TEMPLE

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

Ornithine transcarbamylase deficiency

ADAPTED BY THE DIETITIANS GROUP

BIMDG
British Inherited Metabolic Diseases Group

BASED ON THE ORIGINAL TEMPLE WRITTEN BY BURGARD AND WENDEL

VERSION 3, MARCH 2025

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.

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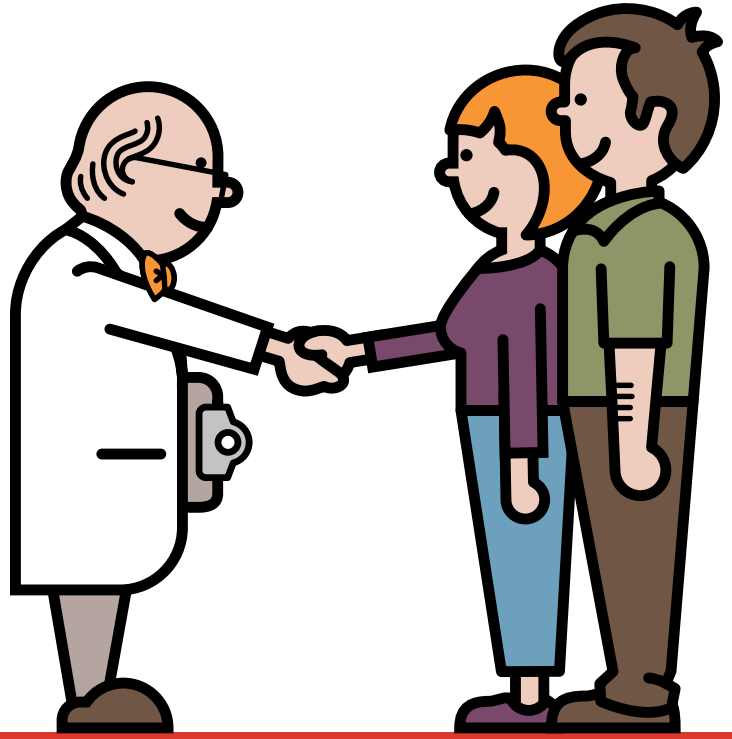
Ornithine transcarbamylase deficiency

Information for families following a new diagnosis



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Tools Enabling Metabolic Parents LEarning

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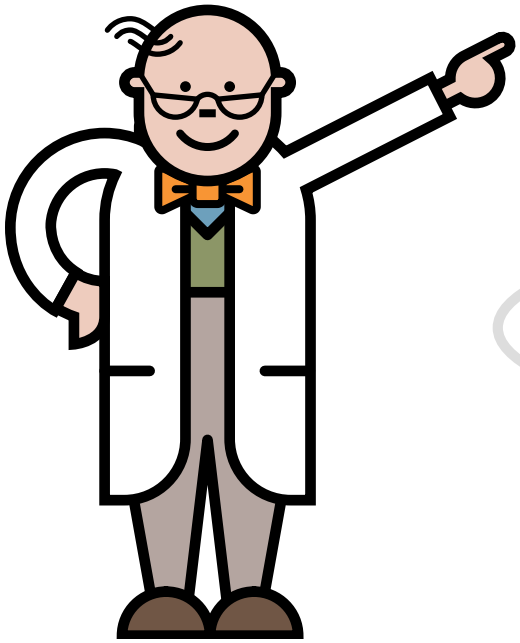
What is Ornithine transcarbamylase deficiency?

It is an inherited metabolic condition.

It is shortened to OTC deficiency.

Ornithine Trans Carbamylase deficiency



Ornithine Trans Carbamylase deficiency

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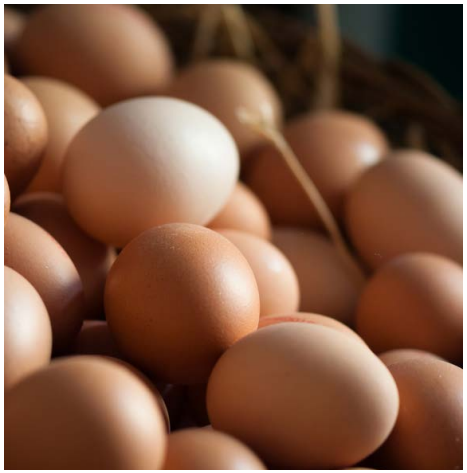
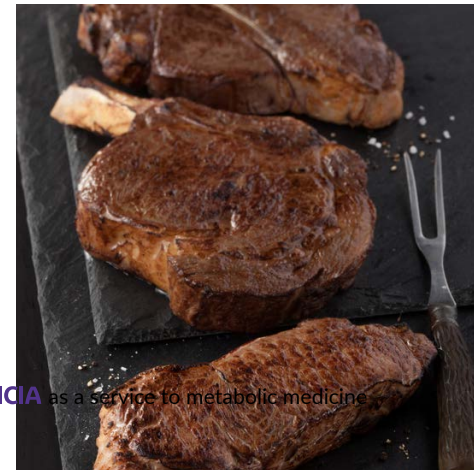
What is protein?

Many foods contain protein.

The body needs protein for growth and repair.

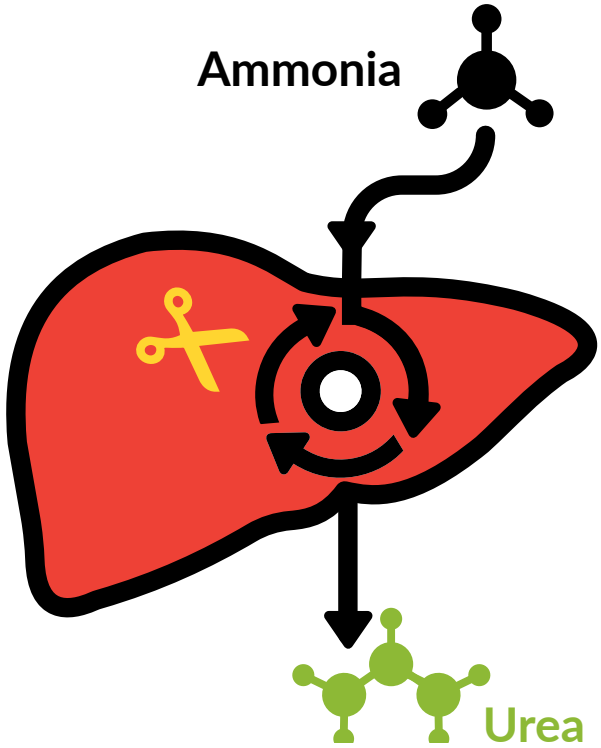
Many people eat more protein than the body needs.





How do we remove waste protein from the body?

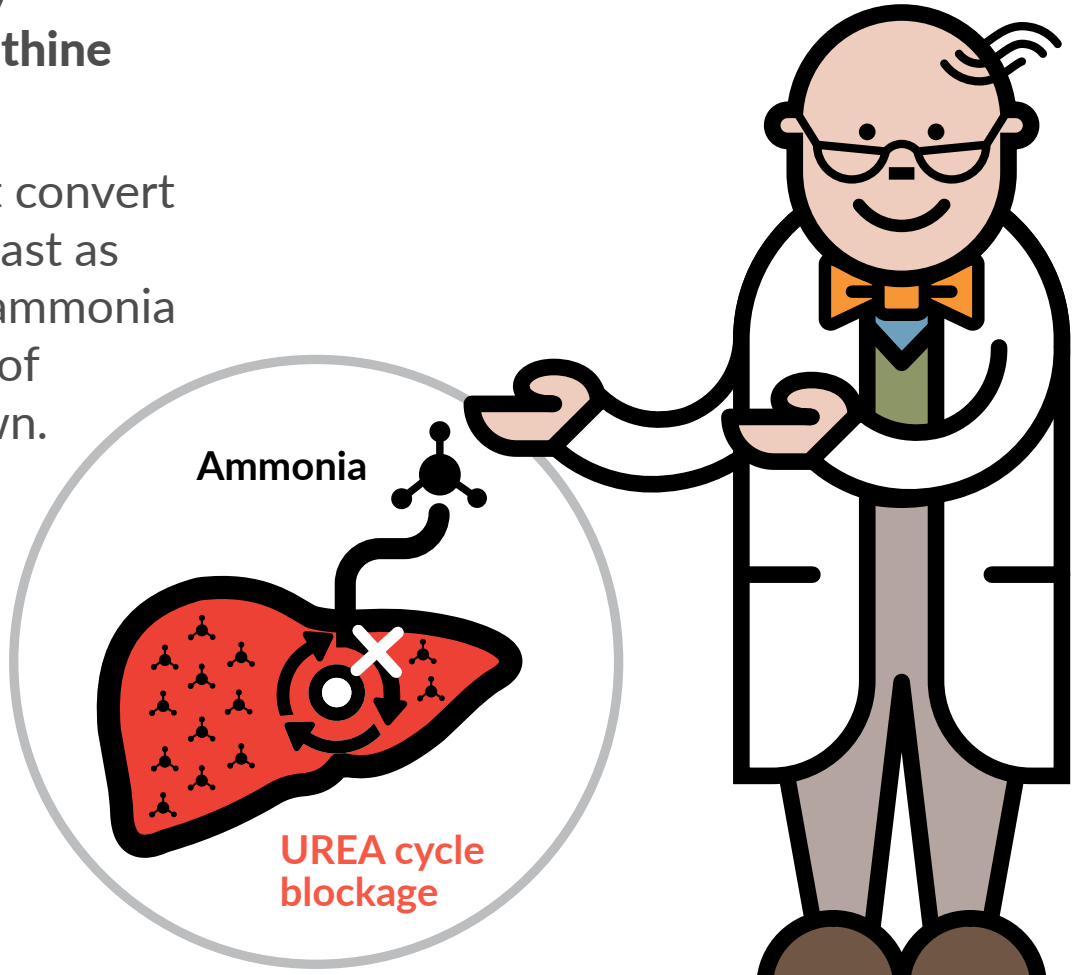
- Firstly, the body converts waste protein to a toxic chemical called **ammonia**
- Ammonia is then converted into a non-toxic chemical **(urea)** in the liver
- This process occurs via the **urea cycle**
- In the urea cycle, several steps have to take place. Each step needs an enzyme (like chemical scissors) for it to work
- Urea is then removed by the kidneys



What happens in OTC deficiency?

In OTC deficiency, the body lacks an enzyme called **ornithine transcarbamylase**.

This means the liver cannot convert waste protein into urea as fast as normal. It can lead to high ammonia levels, particularly at times of increased protein breakdown.

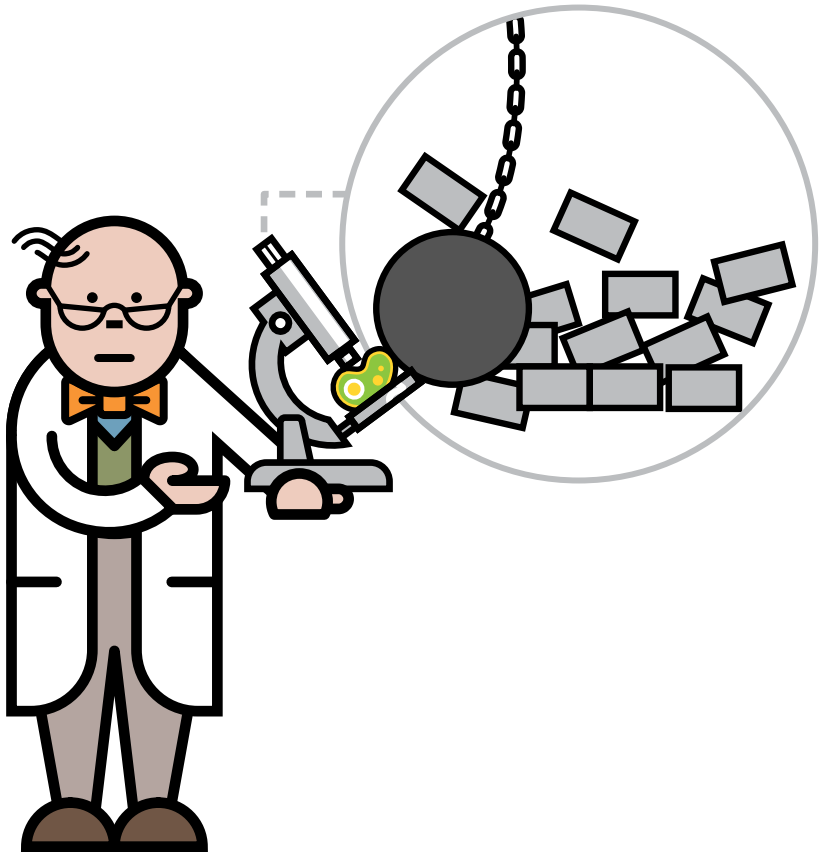


When does OTC deficiency cause high ammonia levels?

Ammonia levels can rise when there is an increased break down of protein. This may happen if too much protein is eaten.

It commonly results from break down of the body's own protein. This is often triggered by infections, particularly if there is vomiting.

This causes **catabolism** which is a break down of body protein and can lead to a metabolic crisis.



What are the symptoms in OTC deficiency?

Some babies become ill in the first few days of life.

Signs and symptoms:

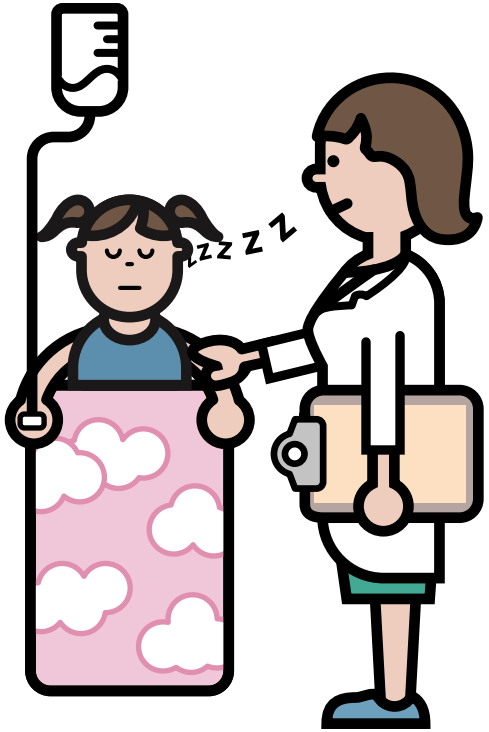
- Poor feeding
- Vomiting
- Floppiness
- Excessive sleepiness
- Rapid breathing
- Dehydration (lack of body fluids)
- Seizures

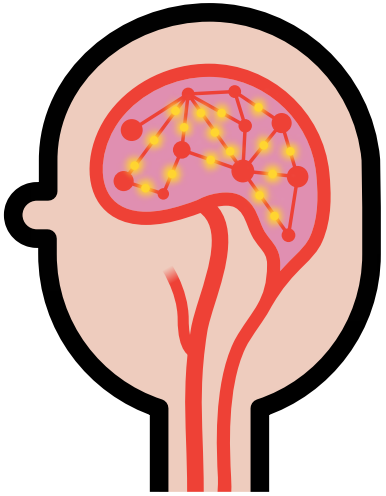
The effects of high ammonia can quickly become life-threatening if unmanaged.

Some children may be diagnosed at a later stage

Signs and symptoms:

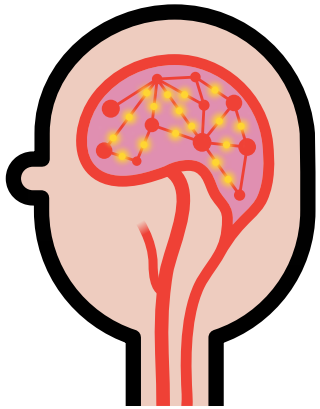
- Repeated episodes of vomiting, which may lead to sleepiness and coma
- Learning difficulties
- Seizures



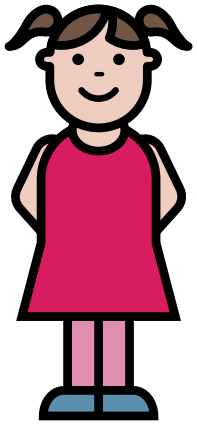


What are the long term effects of OTC deficiency?

It may cause learning difficulties.

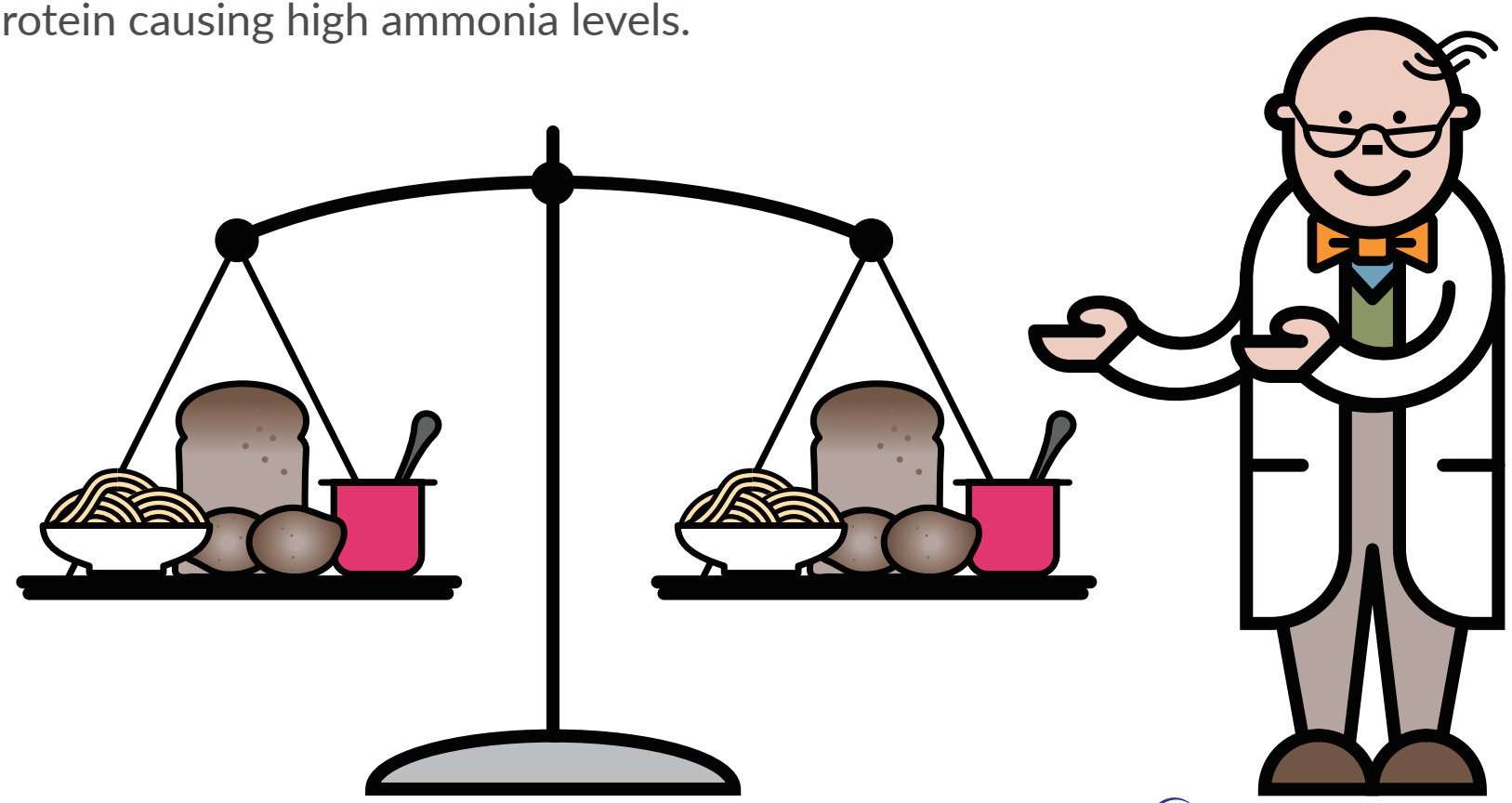


It may also cause delays to normal development like walking and talking.



