TEMPLE

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

Arginase deficiency

ADAPTED BY THE DIETITIANS GROUP

BIMDG
British Inherited Metabolic Diseases Group

BASED ON THE ORIGINAL TEMPLE WRITTEN BY
BURGARD AND WENDEL

VERSION 2, NOVEMBER 2020

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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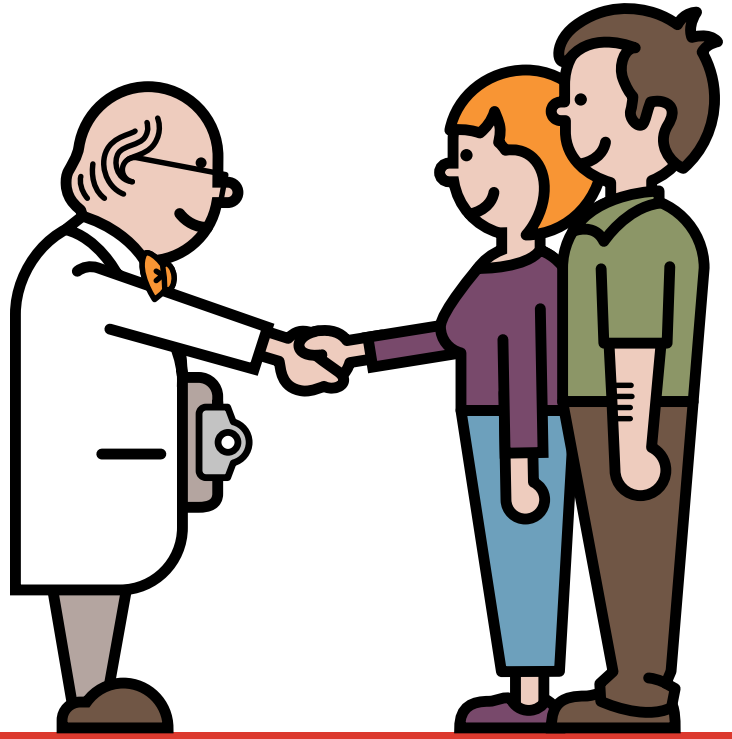
Arginase deficiency

Information for families following a new diagnosis



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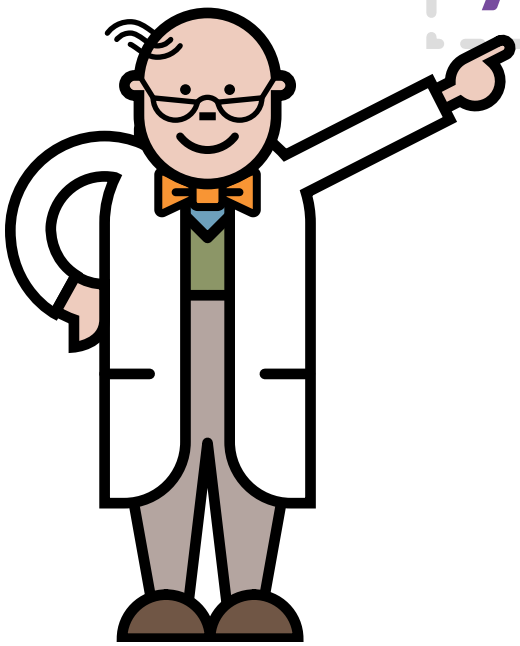
What is Arginase deficiency?

It is an inherited metabolic condition.



Argi nase deficiency

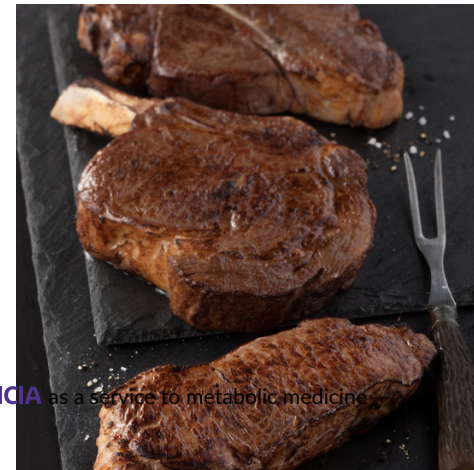
Arginase deficiency

What is protein?

Many foods contain protein.

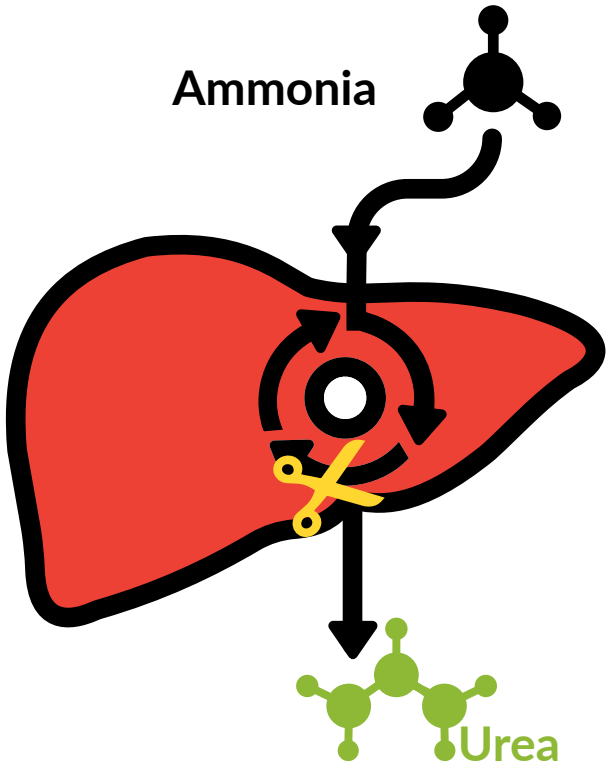
The body needs protein for growth and repair.

Many people eat more protein than the body needs.



How do we remove waste protein from the body?

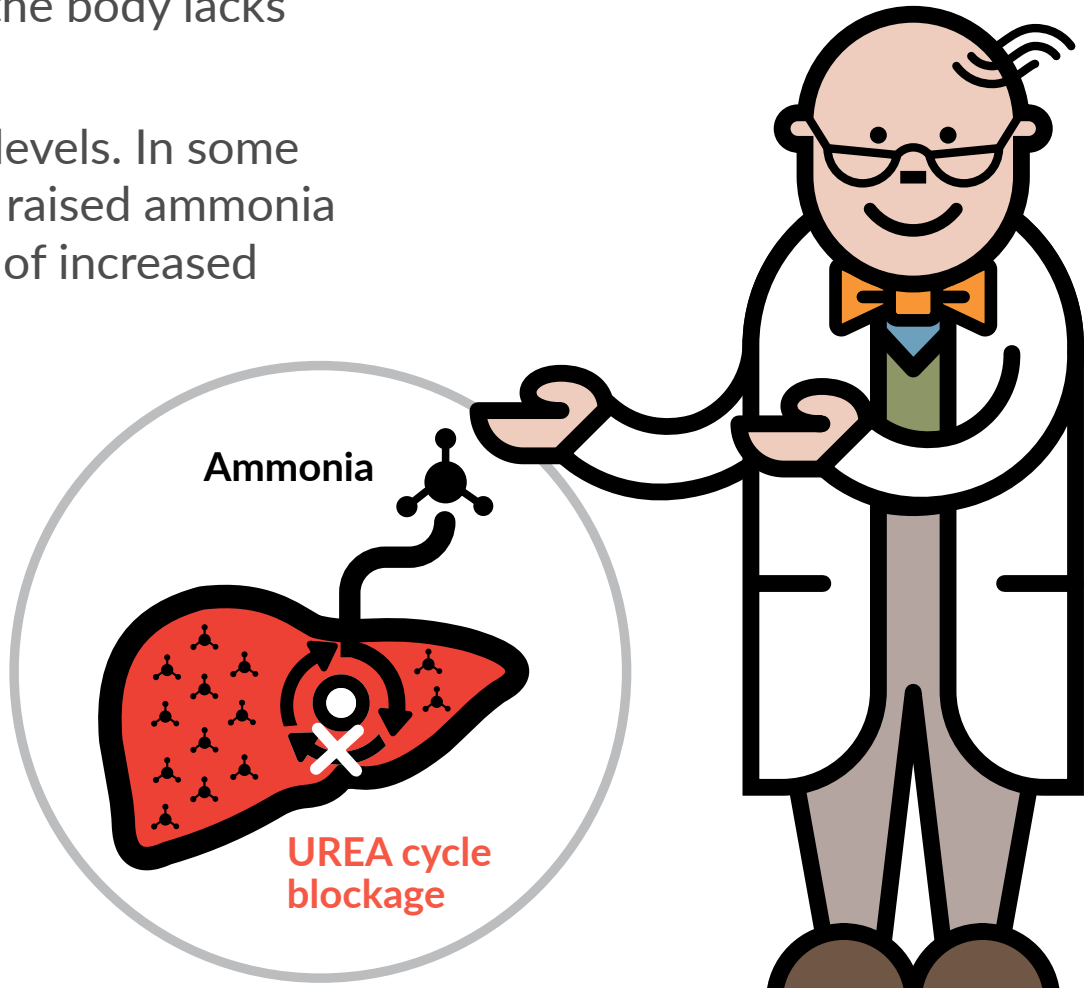
- First, the body converts waste protein to a toxic chemical called **ammonia**
- Ammonia is then converted into a non-toxic chemical **(urea)** in the liver
- This process occurs via the **urea cycle**
- In the urea cycle, several steps have to take place. Each step needs an enzyme (like chemical scissors) for it to work
- Arginine is formed in the urea cycle and then broken down to release urea. The enzyme that **breaks** down arginine is called Arginase 1.



What happens in Arginase deficiency?

In the Arginise deficiency, the body lacks the enzyme **arginase 1**.

This leads to high arginine levels. In some patients, it can also lead to raised ammonia levels, particularly at times of increased protein breakdown.



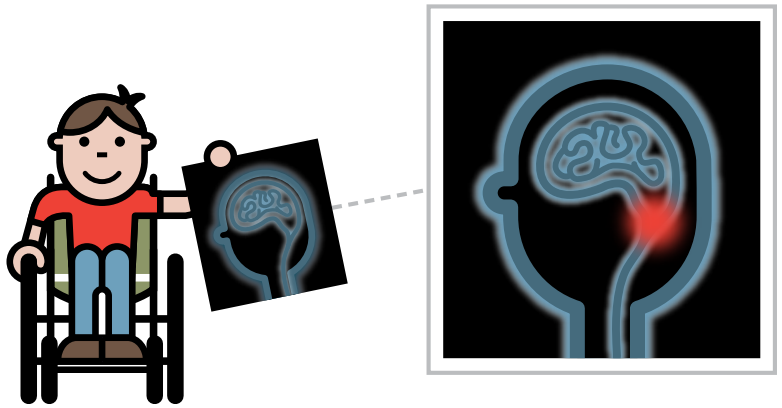
What are the signs of Arginase deficiency?

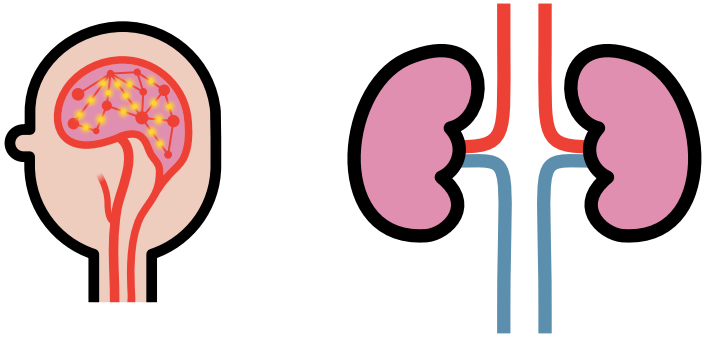
Arginine is one of the building blocks from which protein is made, so we need some arginine in our blood. It is harmful, however, if the levels are much too high.

Children with arginase deficiency may get:

- Stiff legs (spasticity)
- Learning difficulties
- Seizures
- Poor growth

Arginase deficiency sometimes leads to high ammonia levels, causing the patient to become drowsy or even comatose. High ammonia levels are most likely during illnesses.





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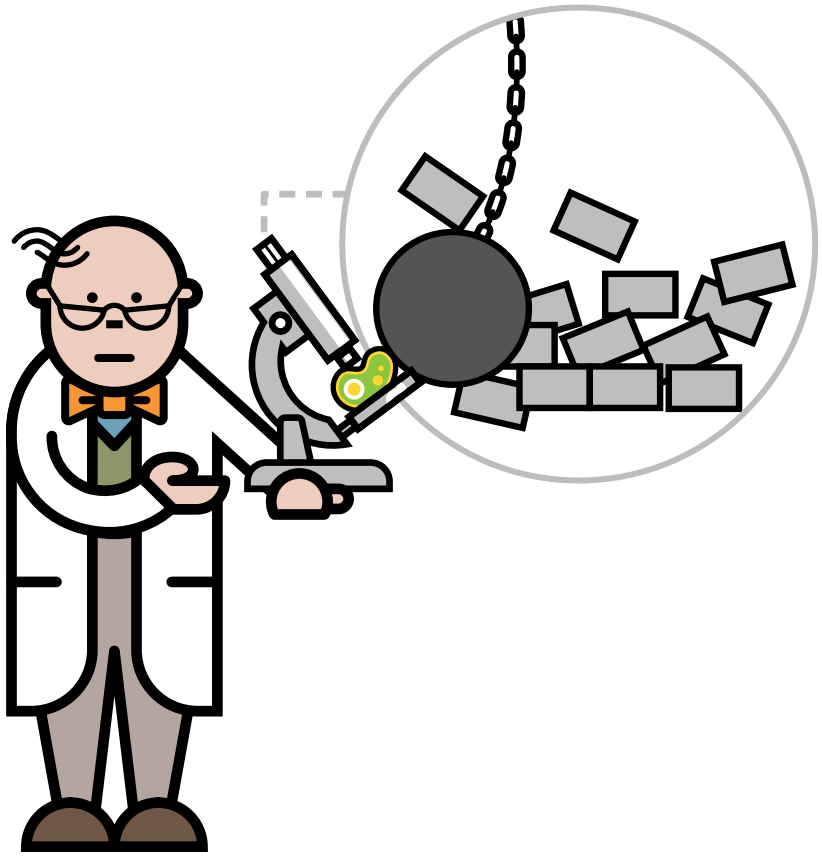
When does Arginase deficiency cause high ammonia levels?

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.

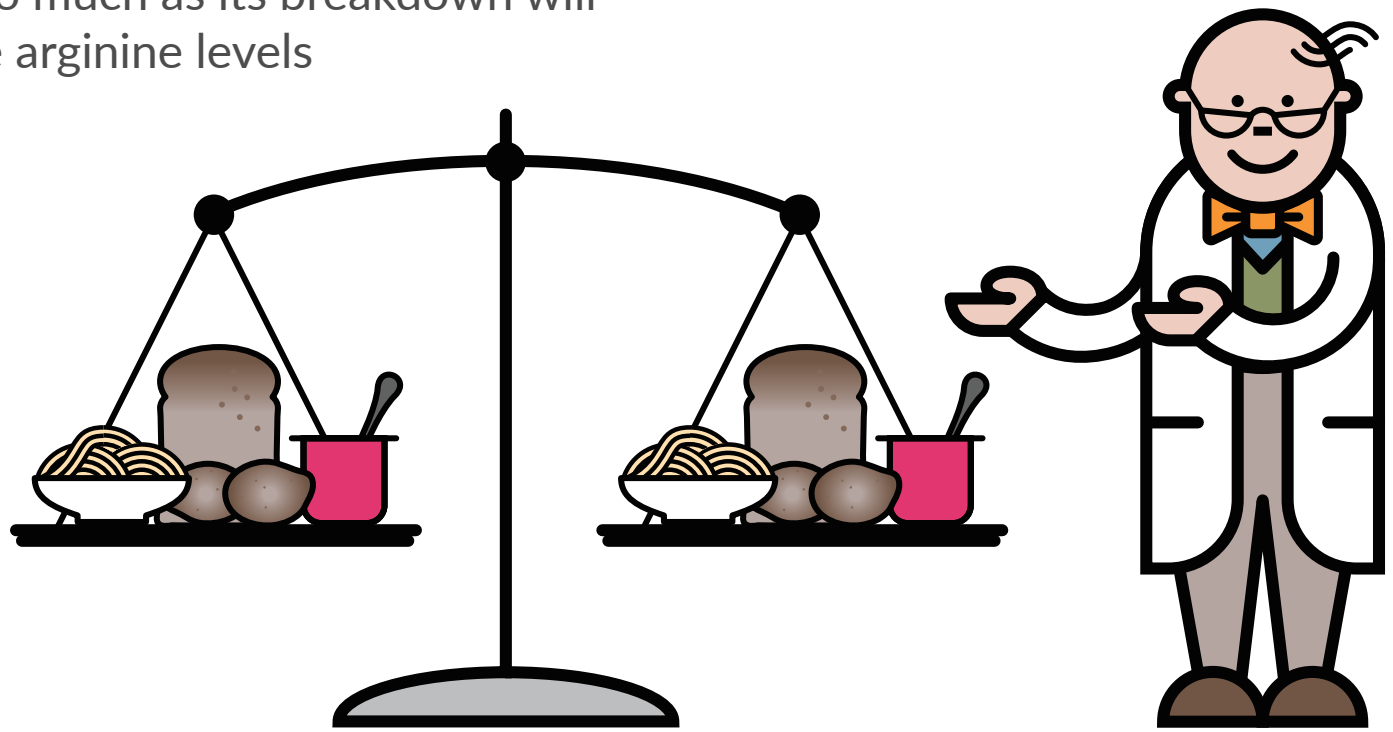
This causes **catabolism** which is a break down of body protein and can lead to a metabolic crisis.



