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

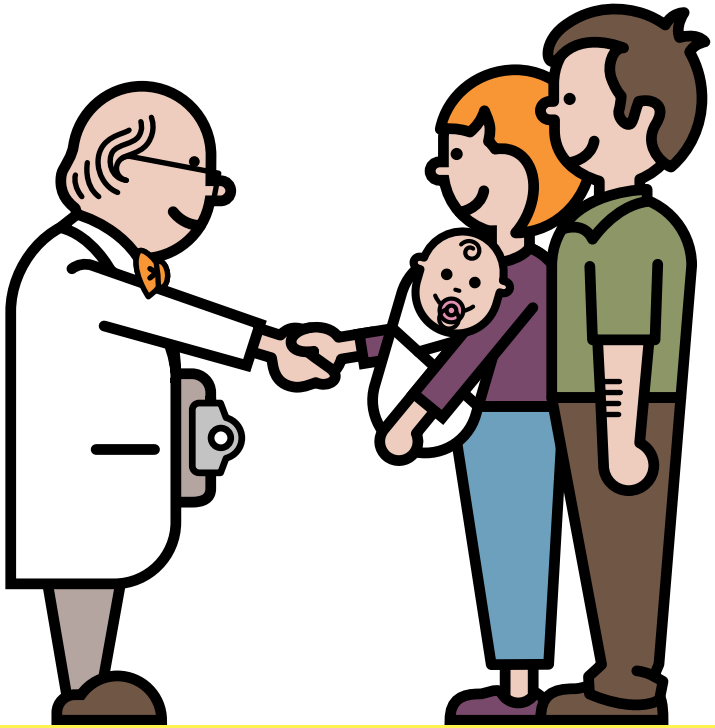
VERSION 1, OCTOBER 2017

Galactosaemia

Supported by NUTRICIA
as a service to metabolic medicine

Galactosaemia

Information for families following a new diagnosis in infancy



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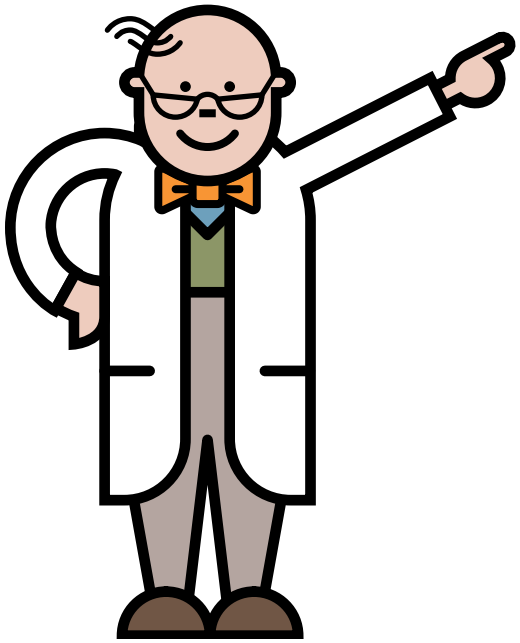
What is Galactosaemia?

It is an inherited metabolic condition.

It affects the way your baby breaks down galactose, a type of sugar found in foods.



Galactos **aemia**

(Too much)

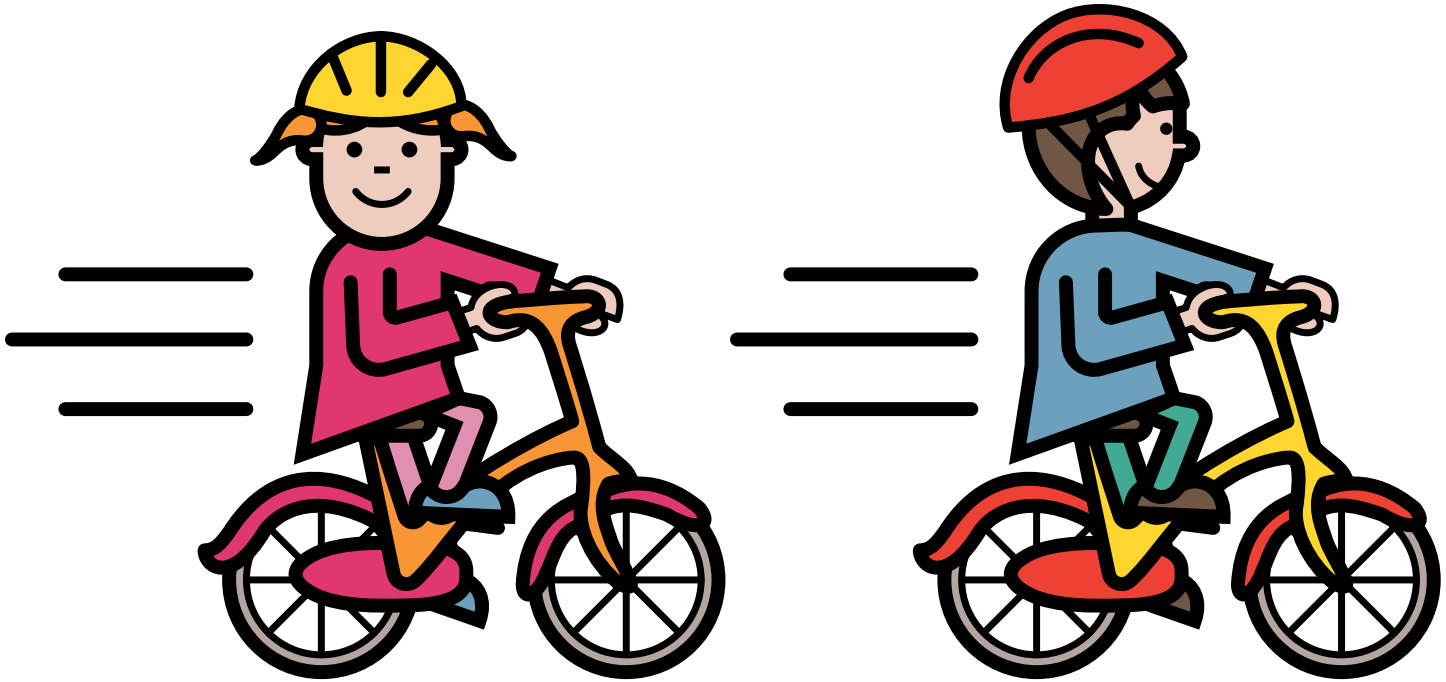
Galactose in blood

Which foods contain galactose?

Galactose mainly comes from lactose. Lactose is the sugar found in milk, milk products, yoghurt and most cheese.

What does galactose do?

Galactose provides energy, but it first needs to be broken down into glucose.

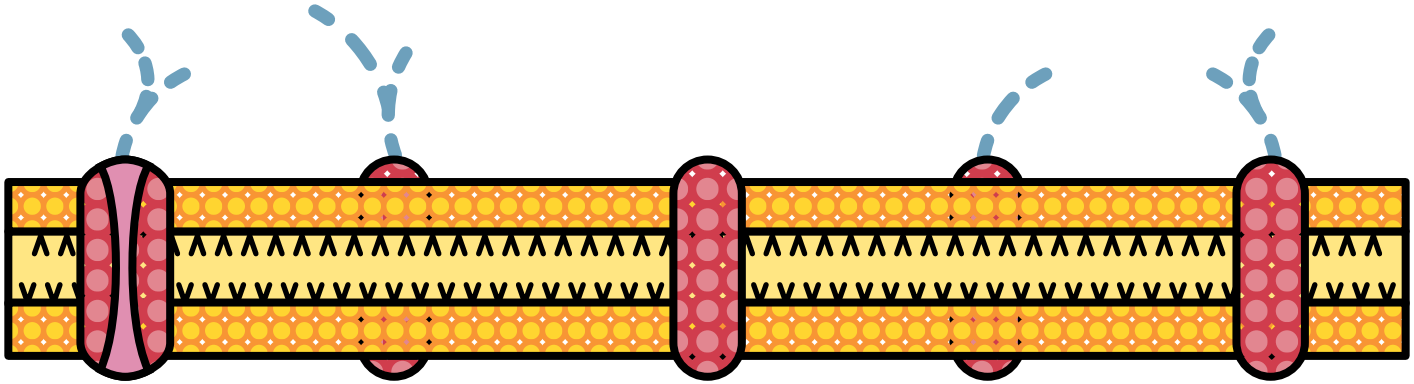


What else does galactose do?

Galactose is a building block of carbohydrate chains.

It joins with **proteins to form glycoproteins** and **fats (lipids) to form glycolipids**.

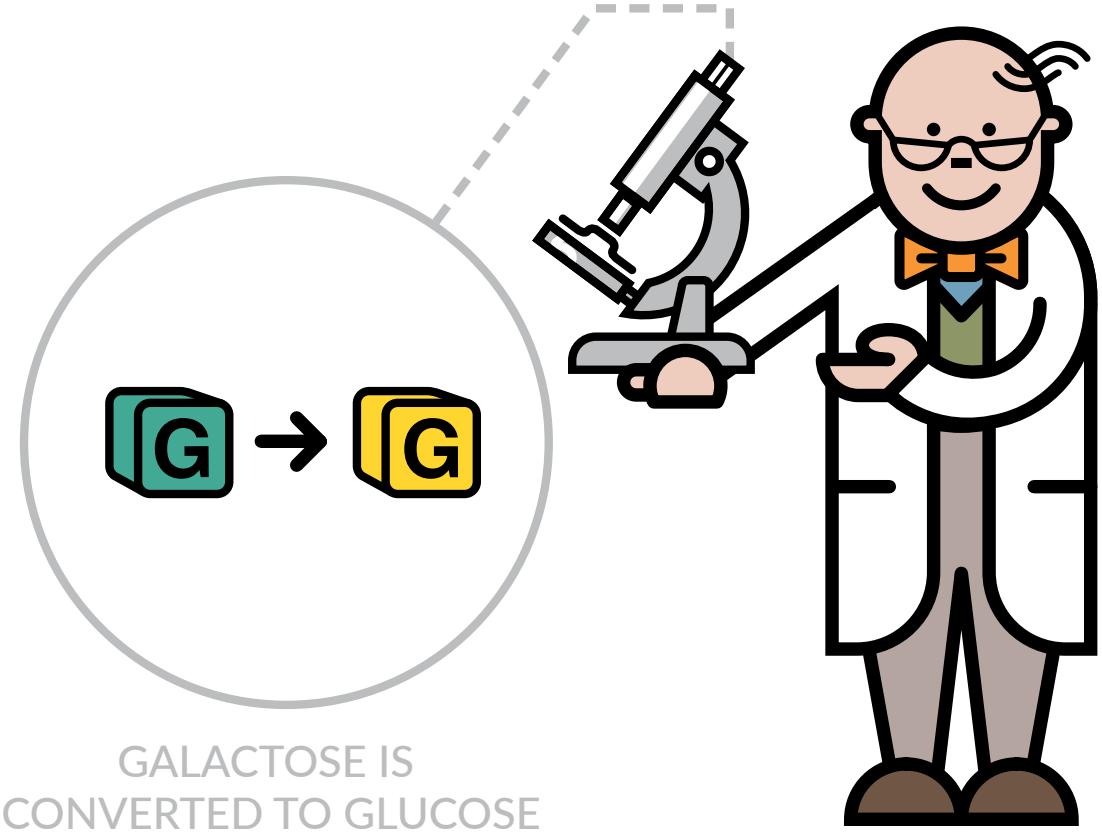
These are important in cell structure.



Carbohydrate chains joining with proteins and lipids in the cell wall

Galactose and enzymes

Galactose is converted into glucose by enzymes (enzymes help chemical reactions).



What happens in Galactosaemia?

In galactosaemia, the body is short of the enzyme that converts galactose into glucose.

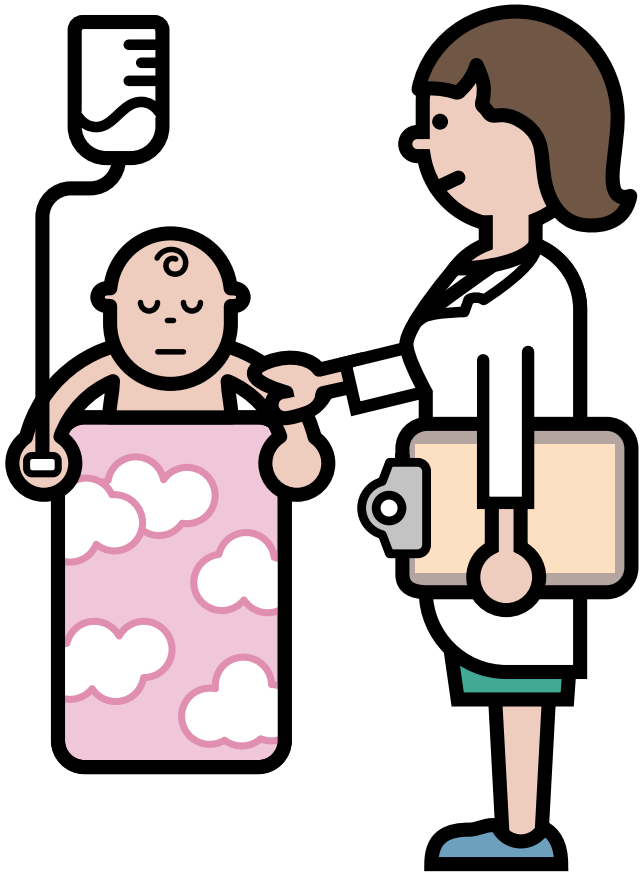
The enzyme is called **galactose-1-phosphate uridyl transferase**.

This leads to a build up of galactose and other chemicals leading to symptoms.

What can go wrong in Galactosaemia?

