meals for your child can still be appealing to the eye and taste good.

There are many low-protein cookbooks to choose from. Your dietitian may be able to recommend a few favorites.

Feeding your baby with metabolic formula

BCAA* are essential for normal development and therefore a limited and controlled amount must be taken daily.

Breast milk or standard infant formula will provide the BCAA required by your baby prior to the introduction of solids, generally around 4-6 months of age.

Your baby will also need a special metabolic formula to provide protein without BCAA.

Your dietitian will determine how much breast milk or standard infant formula and metabolic formula to offer.

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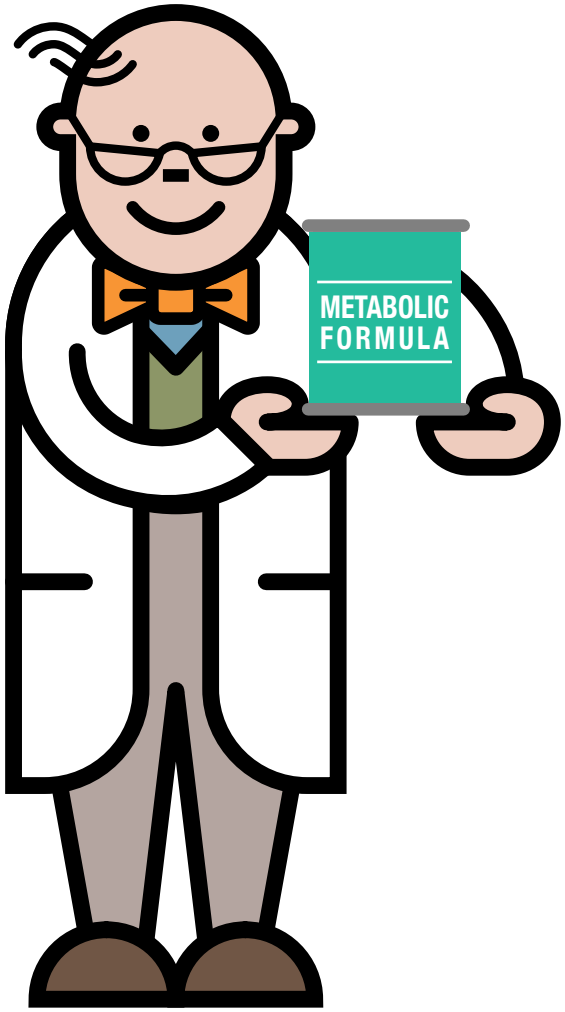
BCAA*-free metabolic formula

Leucine-, Isoleucine-, and Valine-free metabolic formula is an essential part of meeting your baby's nutritional requirements.

Like breast milk or standard infant formula, metabolic formula has carbohydrate, fat, vitamins, minerals and protein in the form of amino acids without BCAA.

Metabolic formula, plus the prescribed amount of BCAA, allows your baby to get all the nutrients he or she needs to grow.



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Tracking BCAA*

As your baby starts to eat solids your clinic will work with you to track BCAA.

Foods must be weighed or measured using household measures (1 cup, 1 tablespoon, etc.) to determine BCAA content.

Your clinic can help you find the best tools to help determine the BCAA content of foods.

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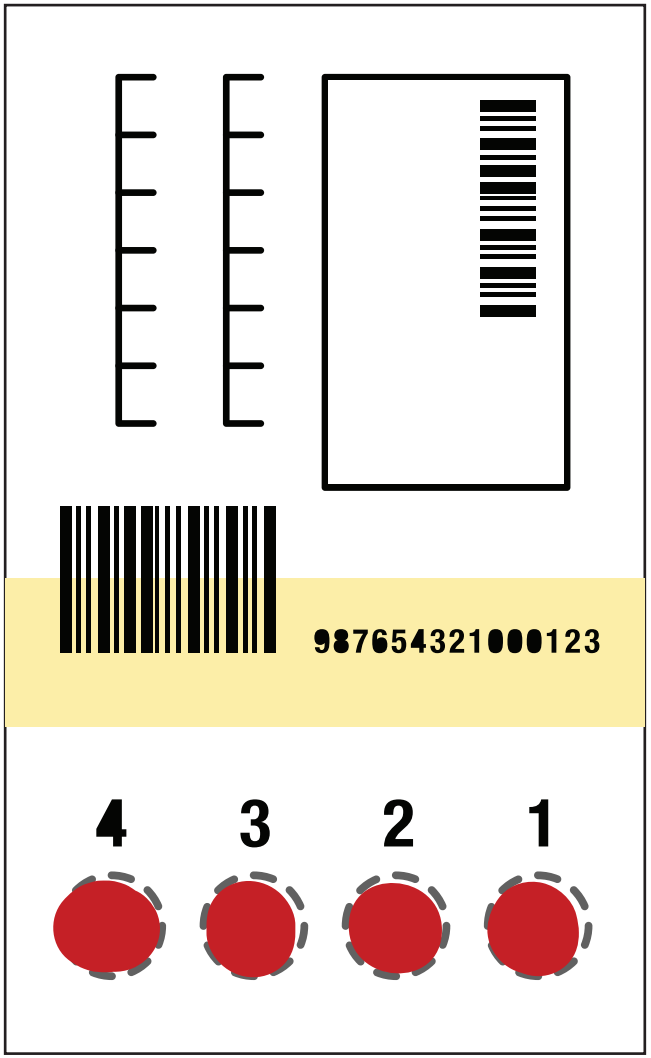
How is MSUD monitored?

