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 2, SEPTEMBER 2020

Argininosuccinic aciduria

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.

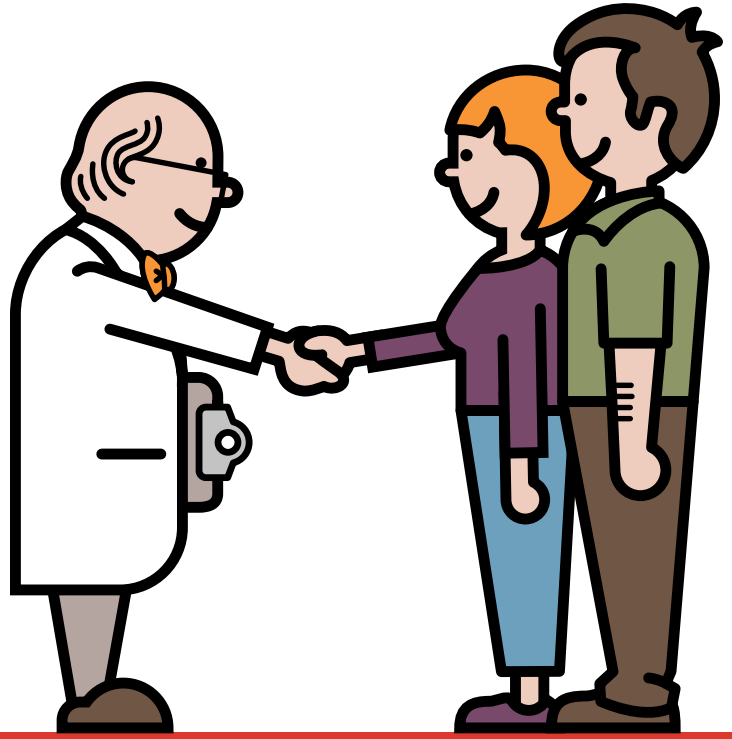
Each module produced is reviewed by a consultant clinician who is a member of the BIMDG.

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

Argininosuccinic aciduria

Information for families following a new diagnosis



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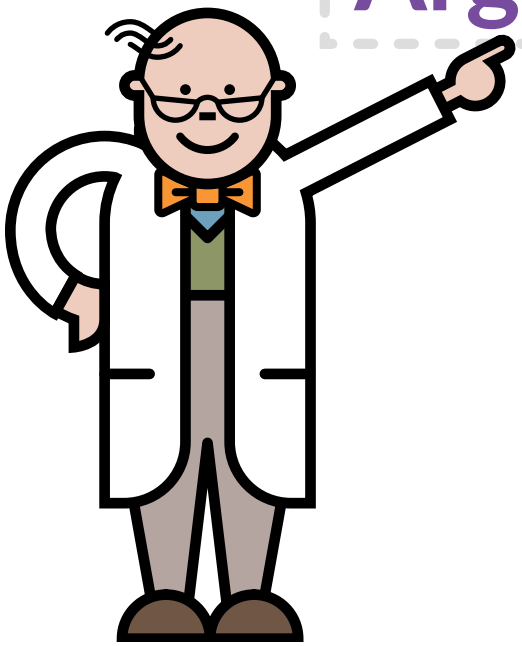
What is Argininosuccinic aciduria?

It is an inherited metabolic condition.

It is sometimes shortened to ASA.



Arginino Succinic Aciduria

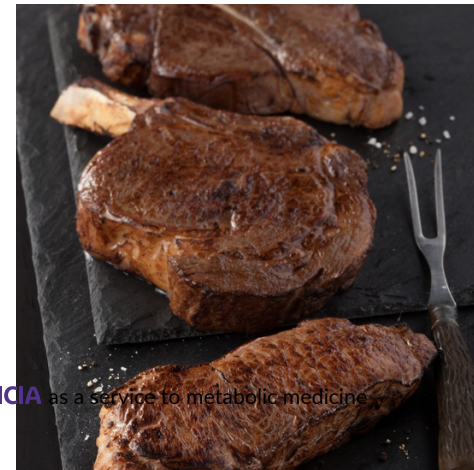
Arginino Succinic Aciduria

What is protein?

Many foods contain protein.

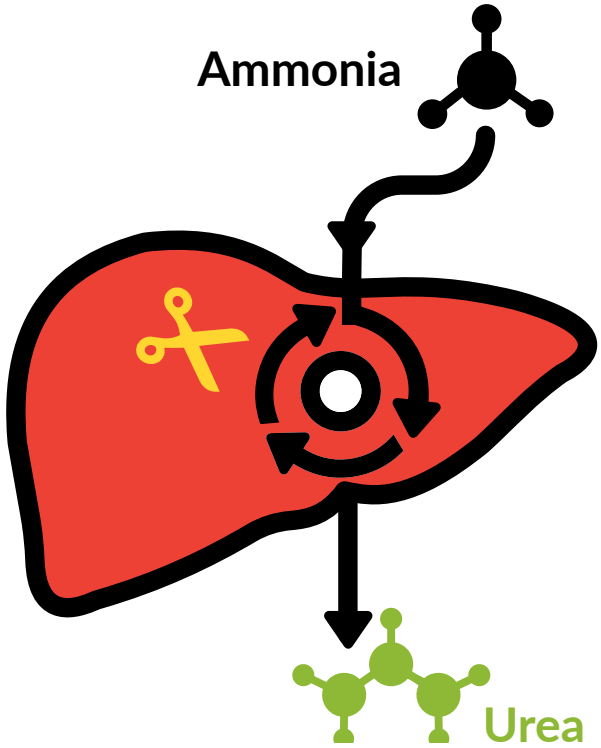
The body needs protein for growth and repair.

Many people eat more protein than the body needs.



How do we remove waste protein from the body?

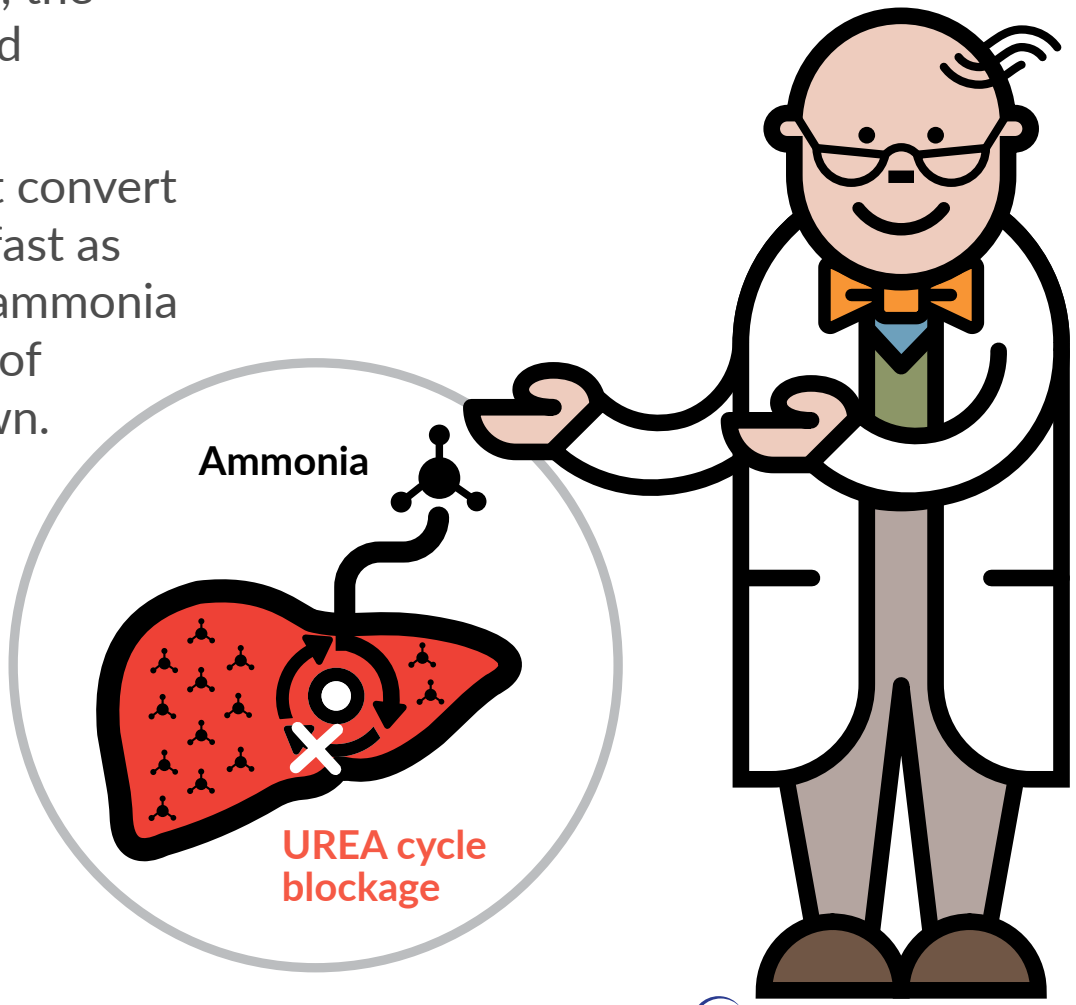
- Firstly, the body converts waste protein to a toxic chemical called **ammonia**
- Ammonia is then converted into a non-toxic chemical **(urea)** in the liver
- This process occurs via the **urea cycle**
- In the urea cycle, several steps have to take place. Each step needs an enzyme (like chemical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



What happens in Argininosuccinic aciduria?

In Argininosuccinic aciduria, the body lacks an enzyme called **argininosuccinate lyase**.

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.

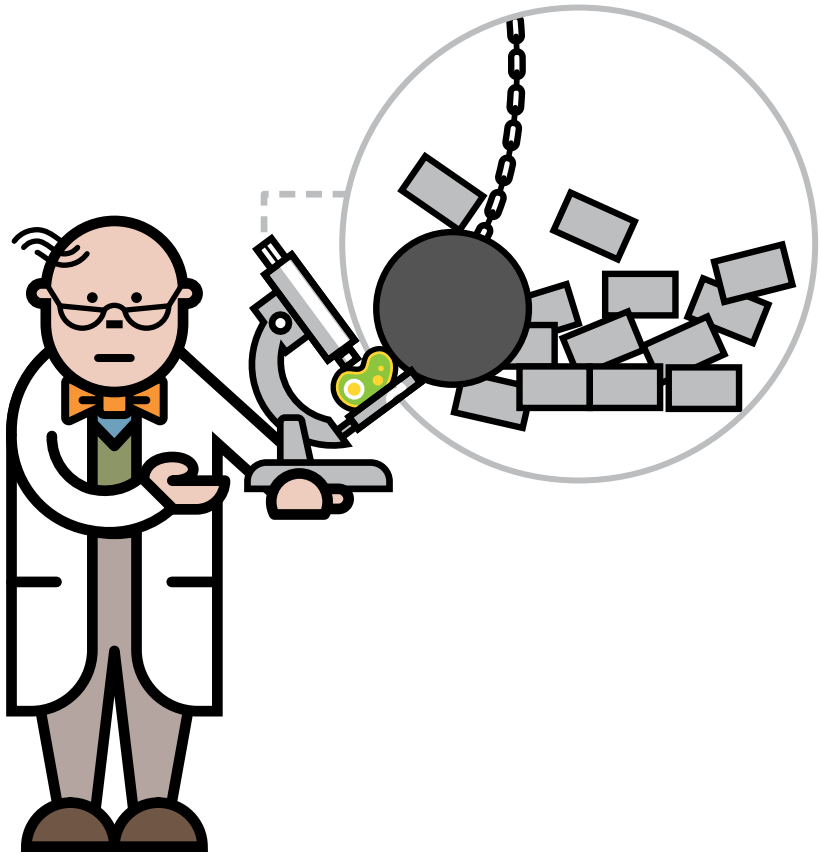


When does Argininosuccinic aciduria cause high ammonia levels?

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 Argininosuccinic aciduria?

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

Signs and symptoms:

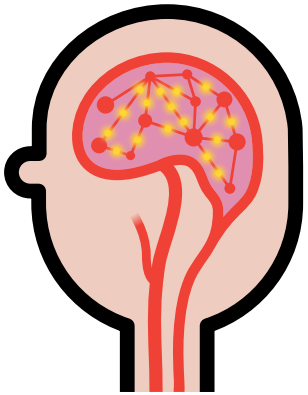
- Poor feeding
- Vomiting
- Floppiness
- Excessive sleepiness
- Rapid breathing
- Dehydration (lack of body fluids)
- Seizures

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

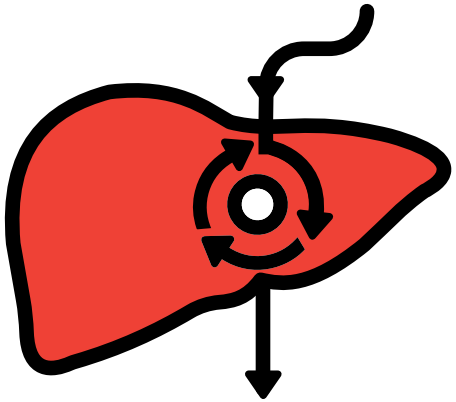
Some babies may present later.

What are the long term effects of Argininosuccinic aciduria?

There may be learning difficulties and delays to normal development, like walking and talking.

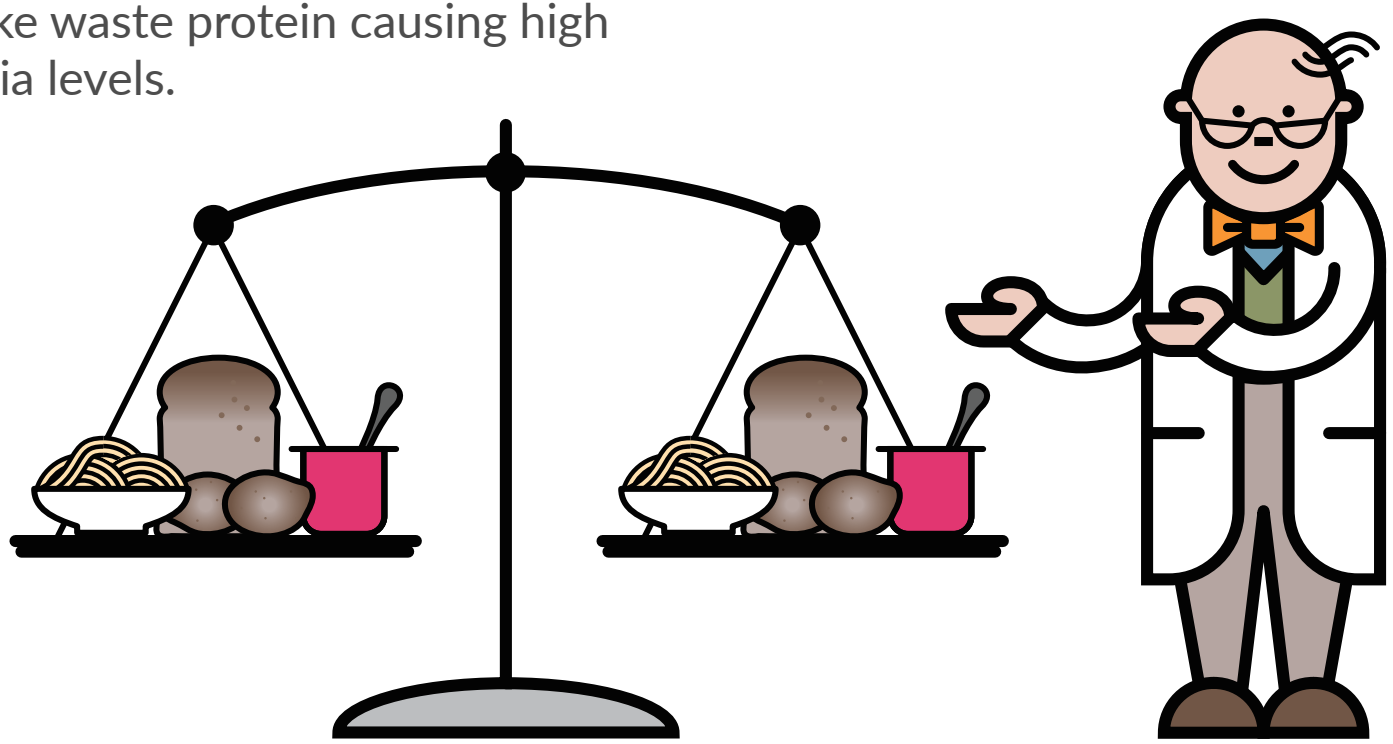


It may also affect the liver or other parts of the body.