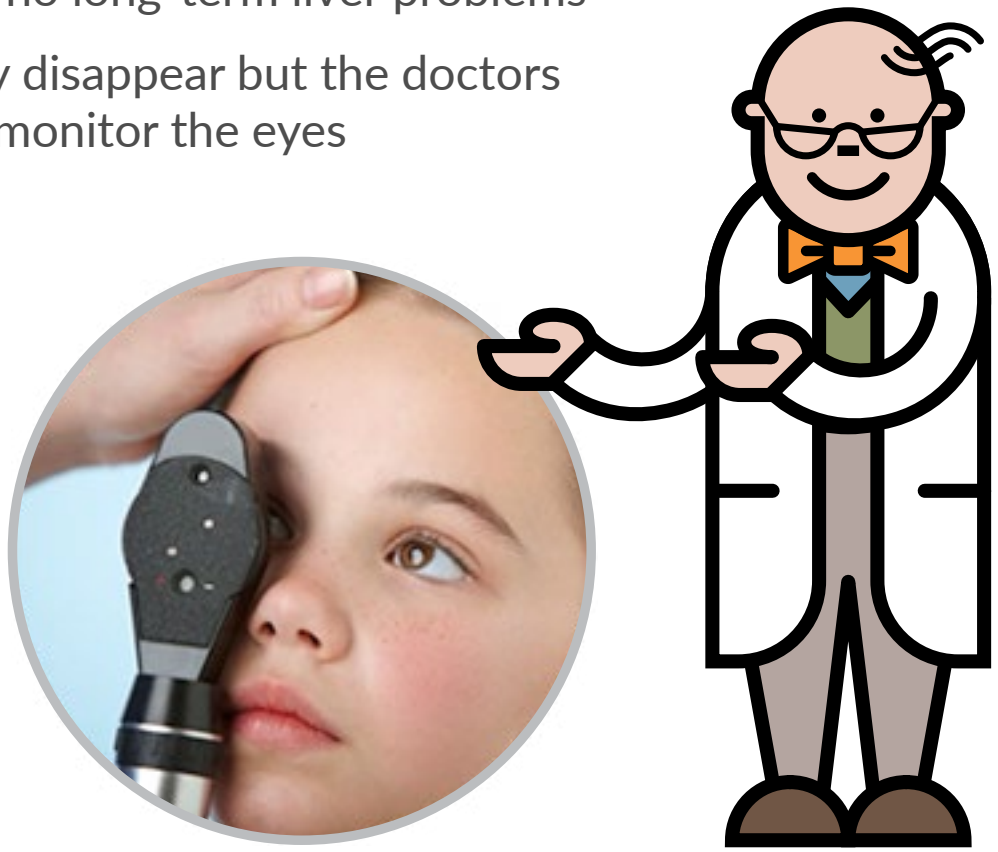
Many babies are very poorly and early symptoms include:

- Vomiting / poor feeding
- Severe jaundice
- Liver dysfunction
- Bacterial infections
- Cataracts (clouding of the lens of the eye)



What happens with treatment?

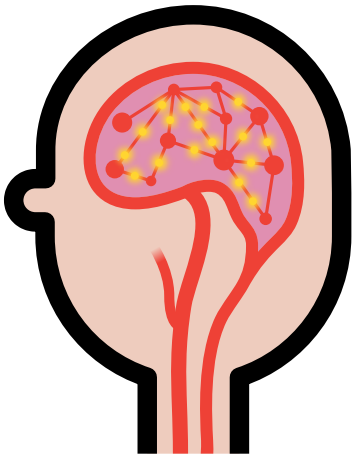
- Once treatment has been started, babies generally start to improve within a few days
- There should be no long-term liver problems
- Cataracts usually disappear but the doctors will continue to monitor the eyes

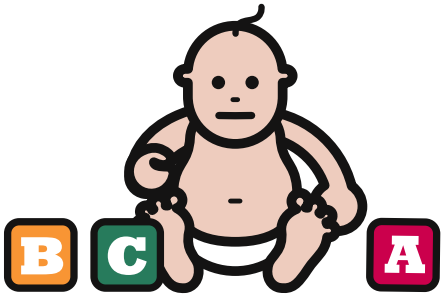


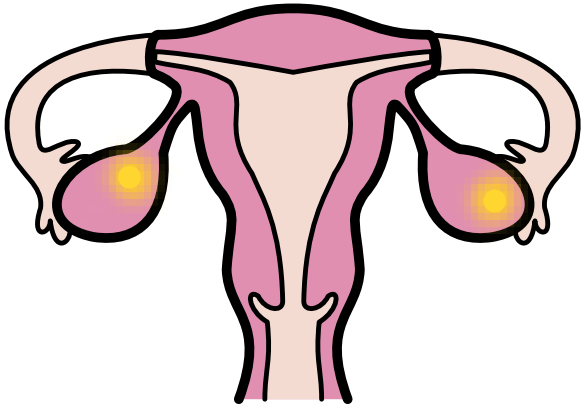
...but even with treatment

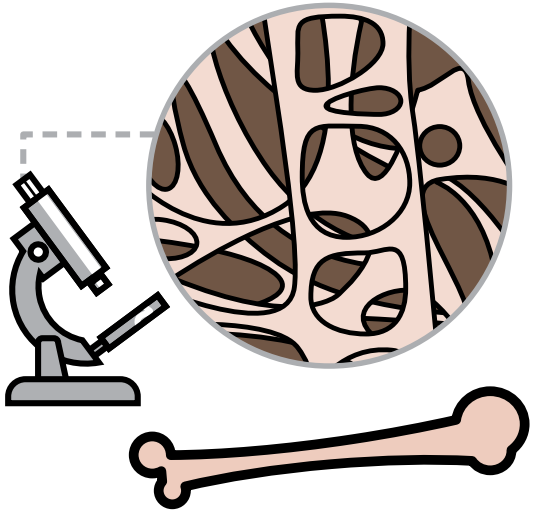
some people have:

- **Learning difficulties**
- **Speech problems**
- **Ovarian problems causing infertility**
- **Lower bone density**
- **A tremor**





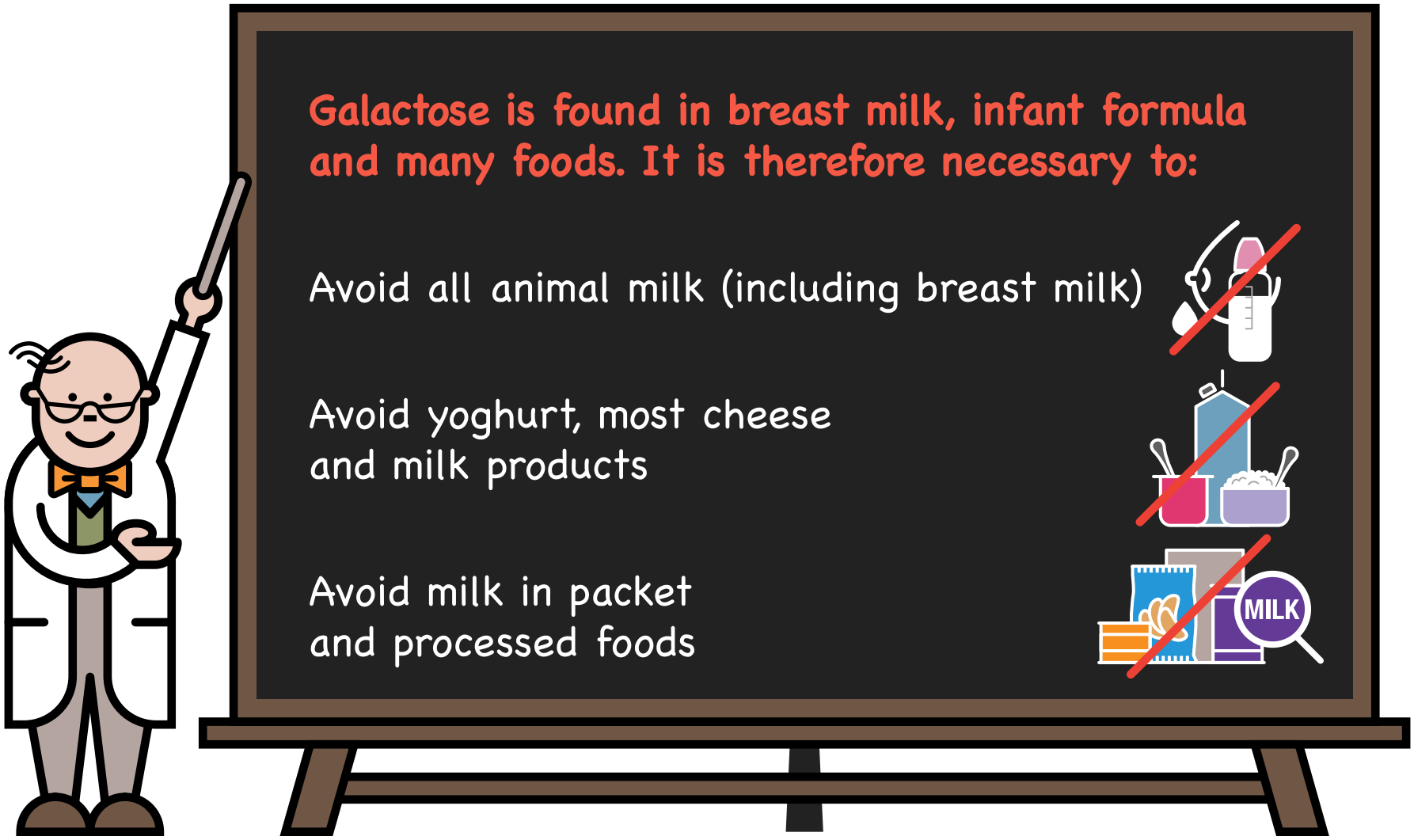




How is Galactosaemia diagnosed?

Galactosaemia is usually diagnosed by looking at enzyme levels in the blood and at the body's genes.

How is Galactosaemia treated?



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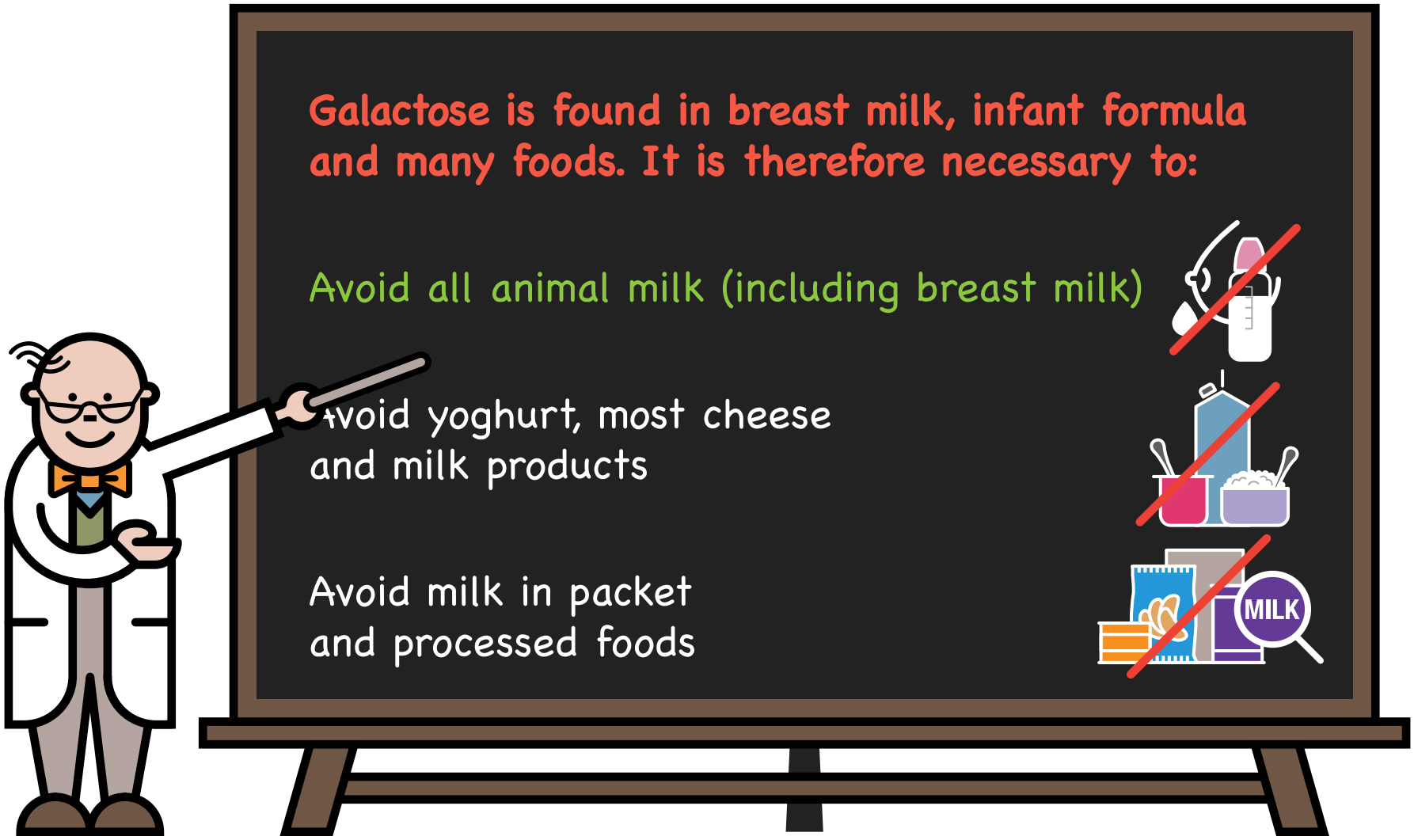