Protein balance is needed in OTC deficiency

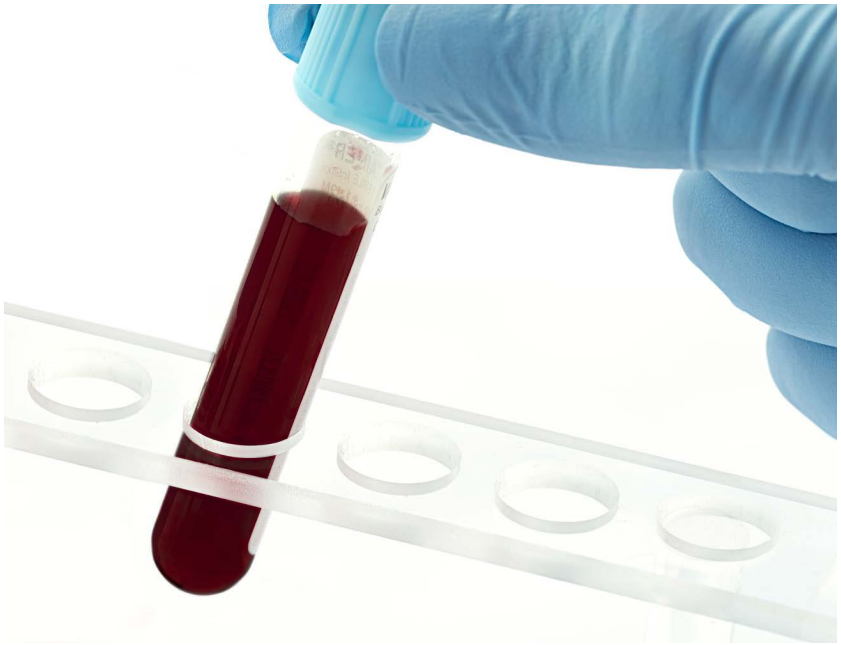
In OTC deficiency it is important that enough protein is given to grow… but not too much as it will make waste protein causing high ammonia levels.



How is OTC deficiency diagnosed?

The diagnosis is suspected in a patient with high ammonia levels because of the pattern of chemicals in the blood and urine.

The diagnosis is confirmed by finding a mutation in the OTC gene.



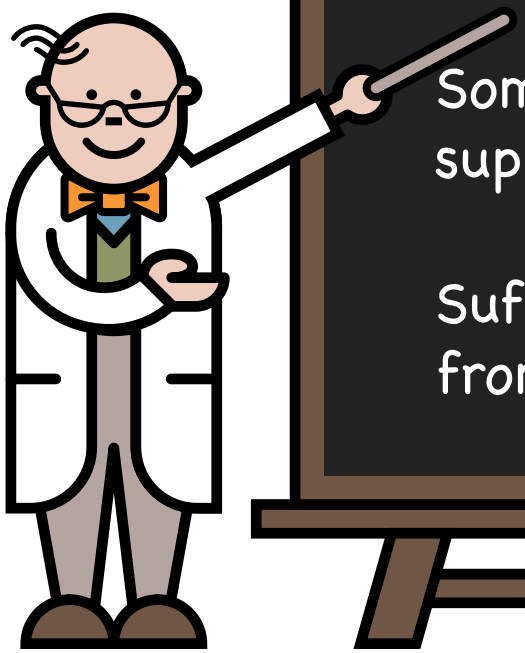
How is OTC deficiency managed day to day?

OTC deficiency is managed with the following:

A protein restricted diet

Sometimes a special amino acid supplement may be needed

Sufficient energy supply from food and feeds



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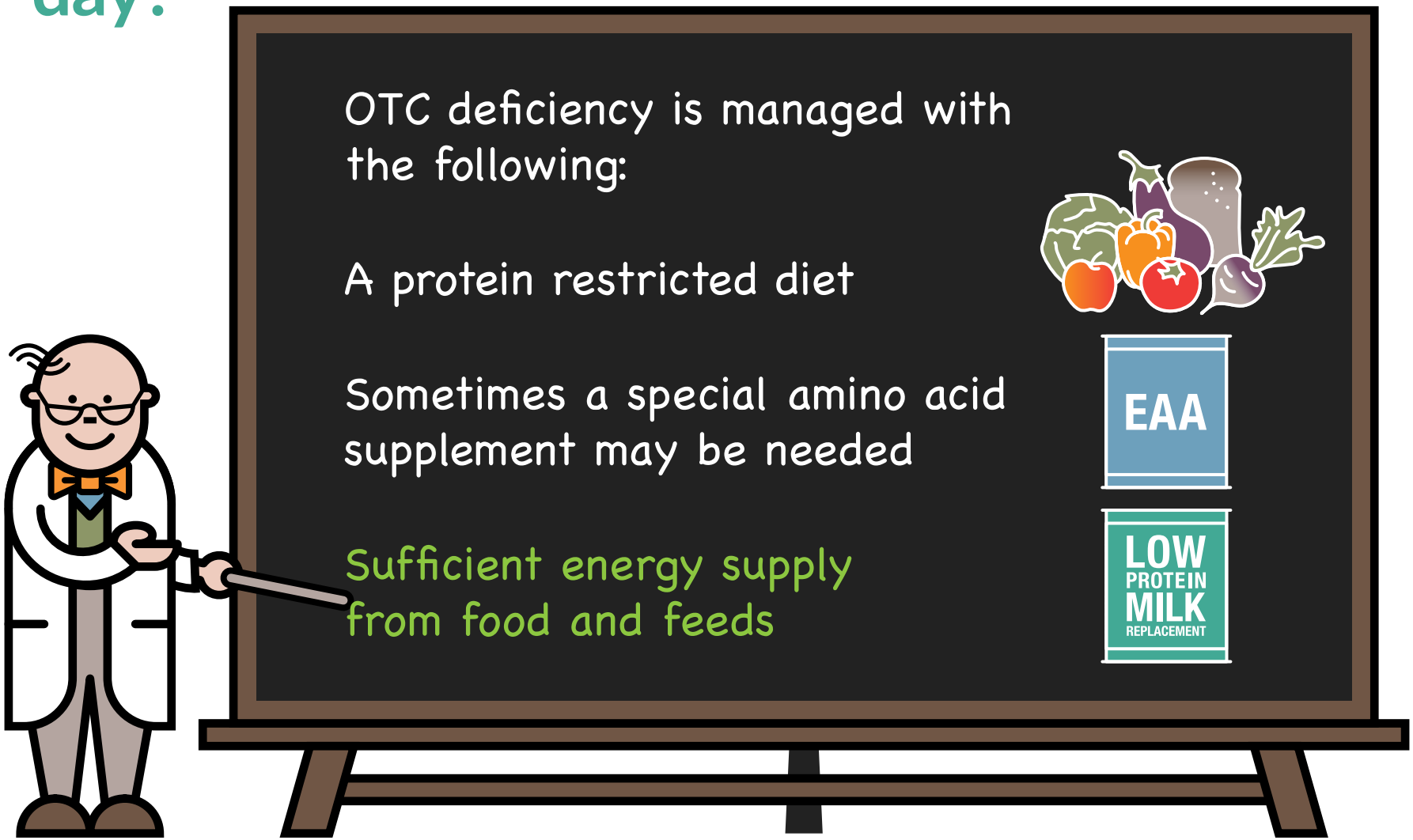

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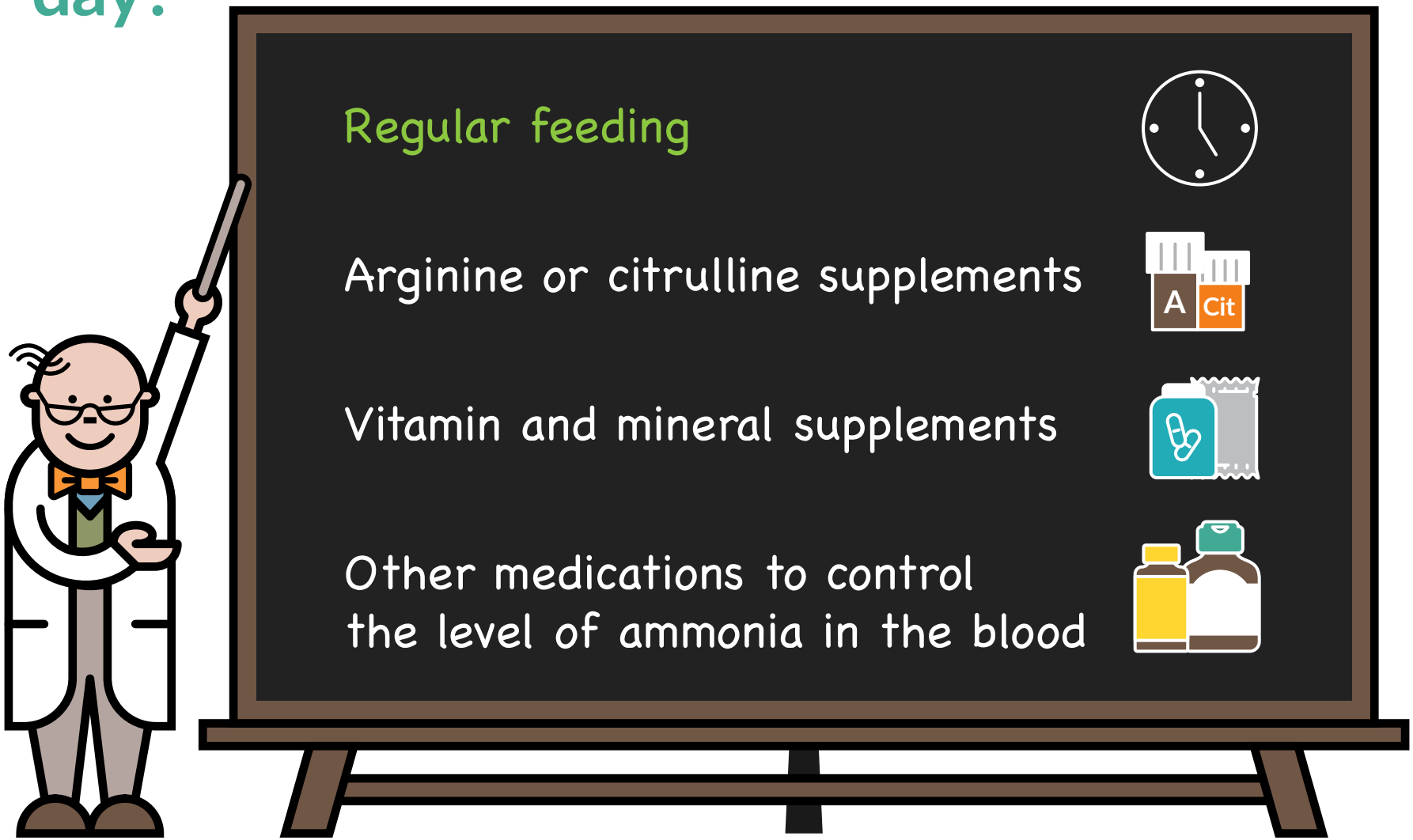

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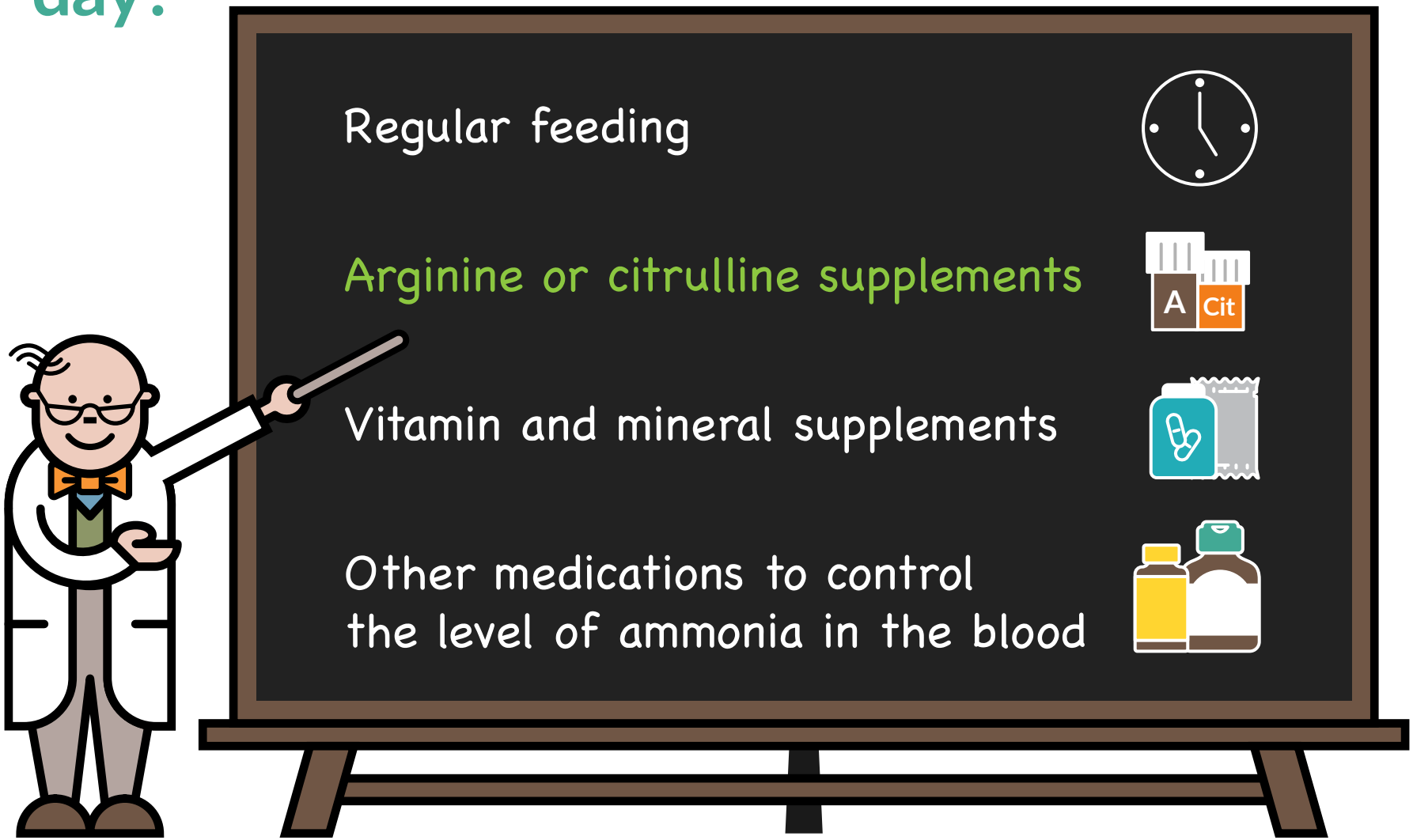

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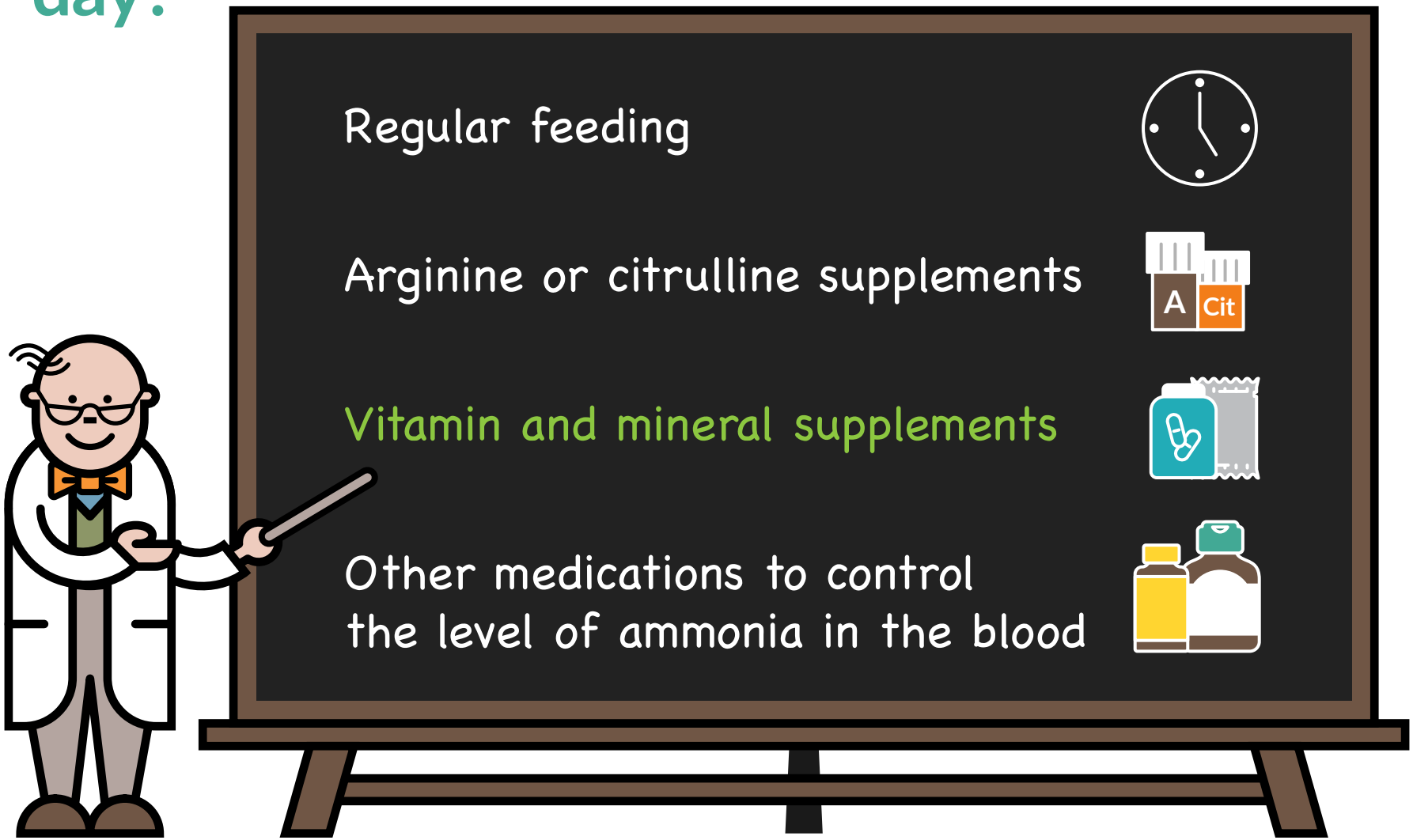