Regular blood tests are taken at home or in the clinic and reviewed by a dietitian.

The sample is tested for the amount of BCAA* it contains.

The metabolic dietitian will contact you with the result and discuss any changes in management.



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Metabolic crisis

A '**metabolic crisis**' causes a buildup of leucine and other toxic substances.

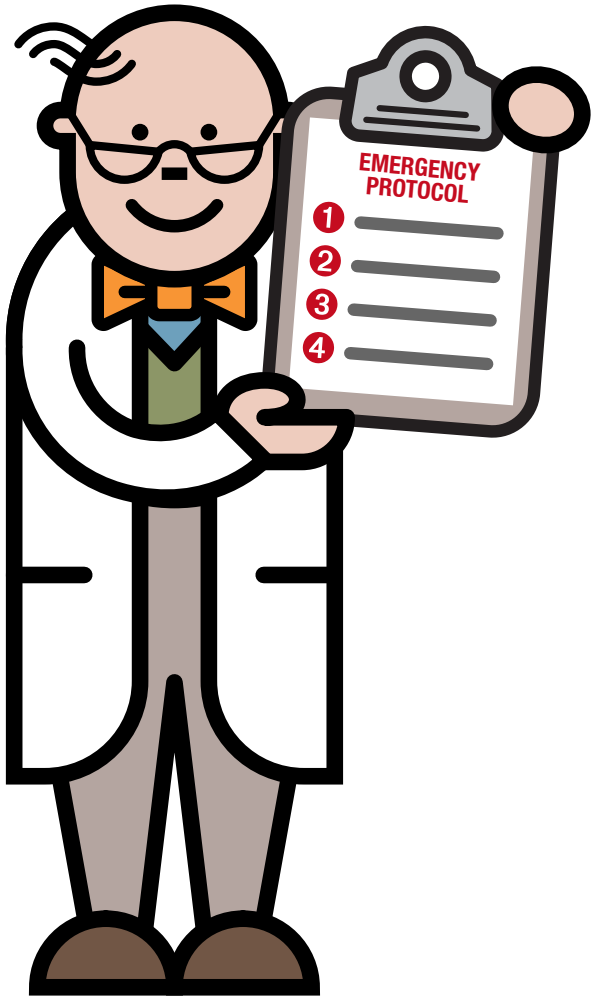
It is usually triggered by childhood infections or viruses causing high temperatures, vomiting, and diarrhea.

It is important to manage a metabolic crisis quickly and properly.

How is MSUD managed during illness?

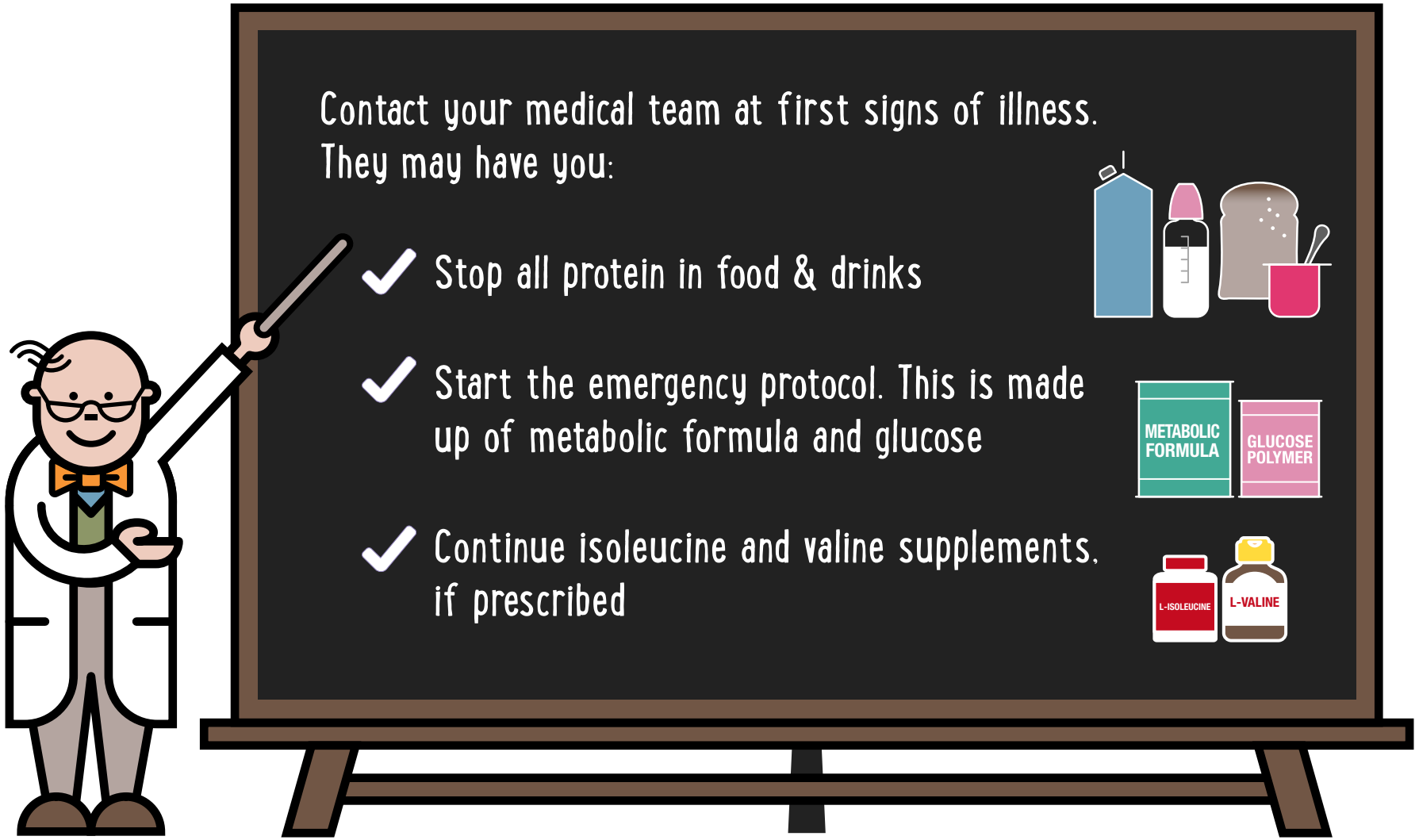
During any illness, our bodies need extra energy. The body will start breaking down cell protein, causing blood leucine levels to increase. This process is also referred to as catabolism.