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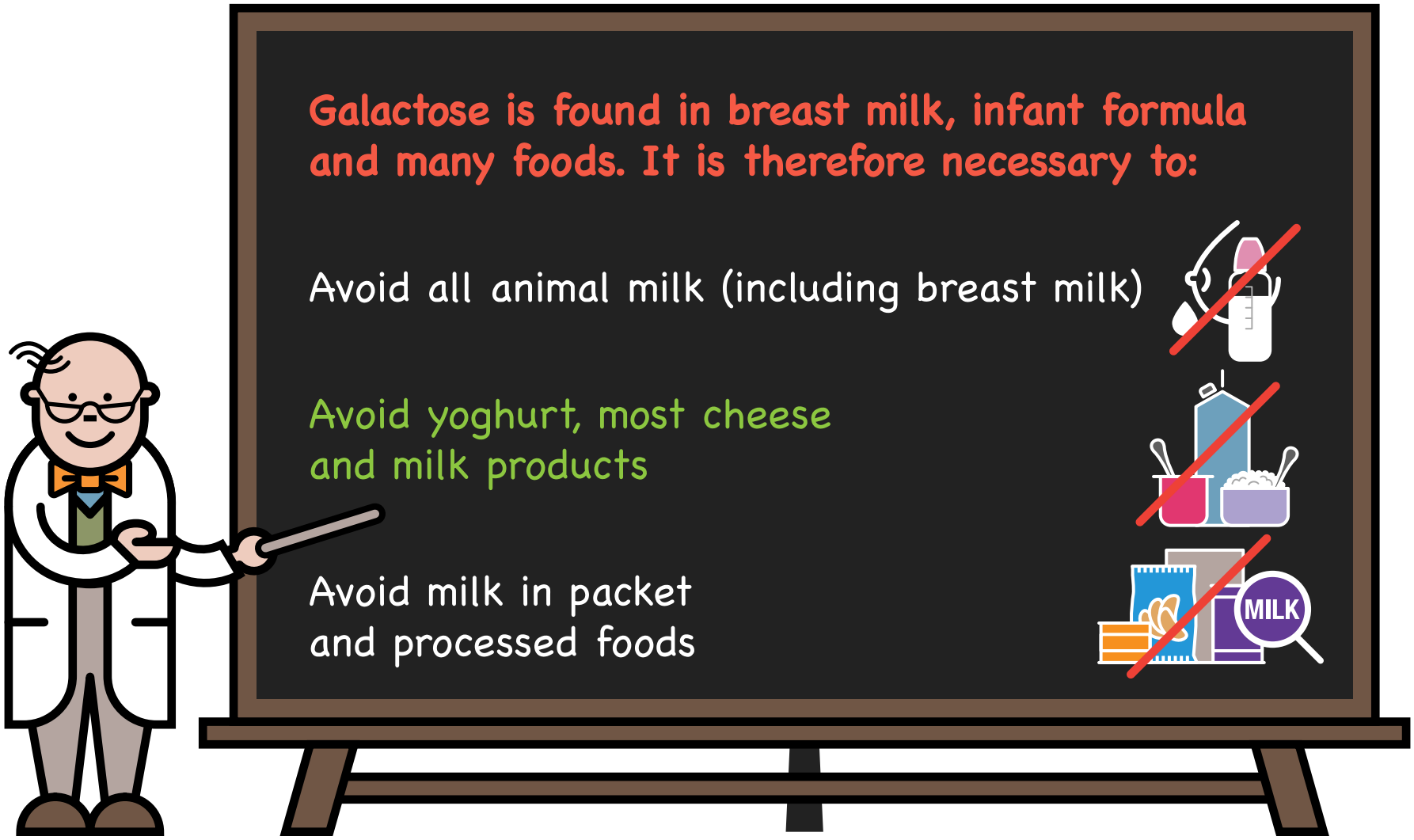

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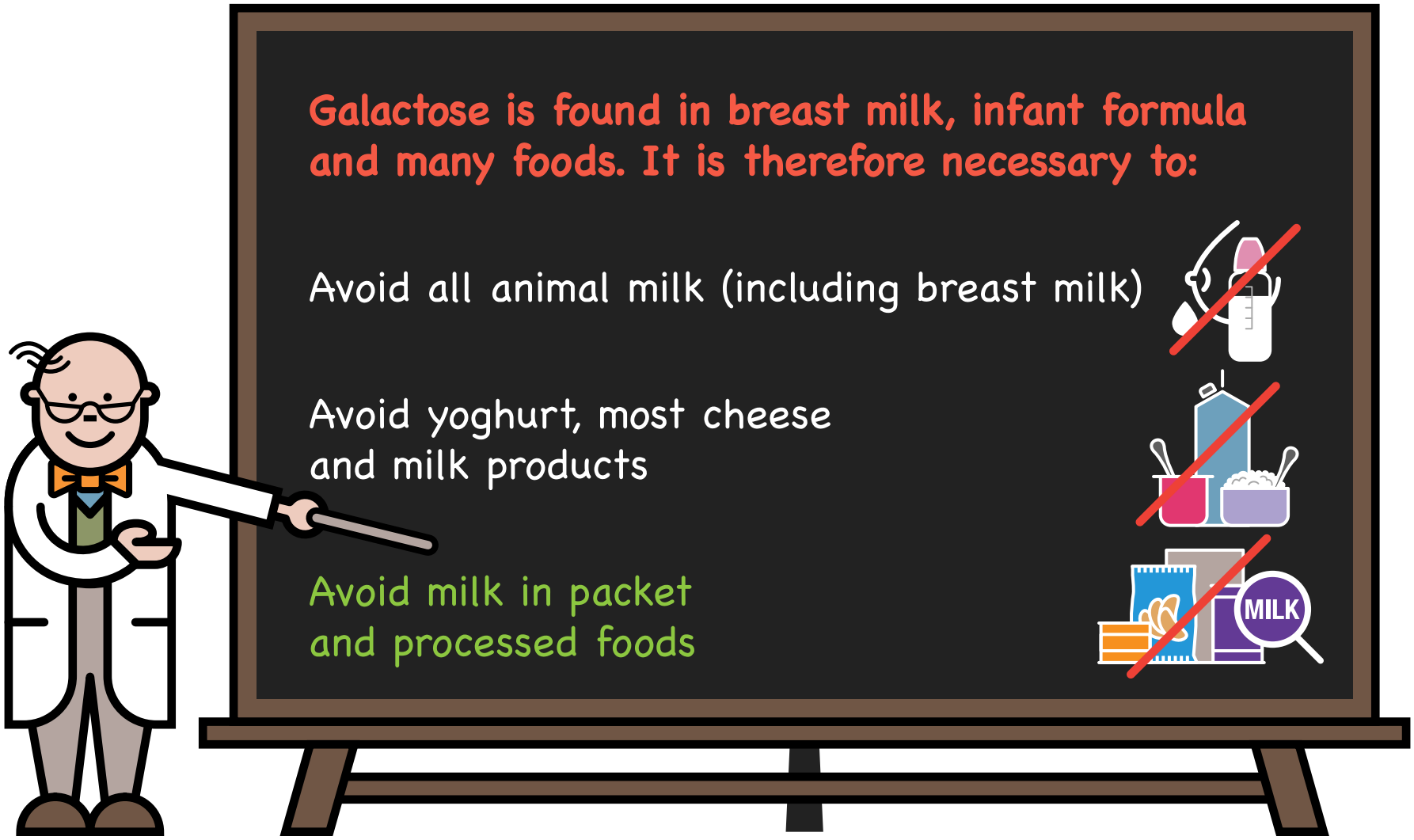

How is Galactosaemia treated?

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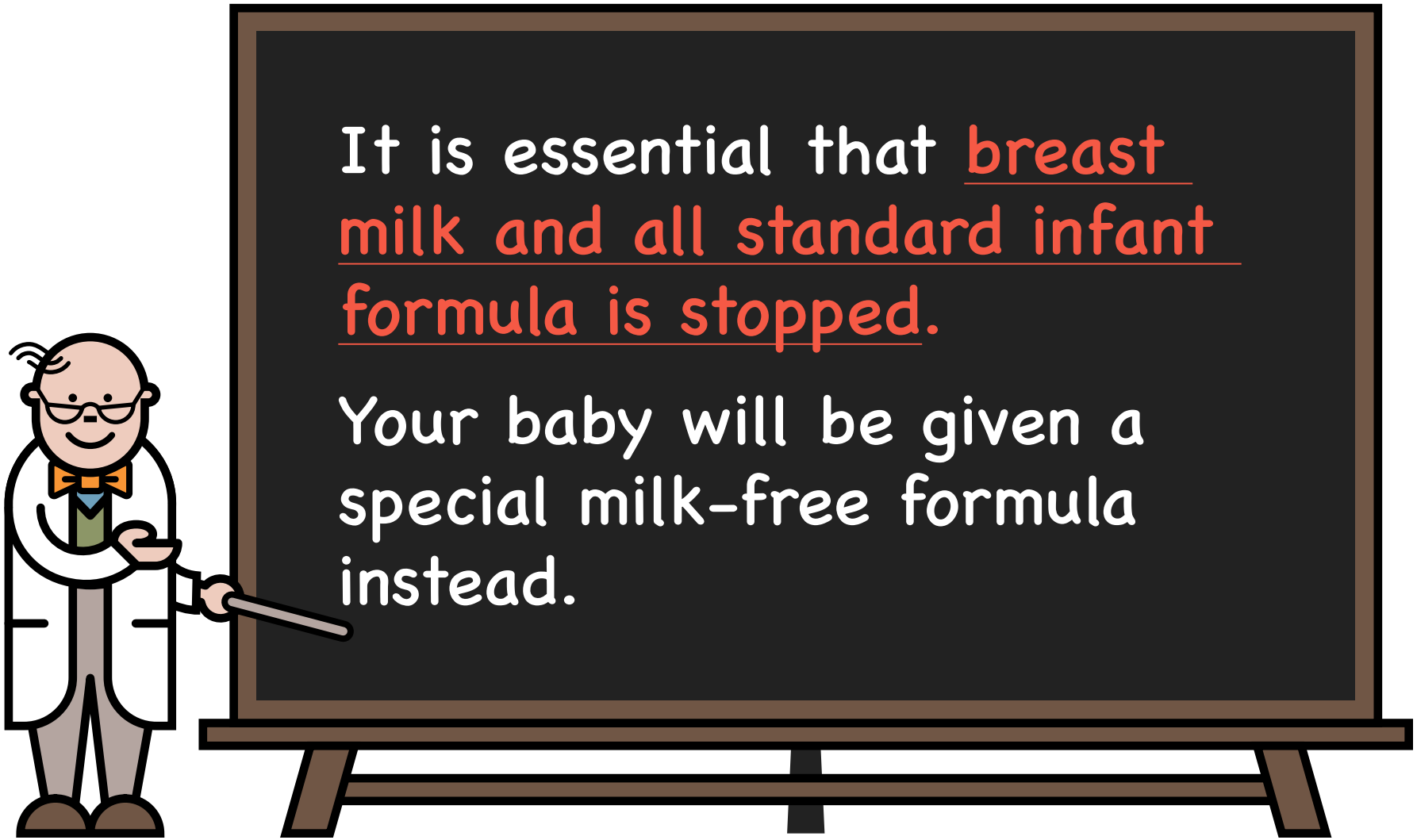
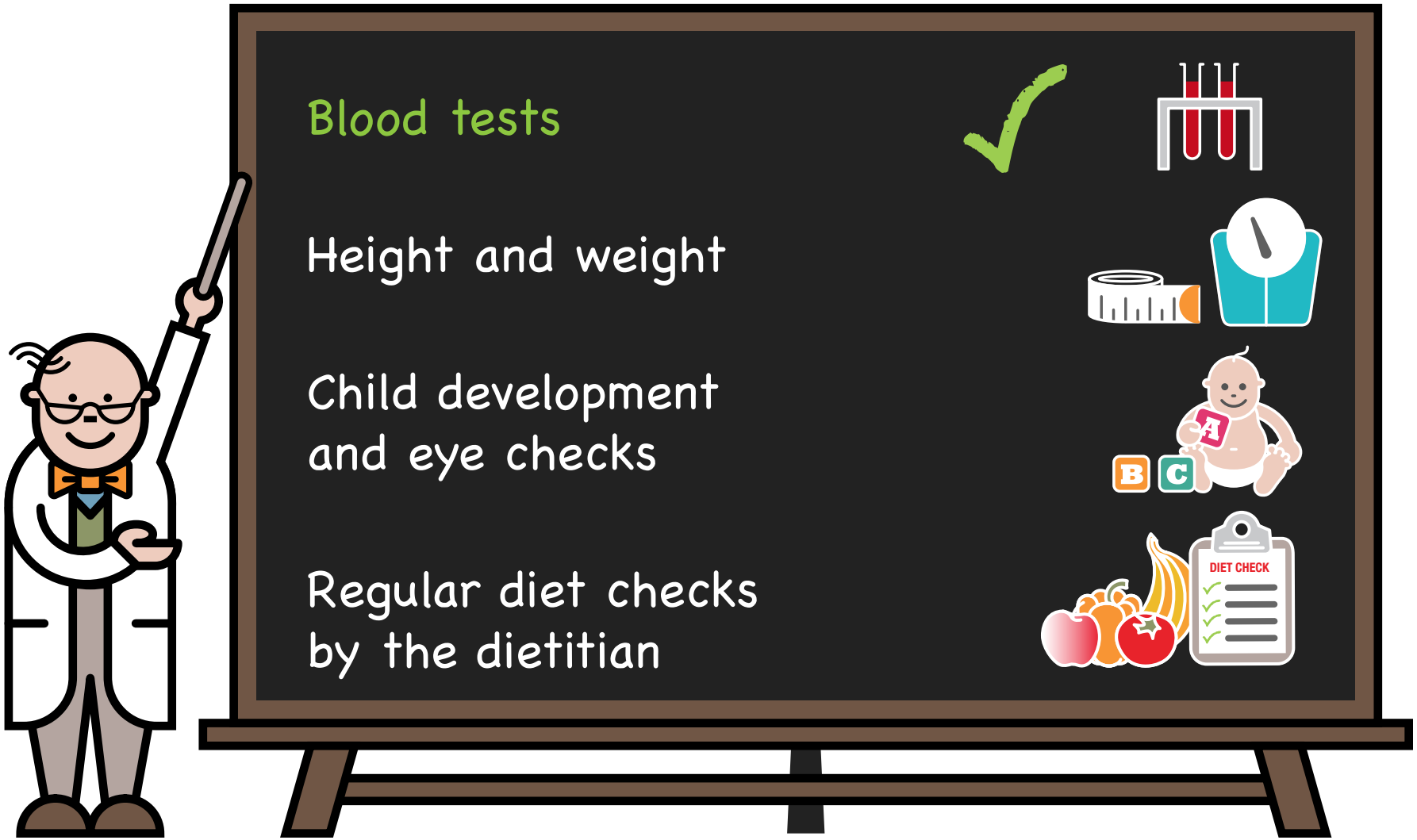
Key message



How is Galactosaemia monitored?