Protein balance is needed in Arginase deficiency

In **Arginase deficiency** it is important that enough protein is given to grow

...but not too much as its breakdown will increase the arginine levels

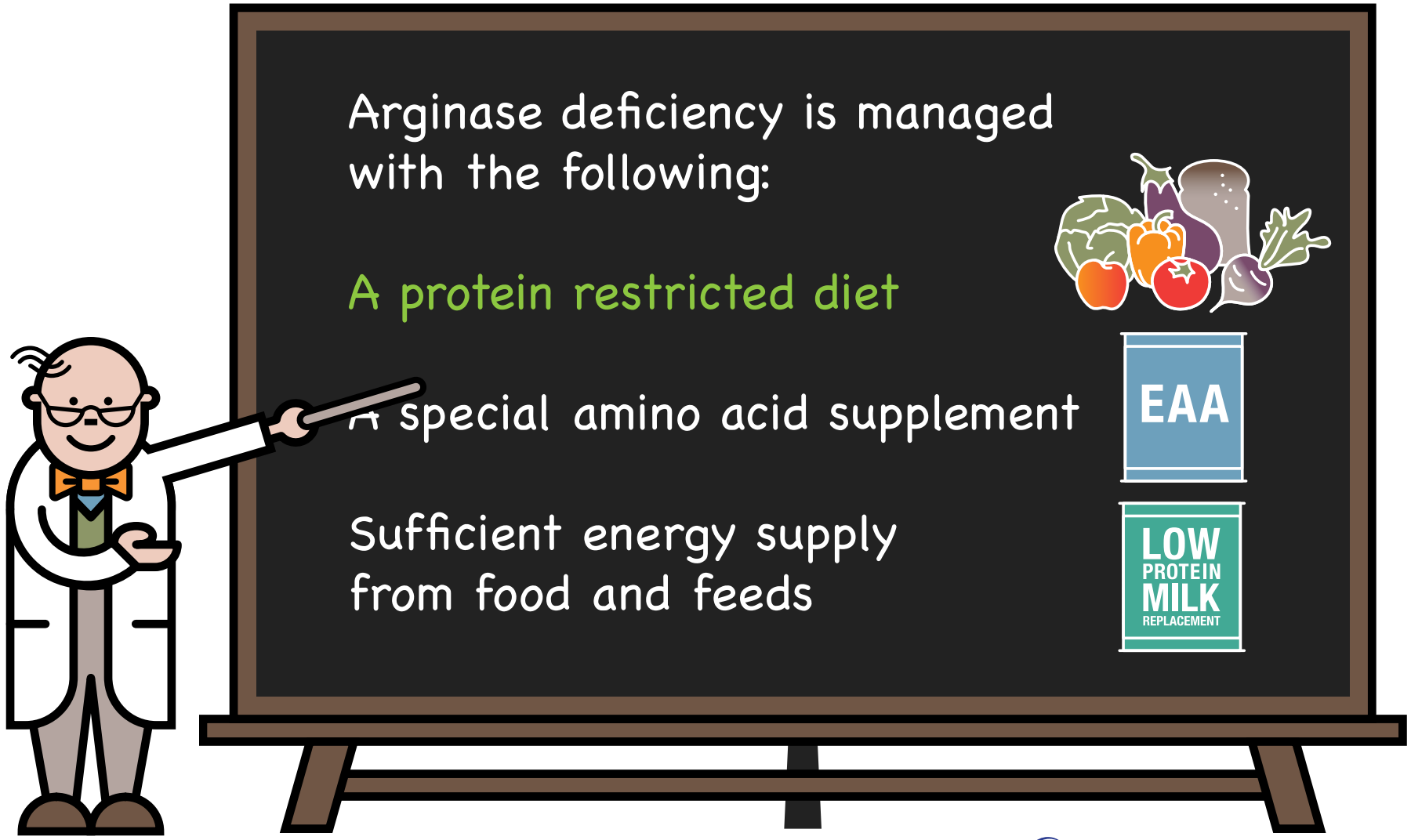


How is Arginase deficiency diagnosed?

The diagnosis is suspected in a patient with high arginine levels in the blood.

The diagnosis is confirmed by finding the mutation in the ARG1 gene .

How is Arginase deficiency managed day to day?



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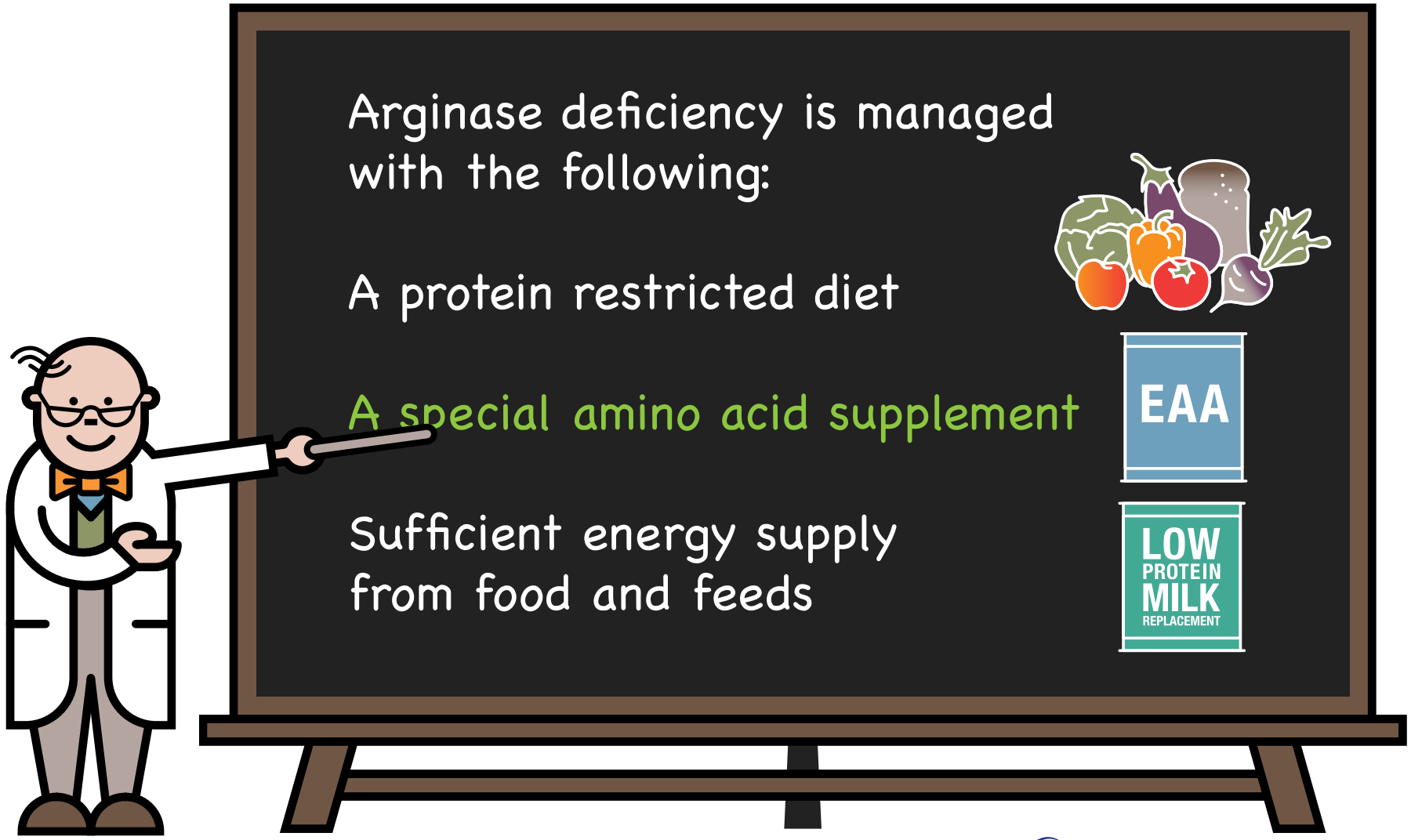

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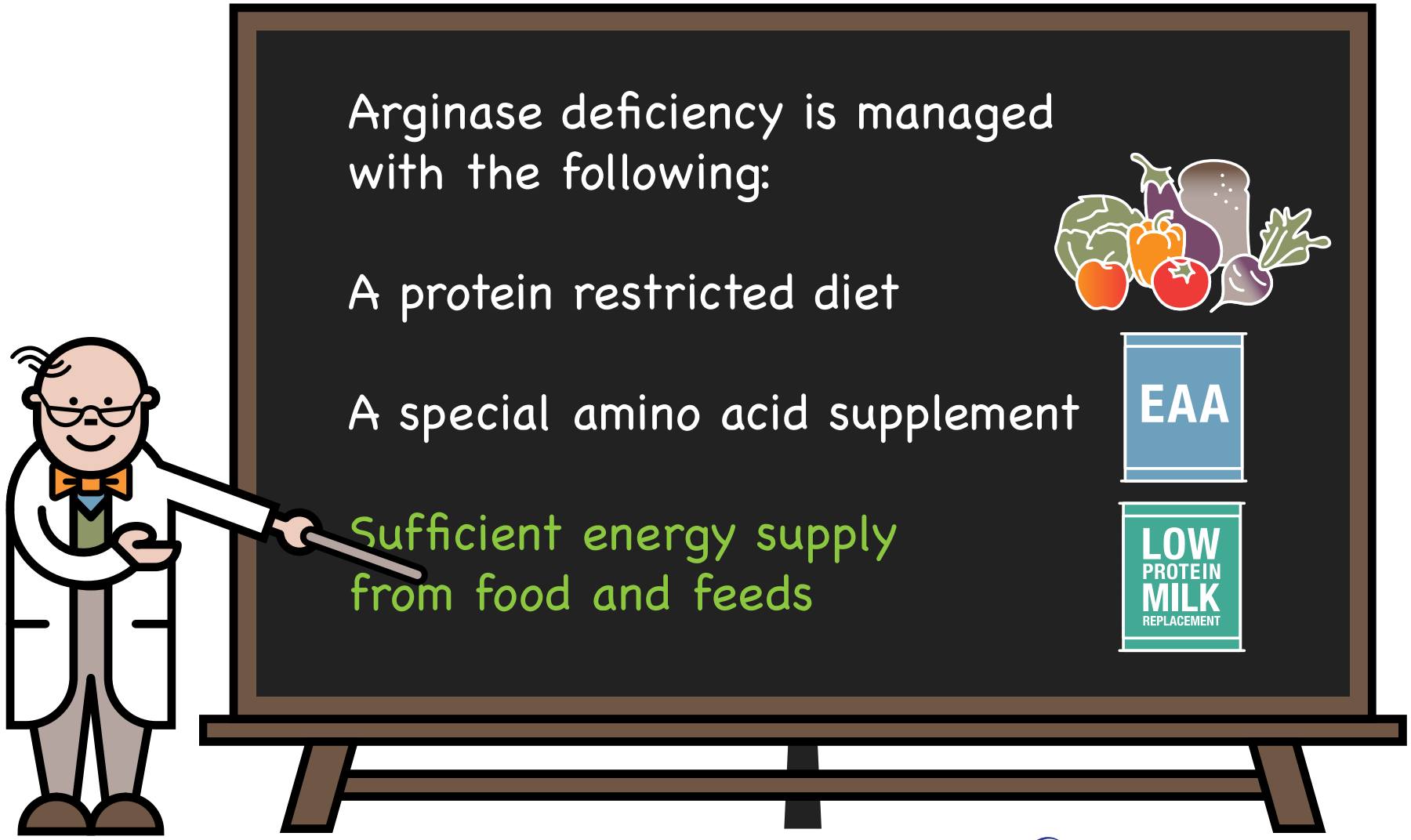

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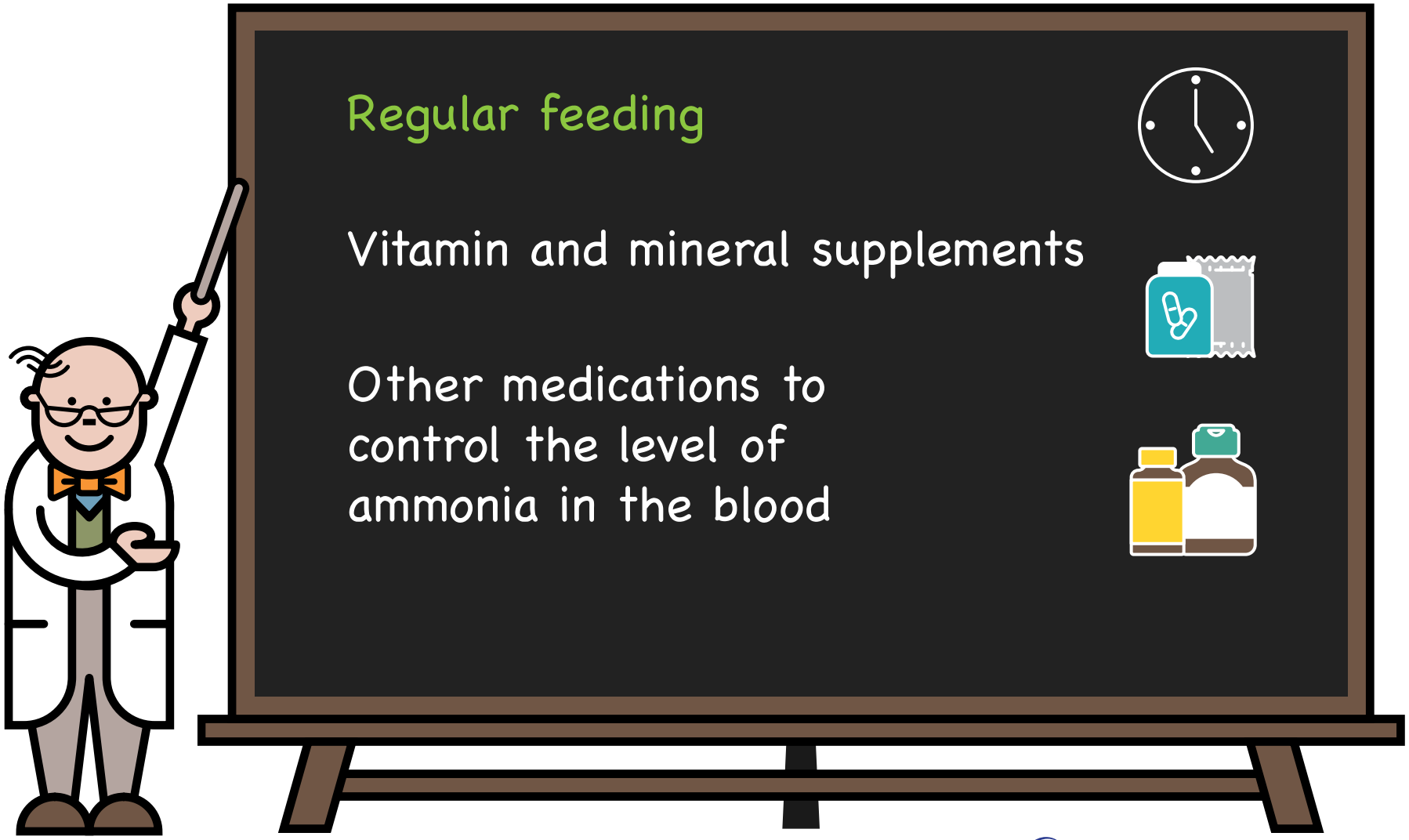

