

TEMPLE



Tools **E**nabling **M**etabolic **P**arents **L**Earning

ADAPTED BY THE DIETITIANS GROUP

BIMDG

British Inherited Metabolic Diseases Group



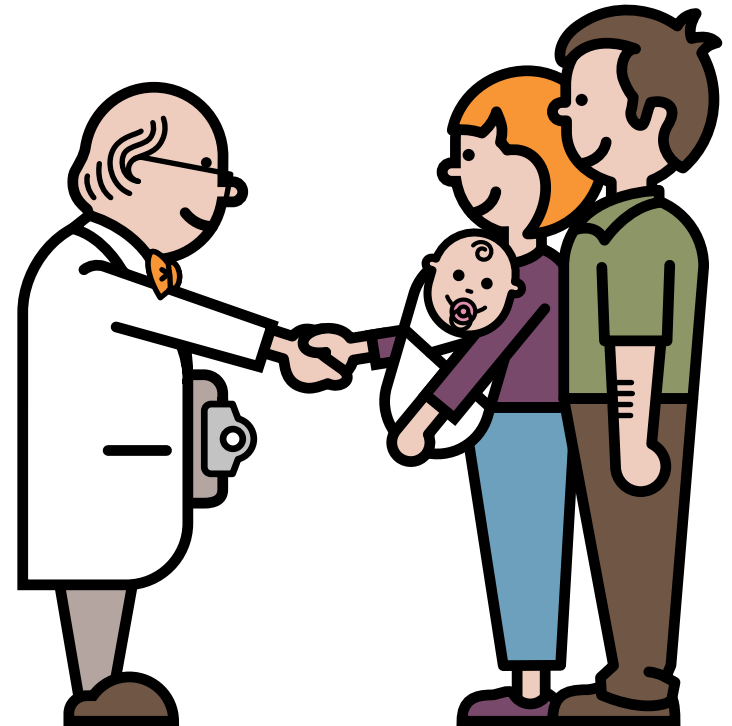
Galactosaemia

BASED ON THE ORIGINAL TEMPLE WRITTEN BY
BURGARD AND WENDEL
VERSION 1, OCTOBER 2017

Supported by  **NUTRICIA**
as a service to metabolic medicine

Galactosaemia

Information for families following
a new diagnosis in infancy



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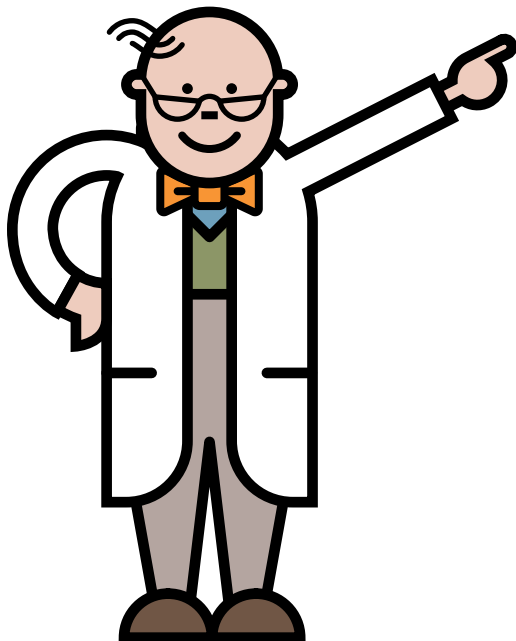
Tools Enabling Metabolic Parents LEarning

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as a service to metabolic medicine