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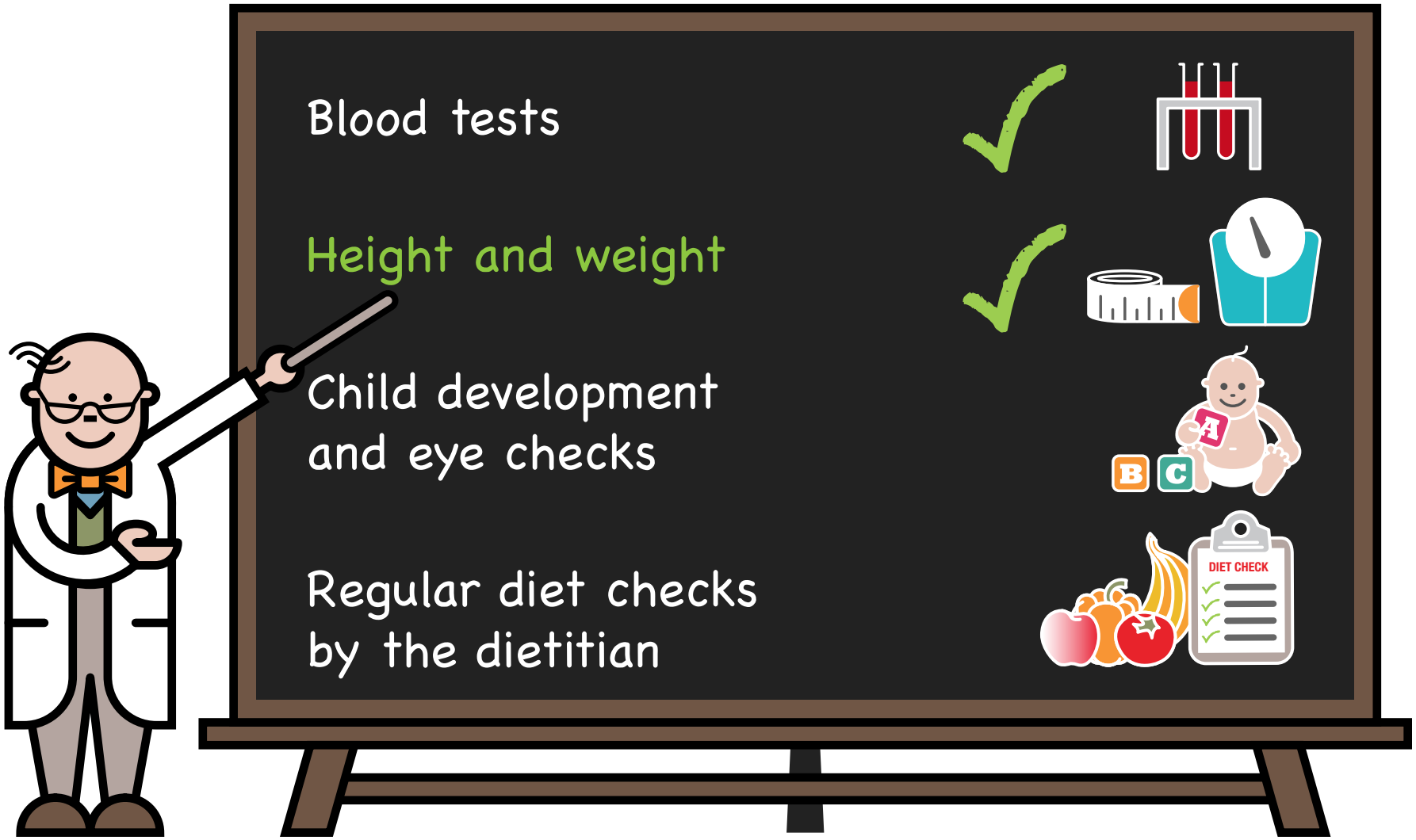

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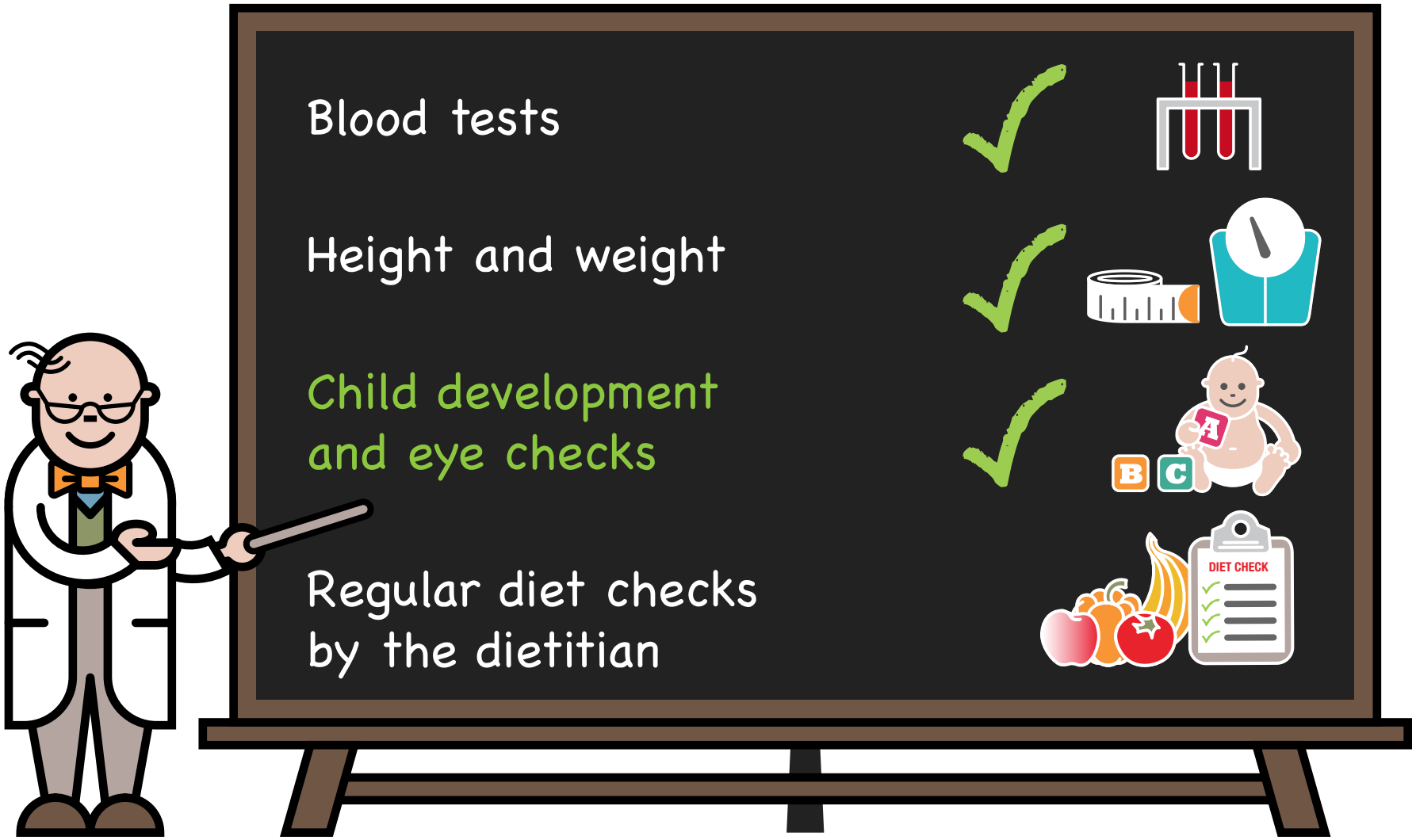

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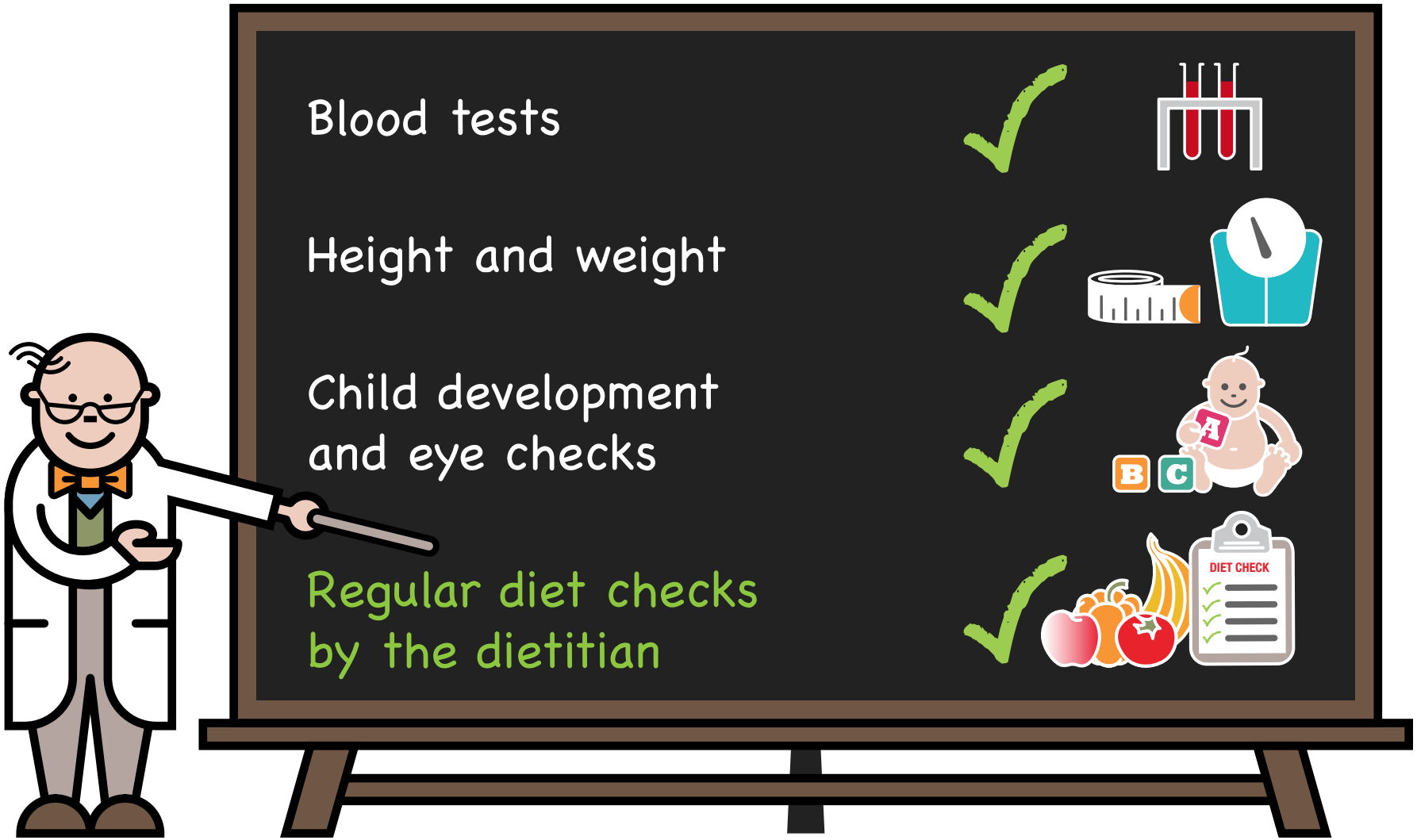
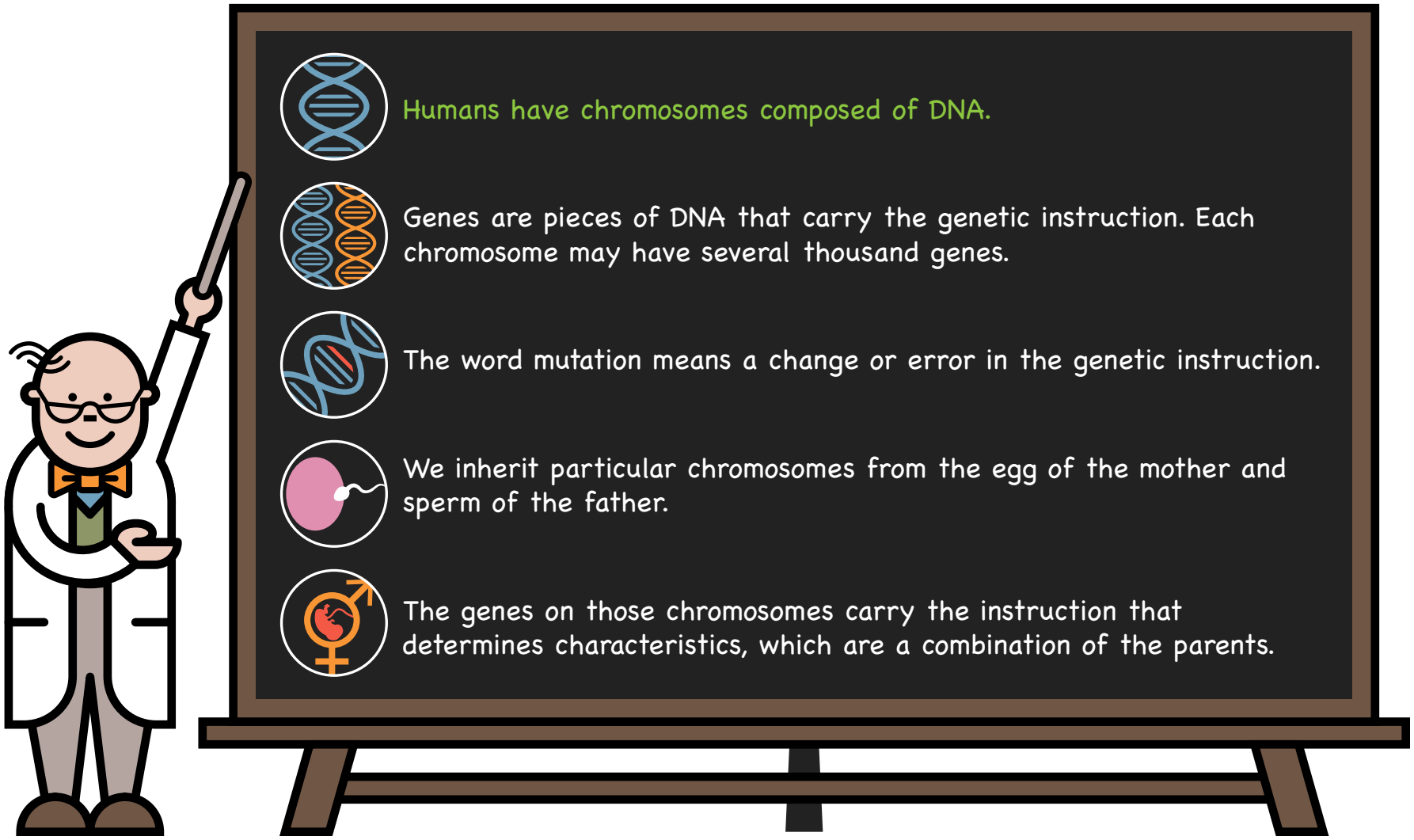