It is extremely important to start the emergency protocol your metabolic team has developed for you and contact them right away.



How is MSUD managed during illness?

Always follow your medical team's guidance.



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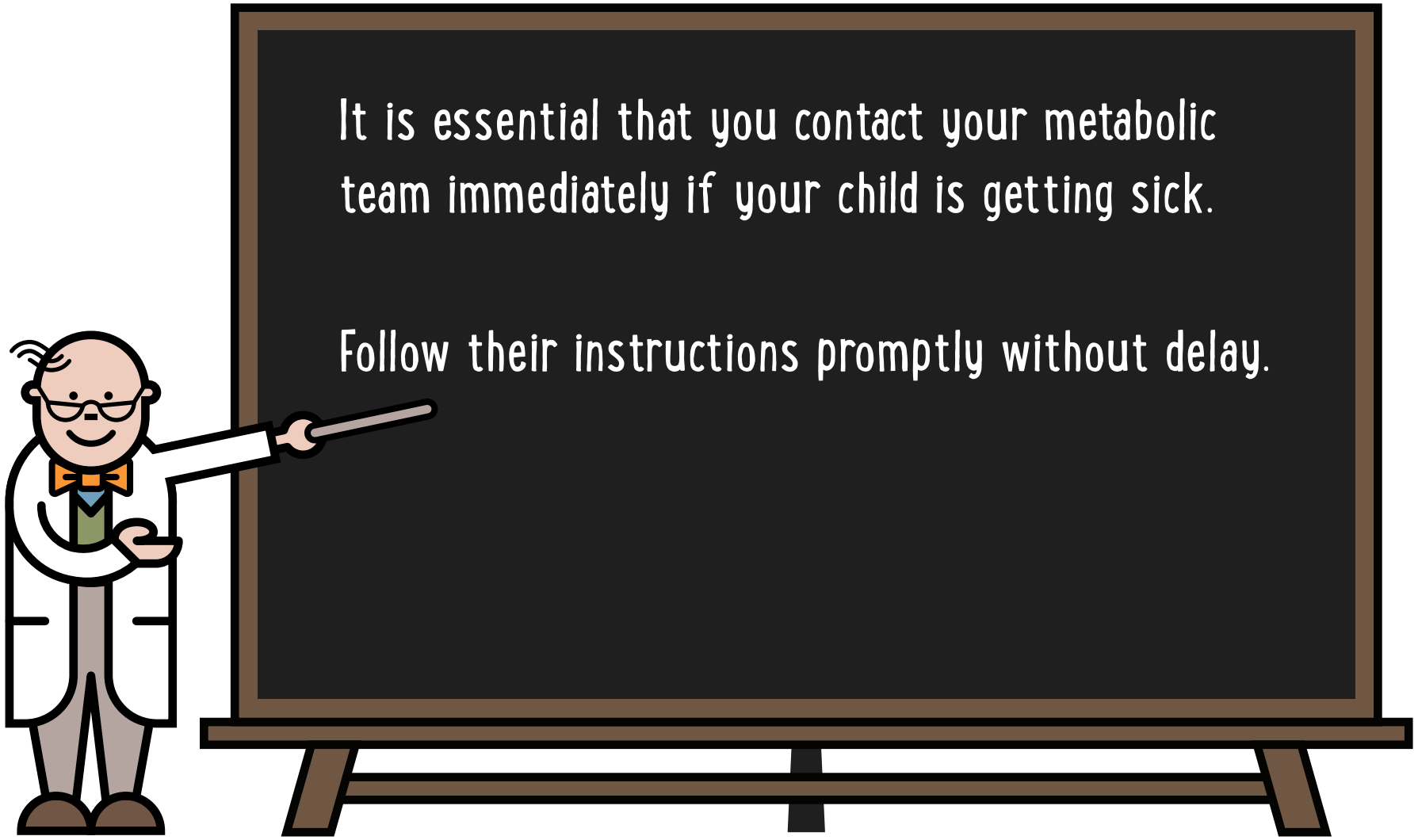
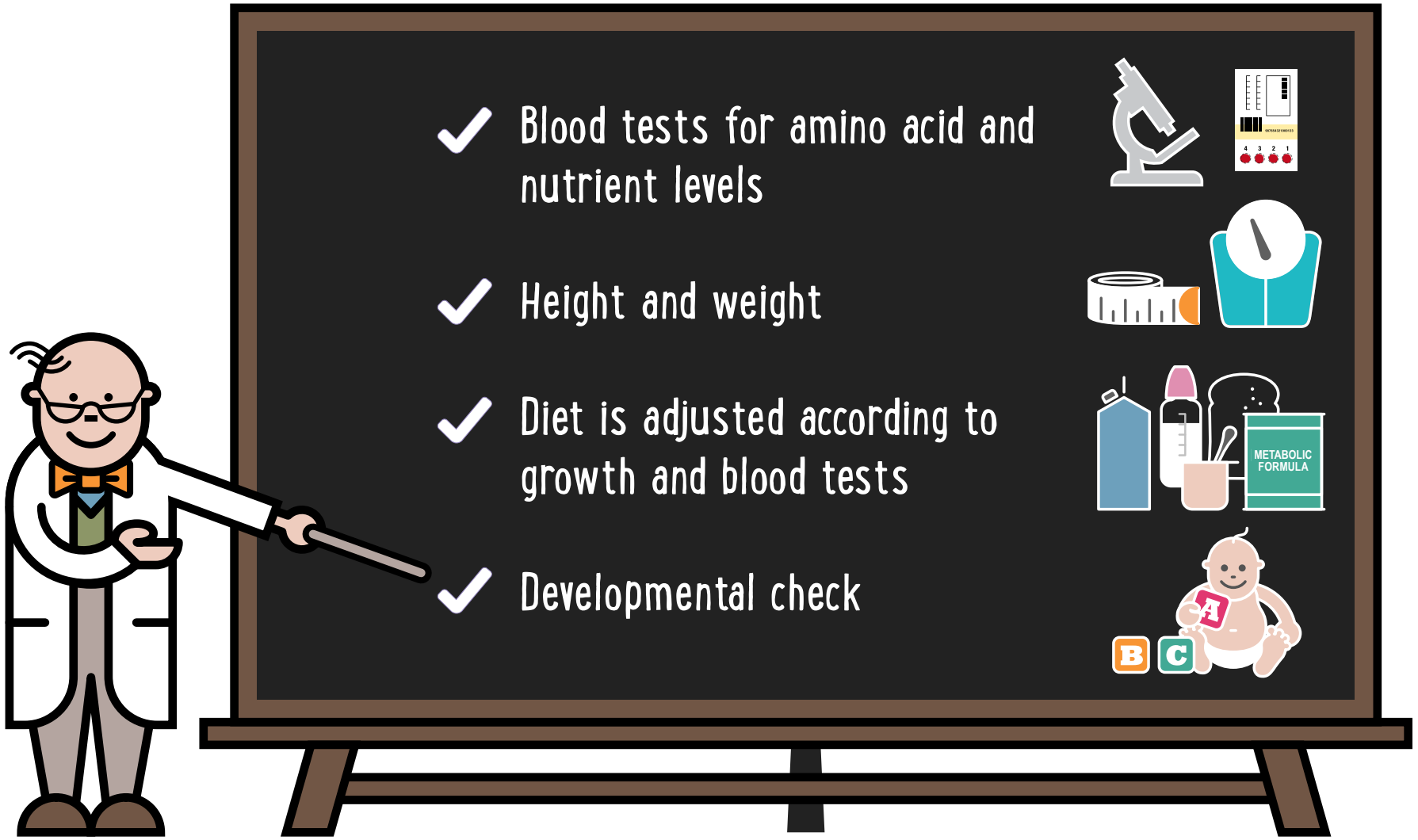
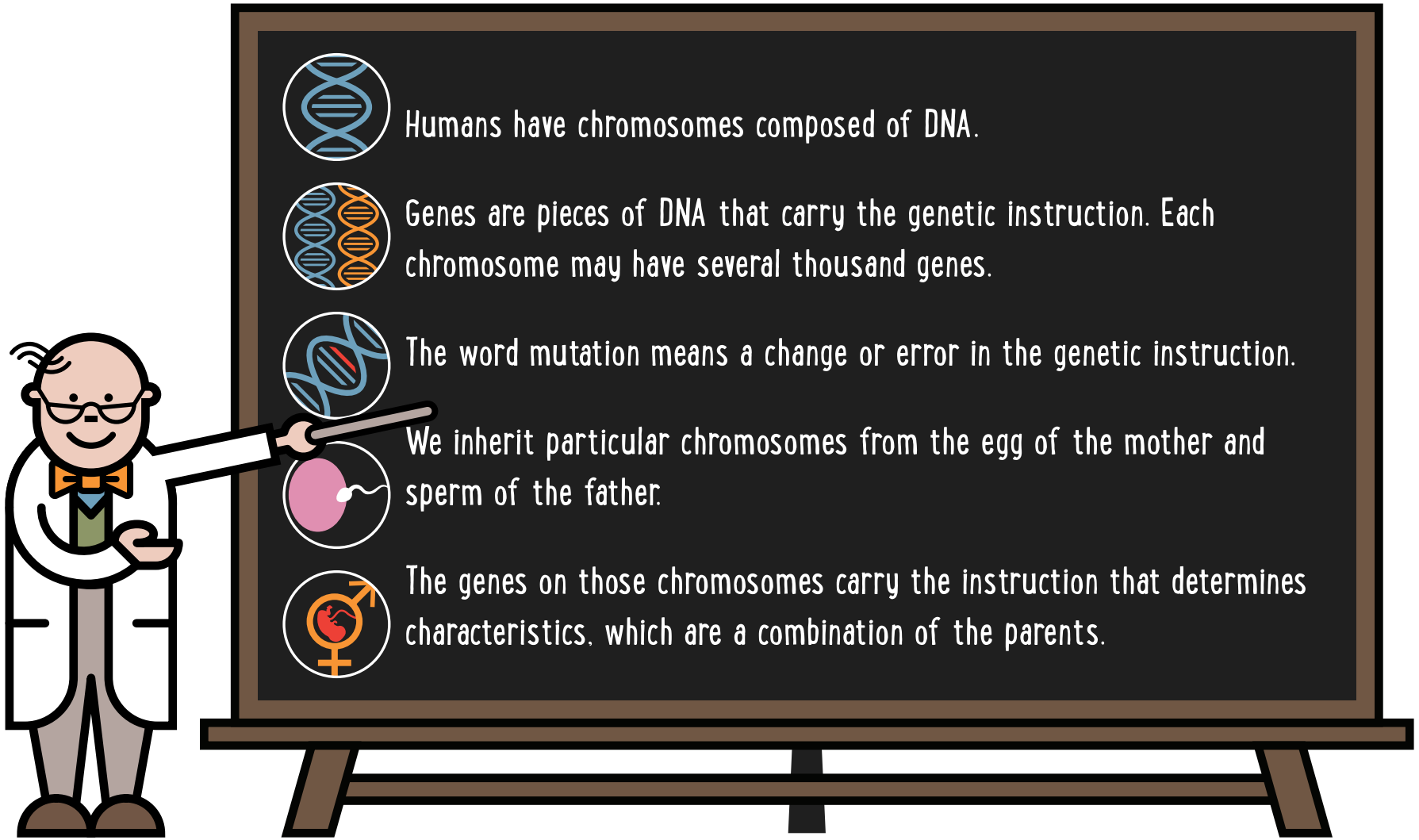
Most importantly