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

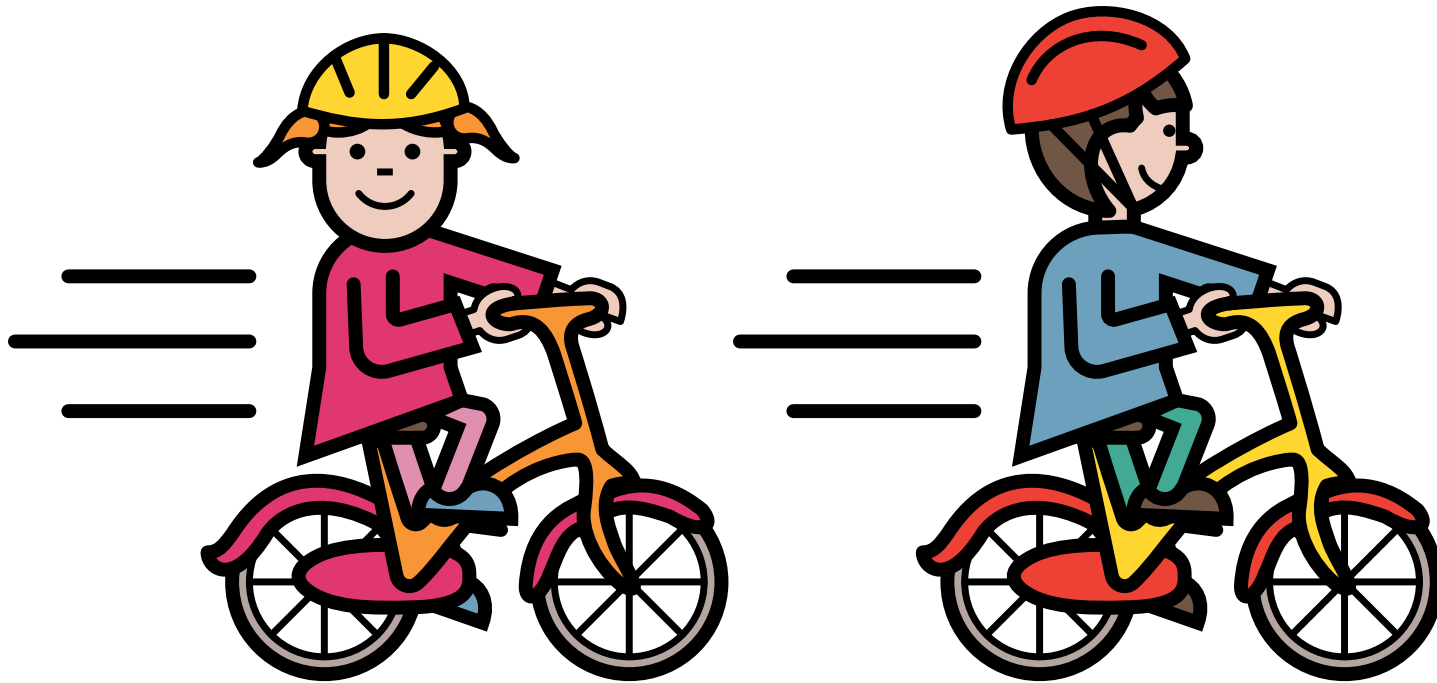
Which foods contain galactose?

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.

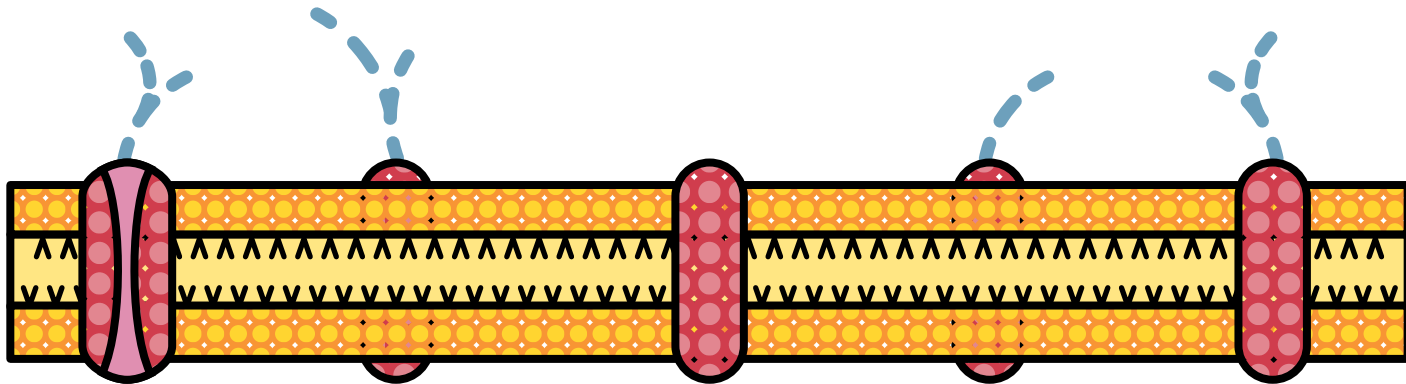


What else does galactose do?

Galactose is a building block of carbohydrate chains.

