TEMPLE

Tools **E**nabling **M**etabolic **P**arents **LE**arning

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

MSUD

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.

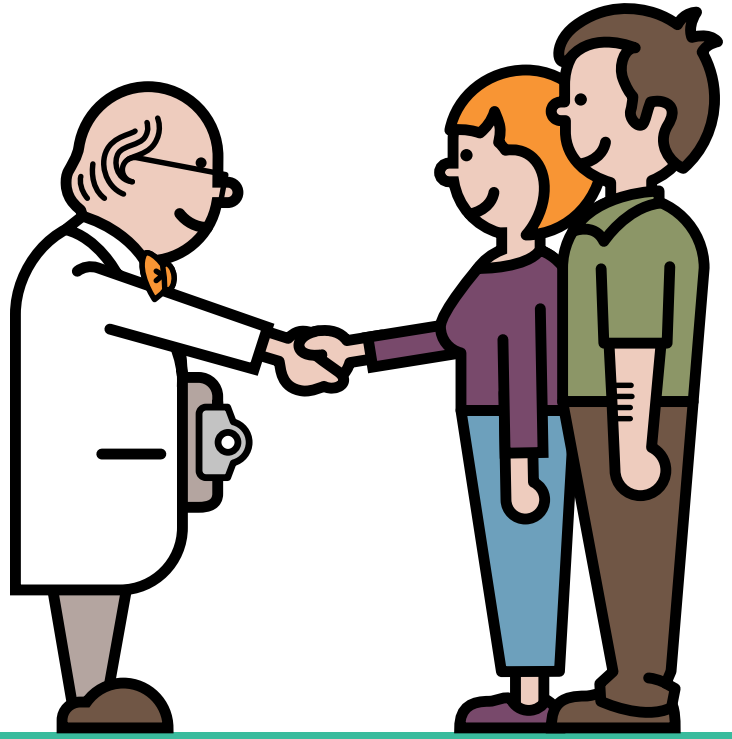
Each module produced is reviewed by a consultant clinician who is a member of the BIMDG.

This teaching tool is not designed to replace dietary information that may be given by a dietitian in clinic.

Supported by NUTRICIA as a service to metabolic medicine

MSUD

Information for families following a positive newborn screening



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Tools Enabling Metabolic Parents LEarning

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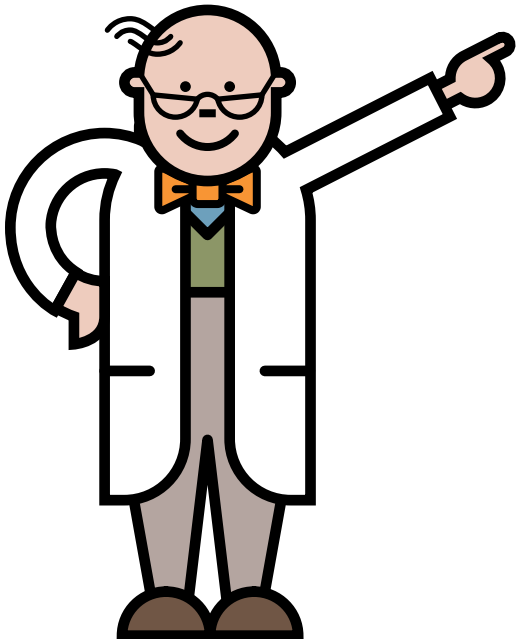
What is MSUD?

MSUD stands for Maple Syrup Urine Disease

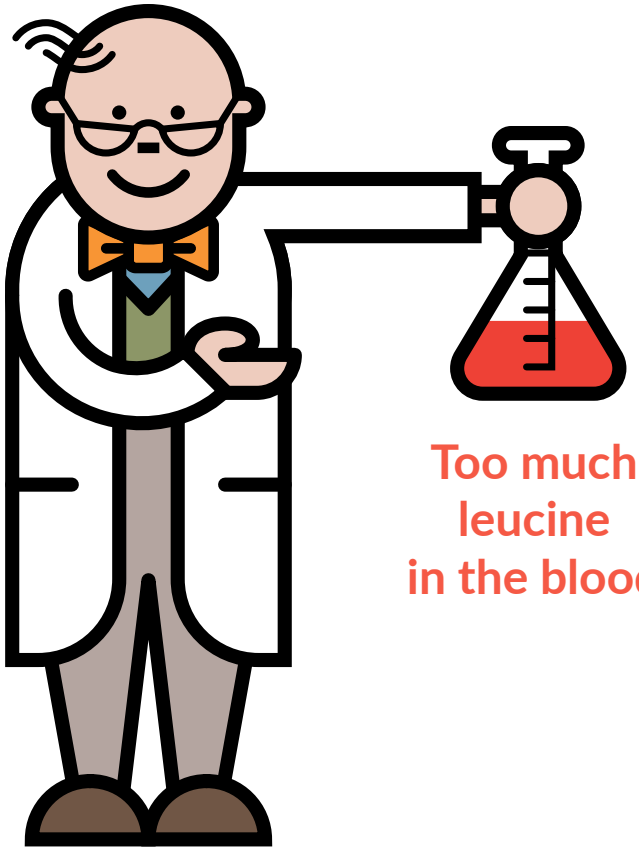
It is an inherited metabolic condition



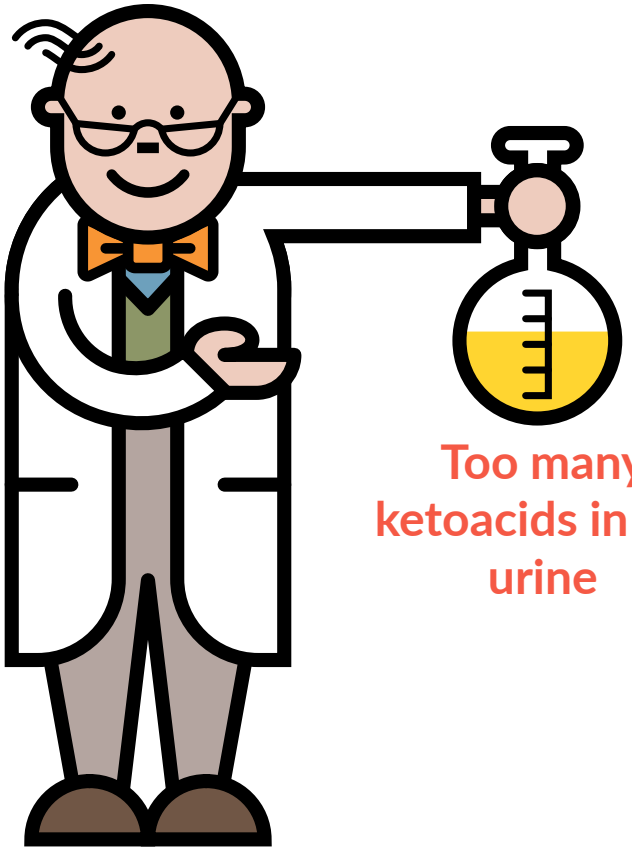
Maple Syrup Urine Disease

MSUD

What is MSUD?



Too much leucine in the blood



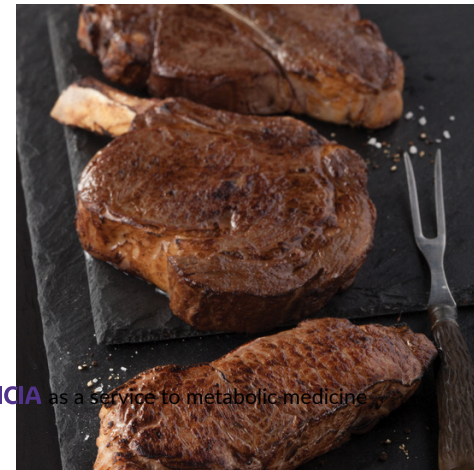
Too many ketoacids in the urine

MSUD and protein

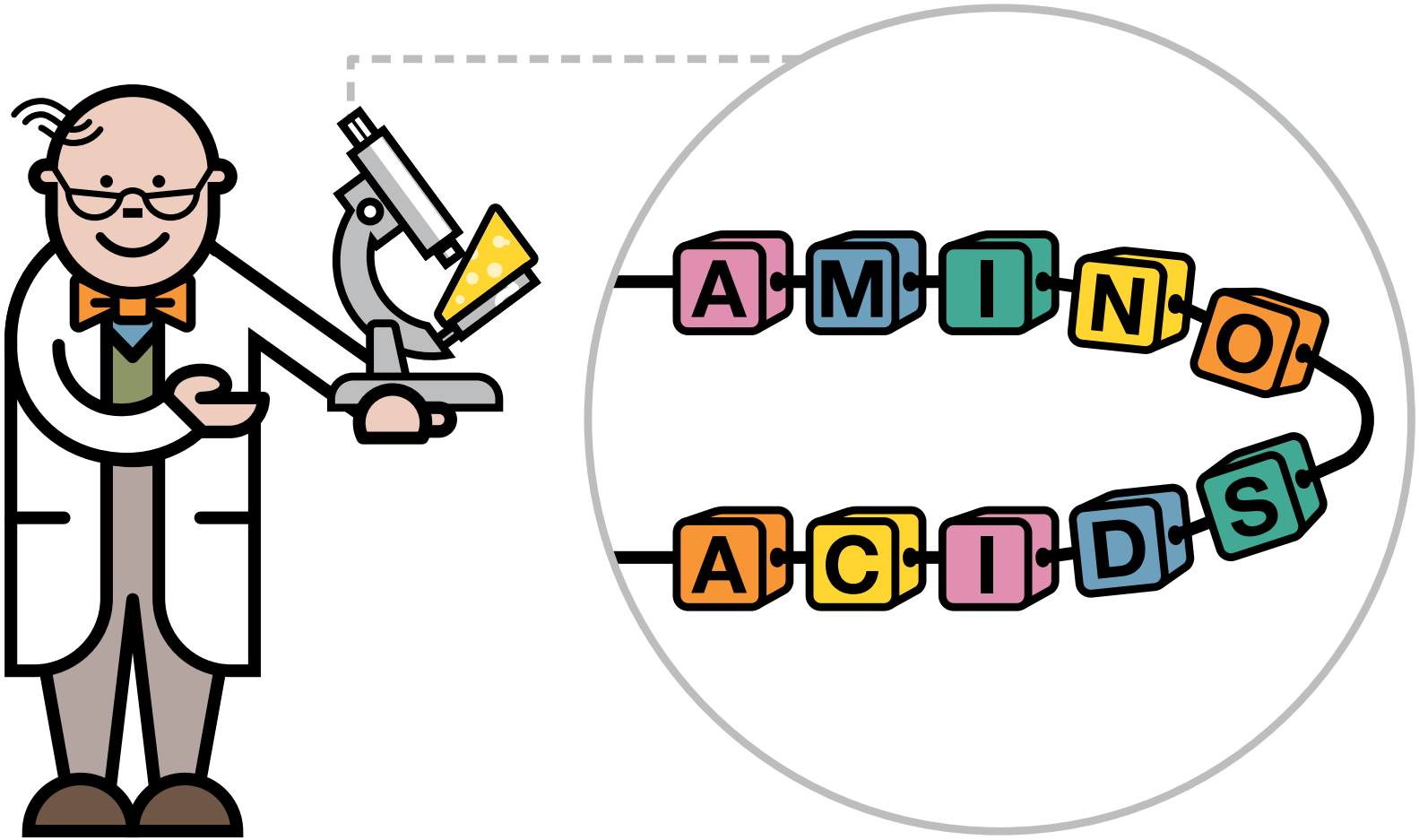
MSUD affects the way your baby breaks down protein

Many foods contain protein

The body needs protein for growth and repair



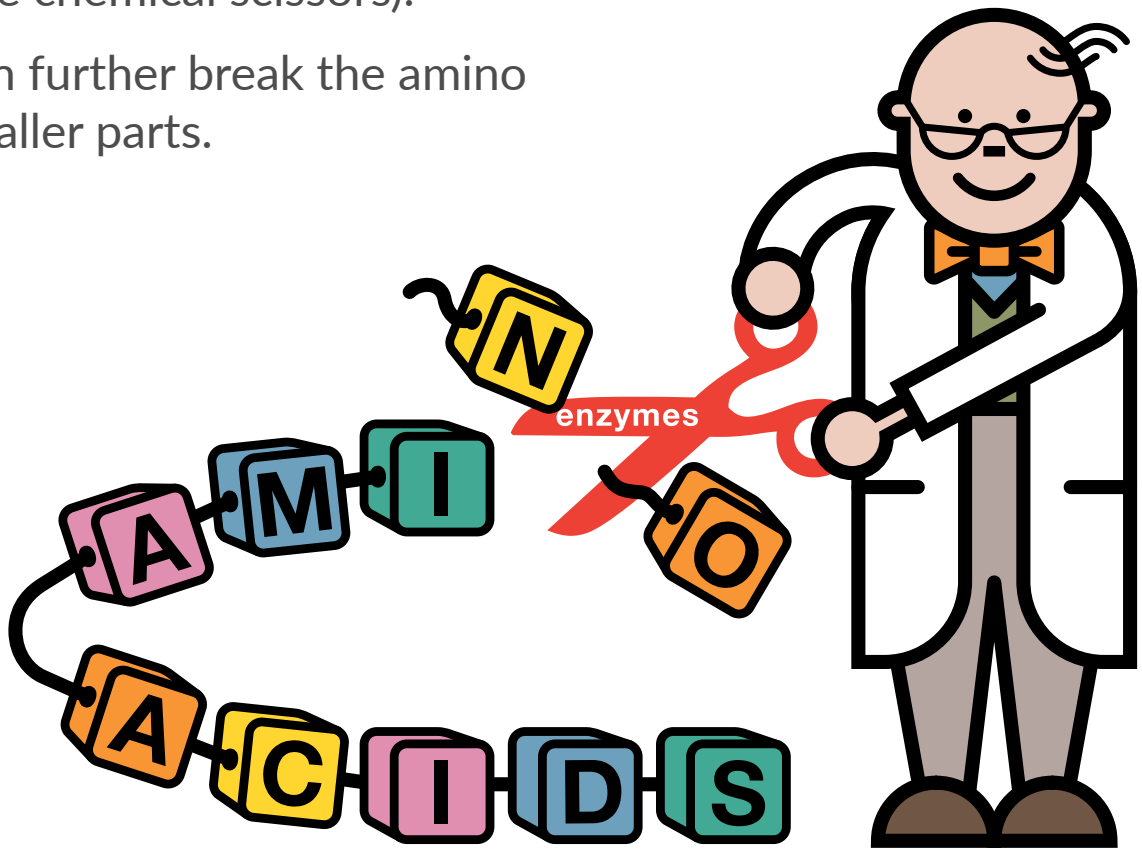
What is protein?



Protein and enzymes

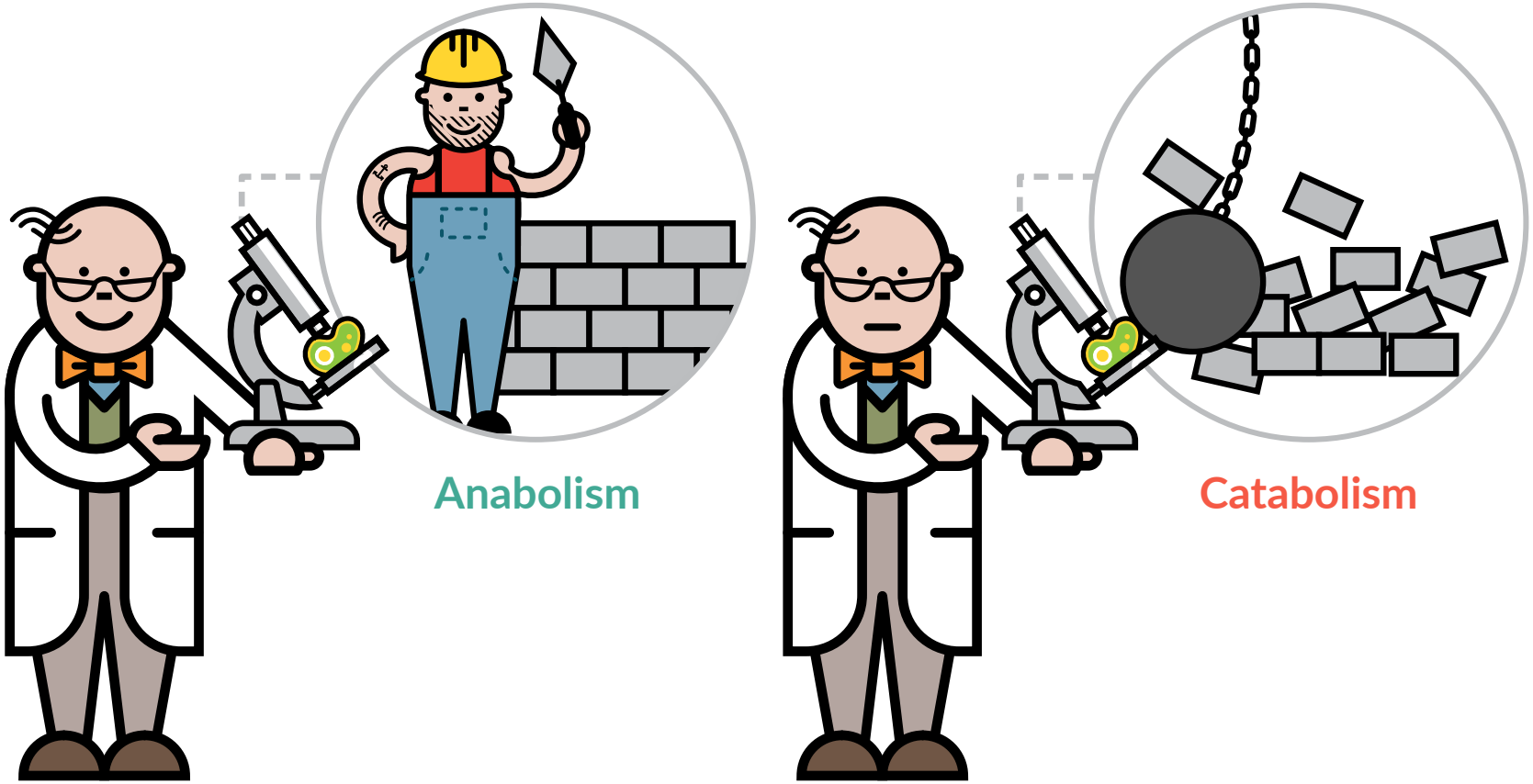
Protein is broken down into amino acids (building blocks of protein) by enzymes (which are like chemical scissors).

