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 1, DECEMBER 2019

CPT 1 DEFICIENCY

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.

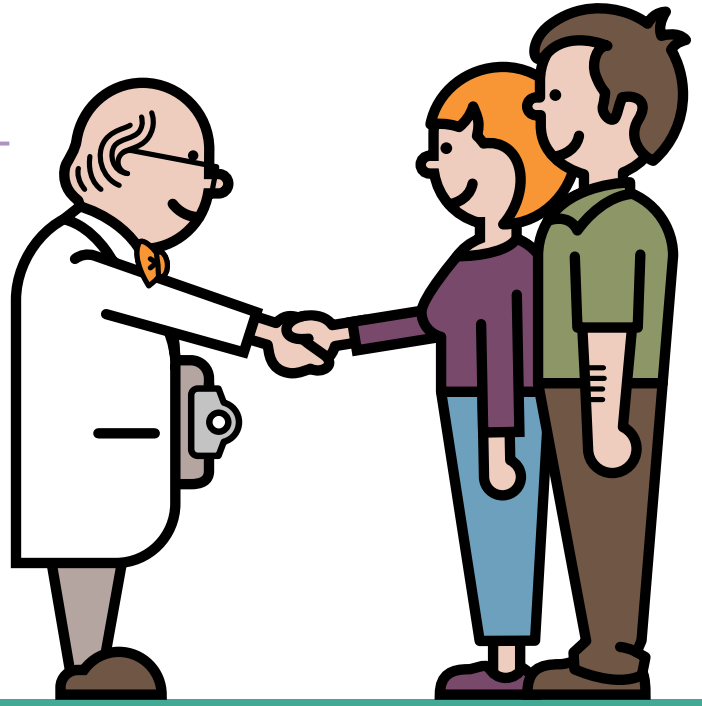
Each module produced is reviewed by a consultant clinician who is a member of the BIMDG.

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

Carnitine Palmitoyl Transferase 1 Deficiency

Information for families following a new diagnosis



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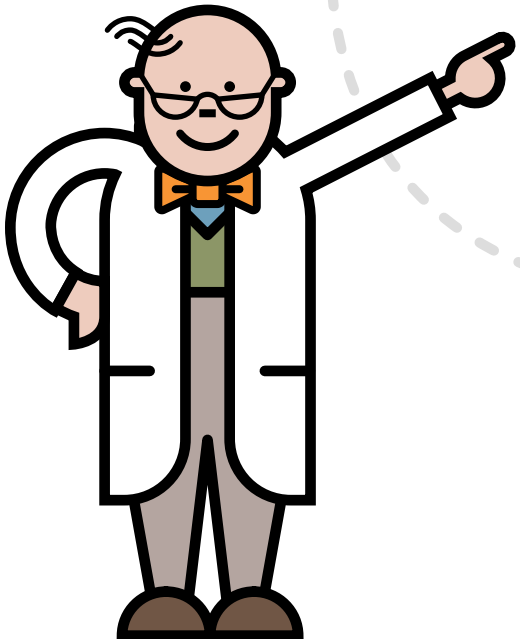
What is CPT 1 deficiency?

CPT 1 stands for Carnitine Palmitoyl Transferase 1 deficiency

It is an inherited metabolic condition

Carnitine **Palmitoyl** **Transferase** **1**

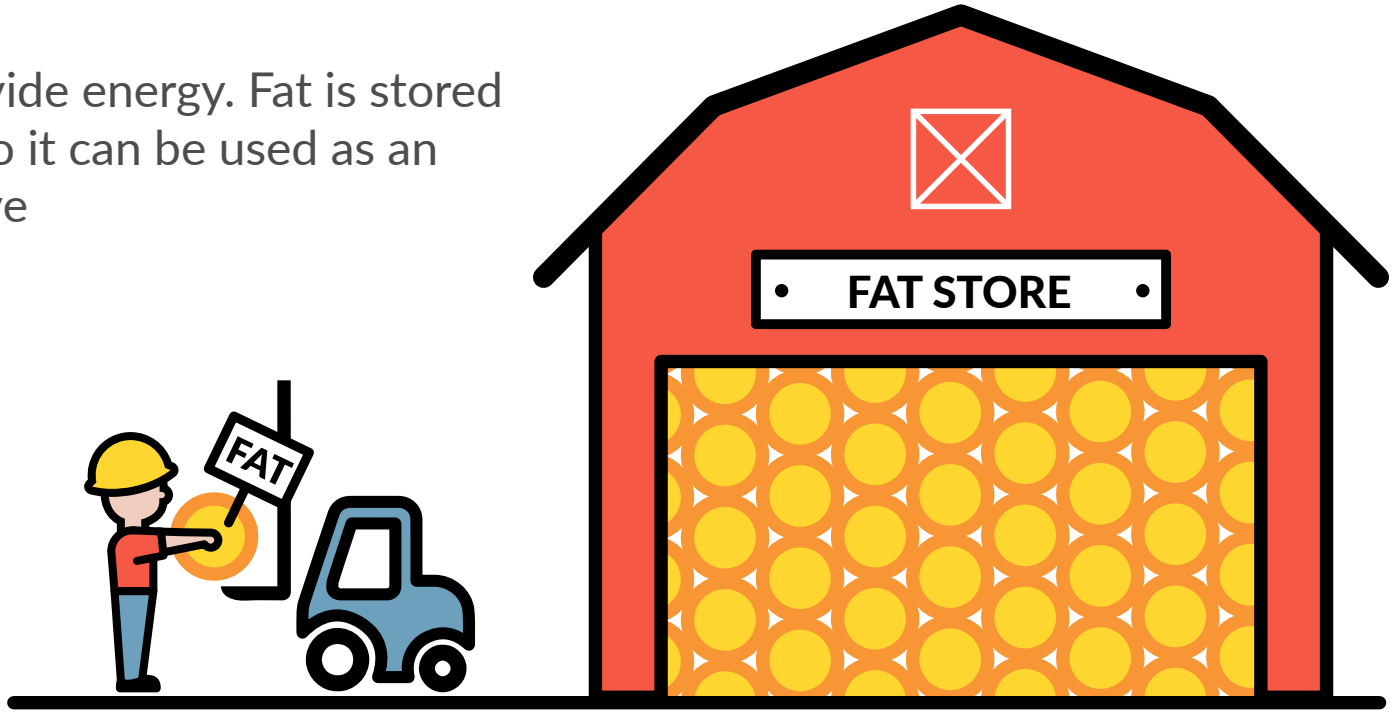


CPT 1 Deficiency

Which foods supply the body with energy?

There are two main food groups that supply the body with energy:

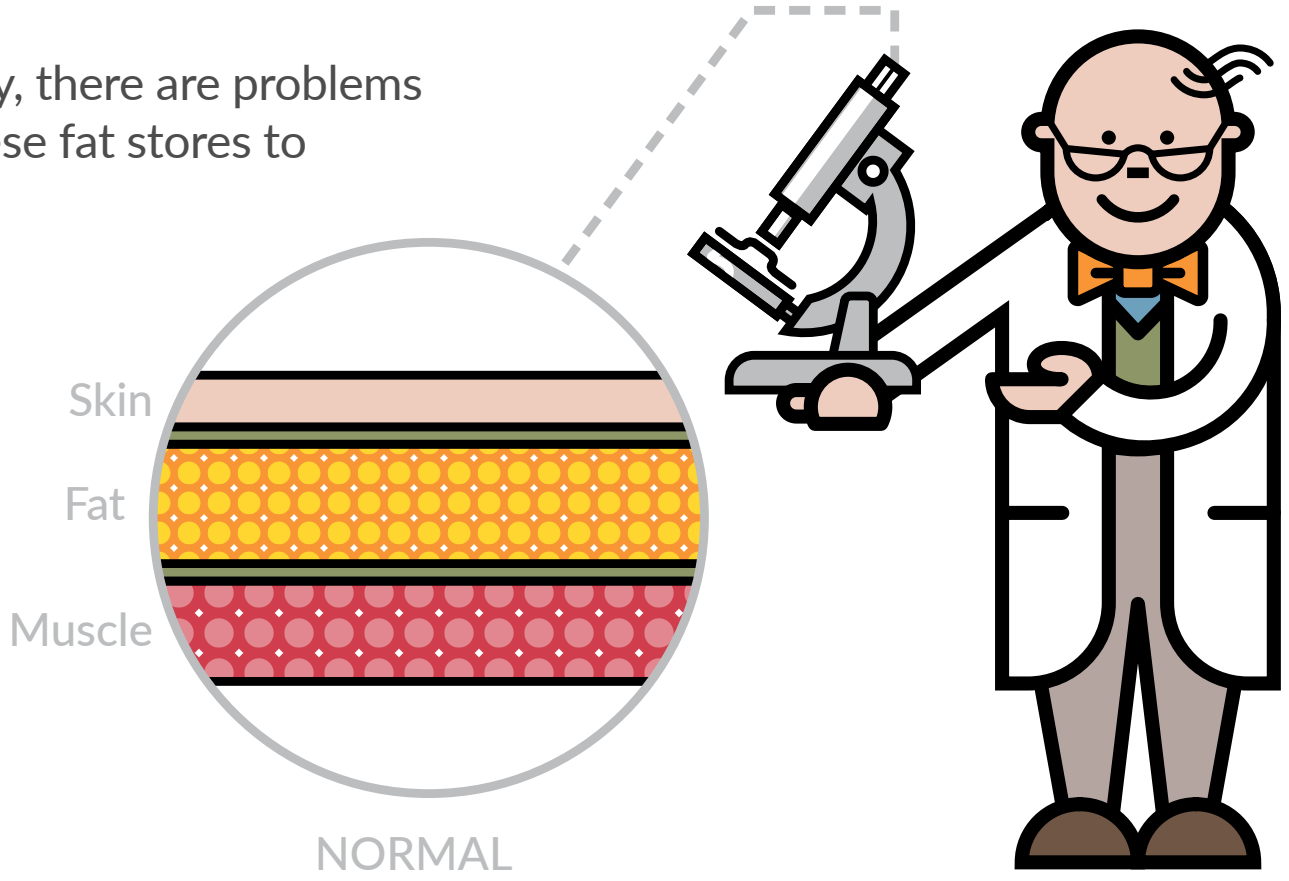
- Carbohydrates (starches and sugars) provide a readily available energy source
- Fats also provide energy. Fat is stored in the body so it can be used as an energy reserve



CPT 1 deficiency and fat

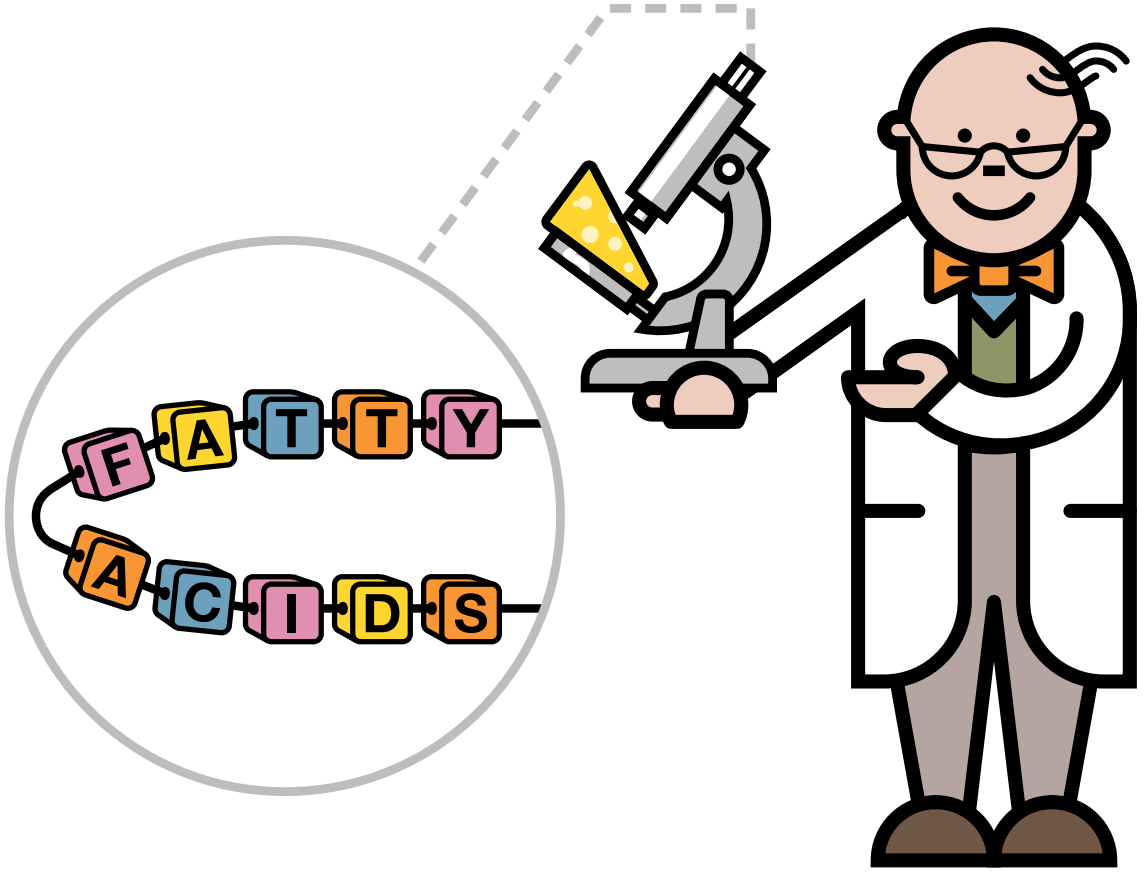
The body uses its own fat stores to provide energy when carbohydrate is depleted.

In CPT 1 deficiency, there are problems breaking down these fat stores to release energy.



Breaking down fat stores for energy

Body fat stores are broken down into fatty acids.



What are fatty acids?

Fatty acids are made up of carbon atoms joined together to form chains of many different lengths.

Short chain

Medium chain

Long chain

Fatty acid transport

Fatty acid chains need to be transported into the cells of the body. This enables the body to produce energy in a form which it can use.

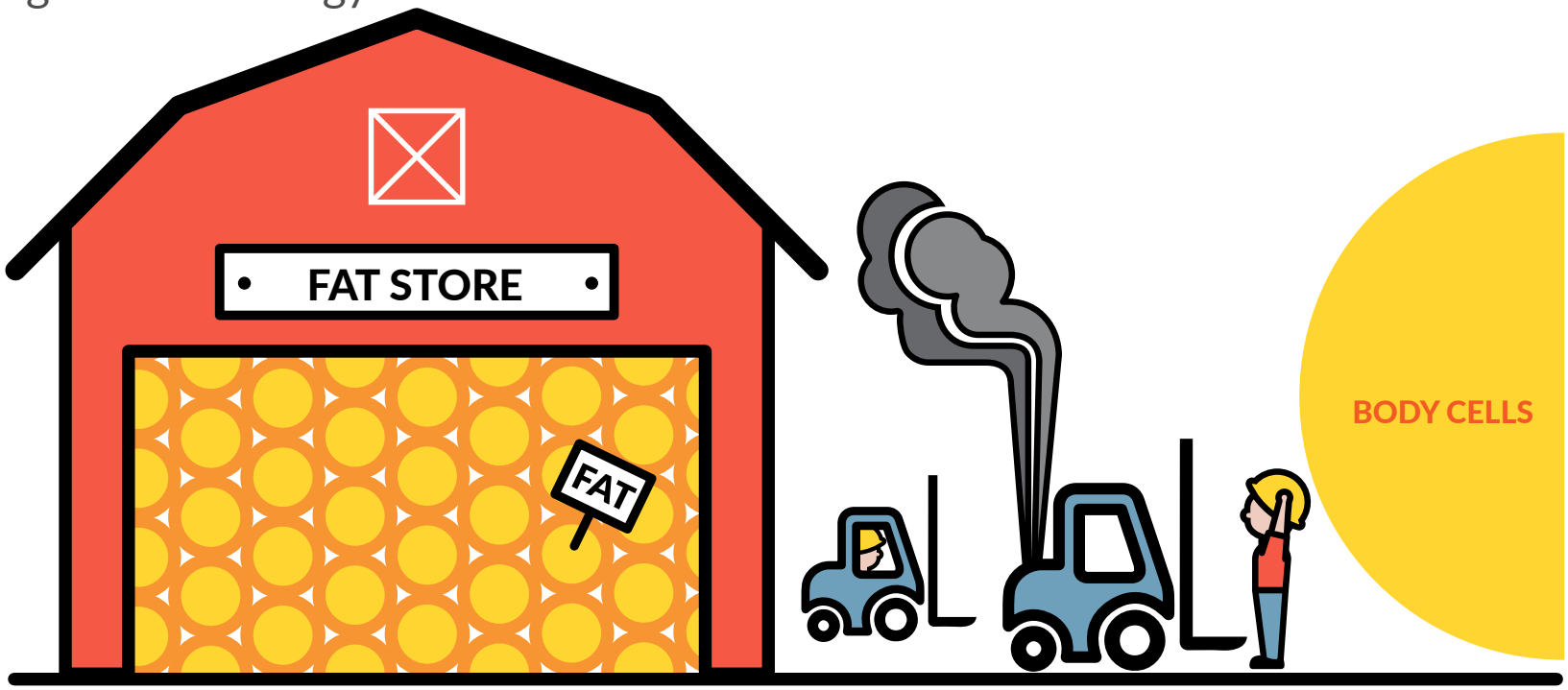
What happens in CPT 1 deficiency?