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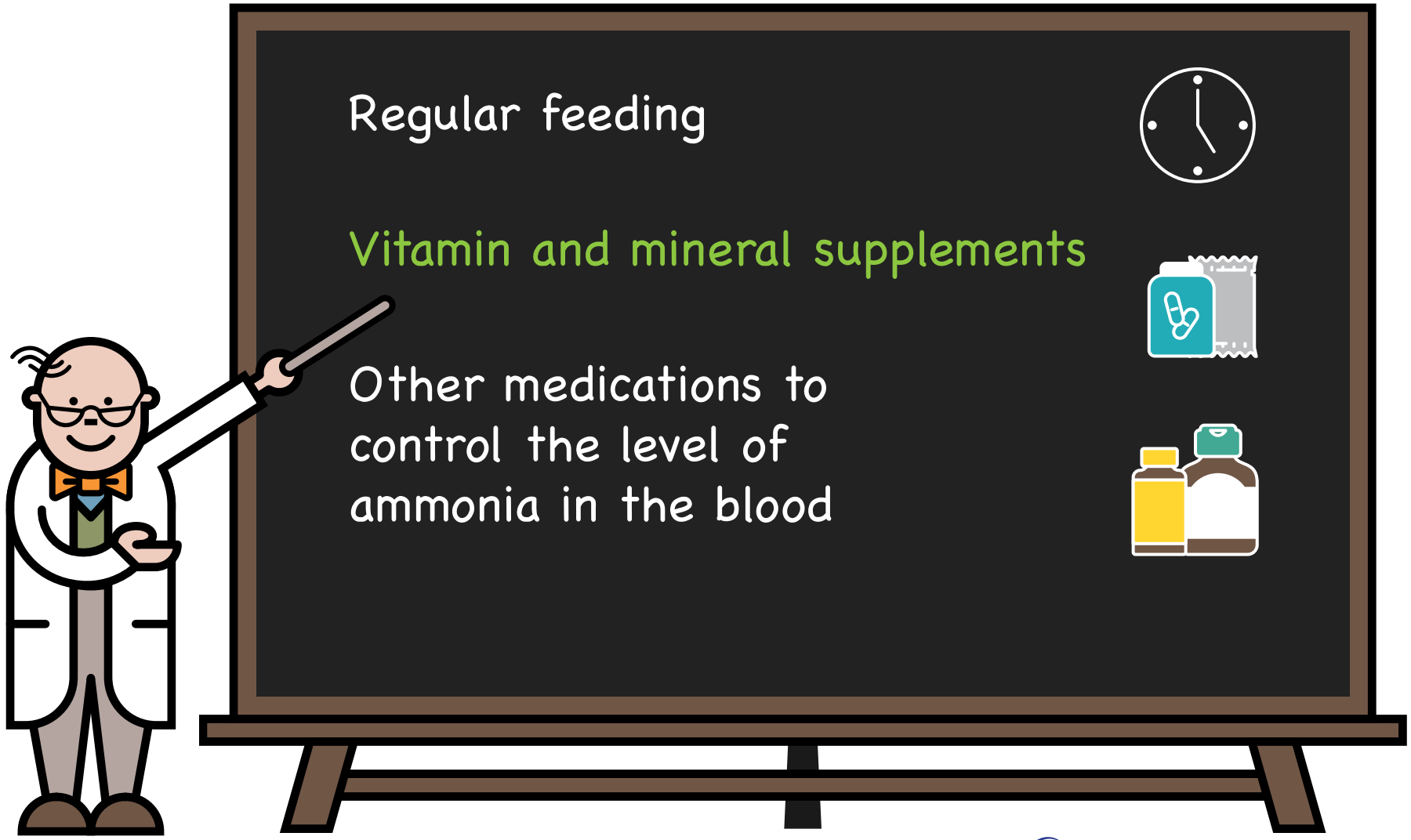

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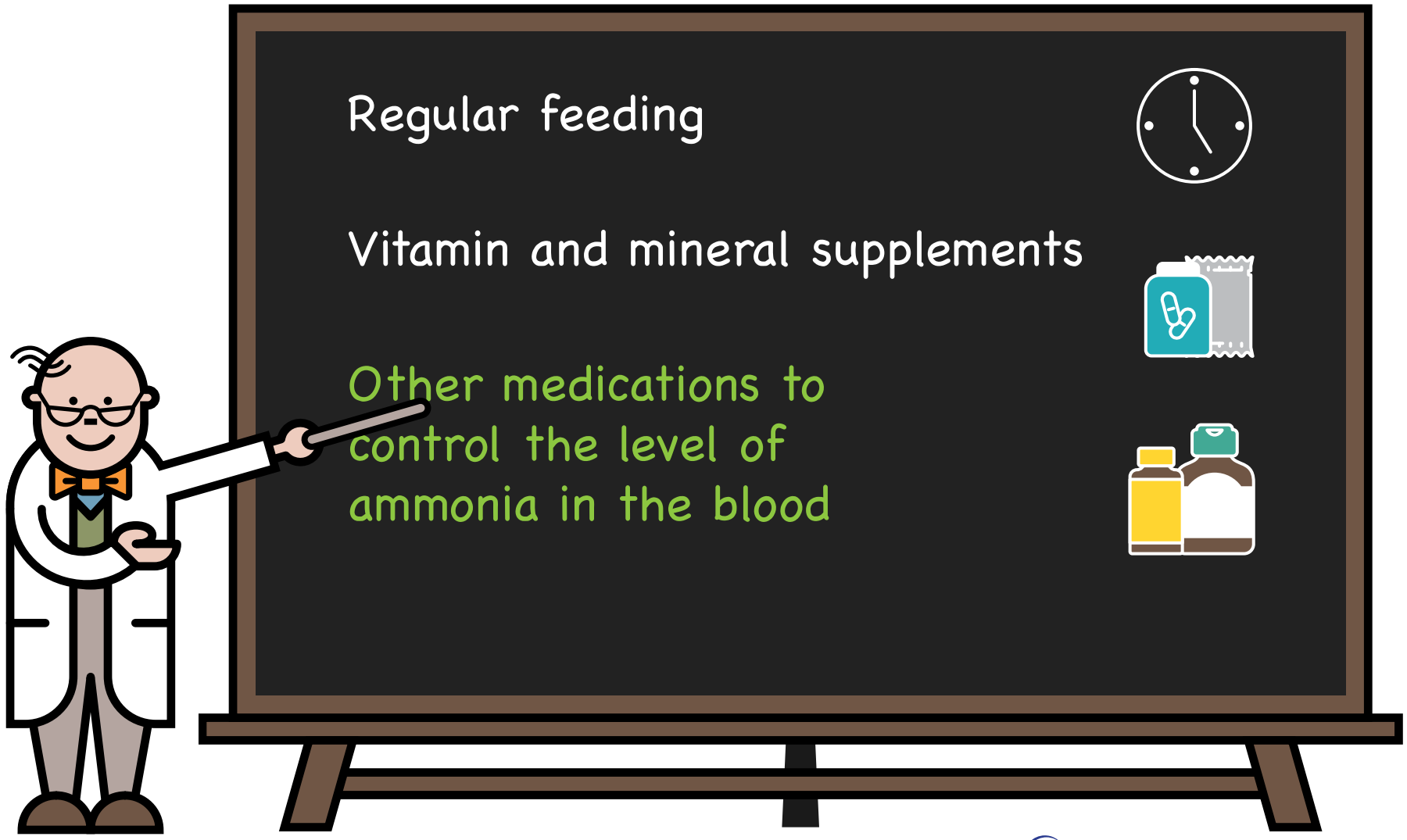
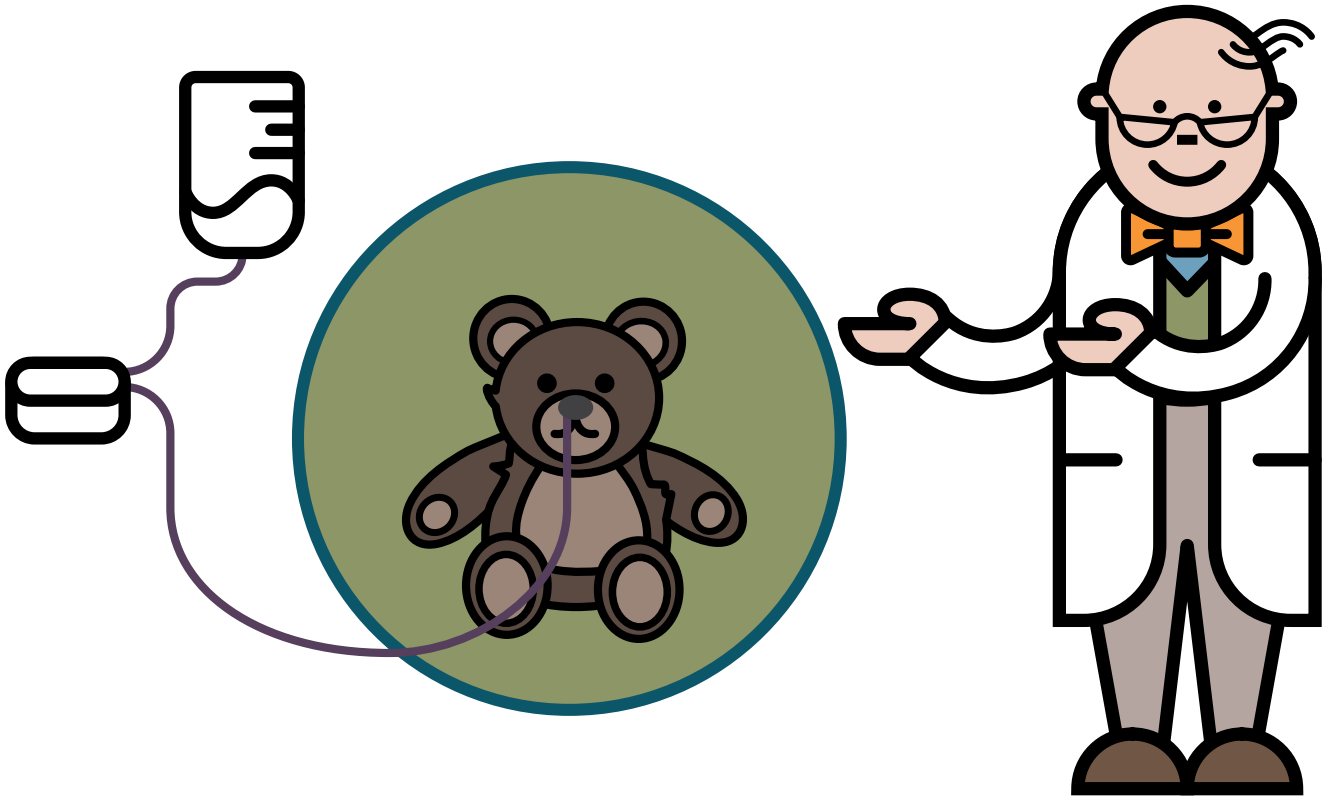


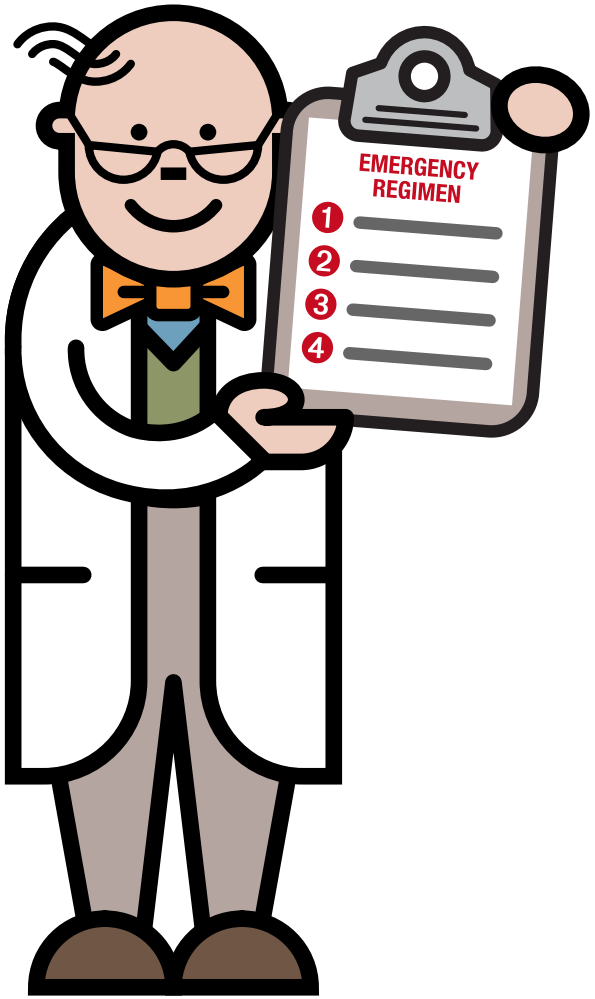
Is tube feeding needed?

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



How is Arginase deficiency managed during illness?

- During any childhood illness, an emergency regimen is given
- This is to avoid a lack of energy supply and build-up of ammonia



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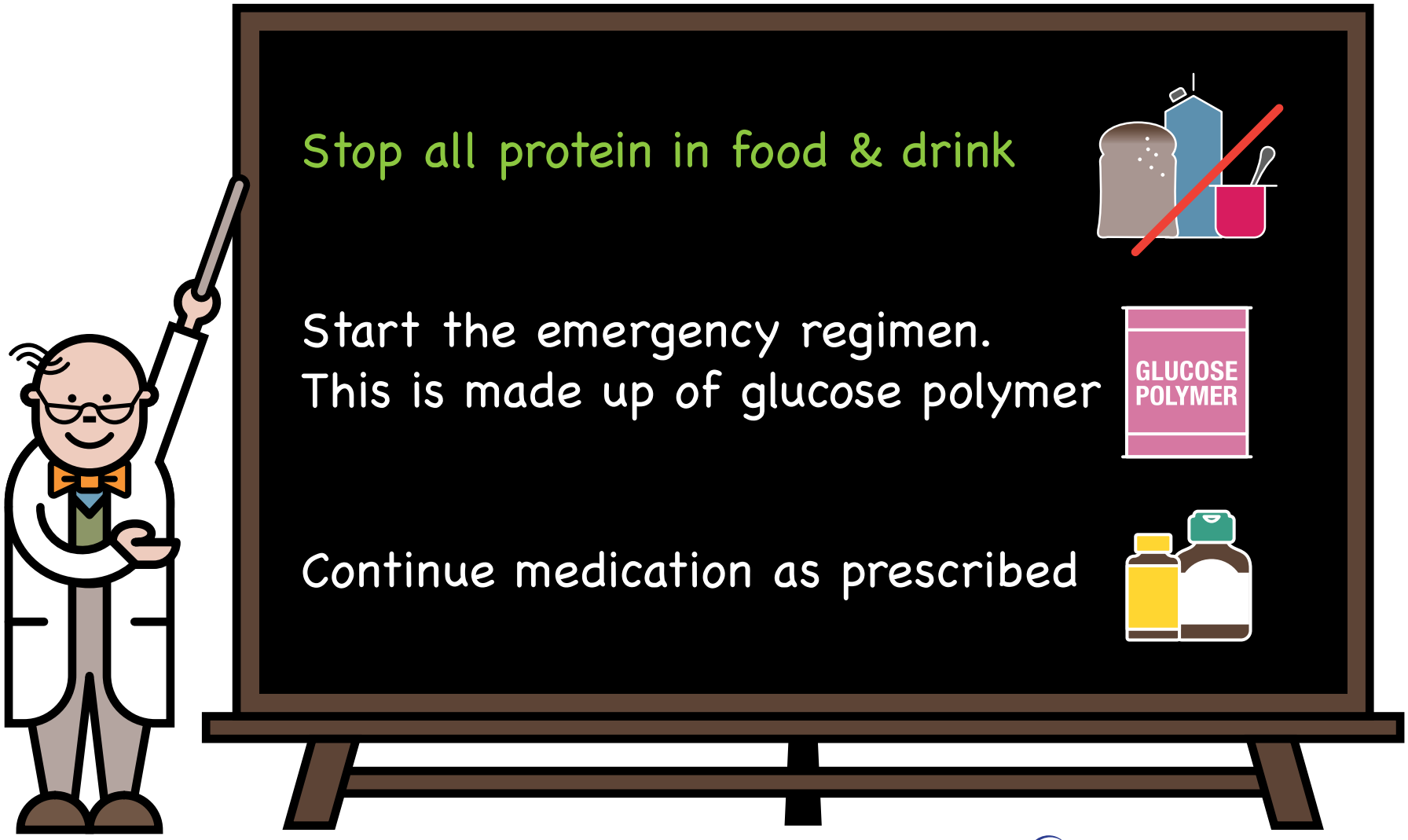

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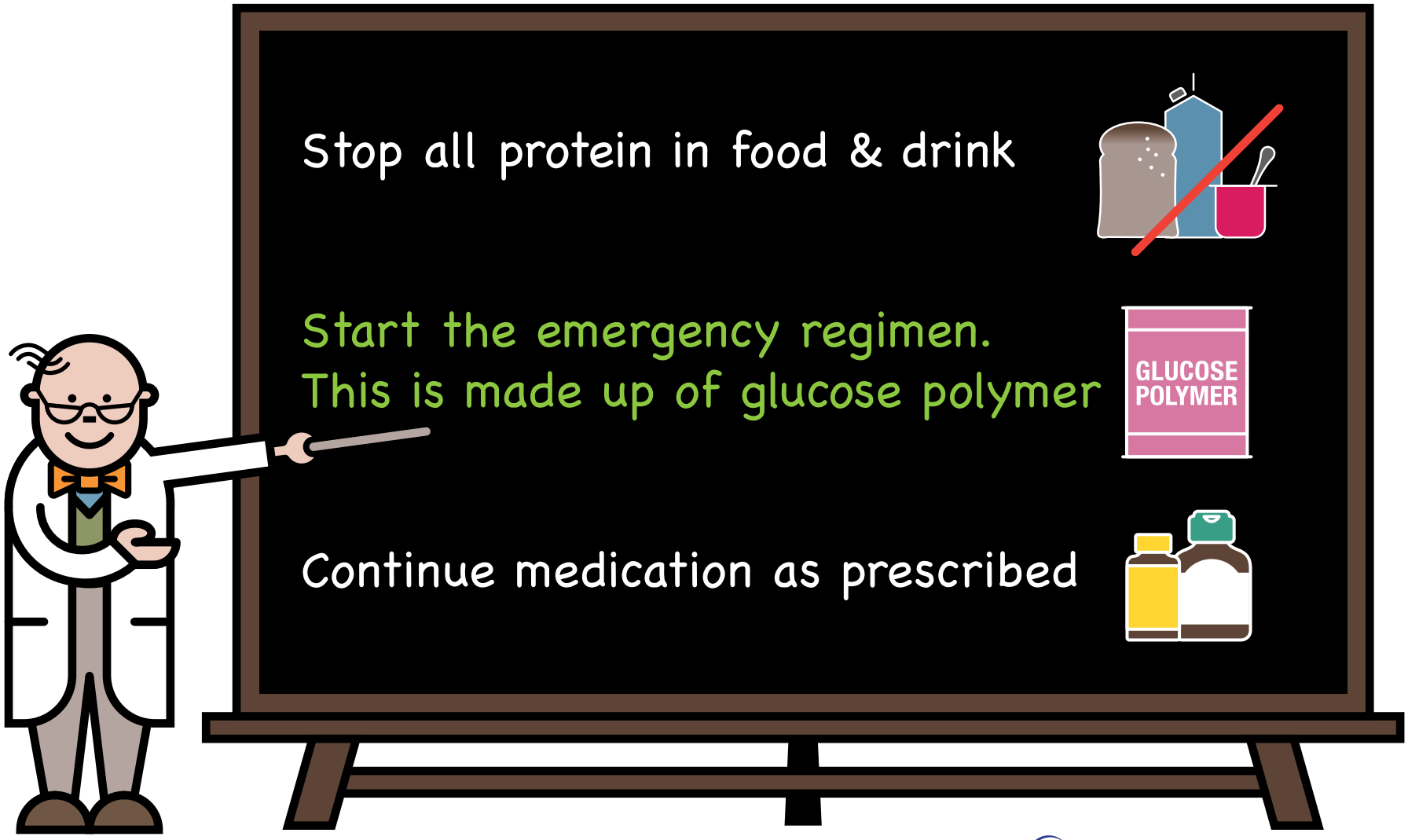