Enzymes then further break the amino acids into smaller parts.



Protein metabolism

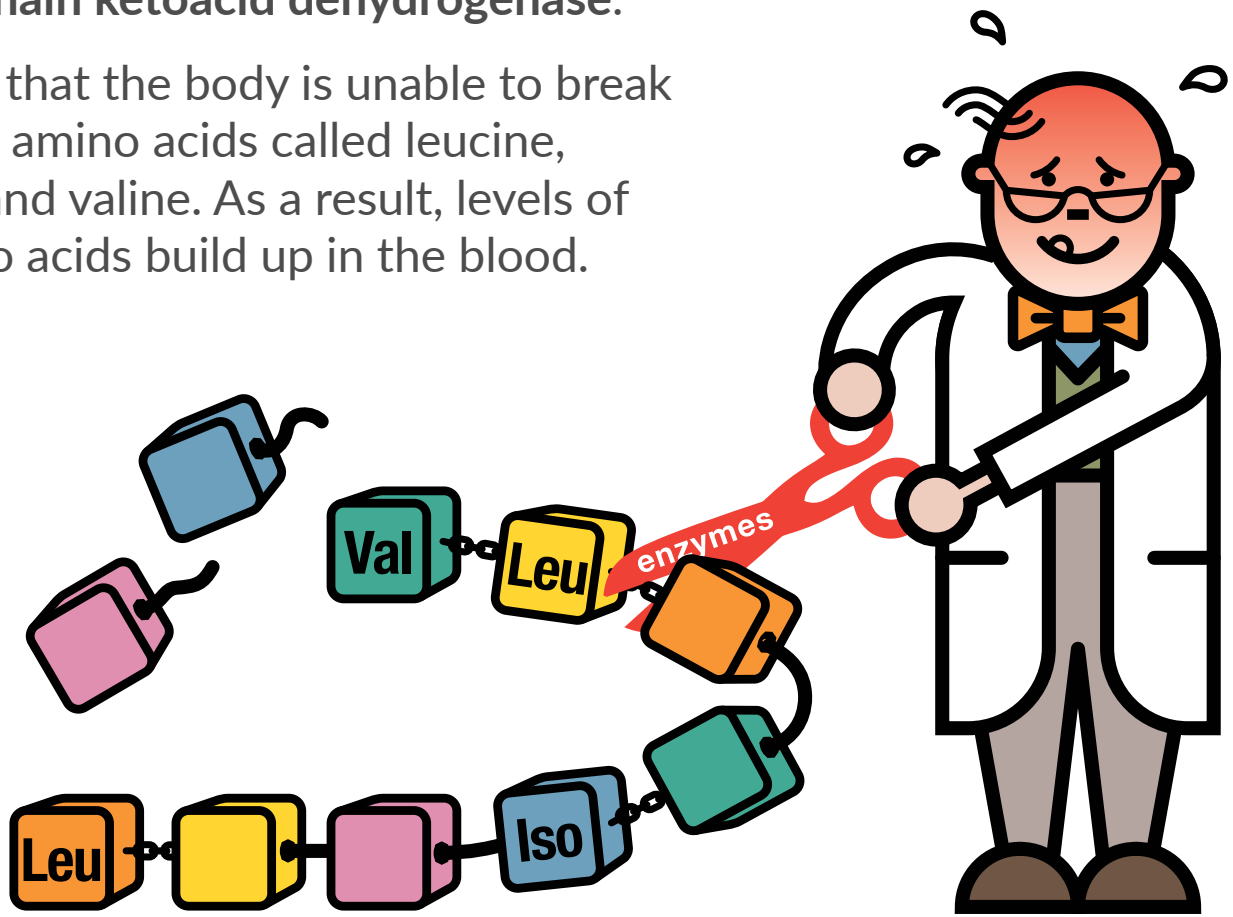
Metabolism refers to the chemical processes that occur inside the cells of the body.



What happens in MSUD?

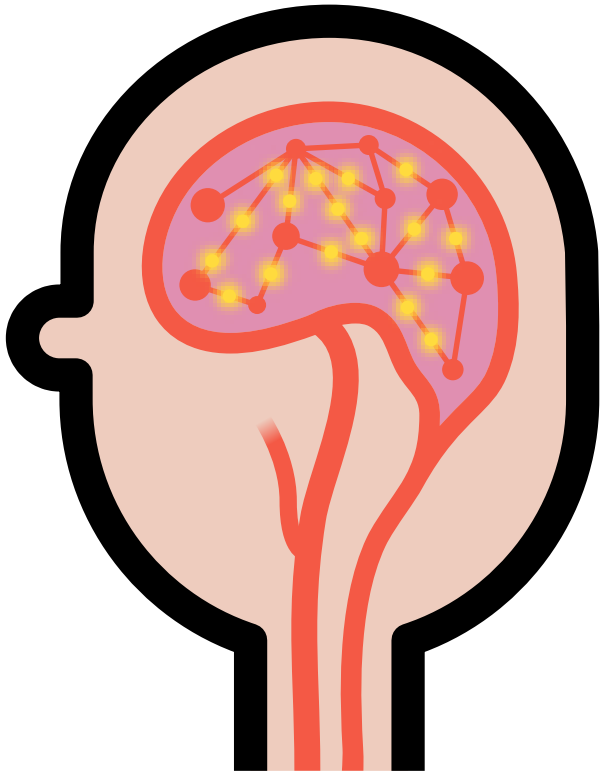
In MSUD, the body lacks an enzyme called **branched chain ketoacid dehydrogenase**.

This means that the body is unable to break down three amino acids called leucine, isoleucine and valine. As a result, levels of these amino acids build up in the blood.



What can go wrong in MSUD?

The build-up of harmful chemicals can cause damage to the brain. It may cause delays to normal development like walking and talking.



Early management can prevent brain damage and learning difficulties

What are the symptoms in MSUD?

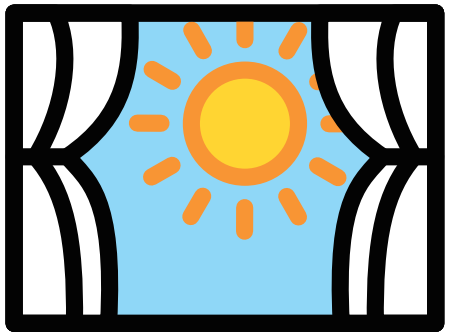
Some babies with MSUD become ill in the first few days of life before the newborn screening result is available.

Symptoms include:

- poor feeding
- vomiting
- dehydration (lack of body fluids)
- floppy baby
- excessively sleepy
- seizures
- rapid breathing
- sweet-smelling urine (like maple syrup)

The effects of MSUD quickly become life-threatening if unmanaged

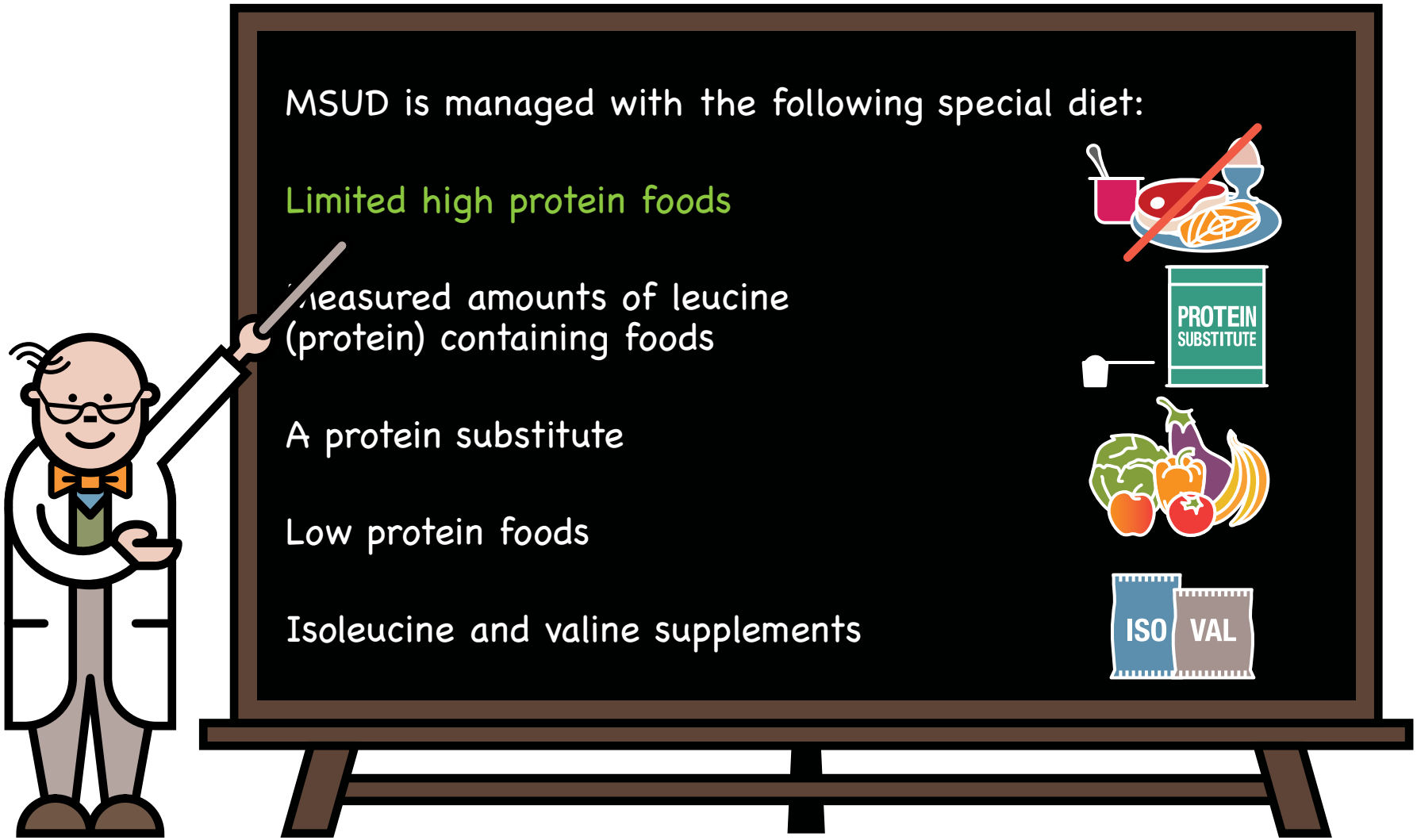
Some children may develop symptoms at a later stage. They may present with developmental delay or with an acute childhood illness such as vomiting or diarrhoea.



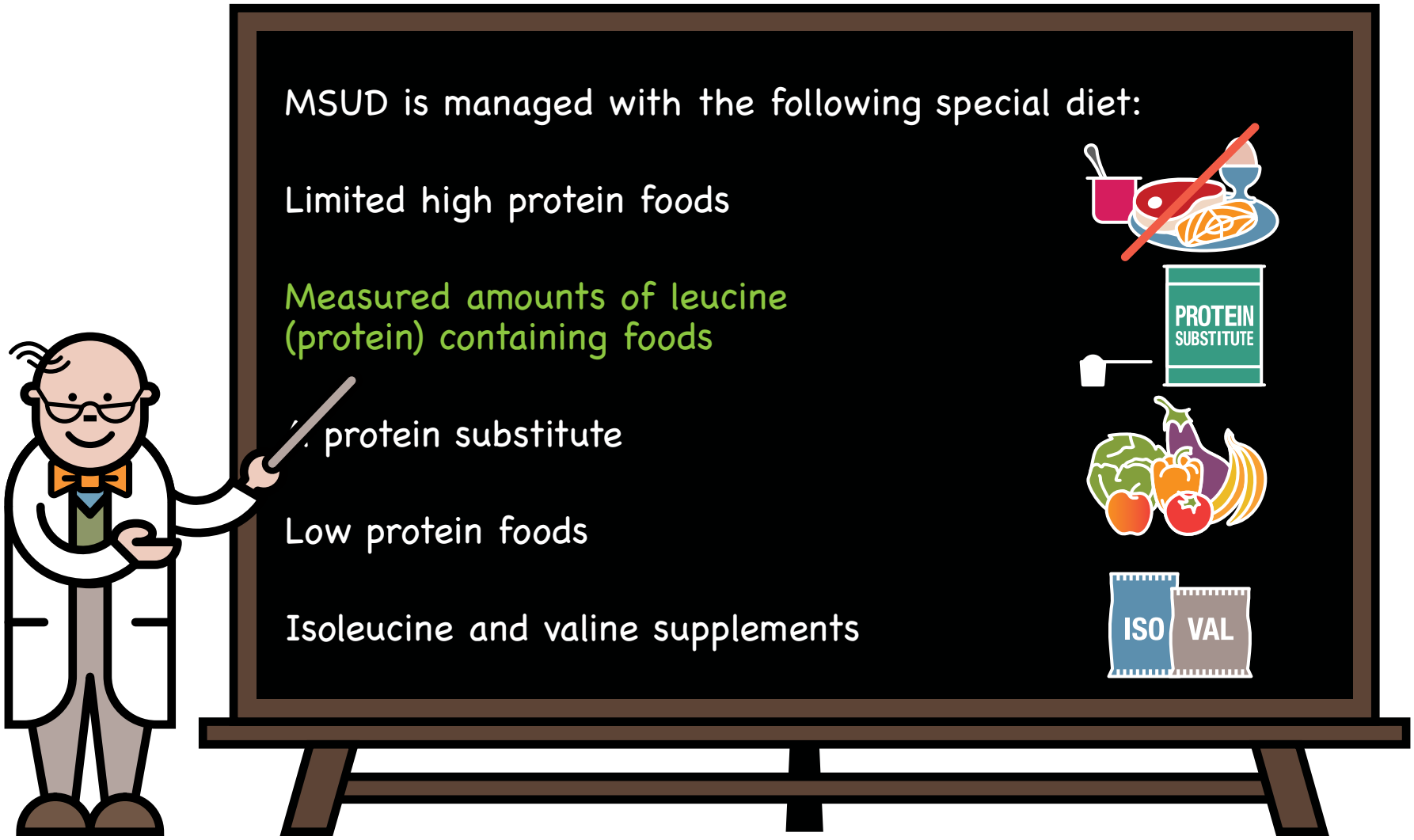
How is MSUD diagnosed?

MSUD is usually diagnosed by newborn screening.
High levels of leucine are found in the blood.

How is MSUD managed?



How is MSUD managed?