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

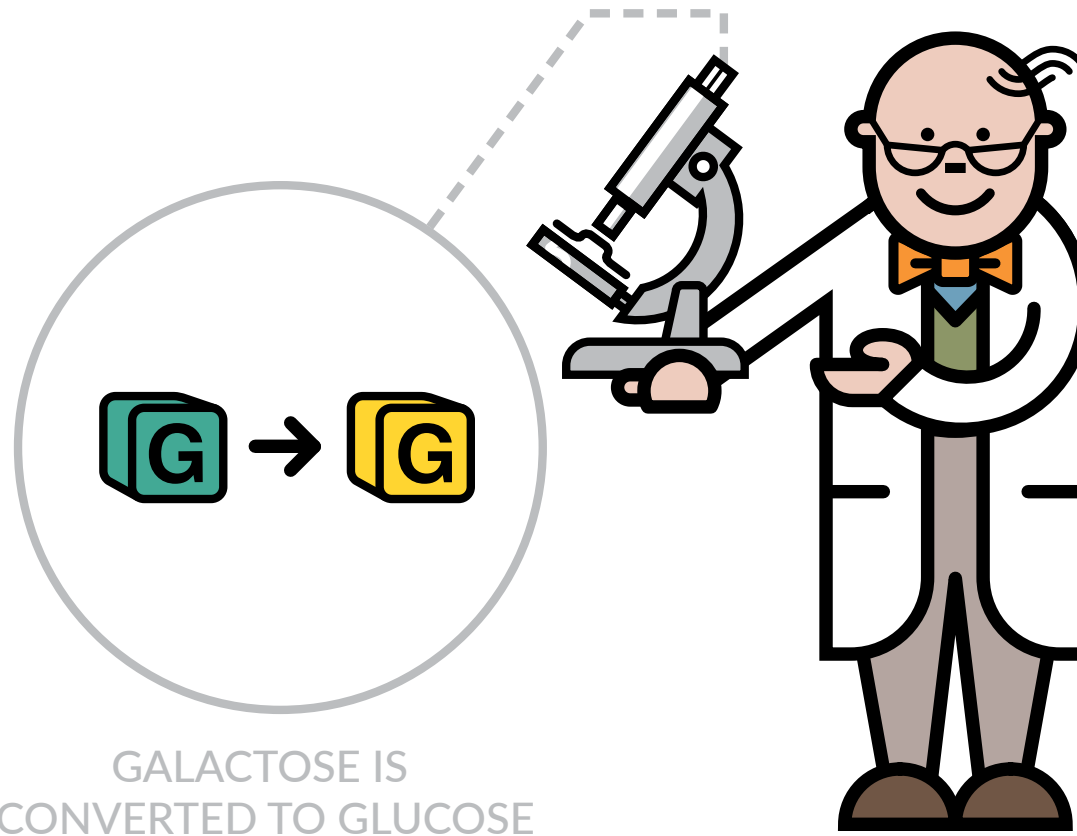
These are important in cell structure.



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**.

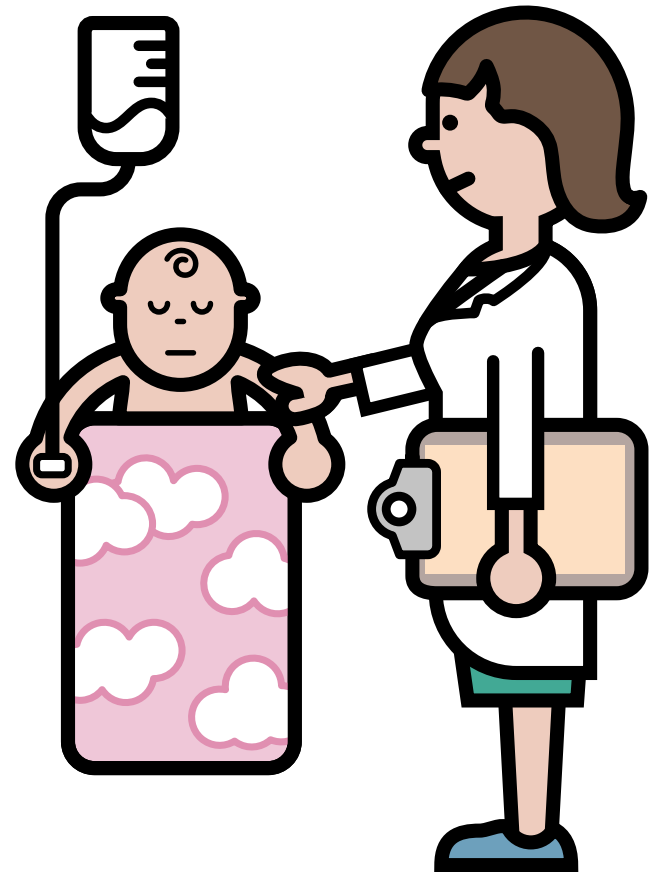
This leads to a build up of galactose and other chemicals leading to symptoms.



What can go wrong in Galactosaemia?

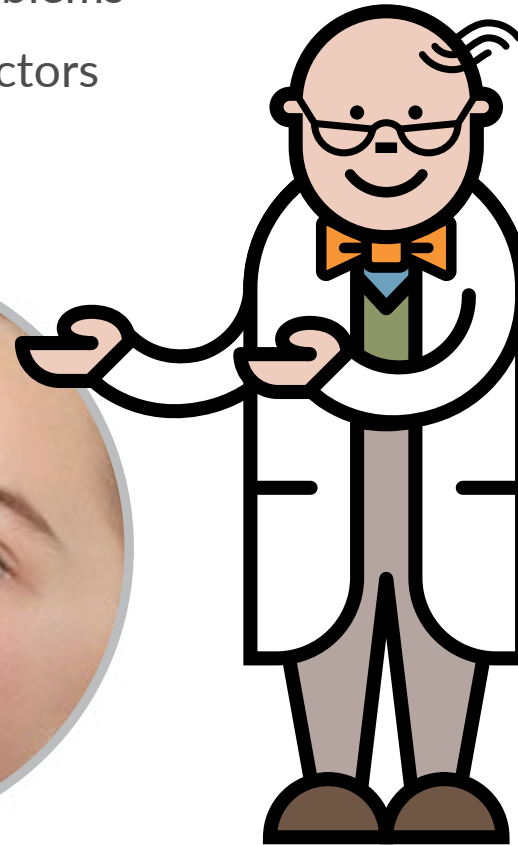
Many babies are very poorly and early symptoms include:

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



What happens with treatment?