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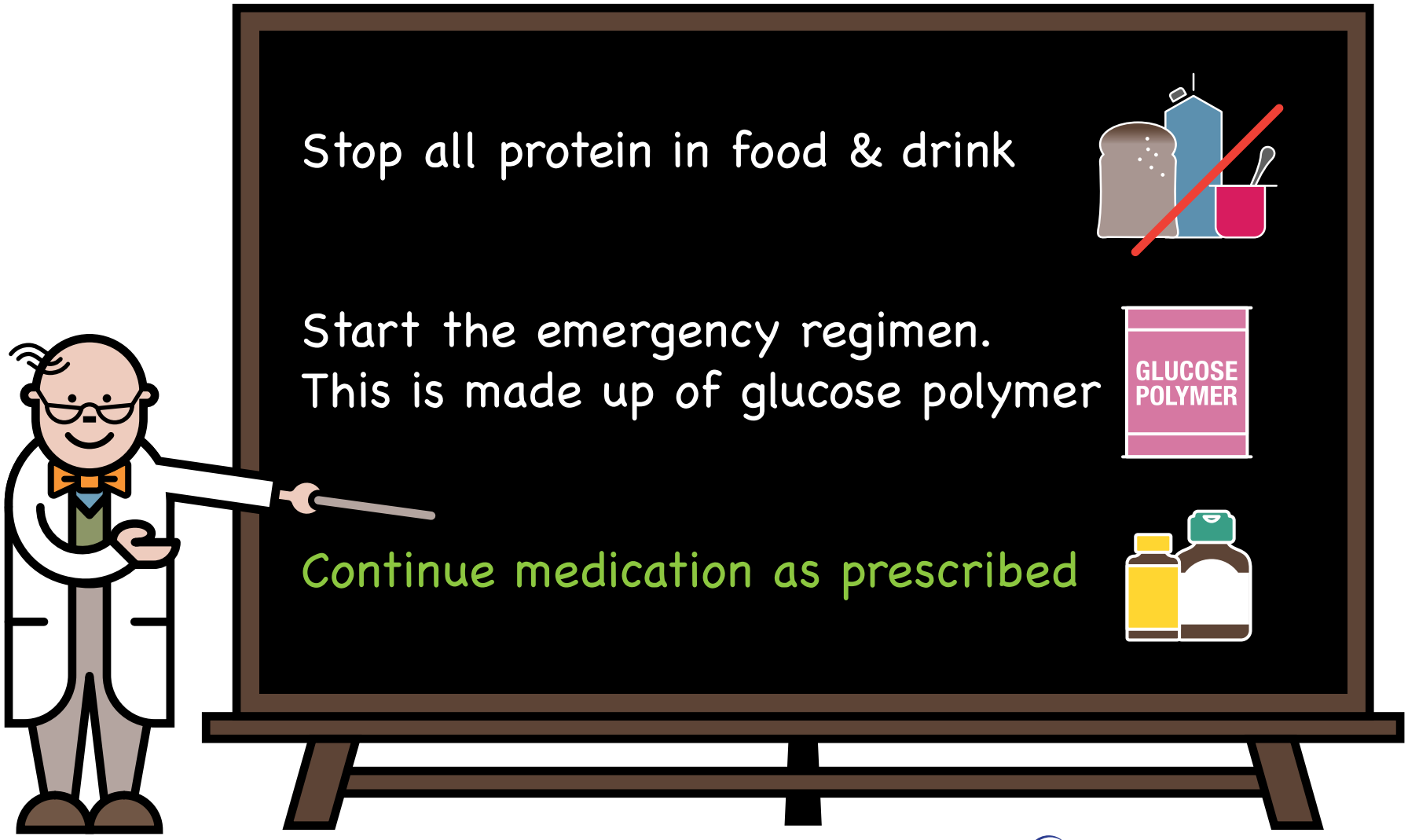
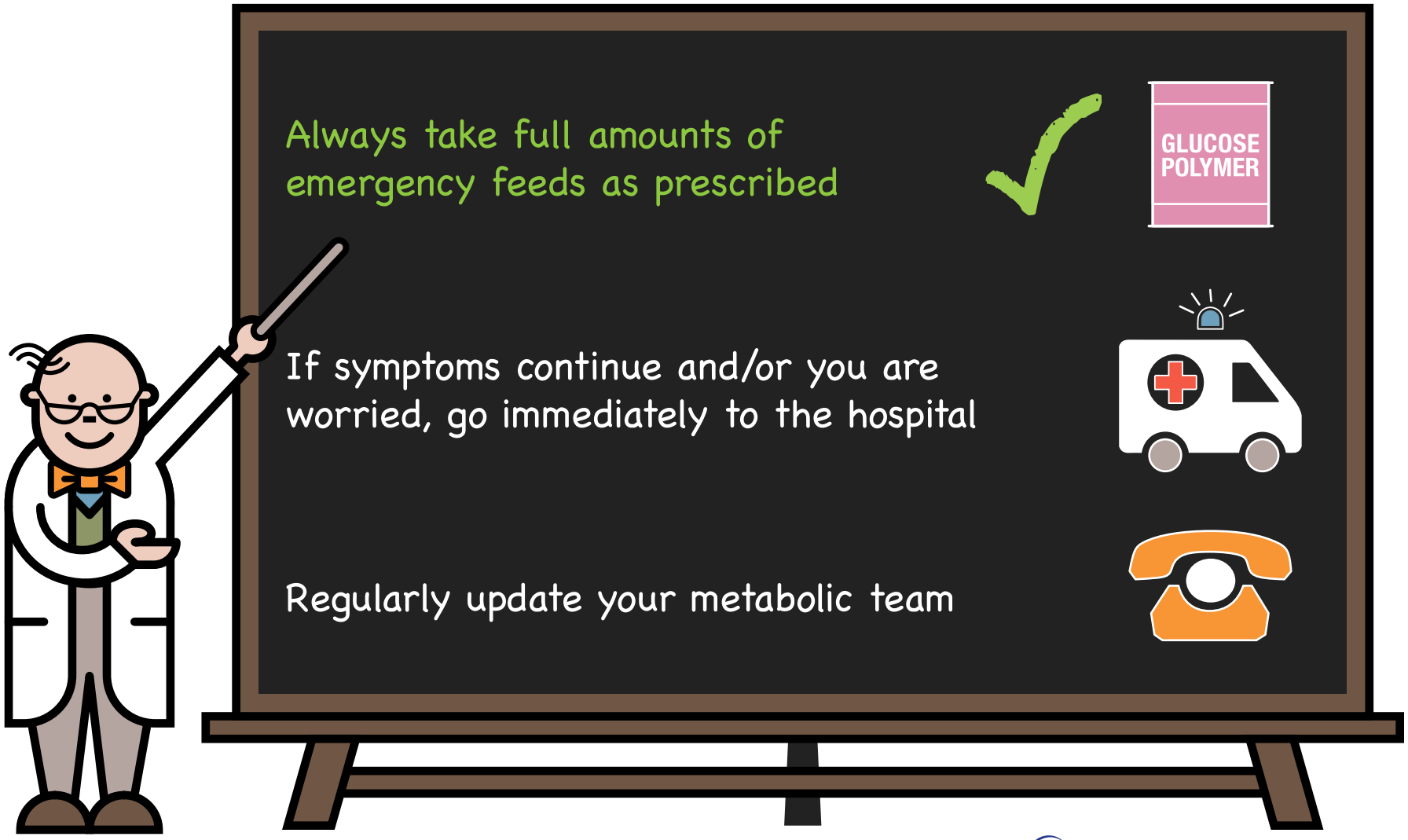


Checklist for illness



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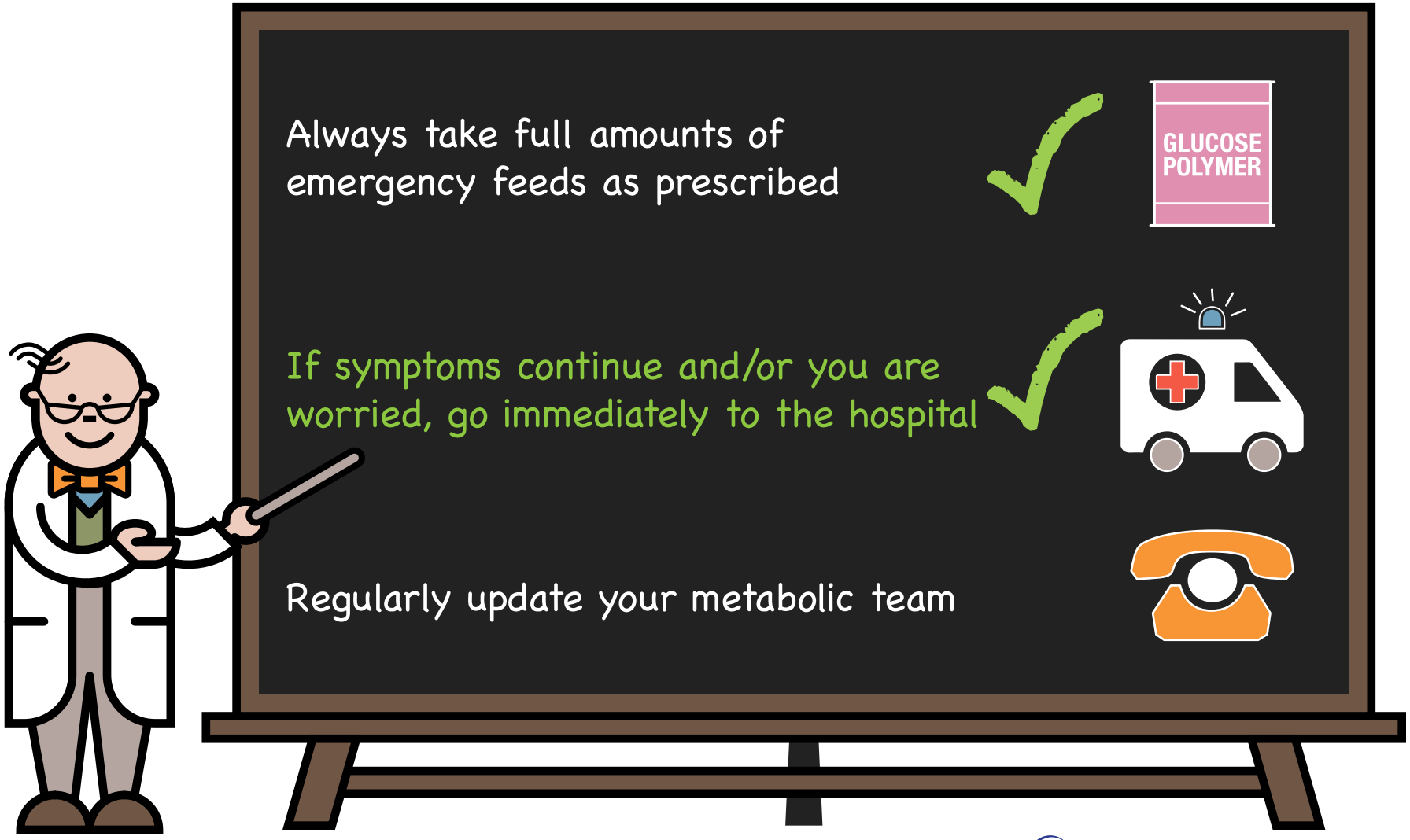