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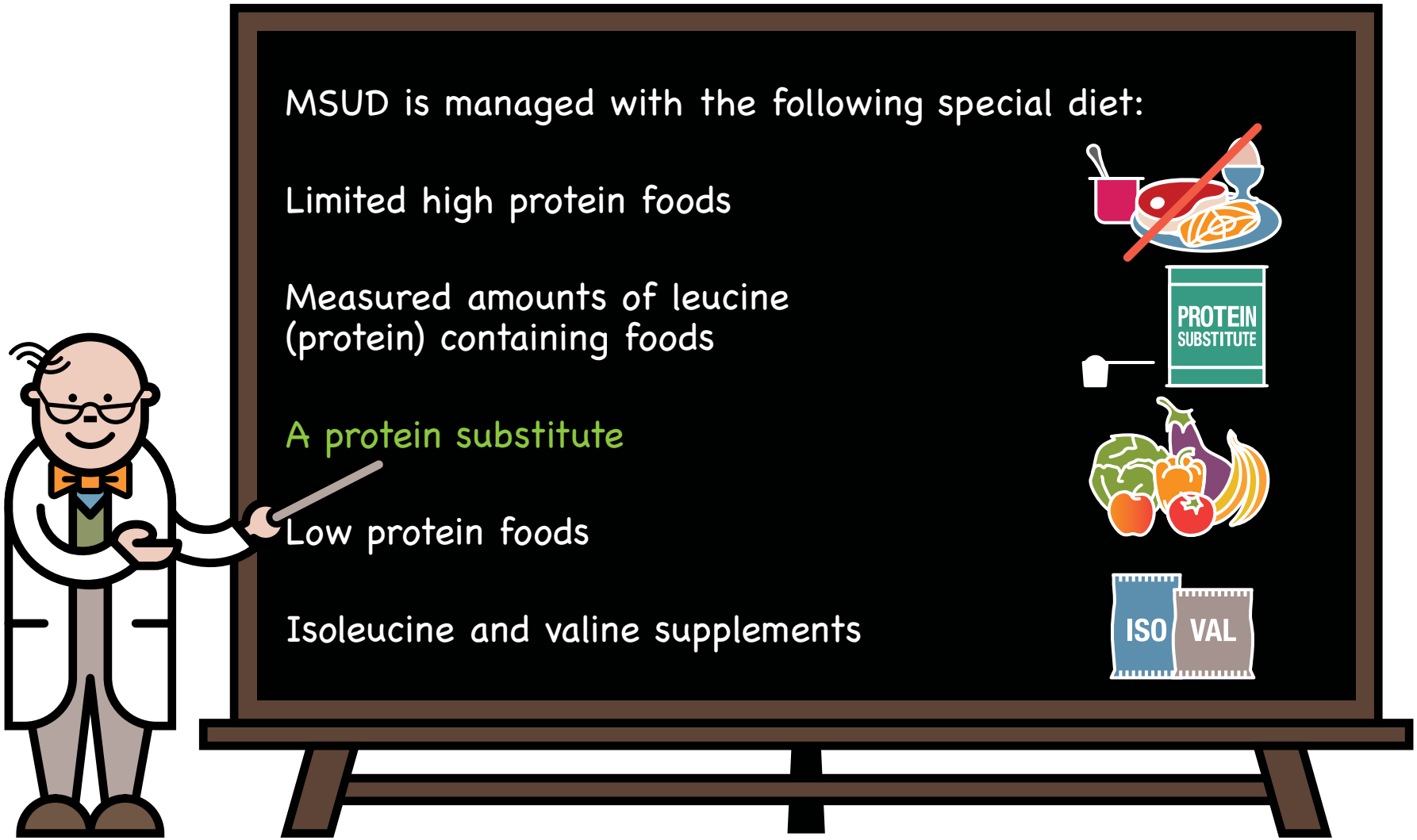

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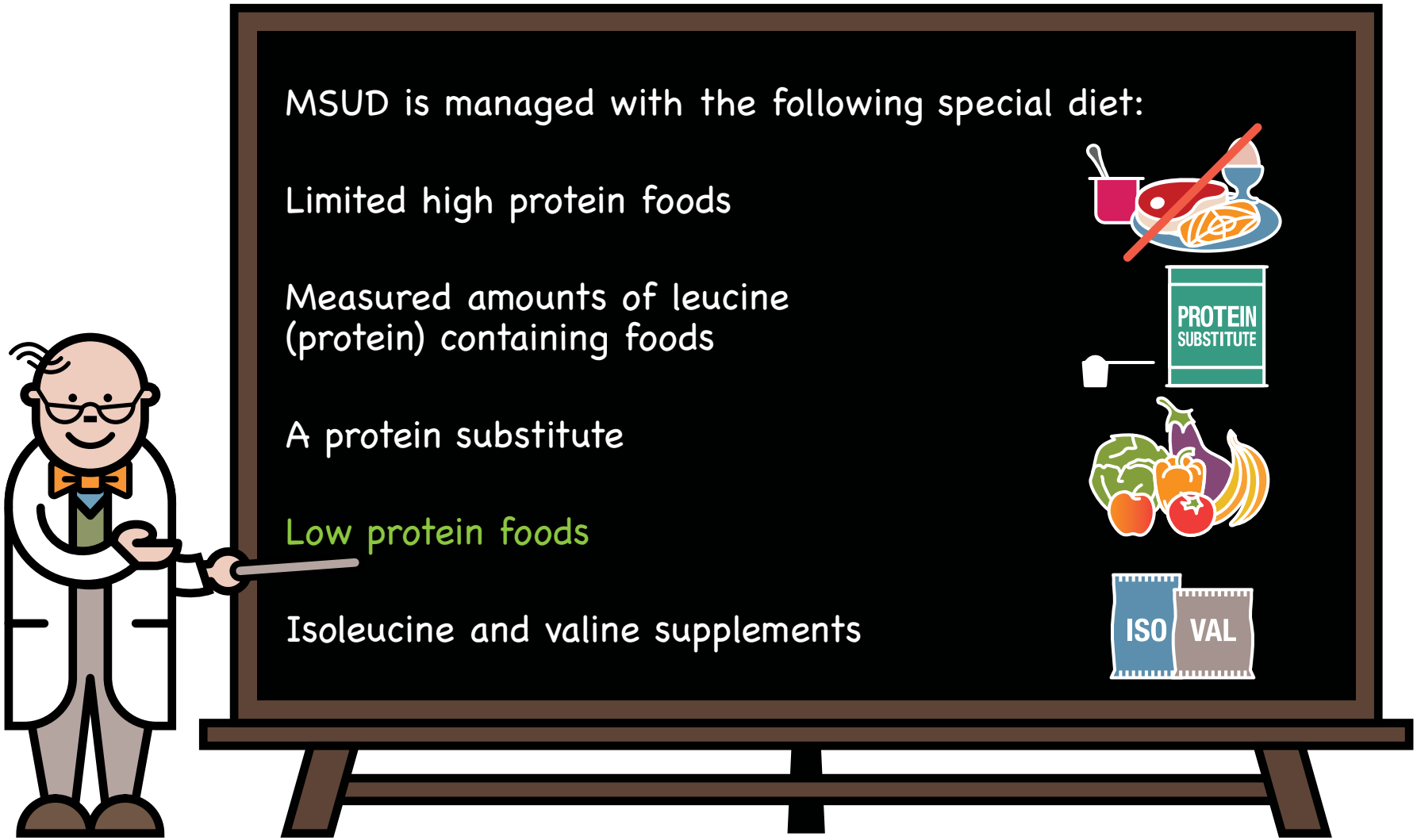

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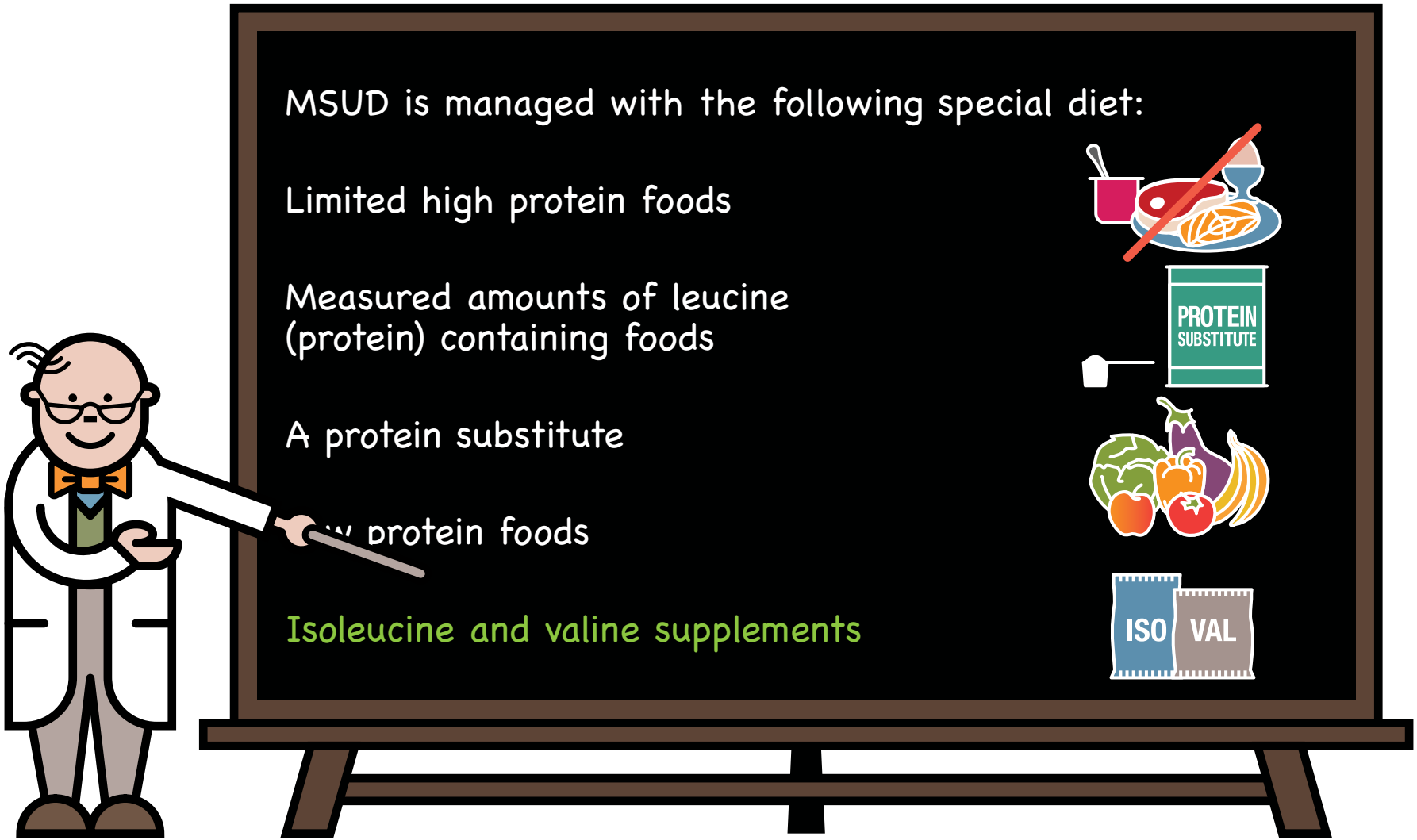


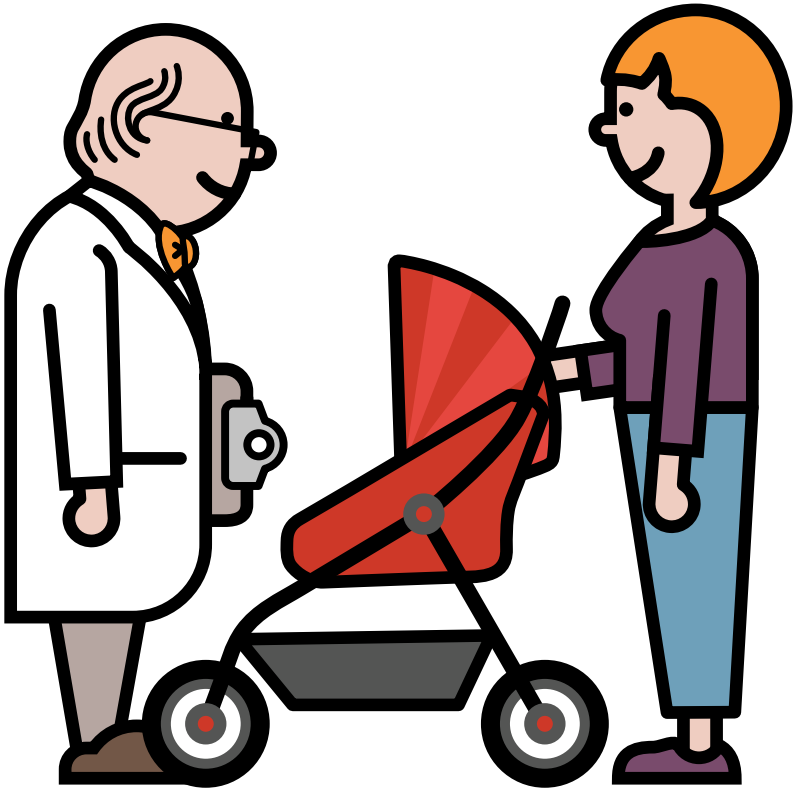
High protein foods

These foods are high in leucine (protein) and must be avoided: **meat, fish, eggs, cheese, bread, pasta, nuts, seeds, soya, Quorn** and **tofu**.

Measured leucine intake

In babies, a restricted amount of leucine (protein) is given from breast milk or measured amounts of infant formula.

The amount given will be monitored regularly by your specialist metabolic dietitian.

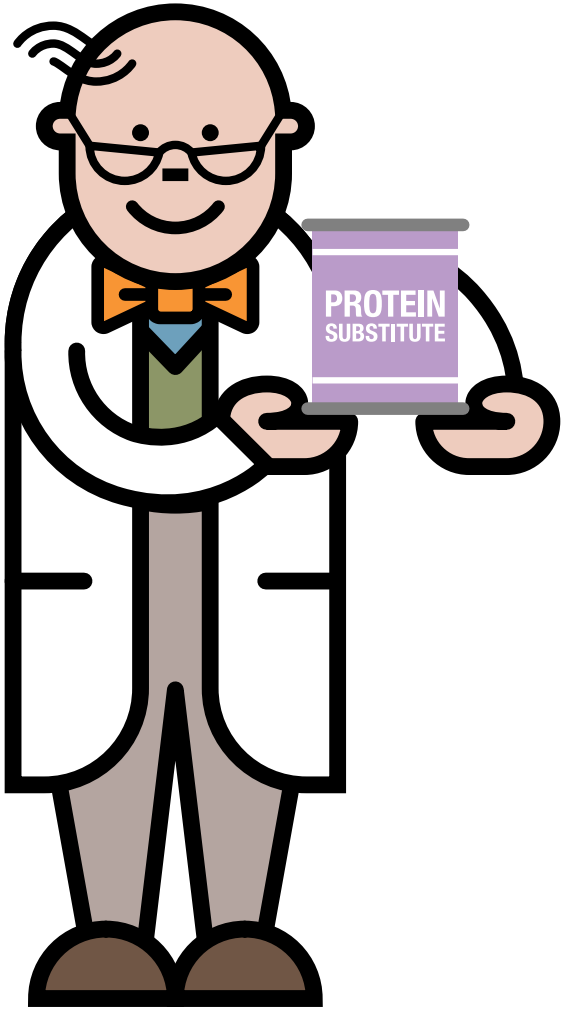


Protein substitute

Protein substitute is essential for metabolic control.

It will help to meet your baby's protein, energy, vitamin and mineral requirements.

It is available on prescription.

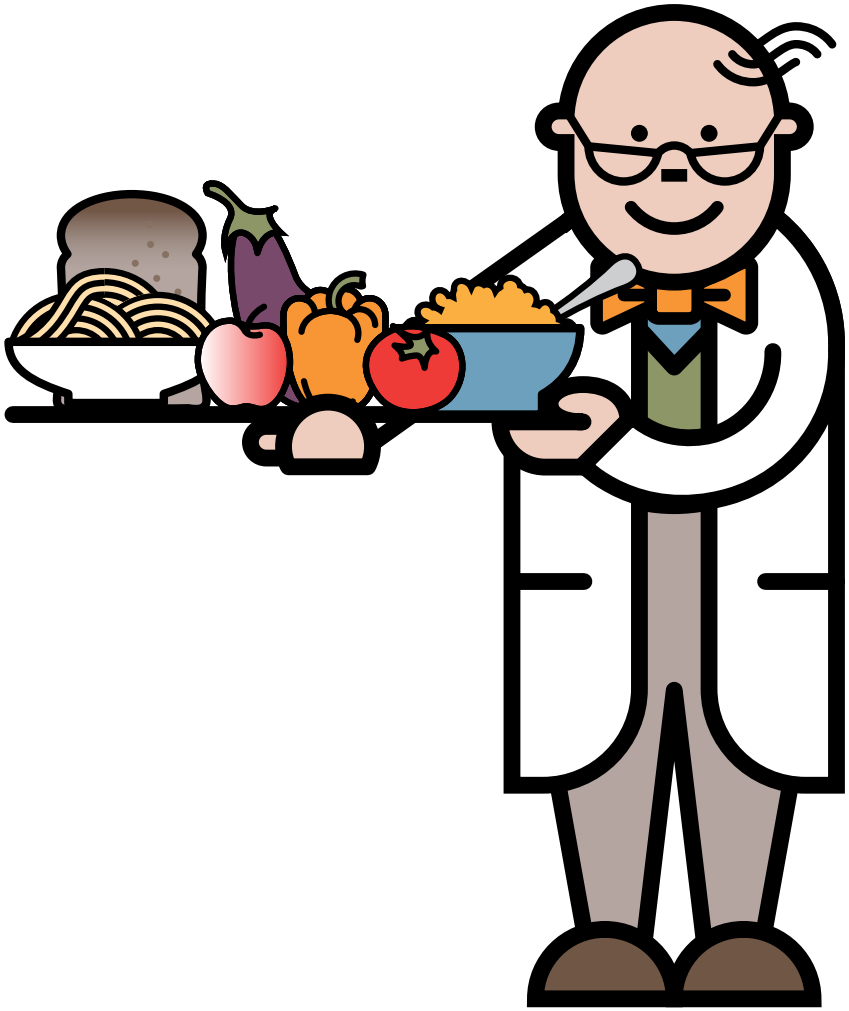


Low protein foods

There are many low protein foods. These include fruit, many vegetables and prescribable low protein foods such as bread and pasta.