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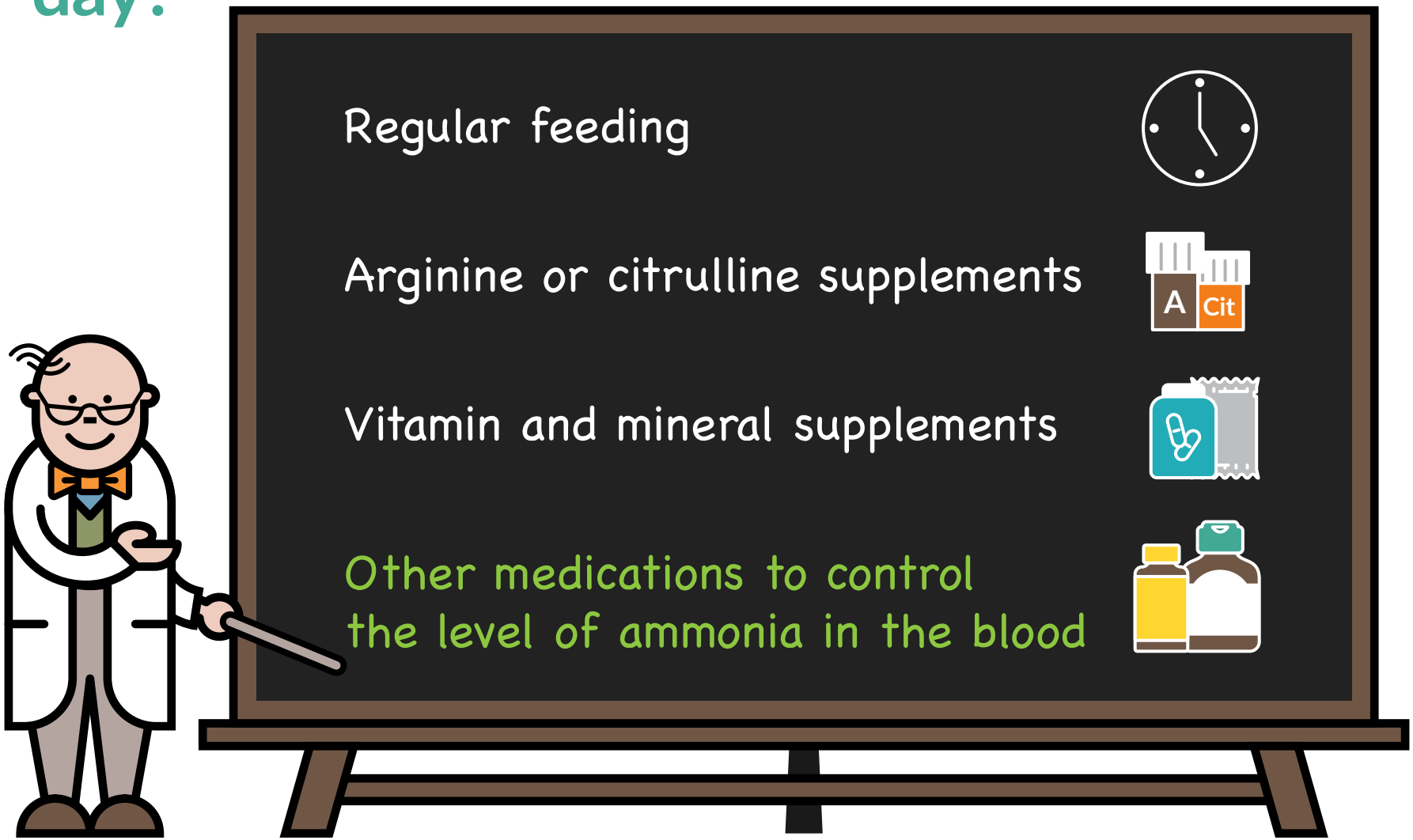
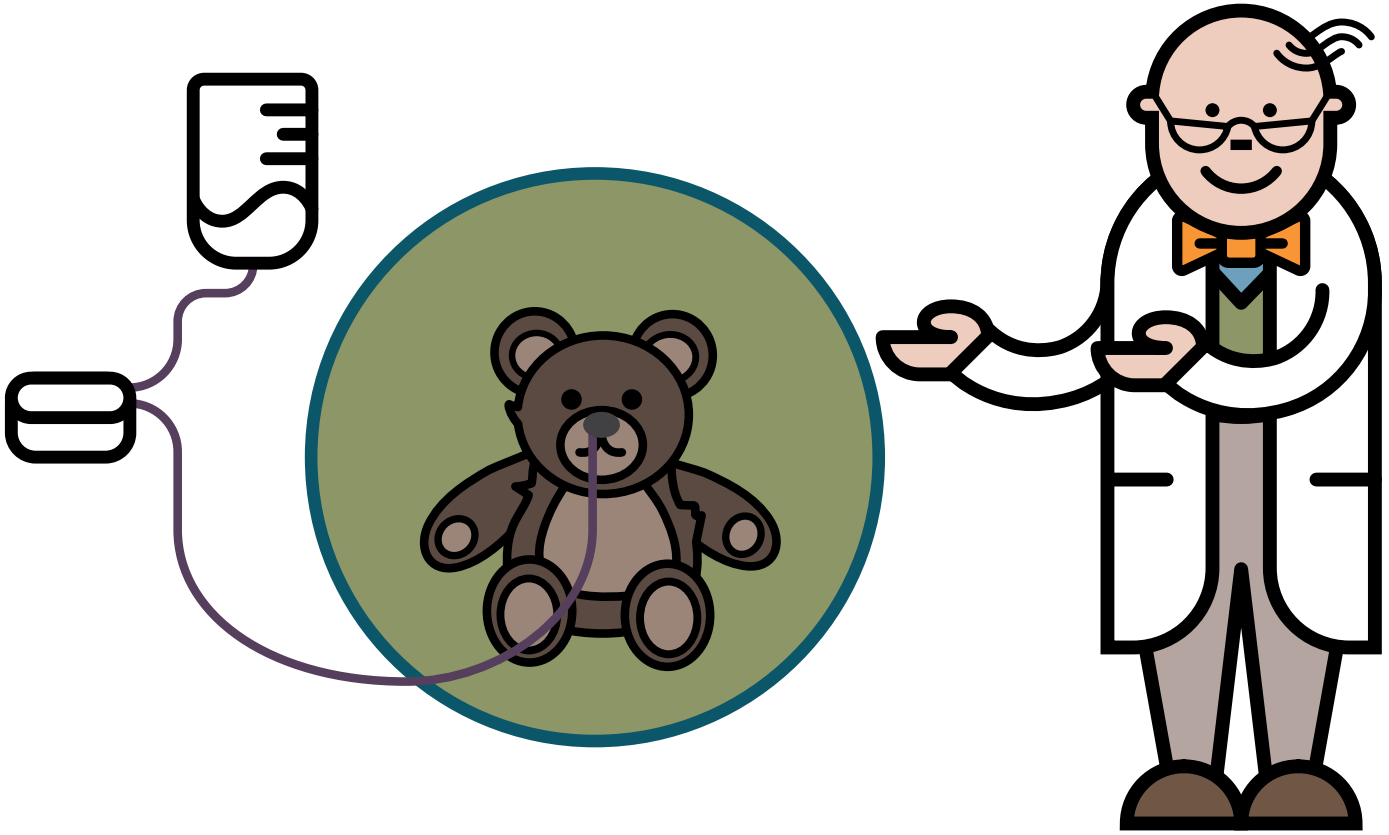


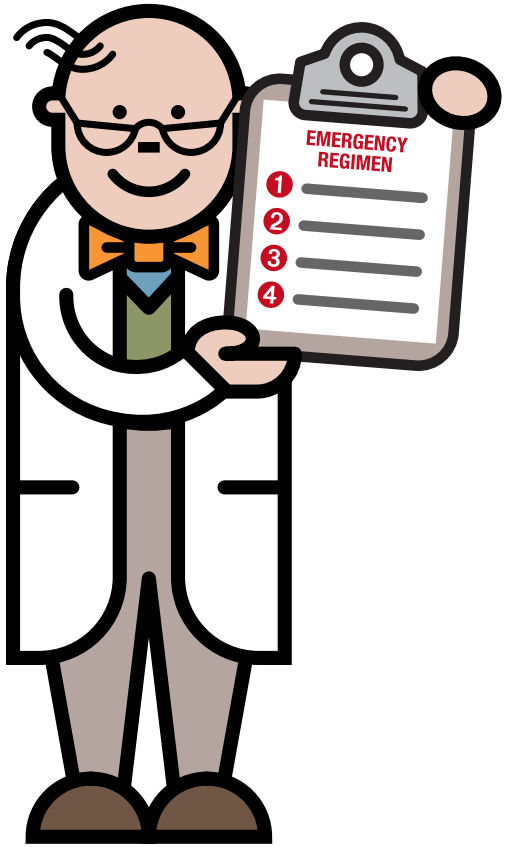
Is tube feeding needed?

Tube feeding may be necessary to give regular feeds. This will ensure energy, nutrient and fluid needs are met.



How is OTC deficiency managed during illness?

- During any childhood illness, an emergency regimen is given
- This will reduce the break down of protein and the build-up of ammonia



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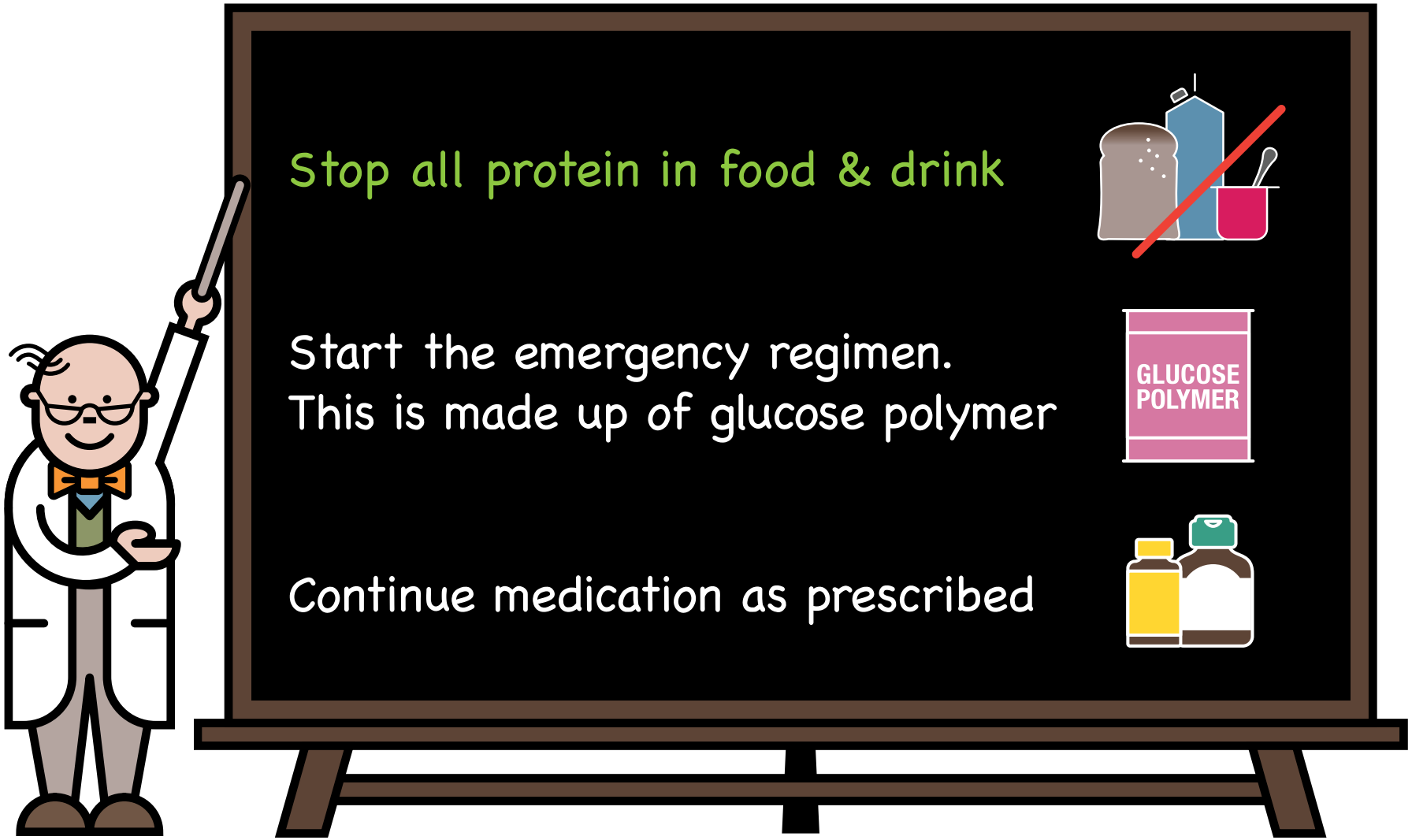

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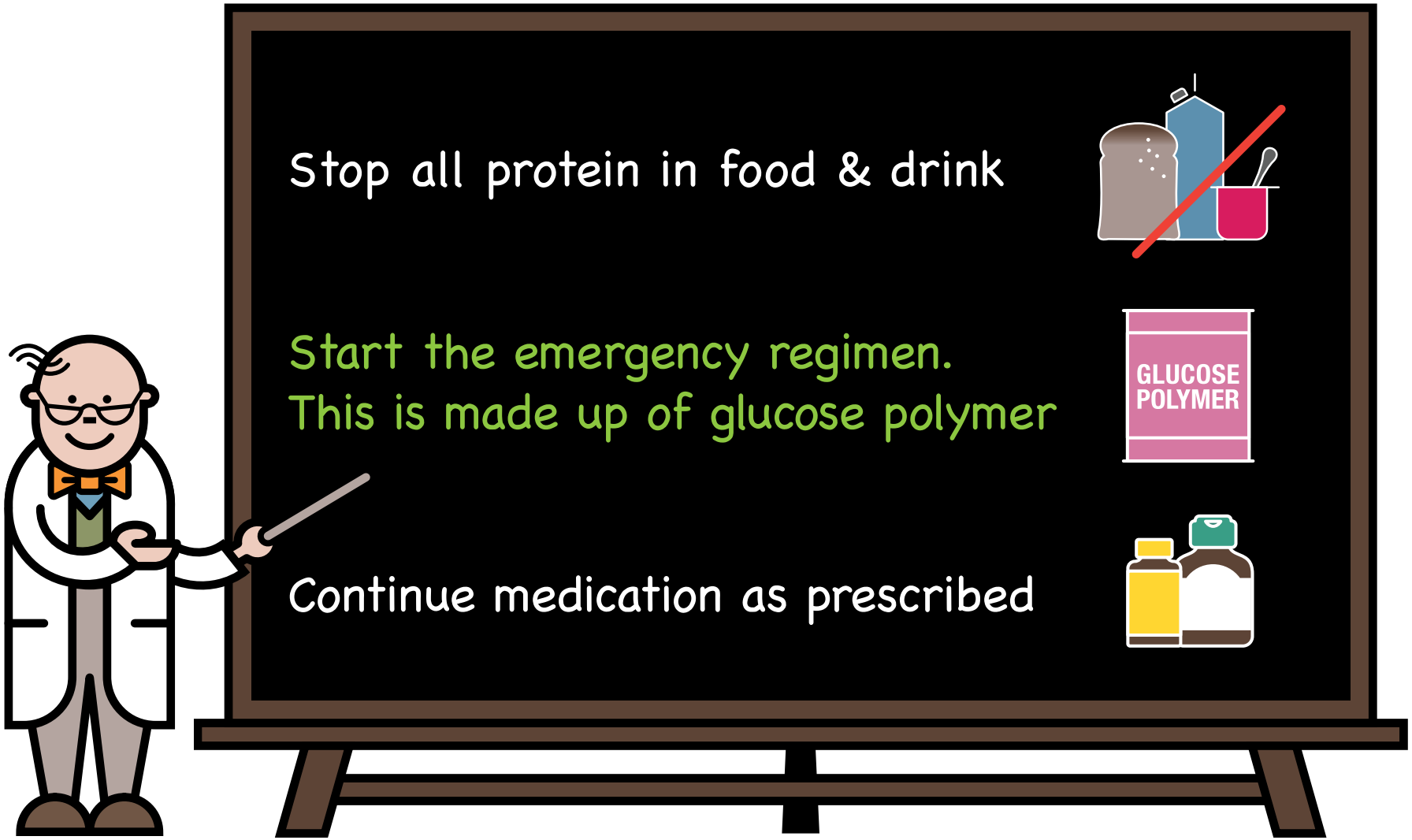