Protein balance is needed in Argininosuccinic aciduria

In **Argininosuccinic** aciduria 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 Argininosuccinic aciduria 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 the mutation in the ASA gene.

How is Argininosuccinic aciduria managed day to day?

Argininosuccinic aciduria 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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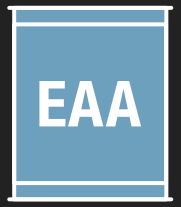
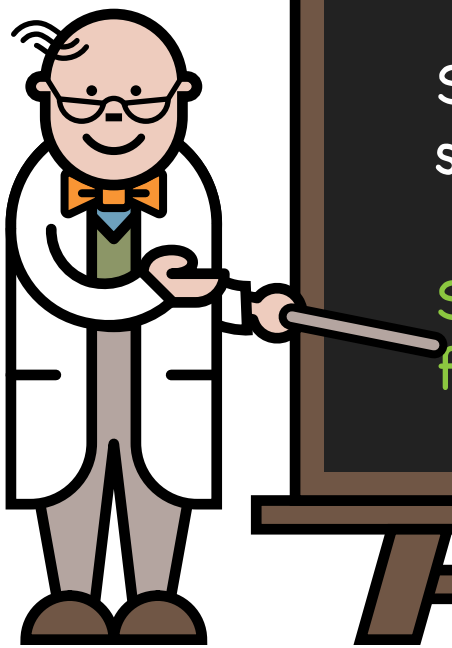
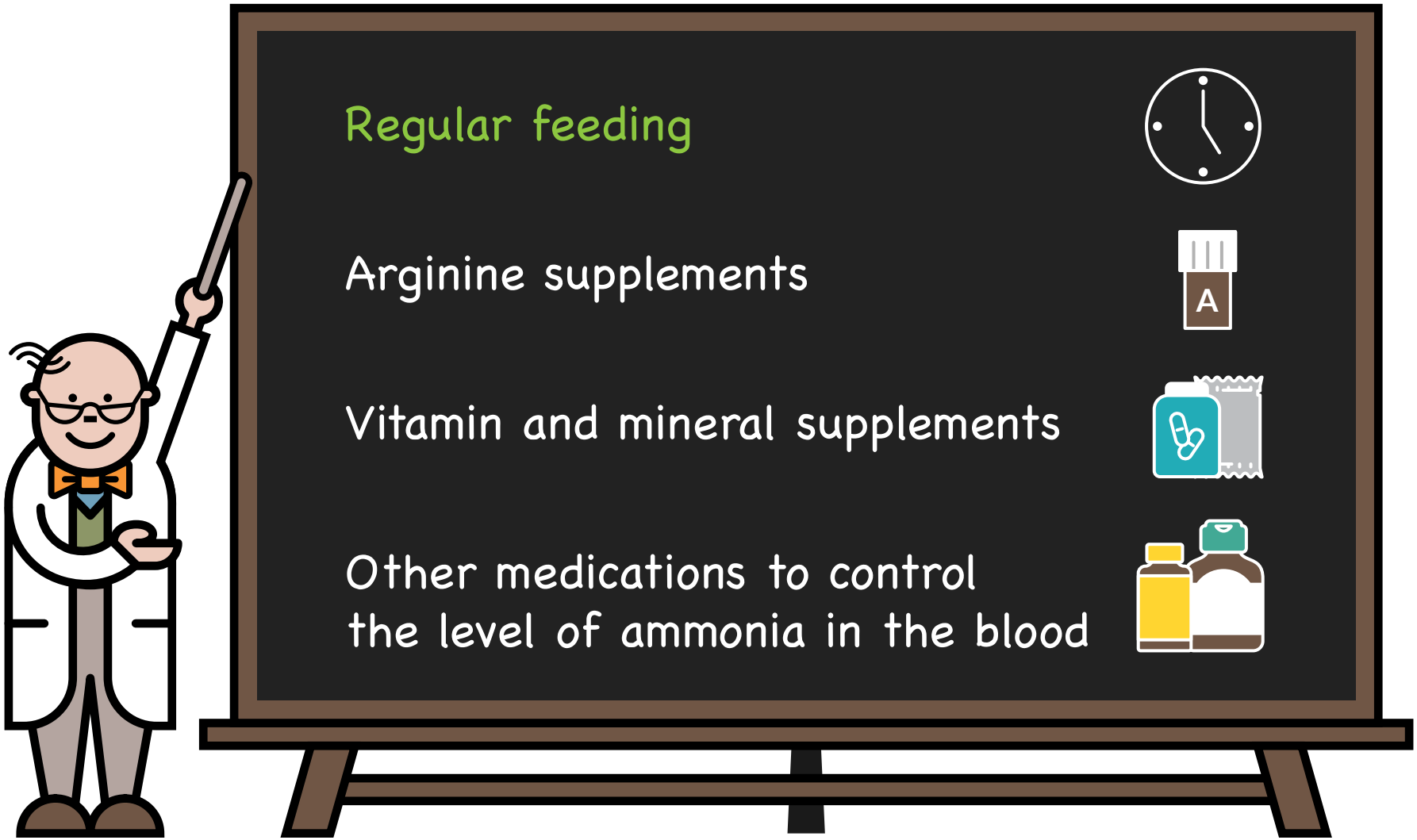


Sufficient energy supply from food and feeds



How is Argininosuccinic aciduria managed day to day?



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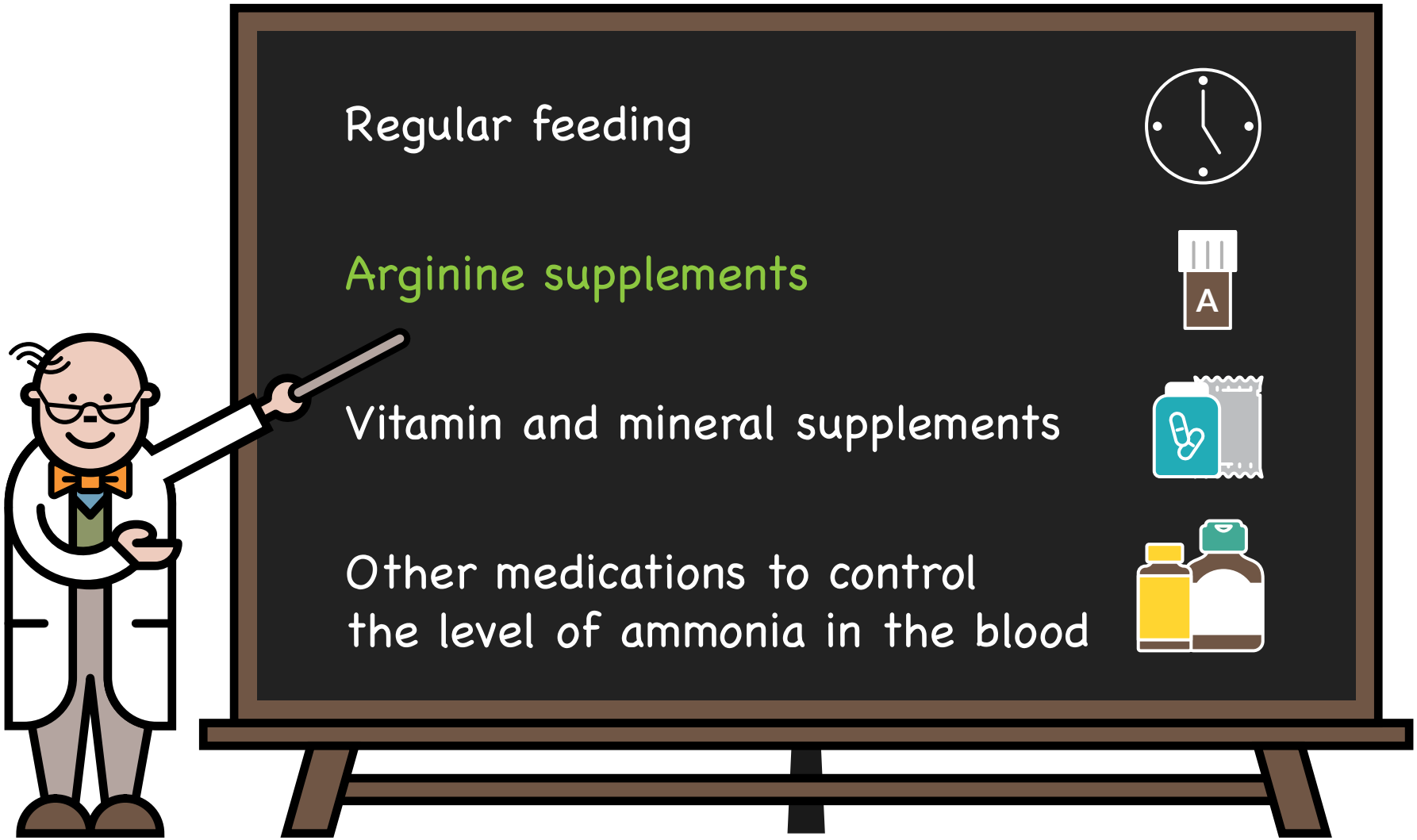

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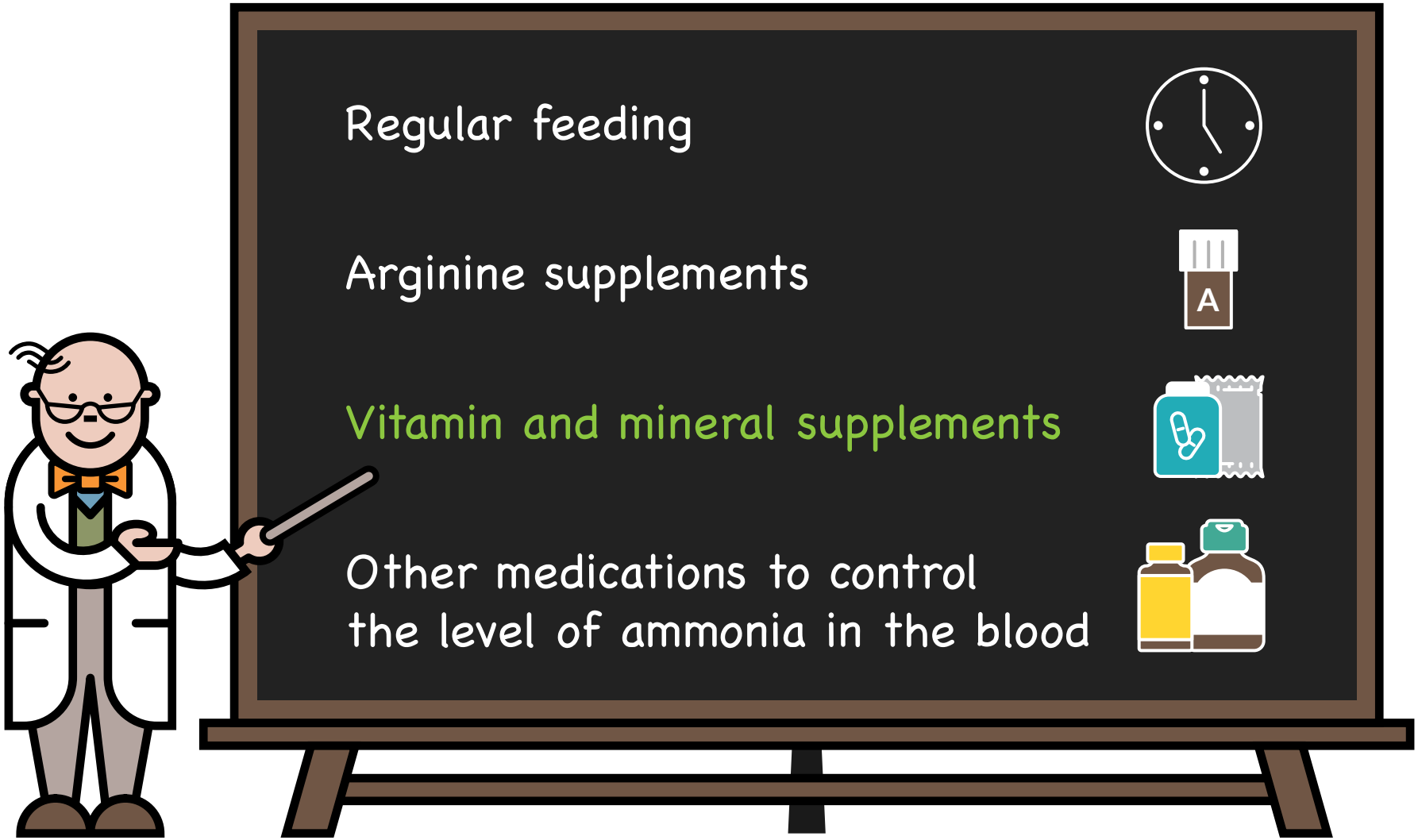

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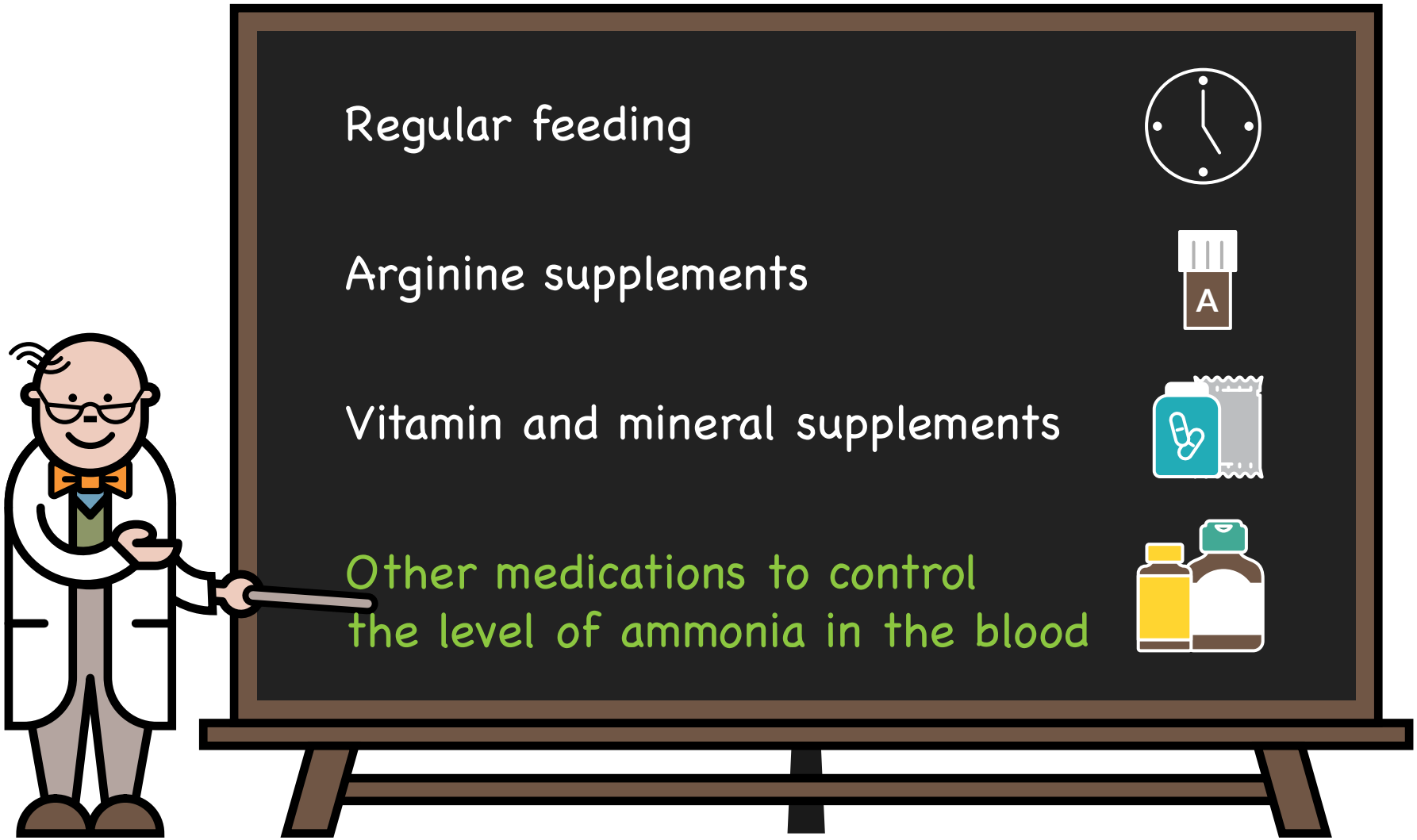
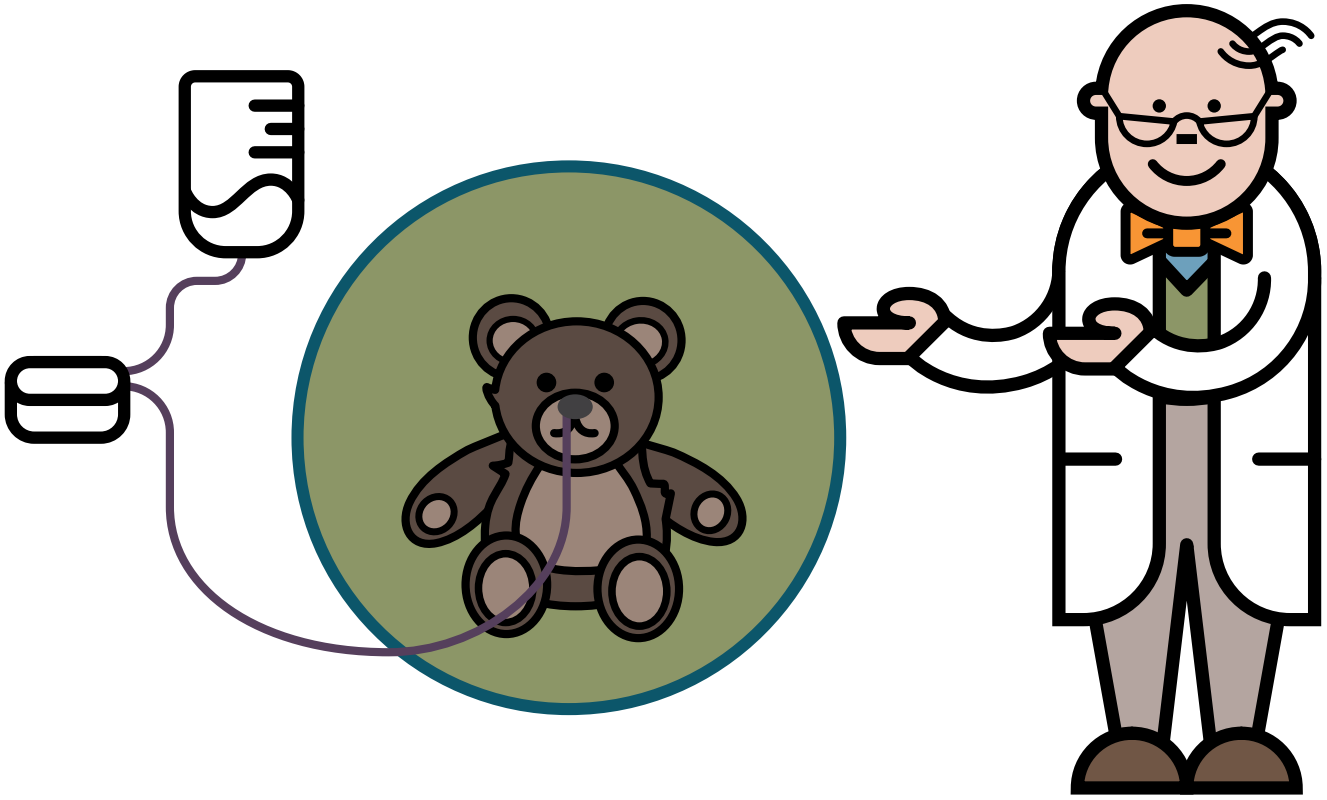