Chromosomes, genes, mutations



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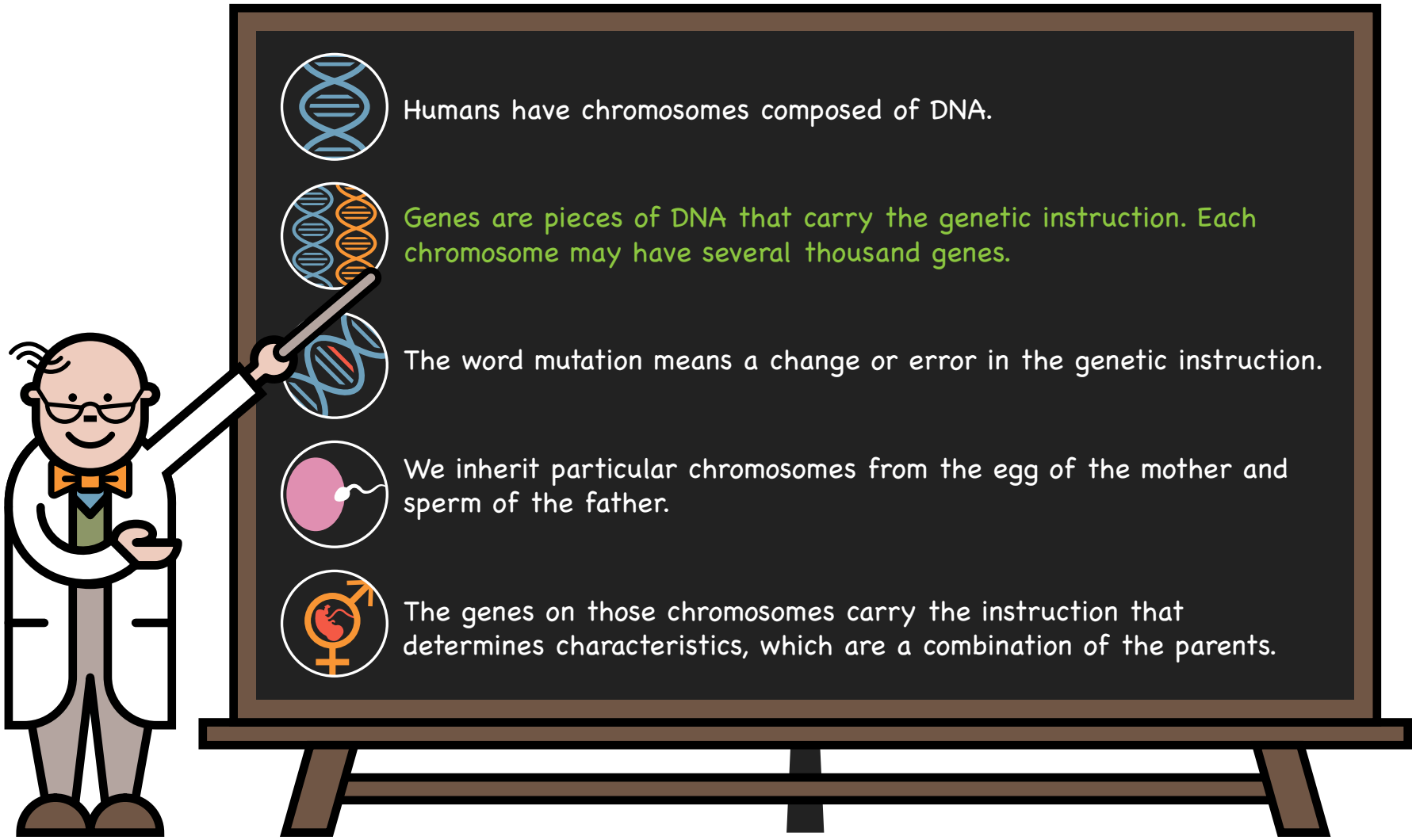
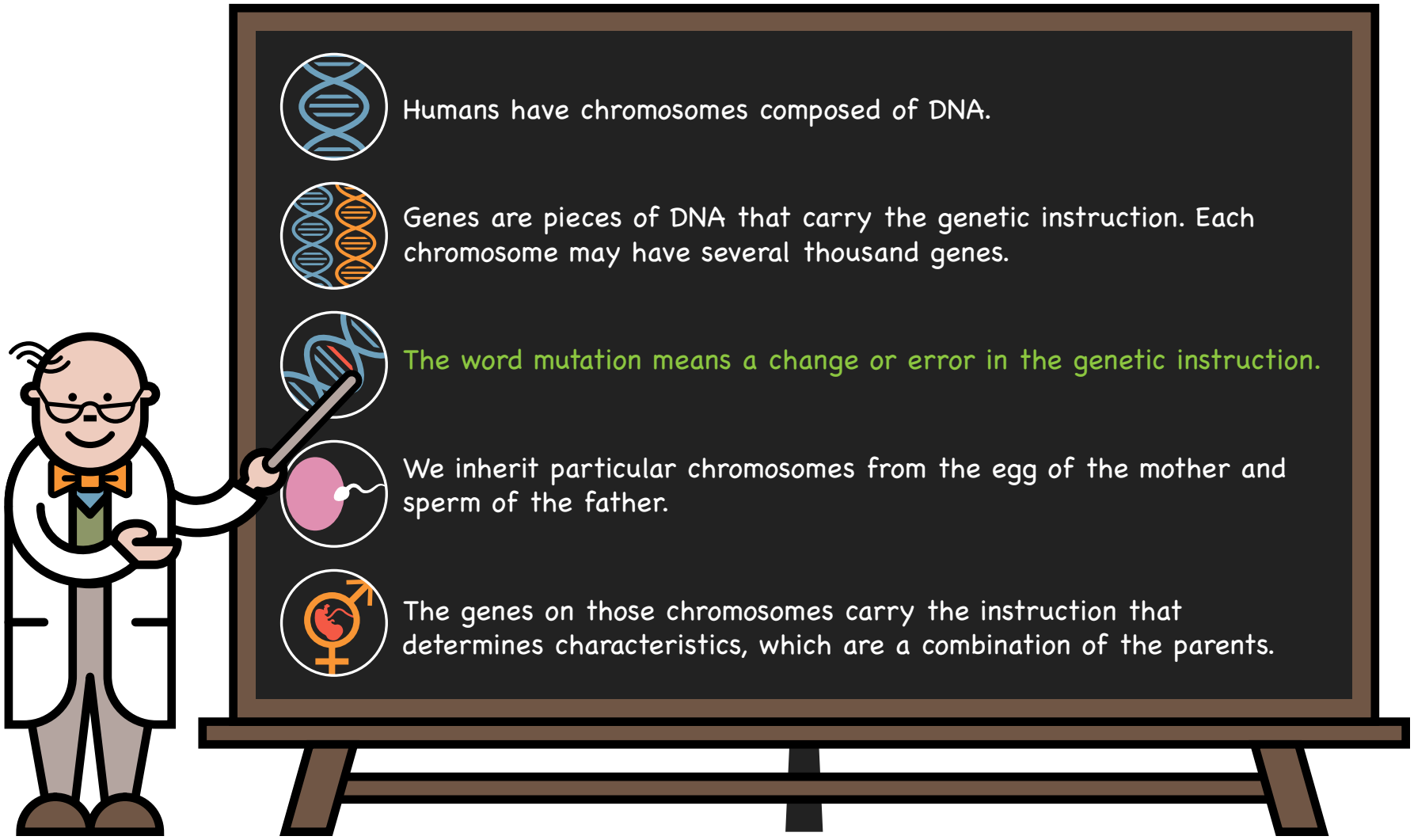


Chromosomes, genes, mutations



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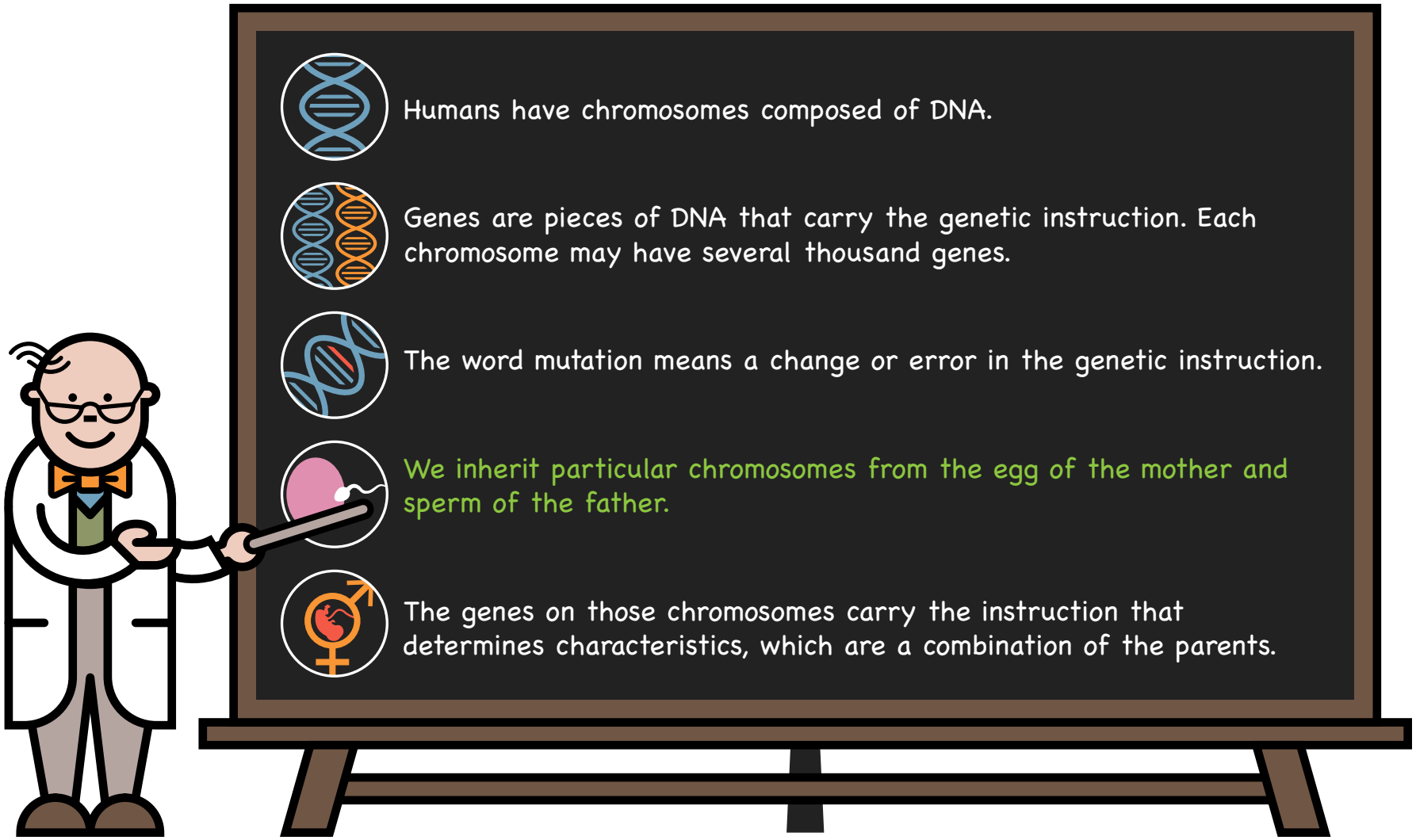
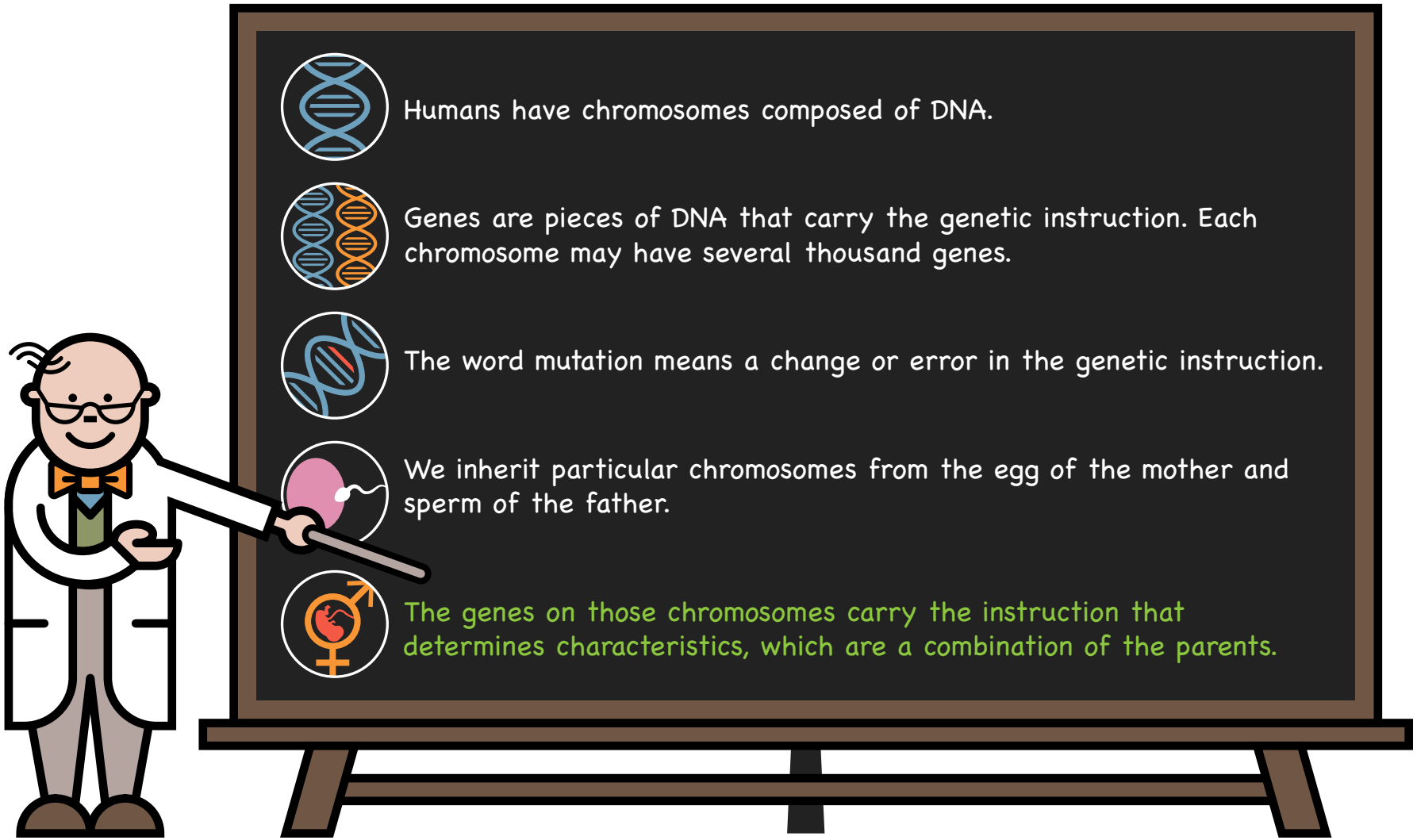
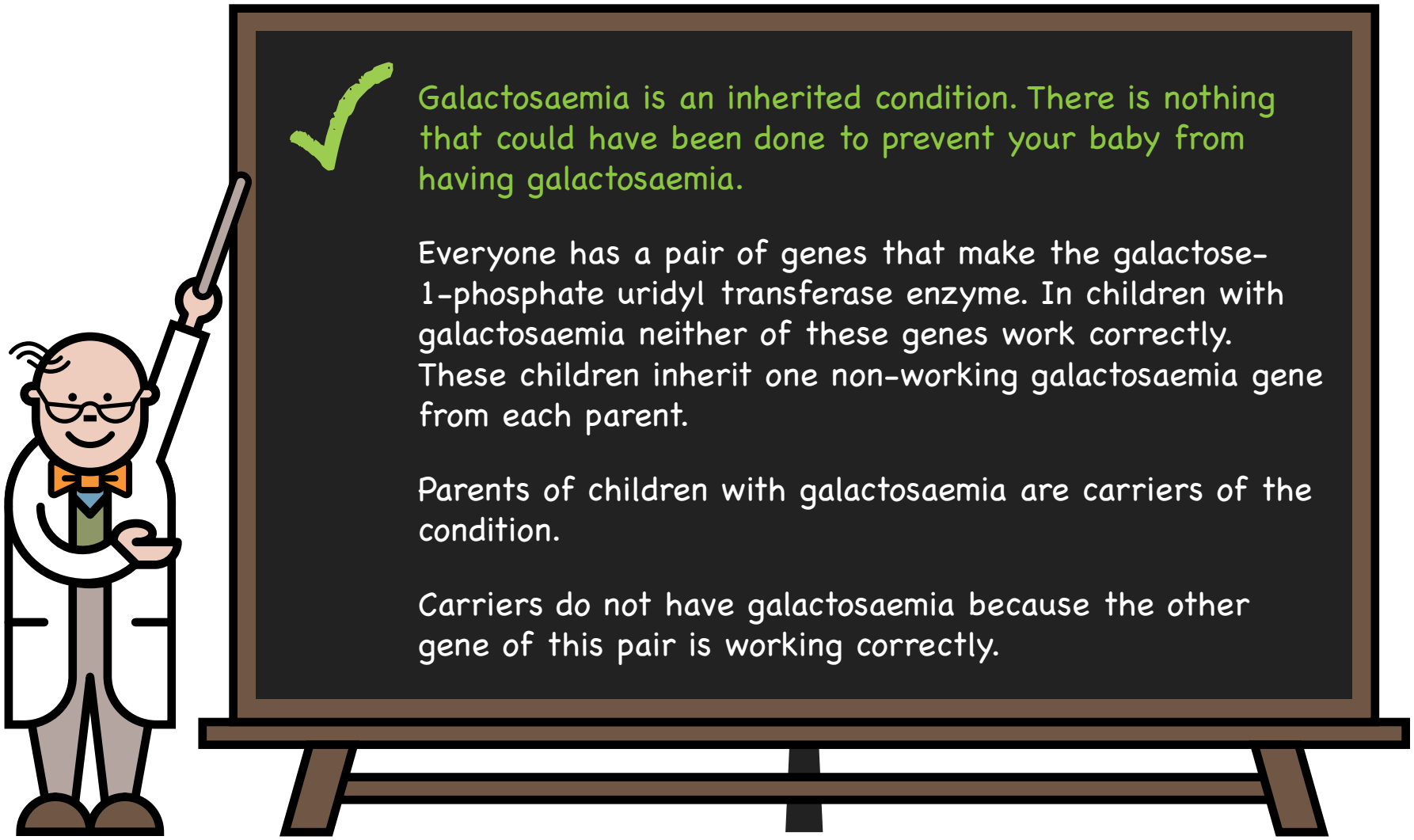