In CPT 1 deficiency, the body lacks a chemical (enzyme) that helps convert fat stores into energy.

This **stops** the long chain fats from being used for energy.

This is a problem when it is necessary to break down fats quickly.

This causes a shortage of energy supply.

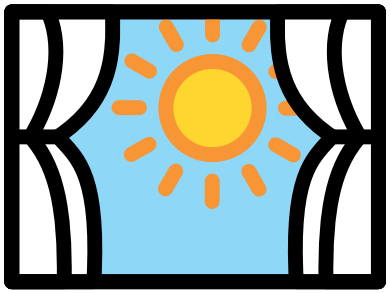


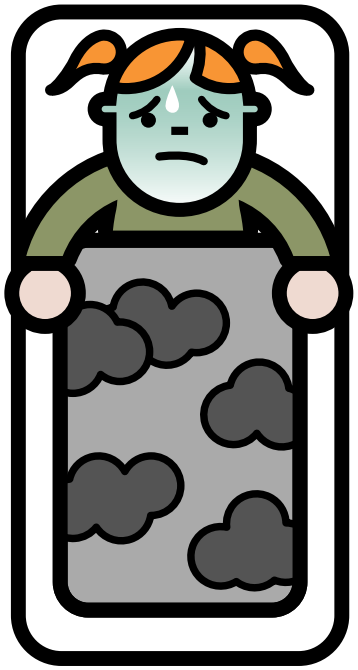
What can go wrong in CPT 1 deficiency in infants?

There can be a shortage of energy supply and a build up of harmful chemicals with illness or lack of food.

Symptoms include:

- poor feeding
- excessive sleepiness
- rapid breathing
- seizures
- low blood sugar
- floppiness
- liver problems
- heart problems





What can go wrong in CPT 1 deficiency?

If there is a shortage of energy and this is not corrected, it can lead to coma and brain damage and it may be life threatening.

However, please remember, this can all be prevented with timely and correct management.

Metabolic crisis

- A **metabolic crisis** triggers the CPT 1 deficiency symptoms
- This leads to a lack of energy and build up of toxic chemicals
- It is usually triggered by childhood infections causing high temperatures, vomiting, and diarrhoea
- It can also be caused by going a very long time without food
- Avoidance of a metabolic crisis is essential

How is CPT 1 deficiency diagnosed?

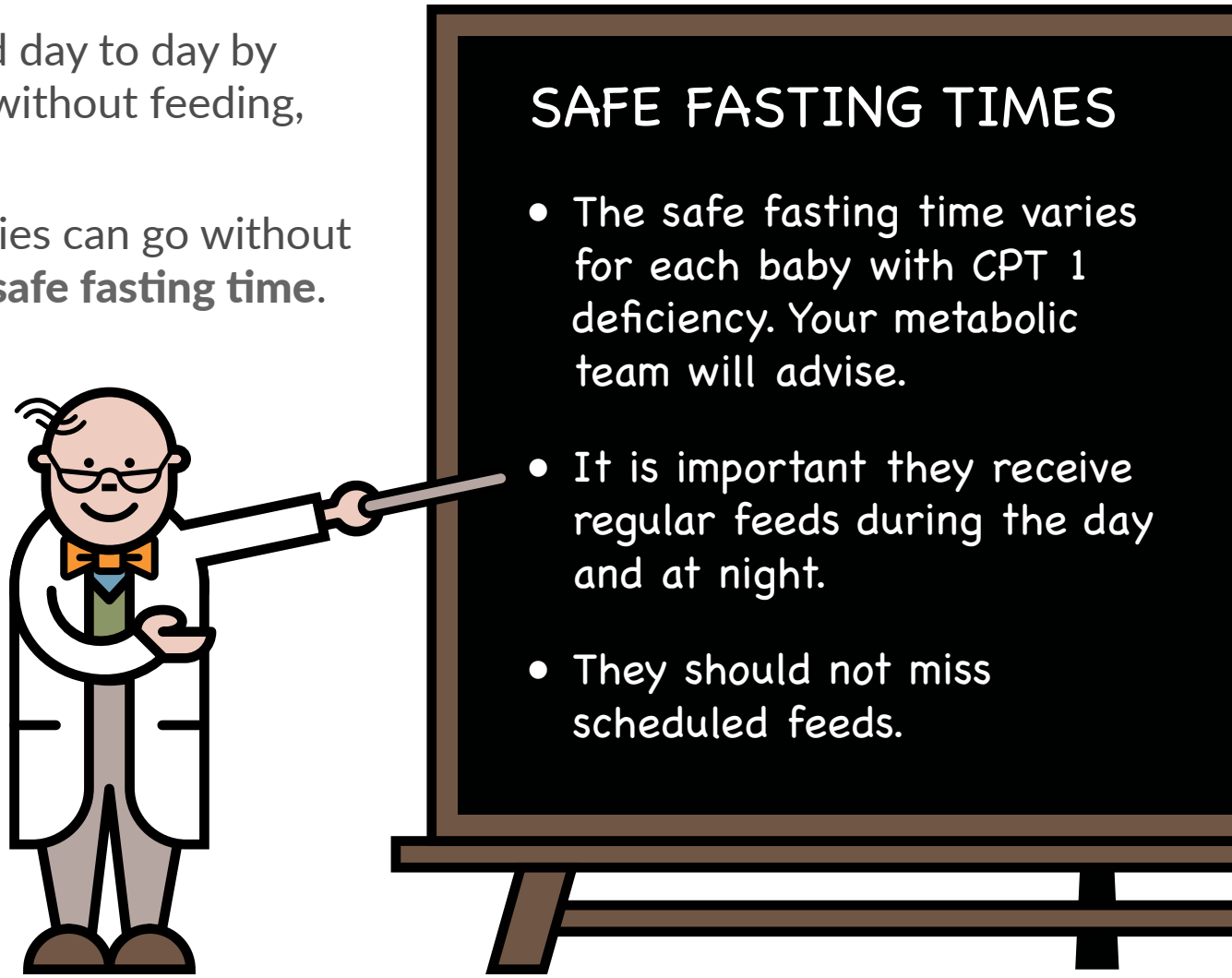
CPT 1 deficiency is suspected because of the pattern of chemicals (acylcarnitines and organic acids) found in the blood and urine.

The diagnosis is confirmed by finding mutations in the CPT 1 gene.

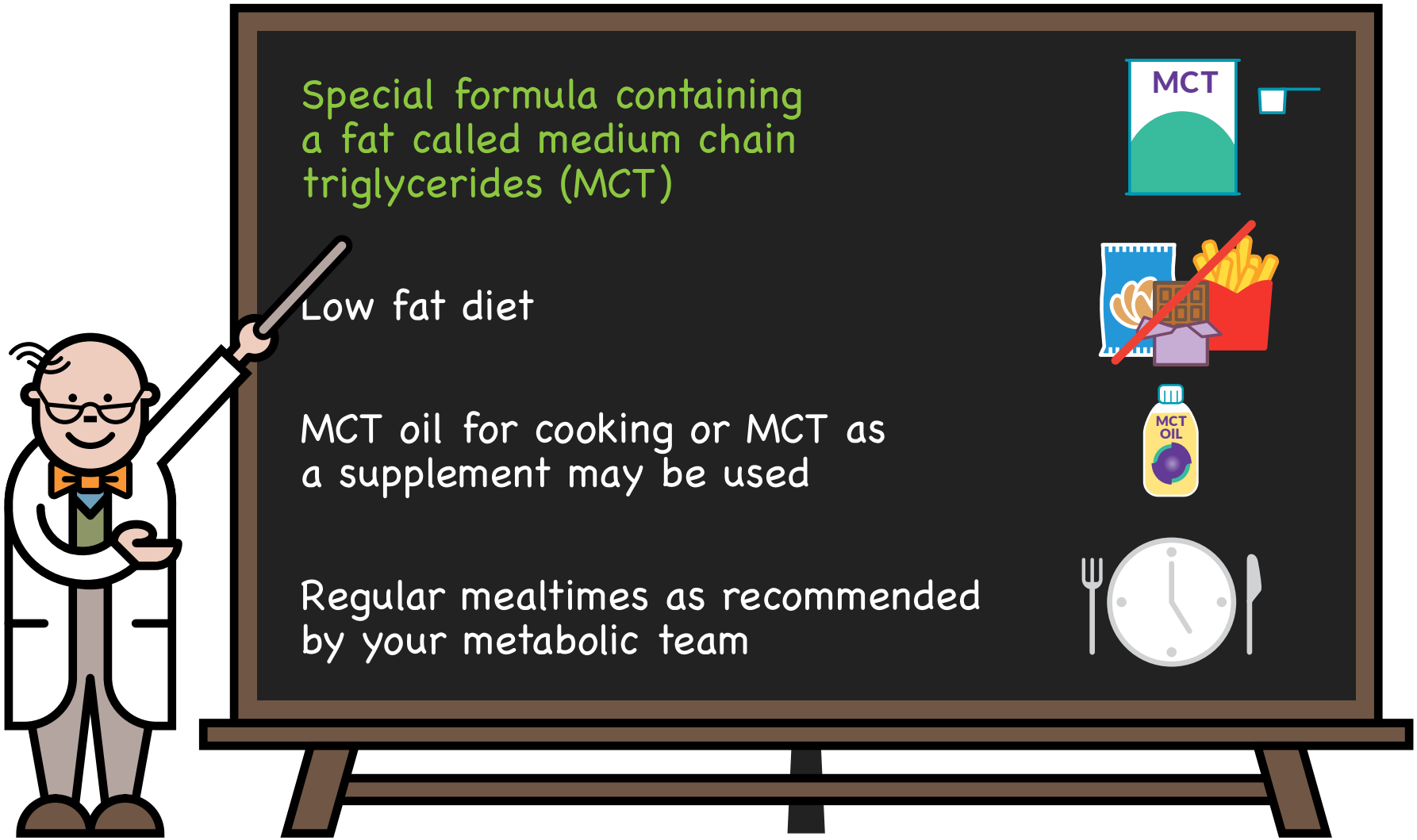
How is CPT 1 deficiency managed?

In babies, it is managed day to day by **avoiding** long periods without feeding, even when well.

The length of time babies can go without feeds is known as **the safe fasting time**.



How is CPT 1 deficiency managed day to day?



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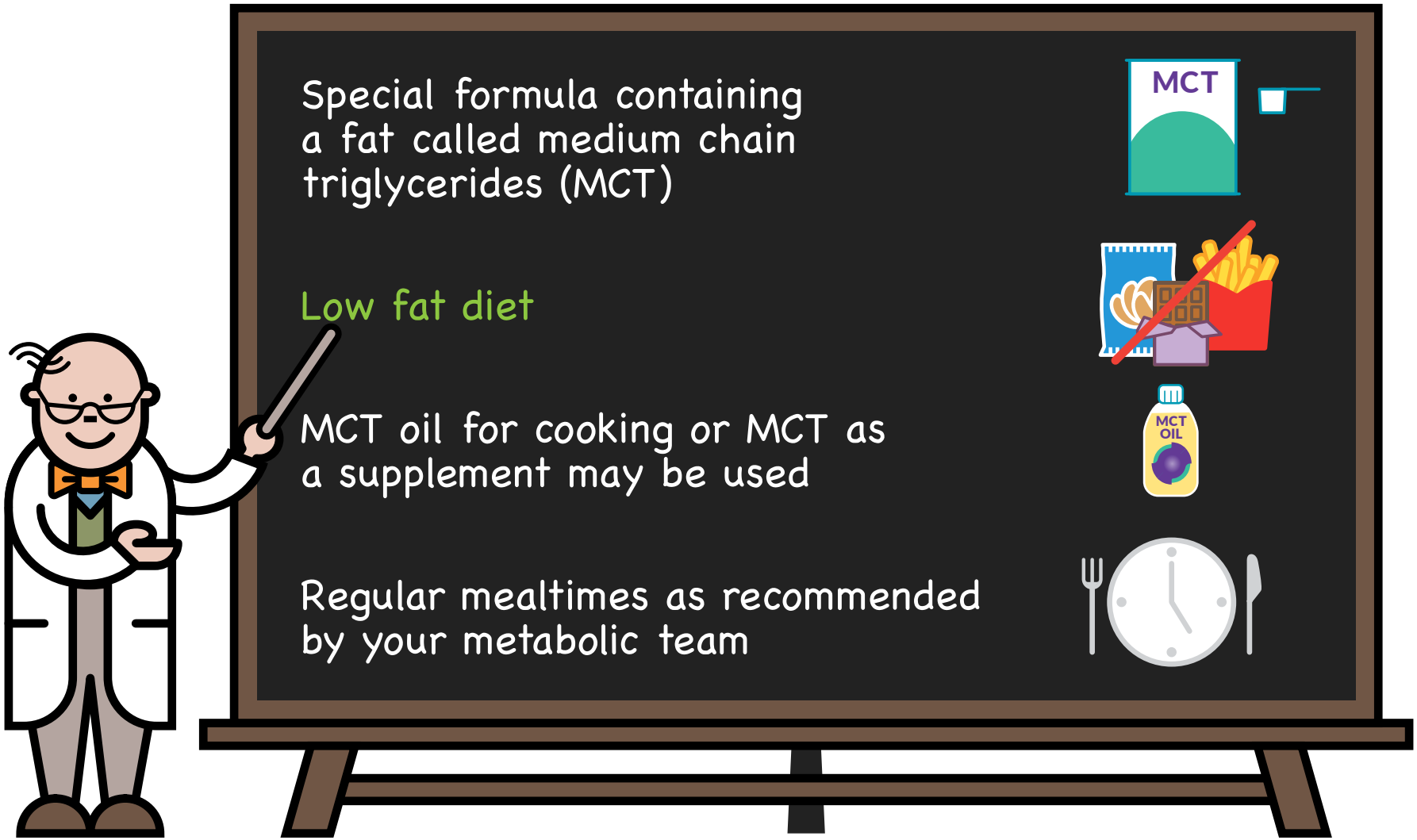

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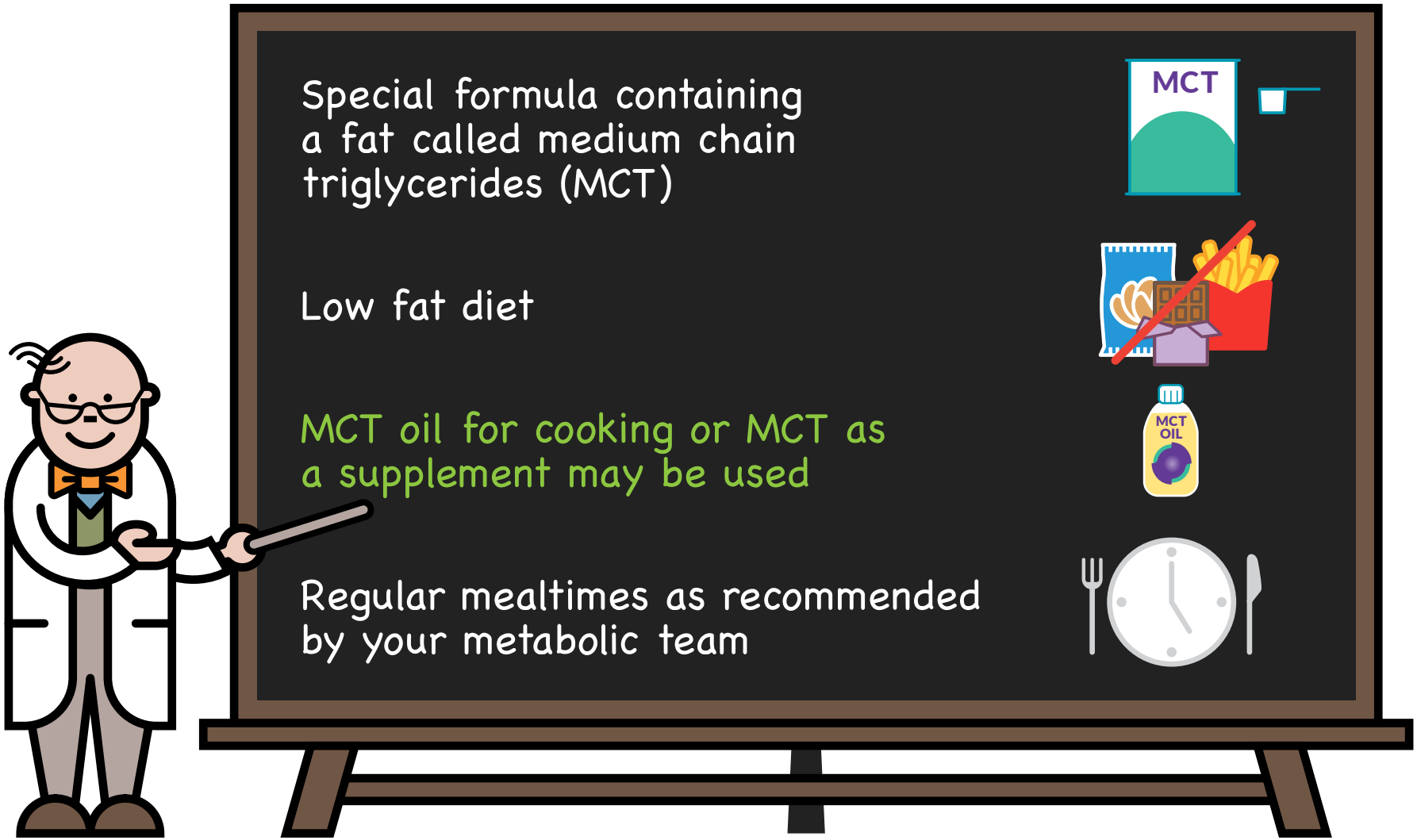