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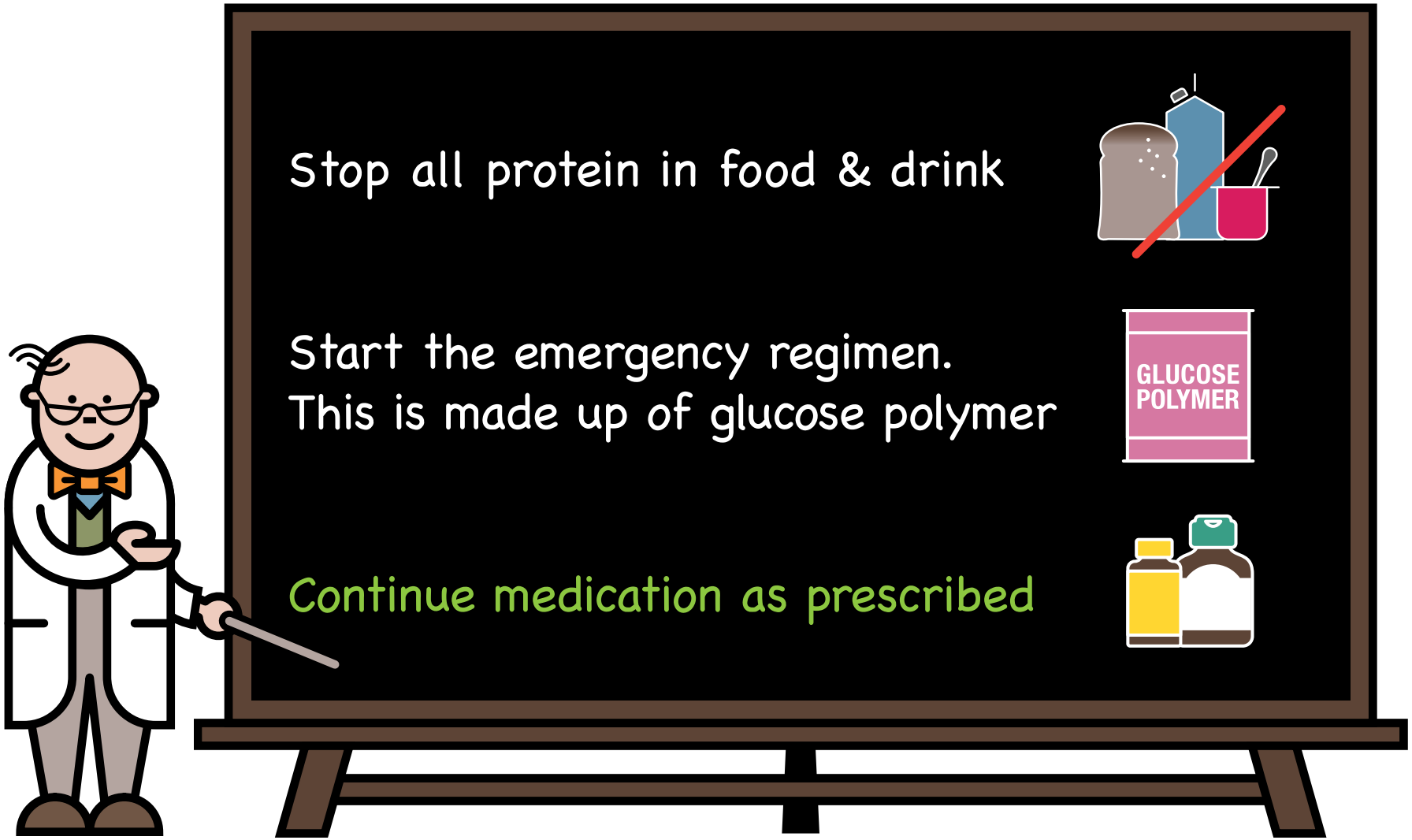
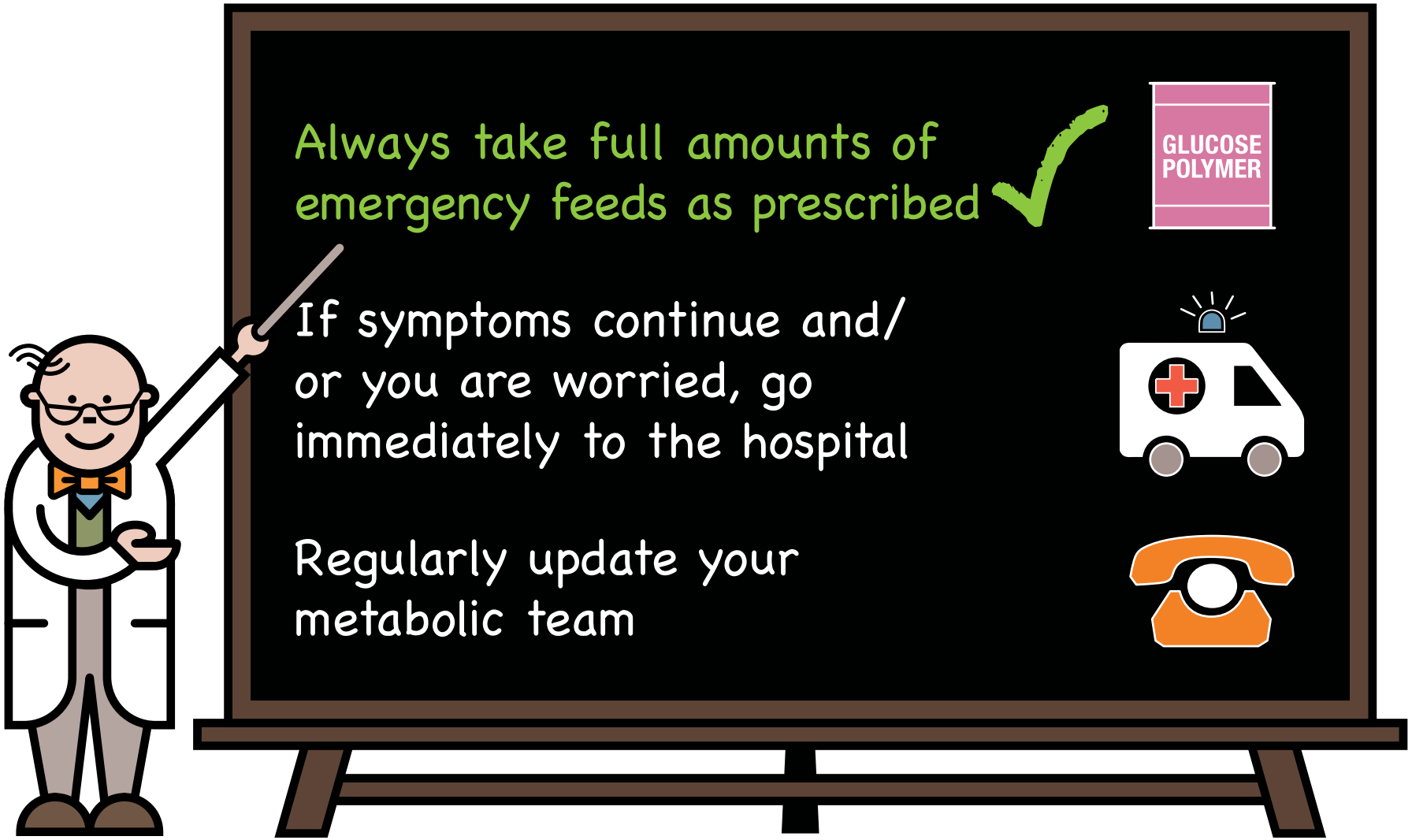


Checklist for illness



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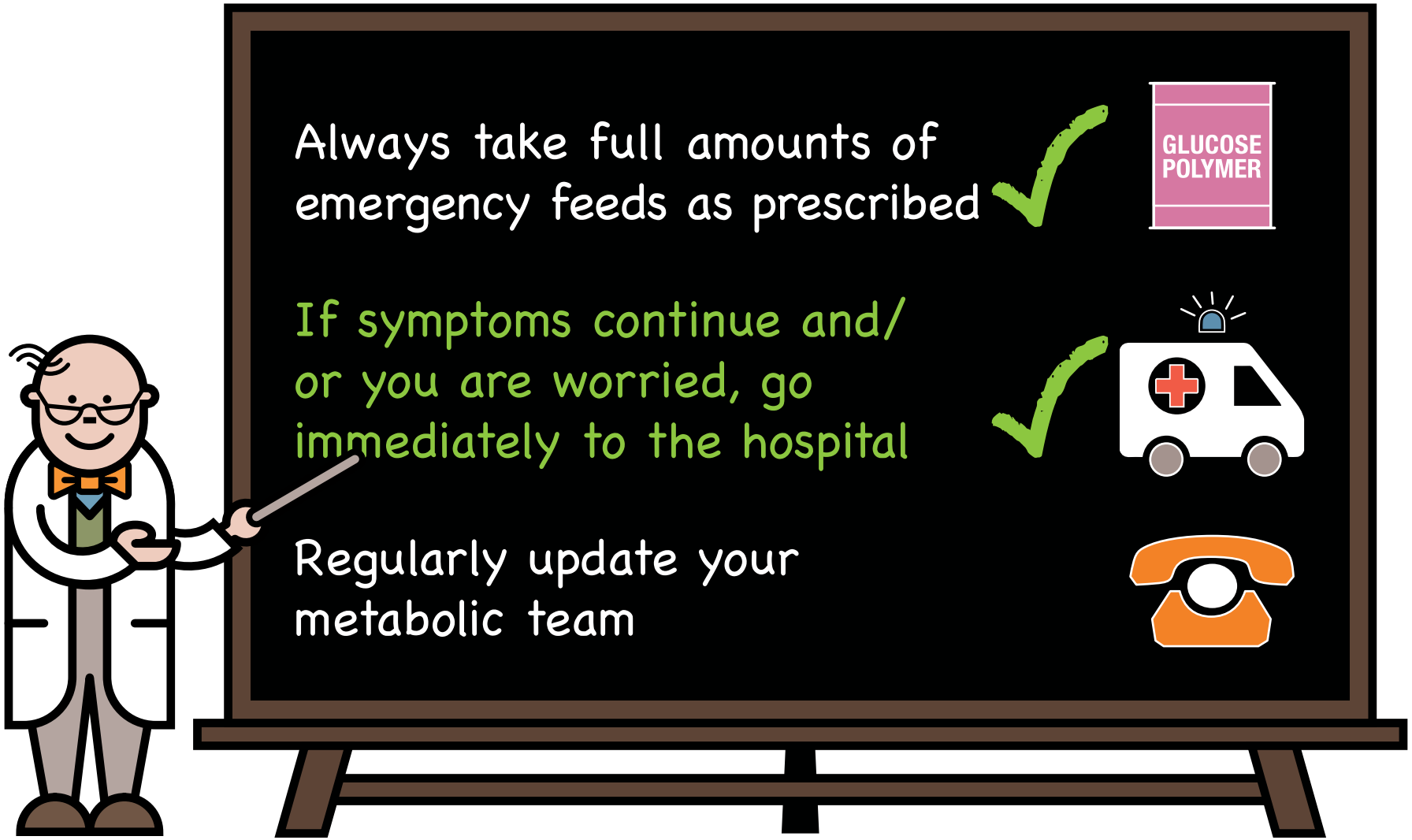

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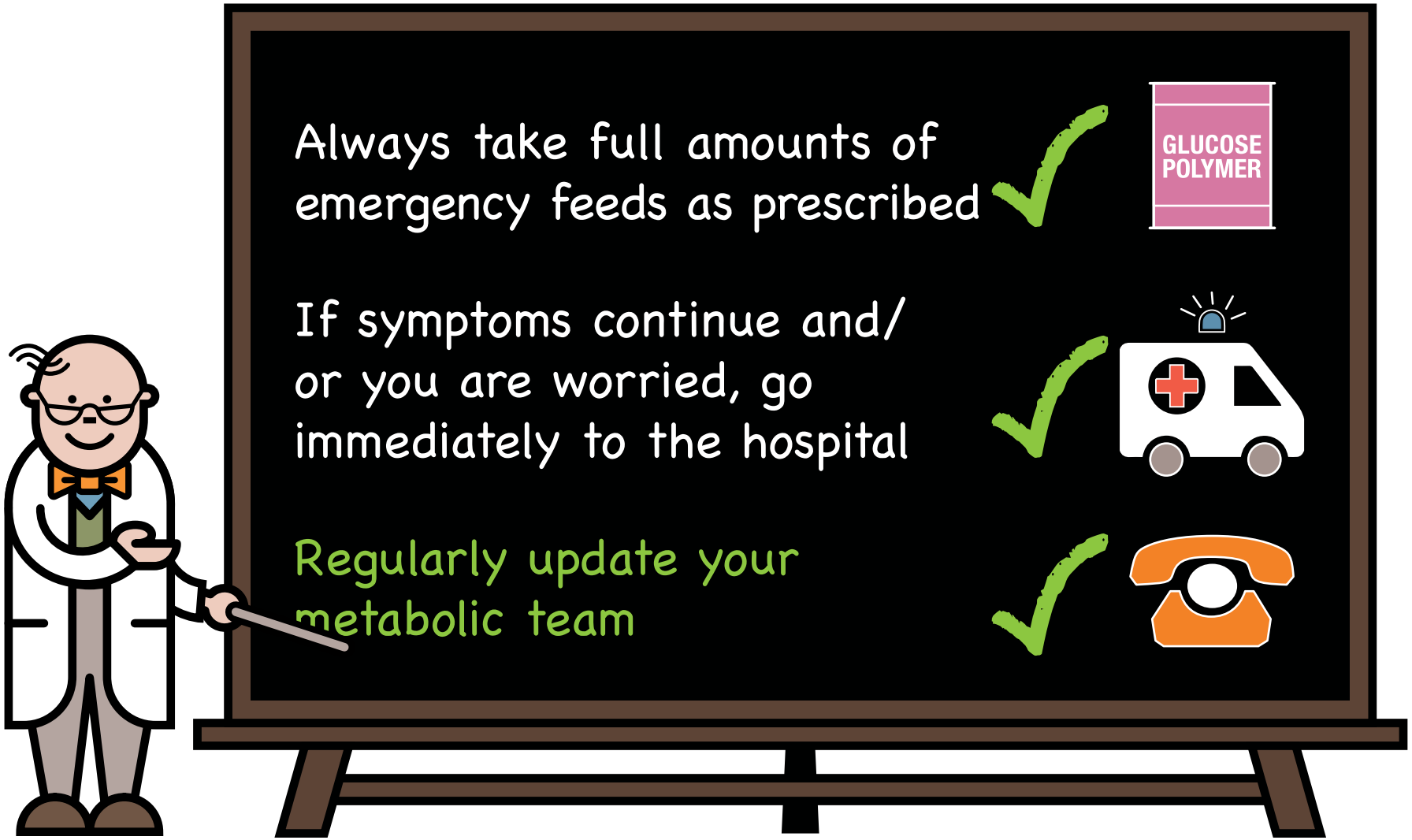
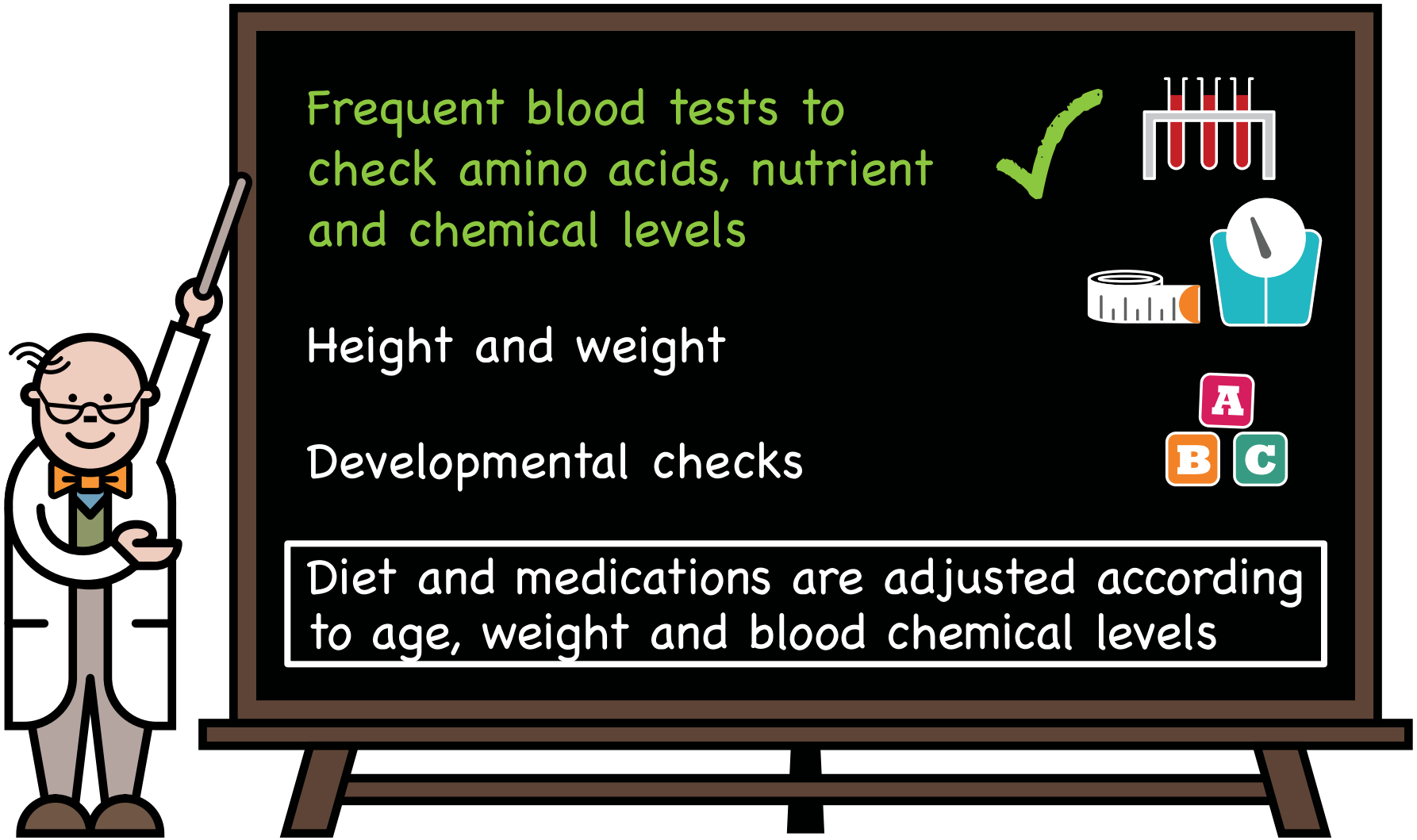


Key message

How is OTC deficiency monitored?



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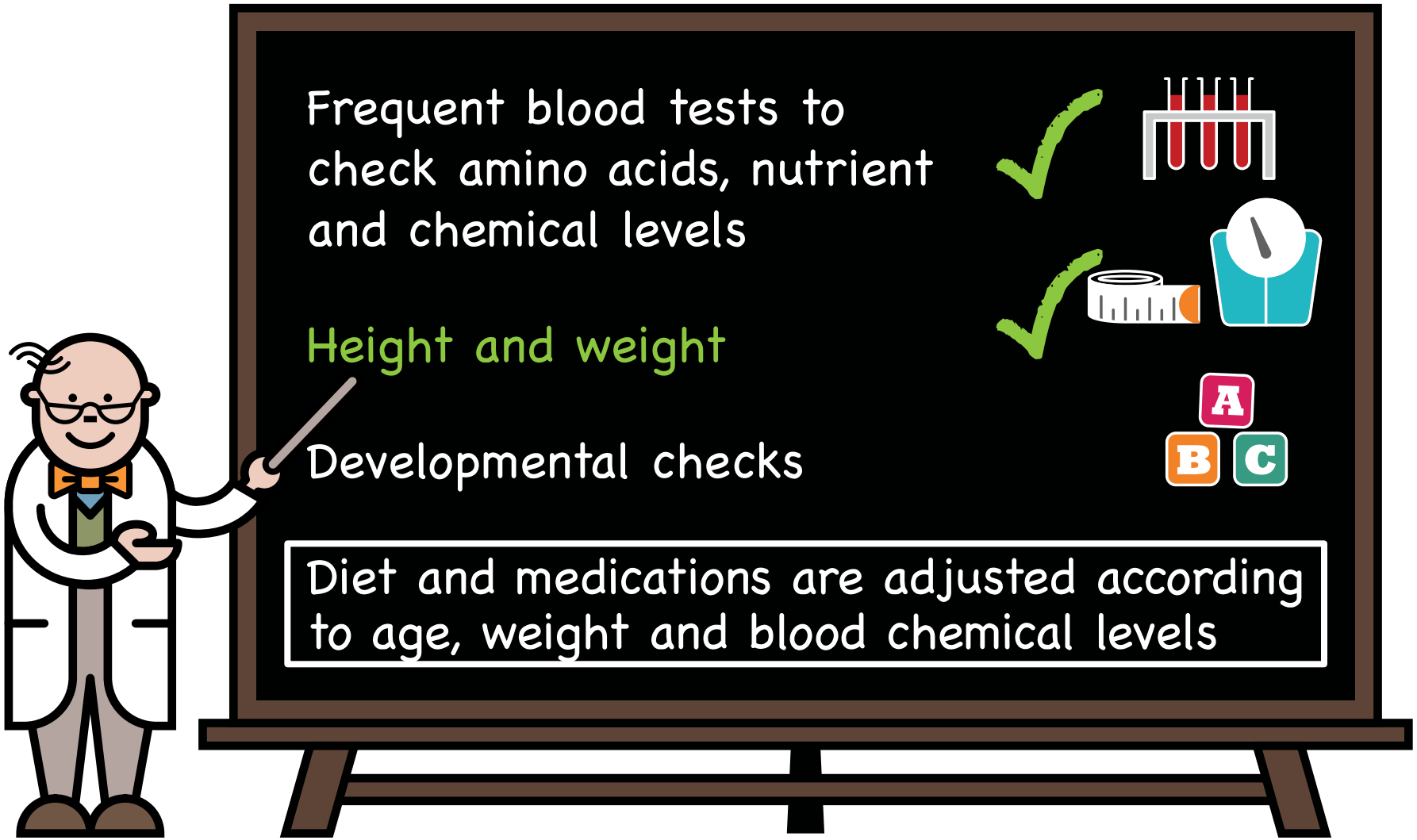