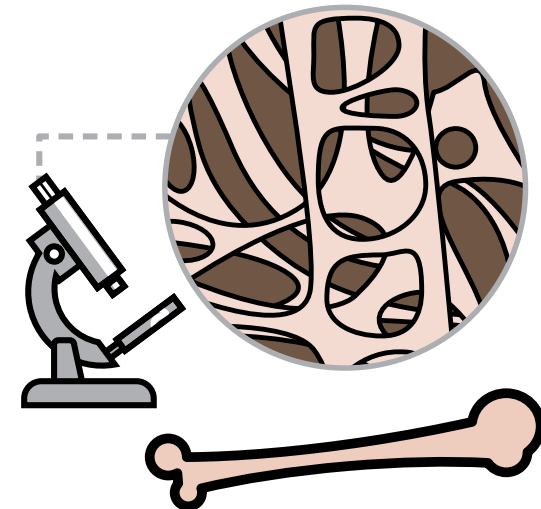
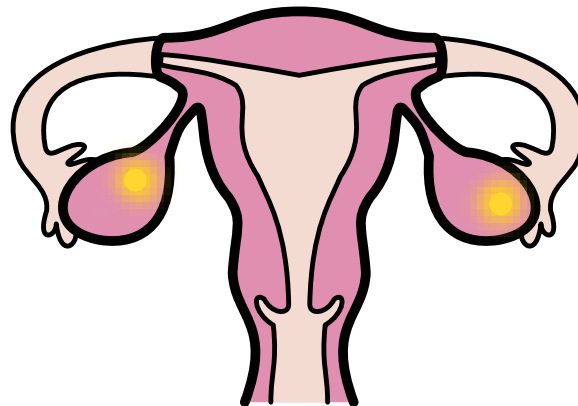
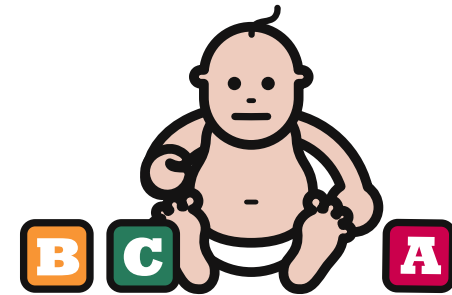
- Once treatment has been started, babies generally start to improve within a few days
- There should be no long-term liver problems
- Cataracts usually disappear but the doctors will continue to monitor the eyes



...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?

Galactose is found in breast milk, infant formula and many foods. It is therefore necessary to:

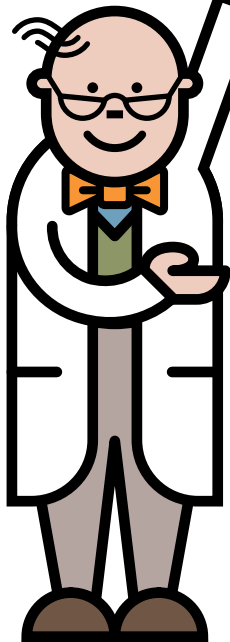
Avoid all animal milk (including breast milk)