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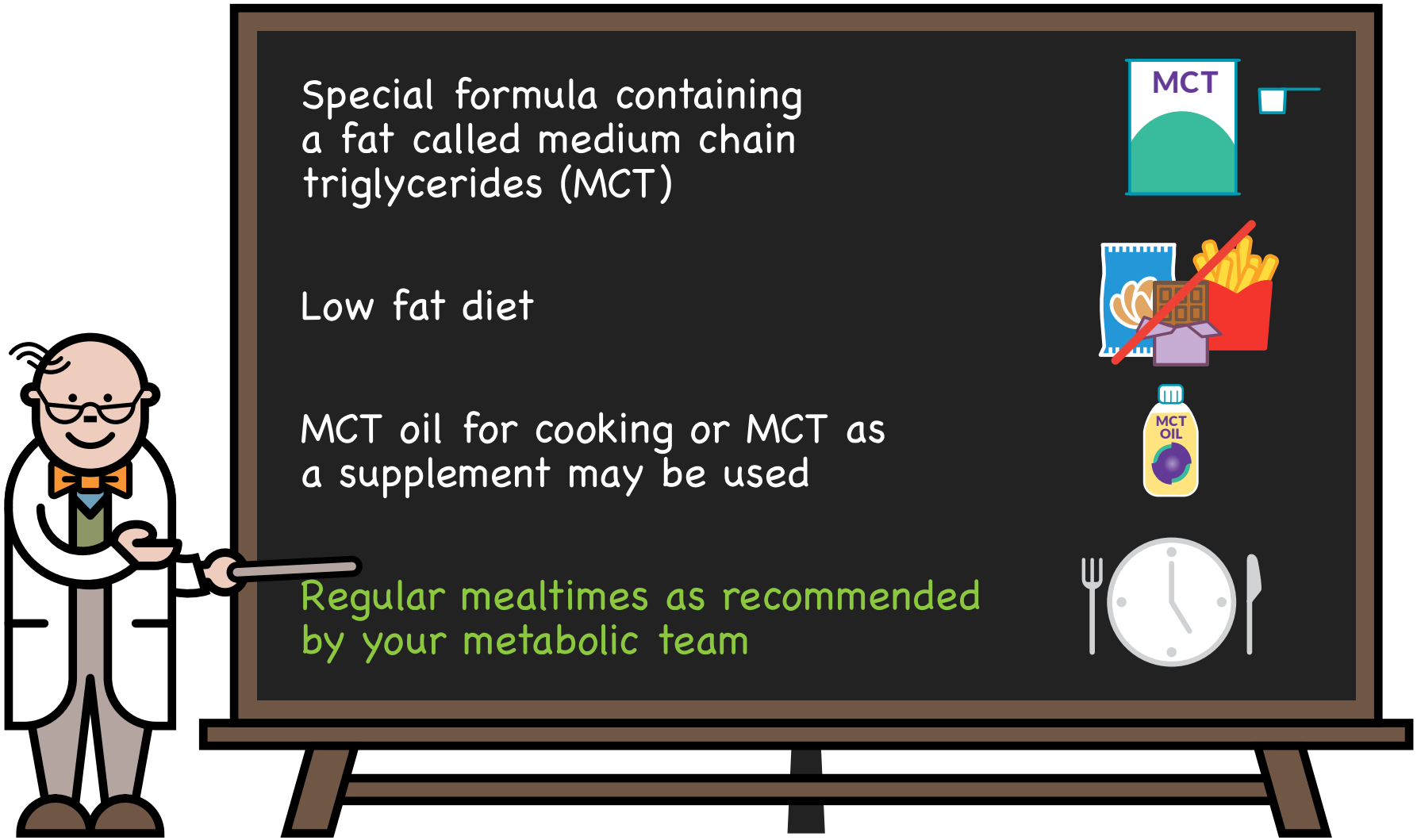


CPT 1 deficiency and fat

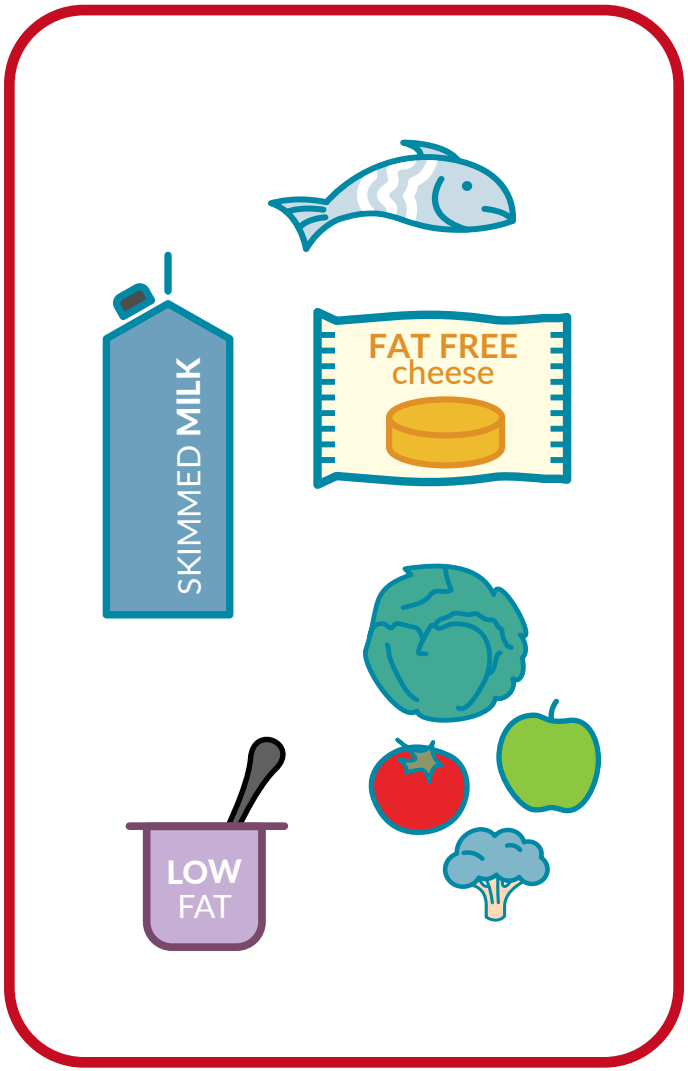
The diet needs to be low in fat.

Foods high in fat are avoided.
Many foods are high in fat e.g. full fat milk, full fat cheese, fatty meat, eggs, ice cream, chips, crisps and chocolate.

Very low fat foods

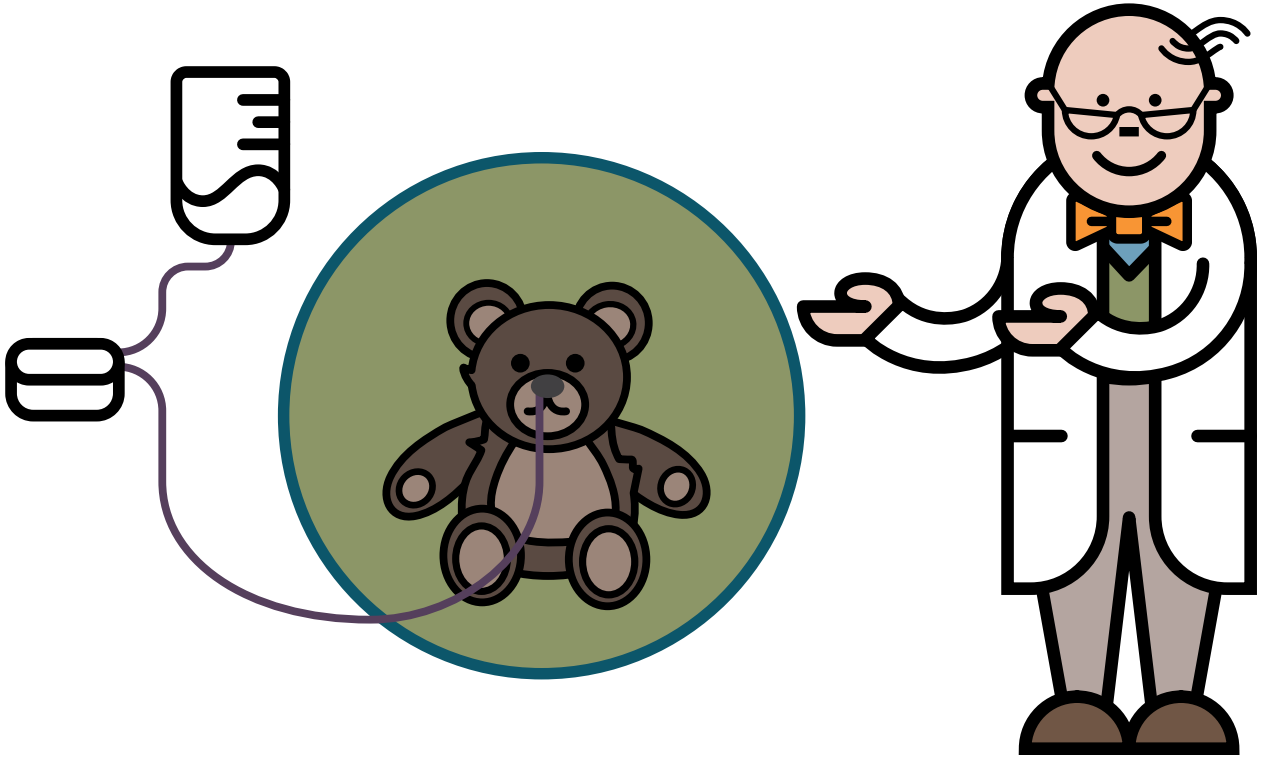
The diet needs to be low in fat.

Very low fat foods are permitted.
There is a wide range of very low fat foods available e.g. skimmed milk, very low fat yoghurt/cheese, white fish, fruit and vegetables.



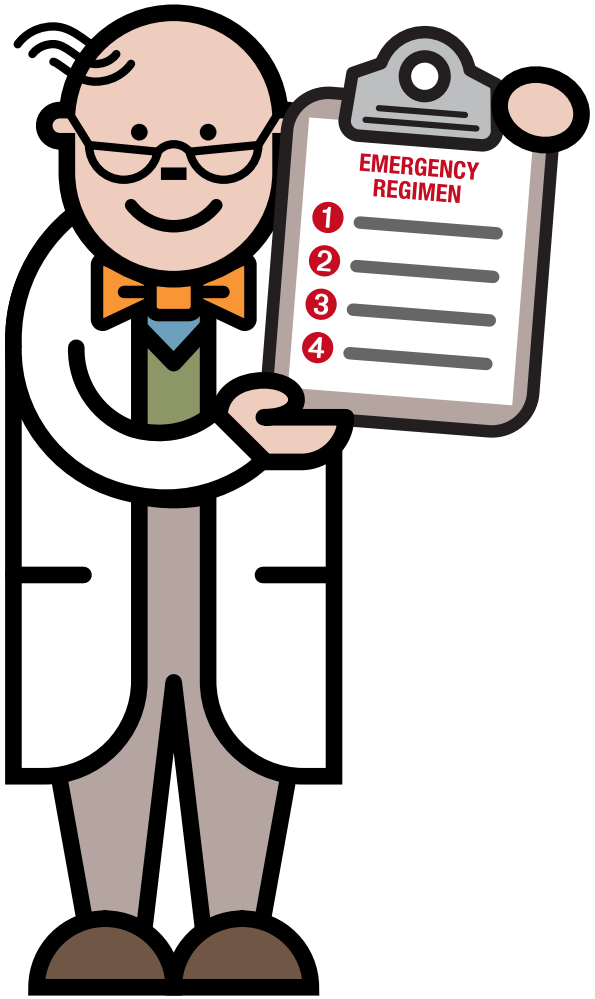
Is tube feeding needed?

In babies with the most severe forms of CPT 1 deficiency, tube feeding may be necessary. This will ensure energy, nutrient and fluid needs are met.

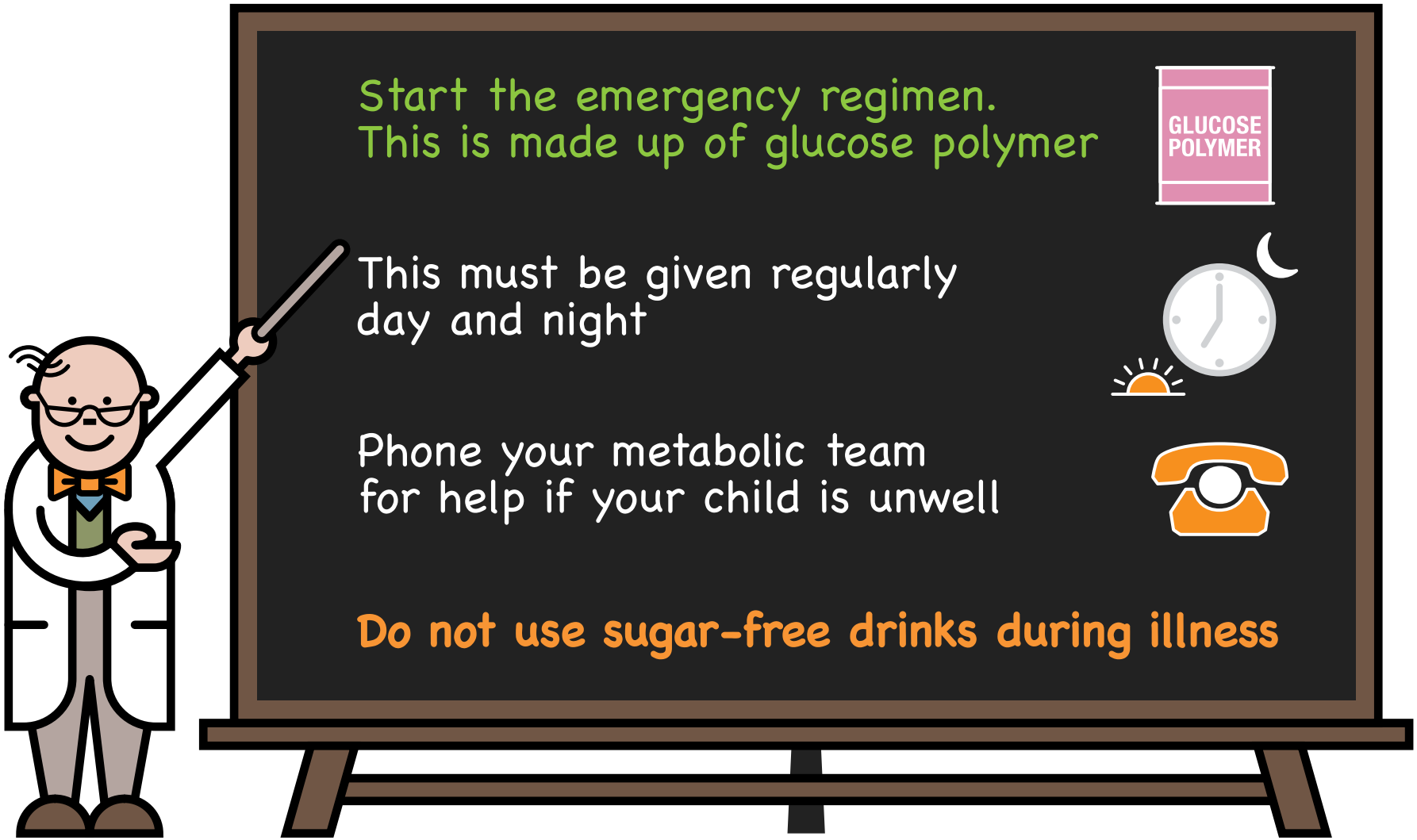


How is CPT 1 deficiency managed during illness?

- During any childhood illness, an emergency regimen is given
- This provides energy and prevents build up of harmful chemicals that cause a metabolic crisis



How is CPT 1 deficiency managed during illness?