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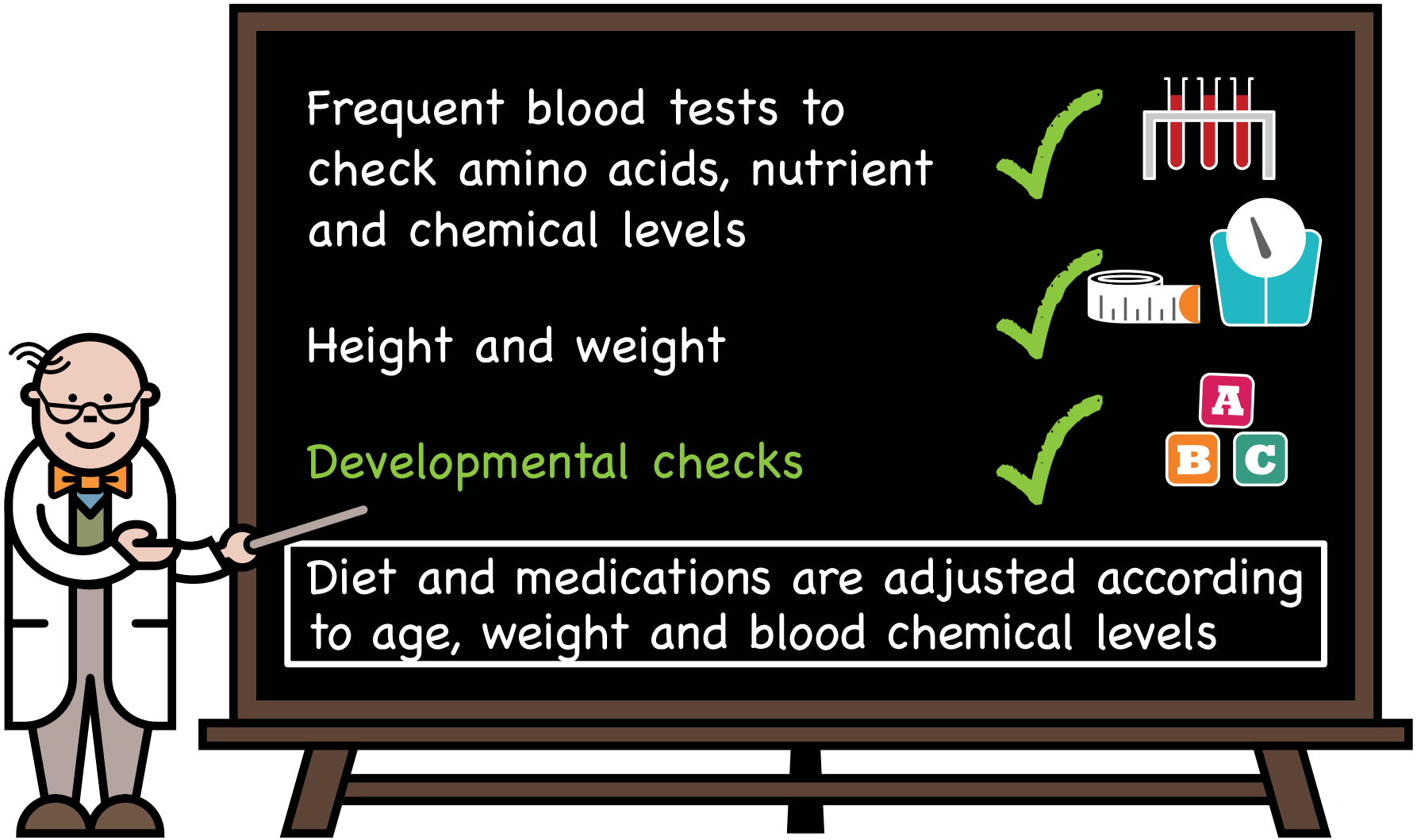
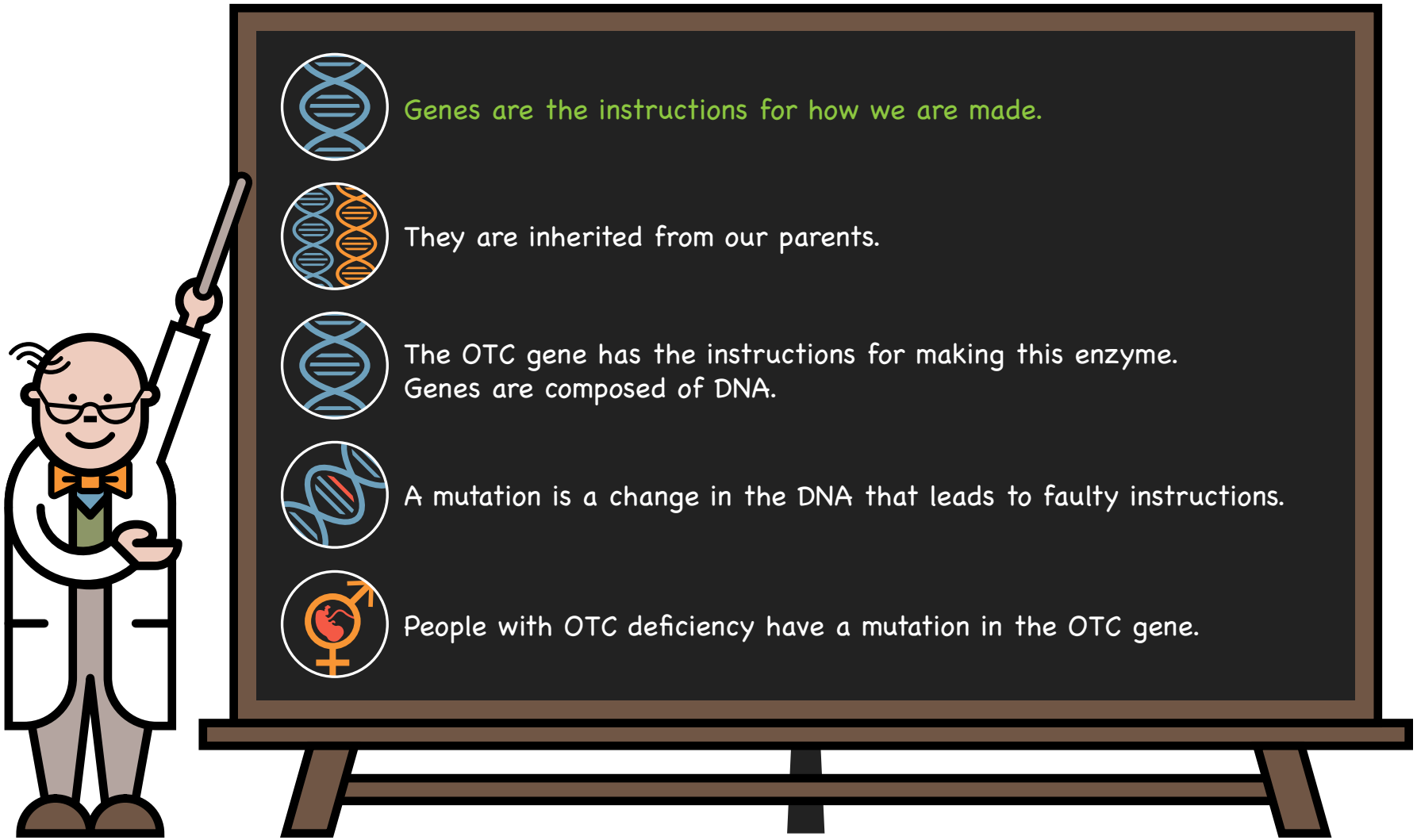


Chromosomes, genes, mutations



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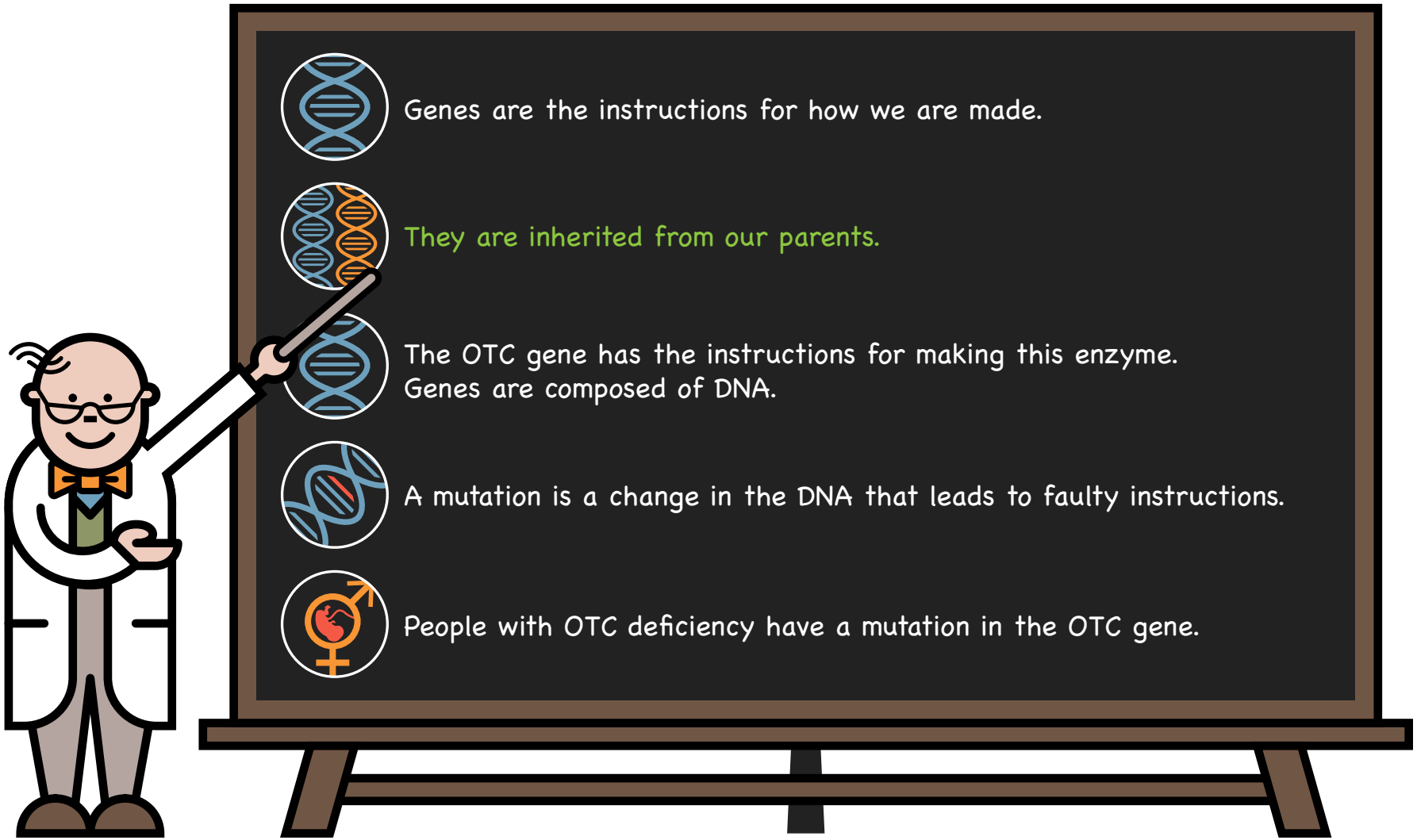

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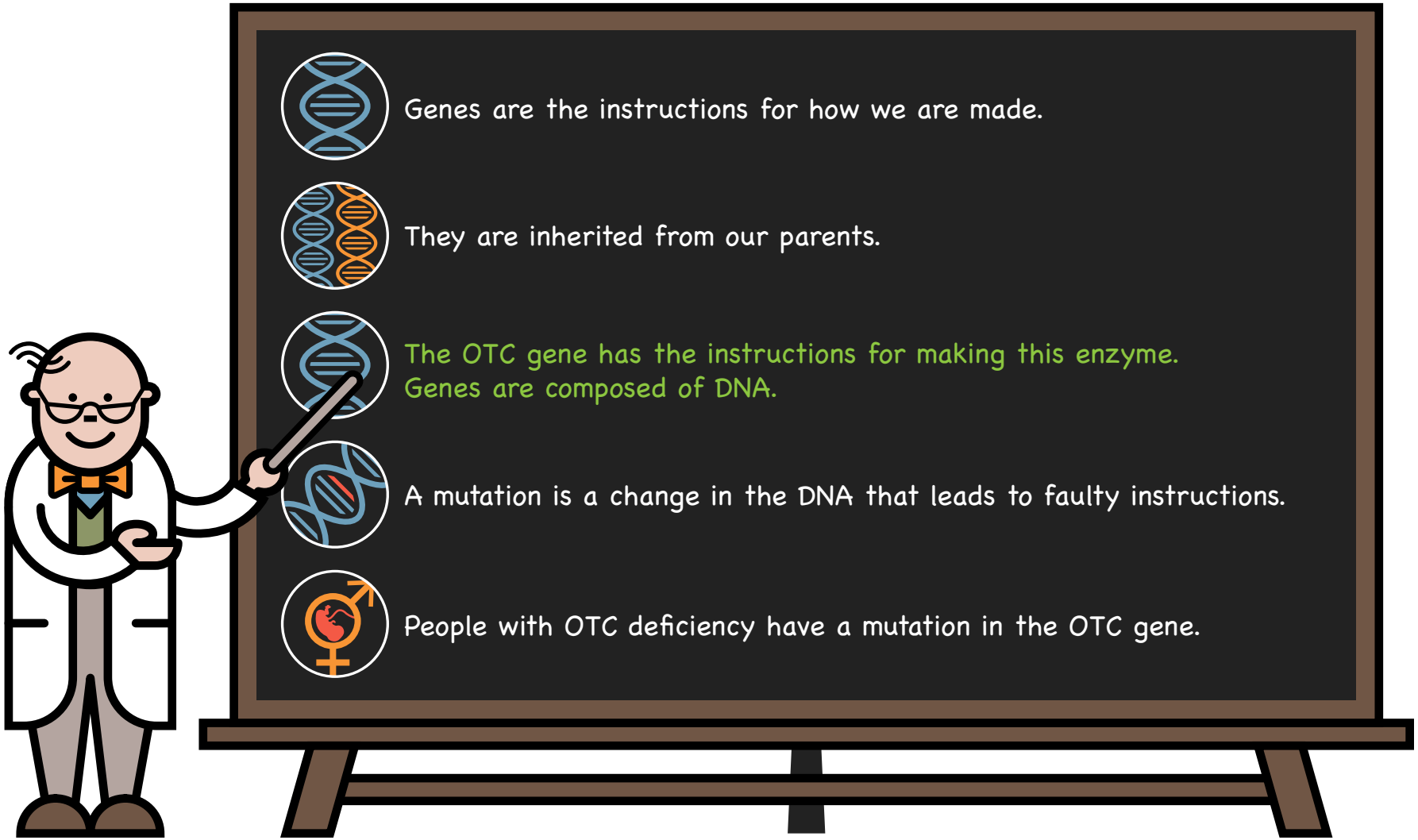

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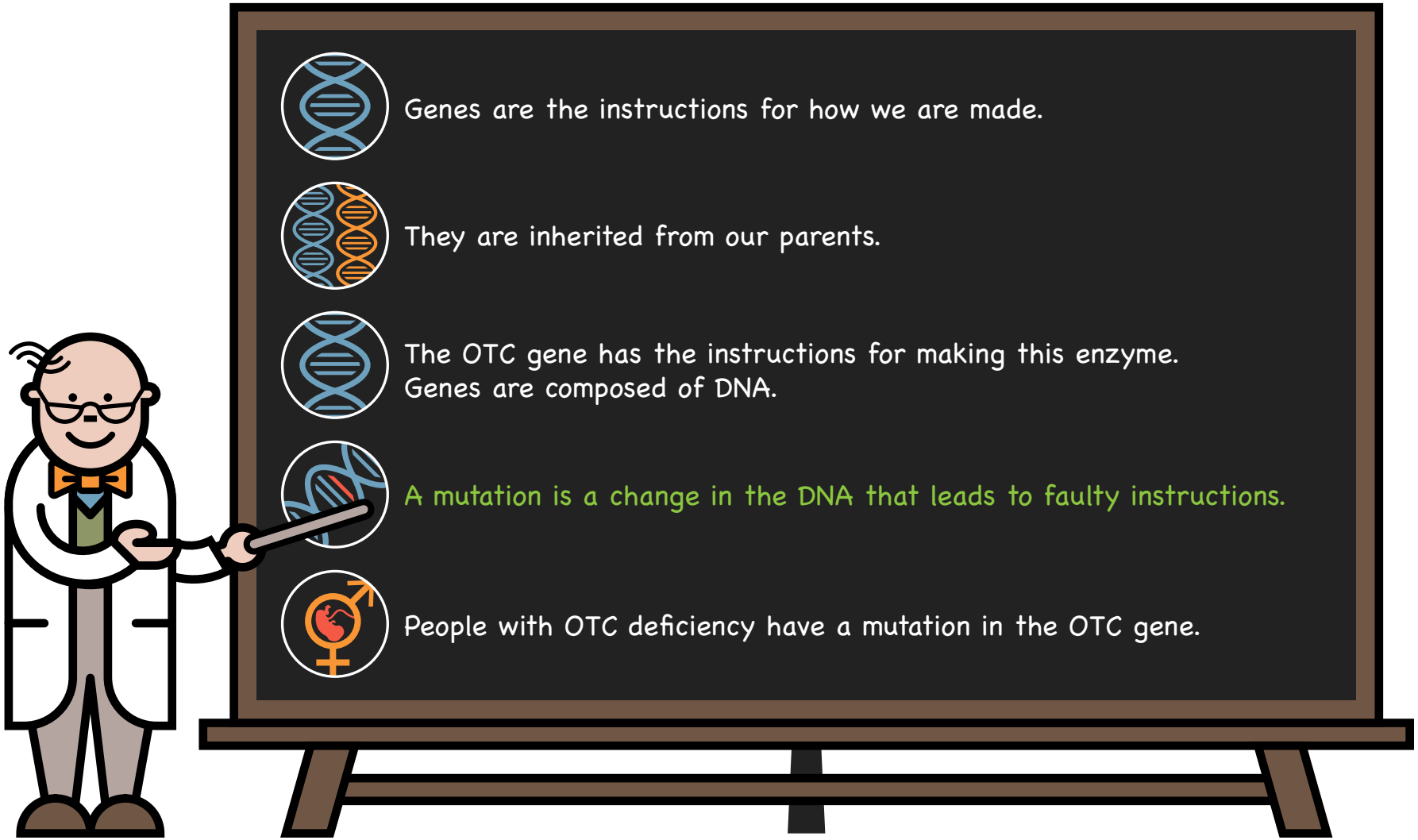