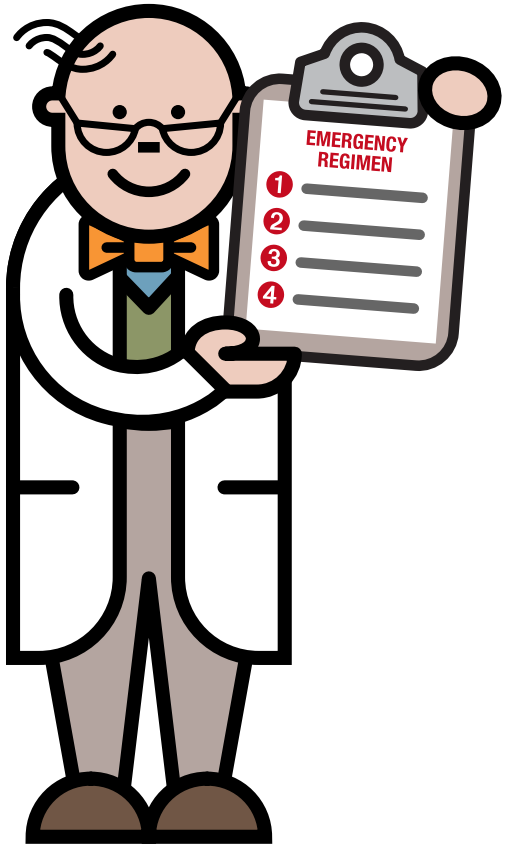
Is tube feeding needed?

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

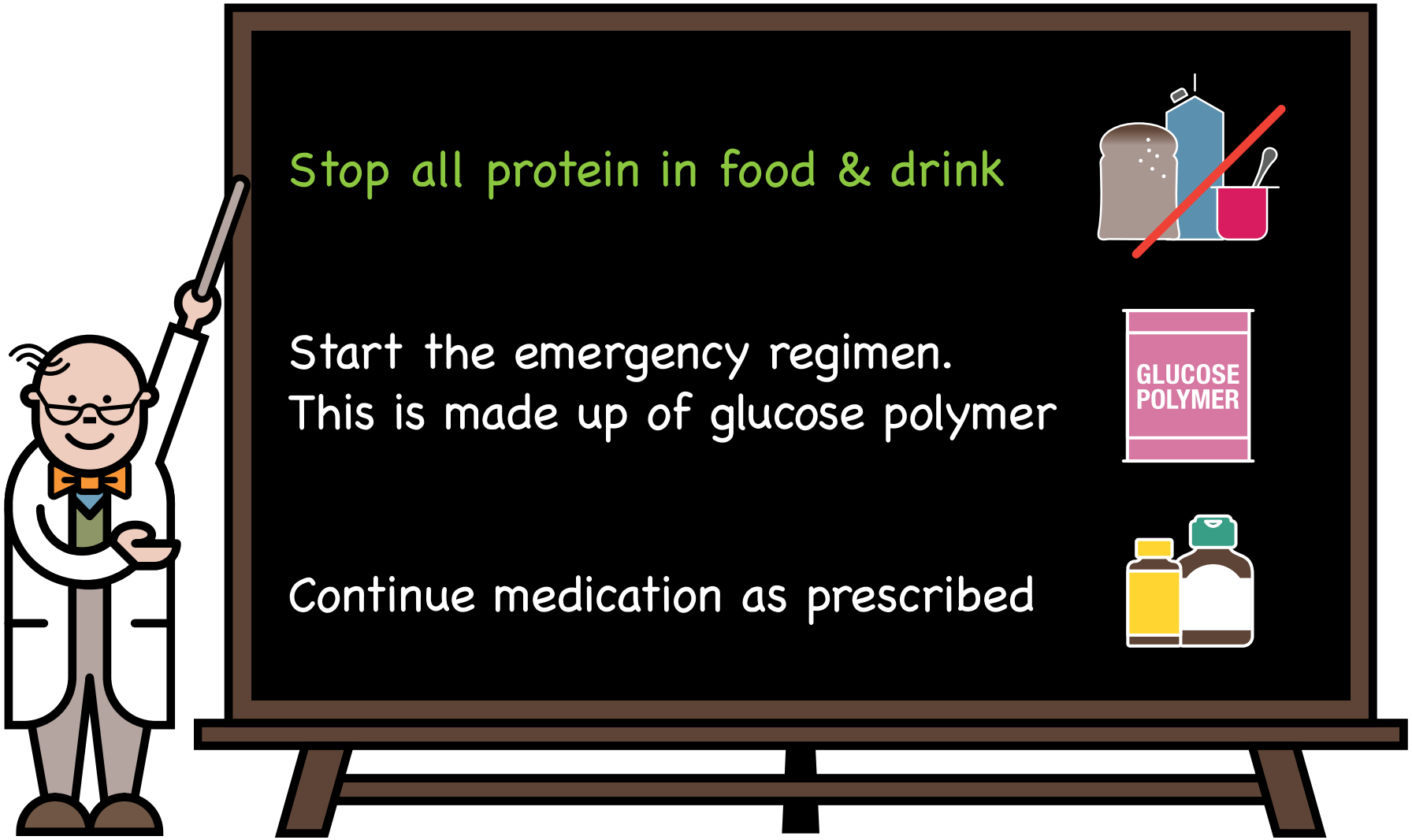


How is Argininosuccinic aciduria managed during illness?

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



How is Argininosuccinic aciduria managed during illness?



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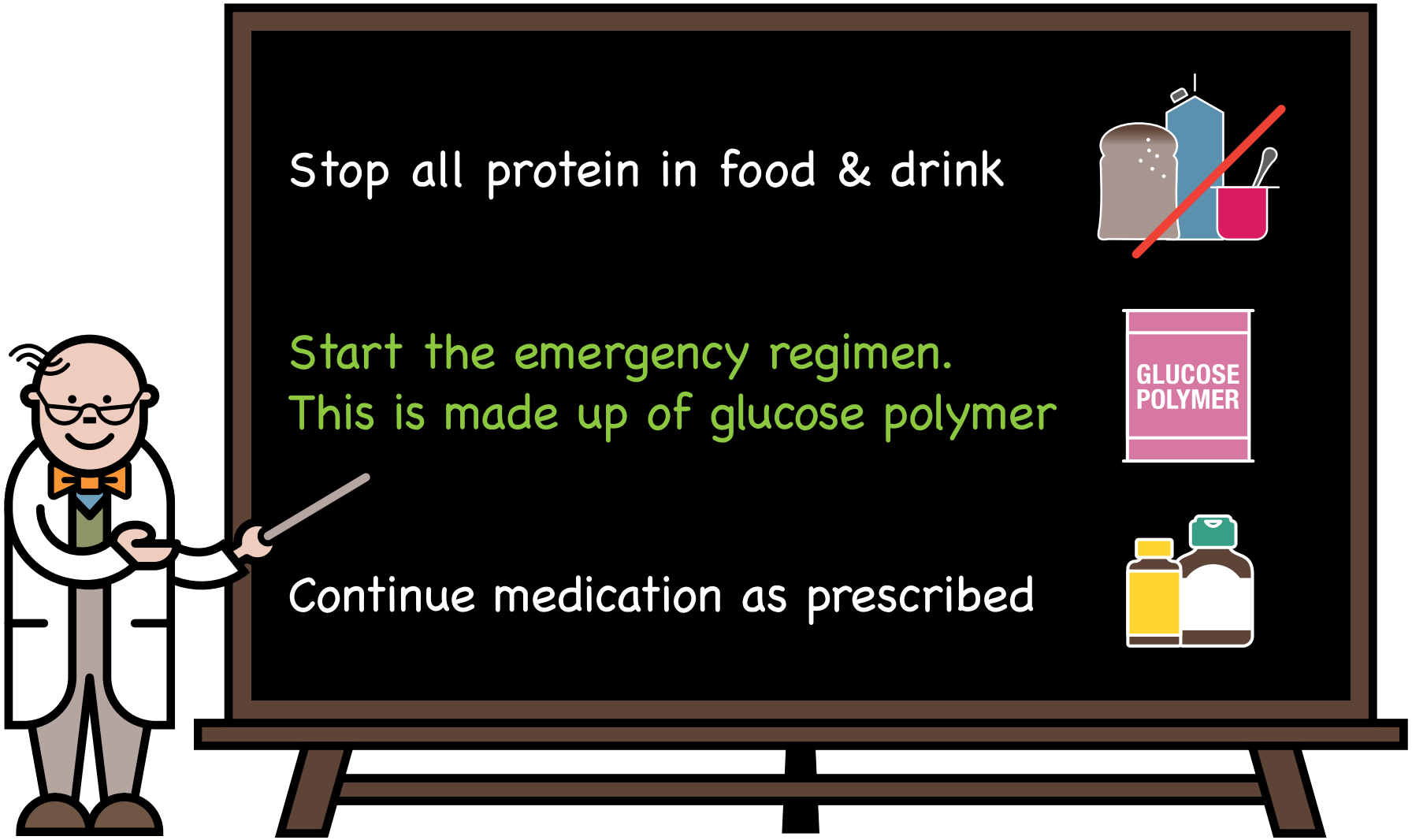

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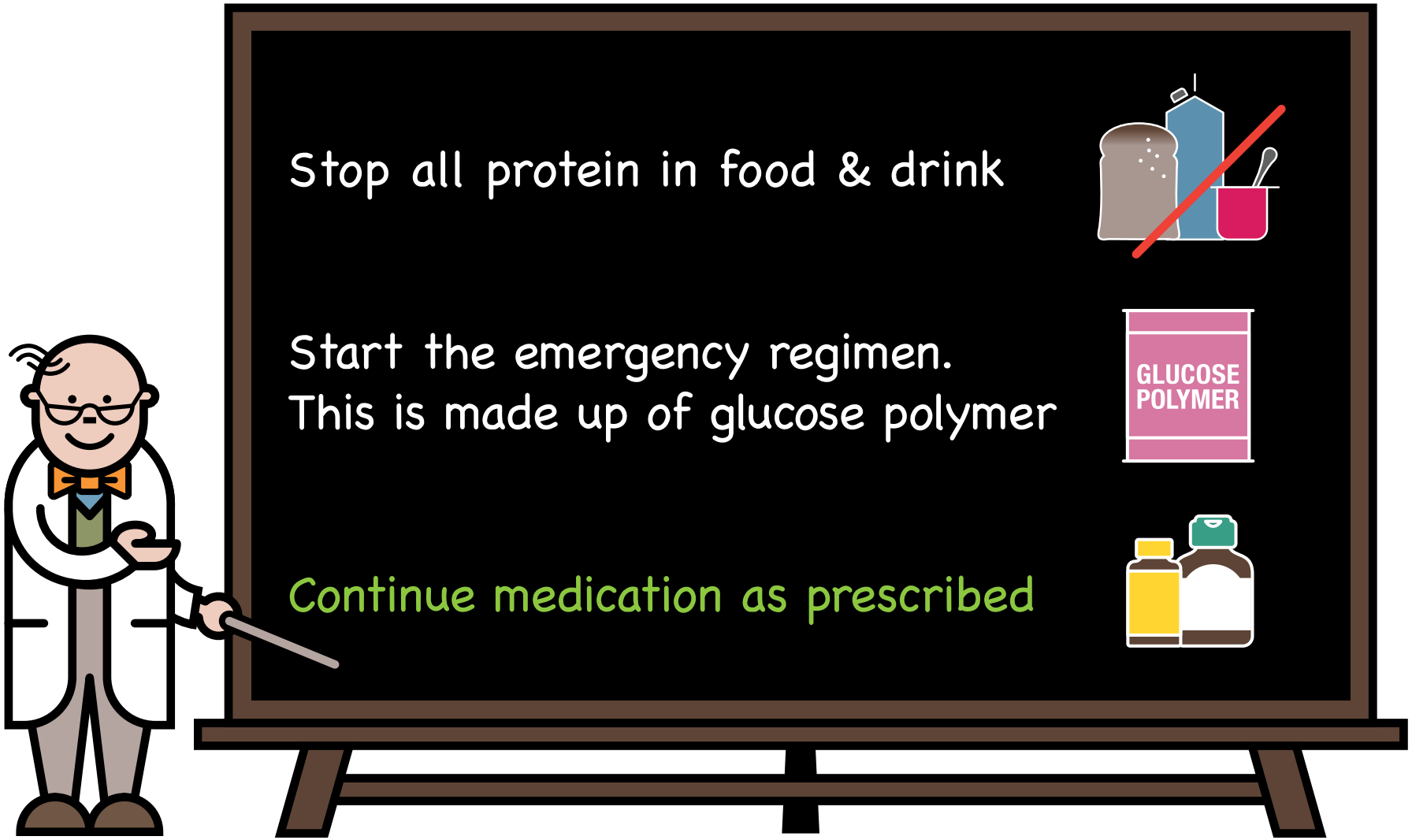
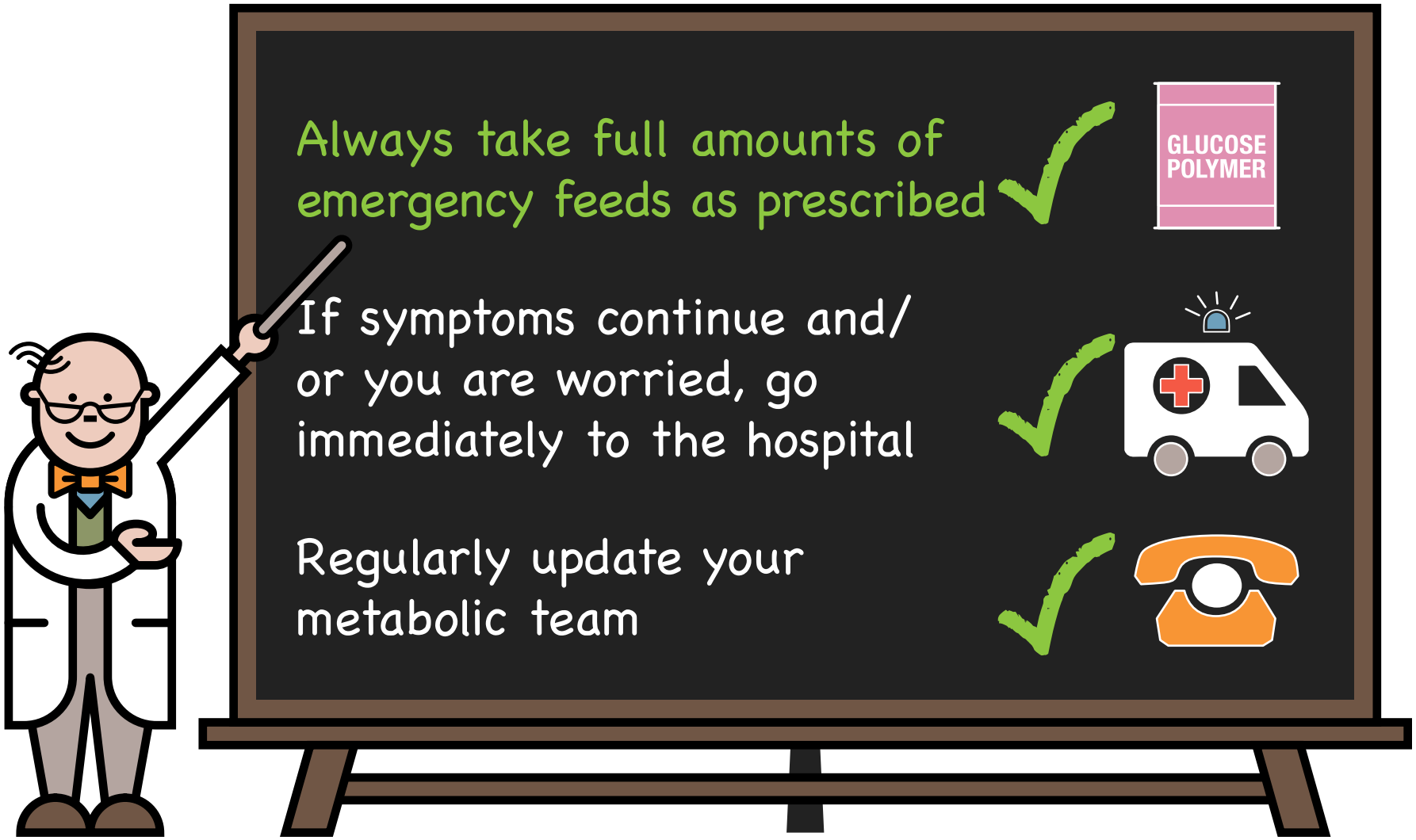