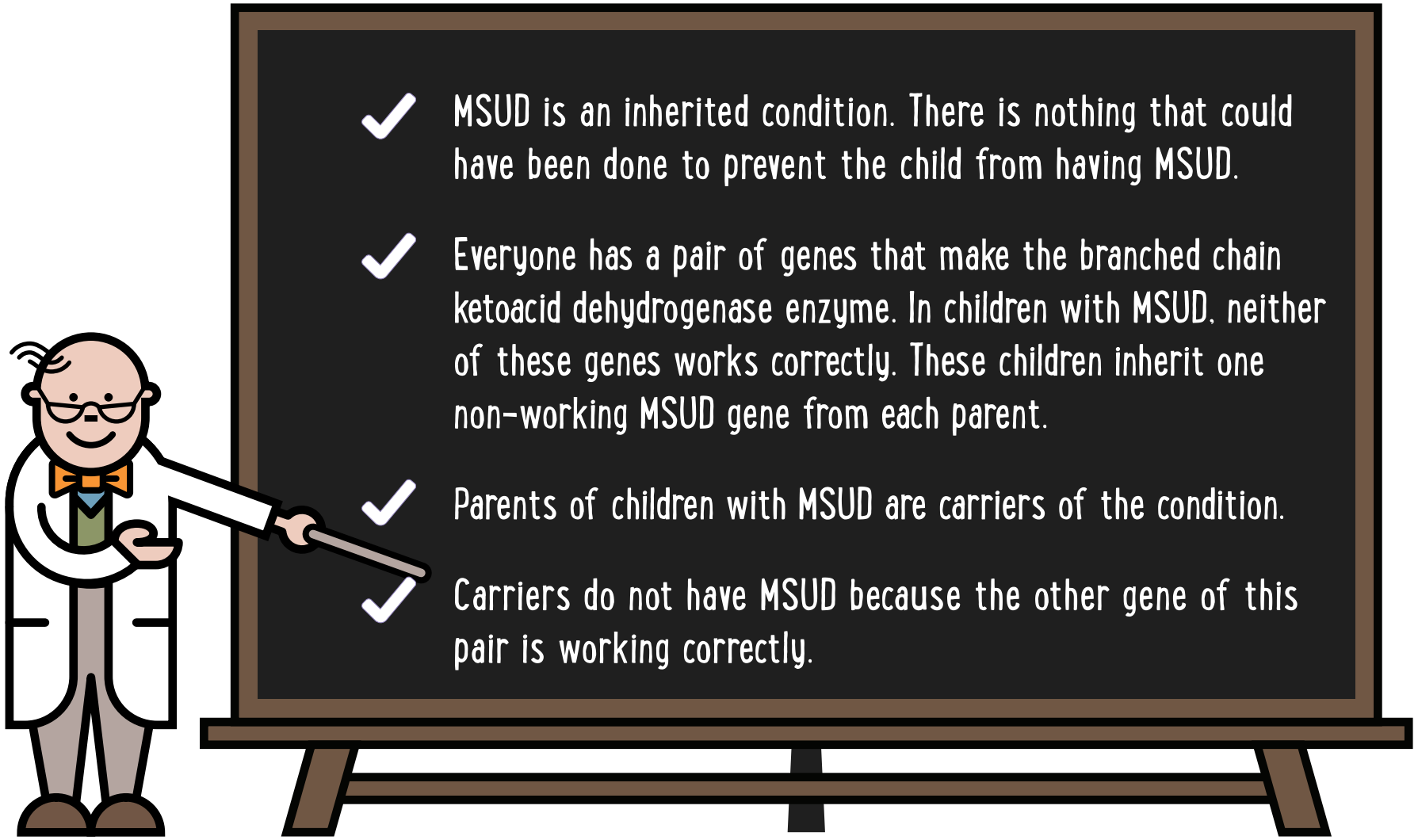
What else is monitored in MSUD?