They provide:

- a source of energy
- variety in the diet

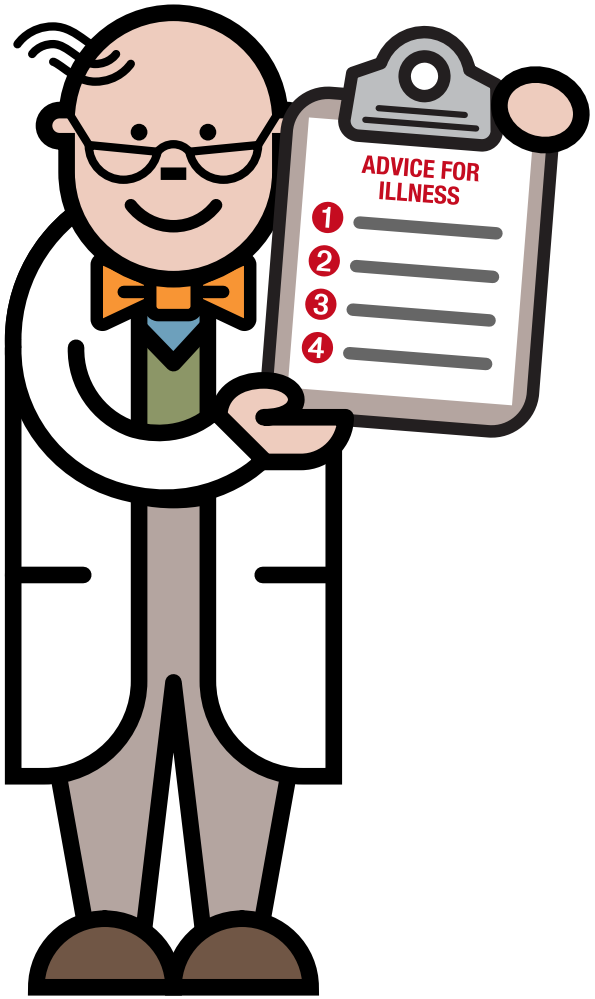


Metabolic crisis

- A '**metabolic crisis**' causes a build-up of leucine and other toxic chemicals
- It is usually triggered by childhood illnesses e.g. vomiting and diarrhoea
- It is important to manage a metabolic crisis quickly and properly

How is MSUD managed during illness?

- During any childhood illness, an emergency regimen is given
- Illnesses can cause catabolism or protein breakdown
- This will lead to a rapid build up of leucine which can cause a metabolic crisis



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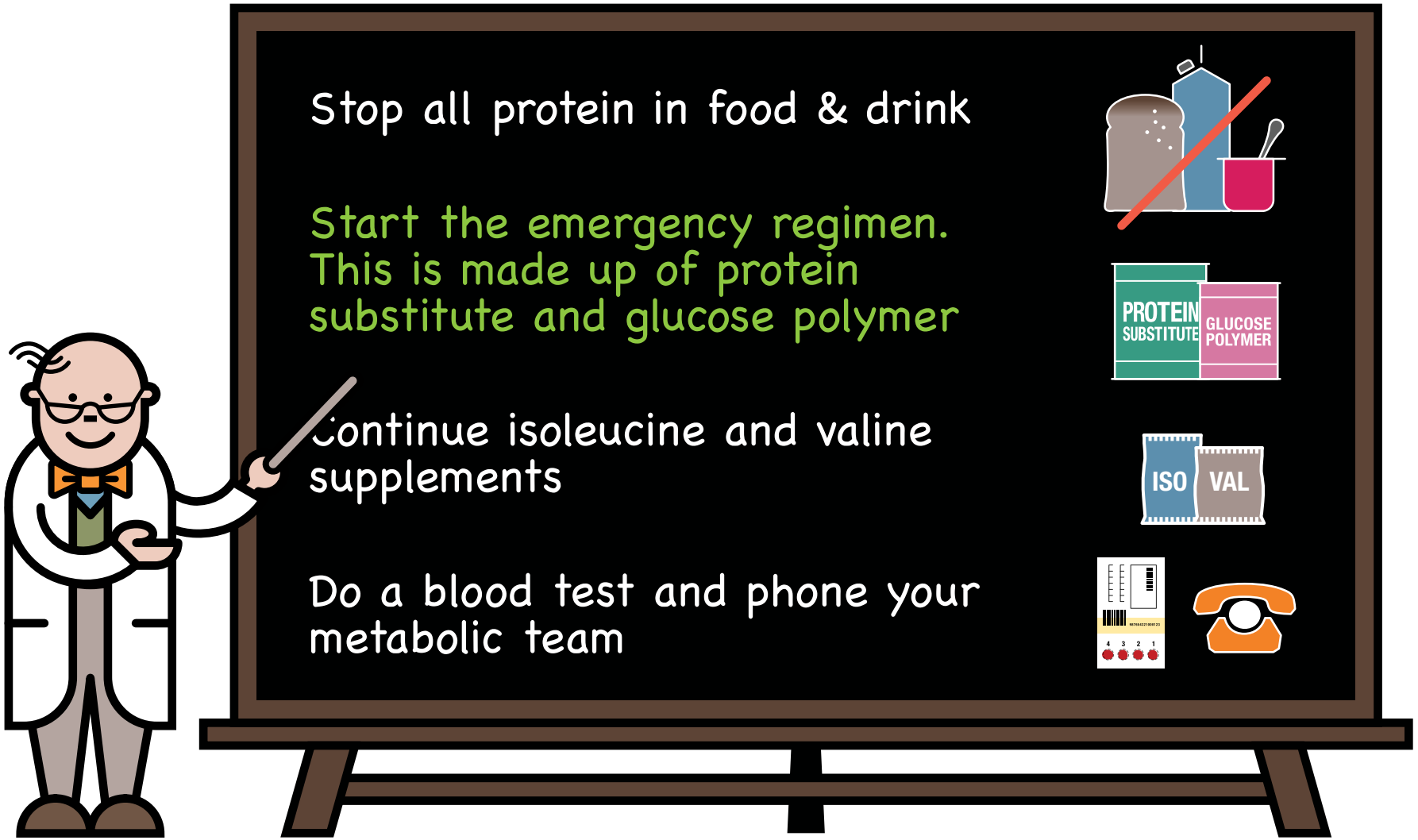

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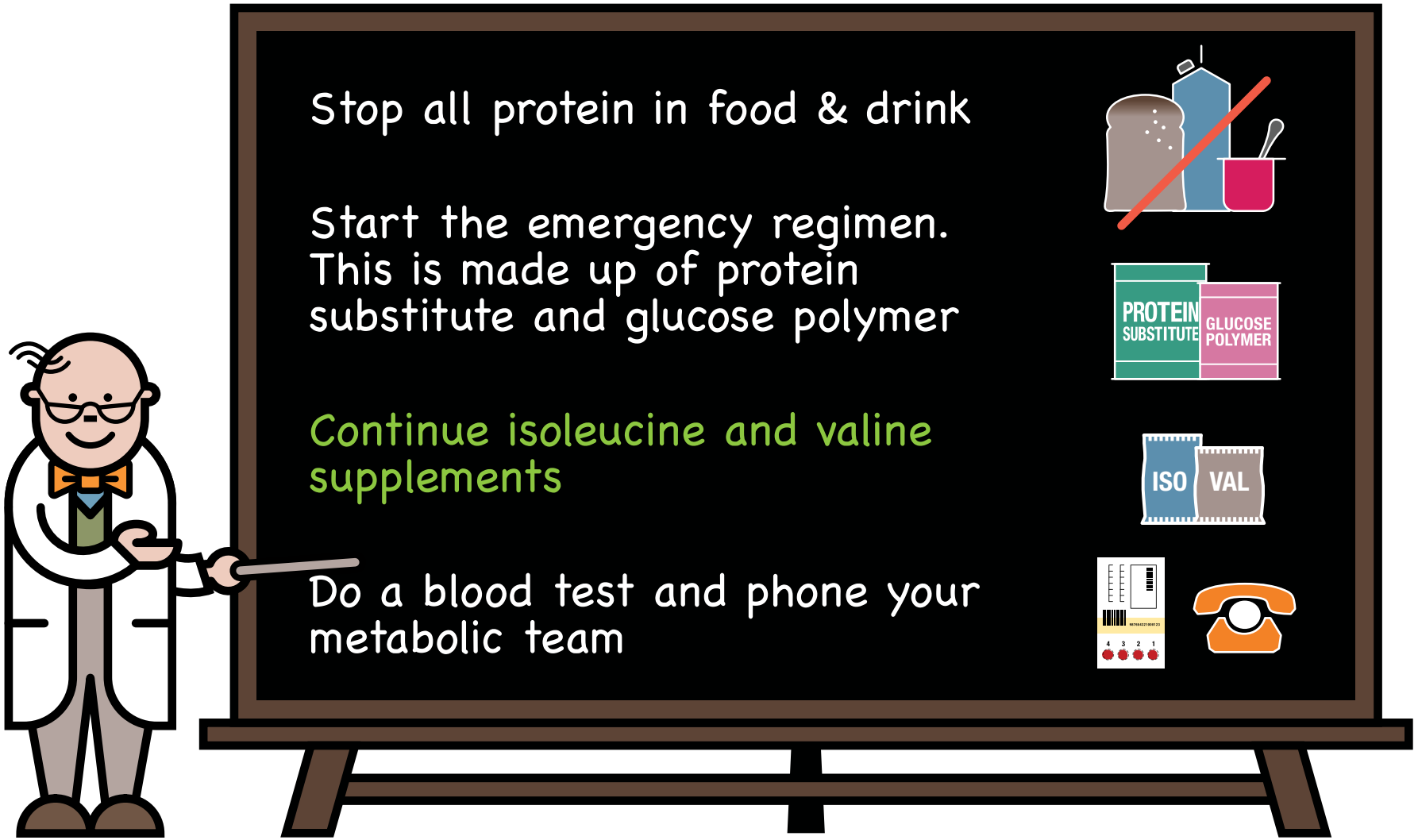


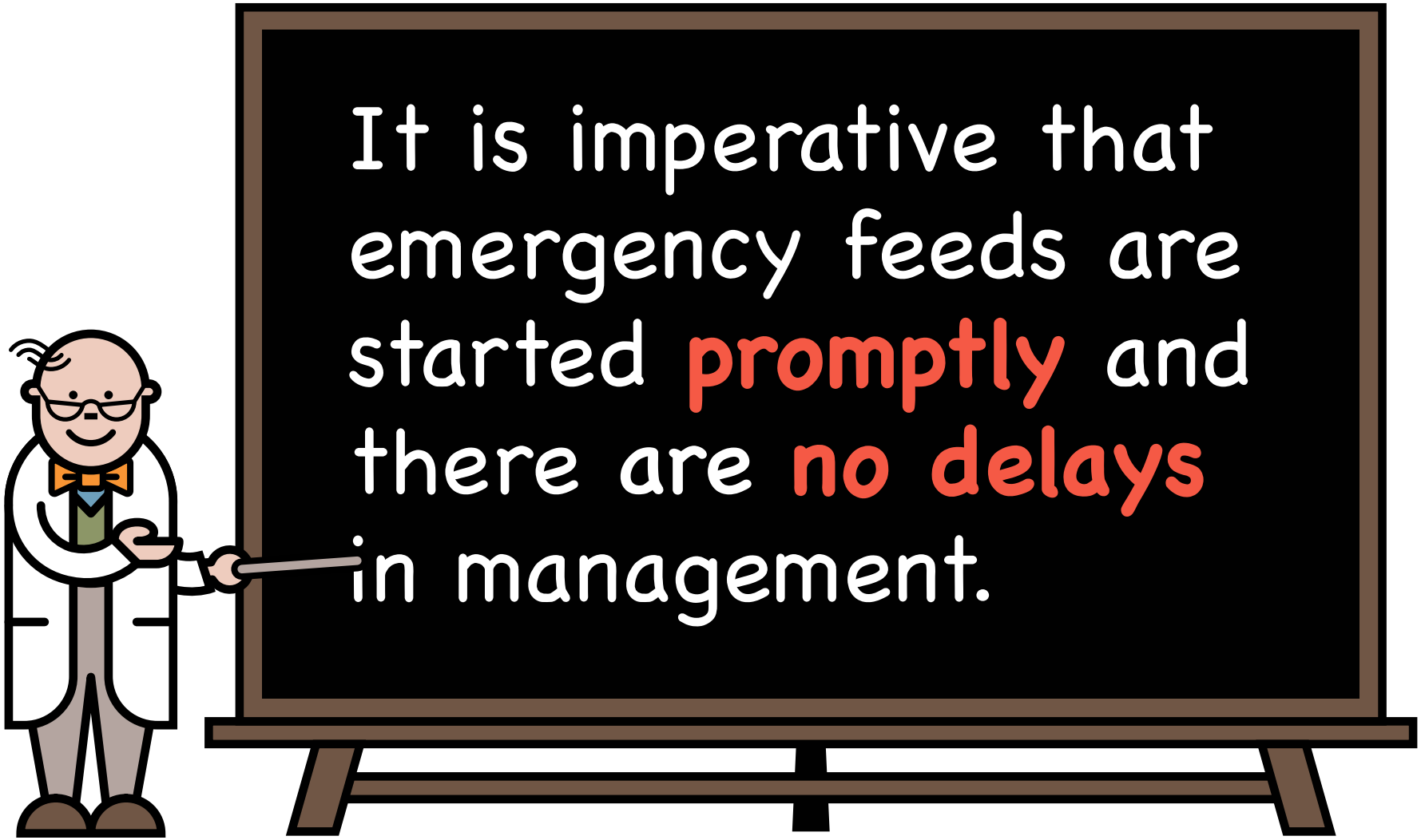
How is MSUD managed during illness?

Checklist for illness

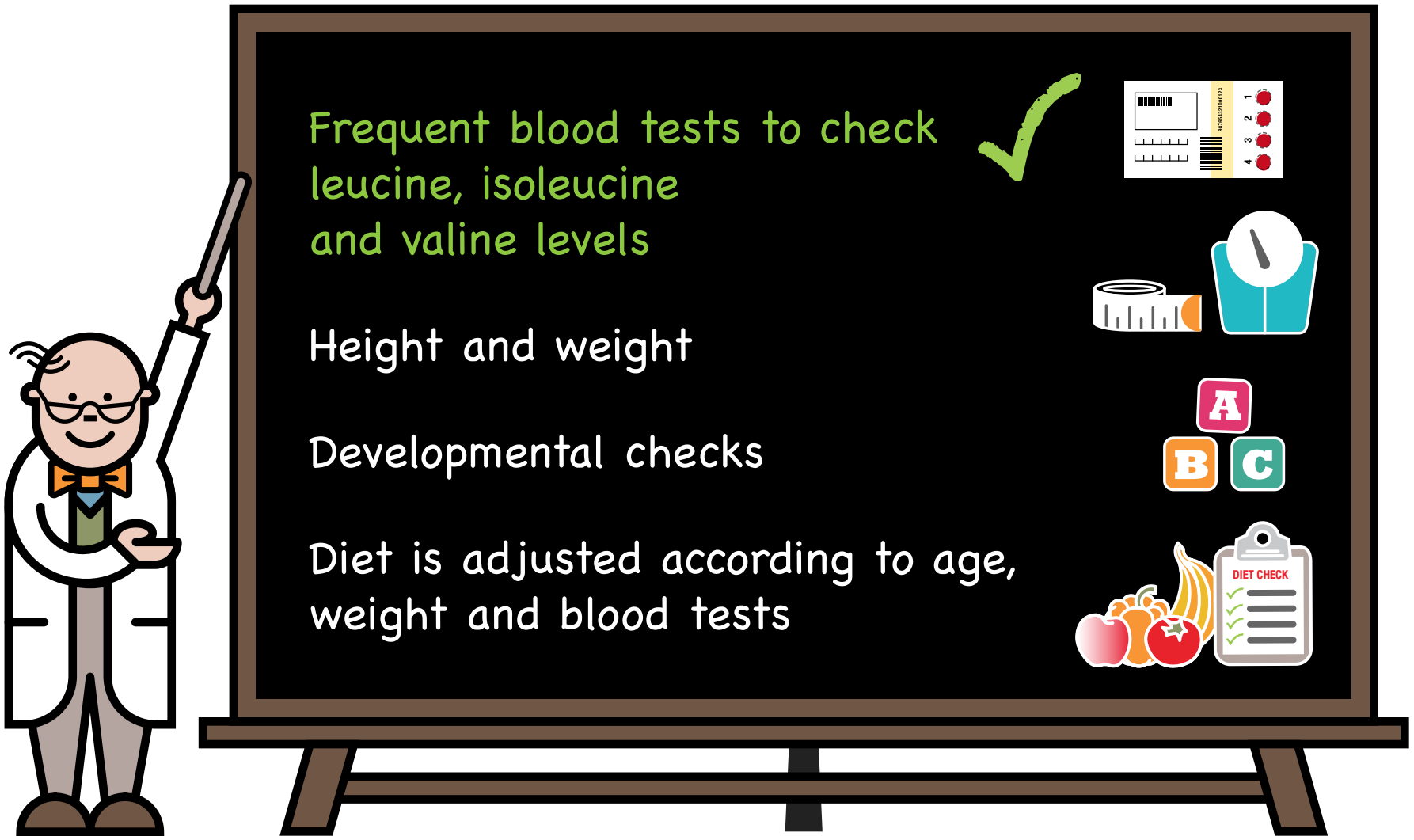
Checklist for illness

Checklist for illness

Key message



How is MSUD monitored?