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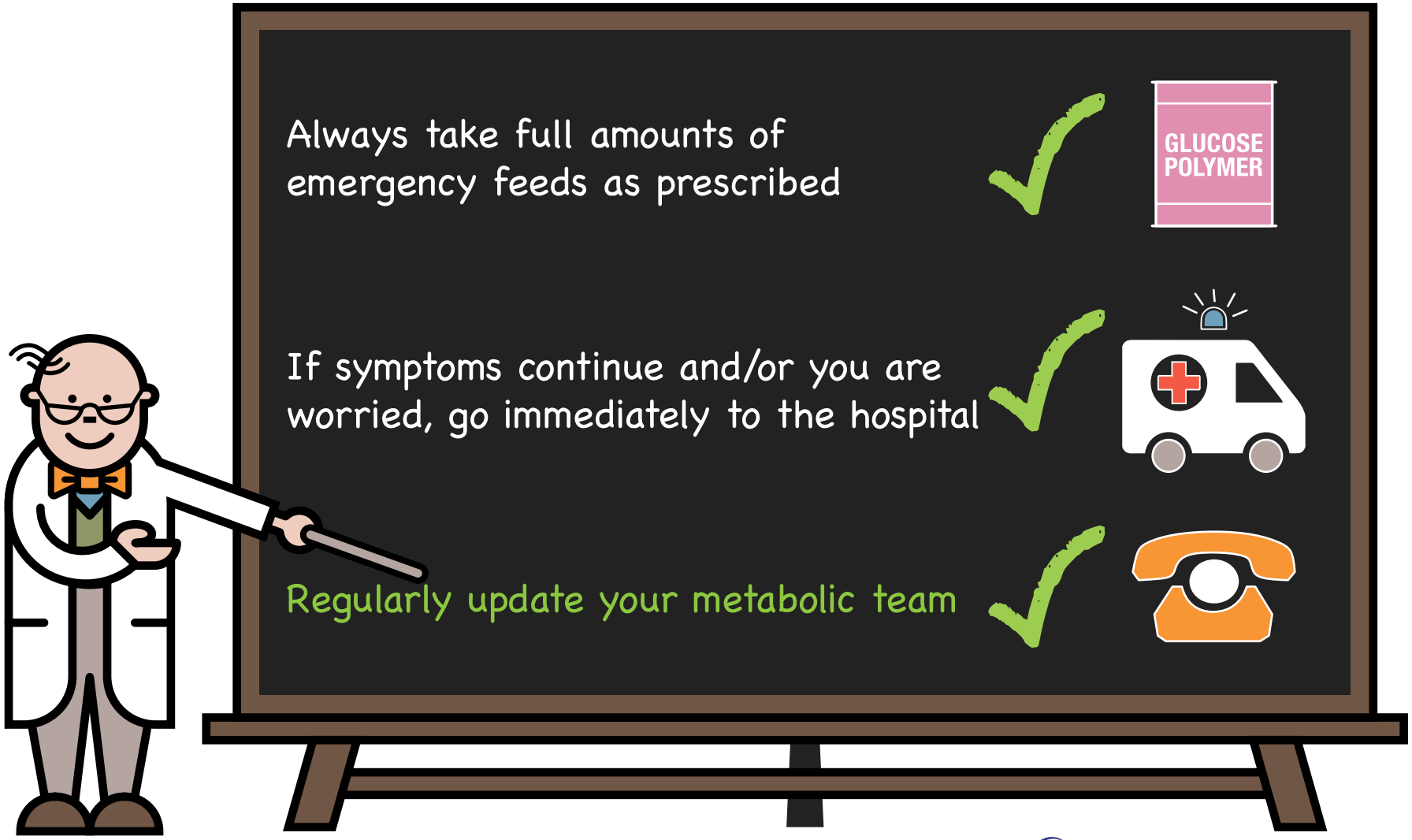
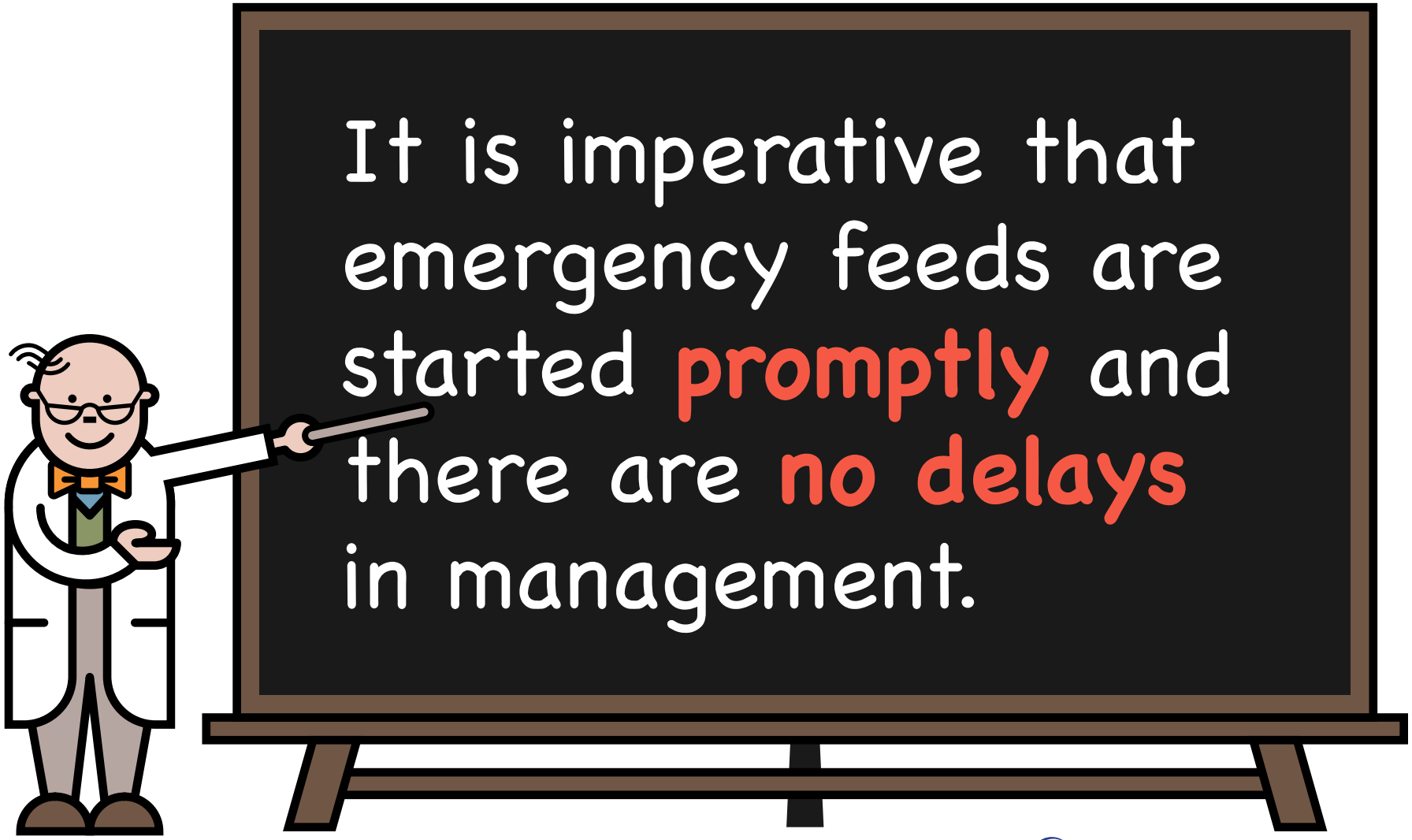
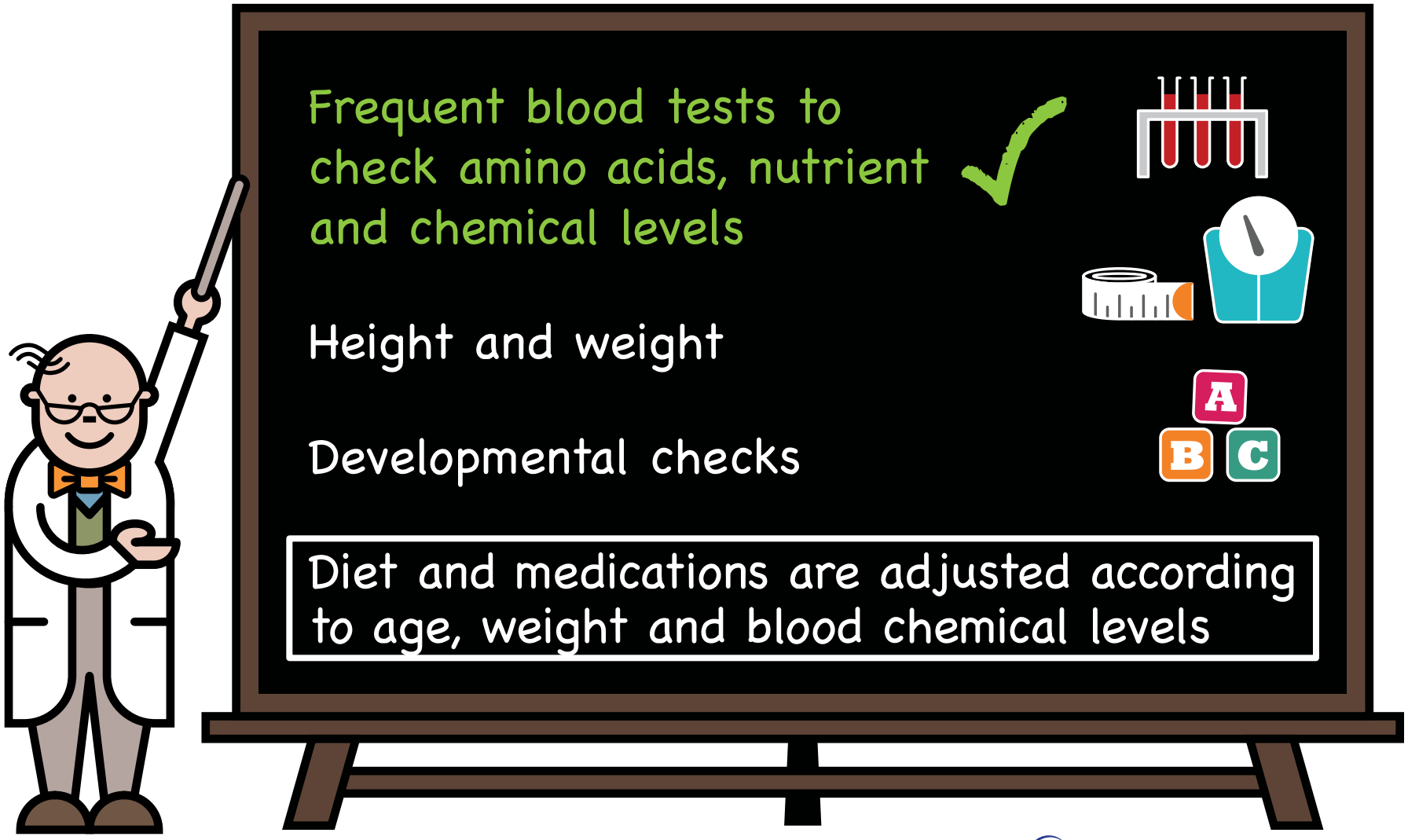


Key message



How is Argininosuccinic aciduria monitored?



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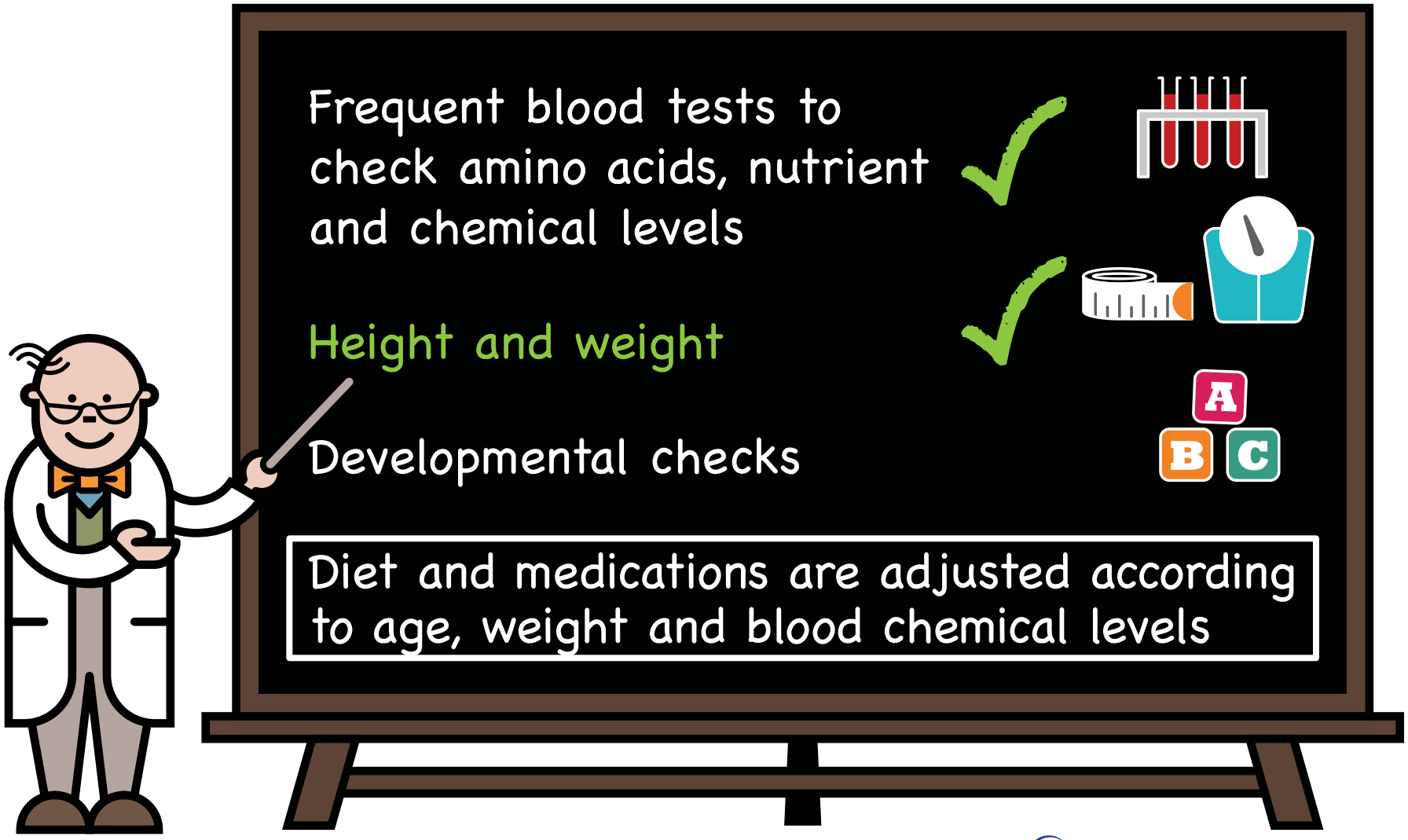

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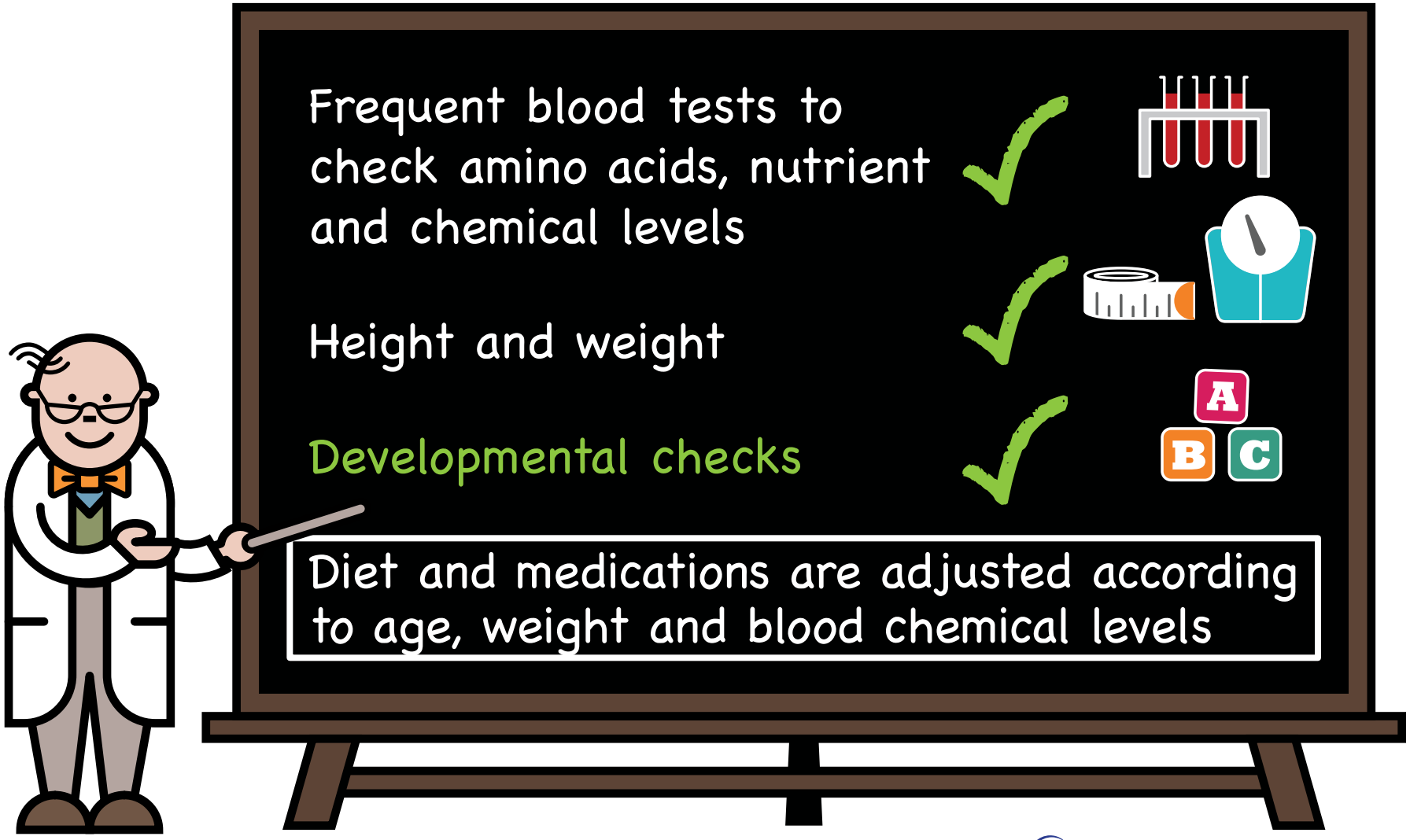
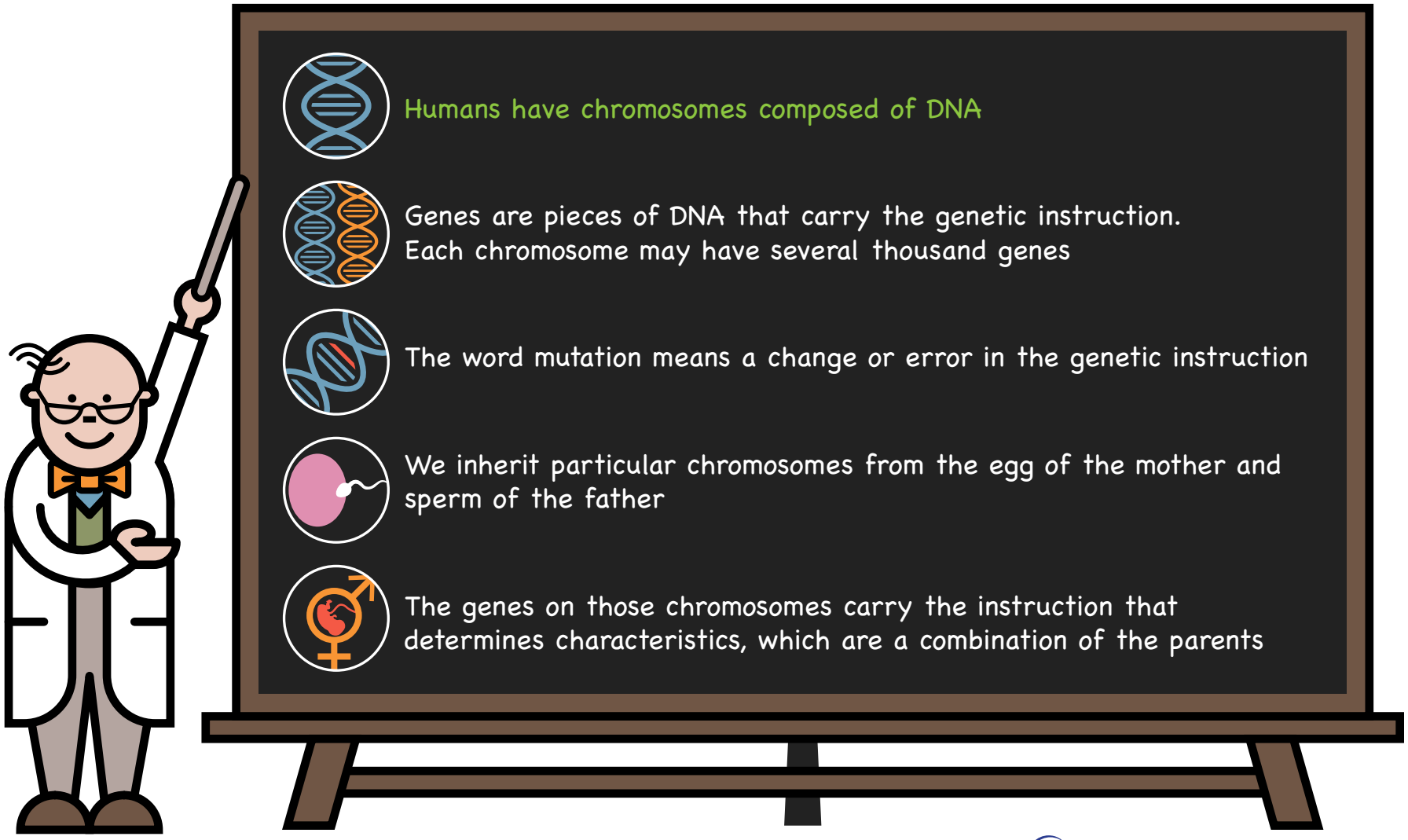