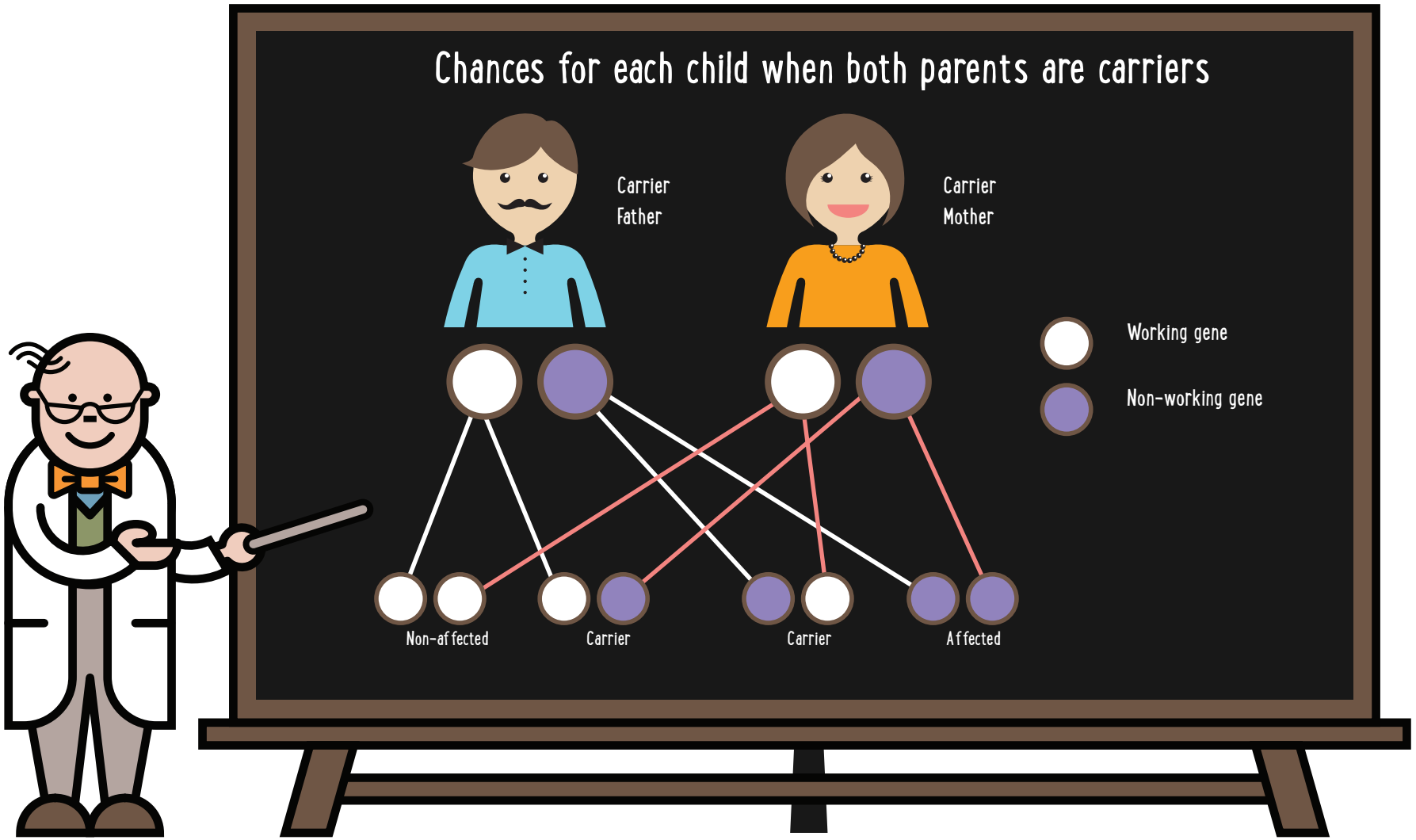
What happens in human genetics?