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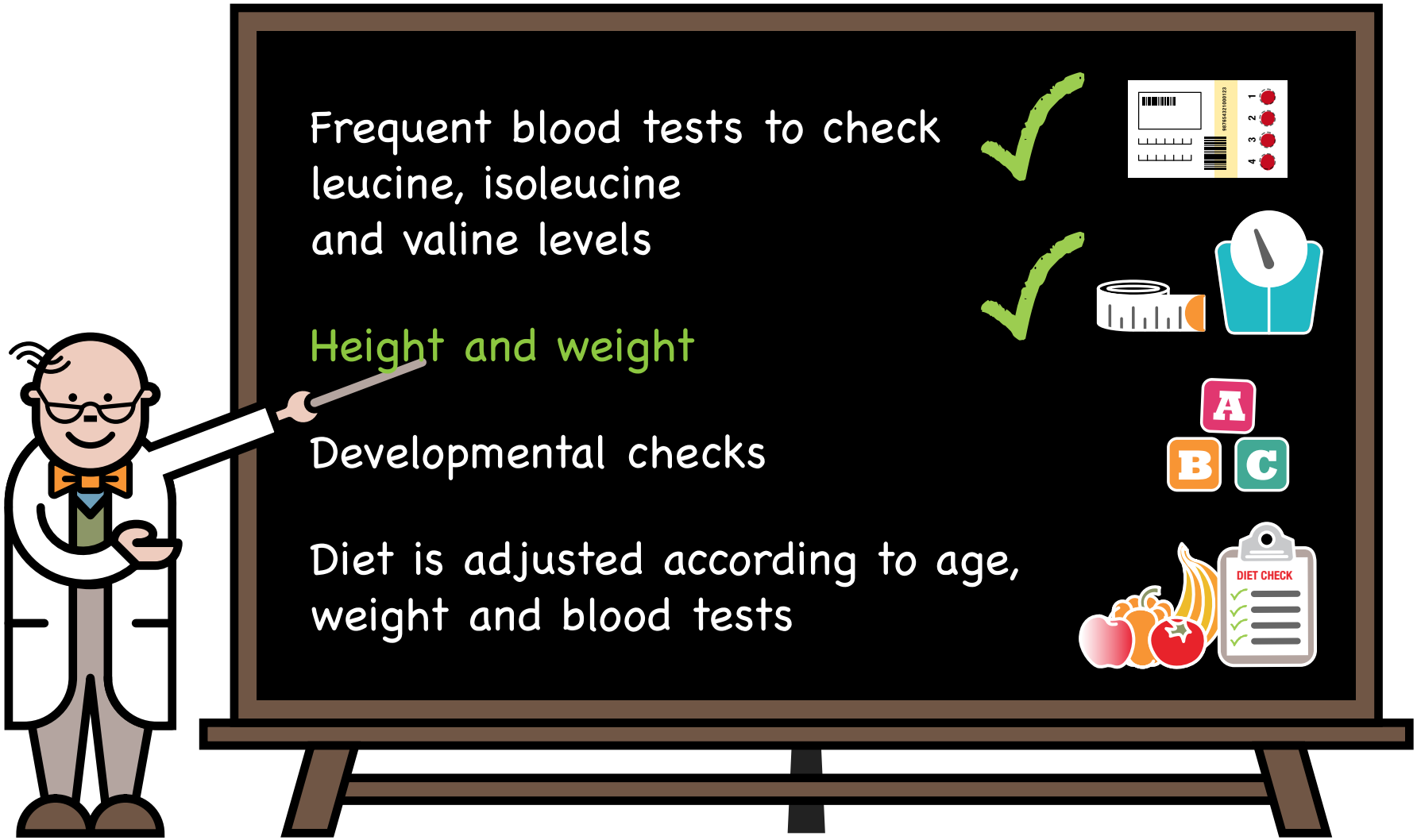

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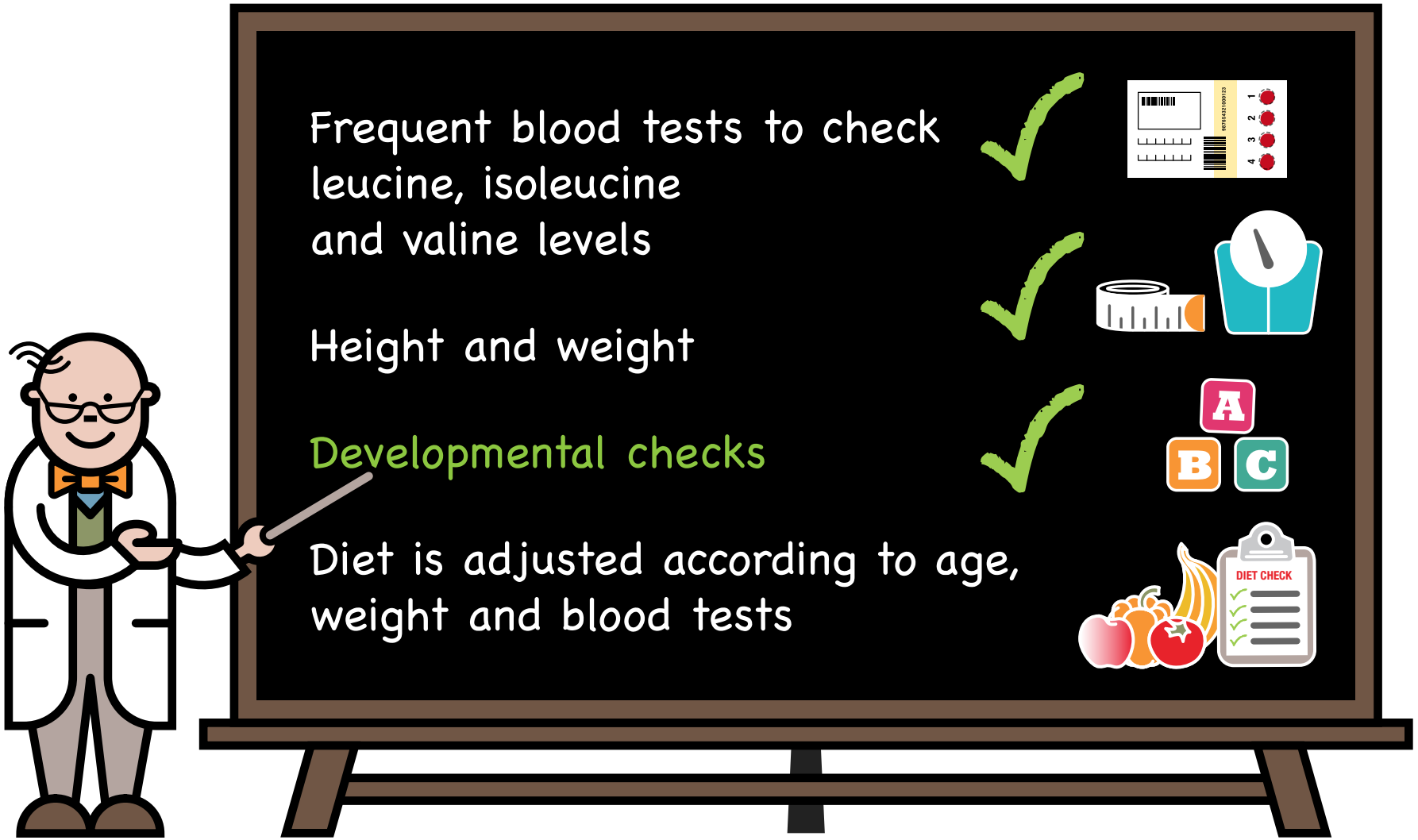

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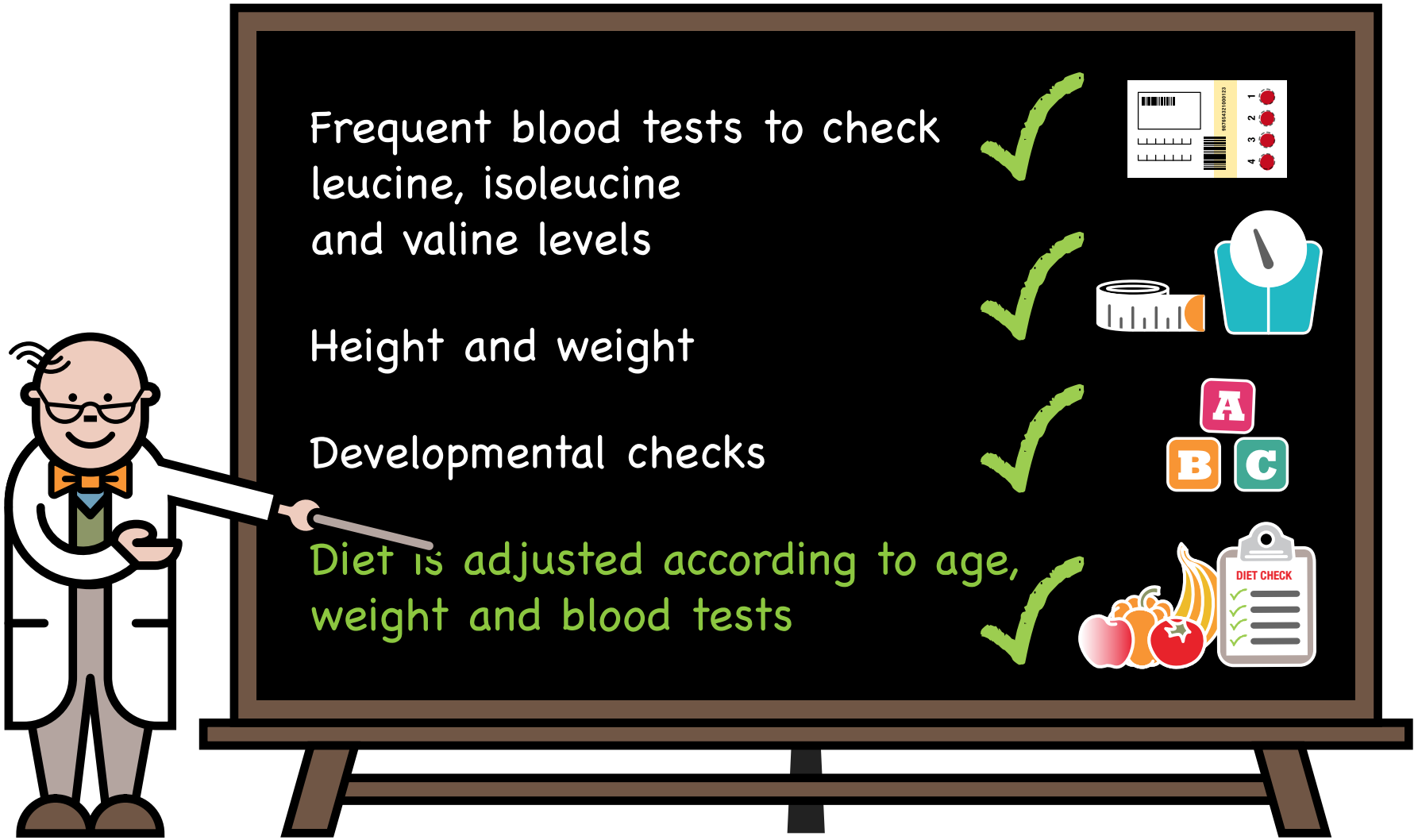
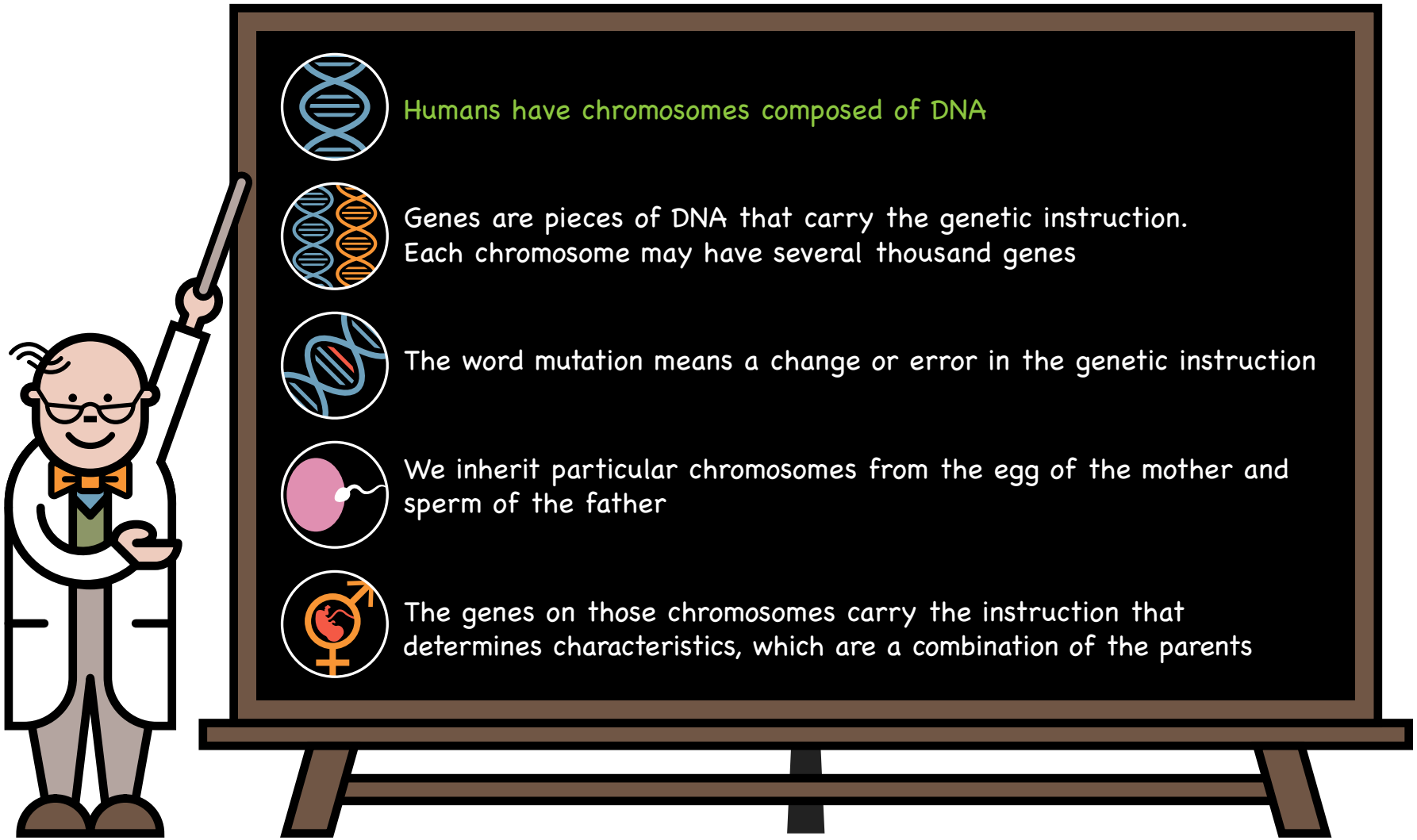