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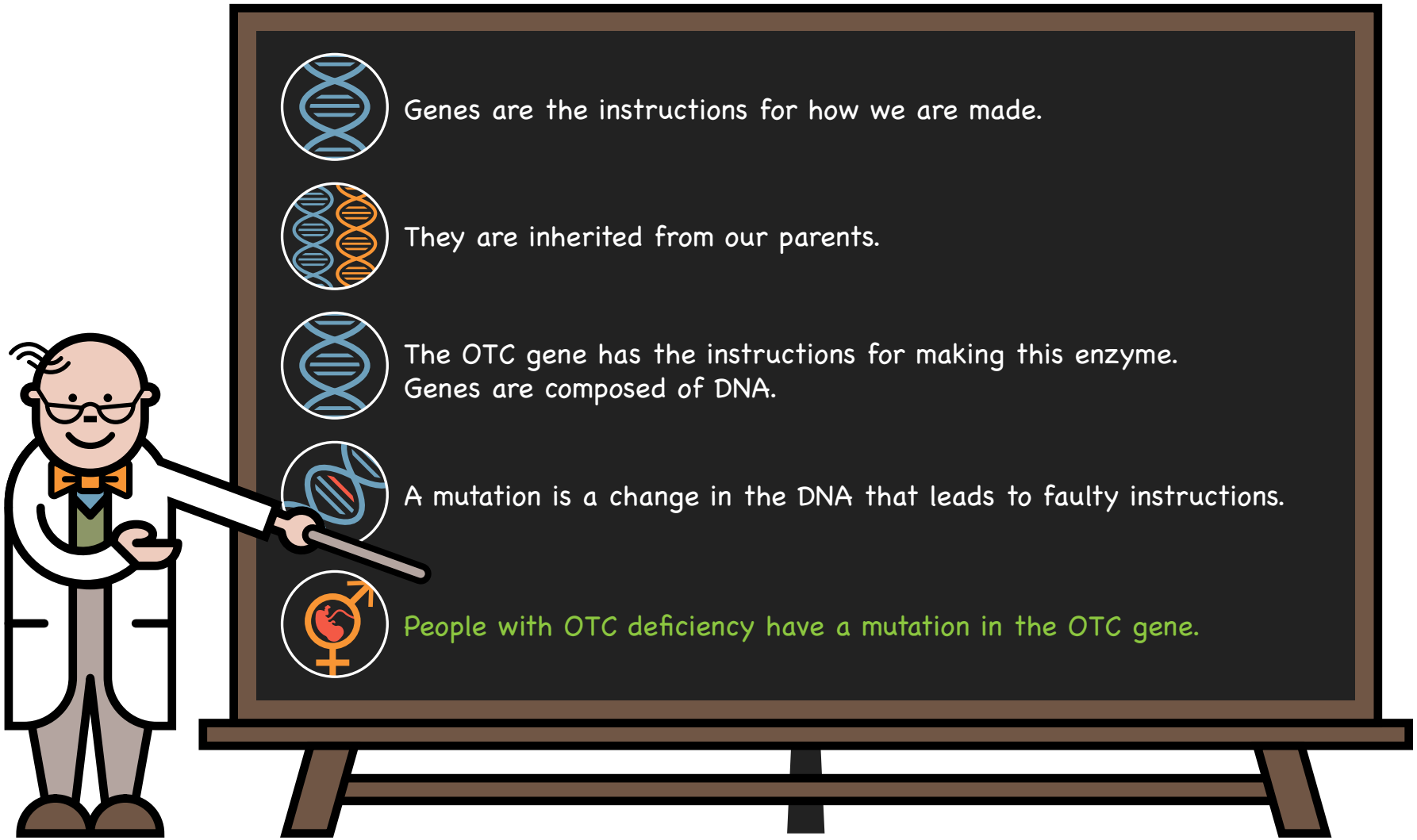
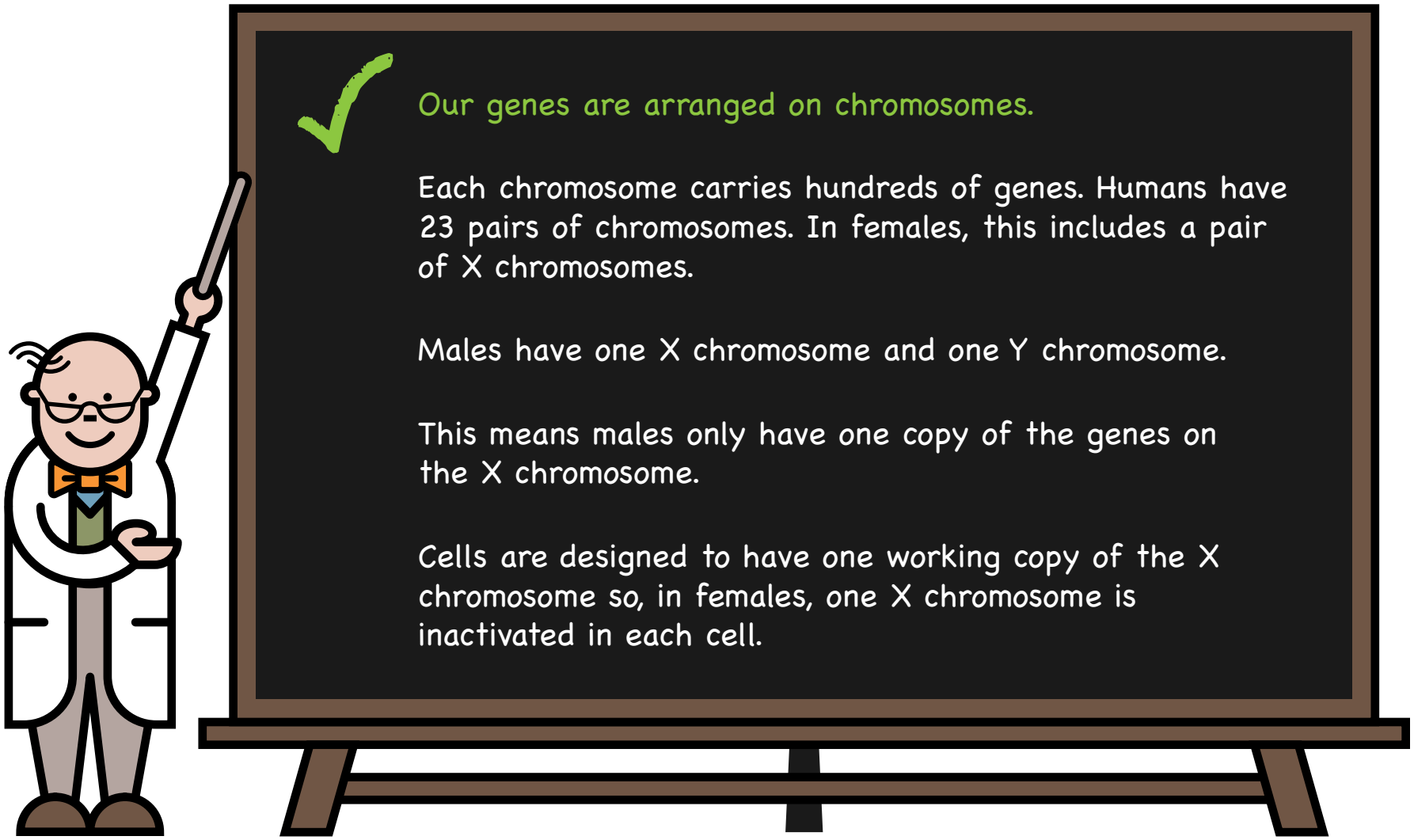


Inheritance



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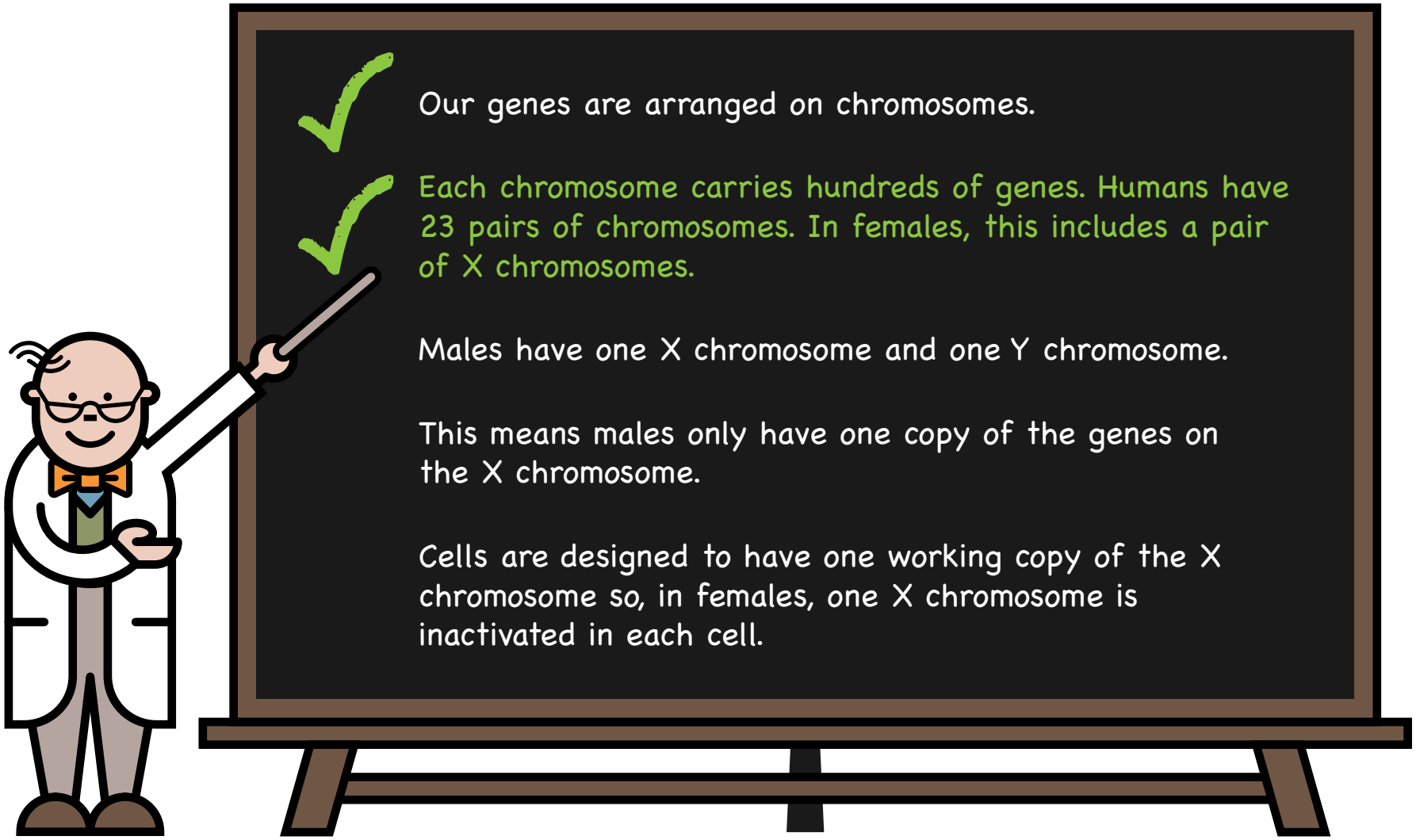

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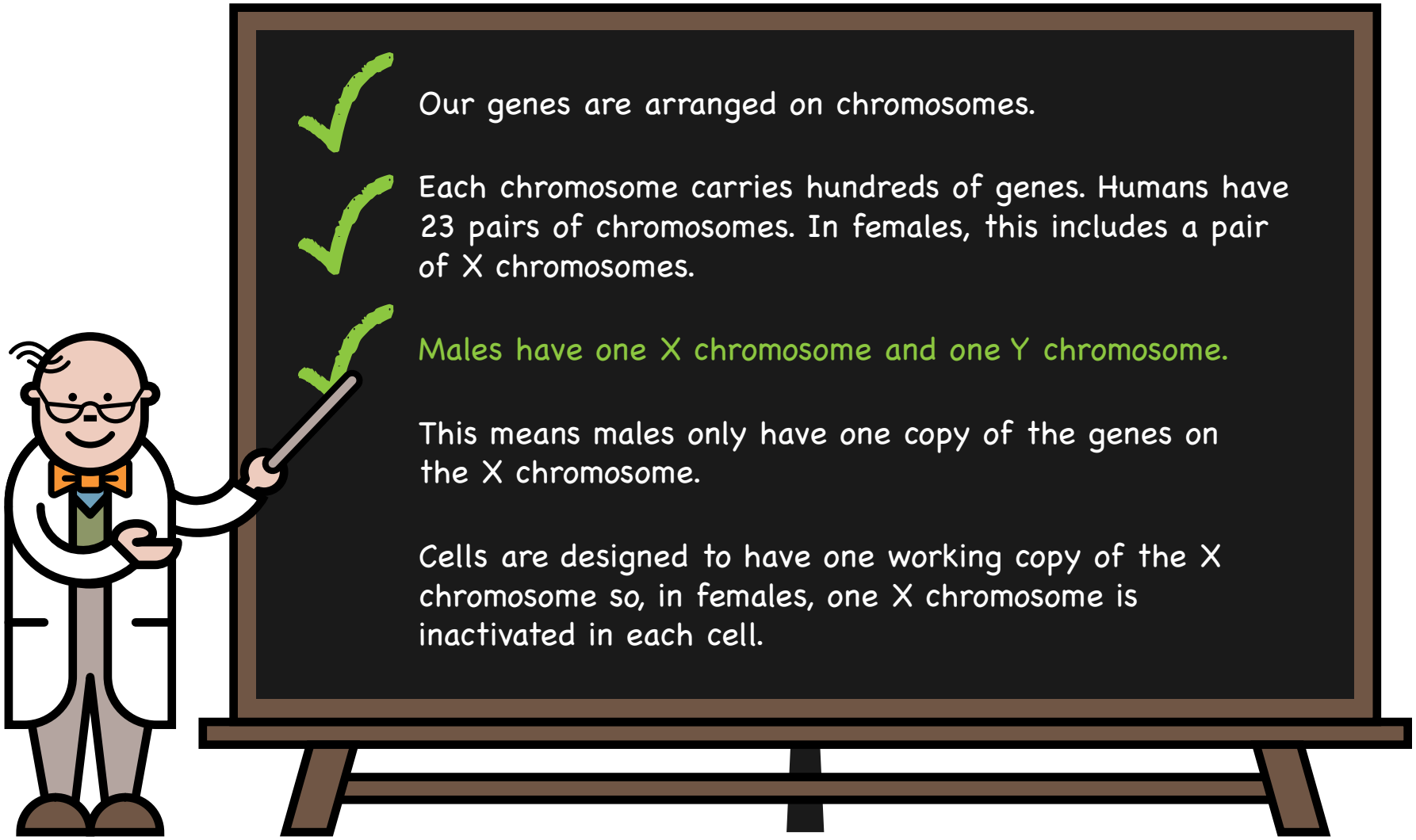

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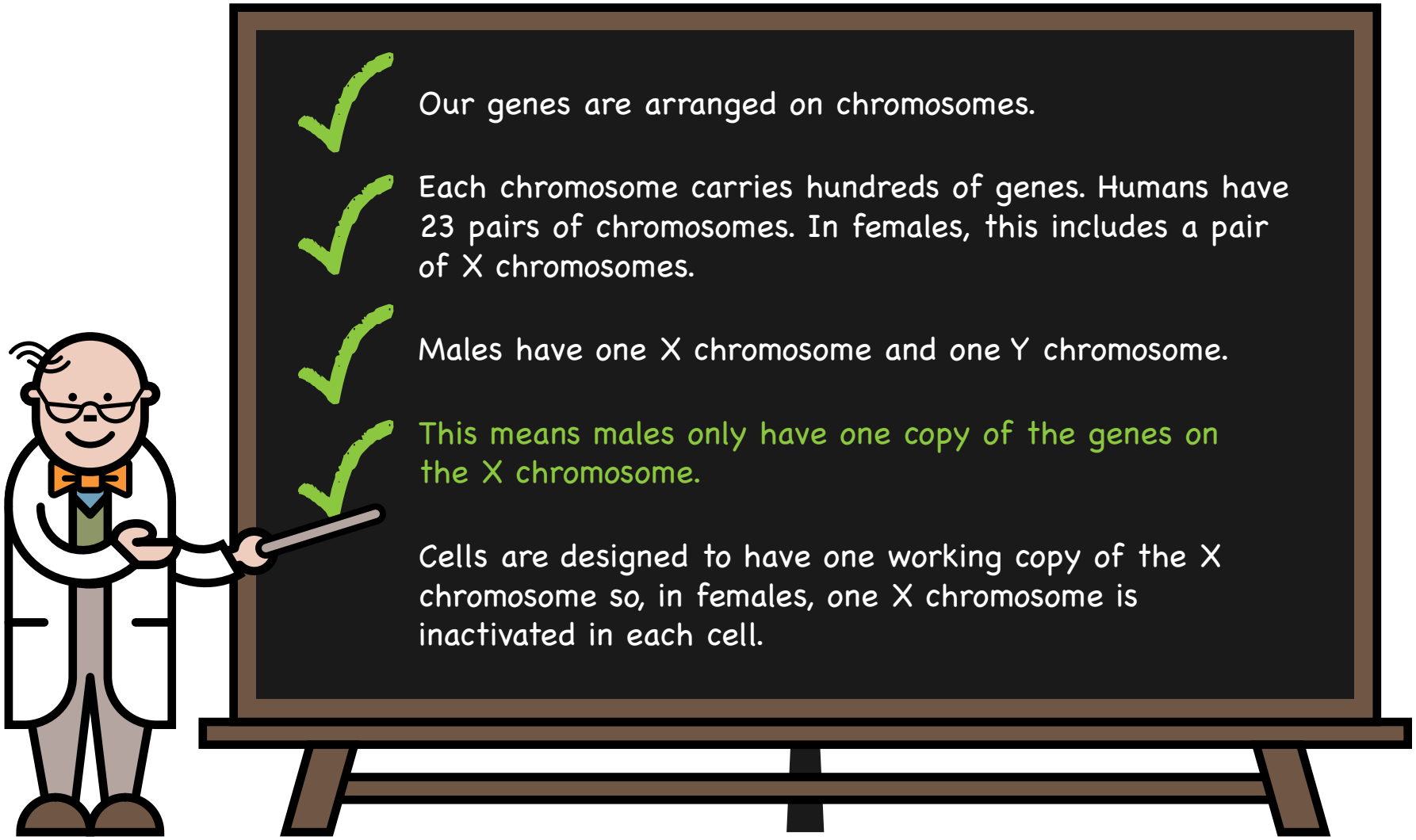