Checklist for illness



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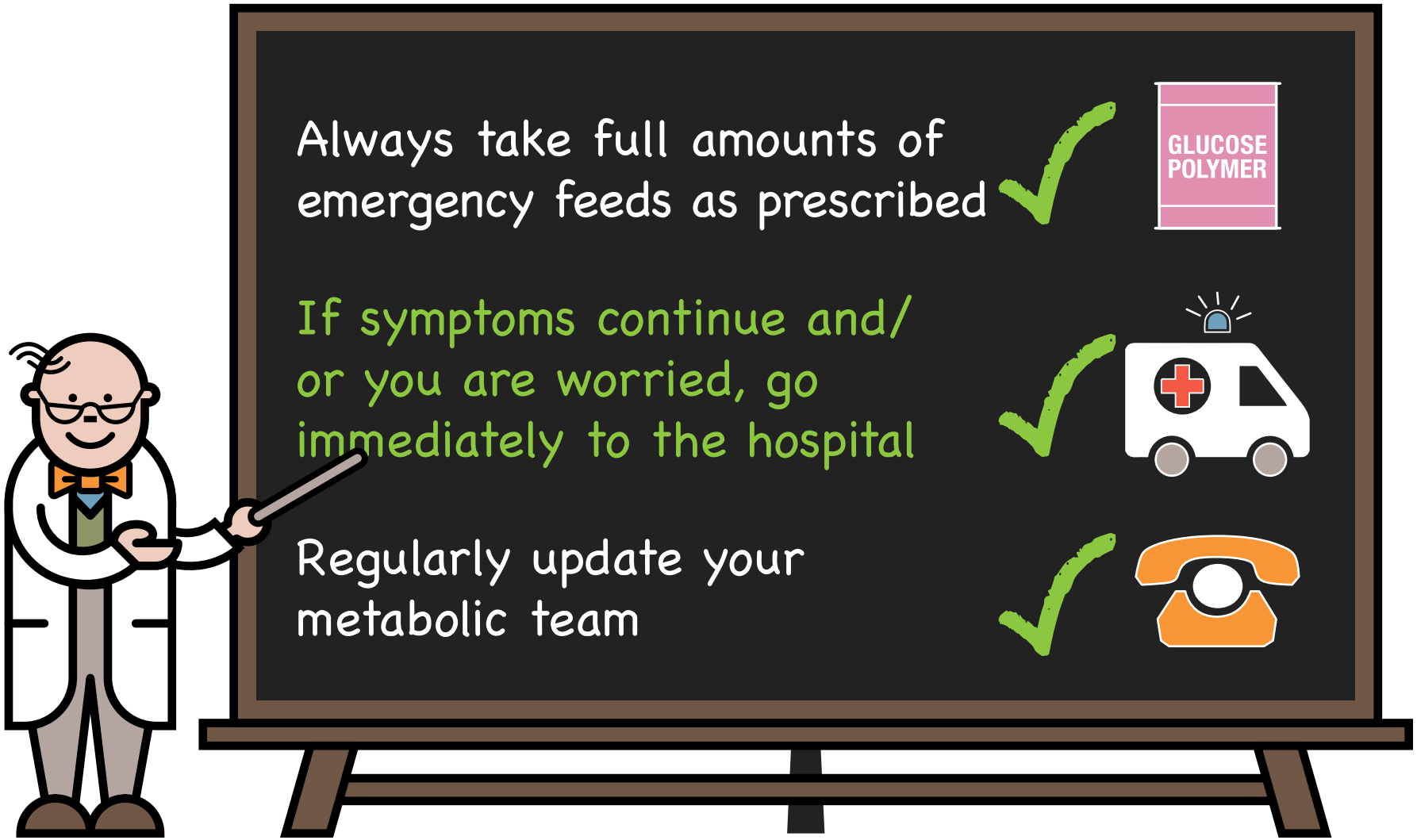

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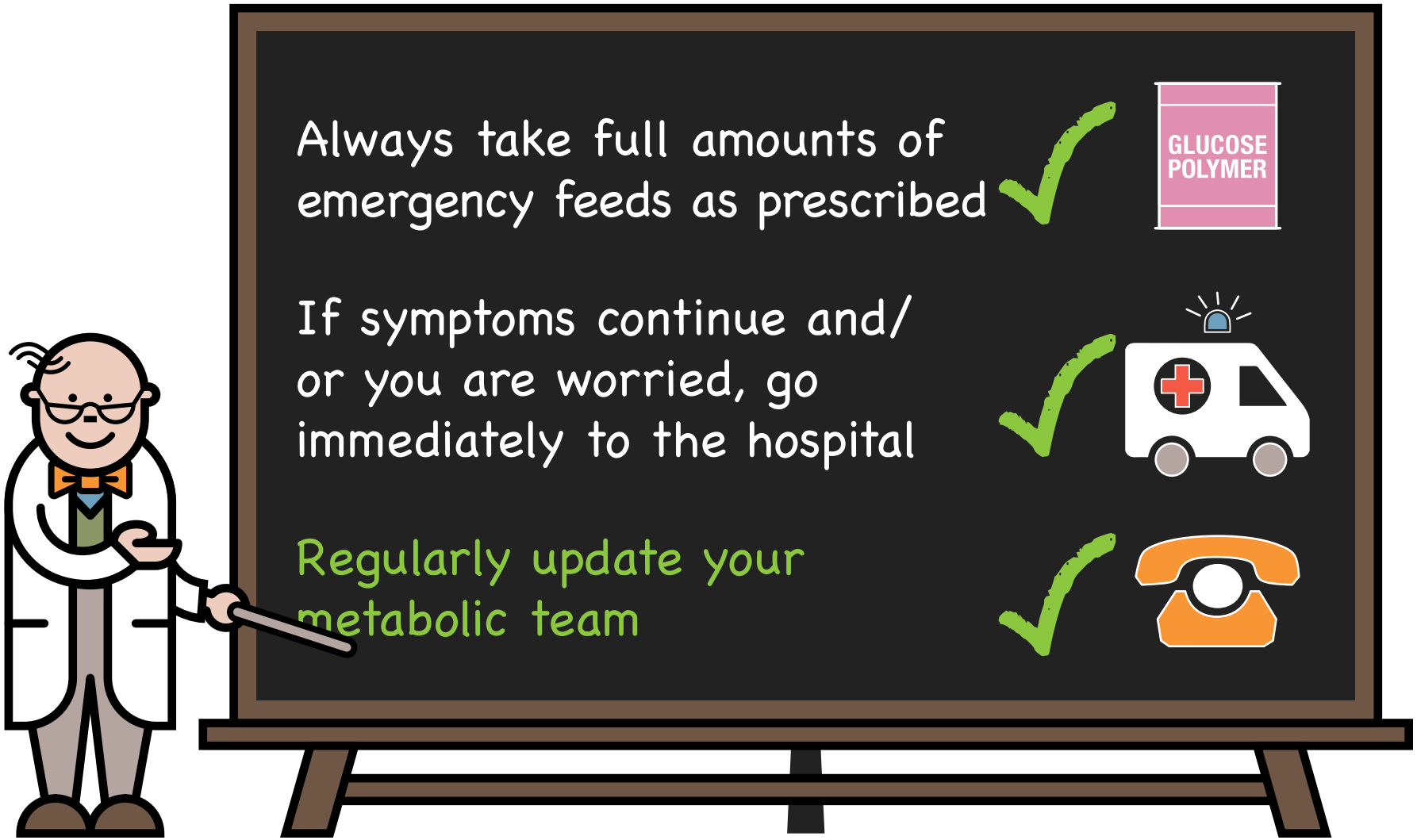
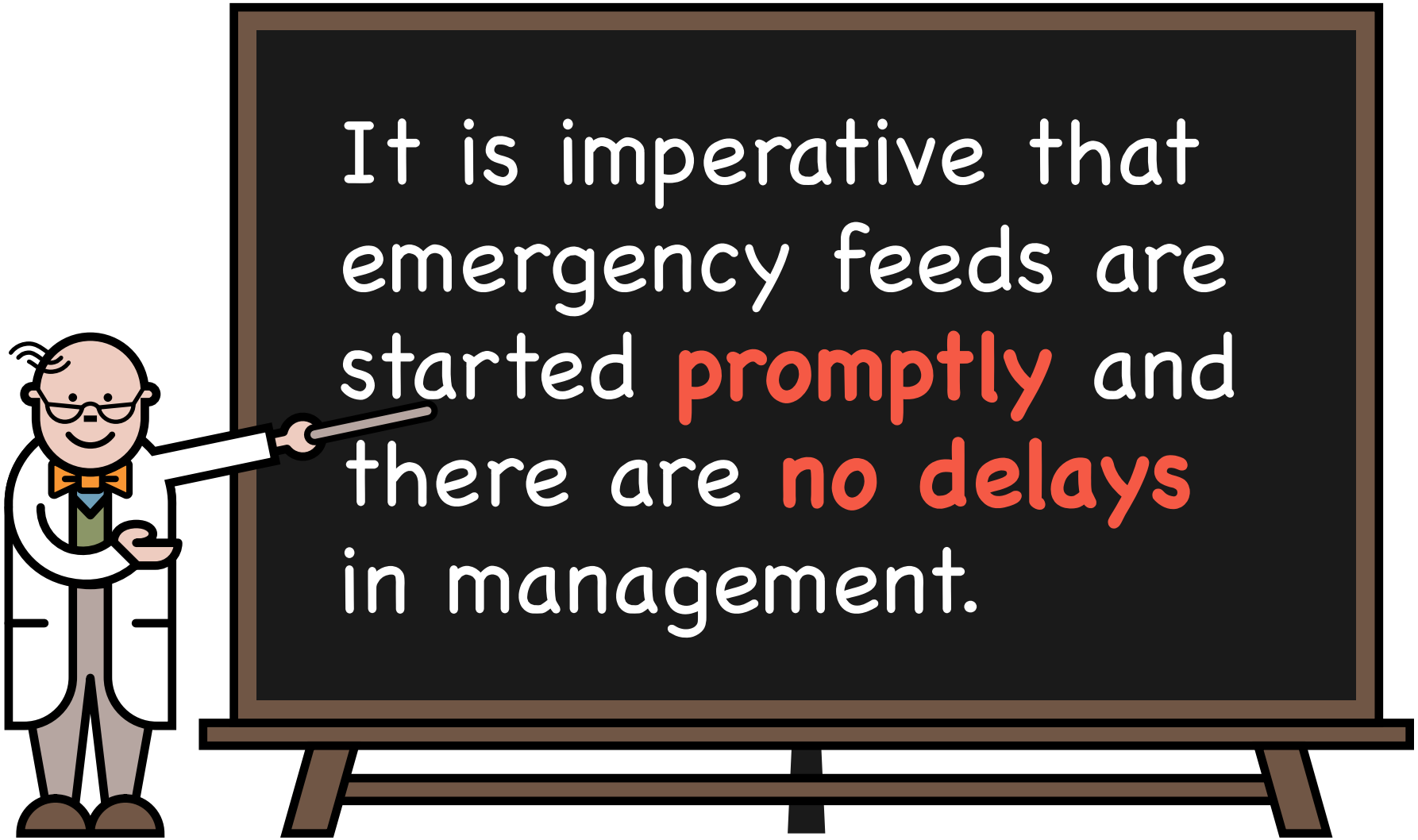
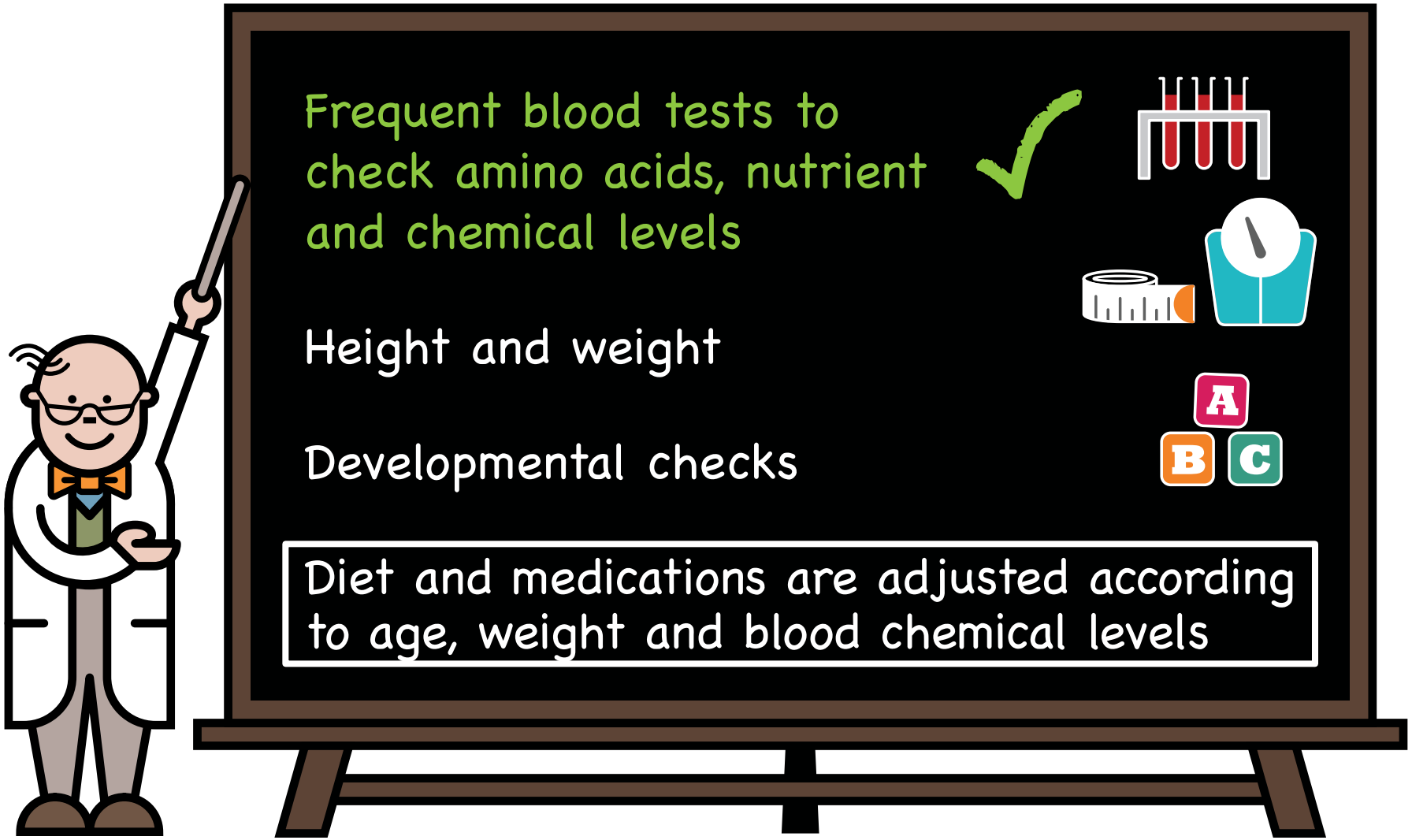


Key message



How is Argininosuccinic aciduria monitored?