Chromosomes, genes, mutations



Inheritance



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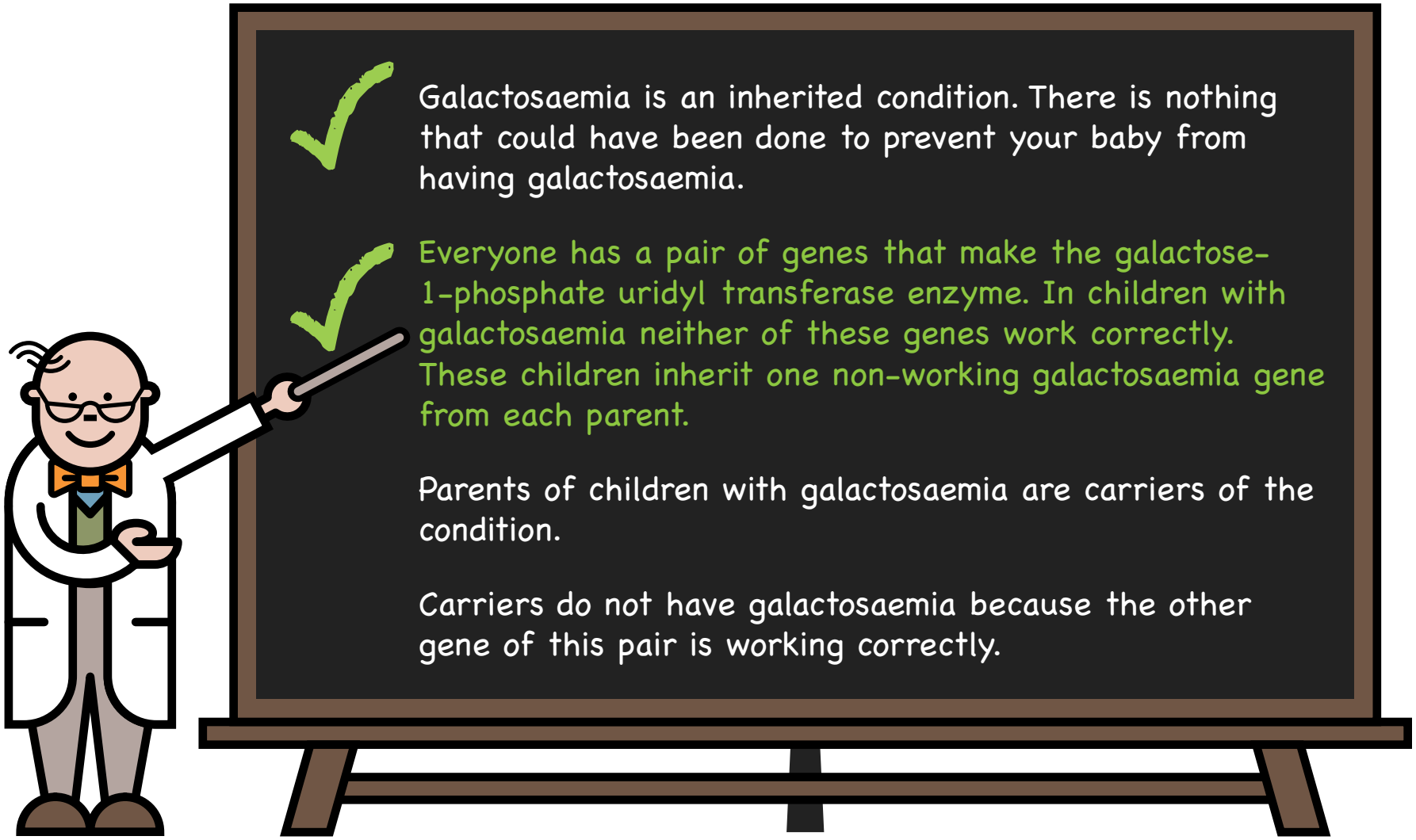