Chromosomes, genes, mutations



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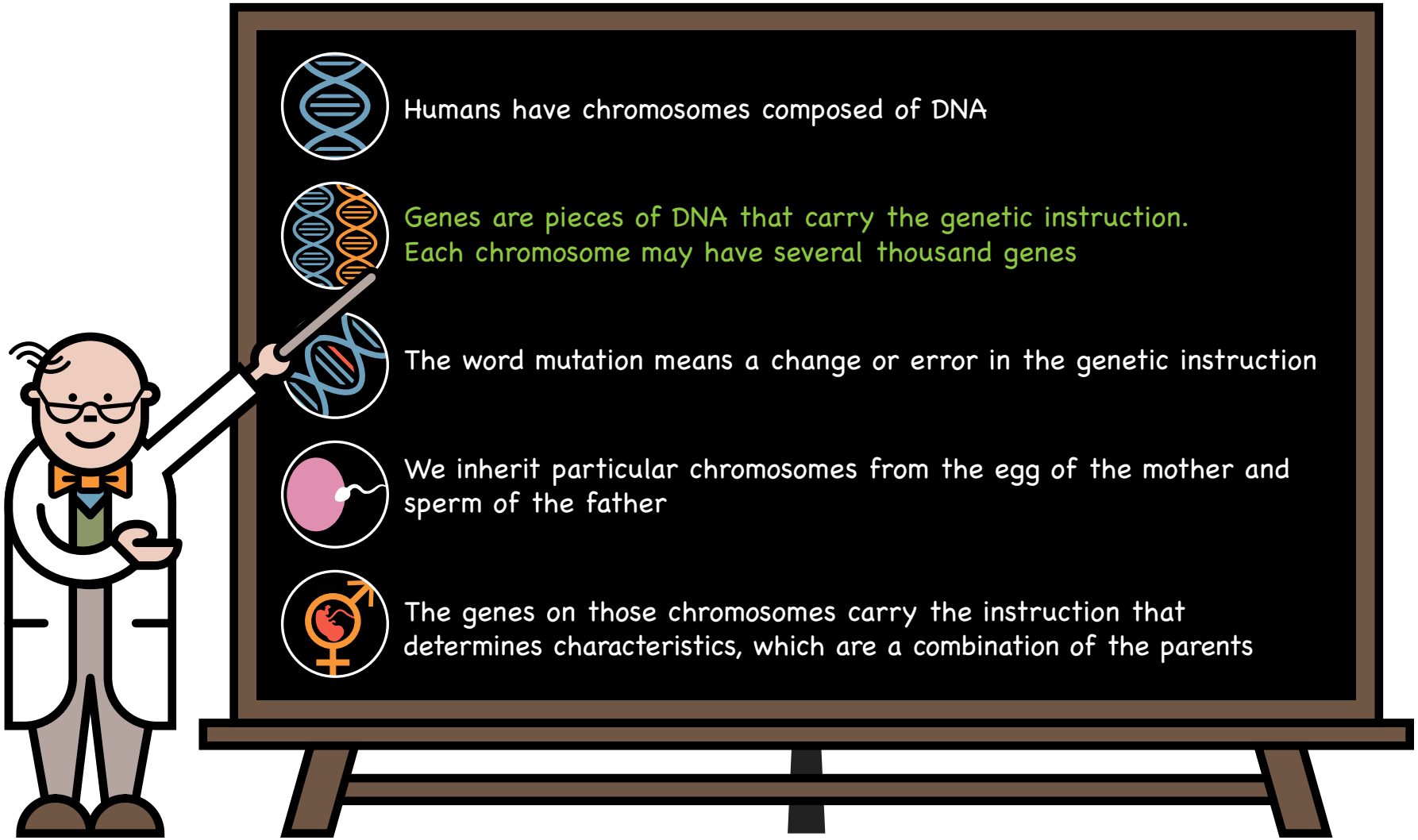
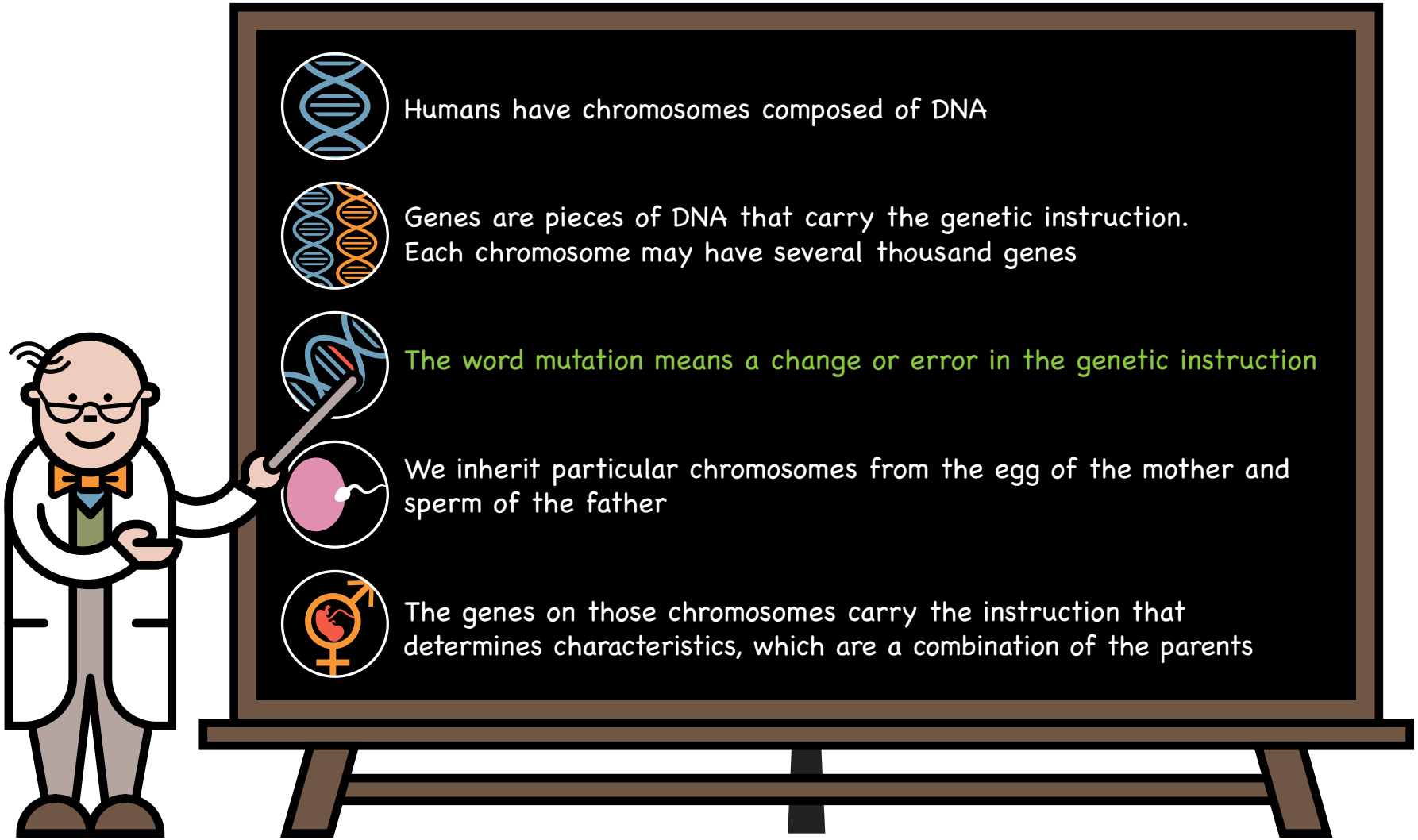


Chromosomes, genes, mutations



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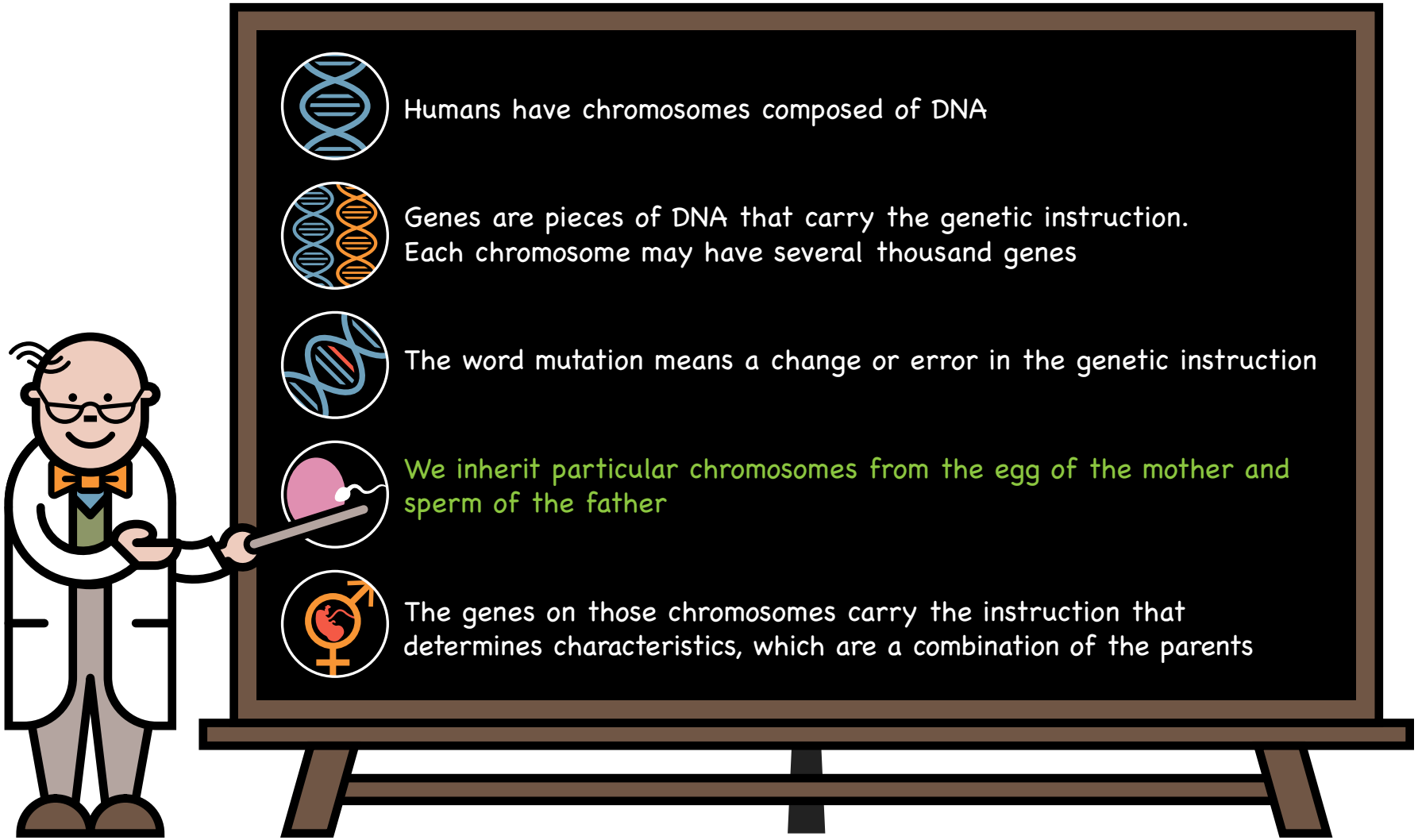

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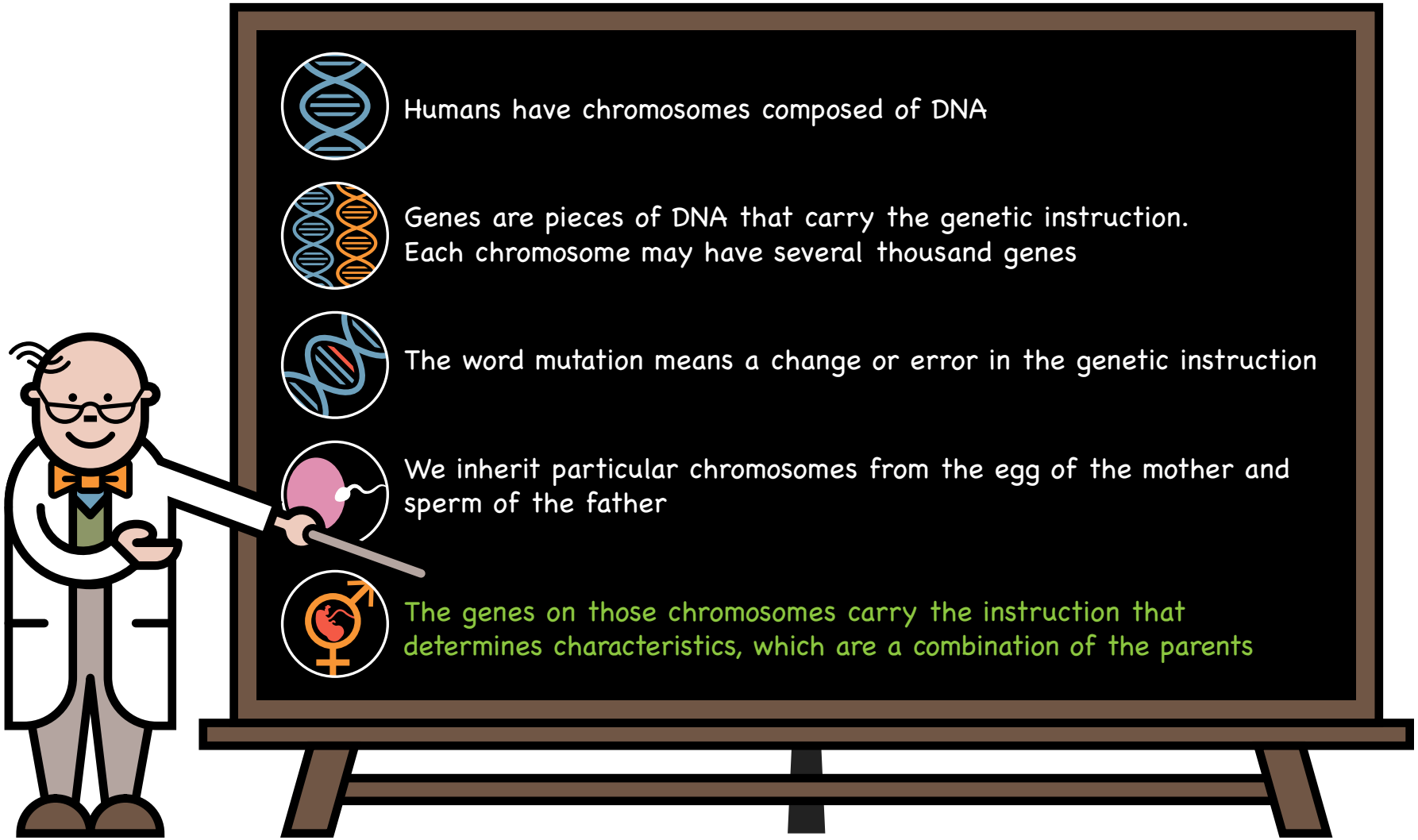
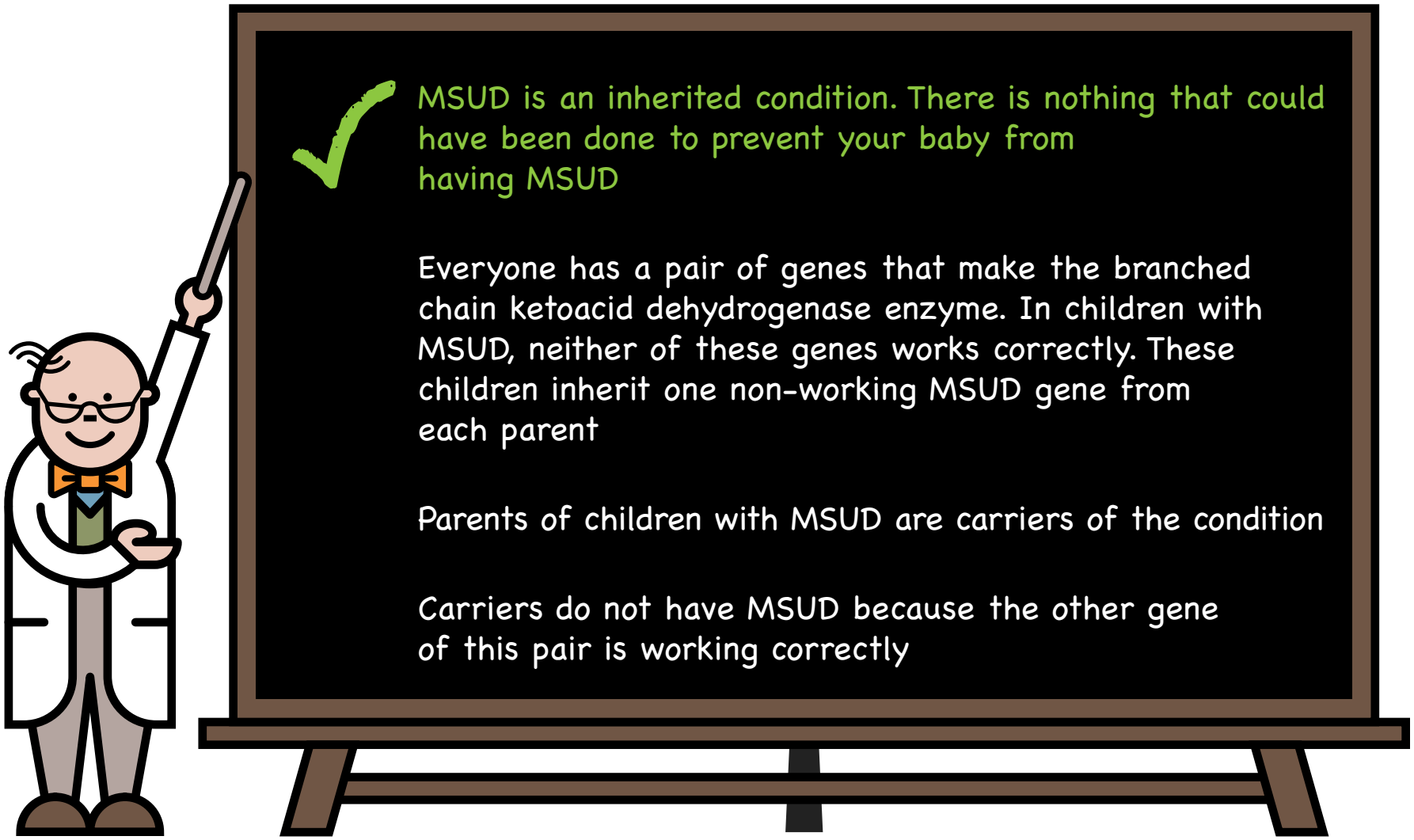


Inheritance



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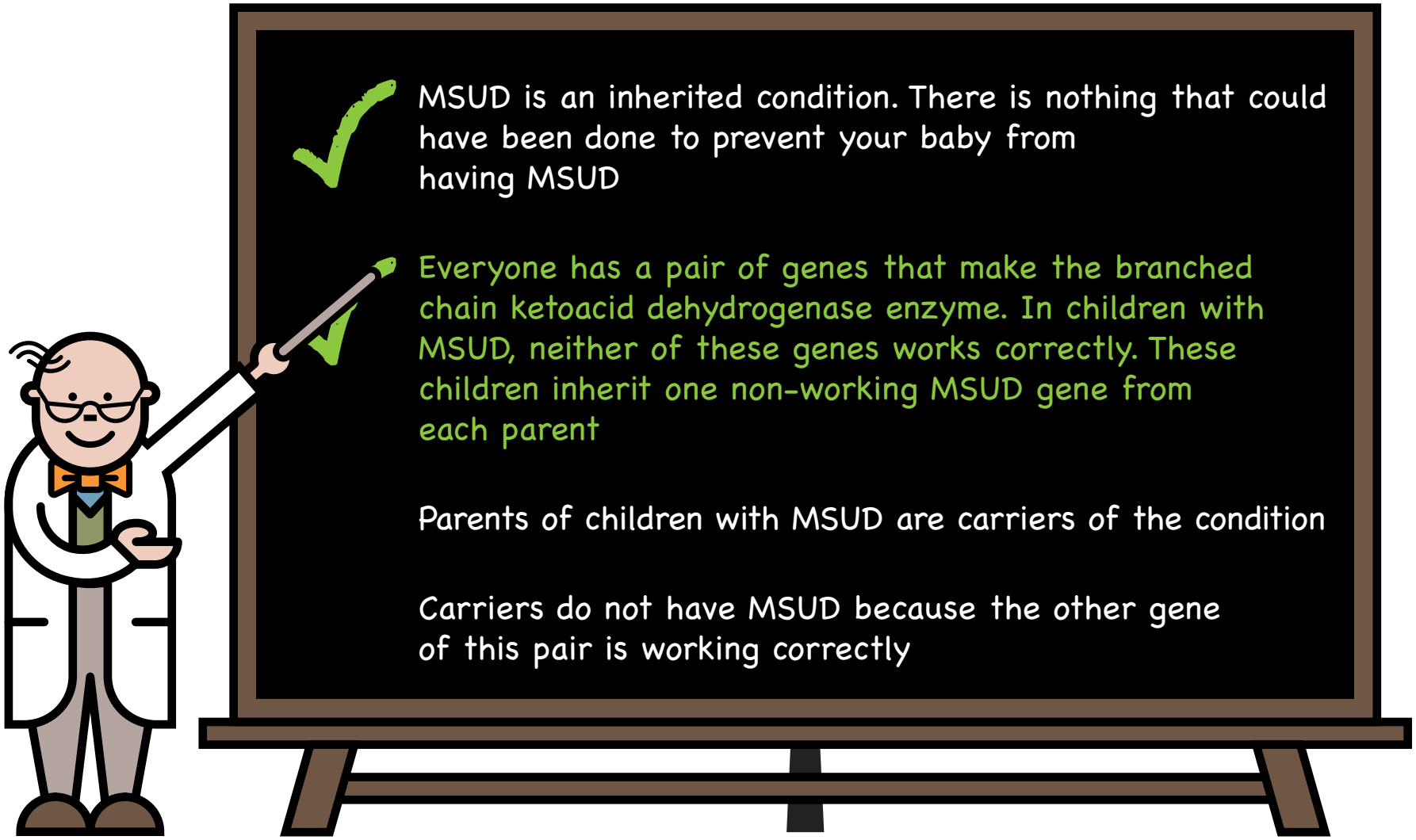