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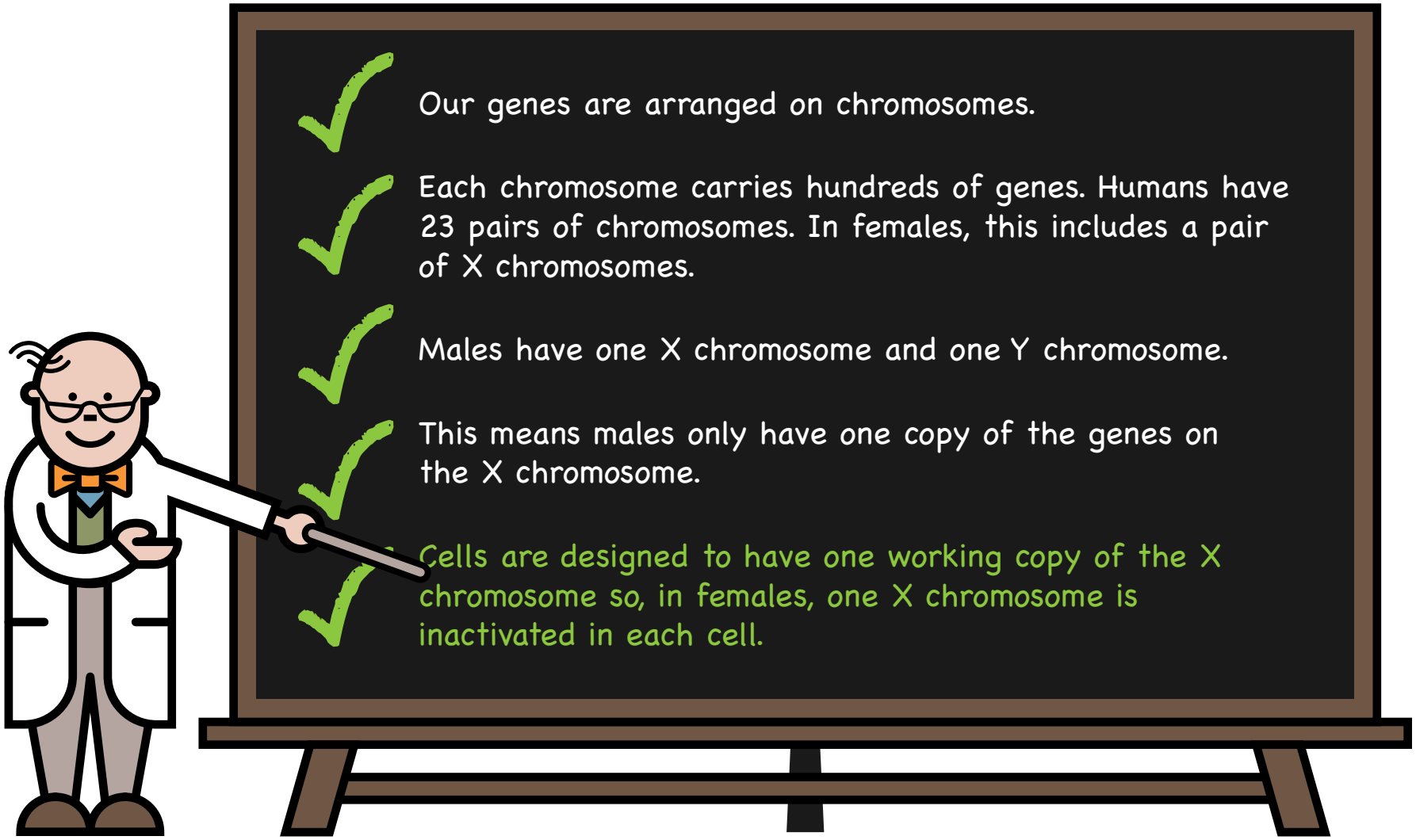

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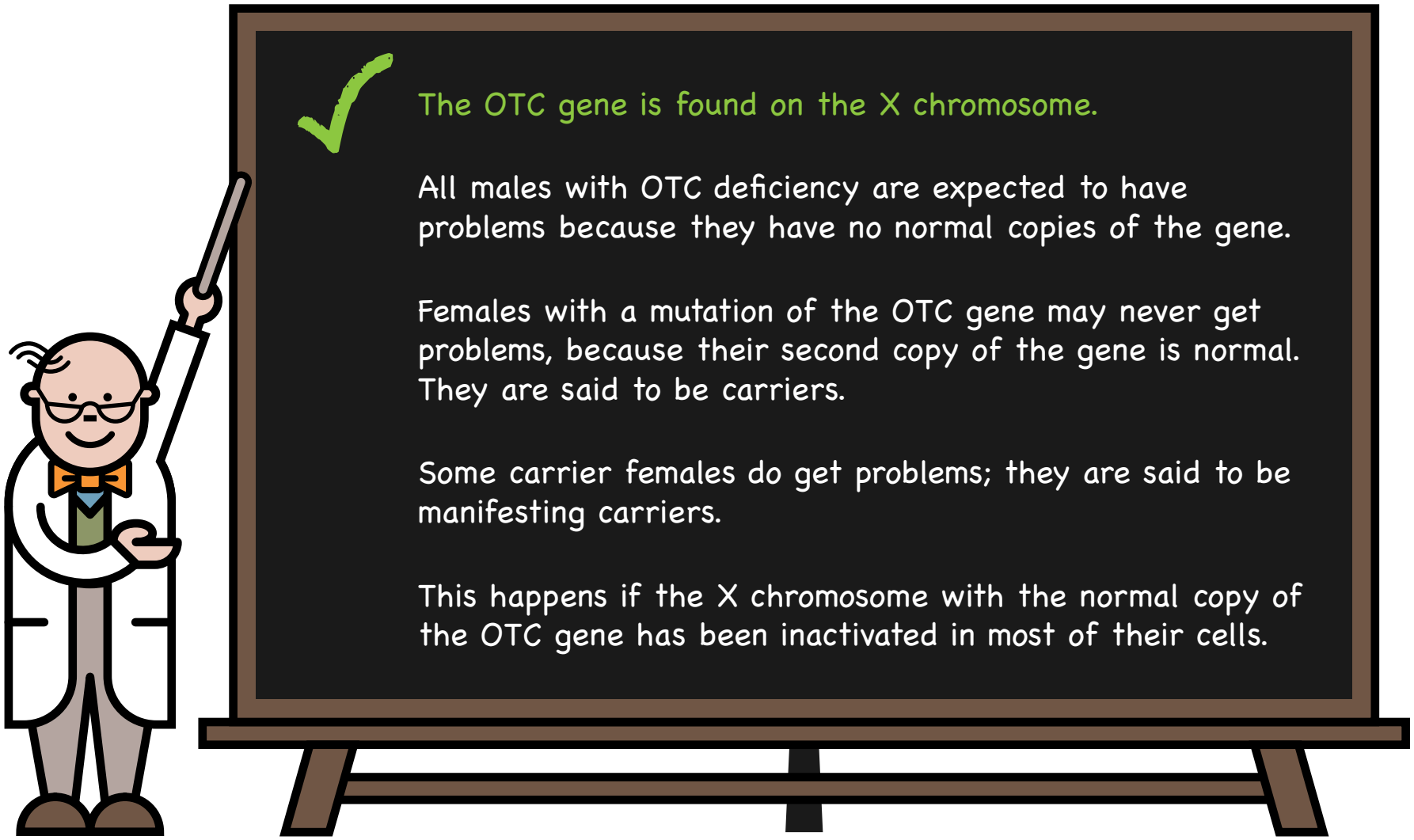

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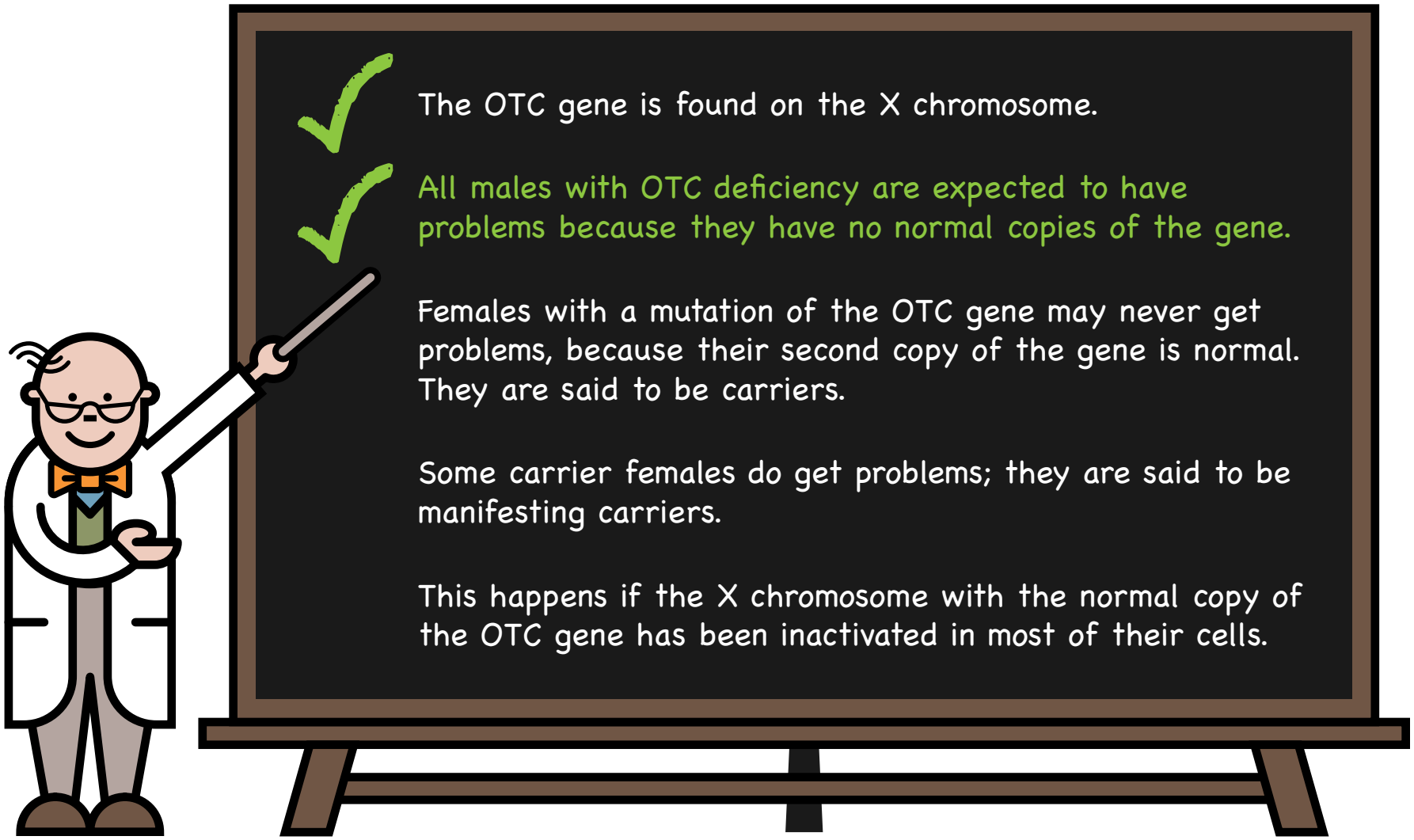

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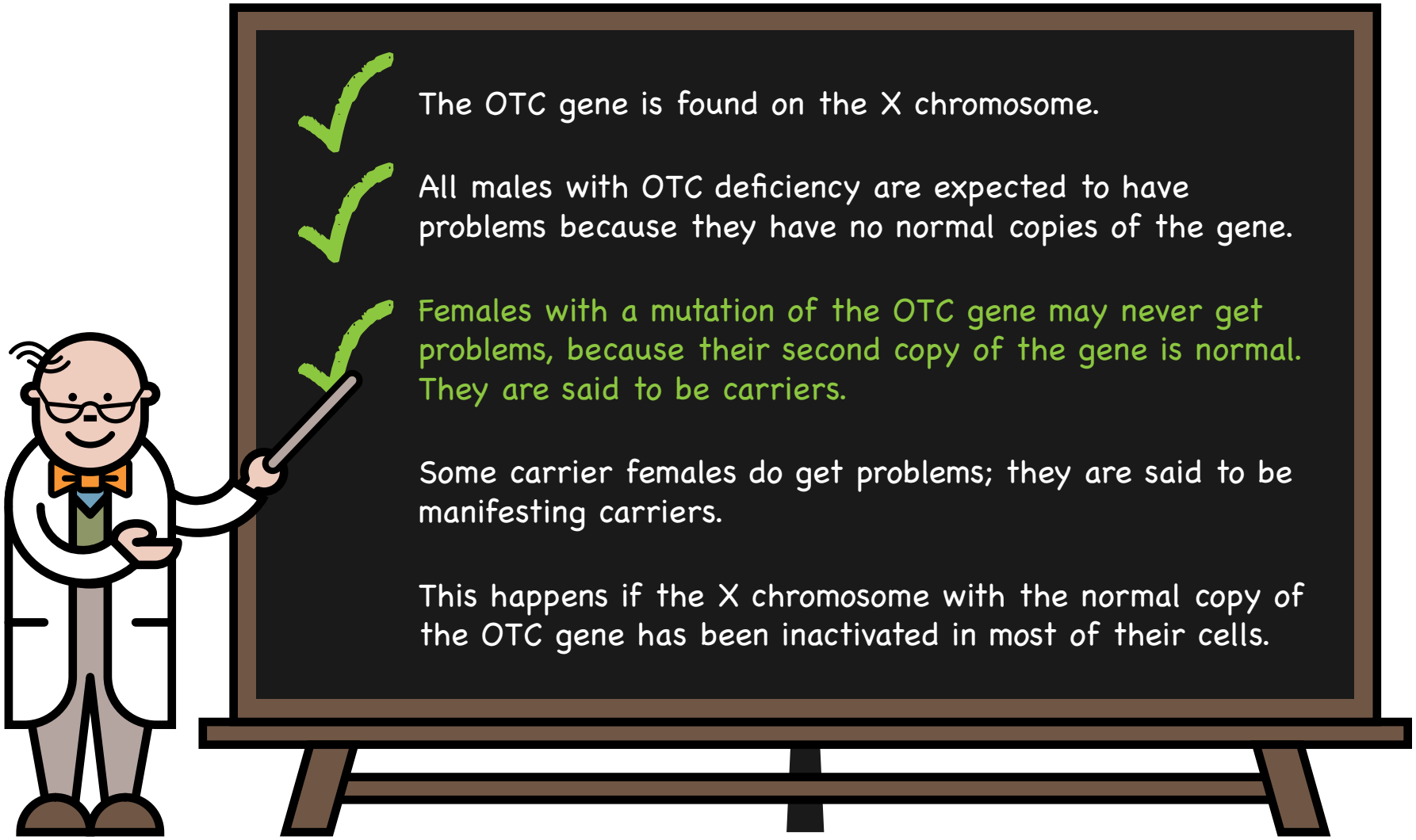

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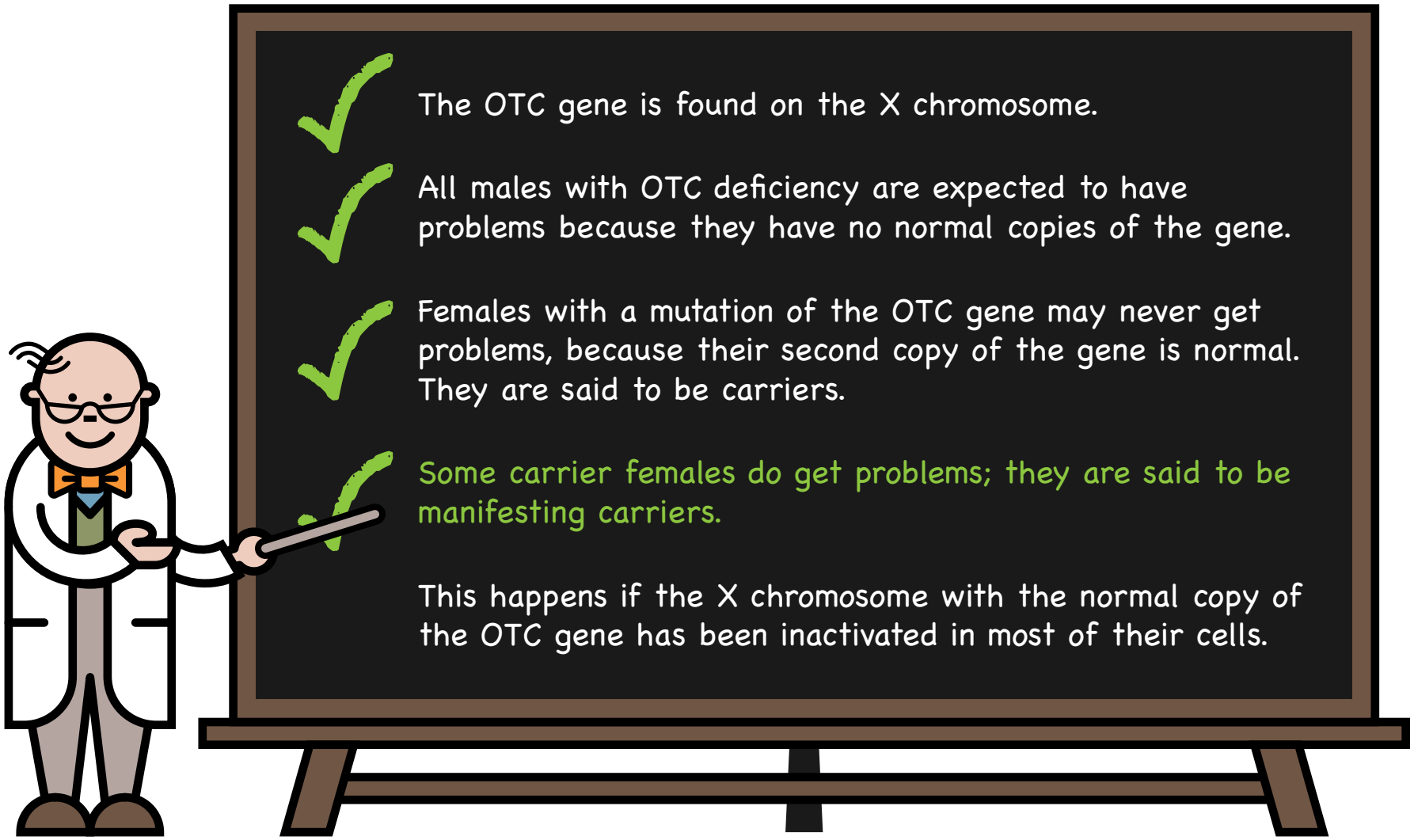