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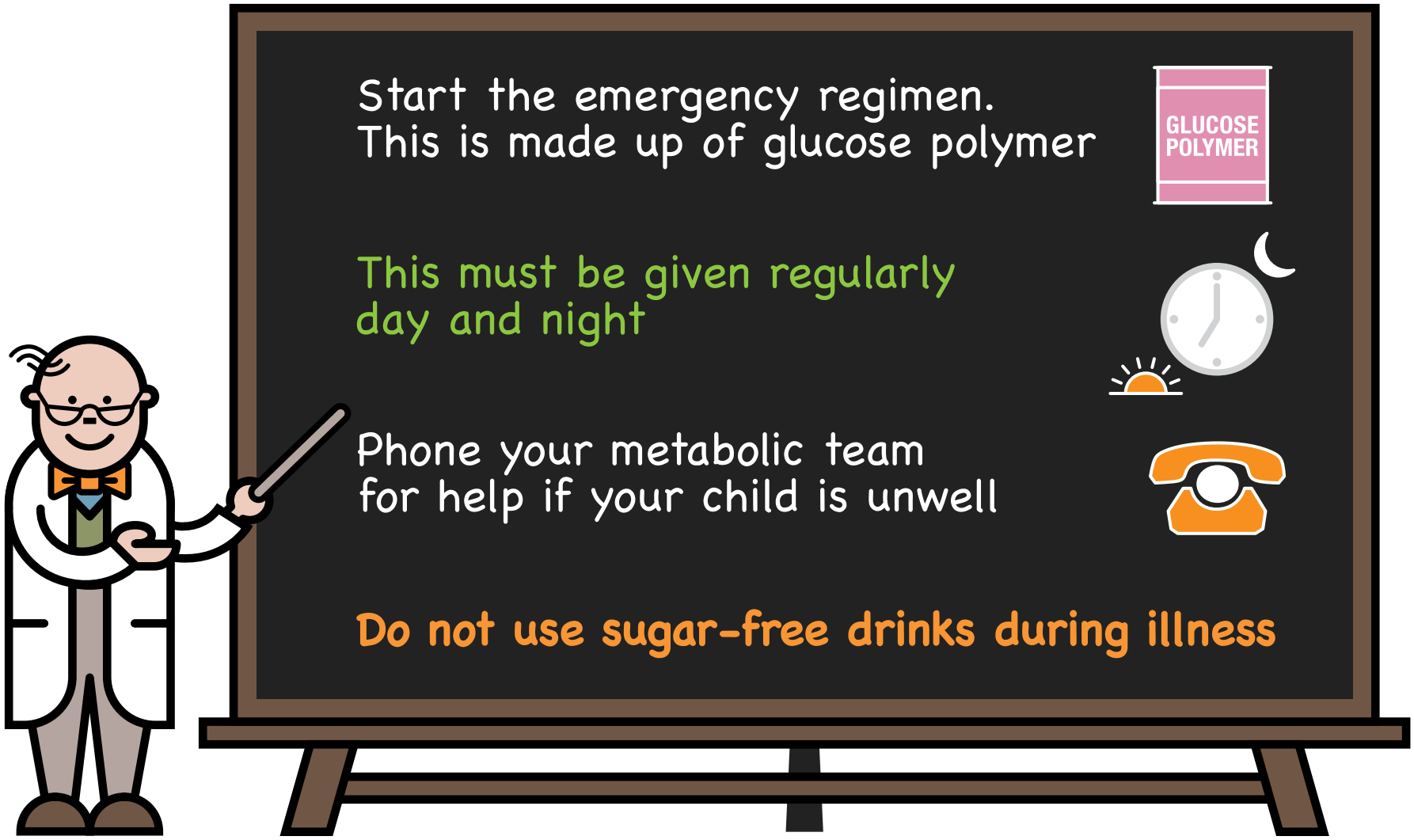

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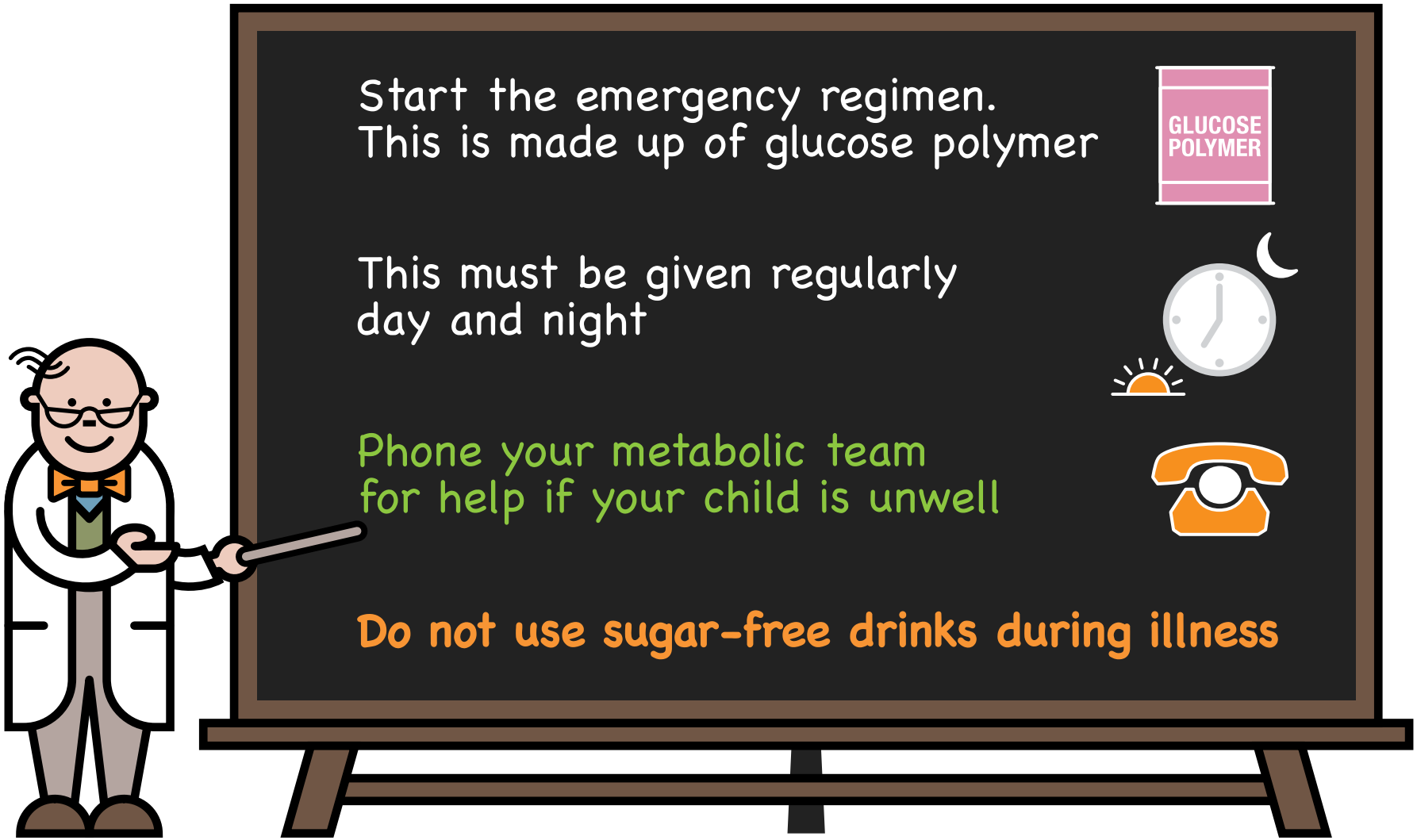


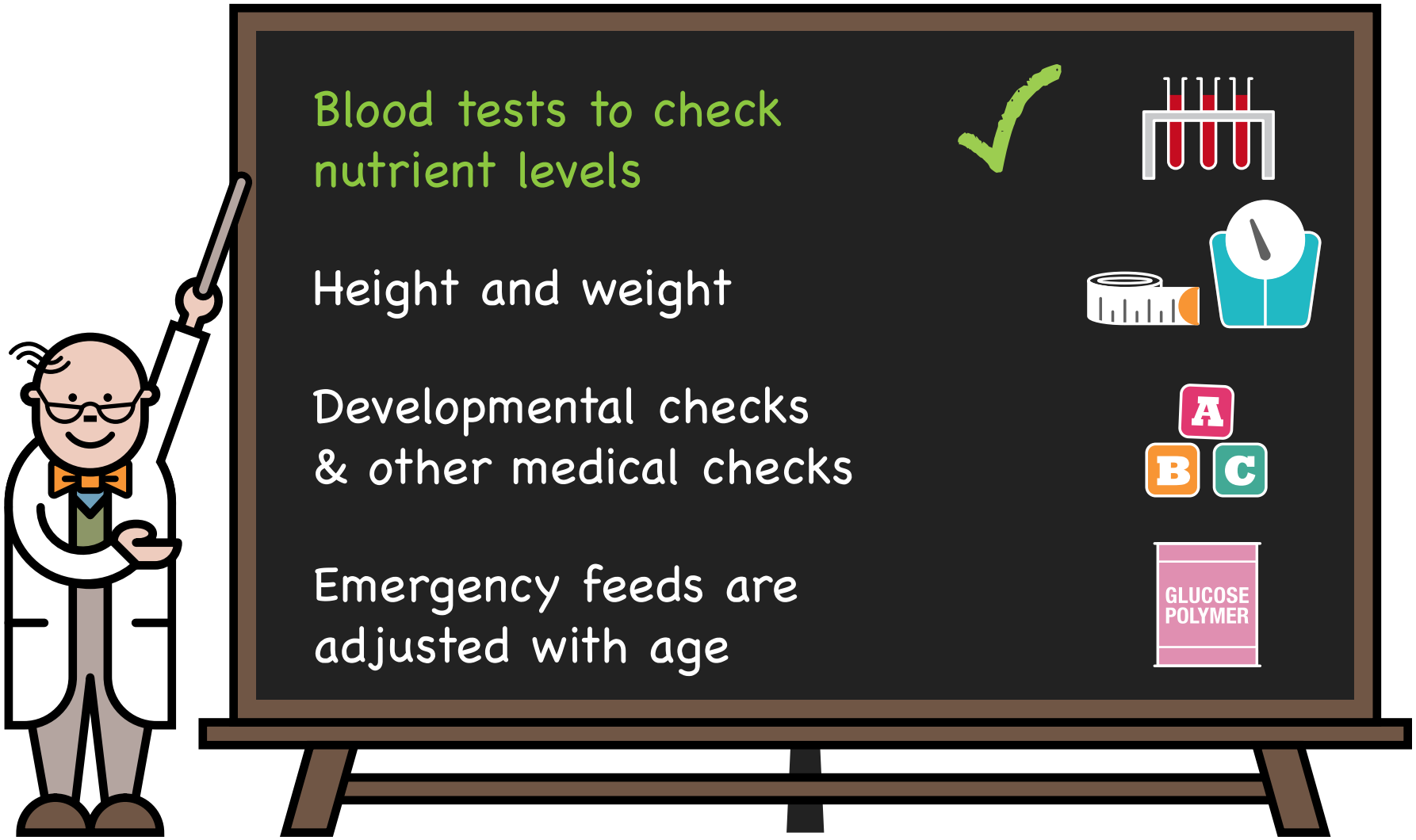
Checklist for illness

Checklist for illness

Checklist for illness

Key message

How is CPT 1 deficiency monitored?



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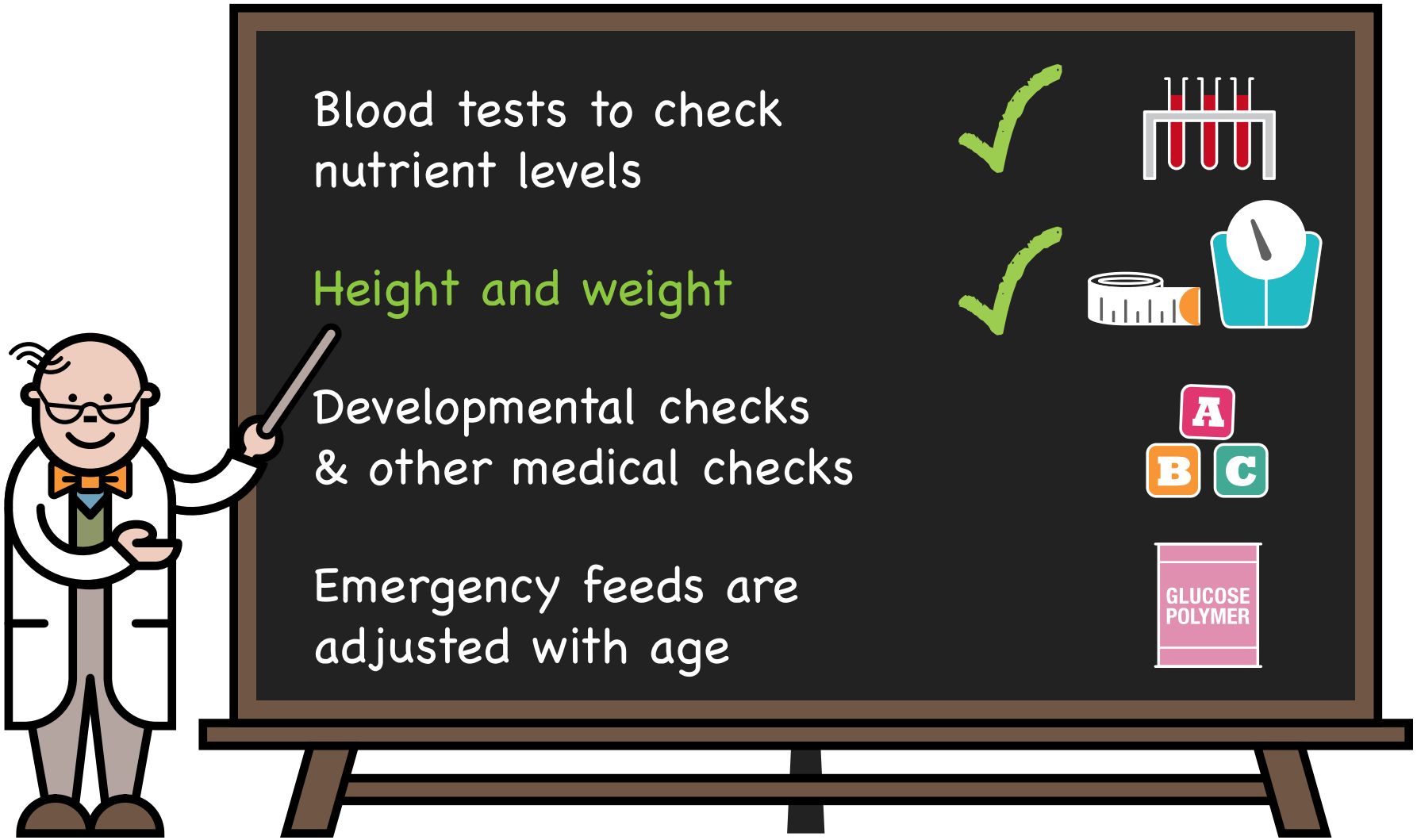

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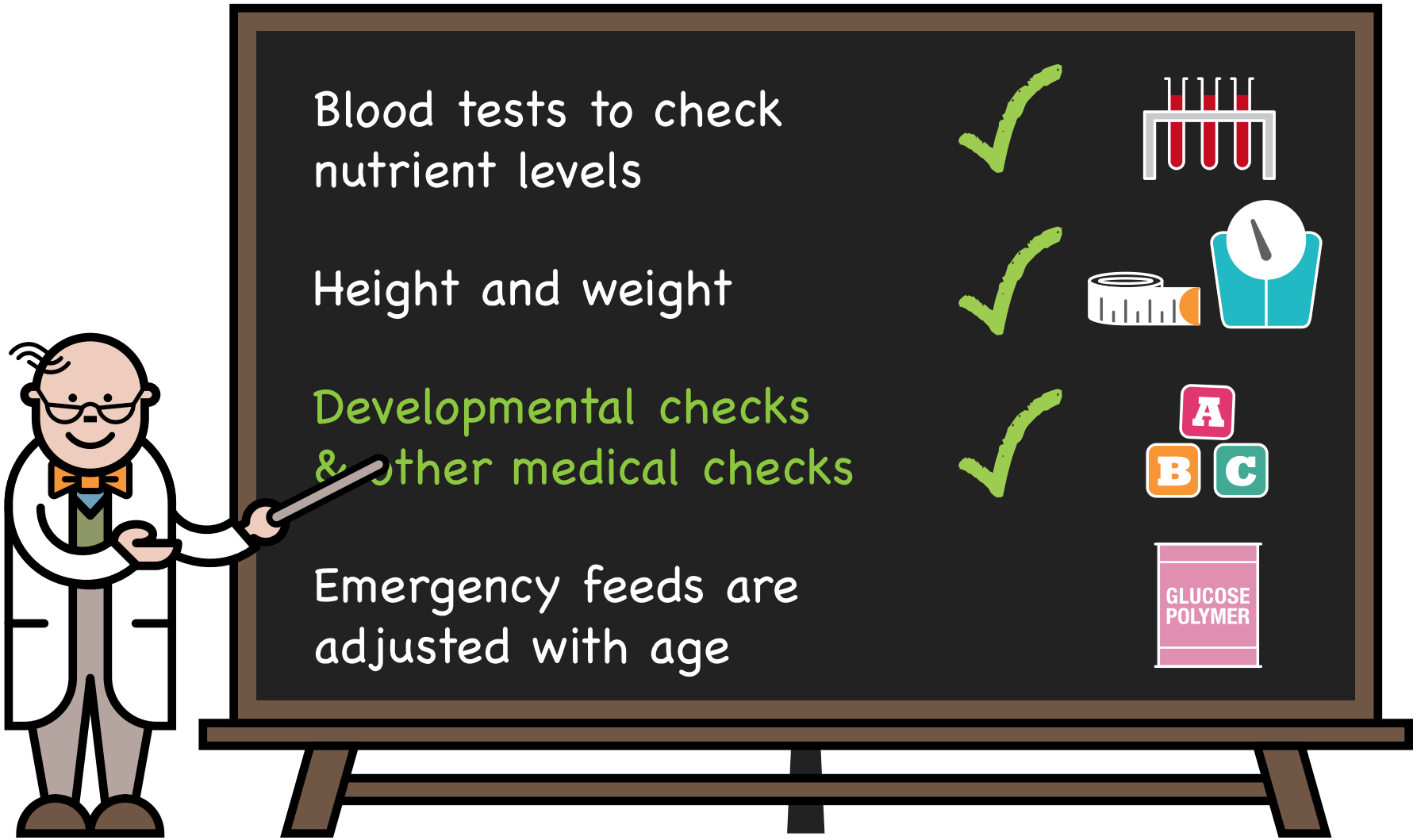