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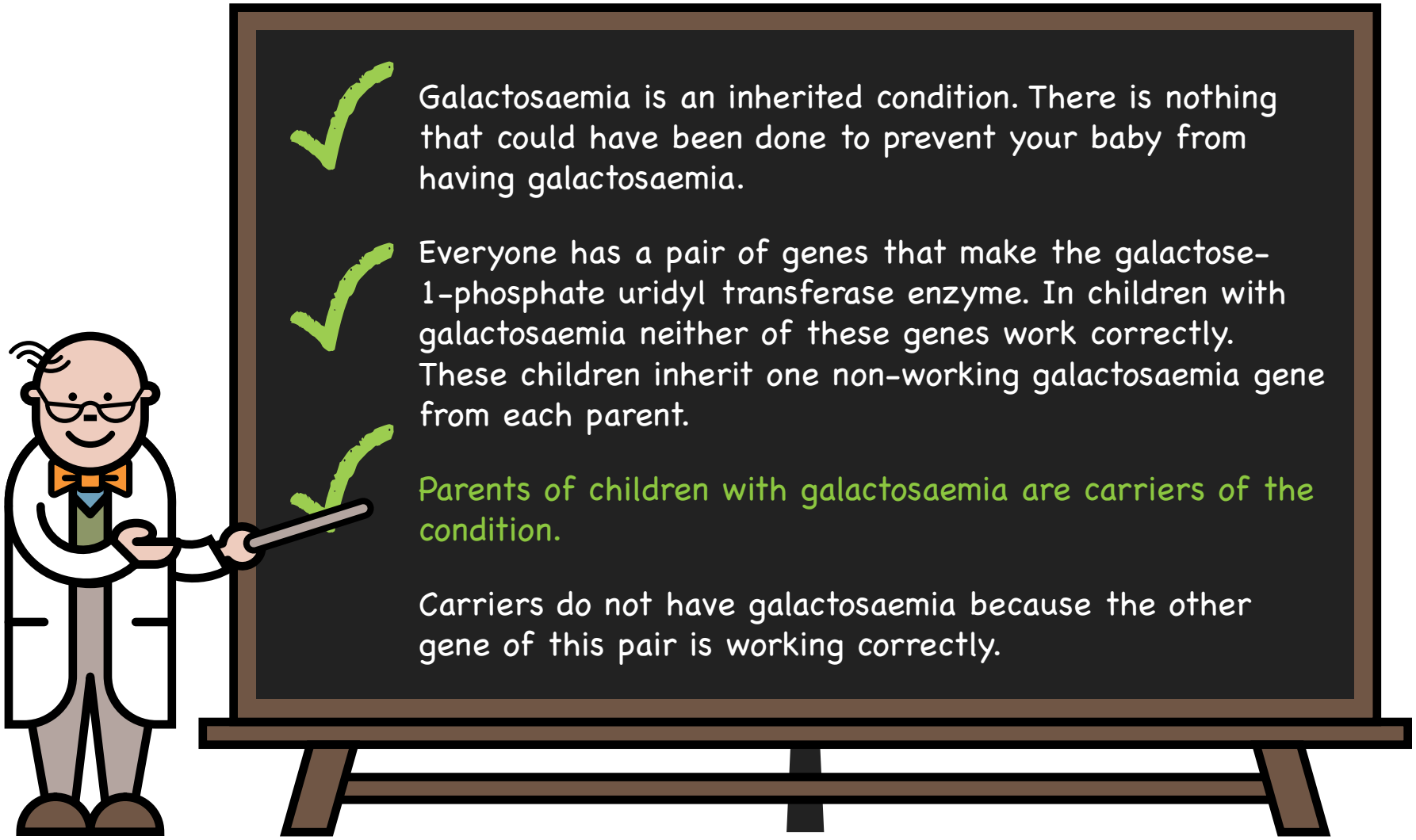
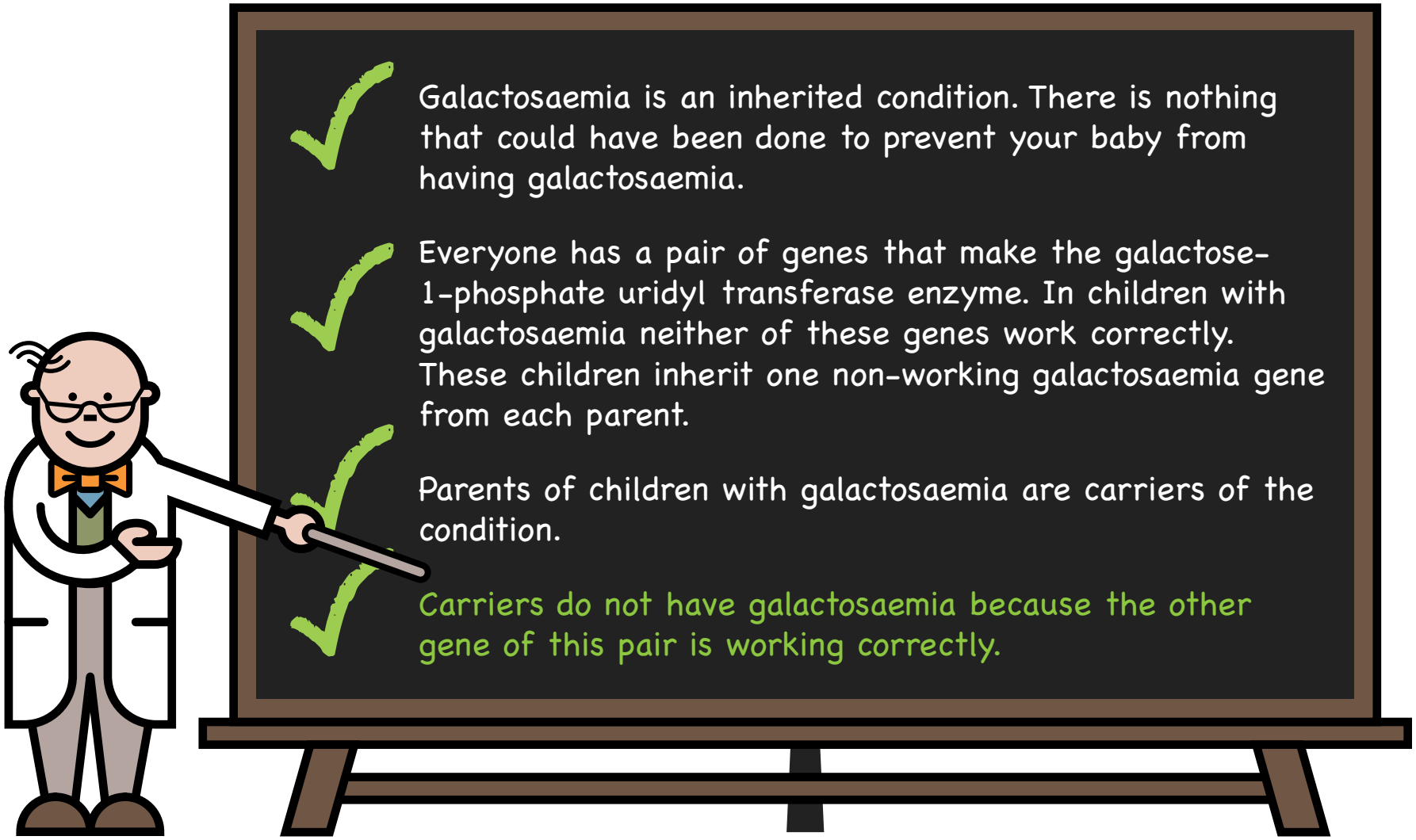
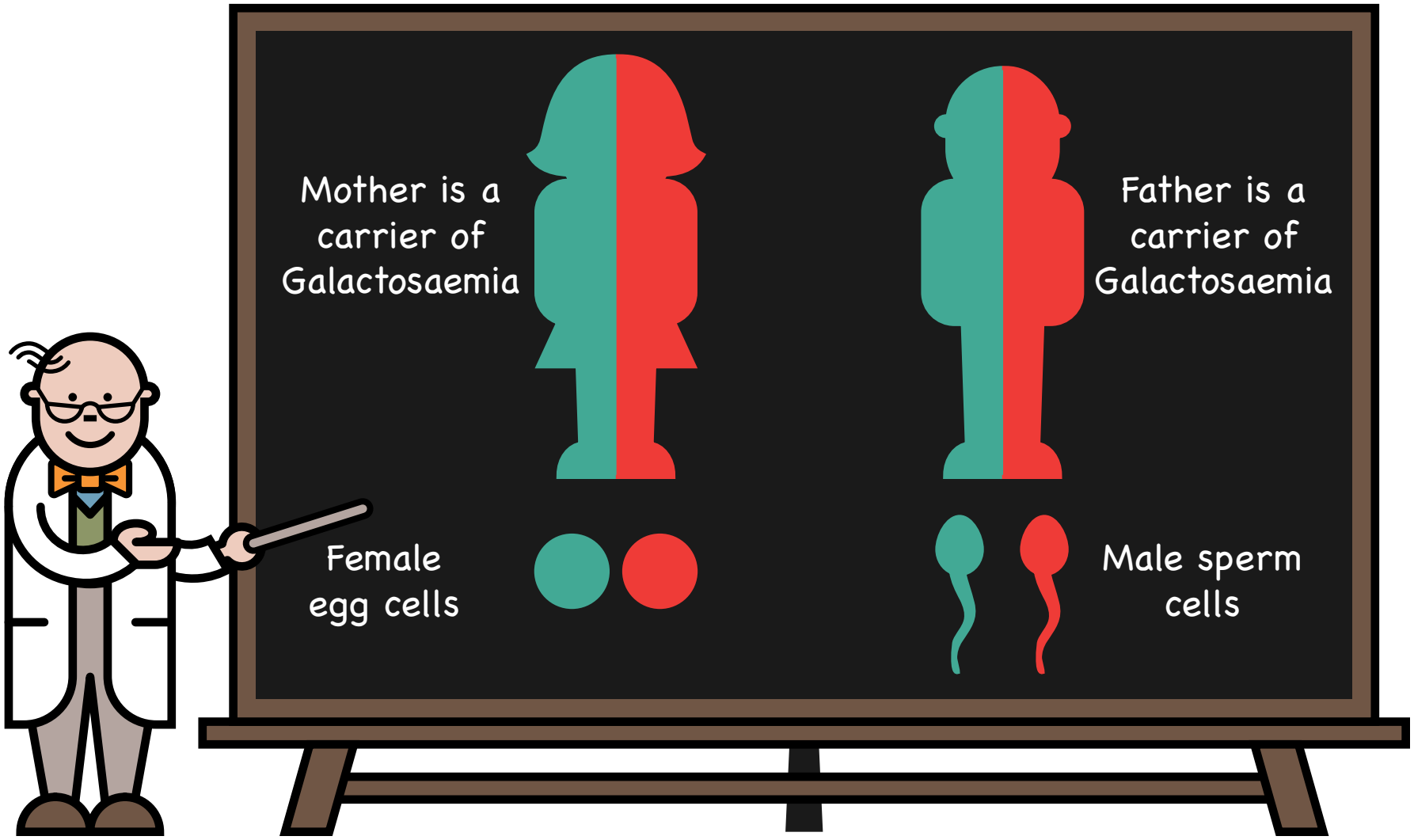


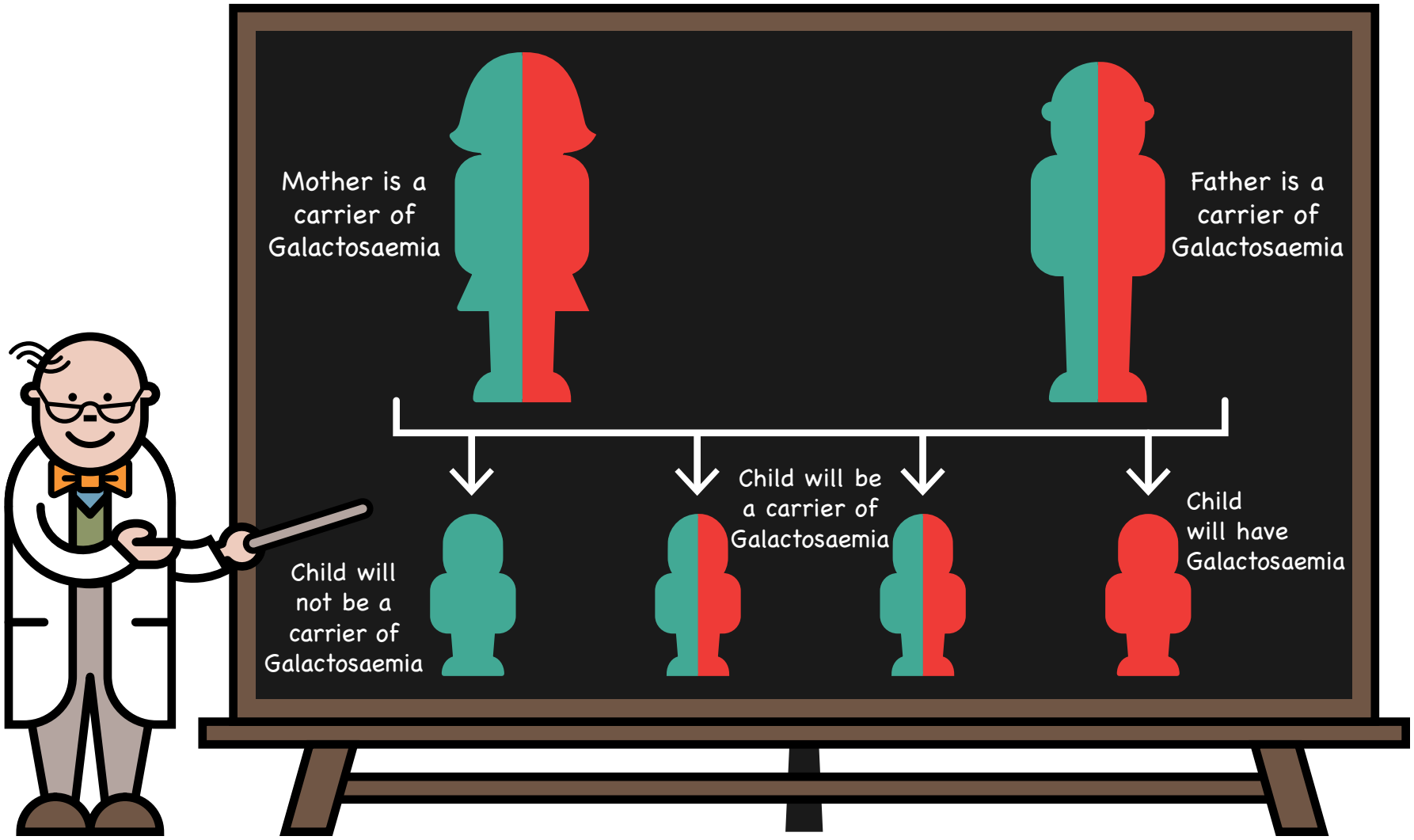
Inheritance



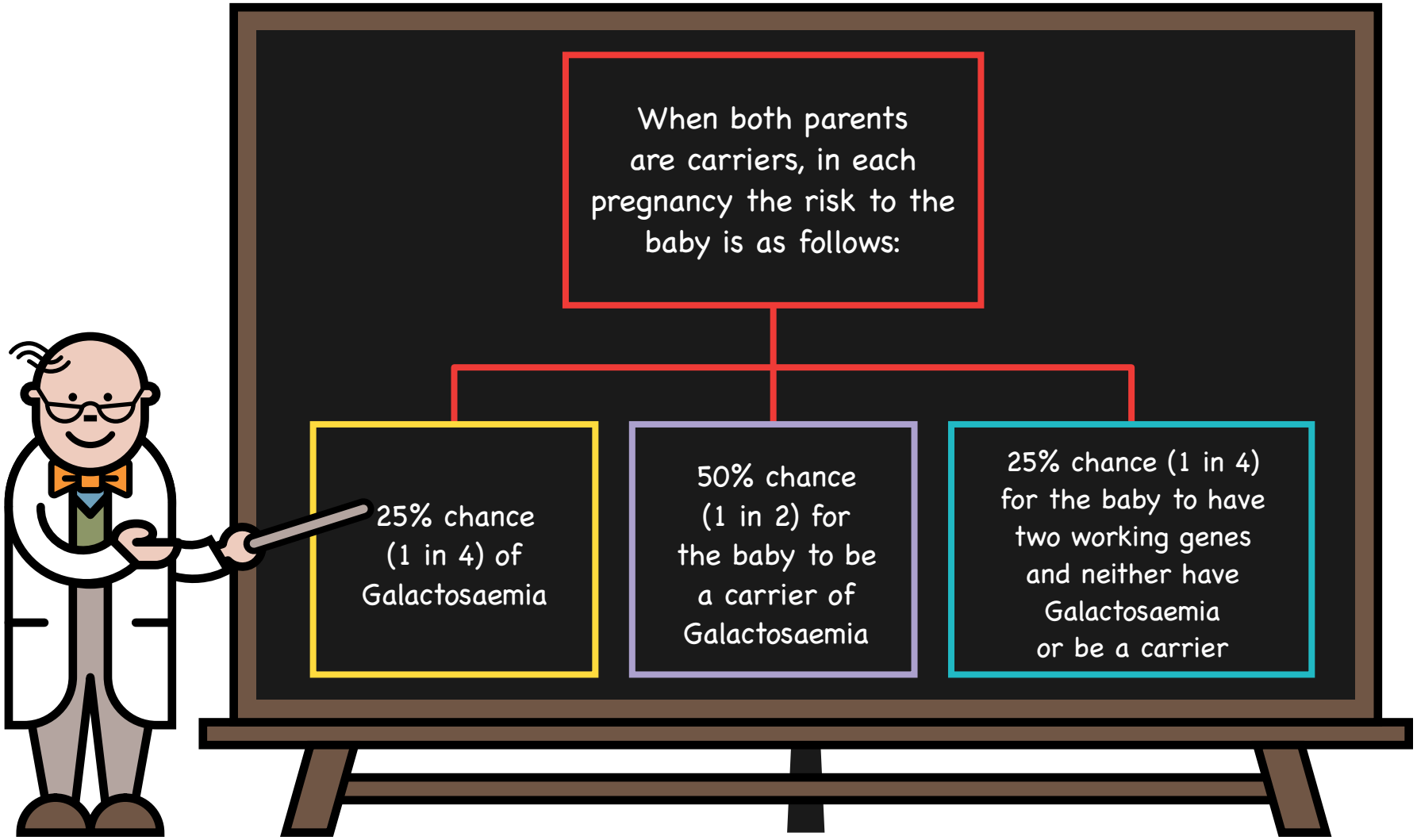
Inheritance – Autosomal-recessive (carriers of Galactosaemia)