Chromosomes, genes, mutations



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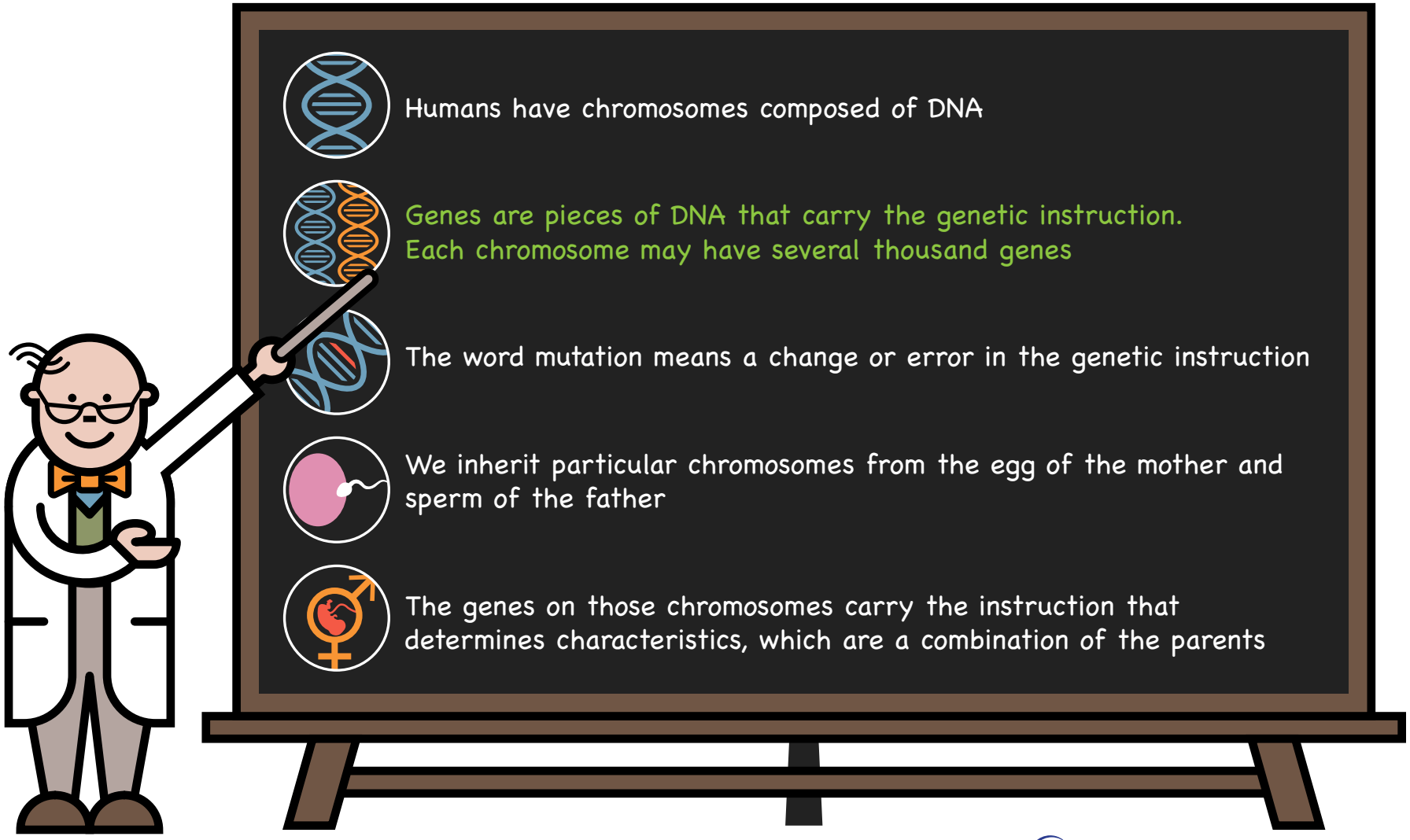
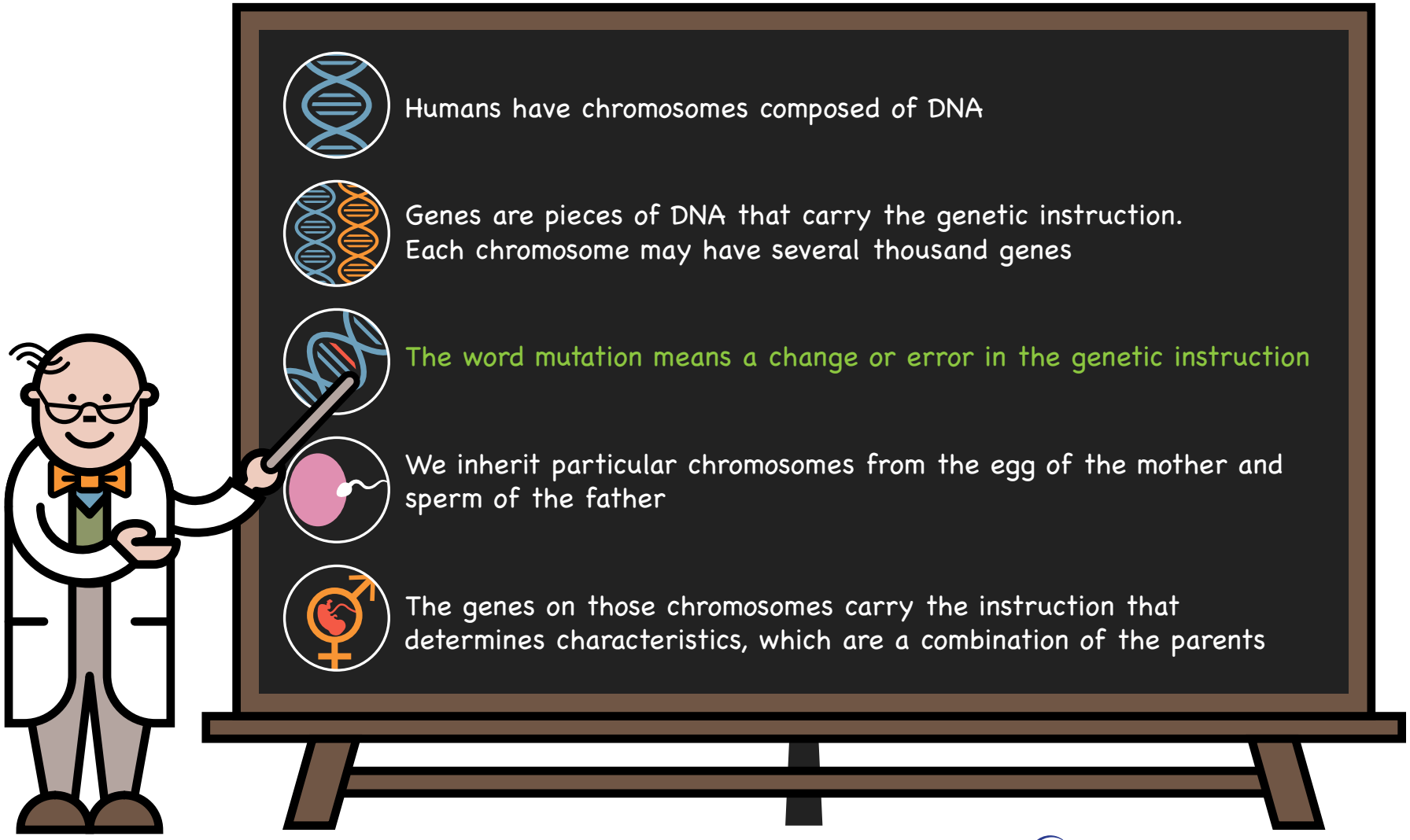
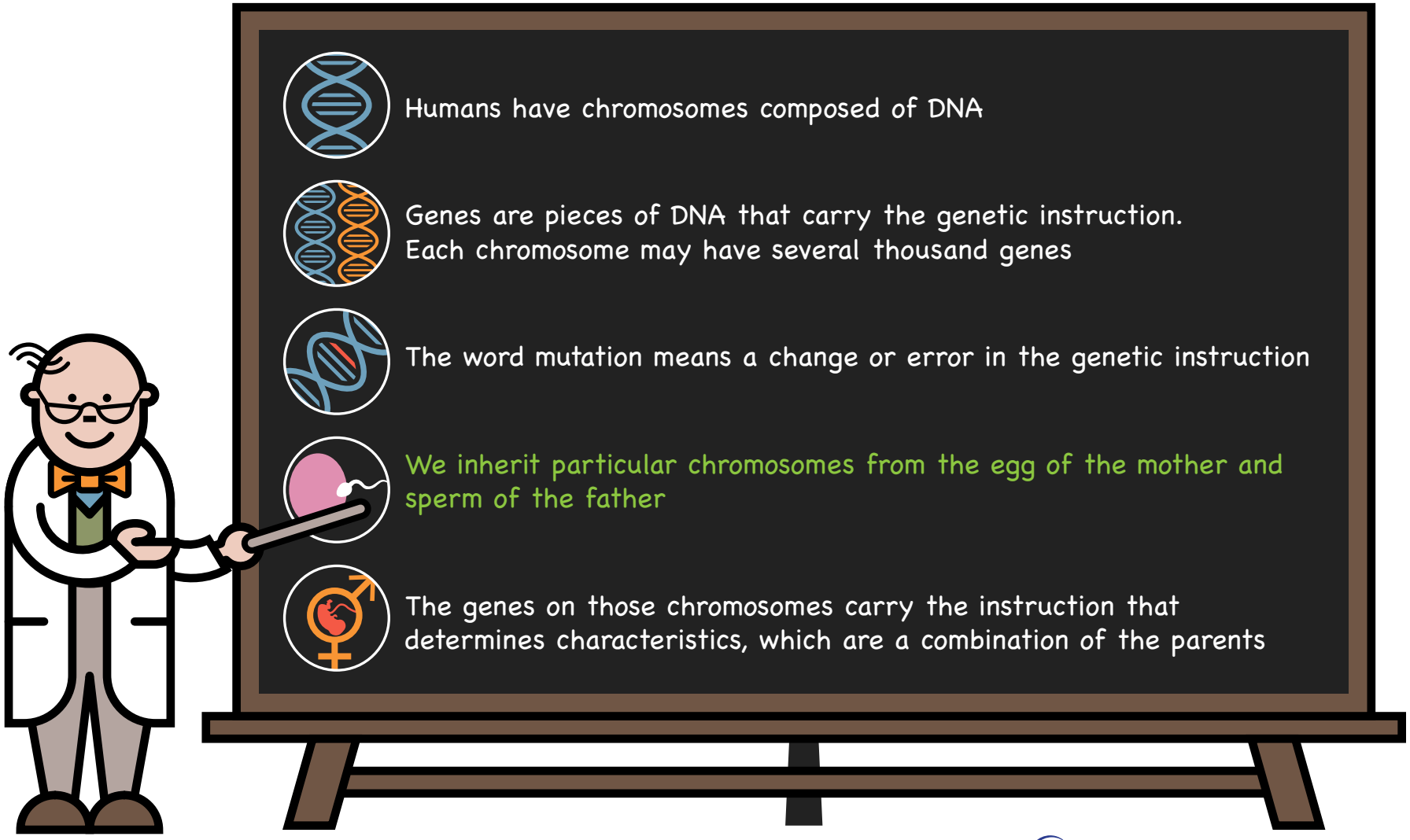


Chromosomes, genes, mutations



Chromosomes, genes, mutations



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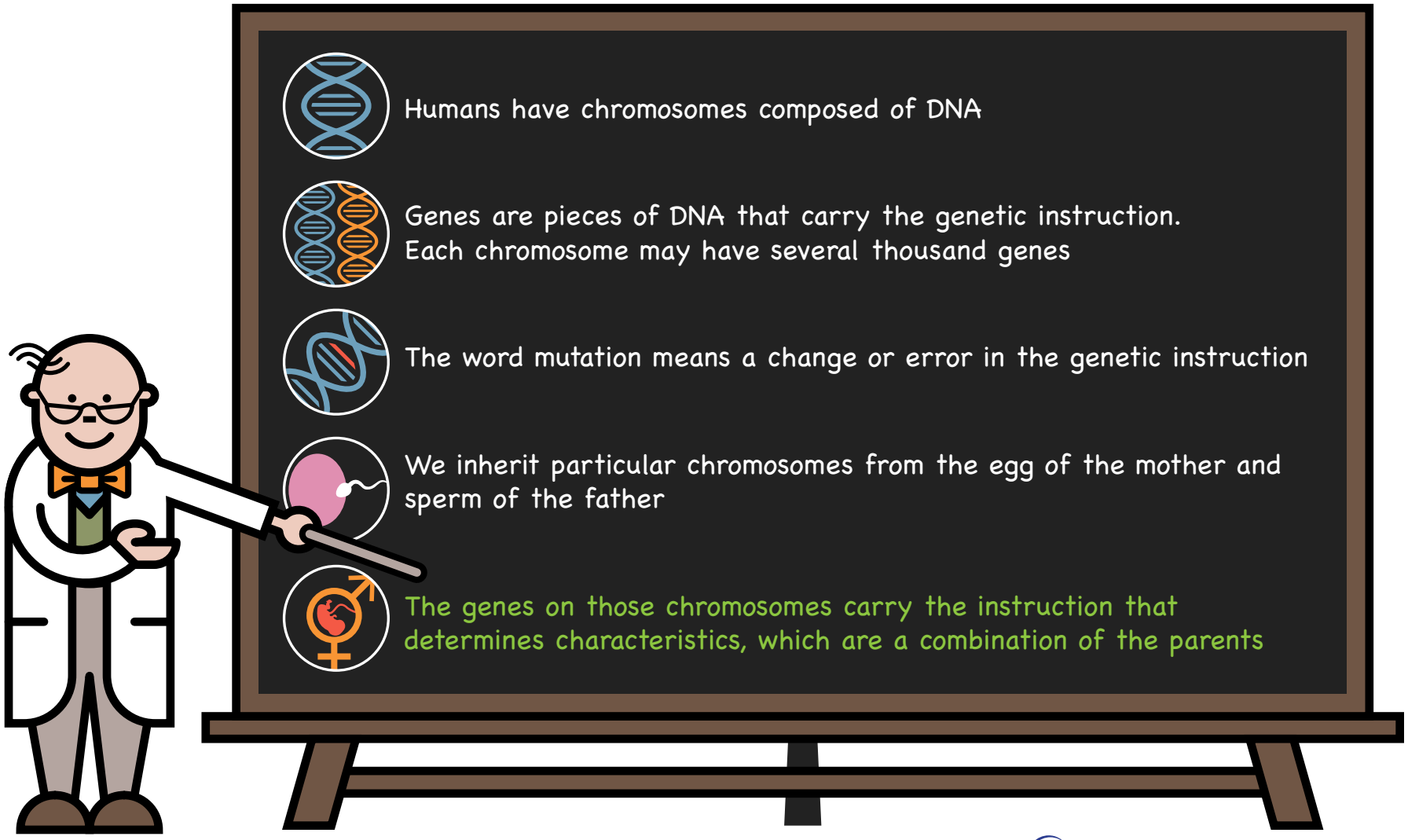
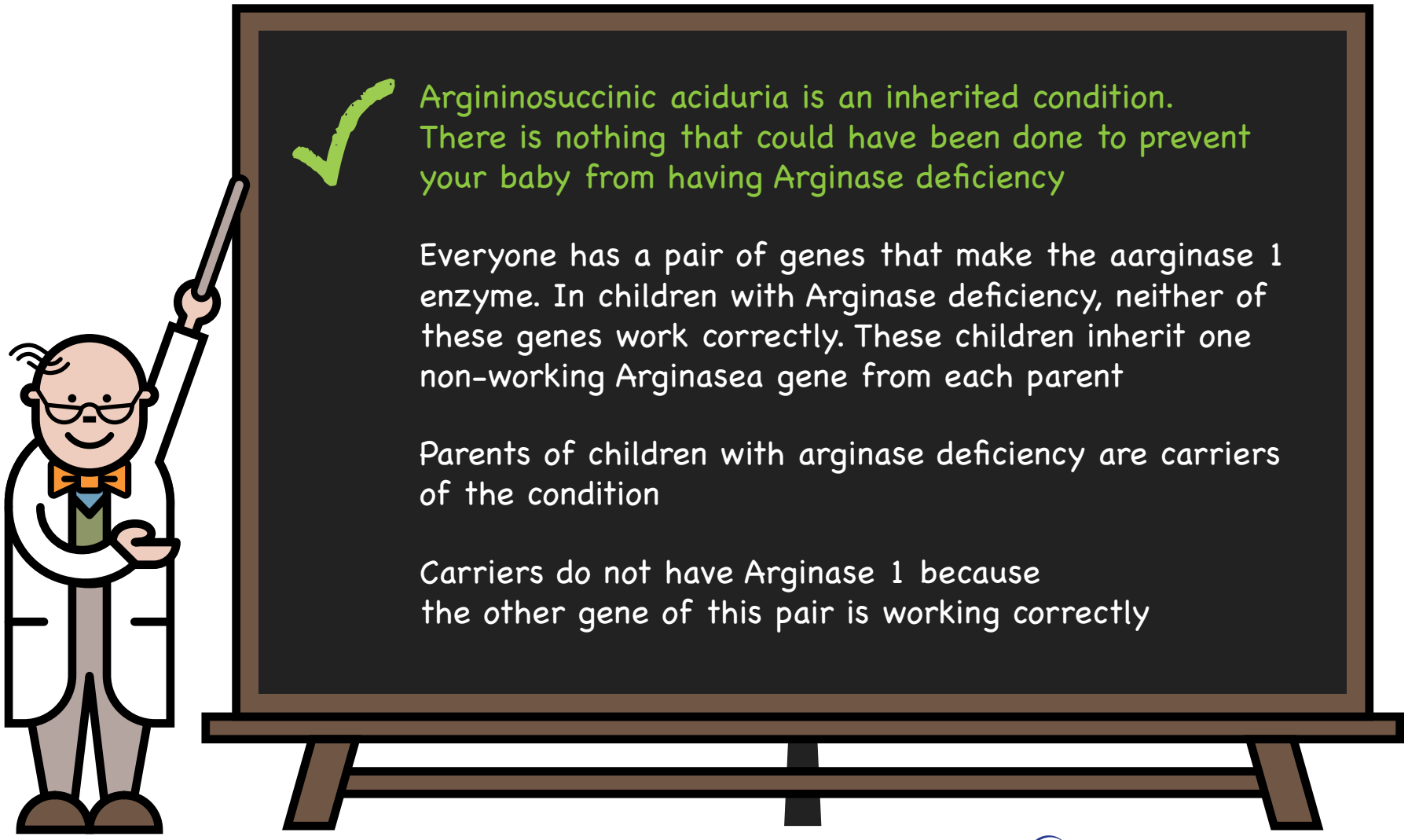