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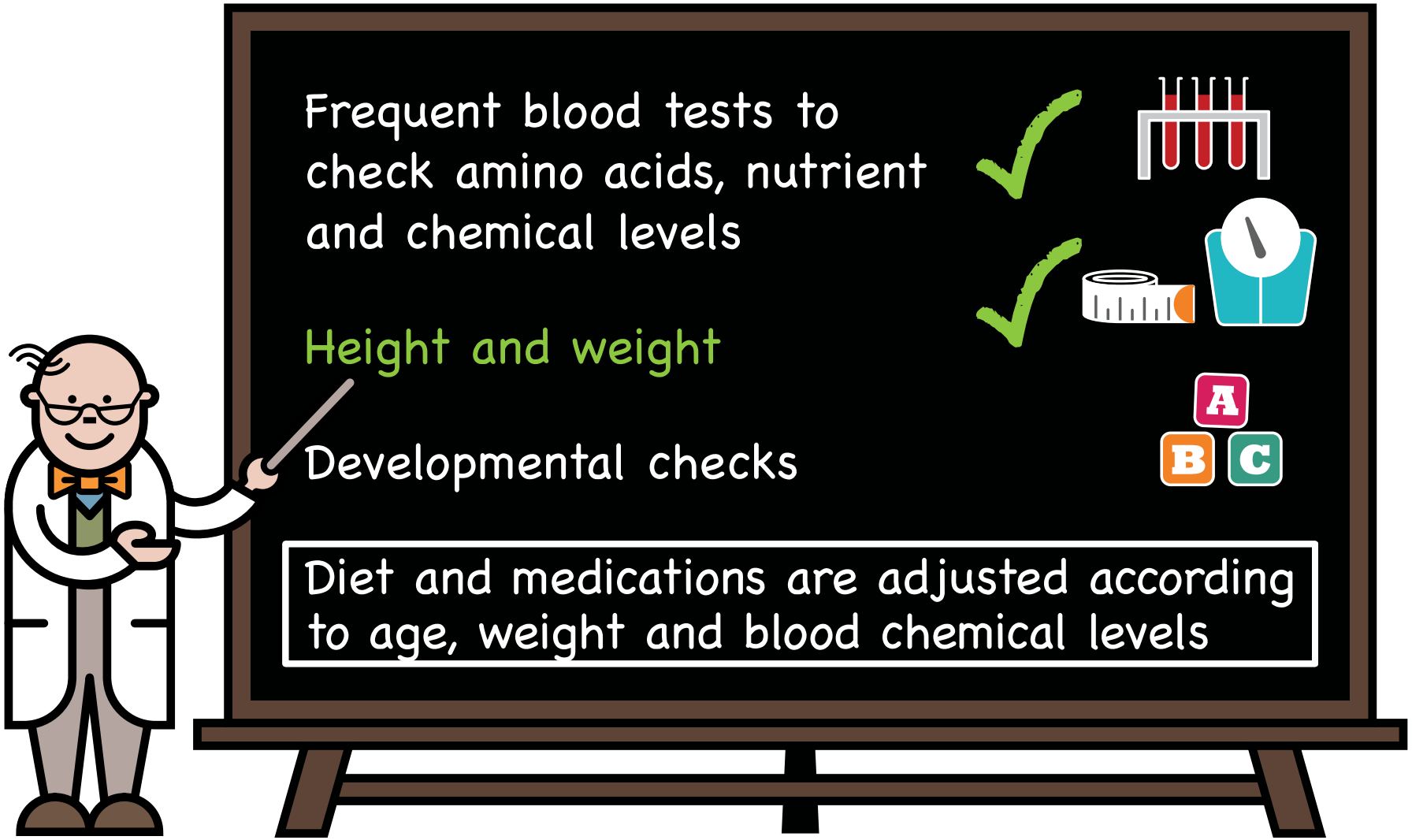

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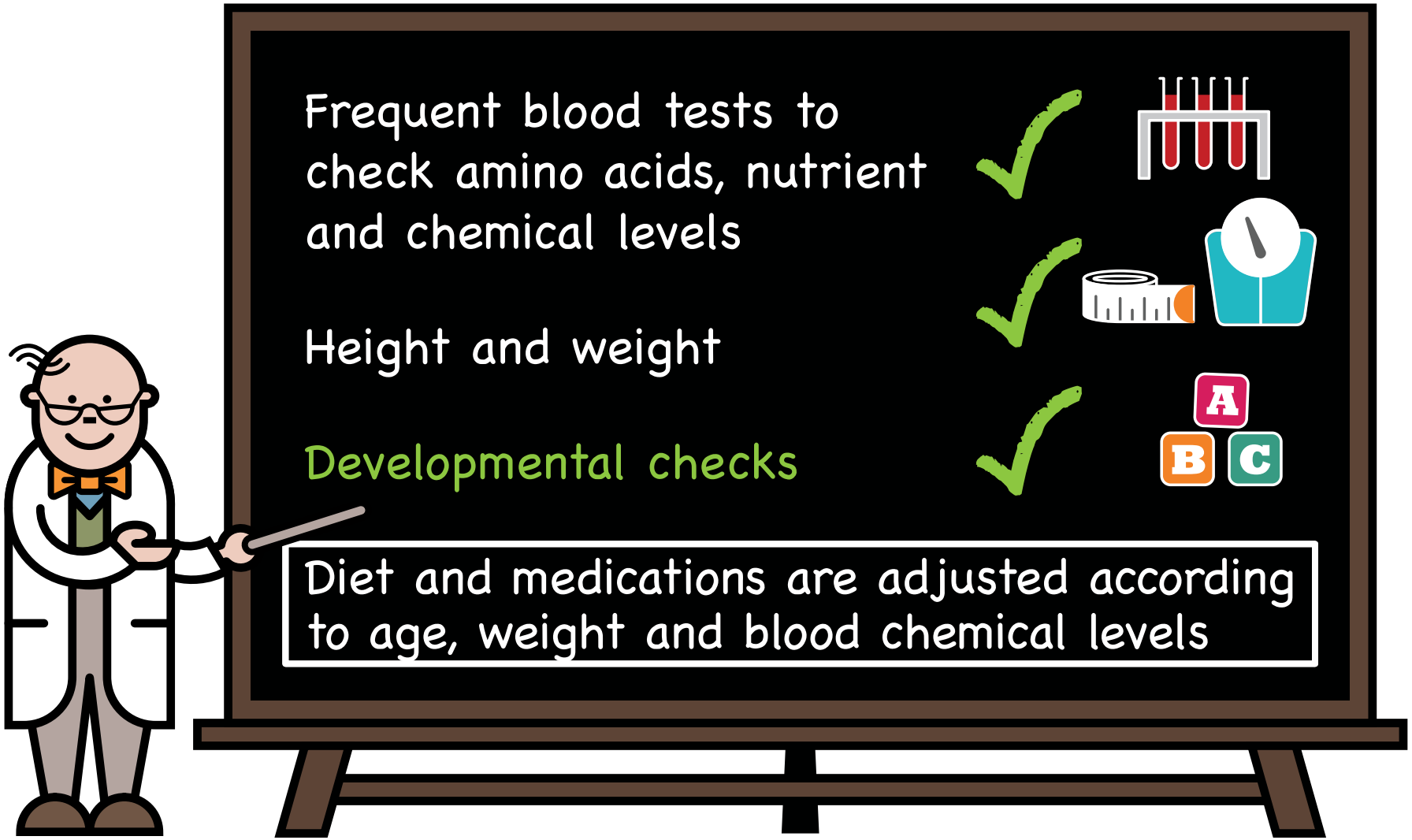
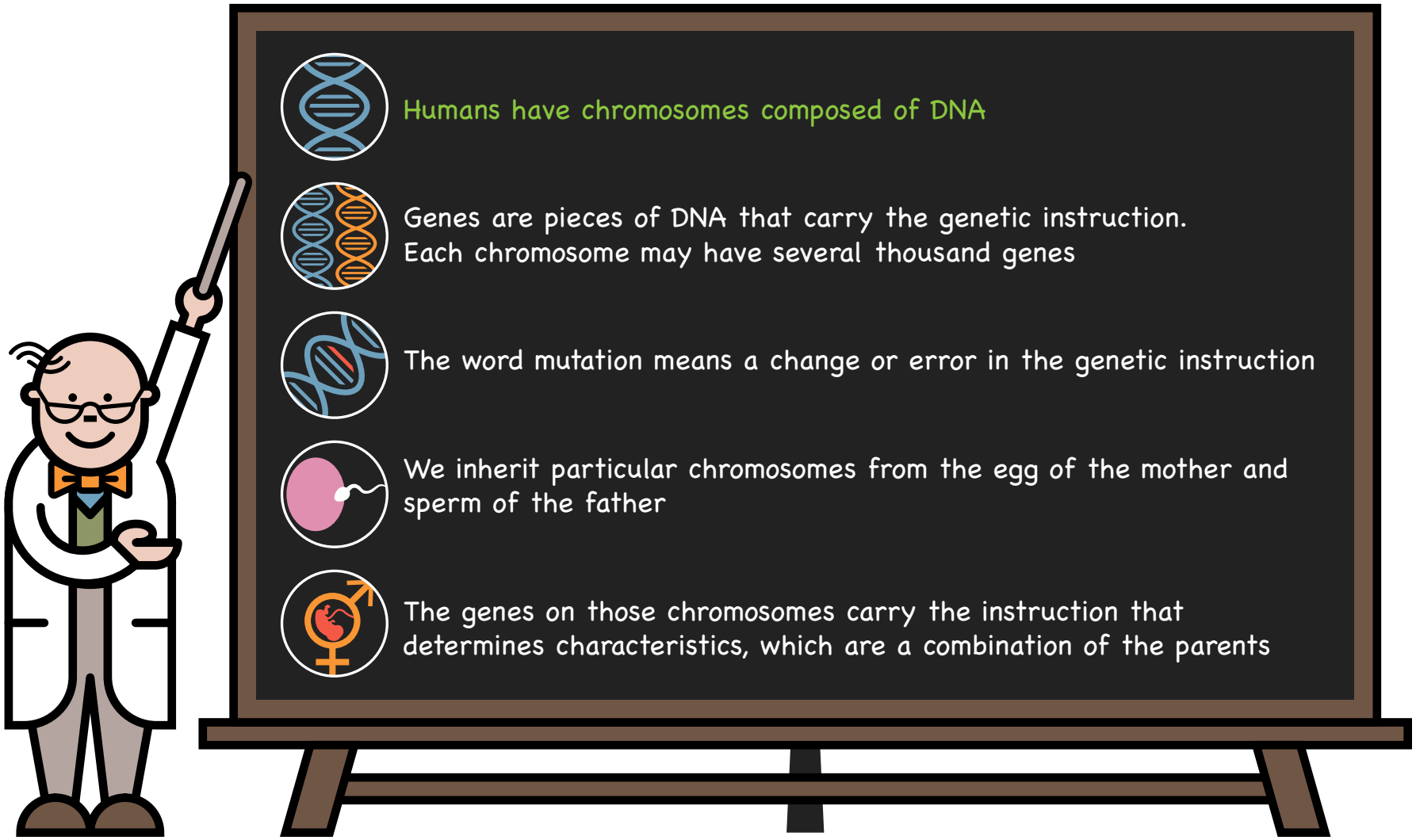


Chromosomes, genes, mutations



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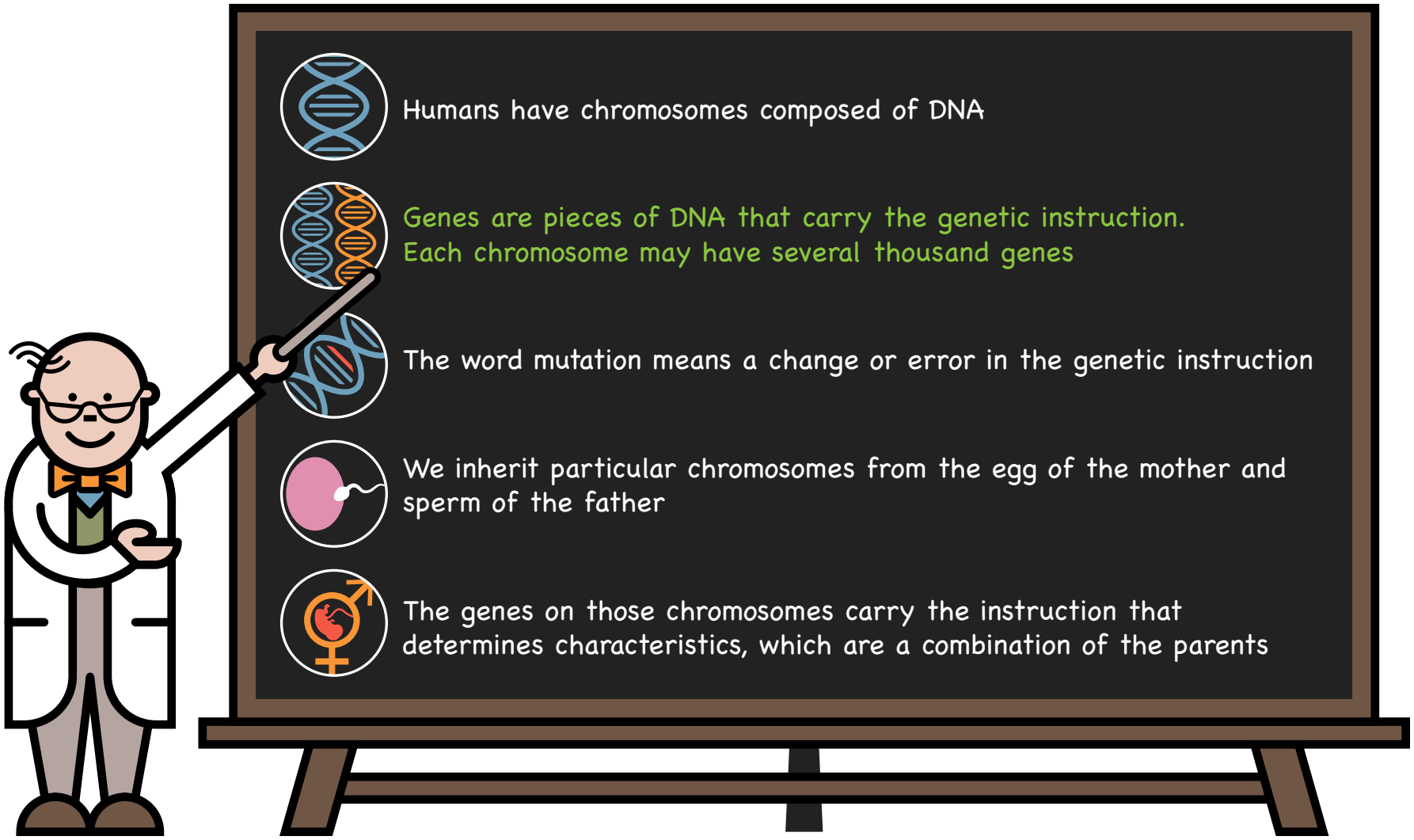