Avoid yoghurt, most cheese and milk products



Avoid milk in packet and processed foods



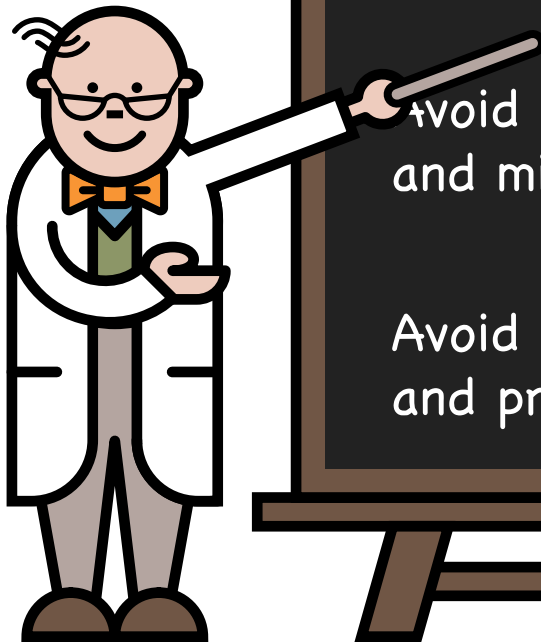
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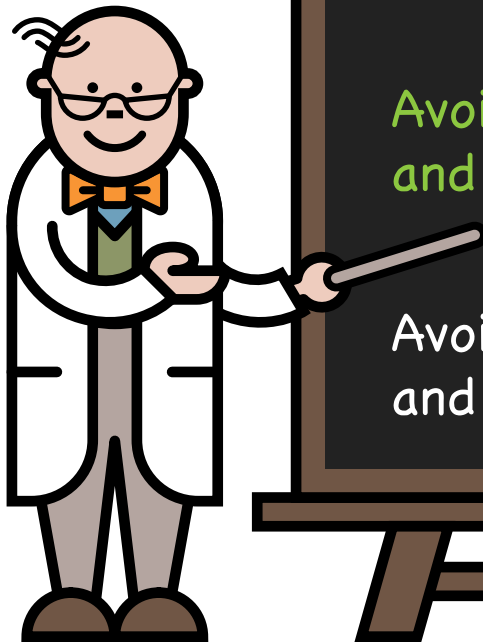
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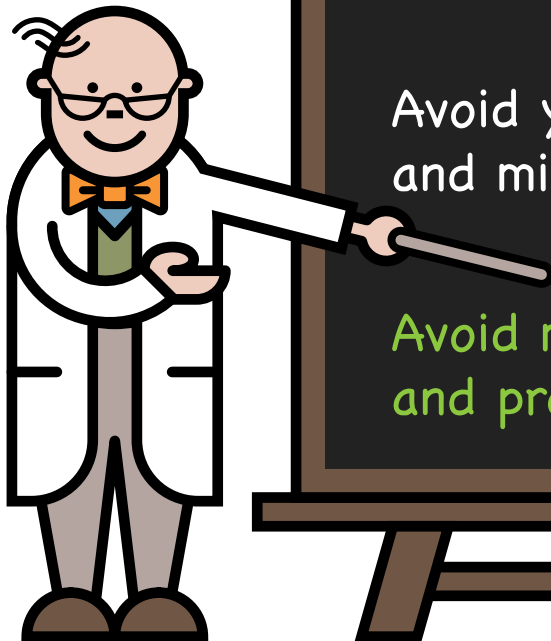
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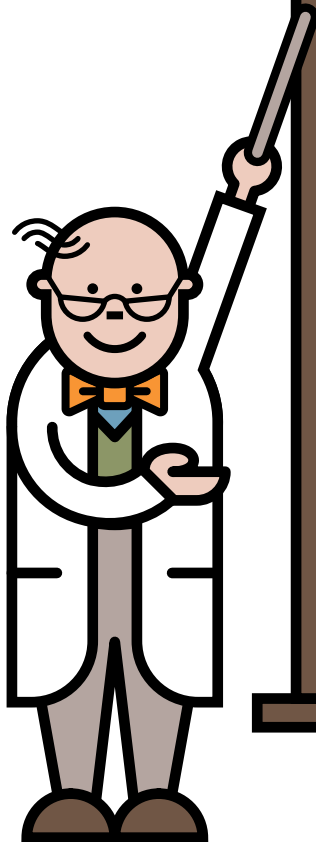
Babies are given a
milk-free infant formula



They are given milk-free
weaning foods



Check all labels to see
foods and medicines are
milk-free



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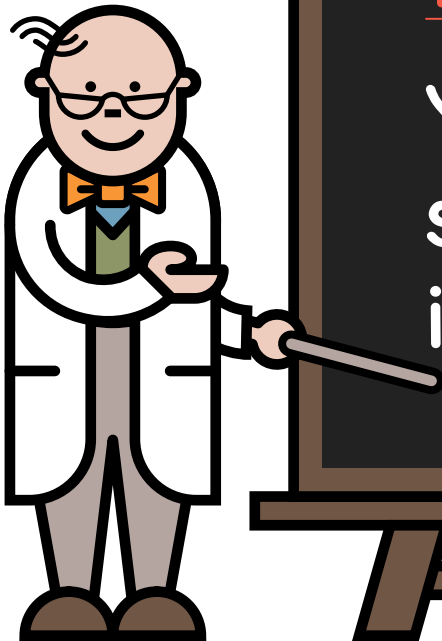
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Key message

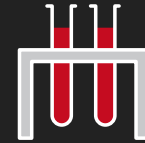
It is essential that breast milk and all standard infant formula is stopped.

Your baby will be given a special milk-free formula instead.



How is Galactosaemia monitored?