Inheritance



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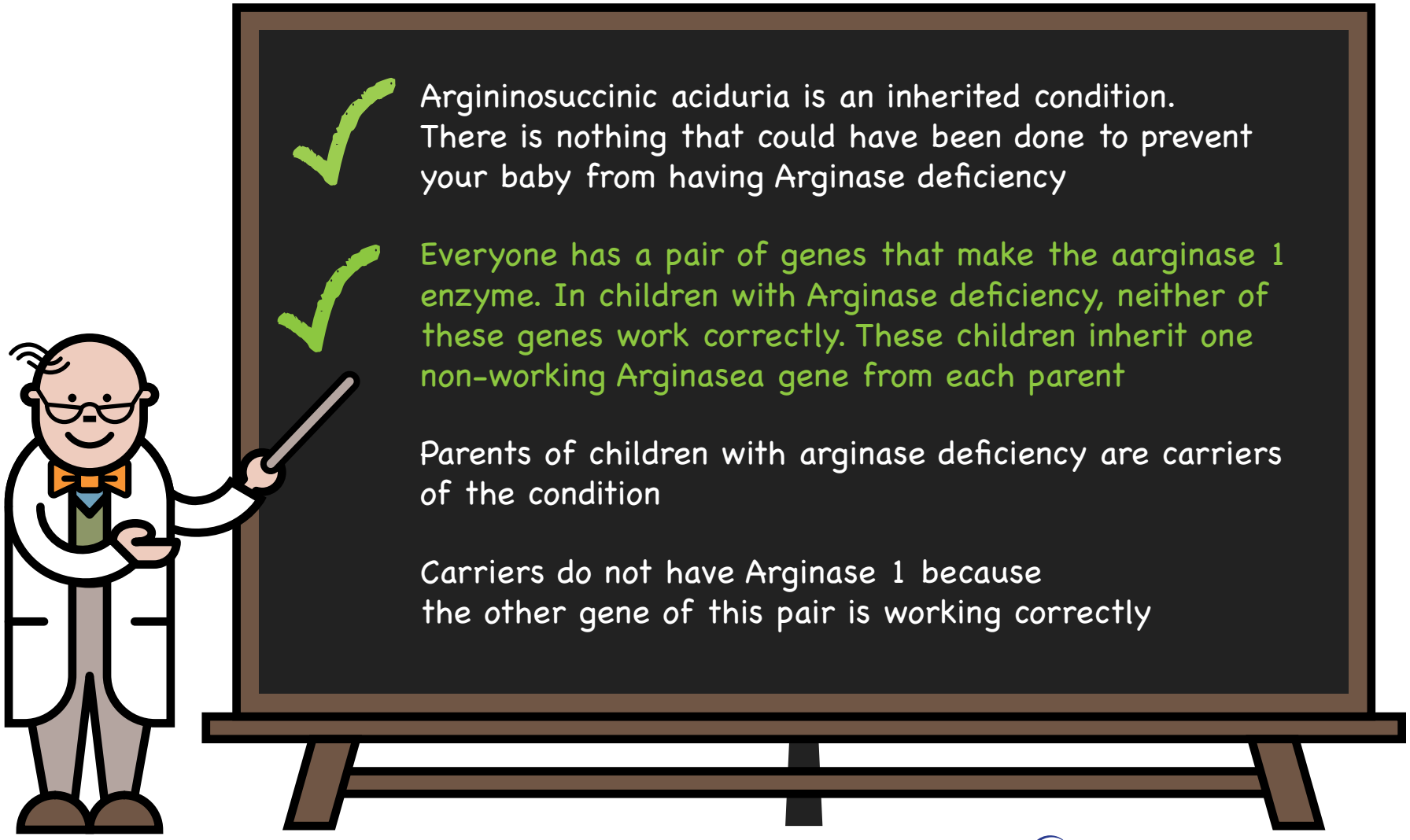

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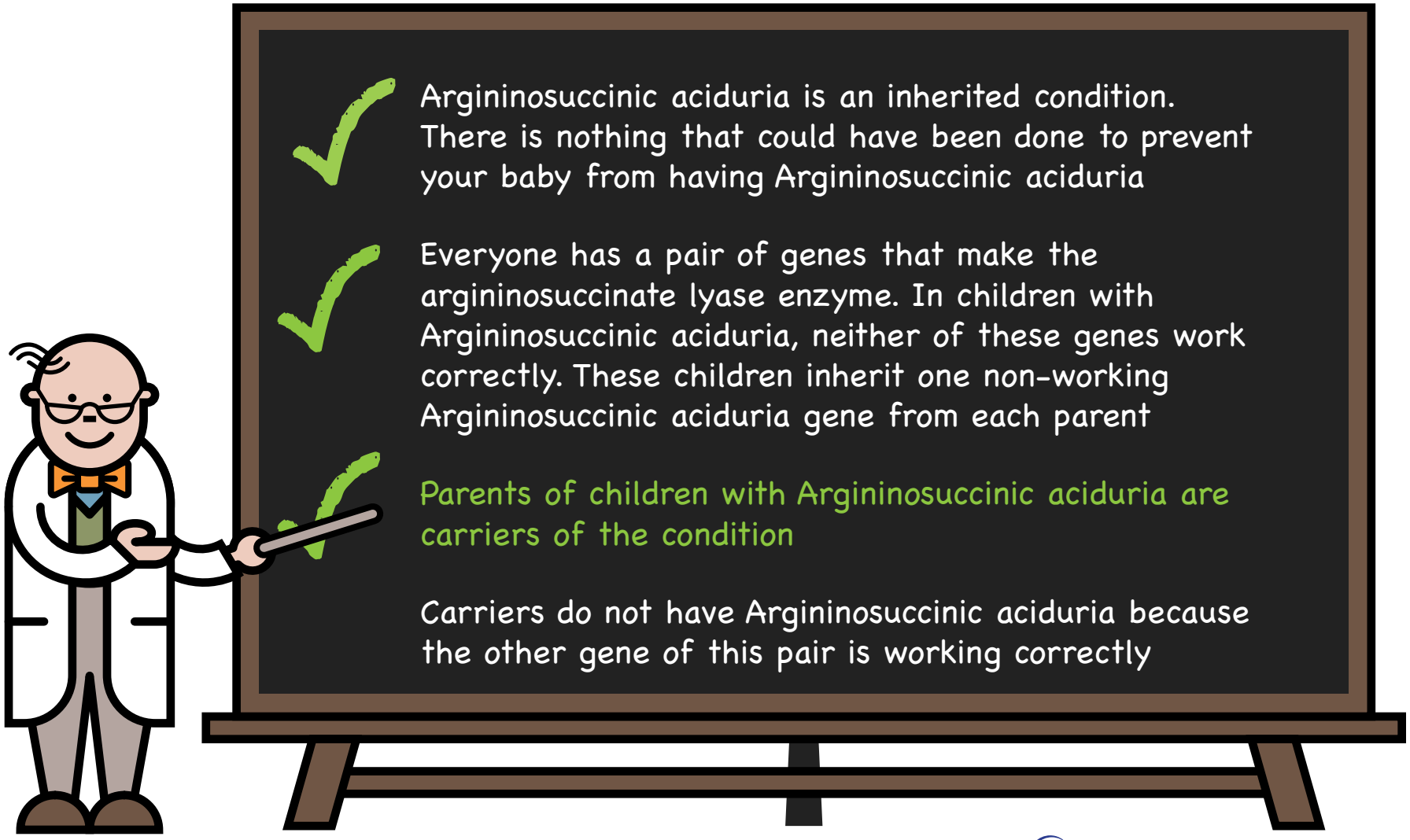

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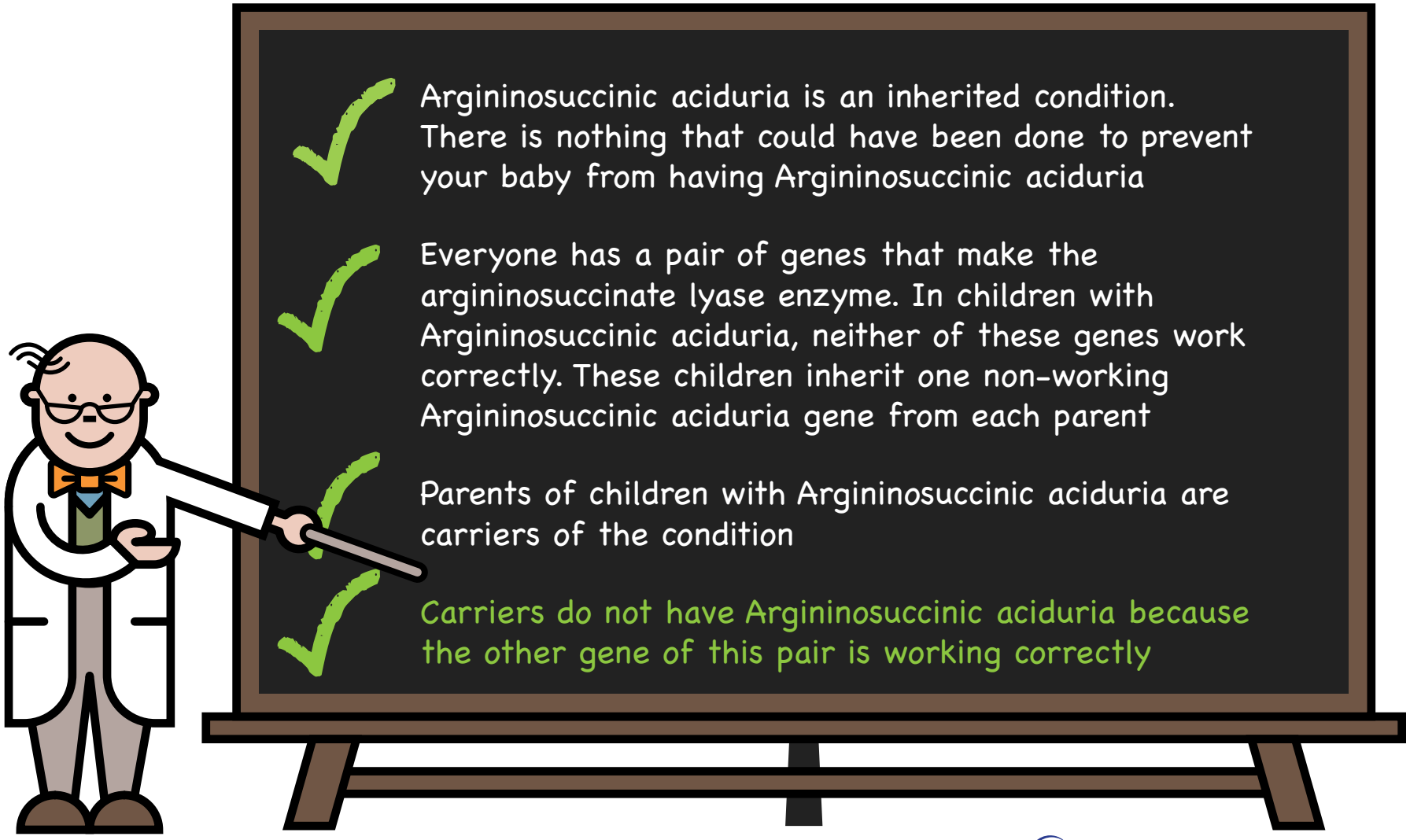
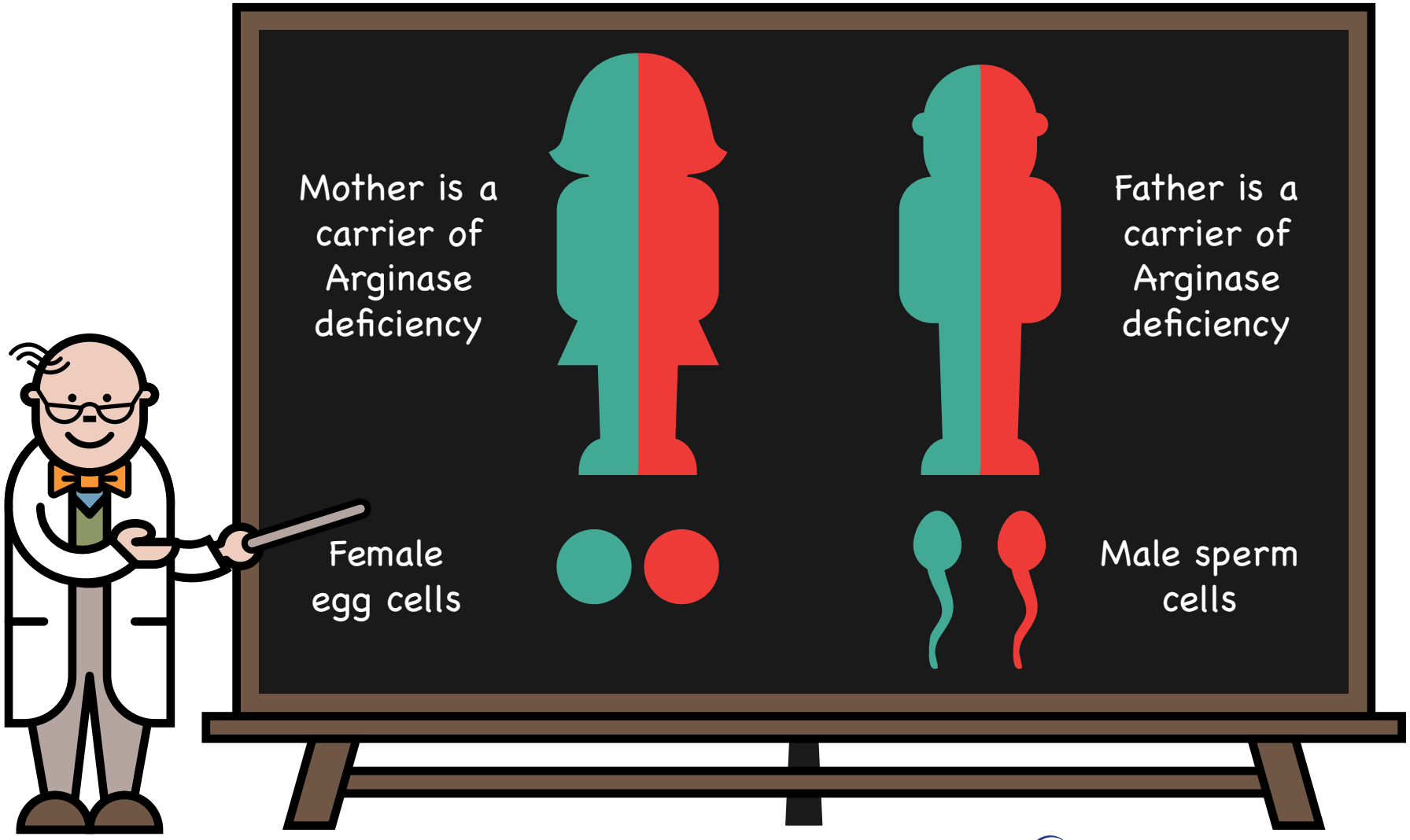
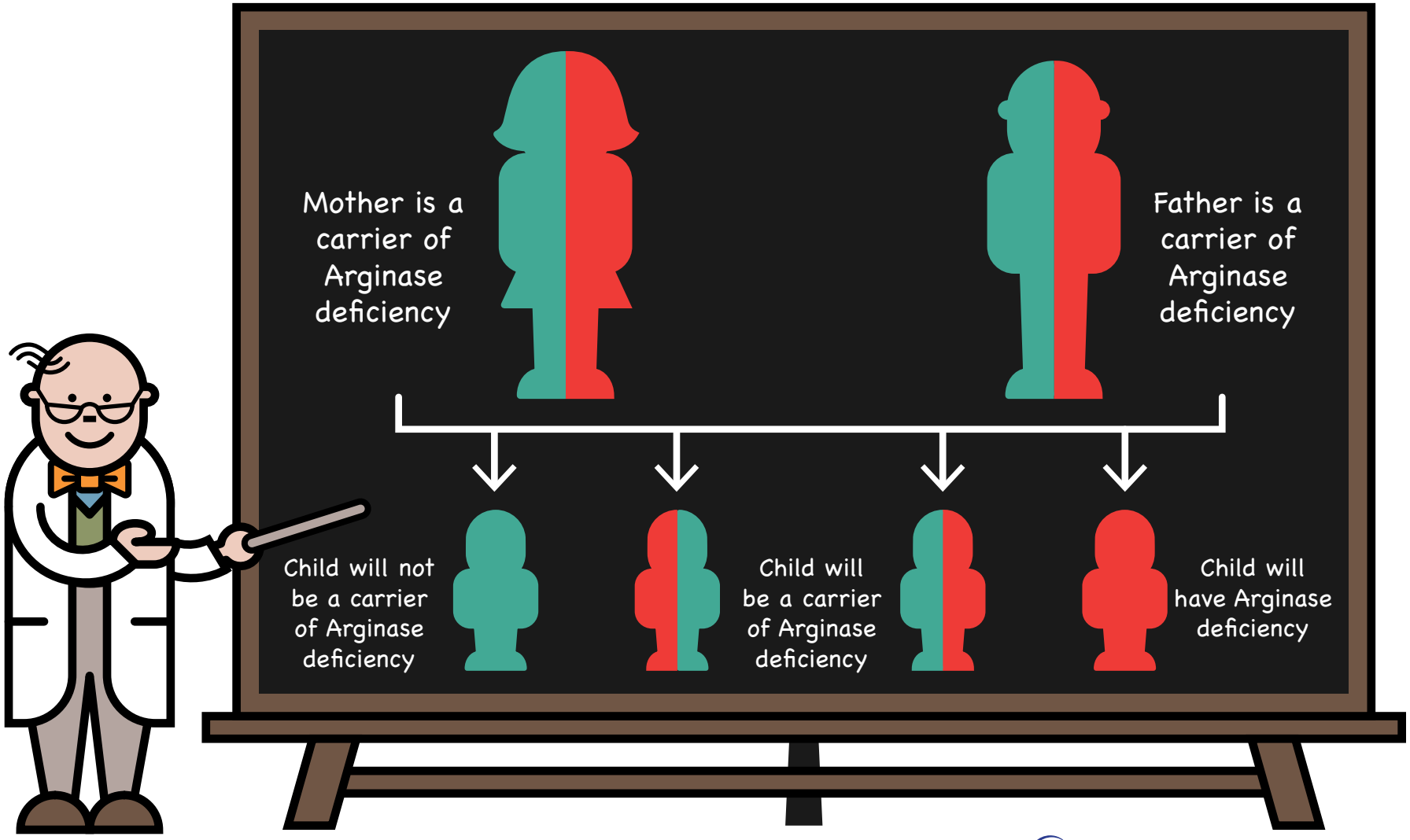