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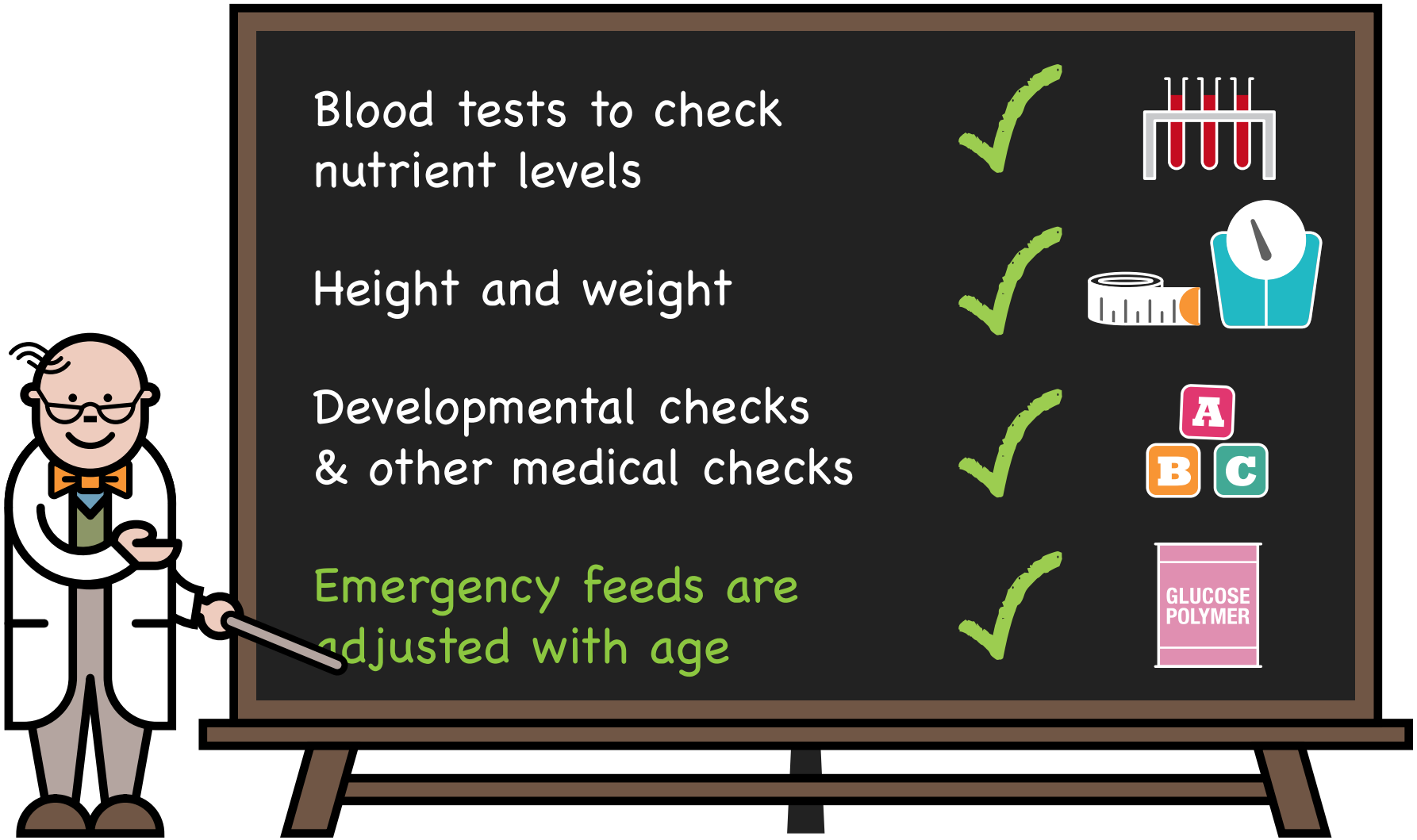
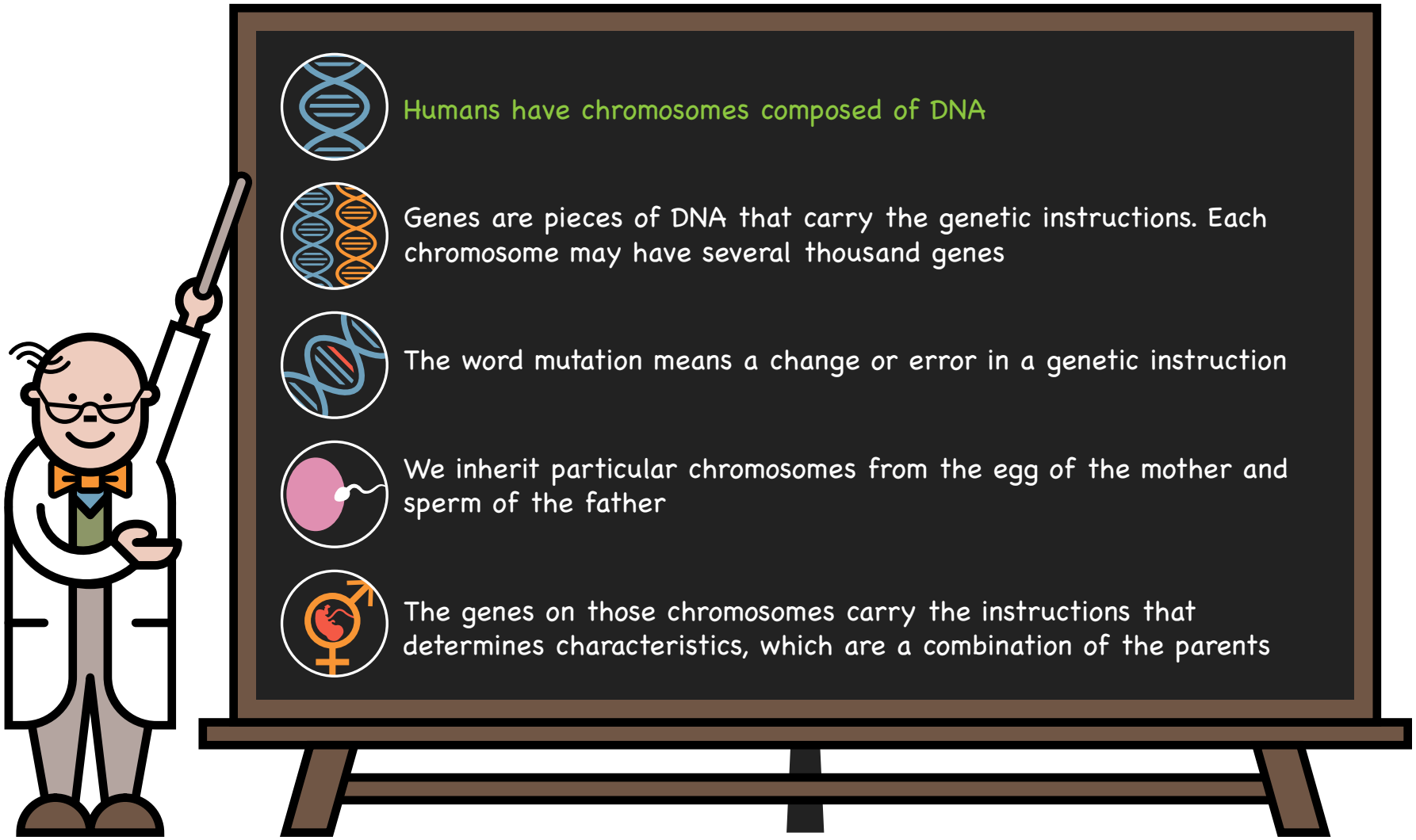


Chromosomes, genes, mutations



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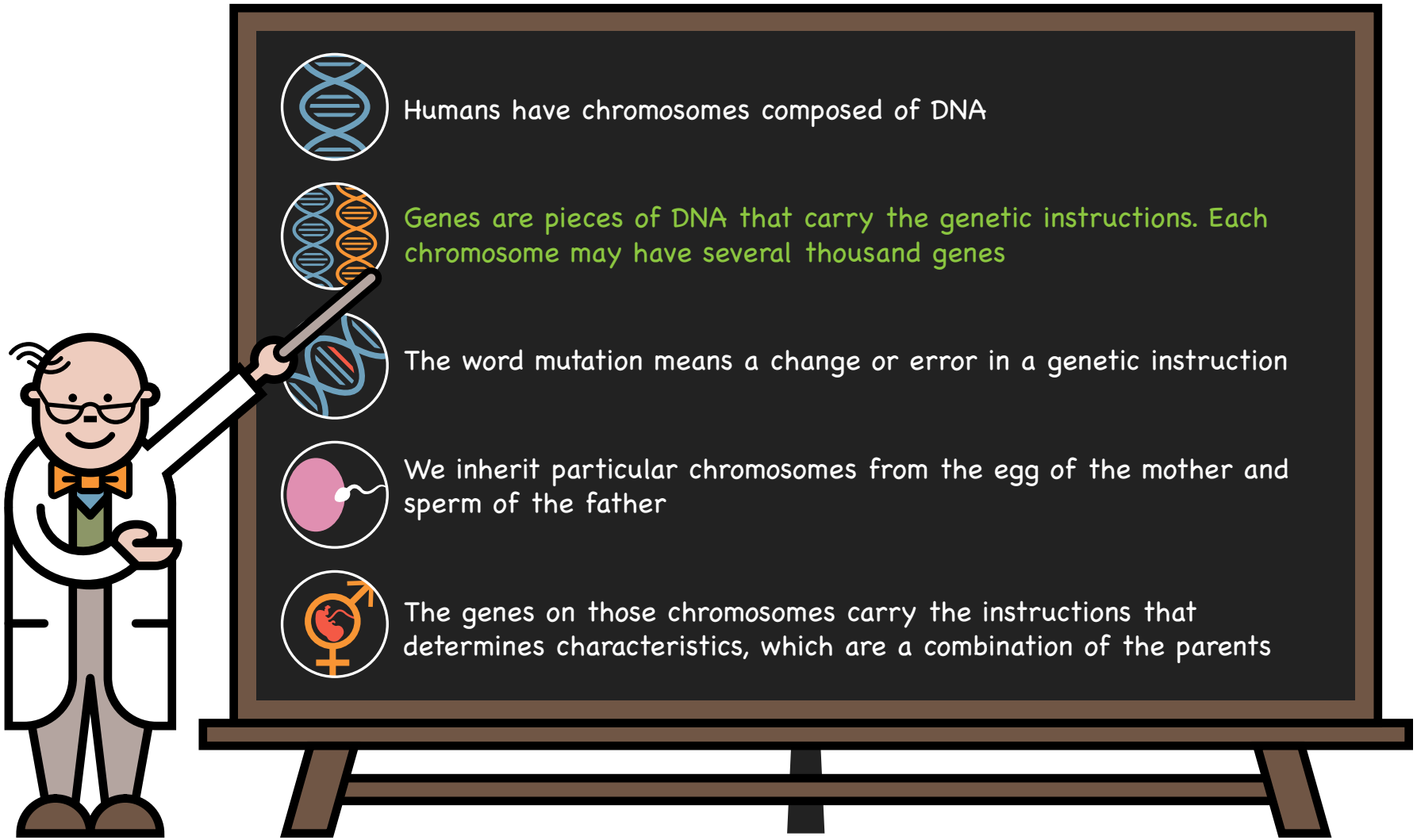
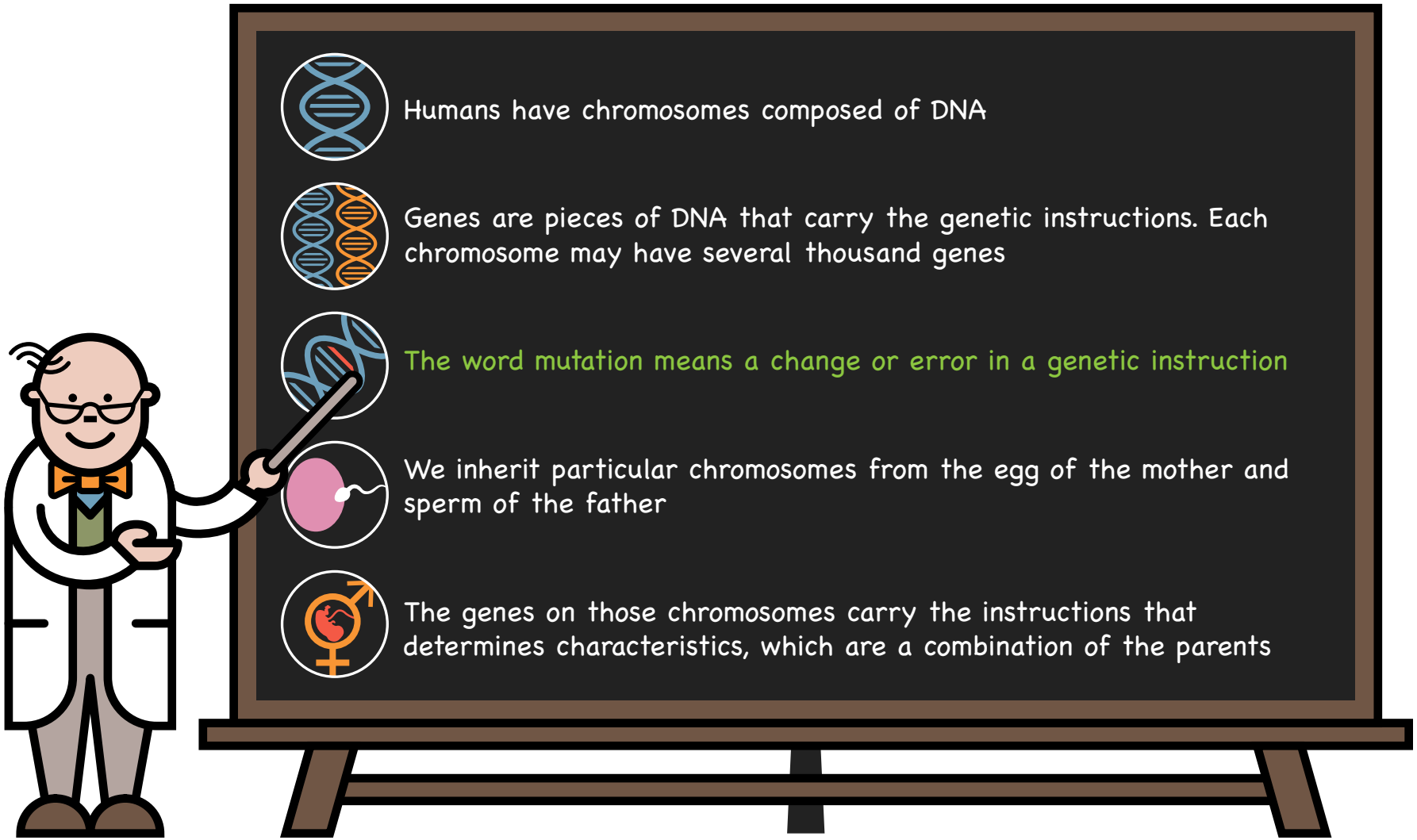
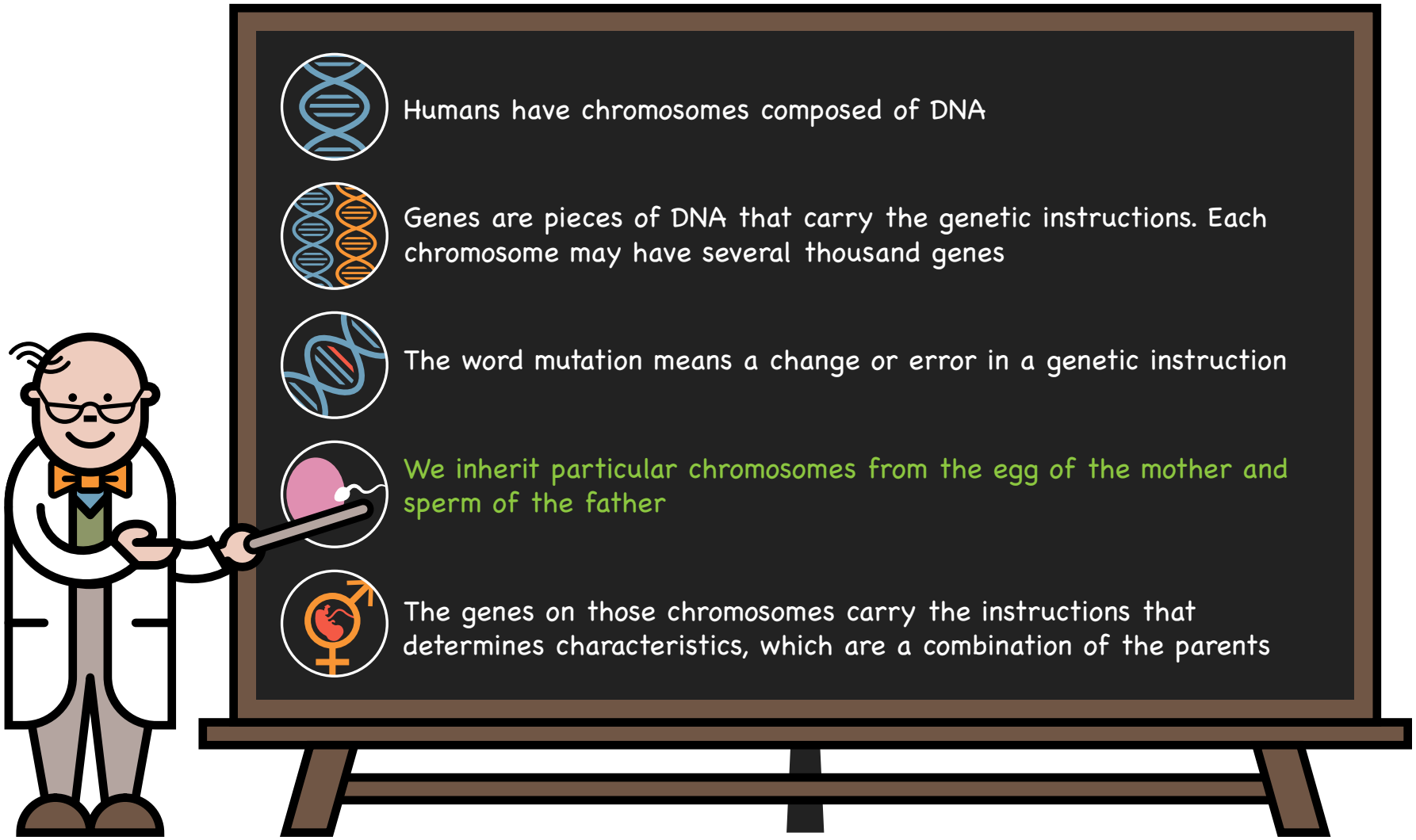