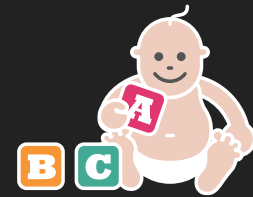
Blood tests



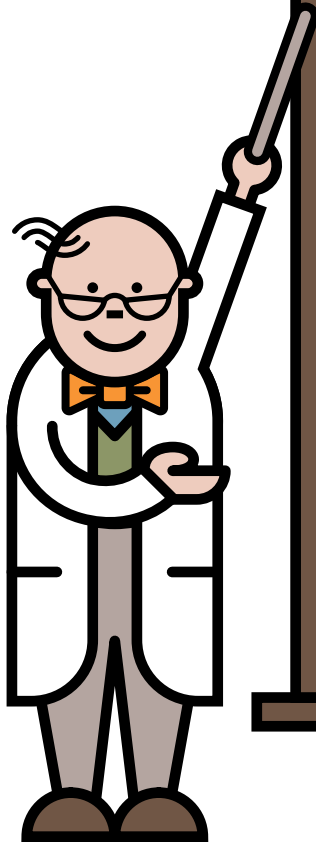
Height and weight



Child development
and eye checks



Regular diet checks
by the dietitian



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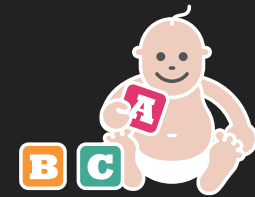
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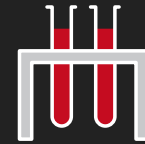
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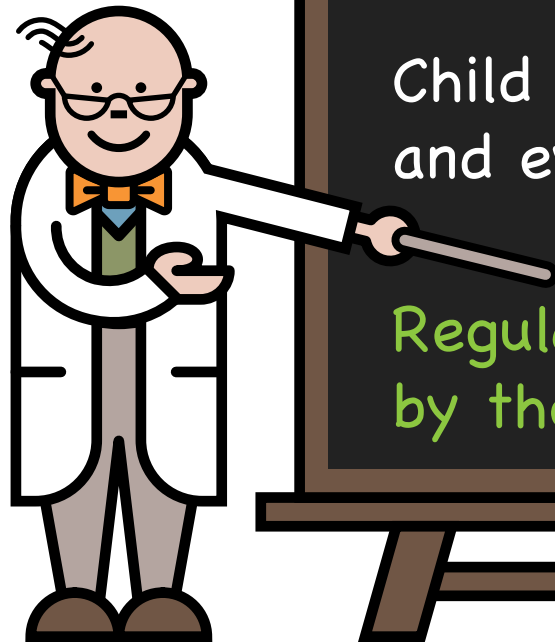
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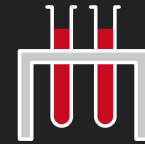
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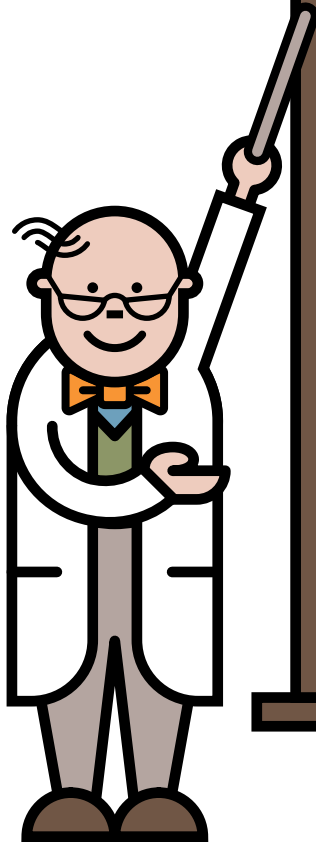
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Chromosomes, genes, mutations



Humans have chromosomes composed of DNA.



Genes are pieces of DNA that carry the genetic instruction. Each chromosome may have several thousand genes.



The word mutation means a change or error in the genetic instruction.