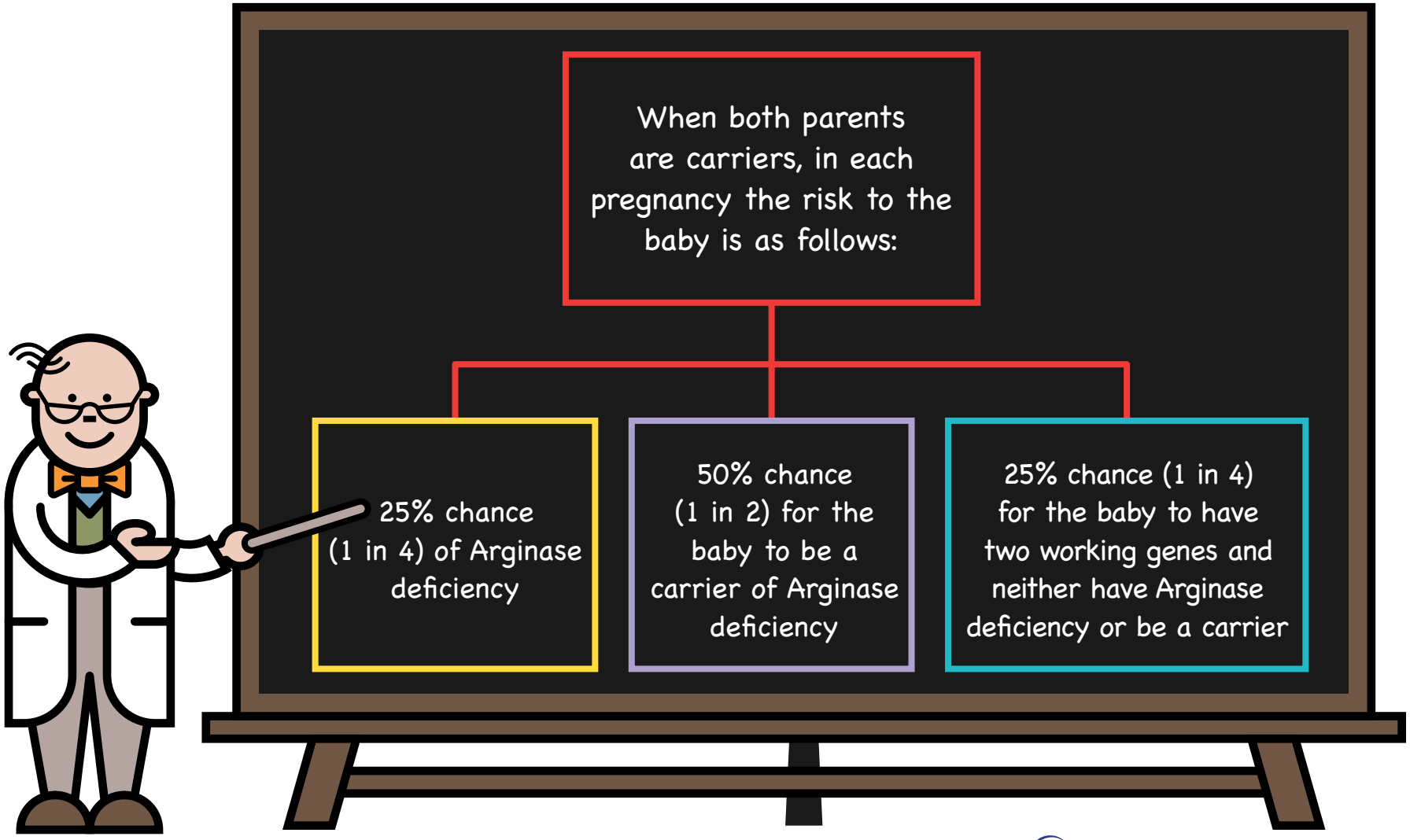
Inheritance – Autosomal recessive (carriers of Arginase deficiency)



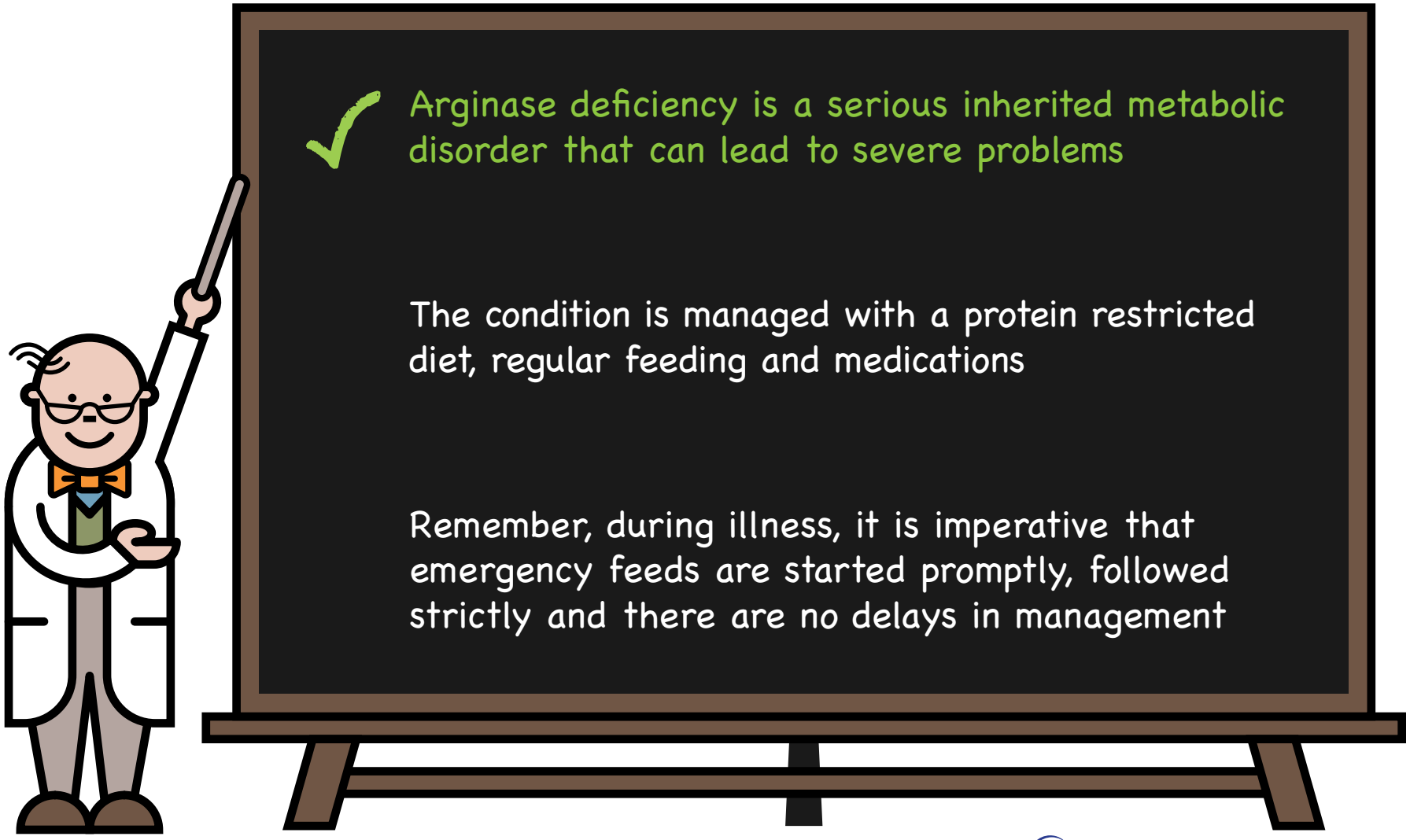
Inheritance – Autosomal recessive – possible combinations



Future pregnancies



Take home messages



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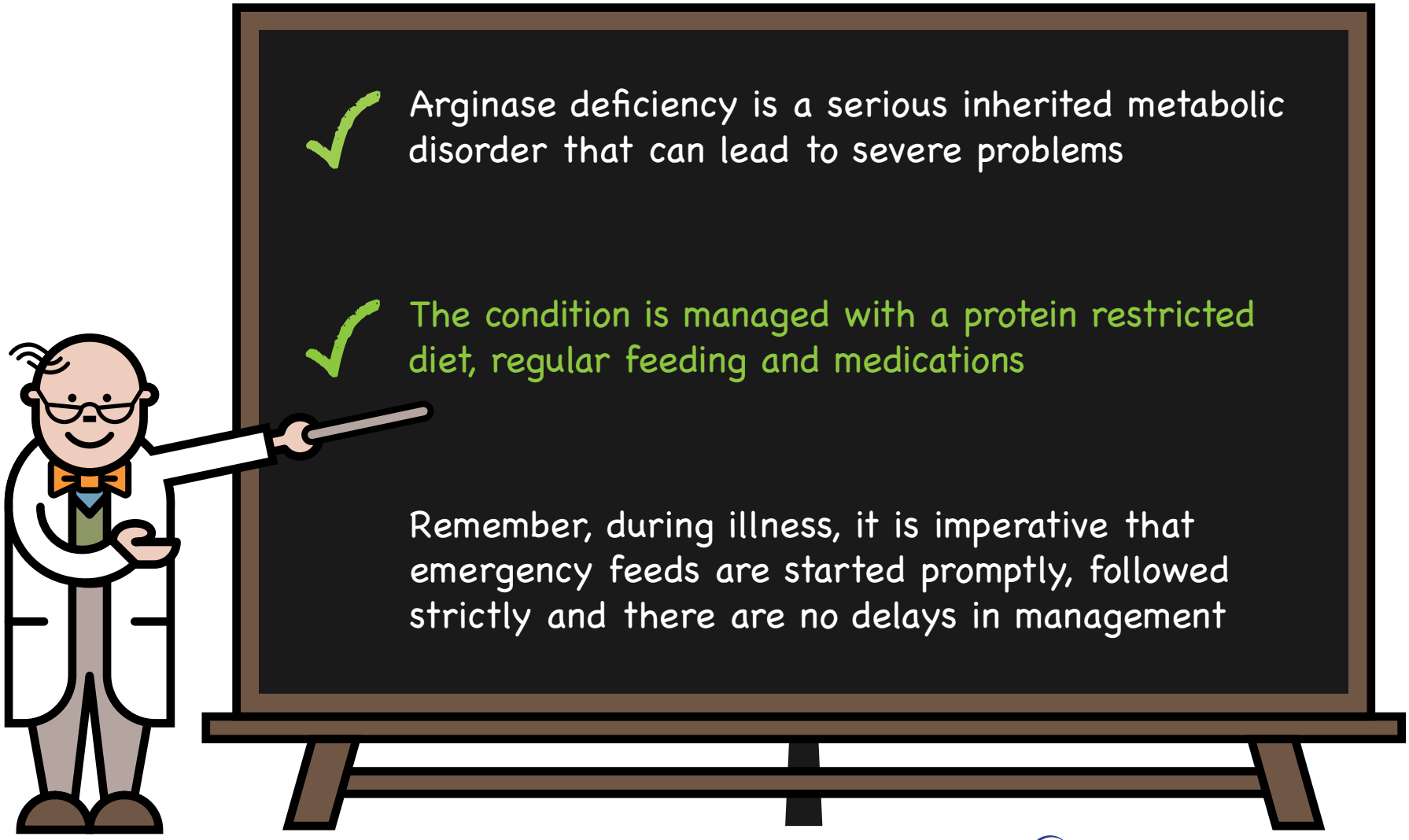

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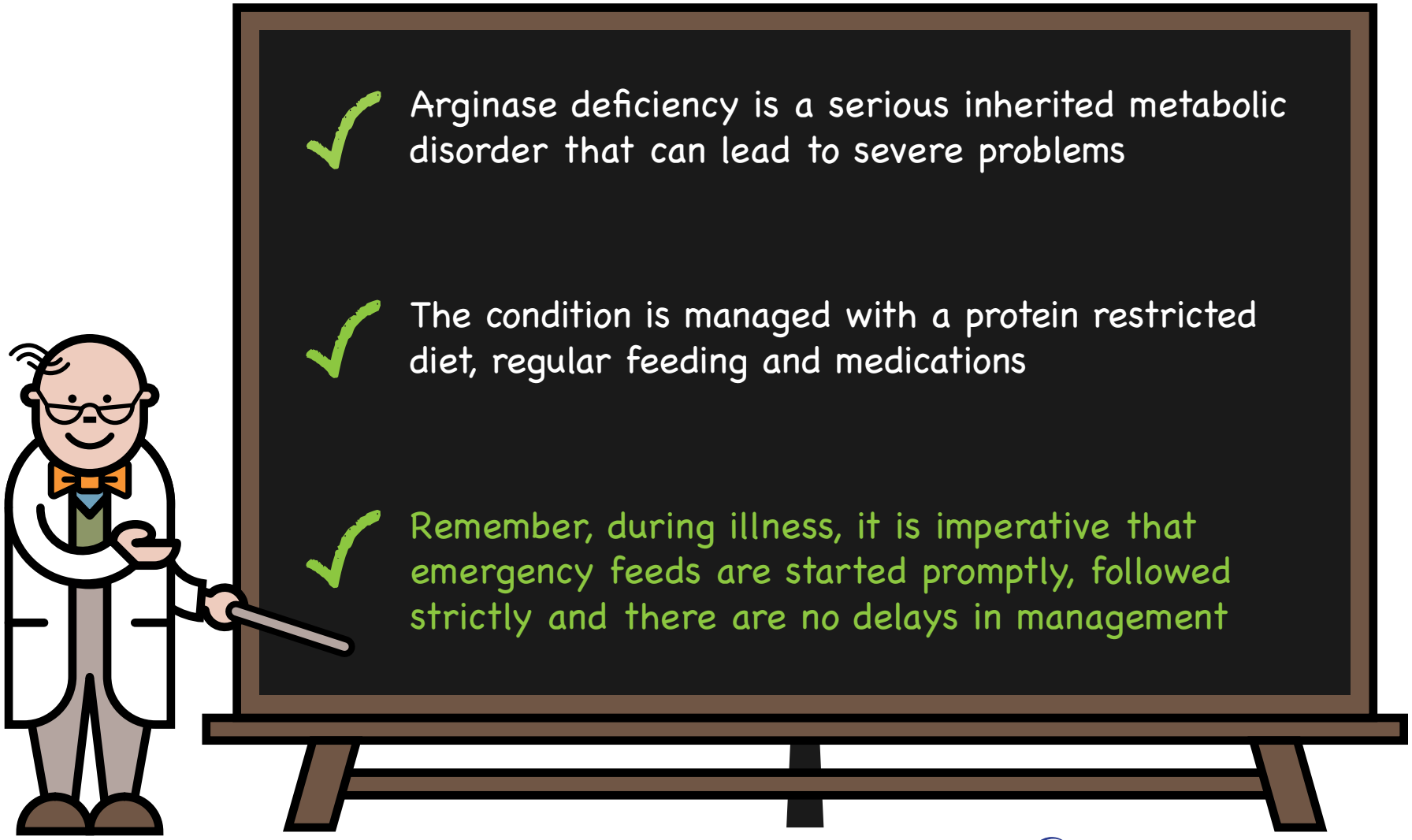

Helpful hints

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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