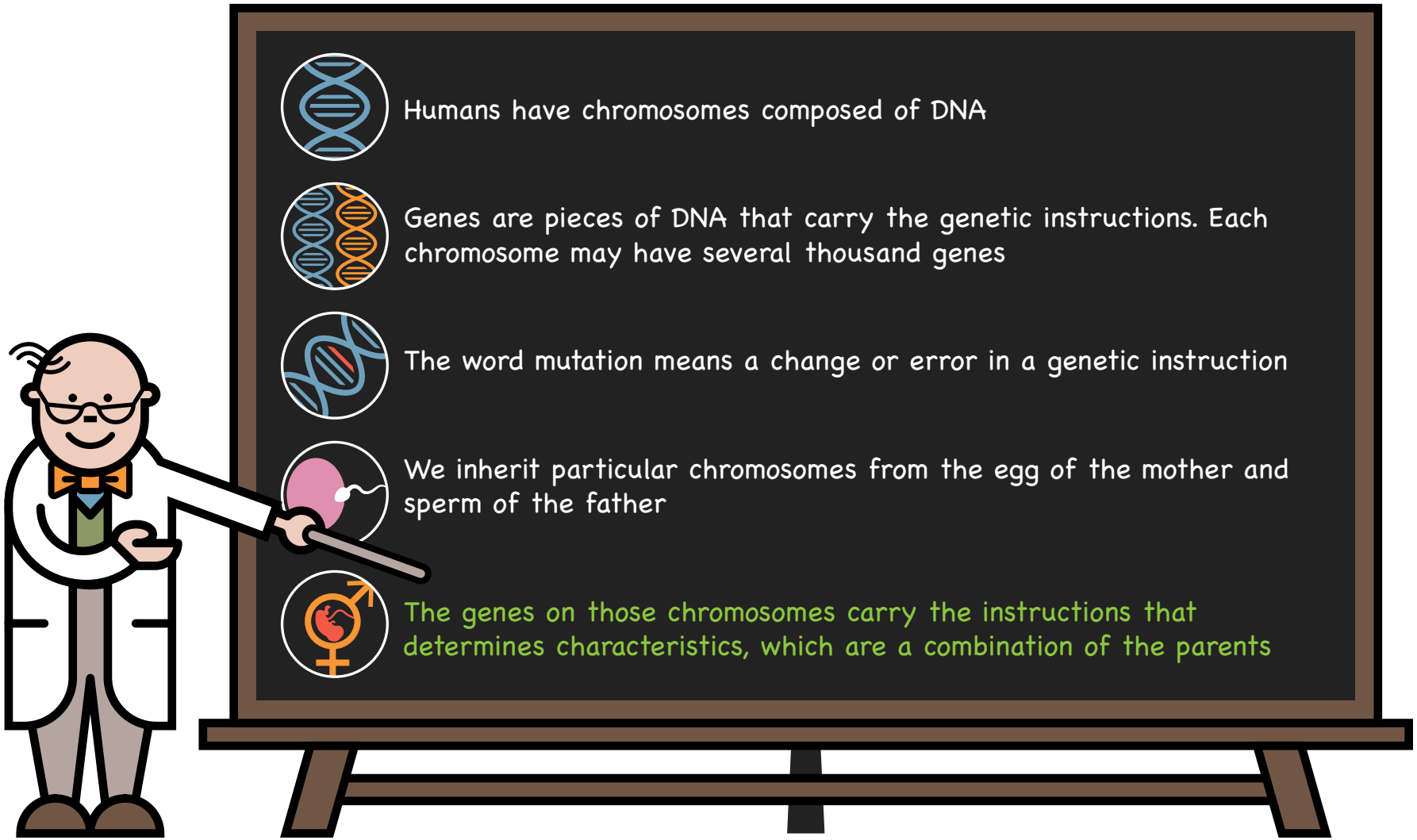
Chromosomes, genes, mutations



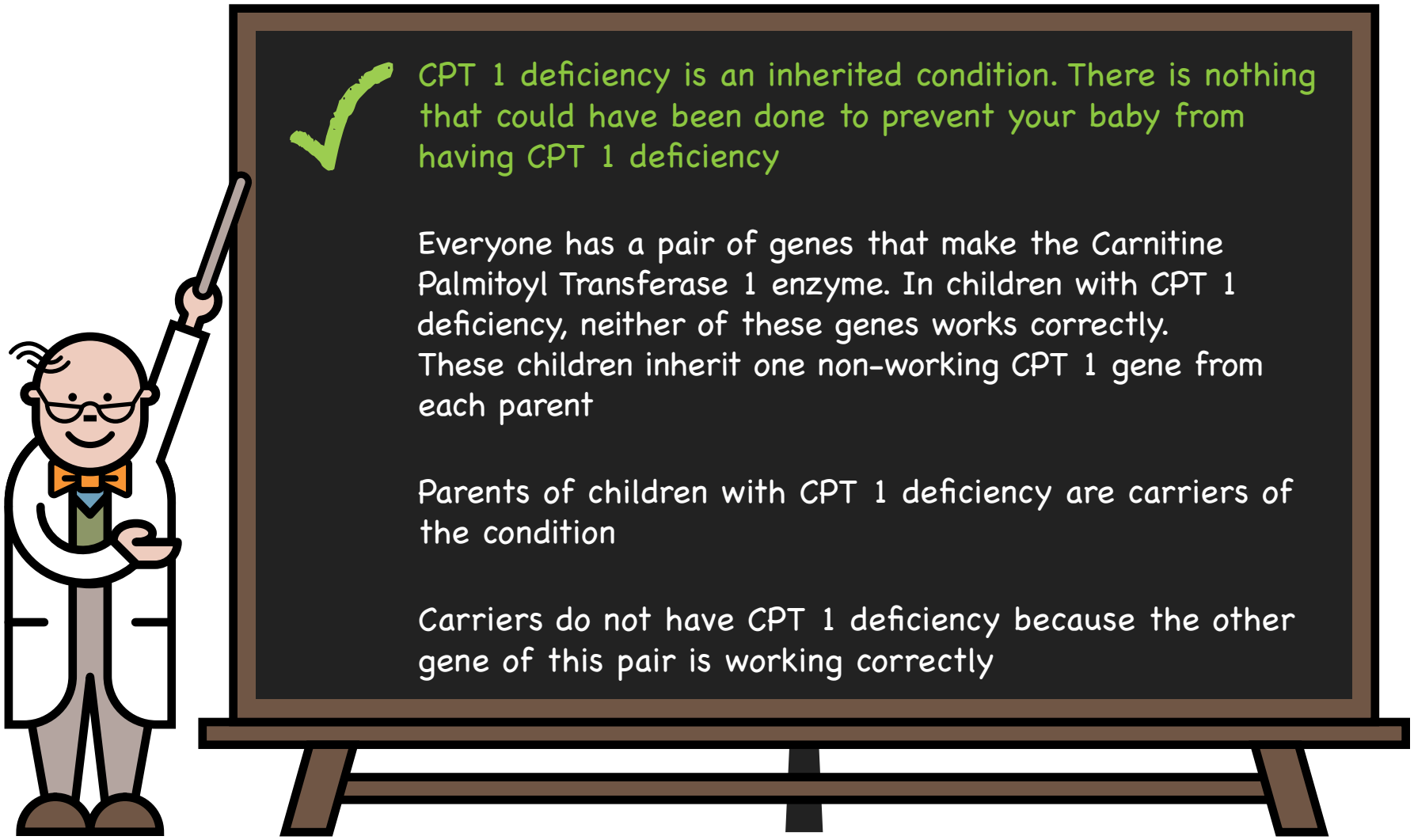
Chromosomes, genes, mutations



Chromosomes, genes, mutations



Inheritance



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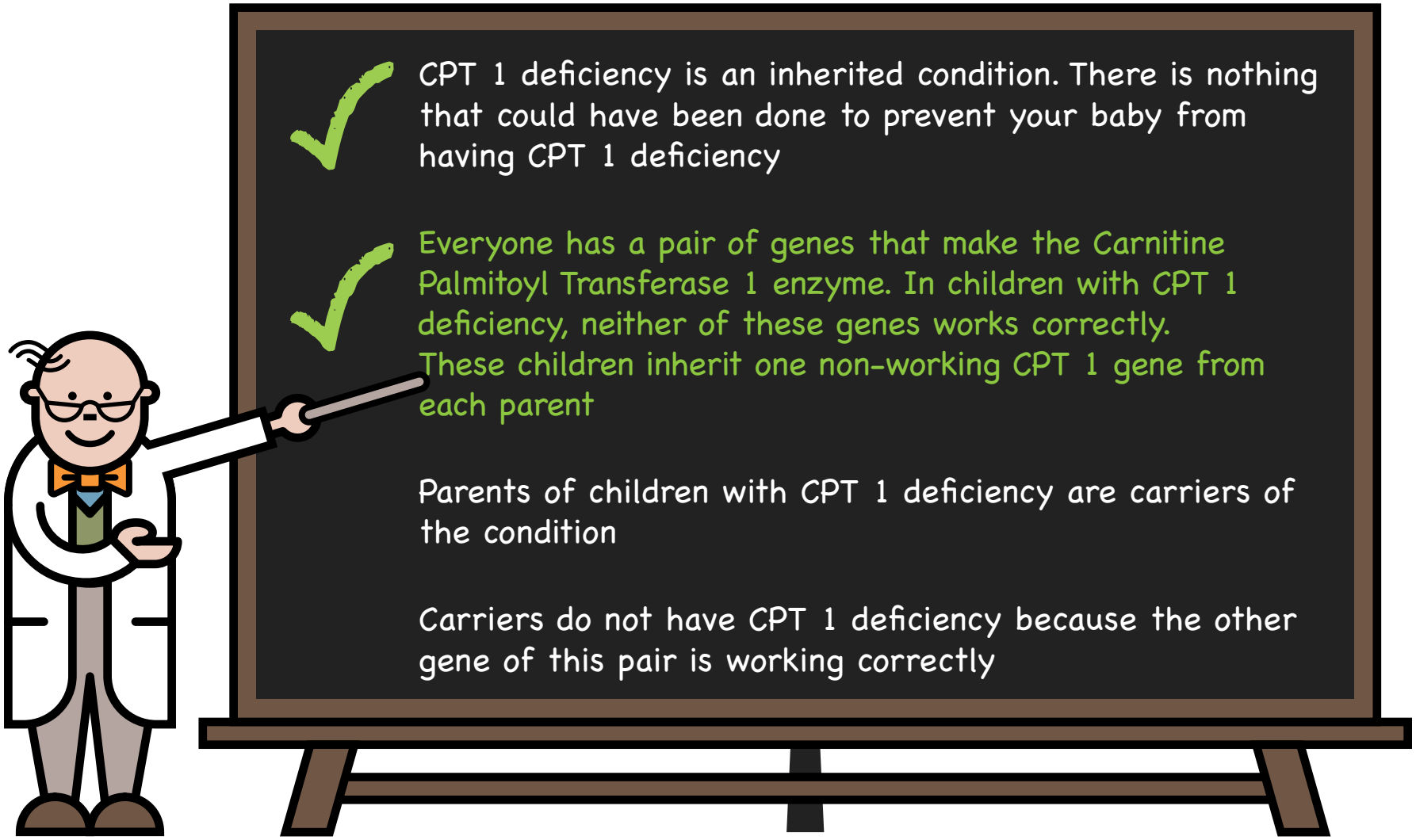

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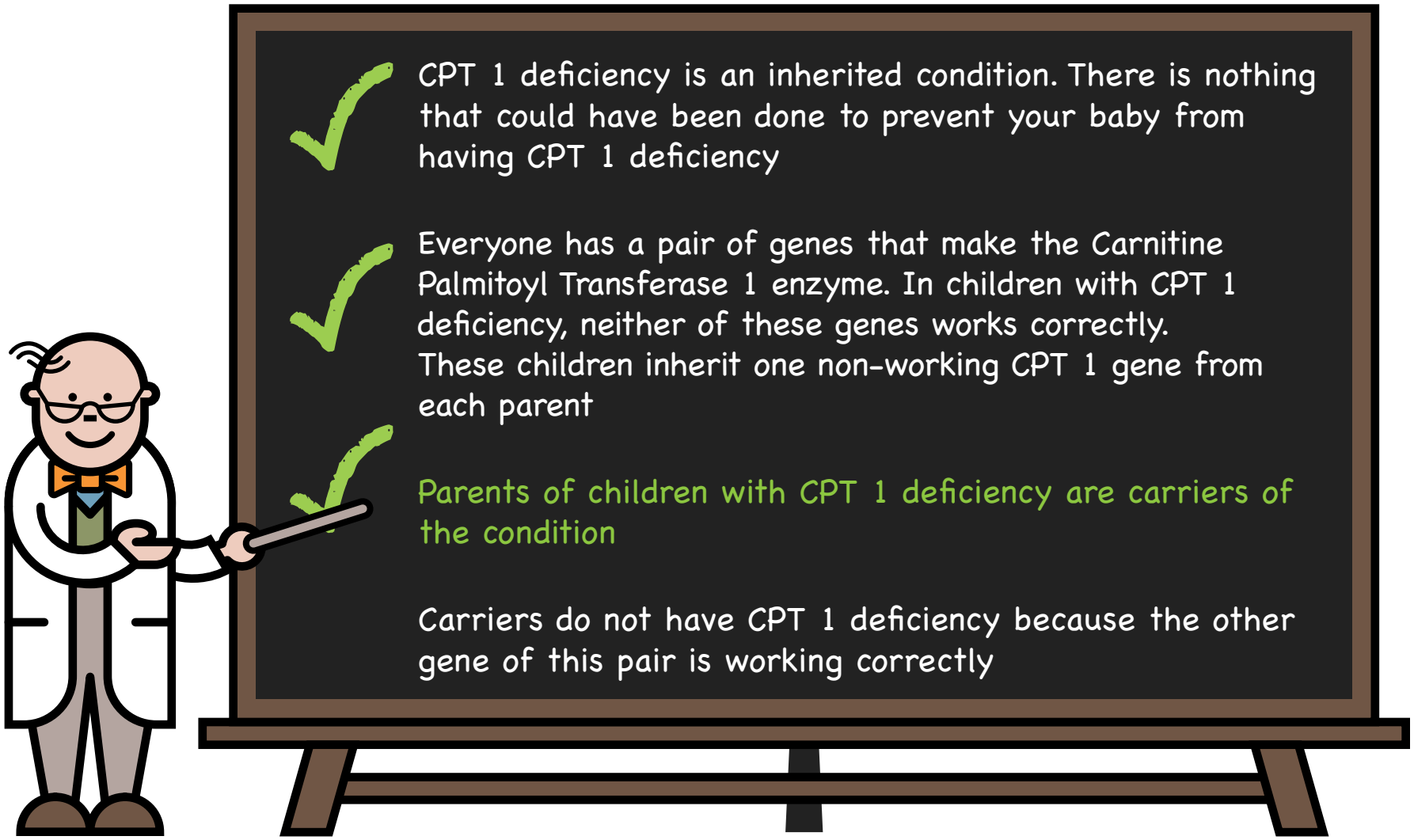