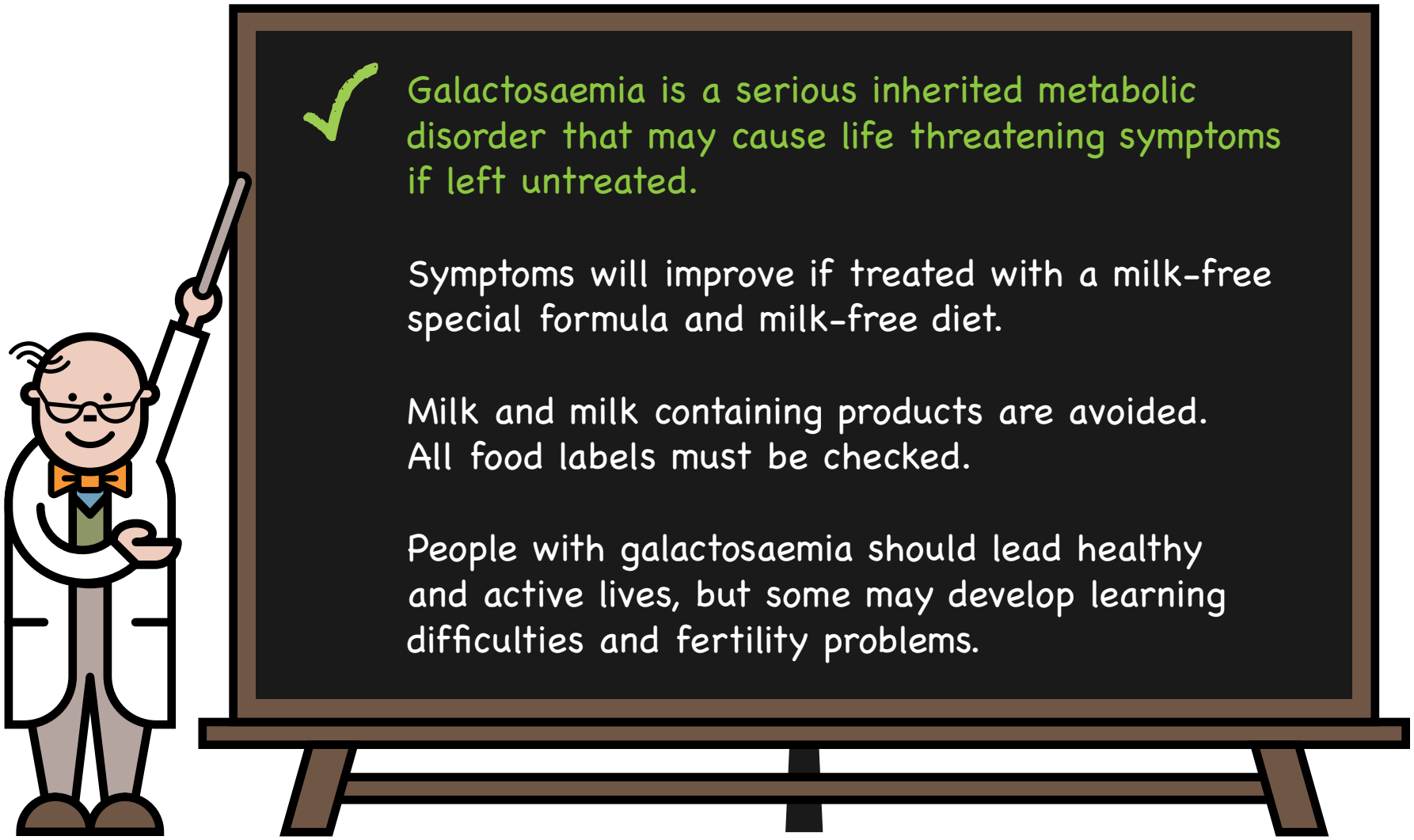
Inheritance – Autosomal recessive – possible combinations



Future pregnancies



Take home messages



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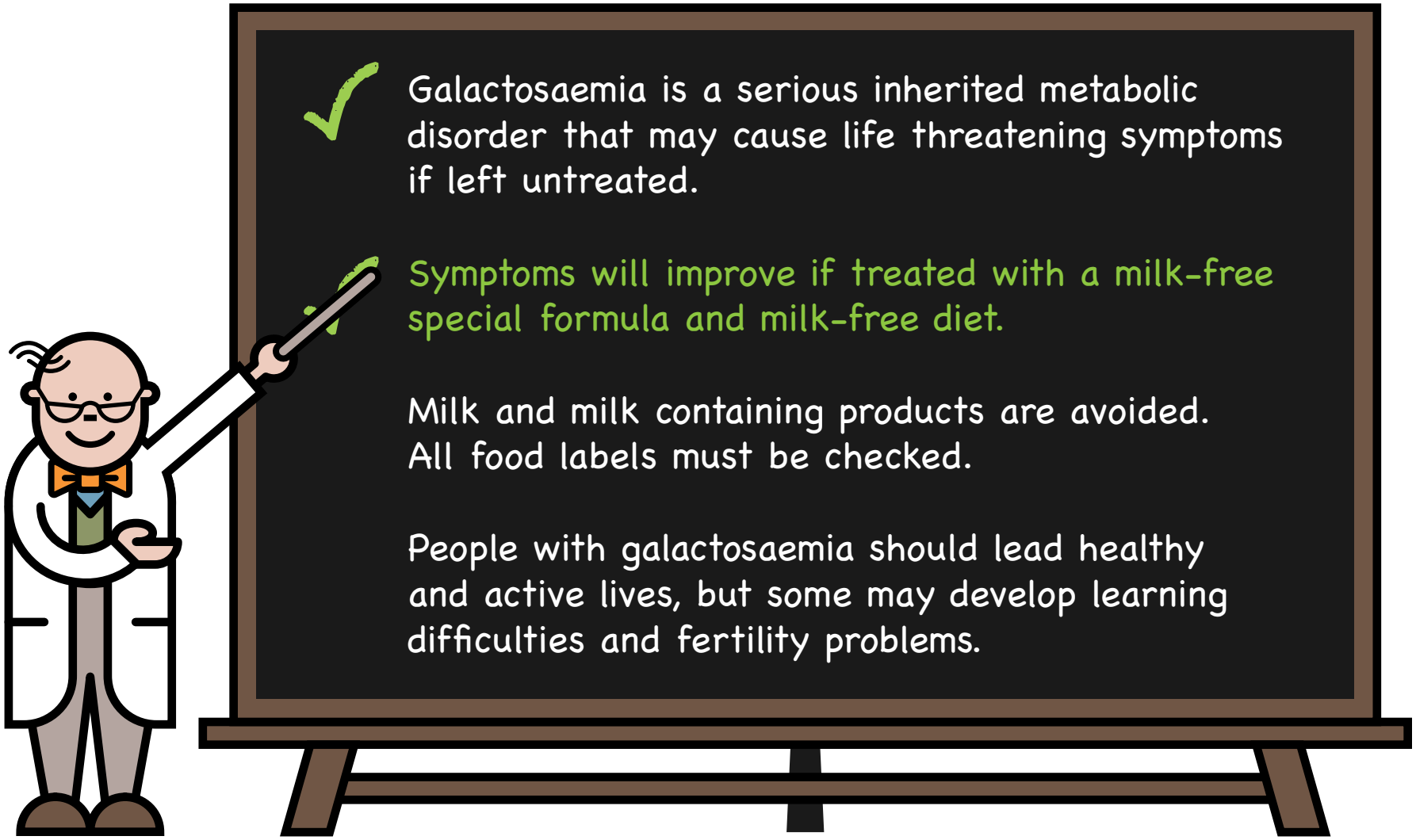

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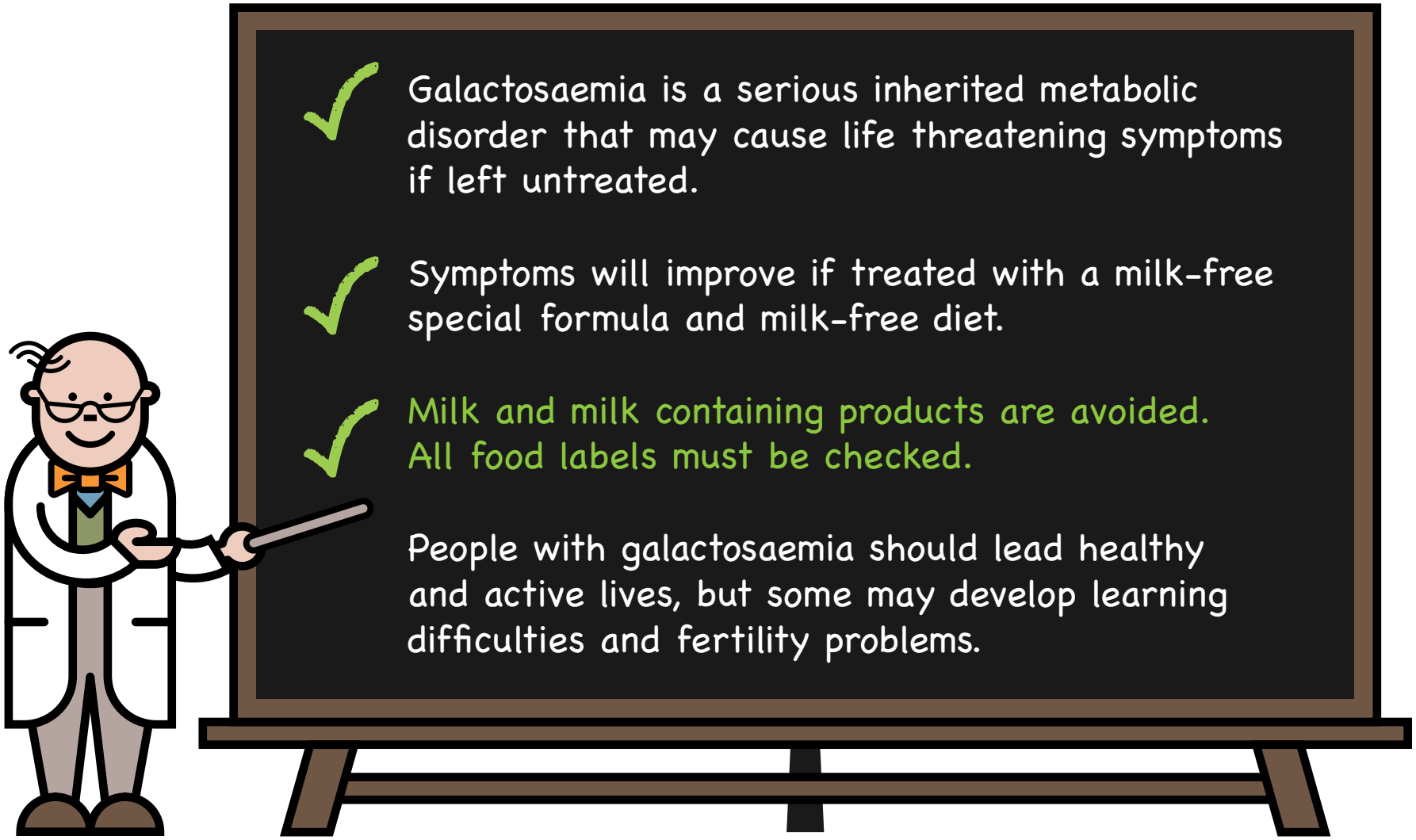

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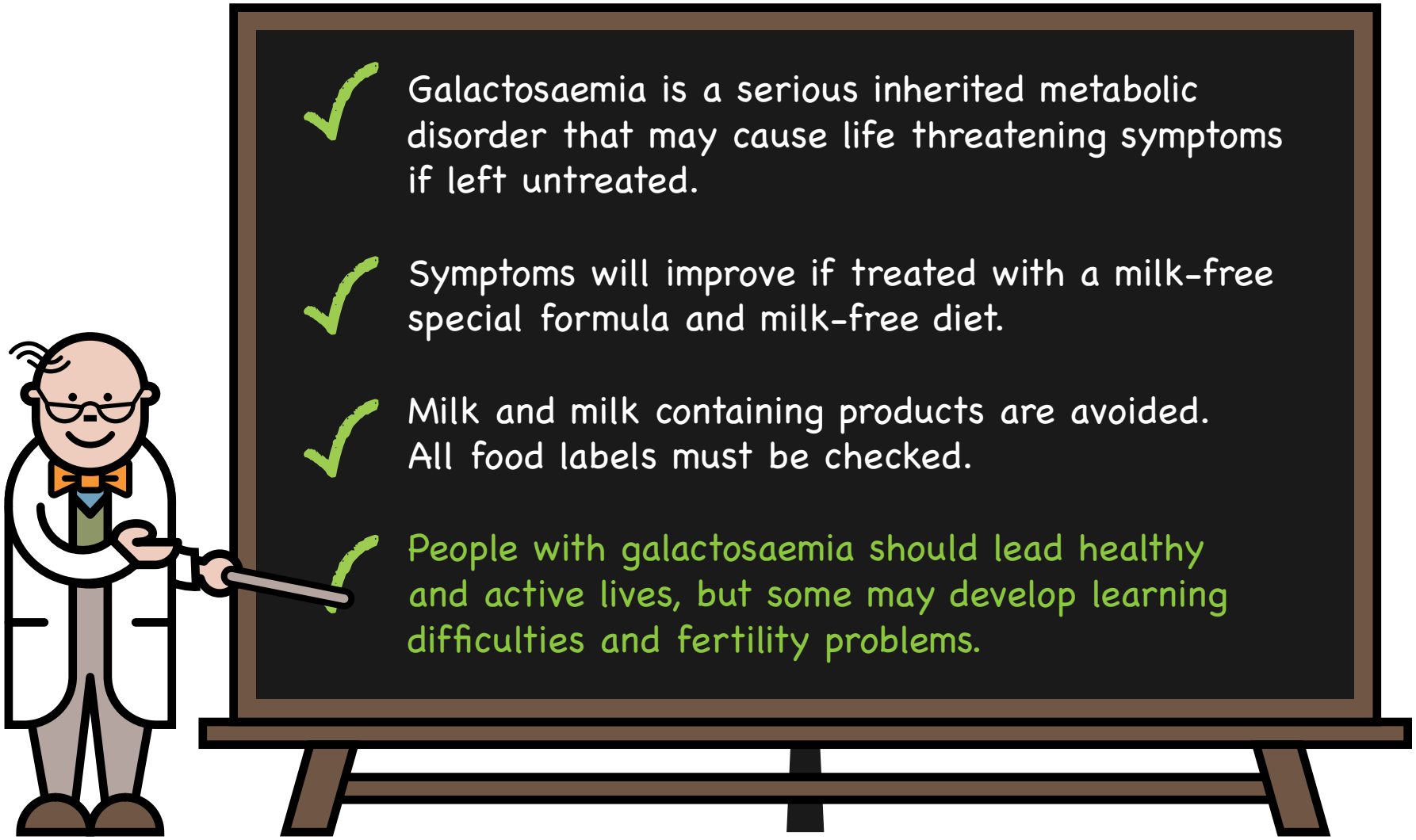

Helpful hints

Helpful hints

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Who's who

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

BIMDG

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