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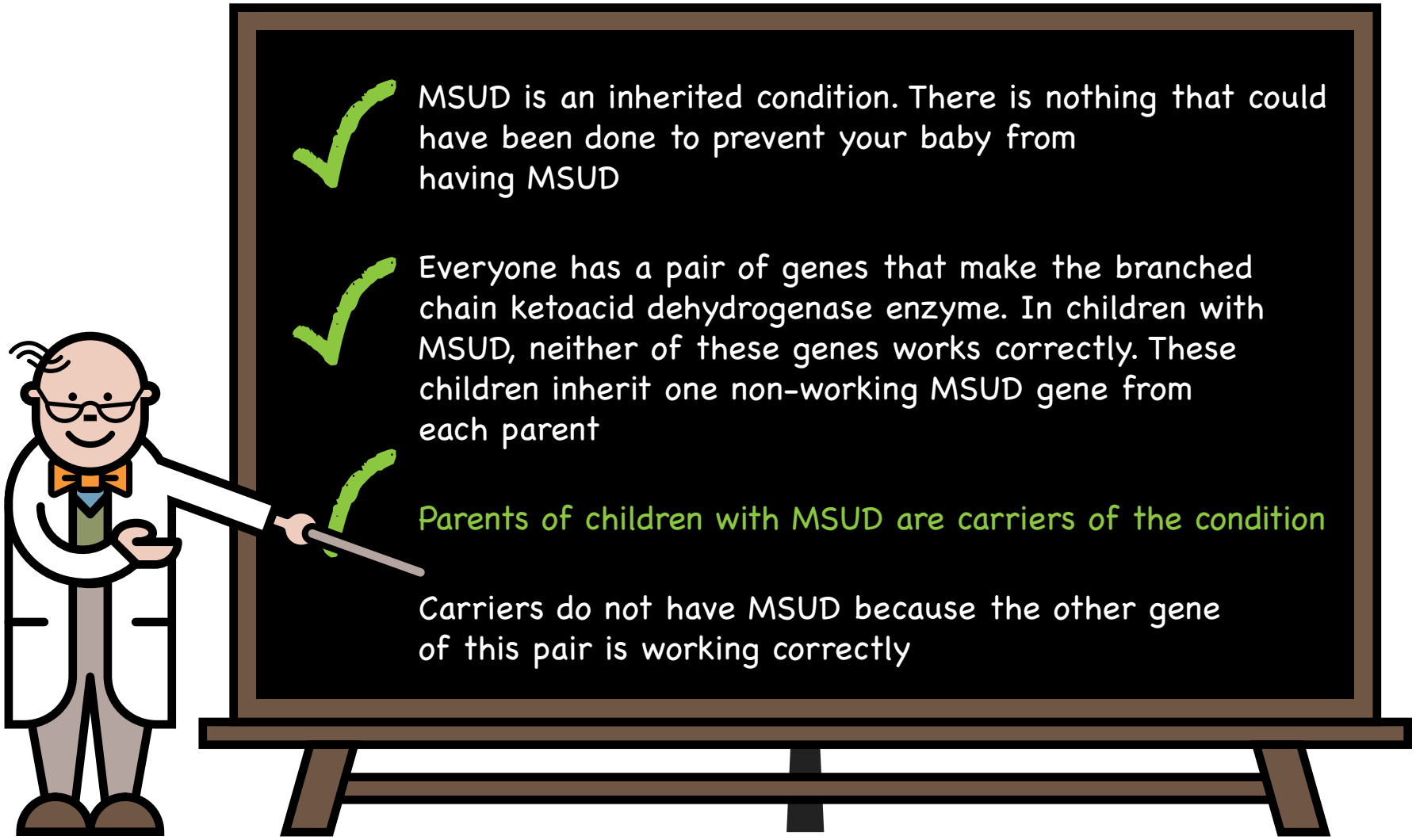

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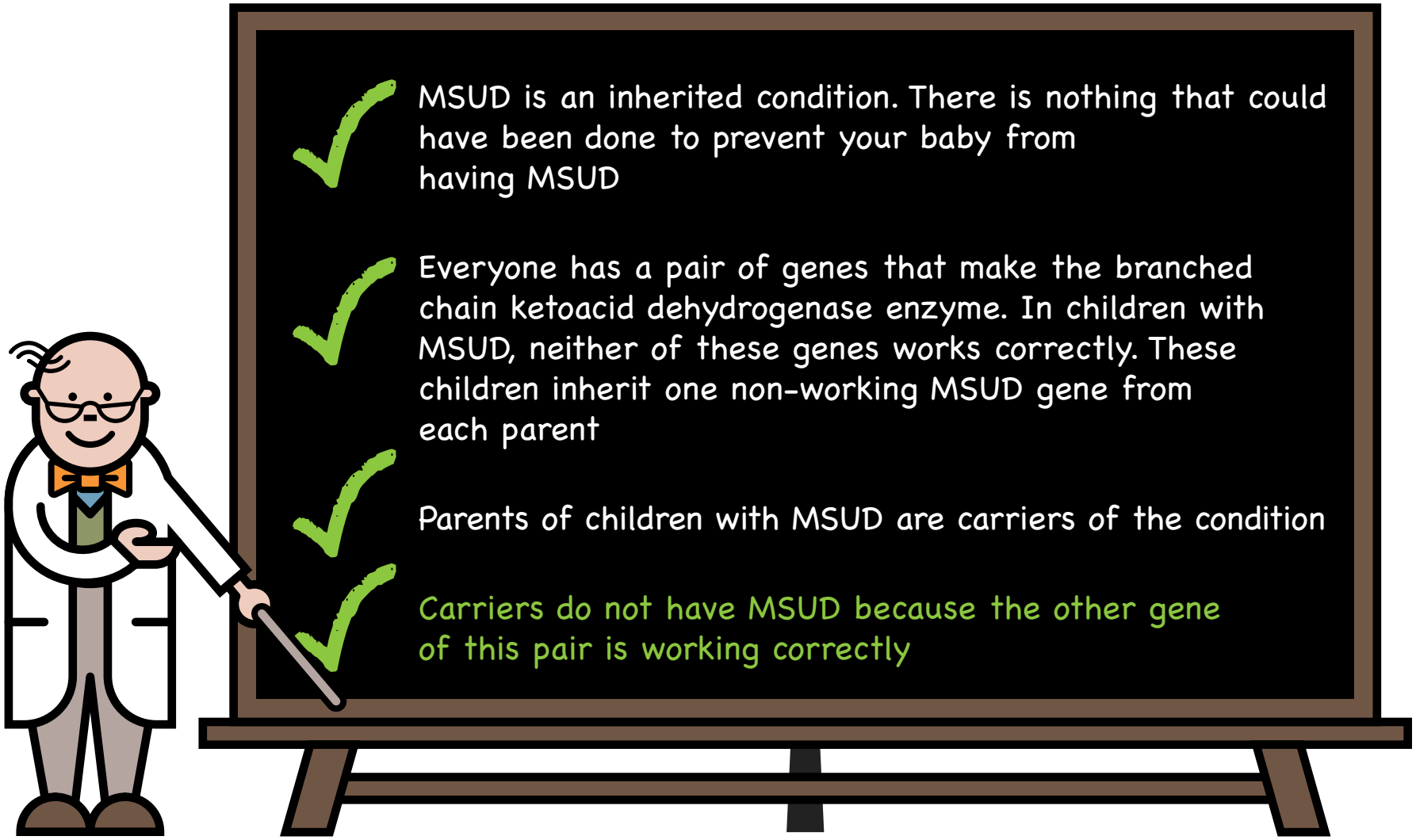
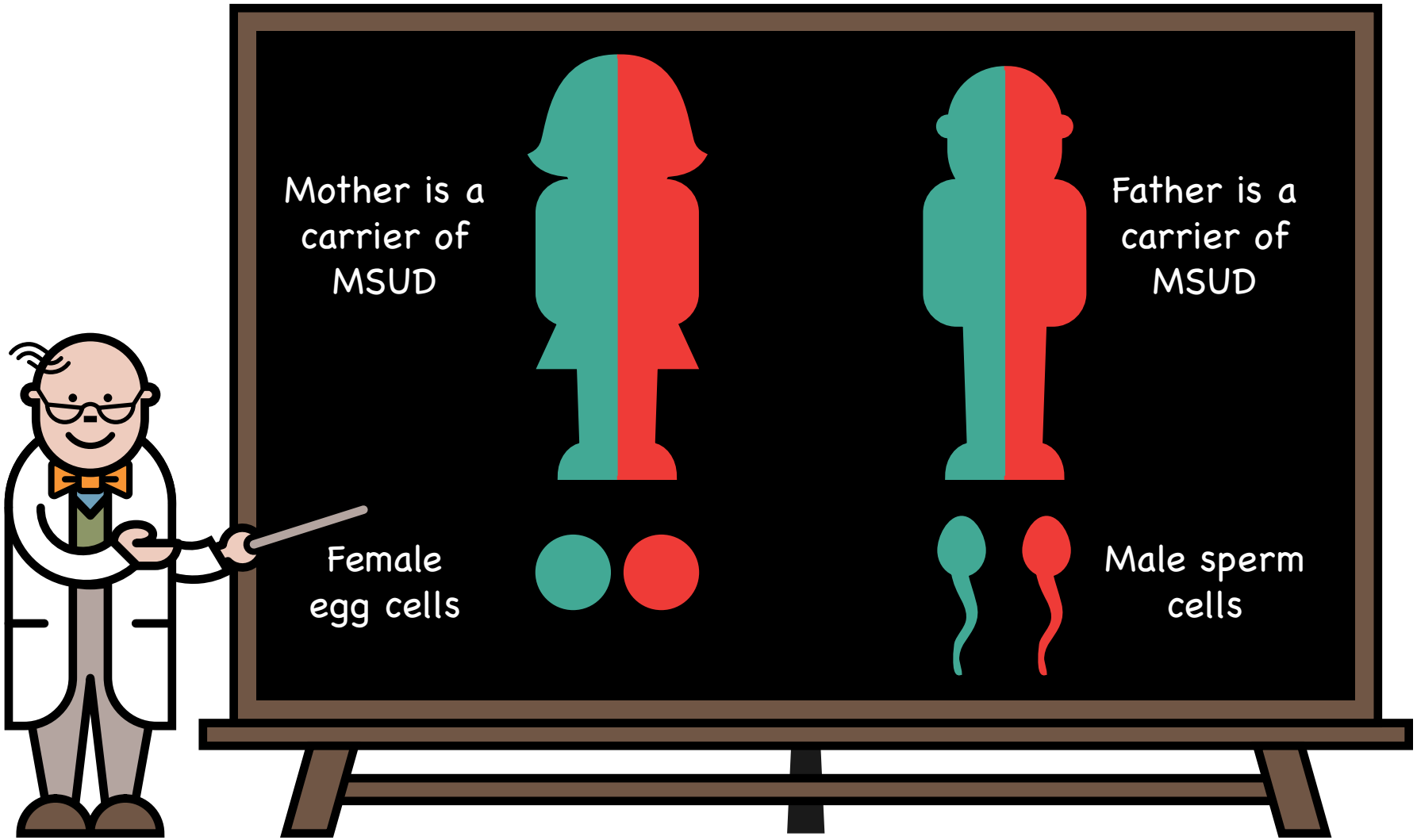
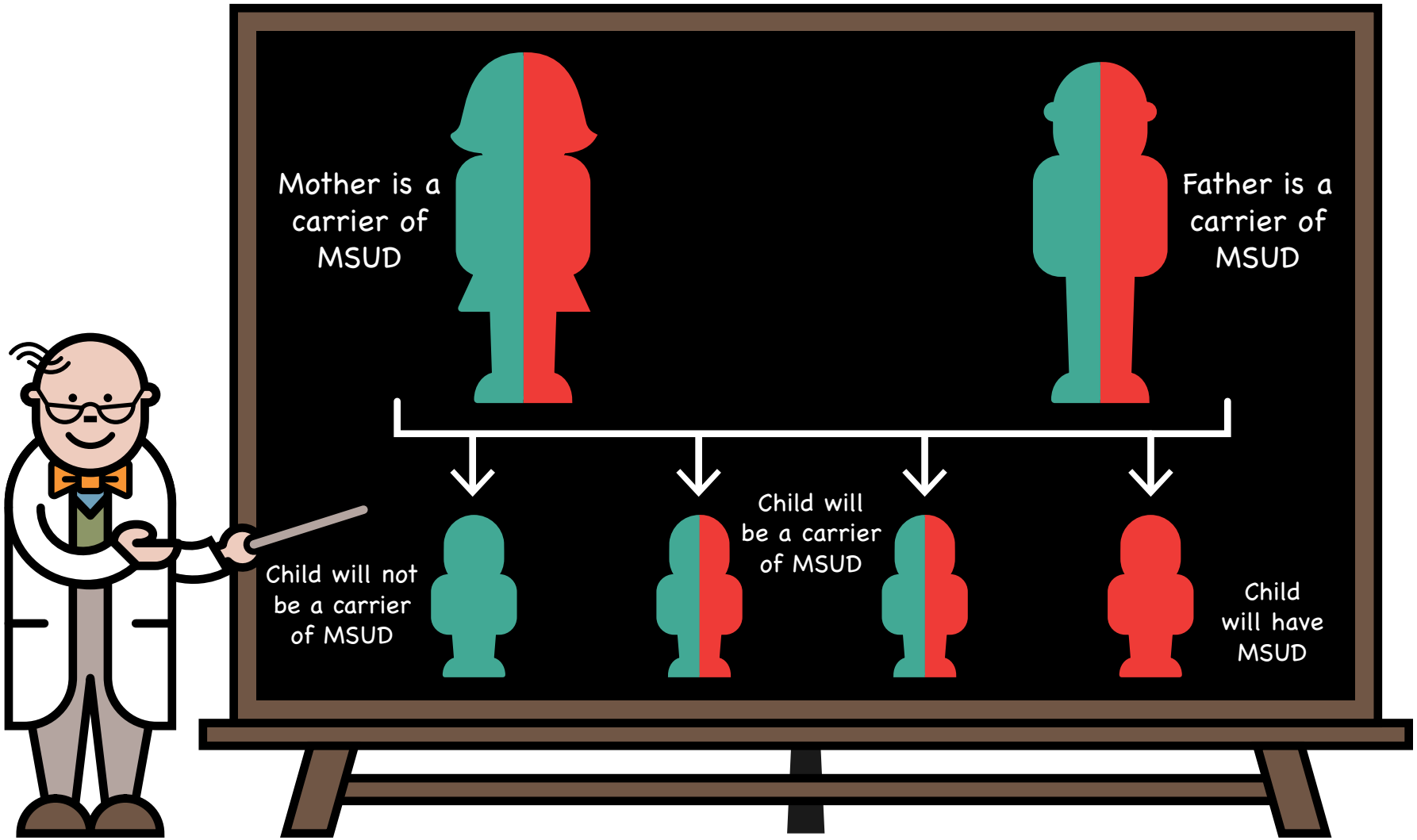