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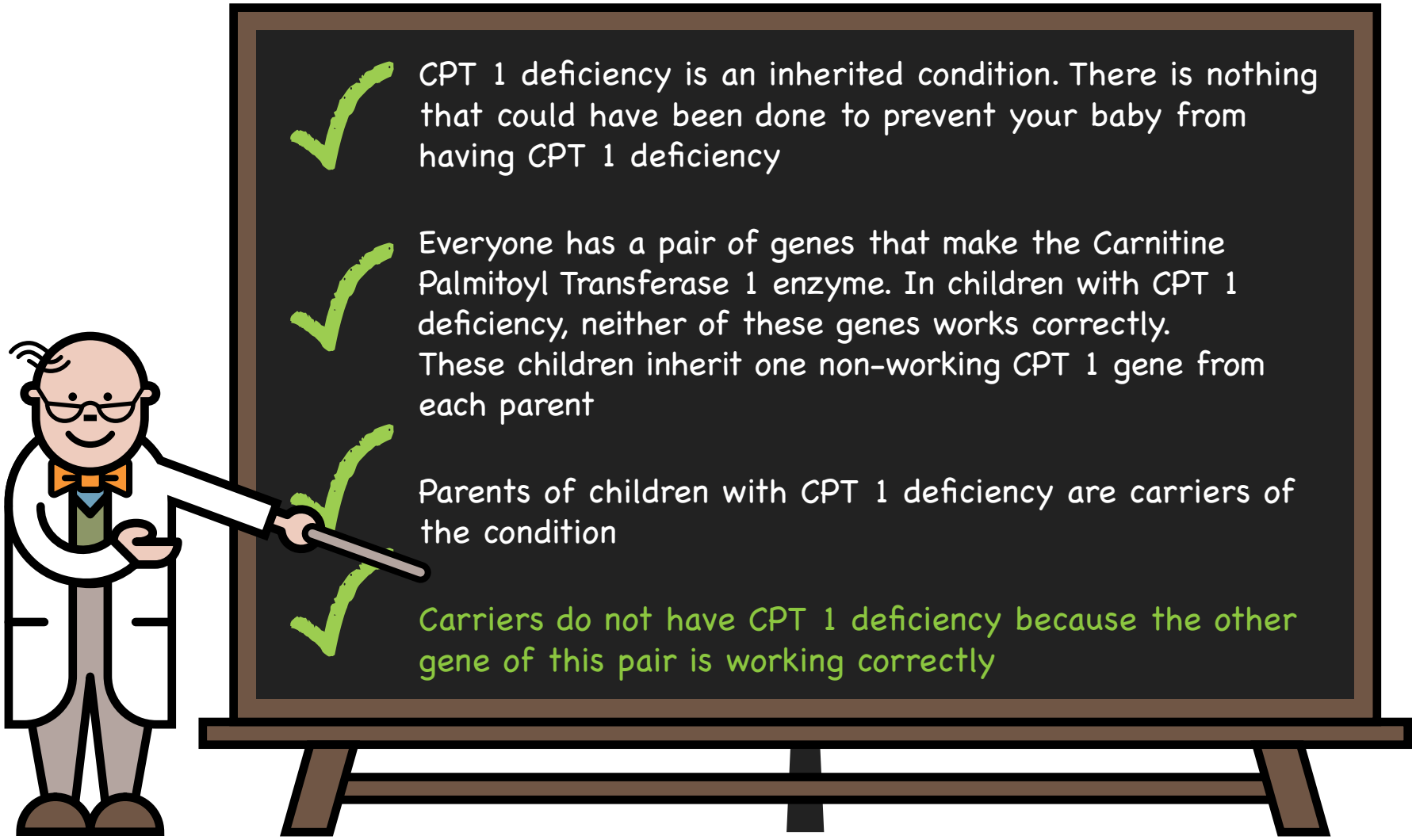
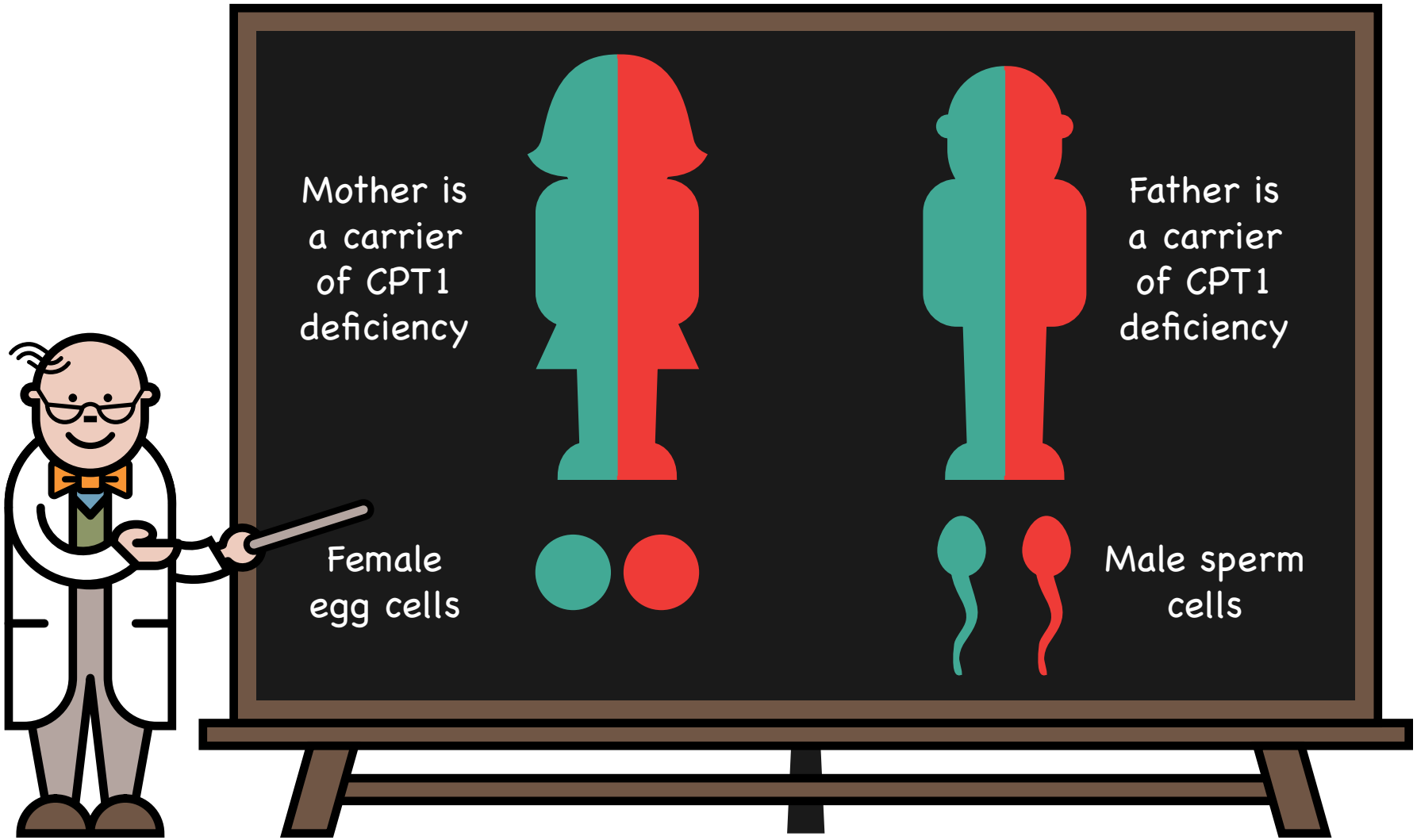
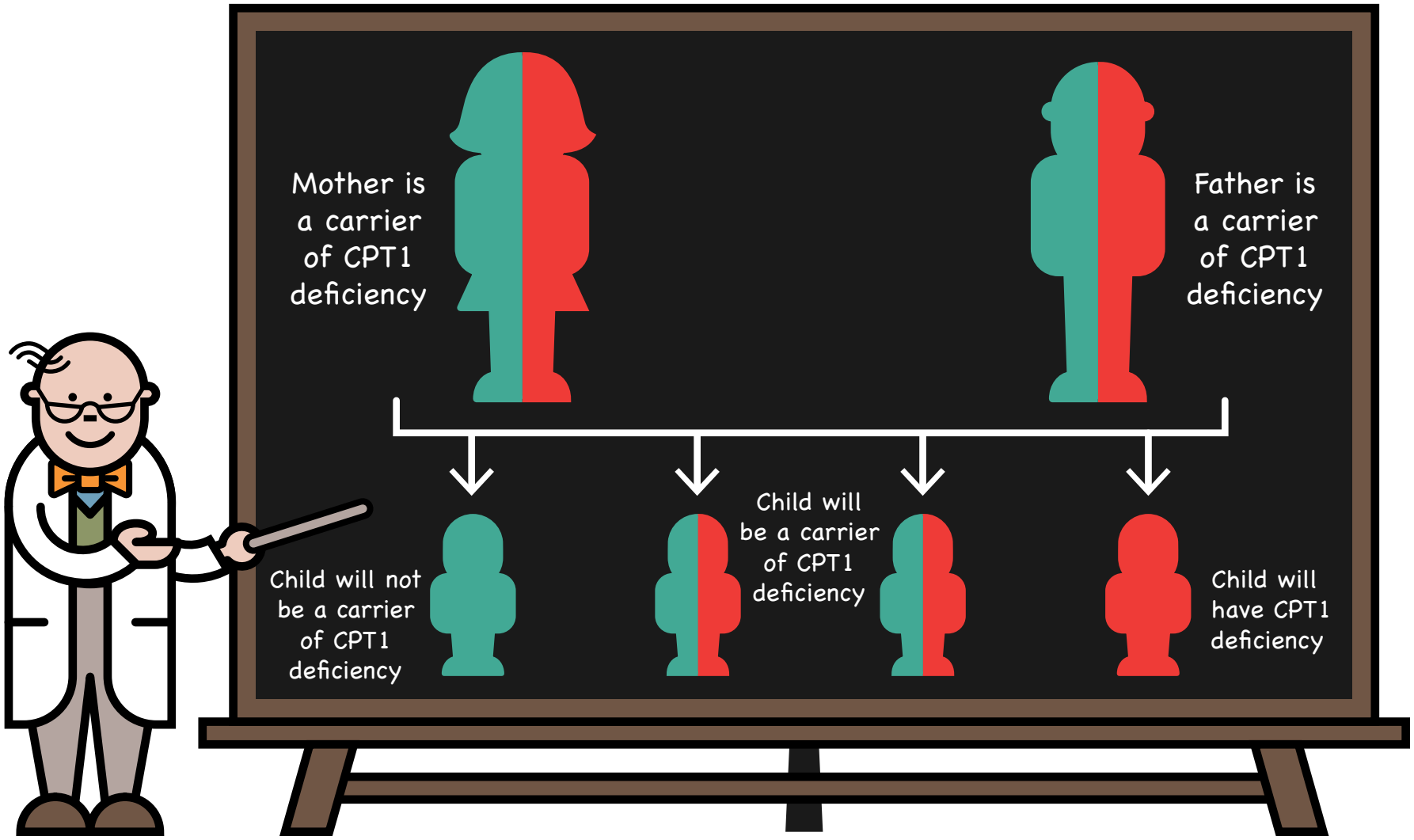


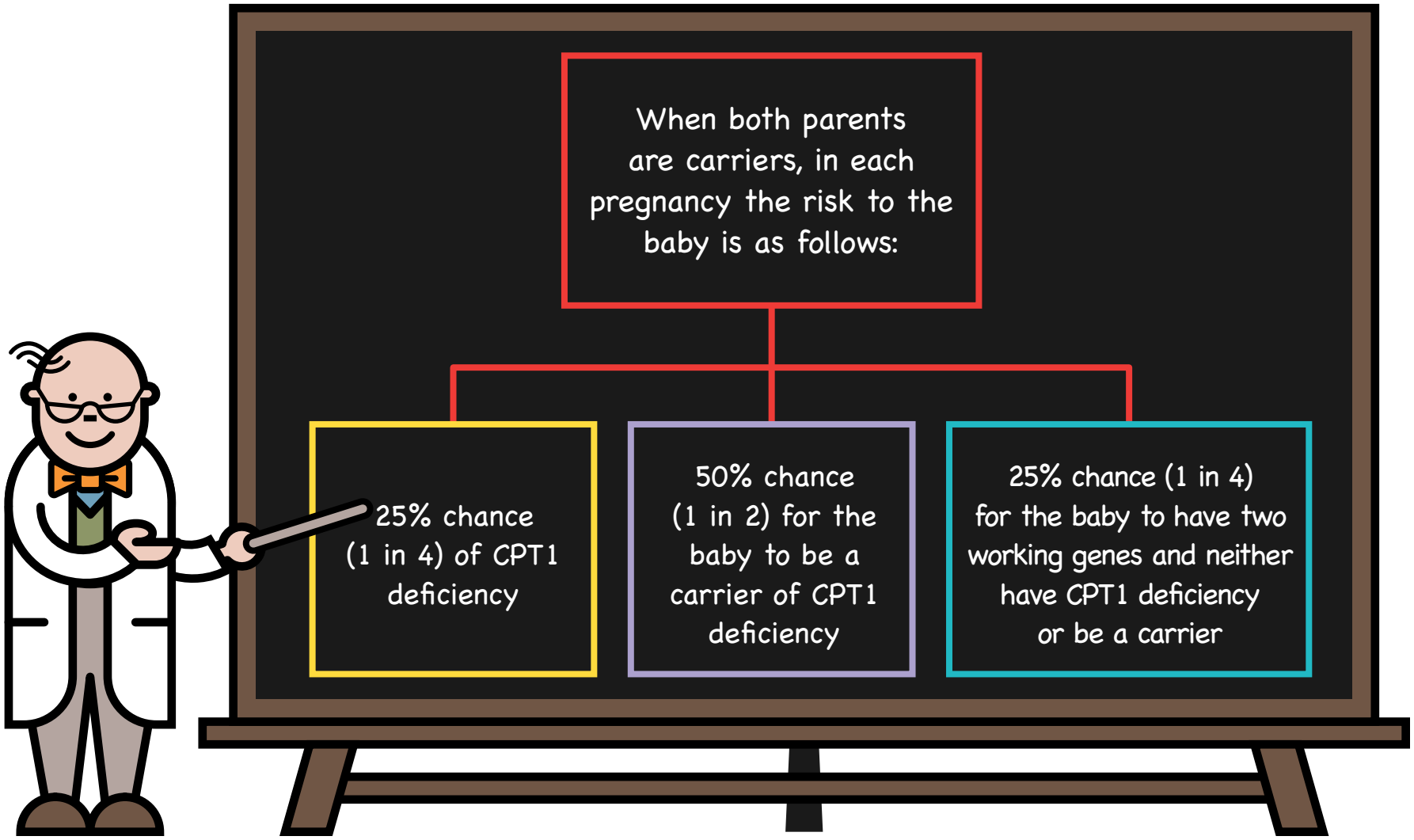
Inheritance – Autosomal recessive (carriers of CPT1 deficiency)