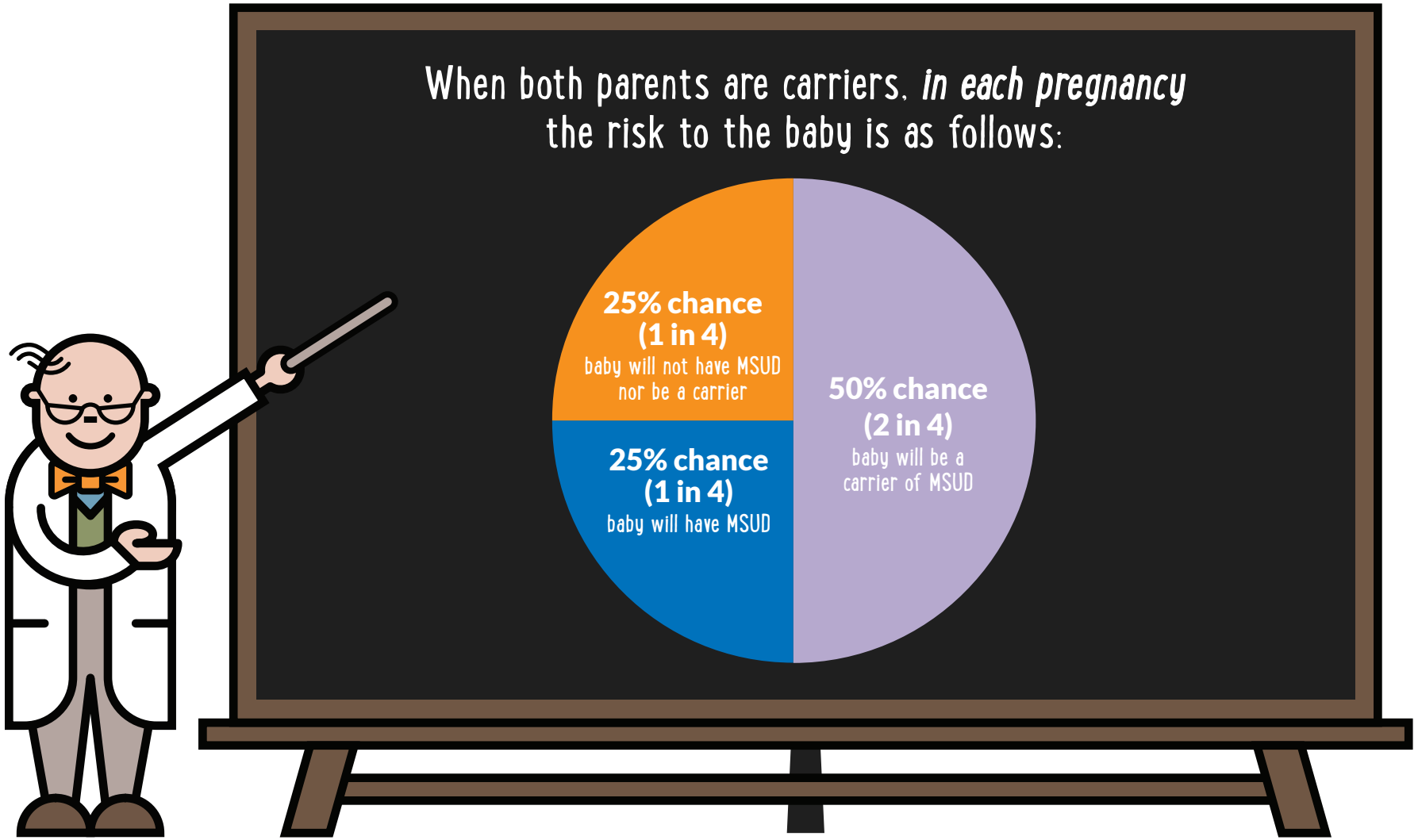
How does one inherit MSUD?



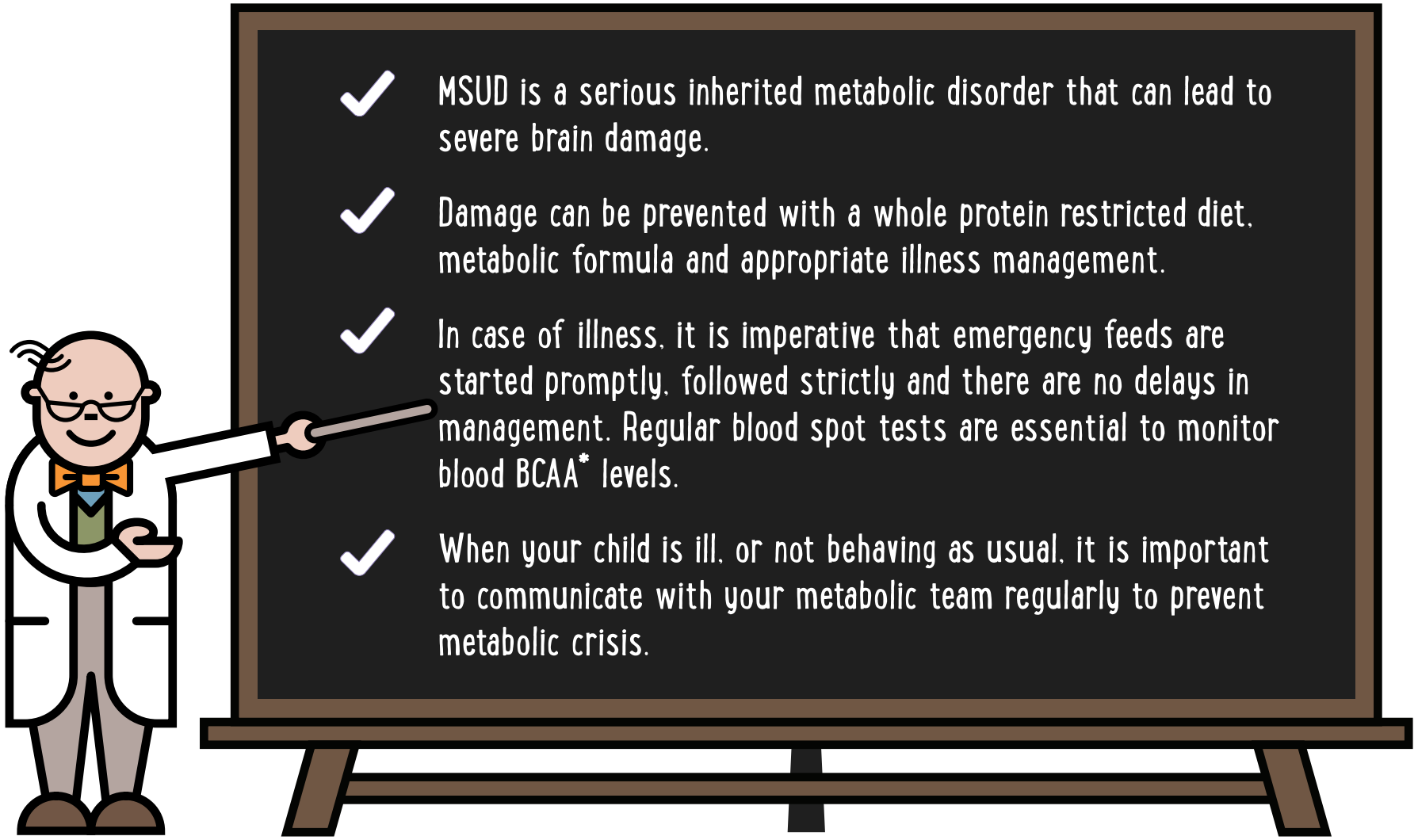
Inheritance – Autosomal recessive – possible combinations