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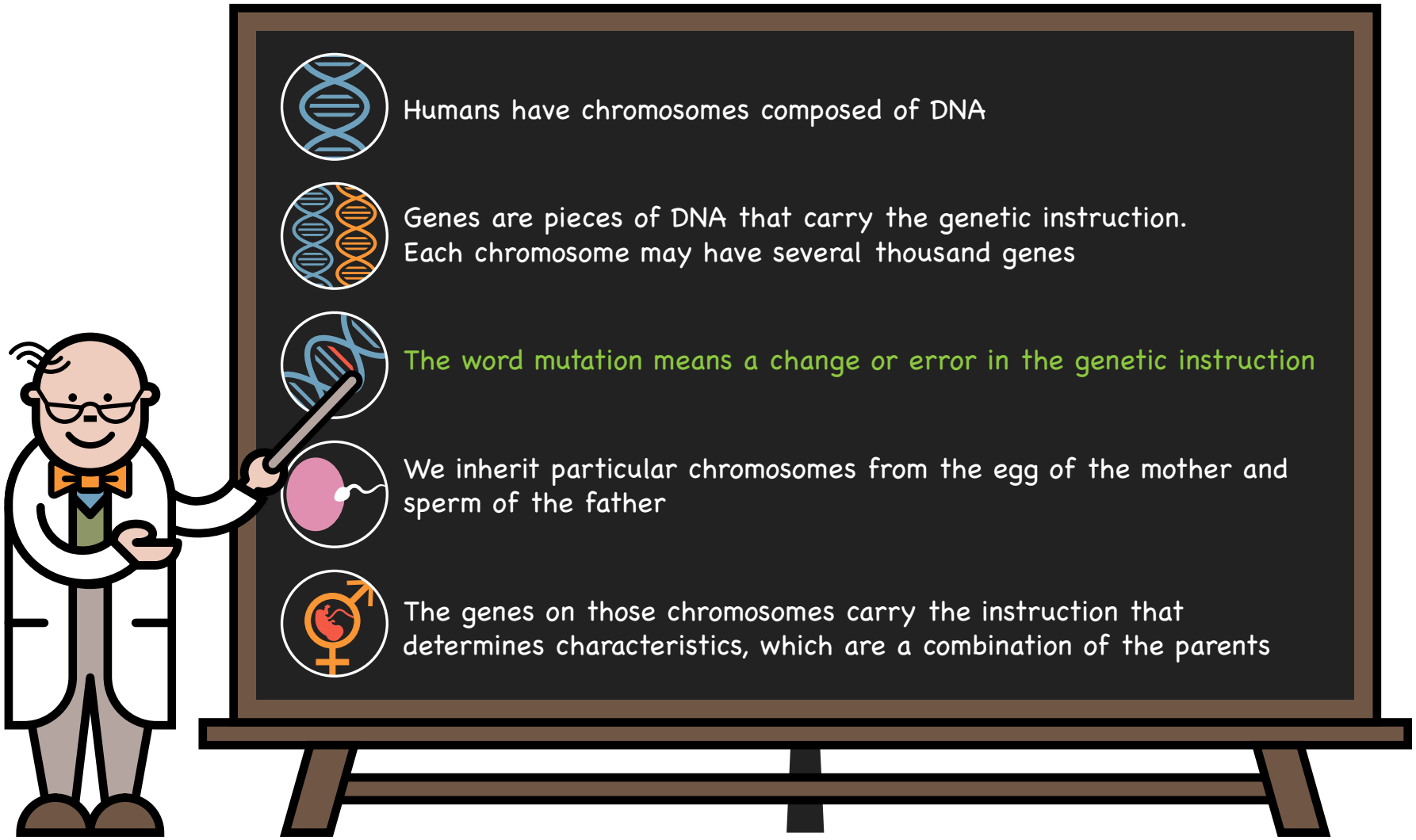

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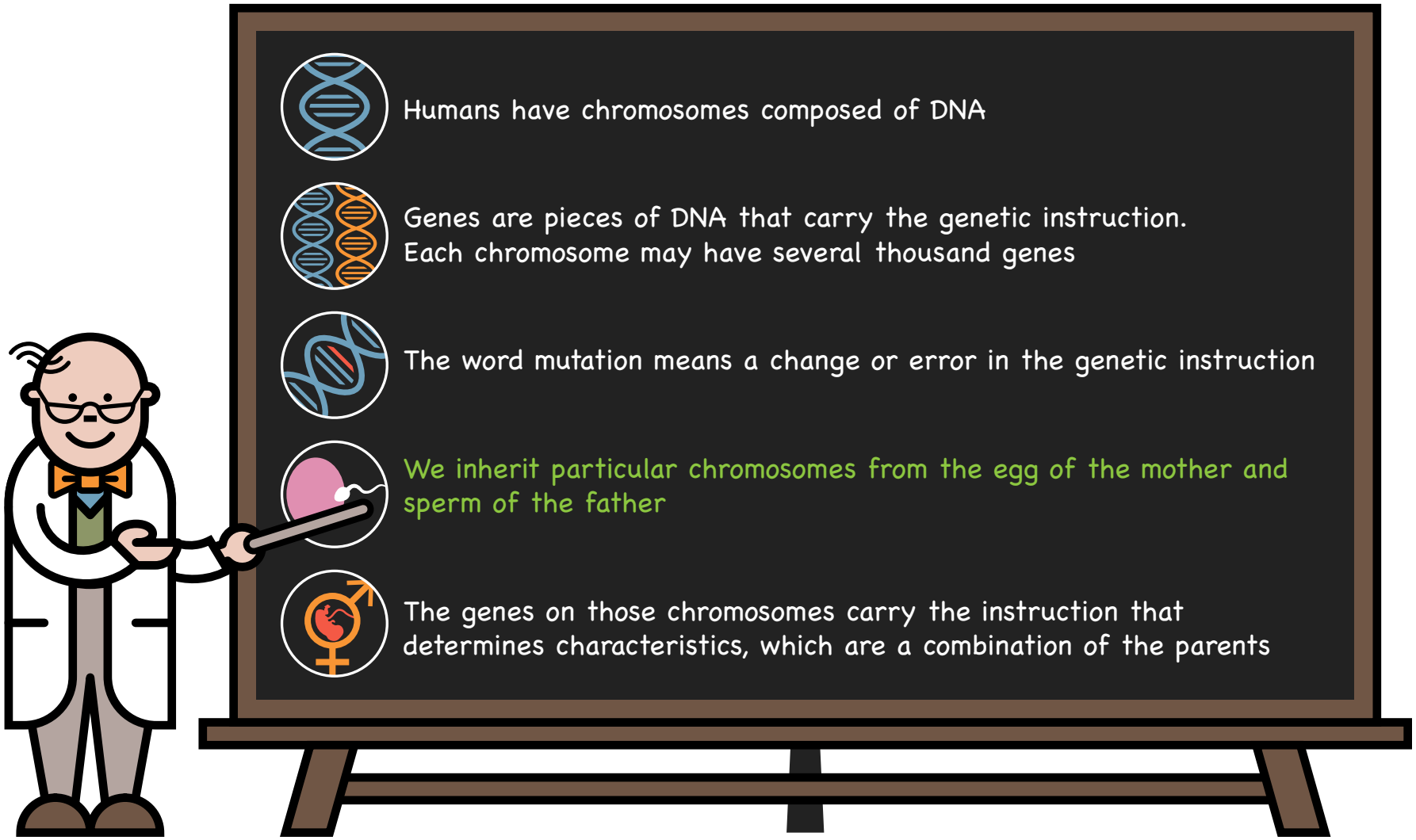

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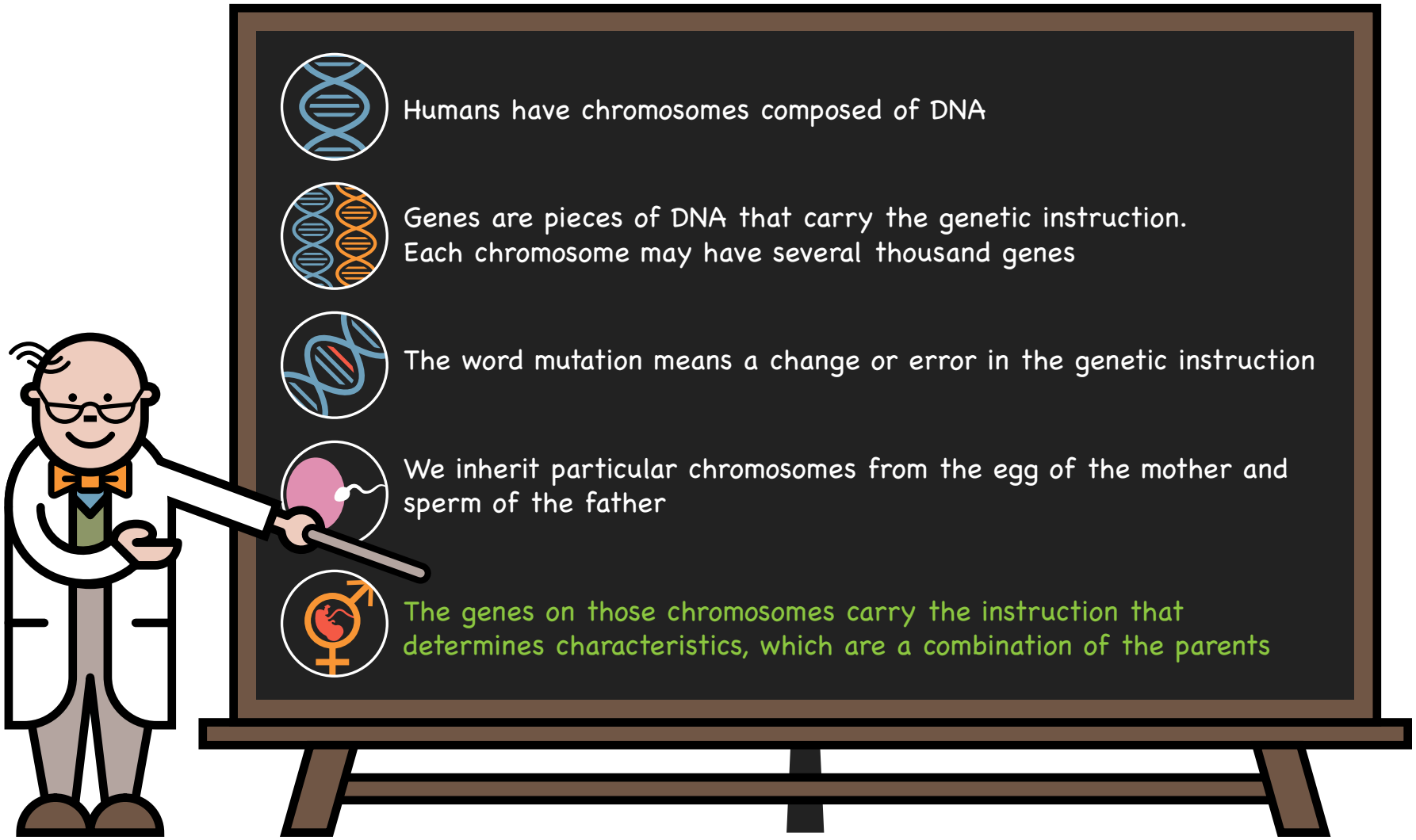
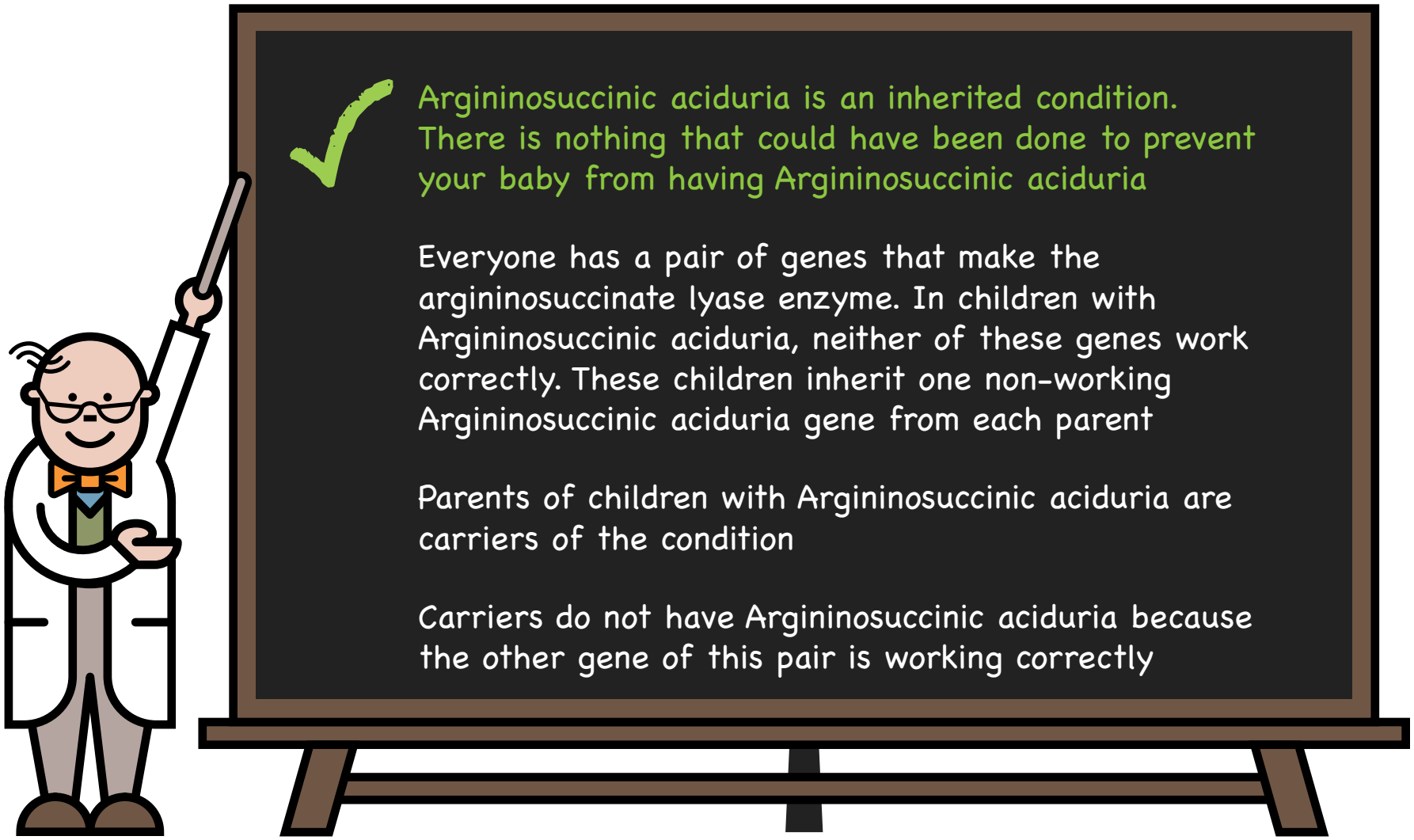