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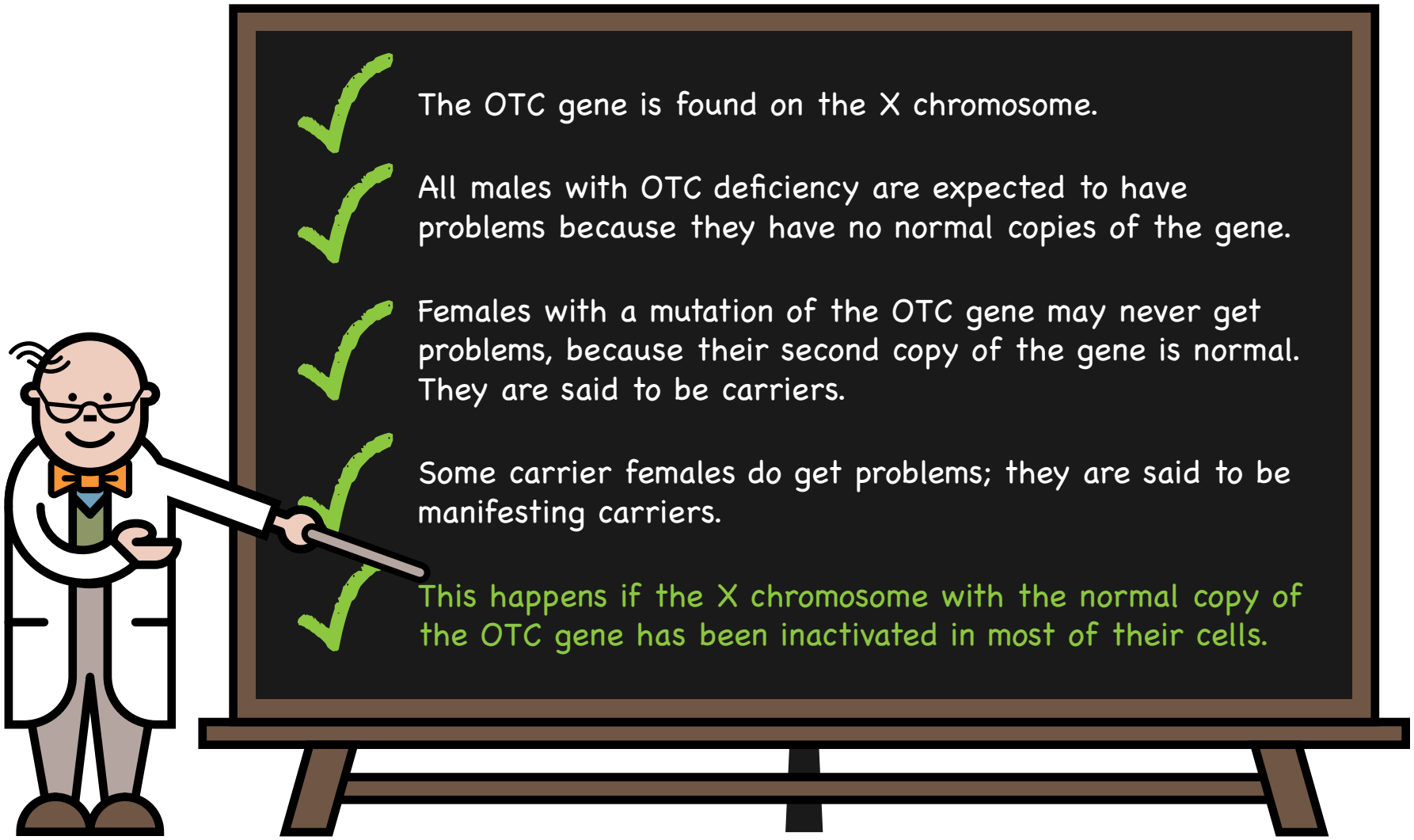
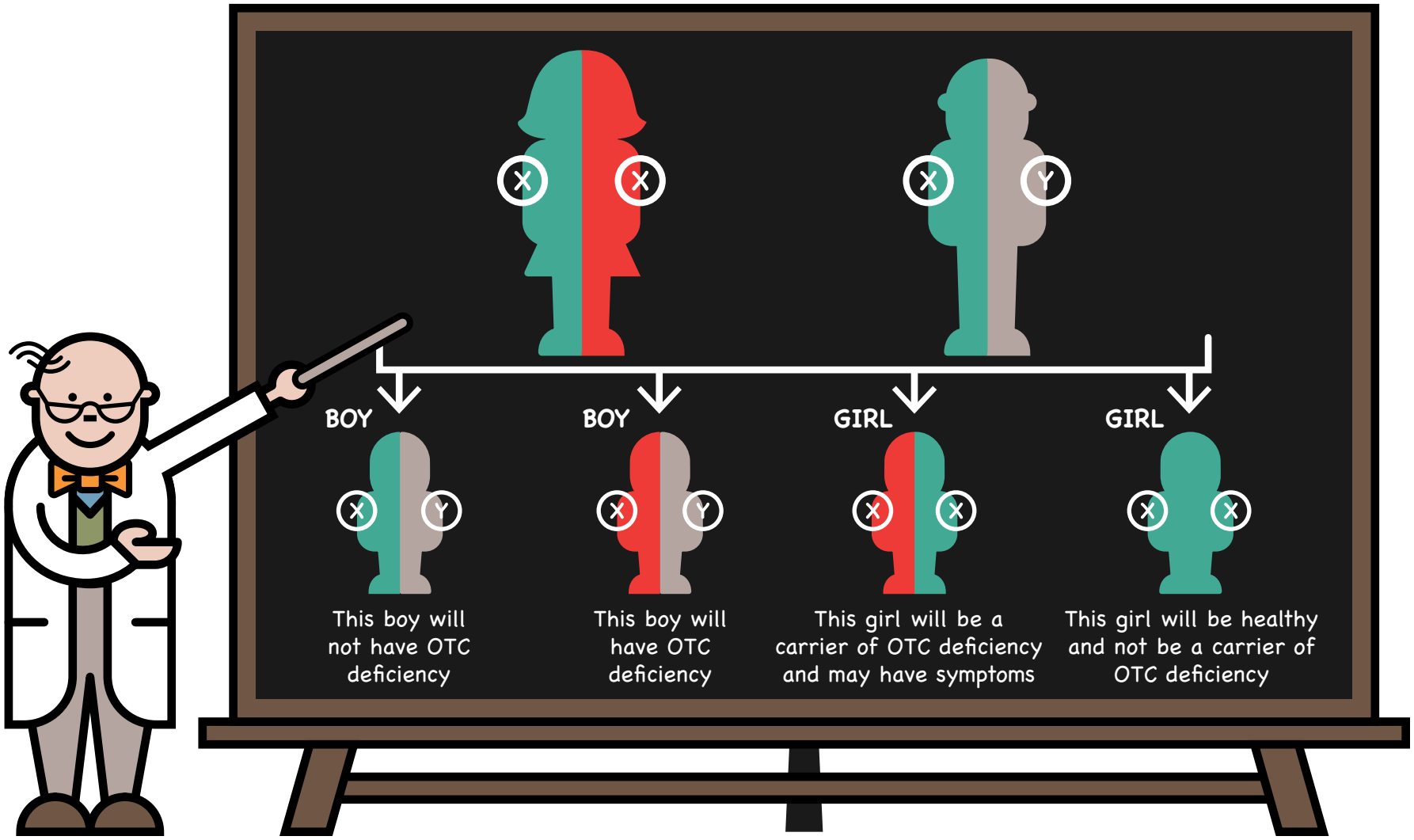
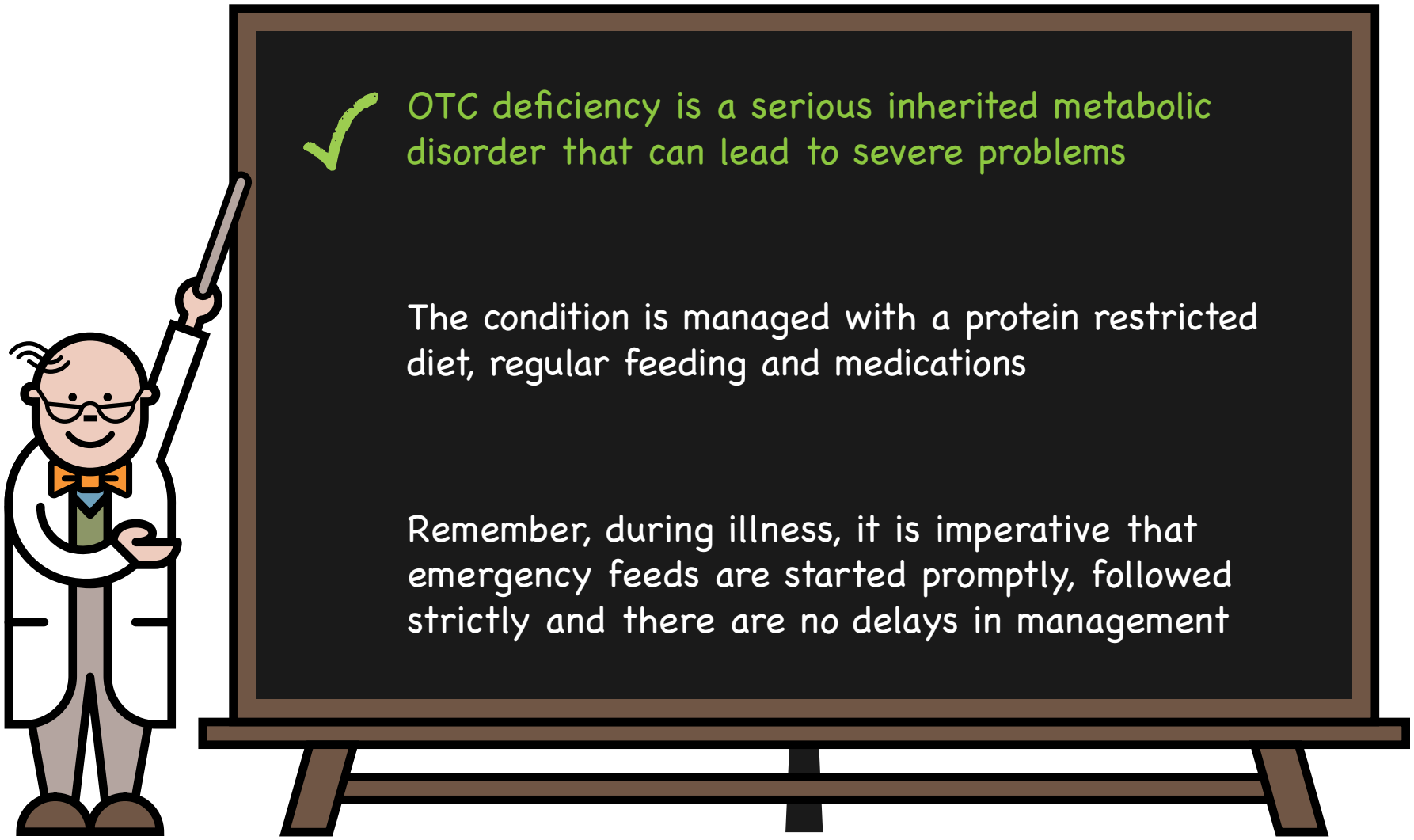


Inheritance —Future pregnancies: possible outcomes if the mother is a carrier



Take home messages



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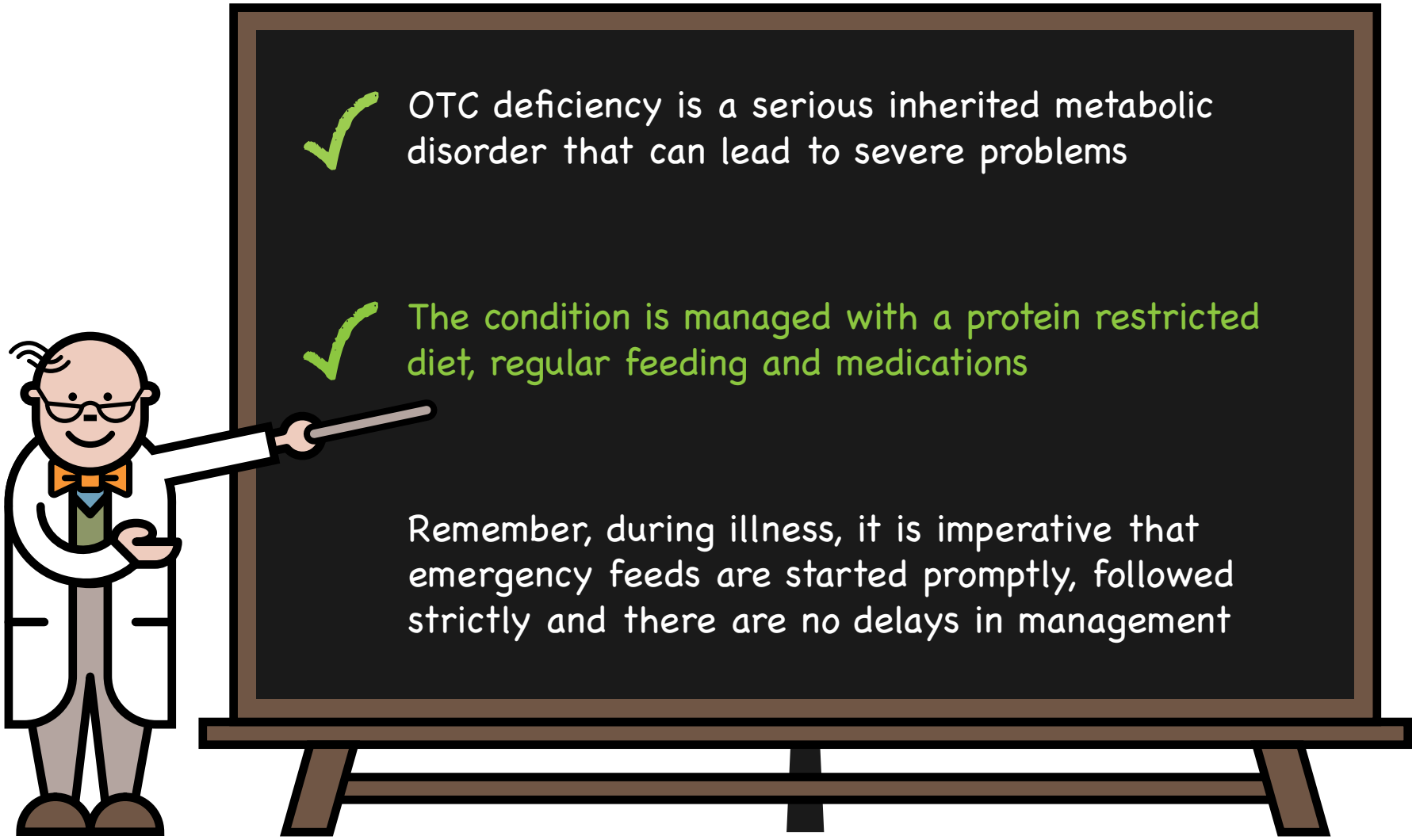

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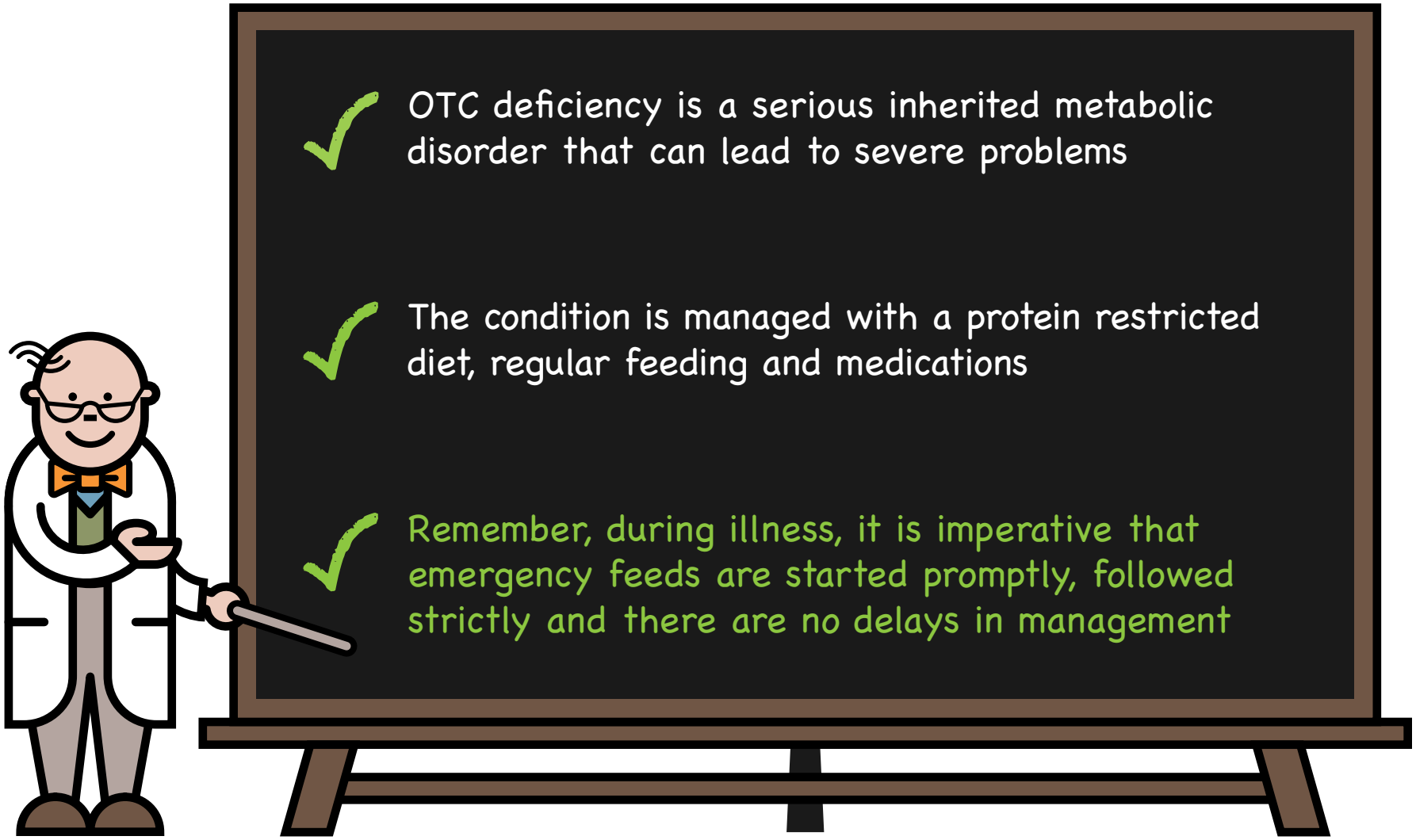

Helpful hints

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Who's who

- My dietitians
- My nurses
- My doctors
  - Contact details, address, photos

Visit **www.nutricia.co.uk/patients-carers/living-with/low-protein-diet.html** 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.

Low Protein Connect

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LowProteinConnect
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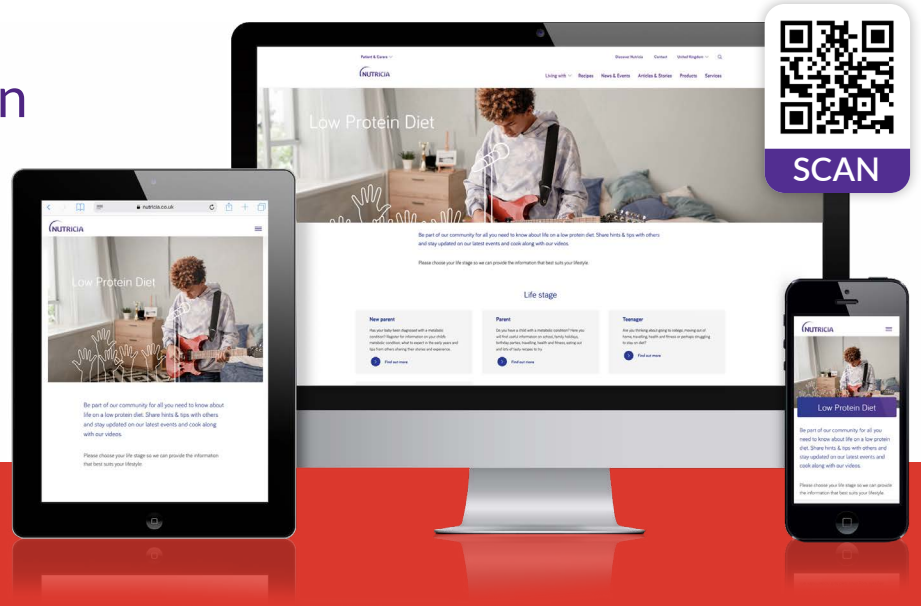

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