Future pregnancies



Take home messages



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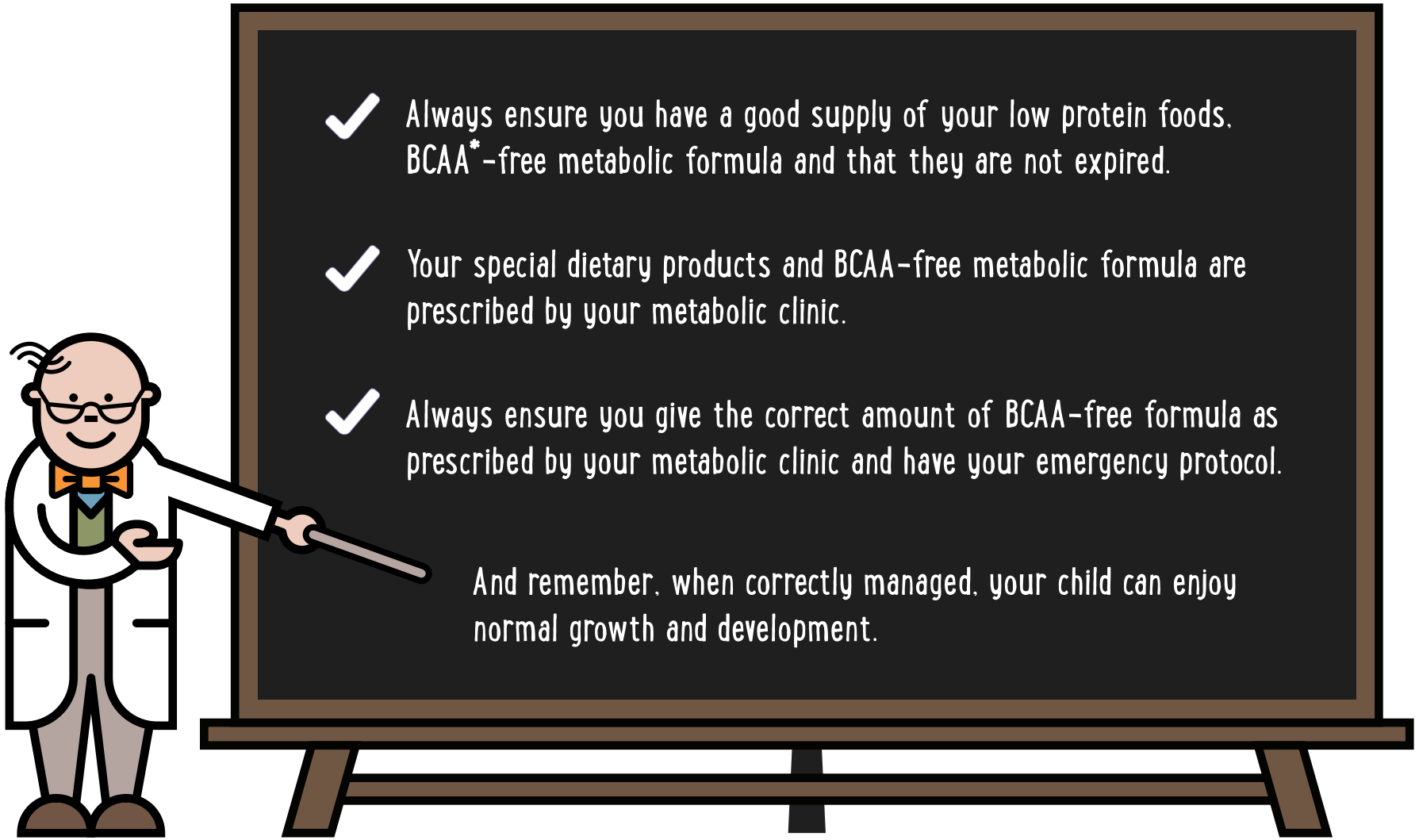
Helpful hints



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Who's who (contact details)

My dietitian

Name: .....

Phone #: .....

Email: .....

My nurse

Name: .....

Phone #: .....

Email: .....

My doctor

Name: .....

Phone #: .....

Email: .....

Notes

Notes

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