Inheritance



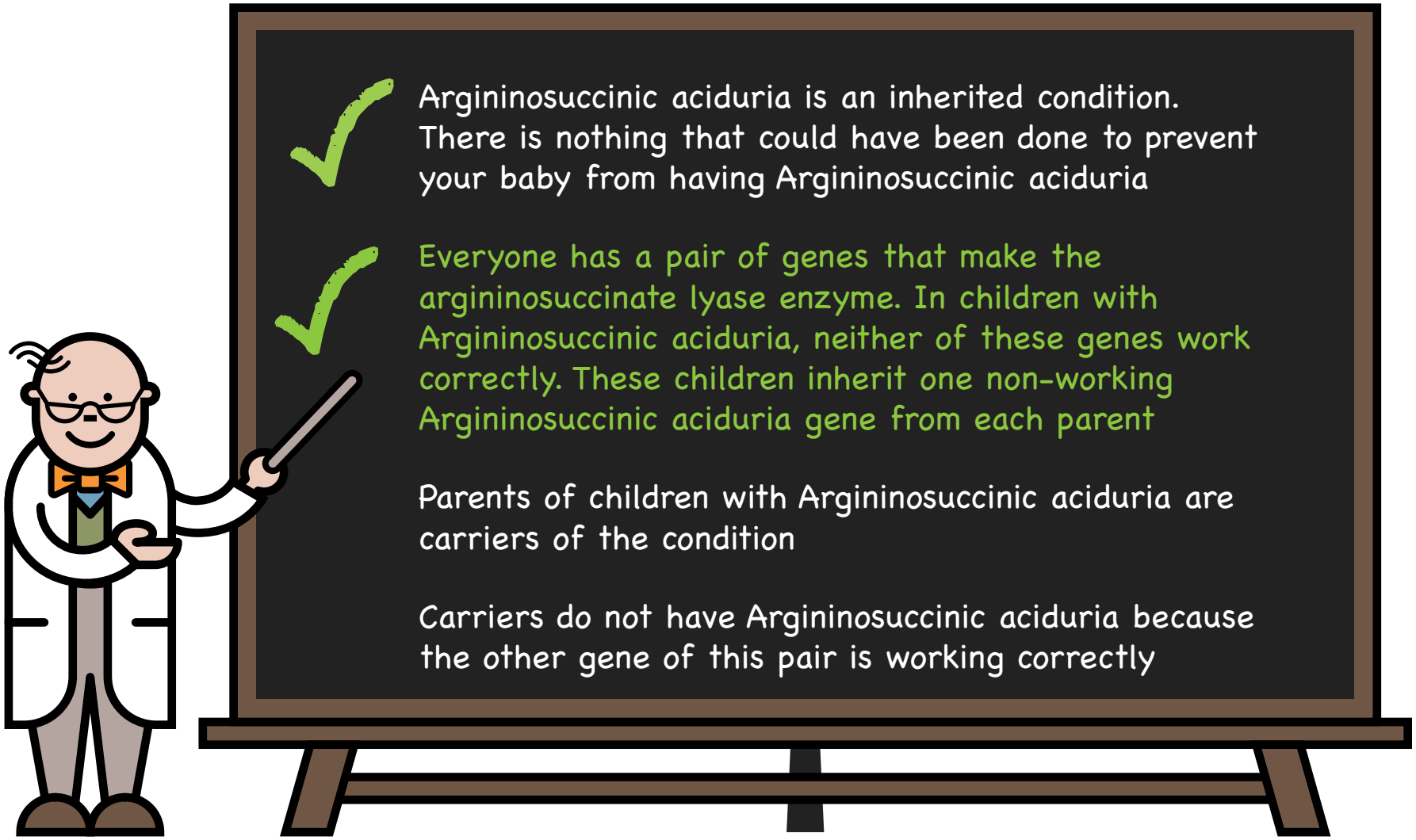
Argininosuccinic aciduria is an inherited condition. There is nothing that could have been done to prevent your baby from having Argininosuccinic aciduria

Everyone has a pair of genes that make the argininosuccinate lyase enzyme. In children with Argininosuccinic aciduria, neither of these genes work correctly. These children inherit one non-working Argininosuccinic aciduria gene from each parent

Parents of children with Argininosuccinic aciduria are carriers of the condition

Carriers do not have Argininosuccinic aciduria because the other gene of this pair is working correctly

Inheritance



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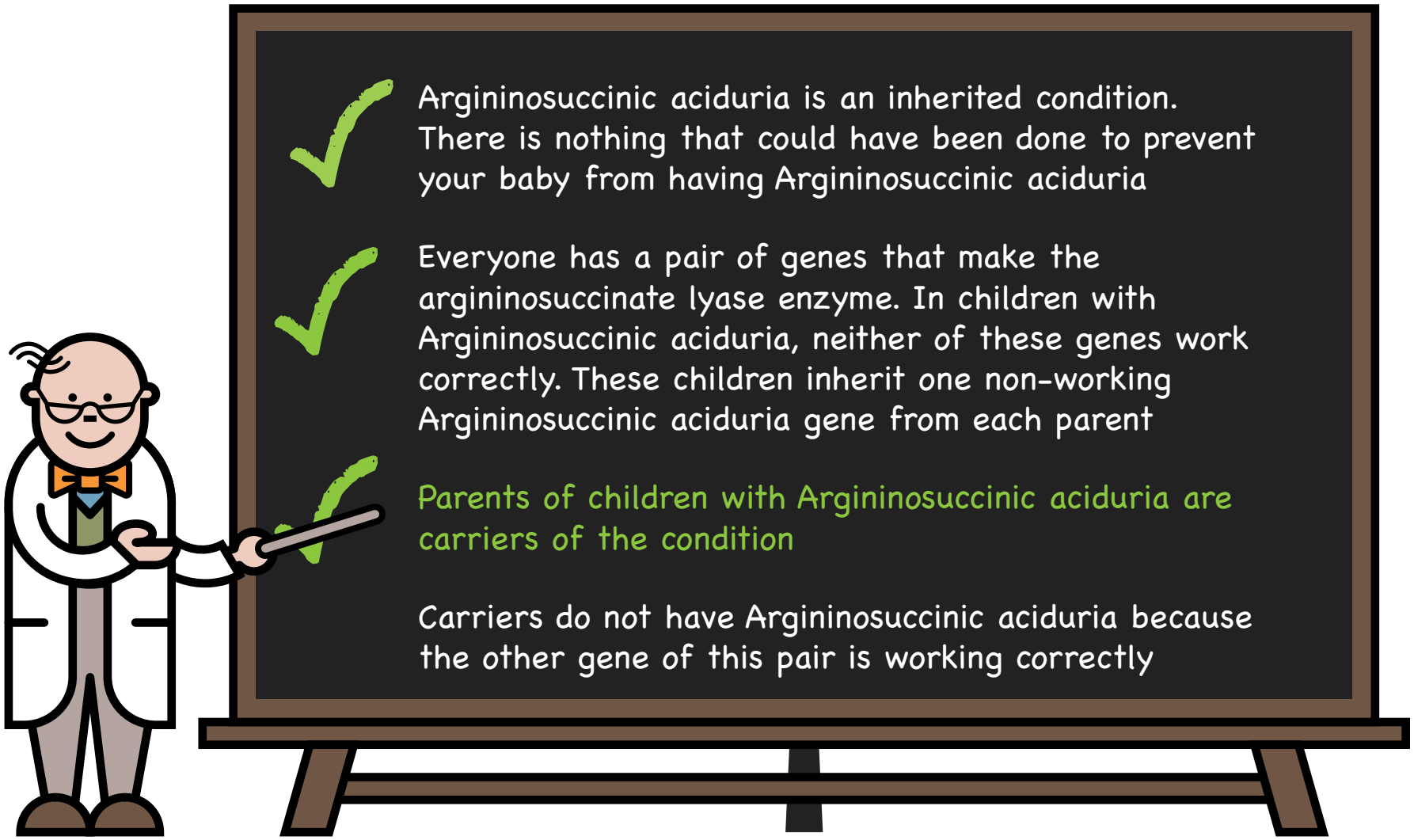
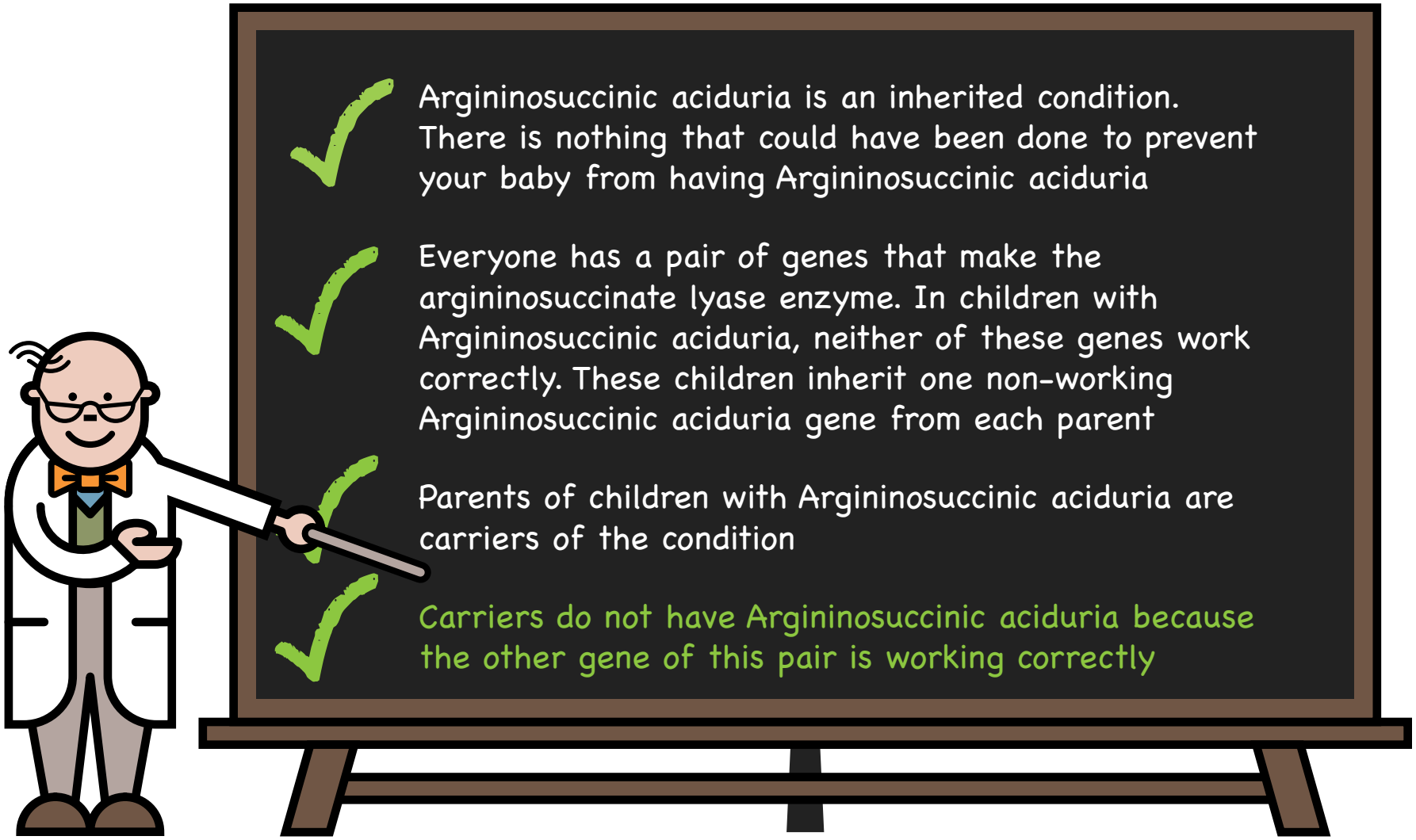
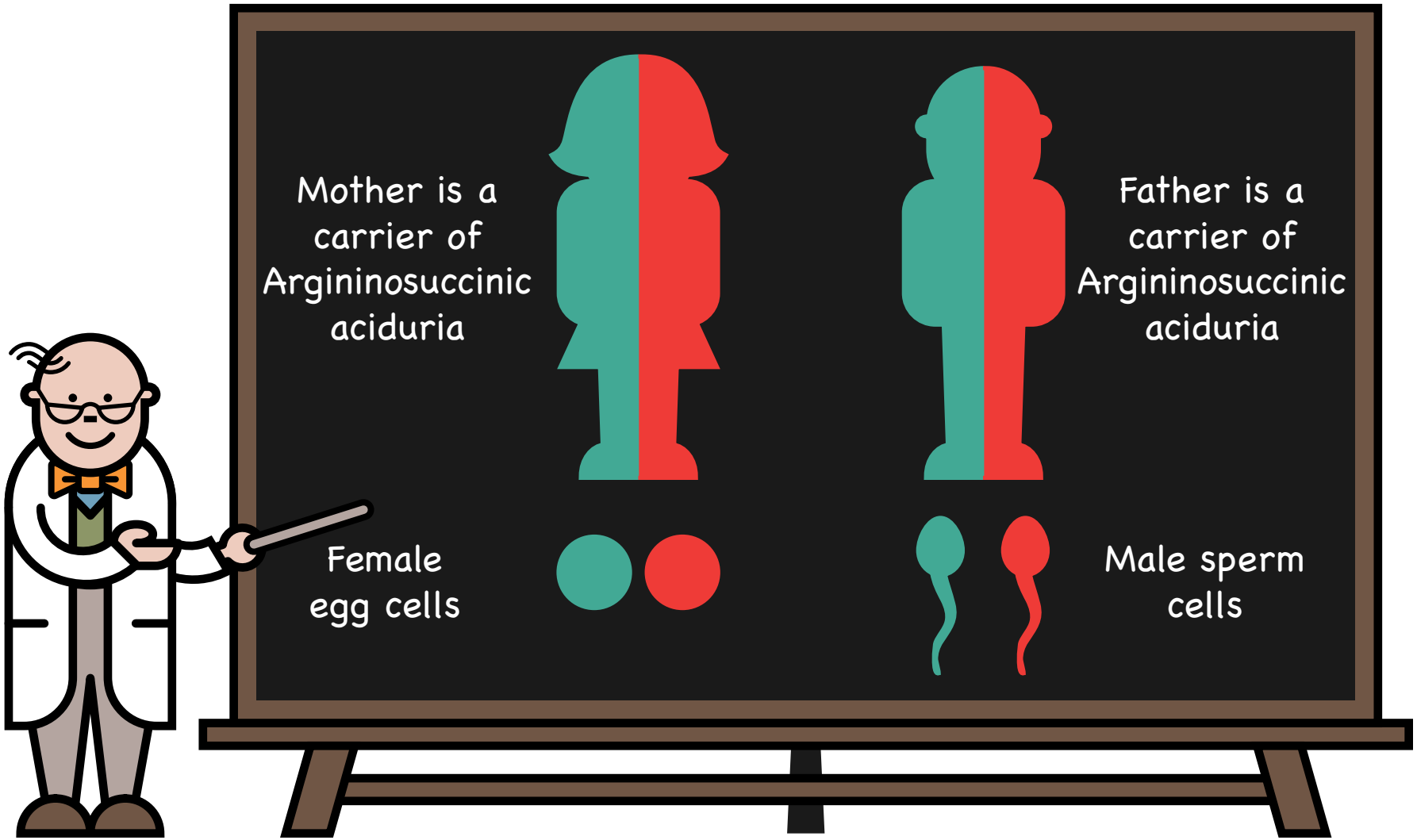