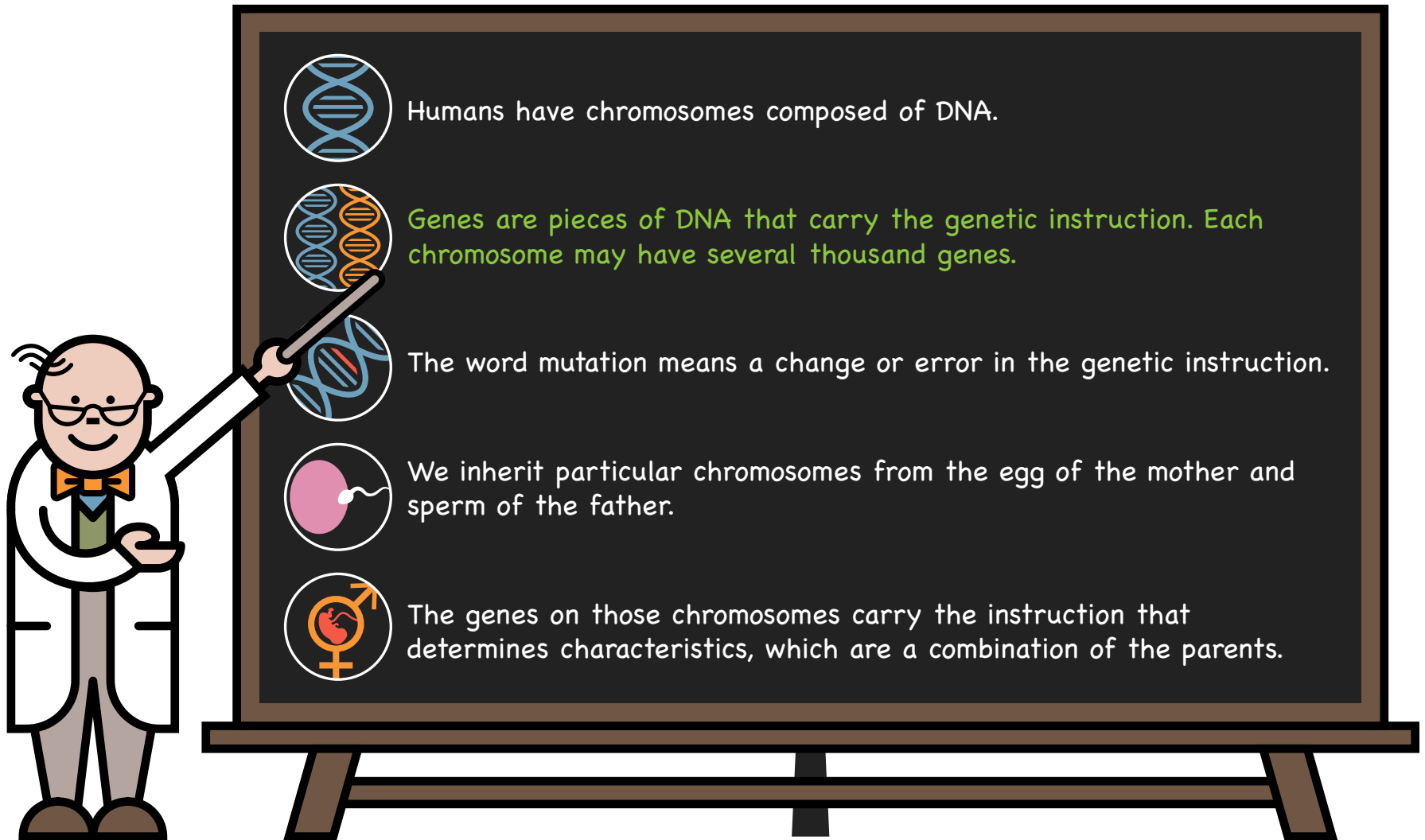



We inherit particular chromosomes from the egg of the mother and sperm of the father.





The genes on those chromosomes carry the instruction that determines characteristics, which are a combination of the parents.


Chromosomes, genes, mutations




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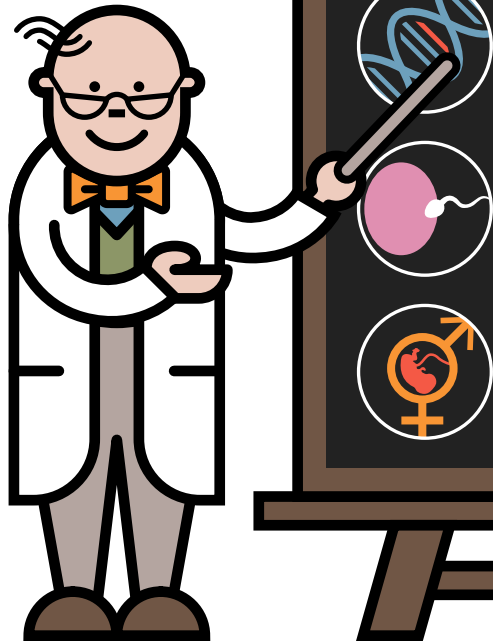
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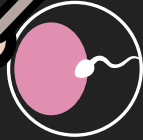
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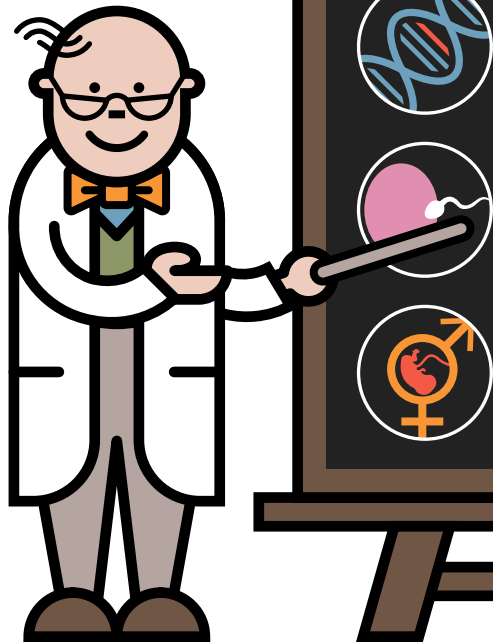


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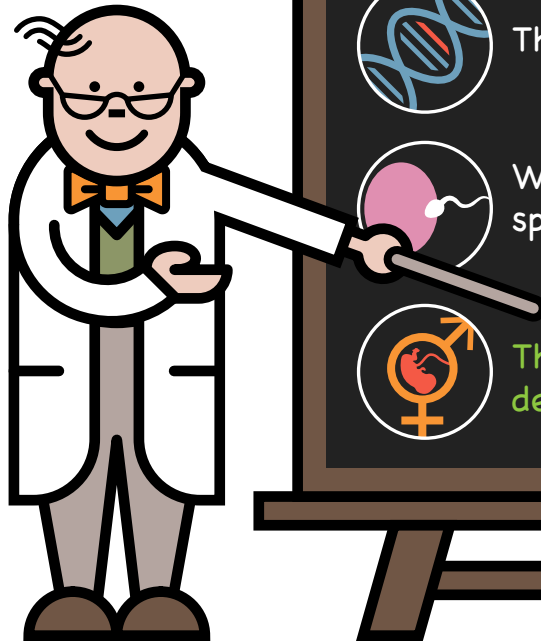