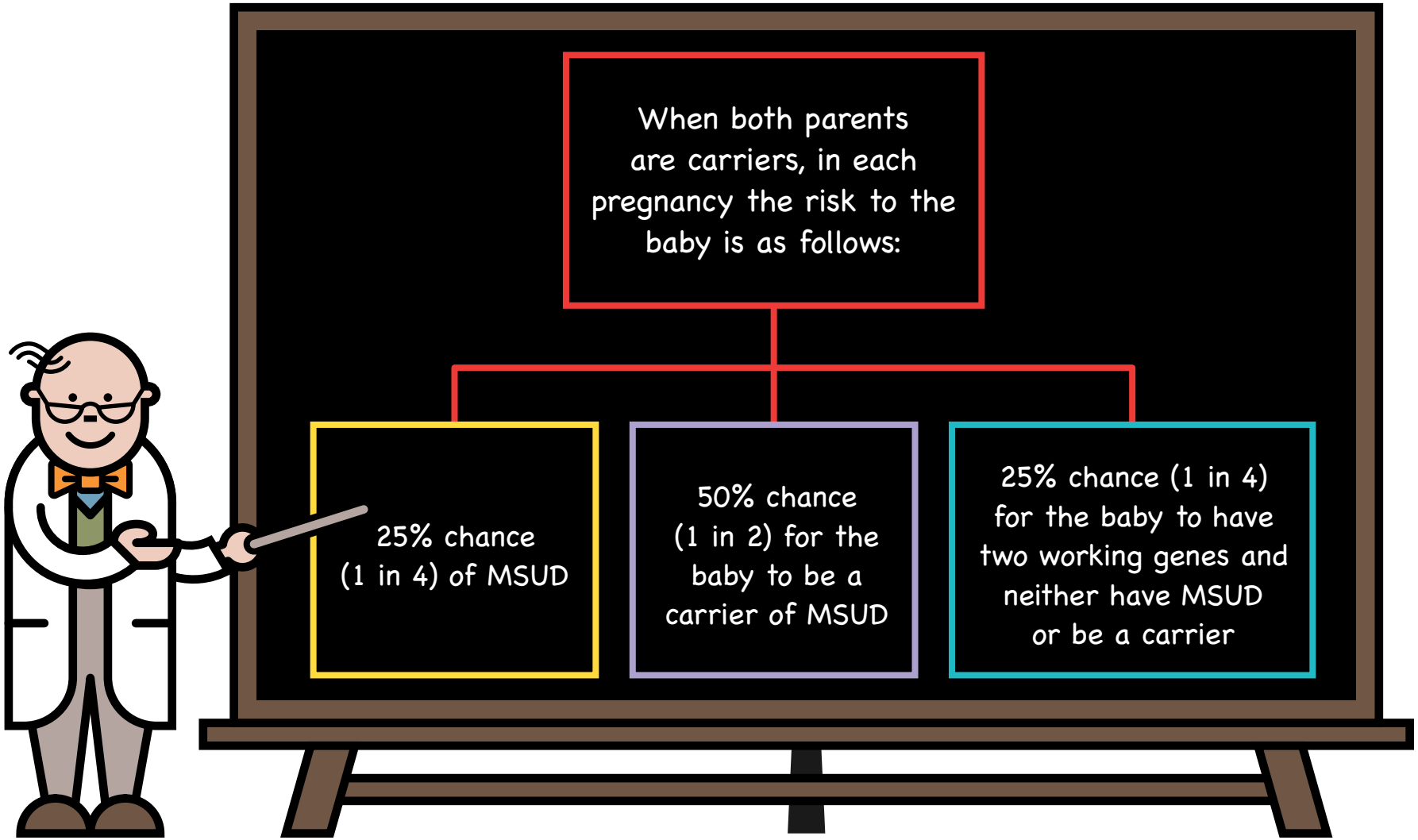
Inheritance – Autosomal recessive (carriers of MSUD)



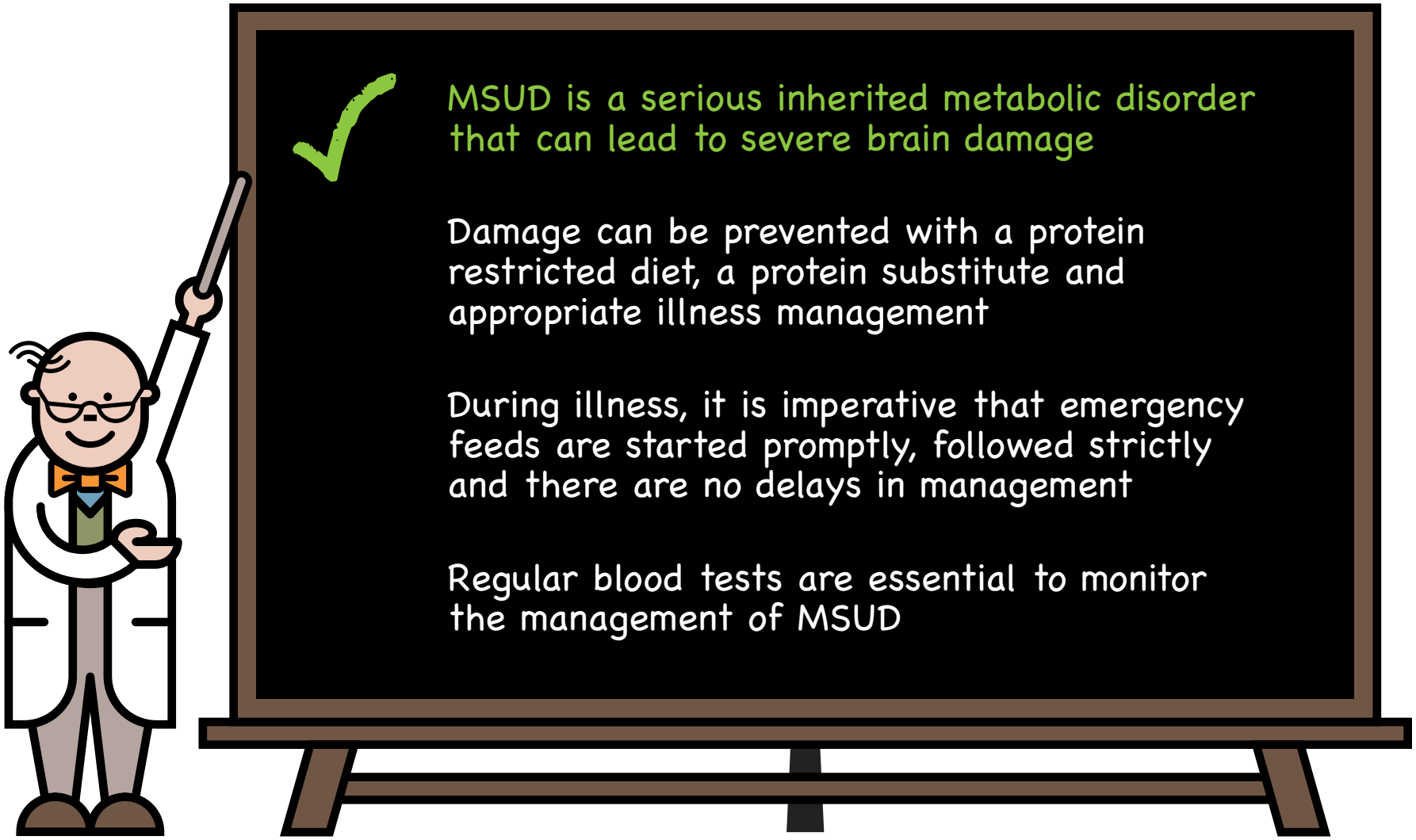
Inheritance – Autosomal recessive – possible combinations



Future pregnancies



Take home messages



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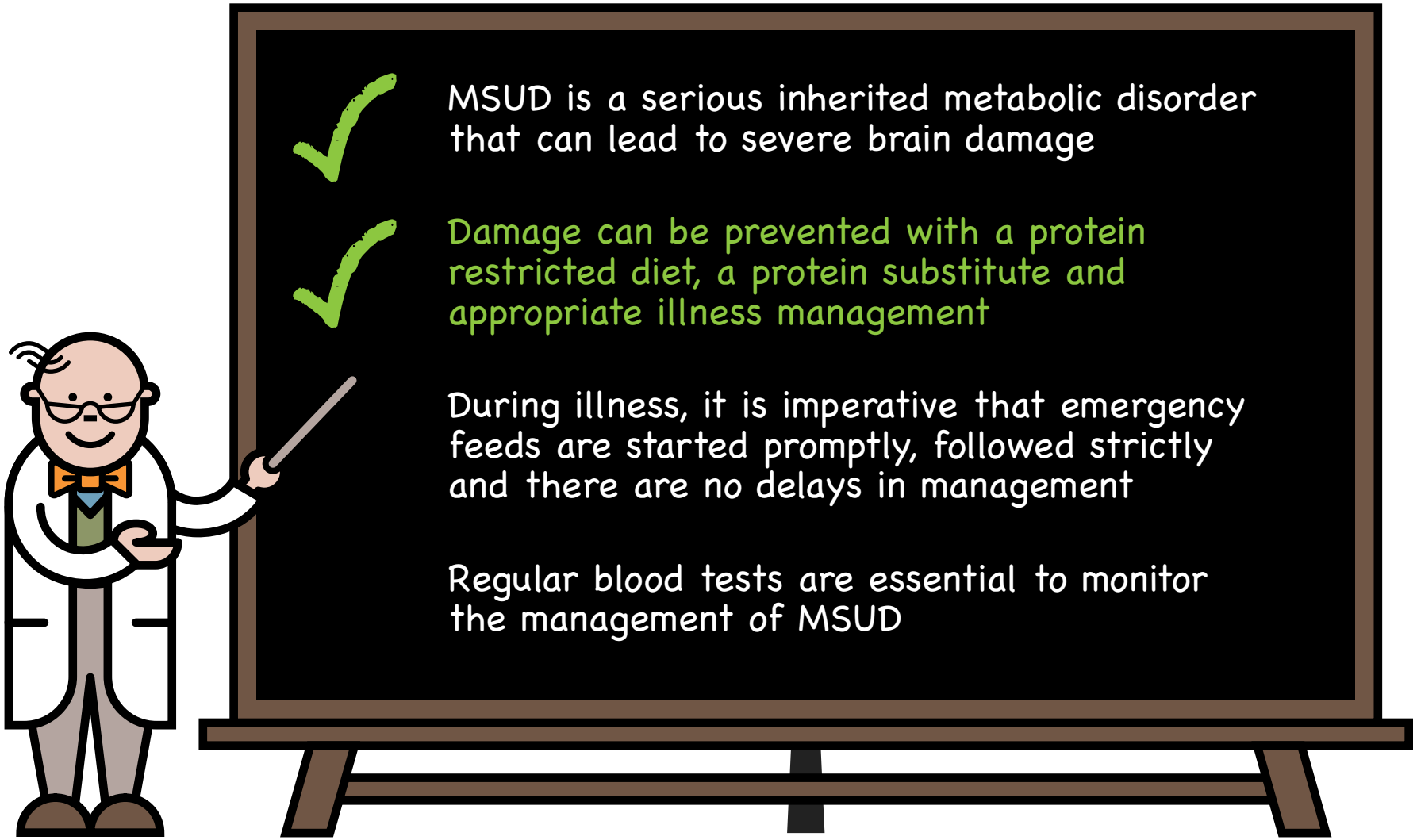

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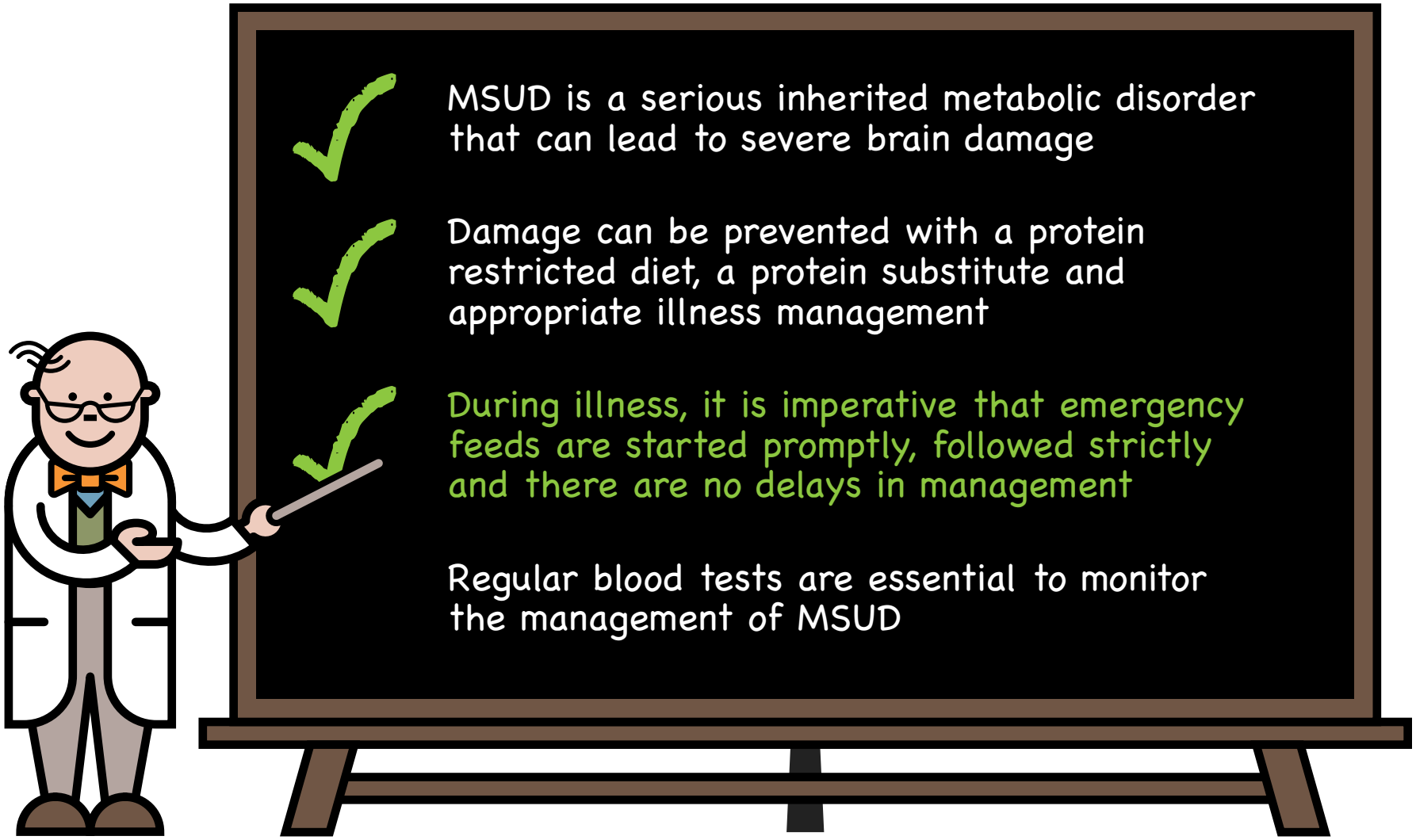

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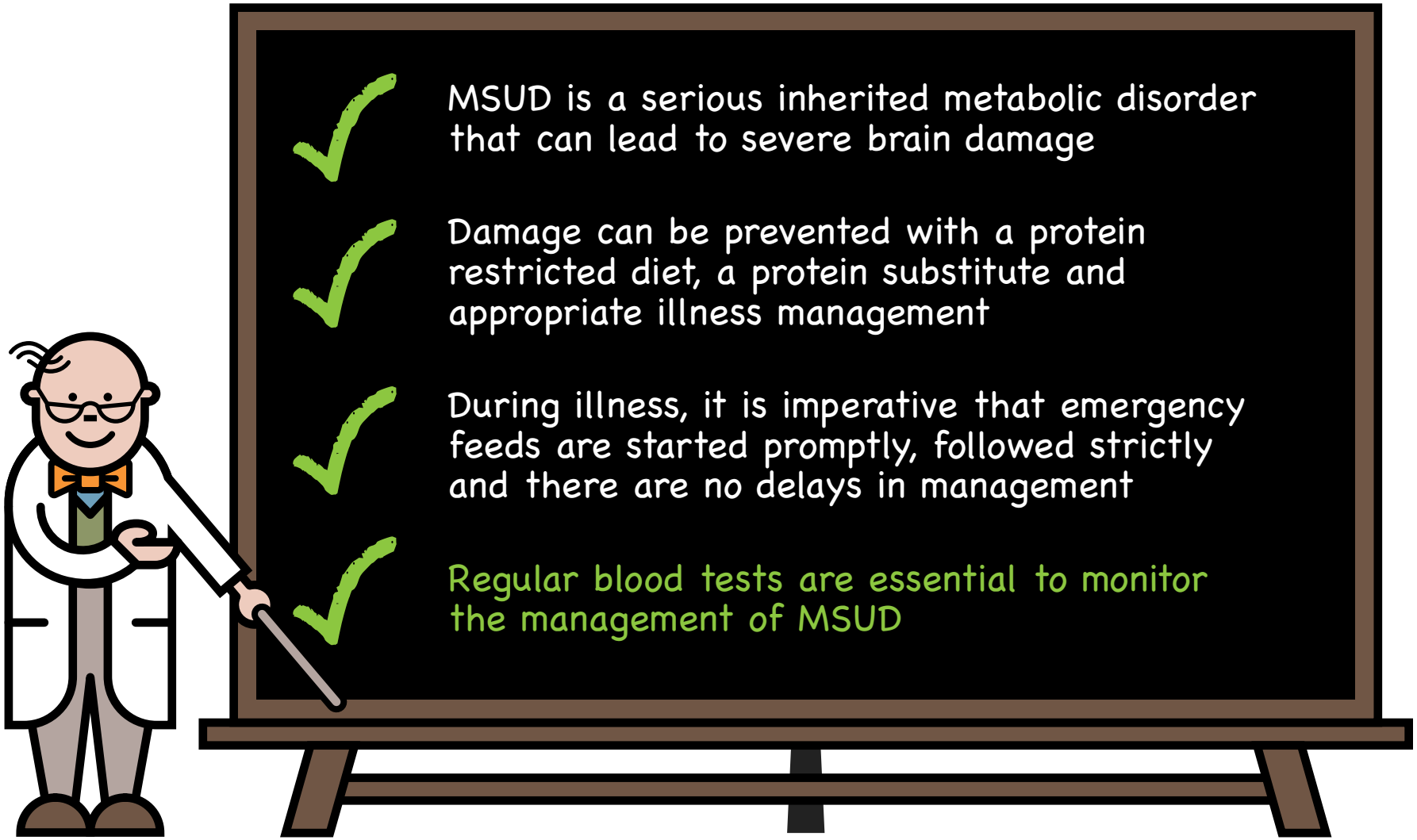

Helpful hints

Helpful hints

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Helpful hints

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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