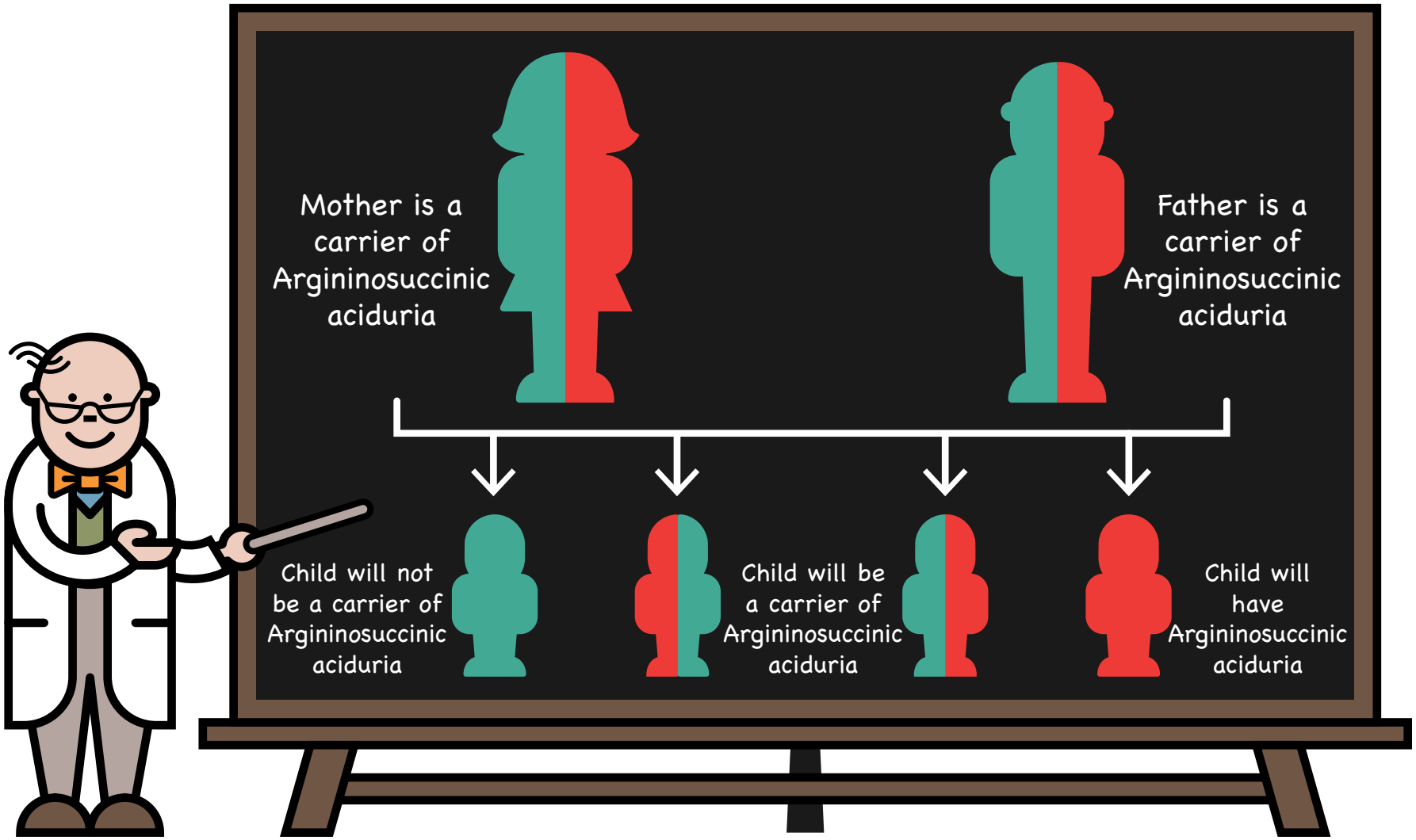
Inheritance



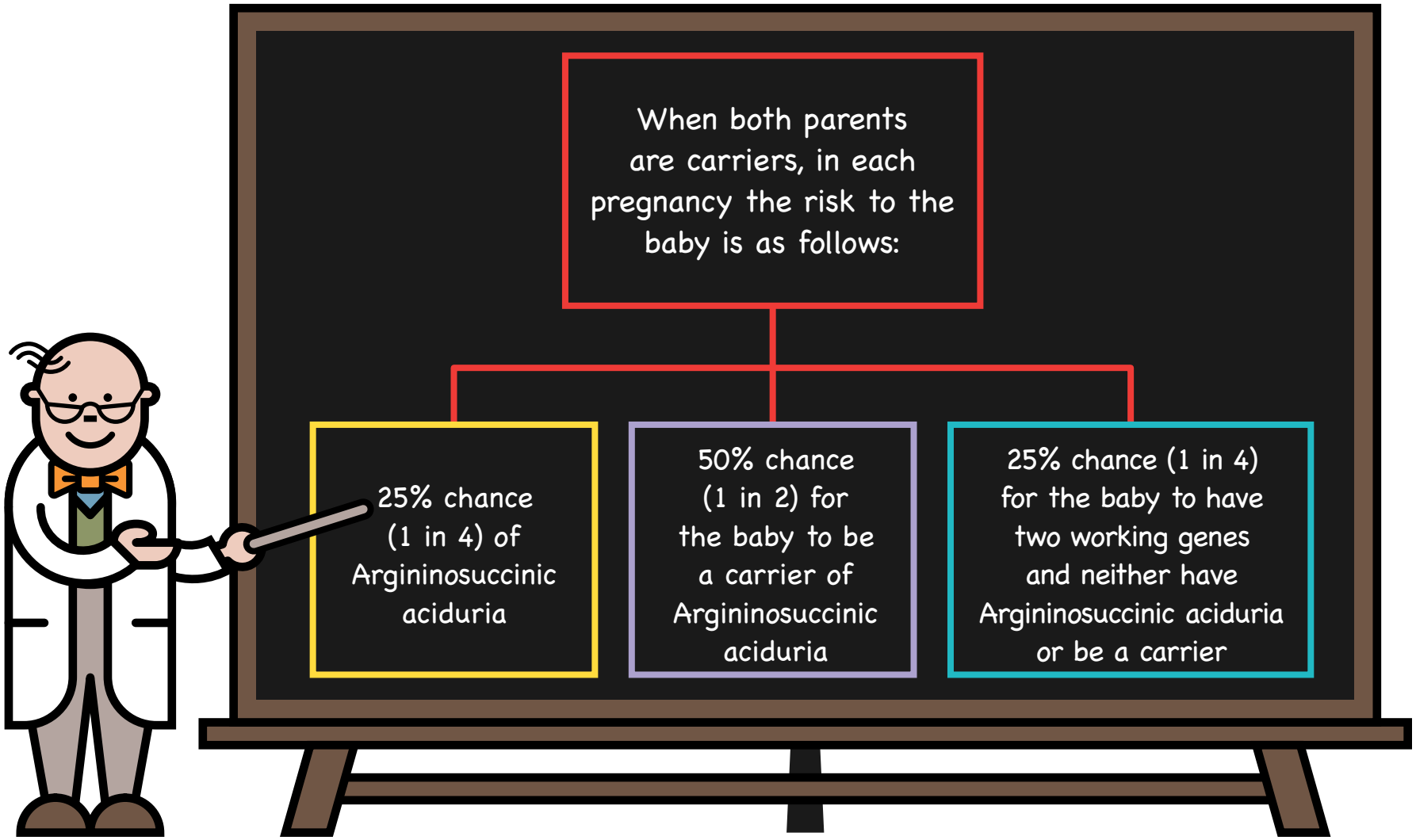
Inheritance – Autosomal recessive (carriers of Argininosuccinic aciduria)



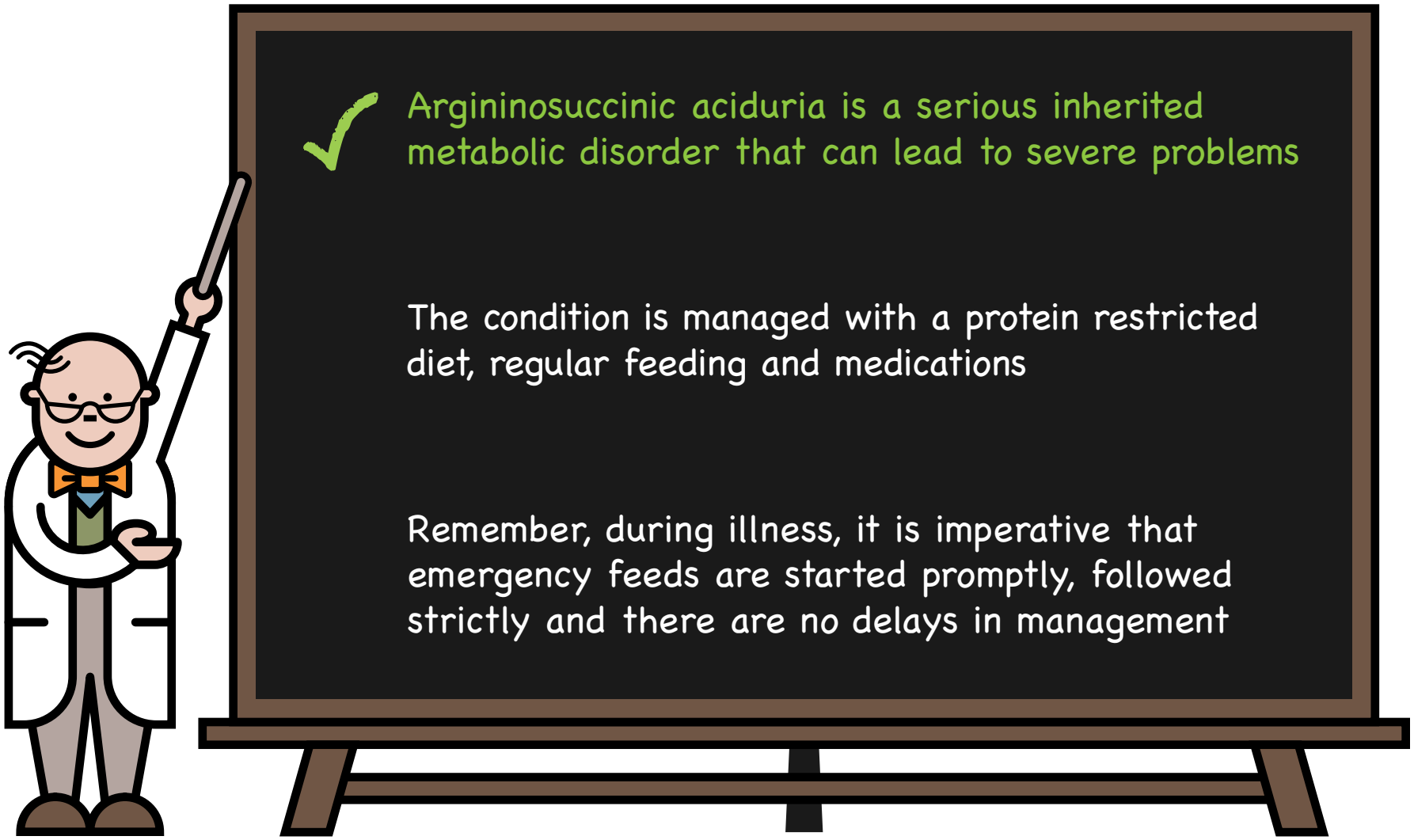
Inheritance – Autosomal recessive – possible combinations



Future pregnancies



Take home messages



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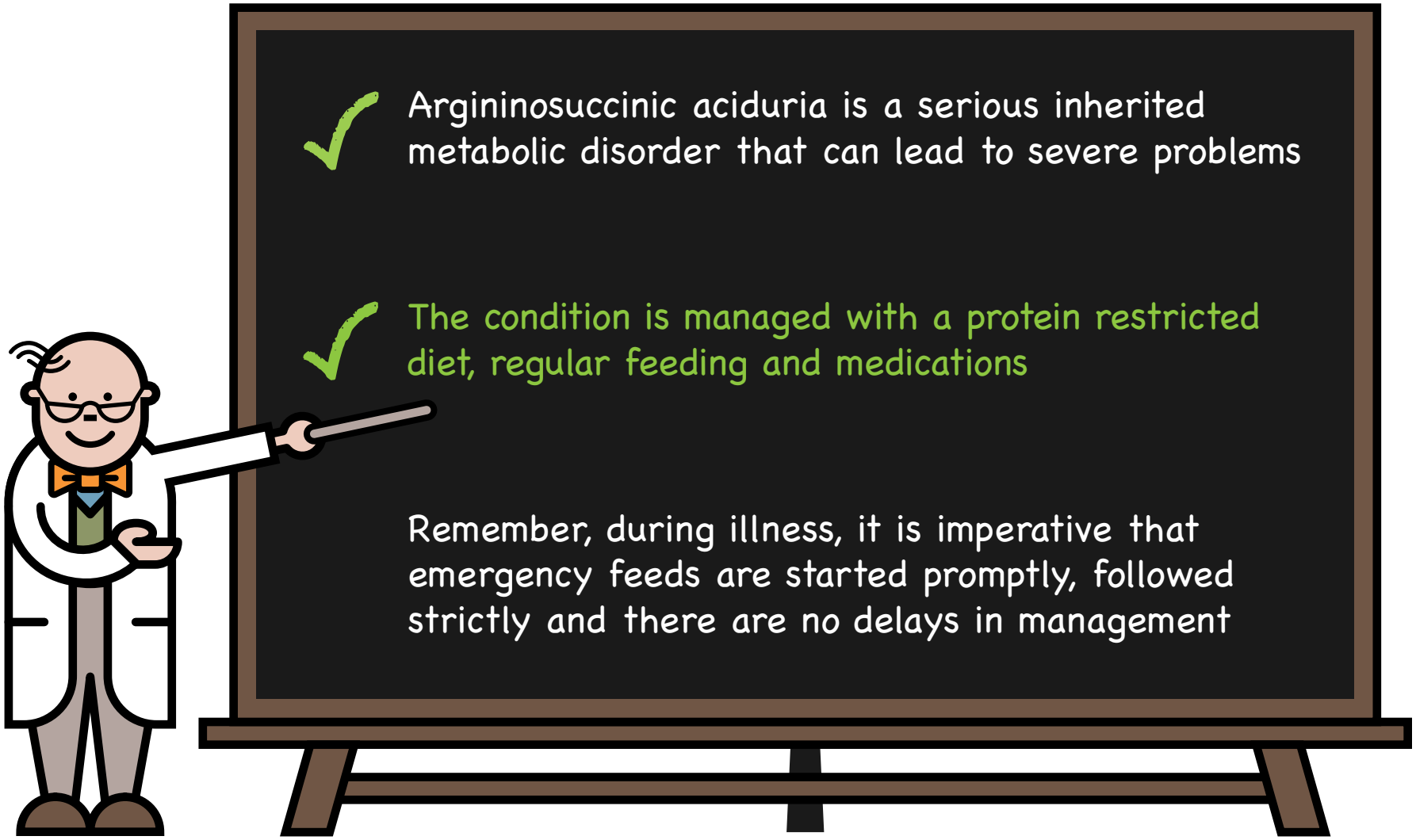

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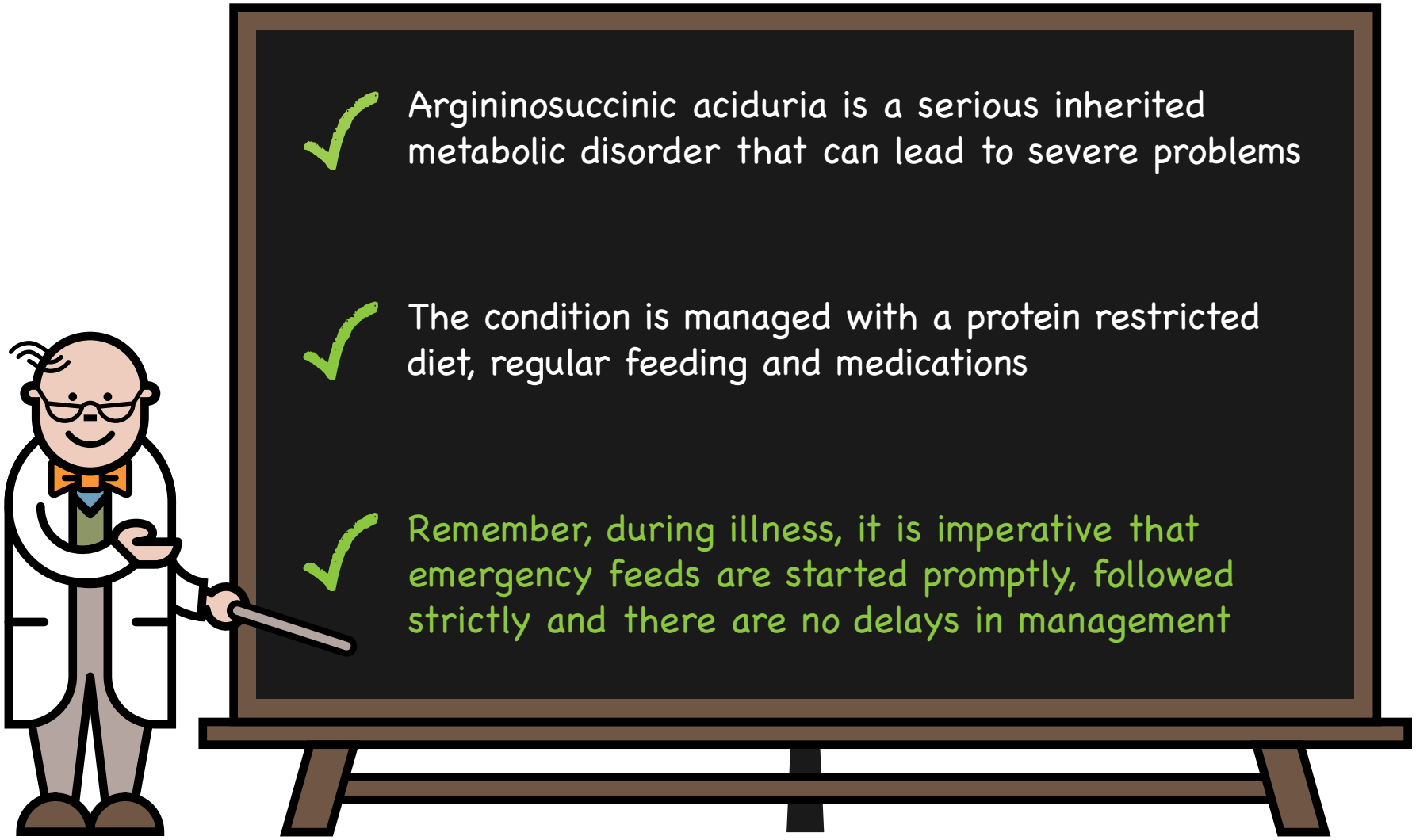

Helpful hints

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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