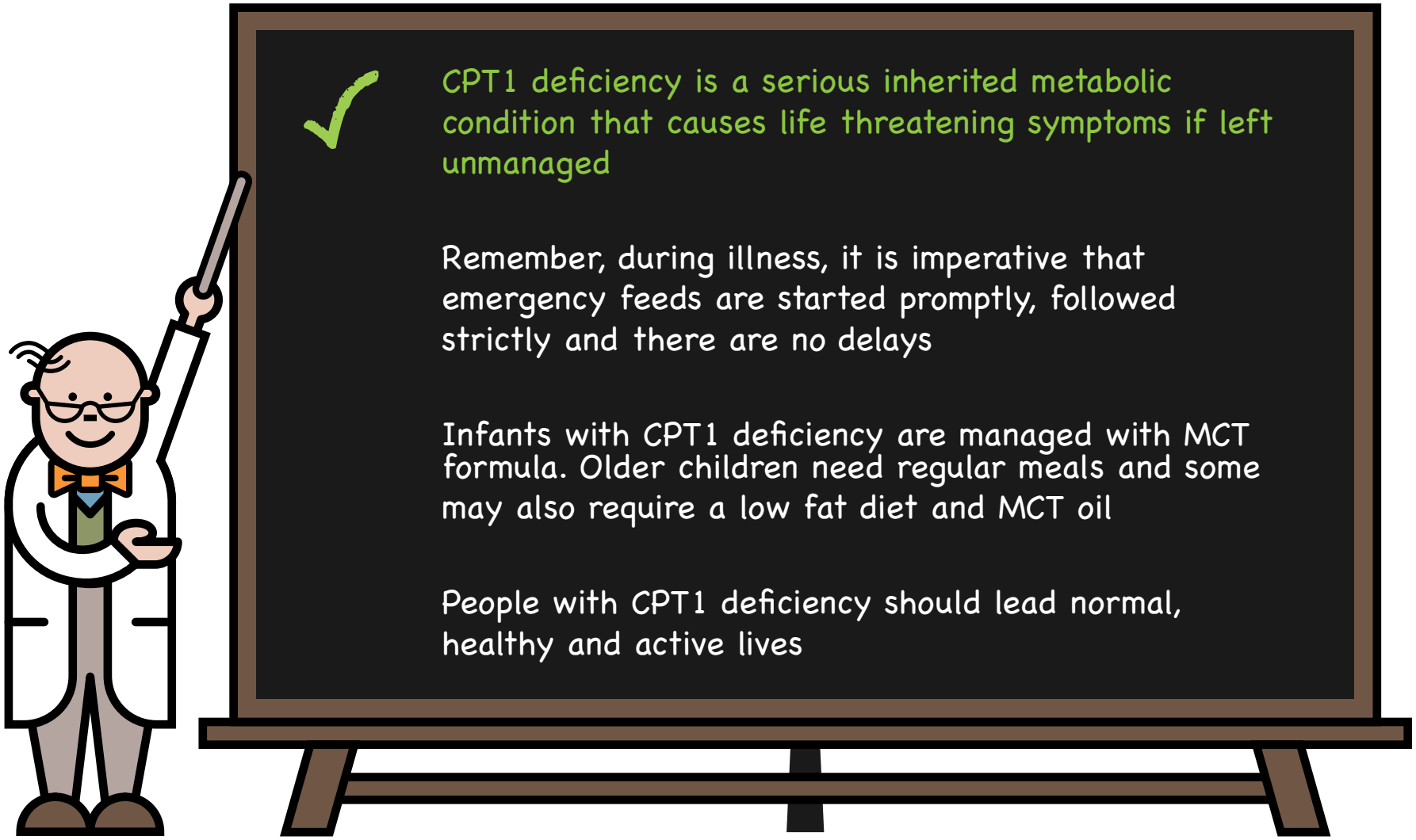
Inheritance – Autosomal recessive – possible combinations



Future pregnancies



Take home messages



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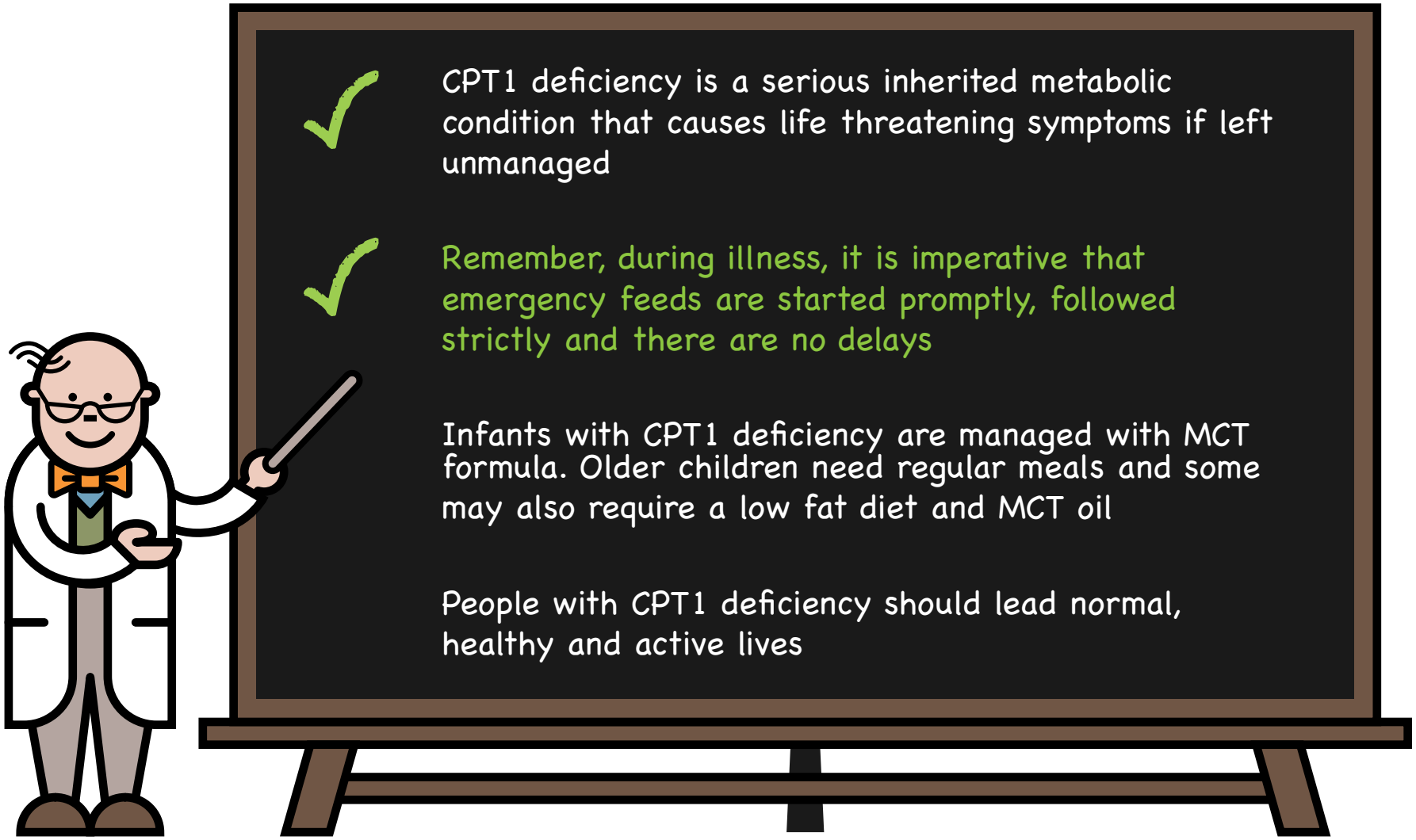

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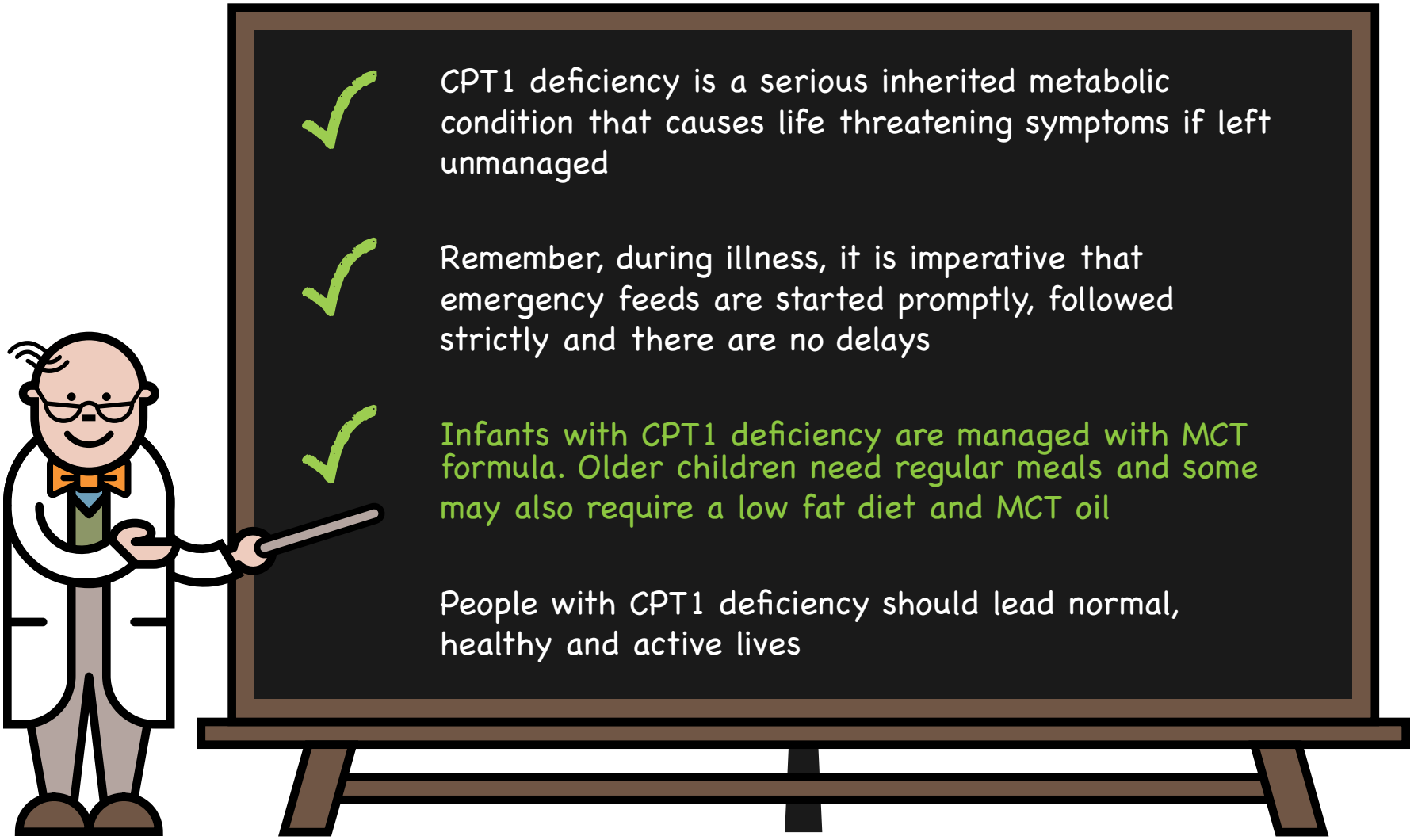

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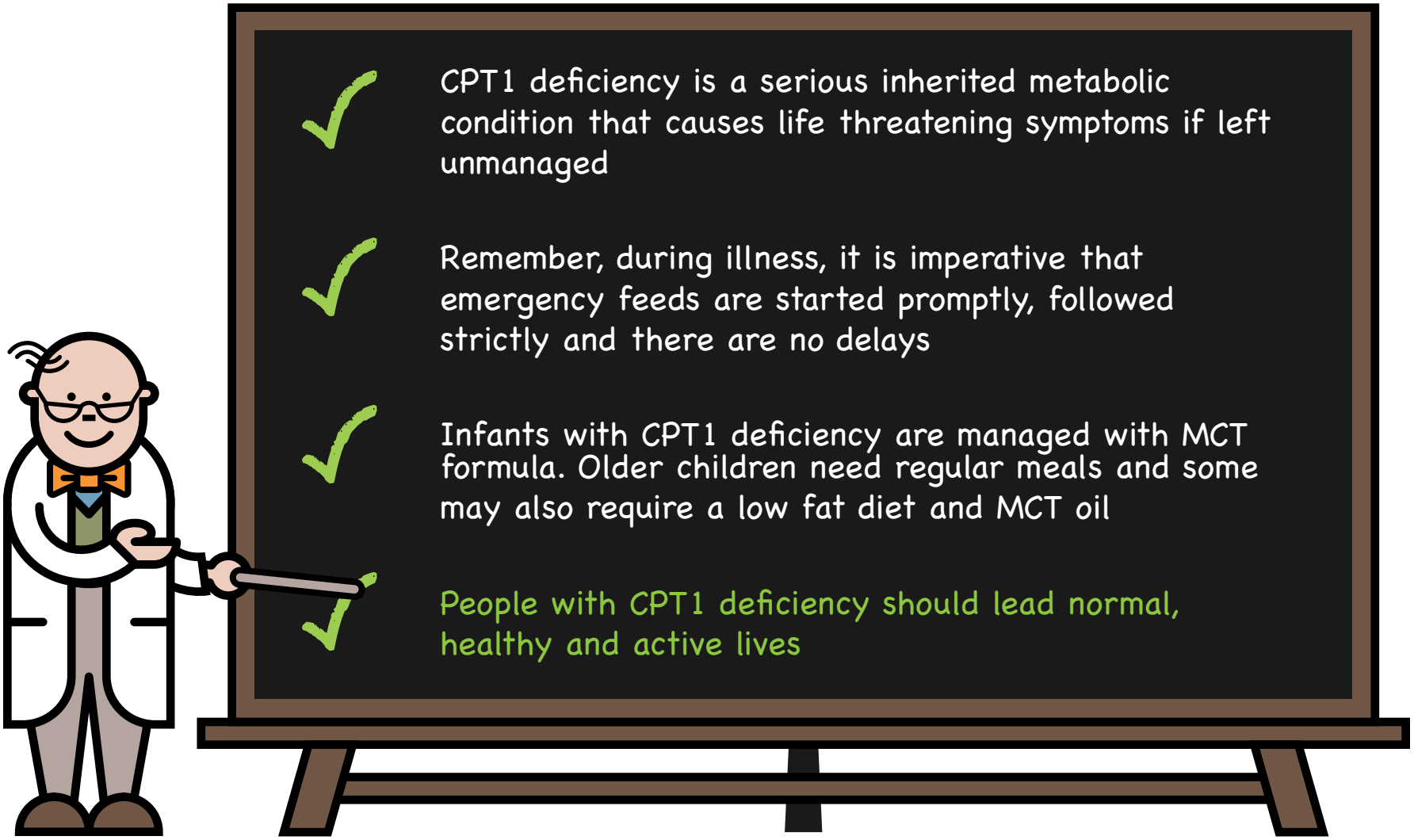
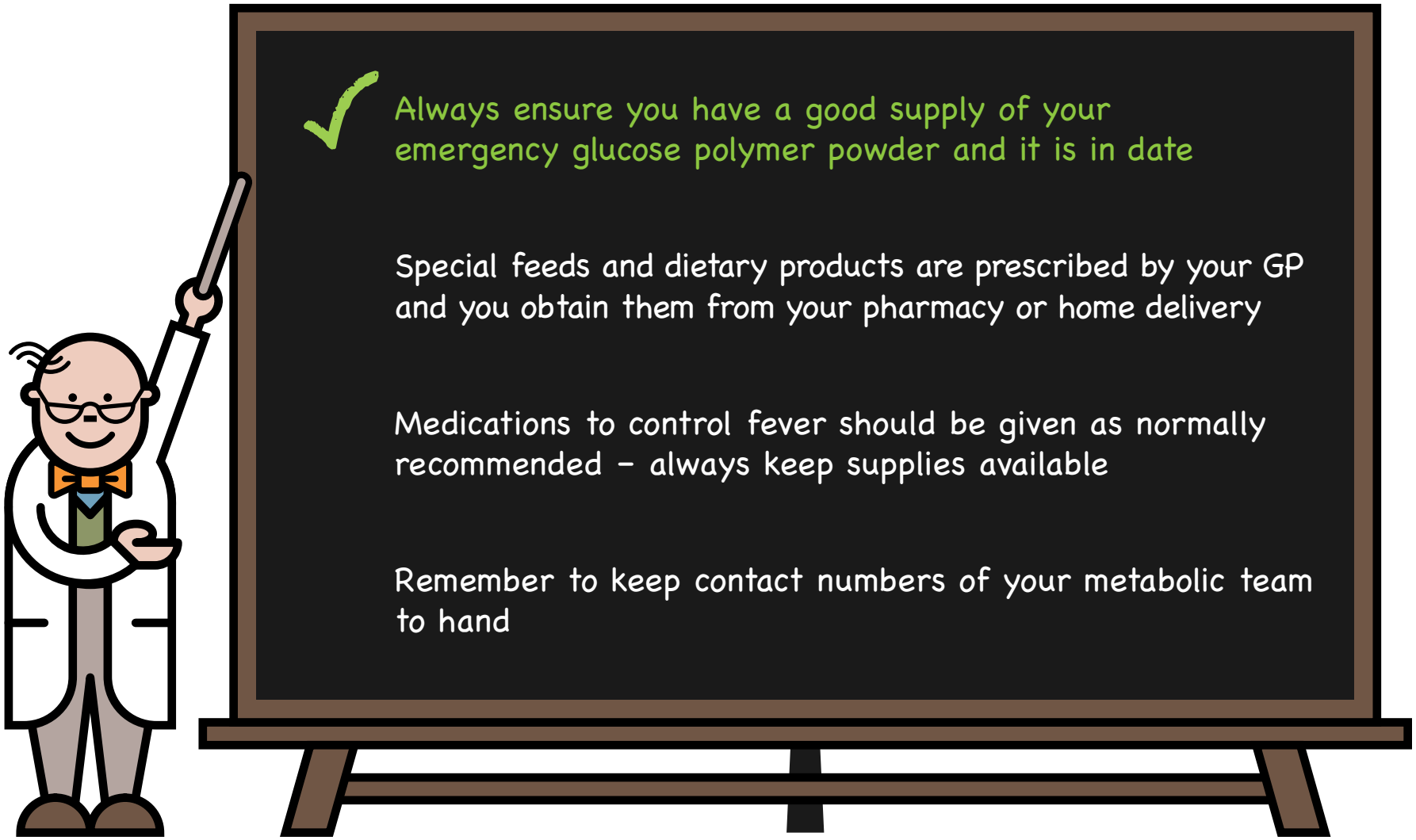


Helpful hints



Helpful hints

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Helpful hints

Who's who

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

BIMDG

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Our common fight.

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