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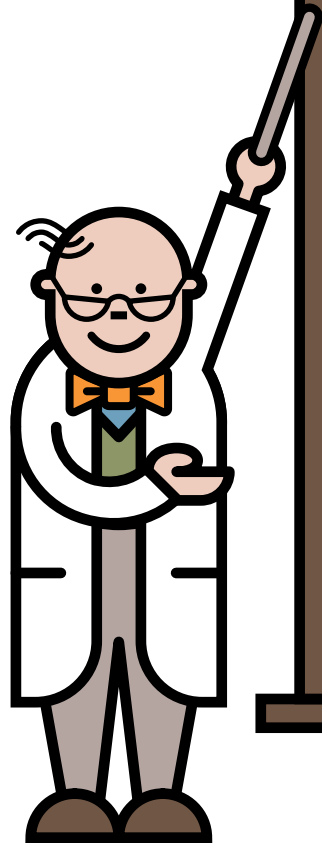


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Inheritance



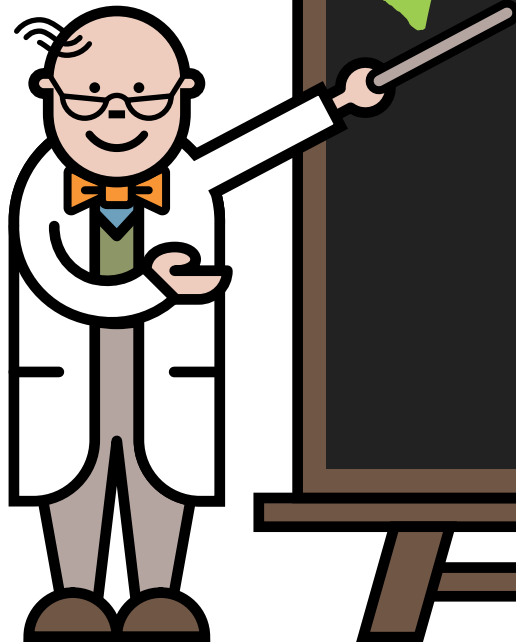
Galactosaemia is an inherited condition. There is nothing that could have been done to prevent your baby from having galactosaemia.

Everyone has a pair of genes that make the galactose-1-phosphate uridyl transferase enzyme. In children with galactosaemia neither of these genes work correctly. These children inherit one non-working galactosaemia gene from each parent.

Parents of children with galactosaemia are carriers of the condition.

Carriers do not have galactosaemia because the other gene of this pair is working correctly.

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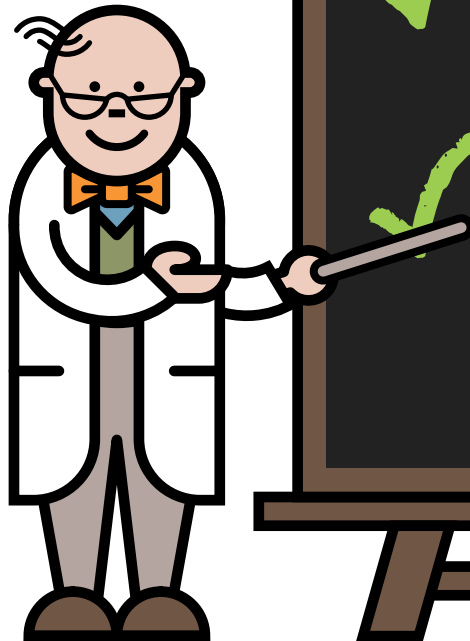


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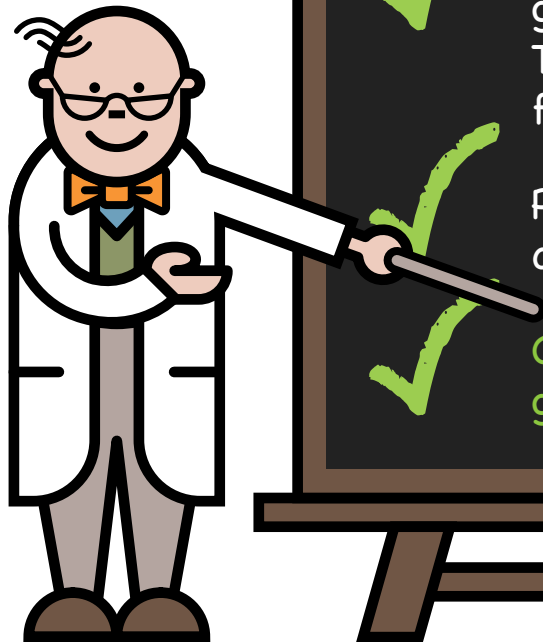
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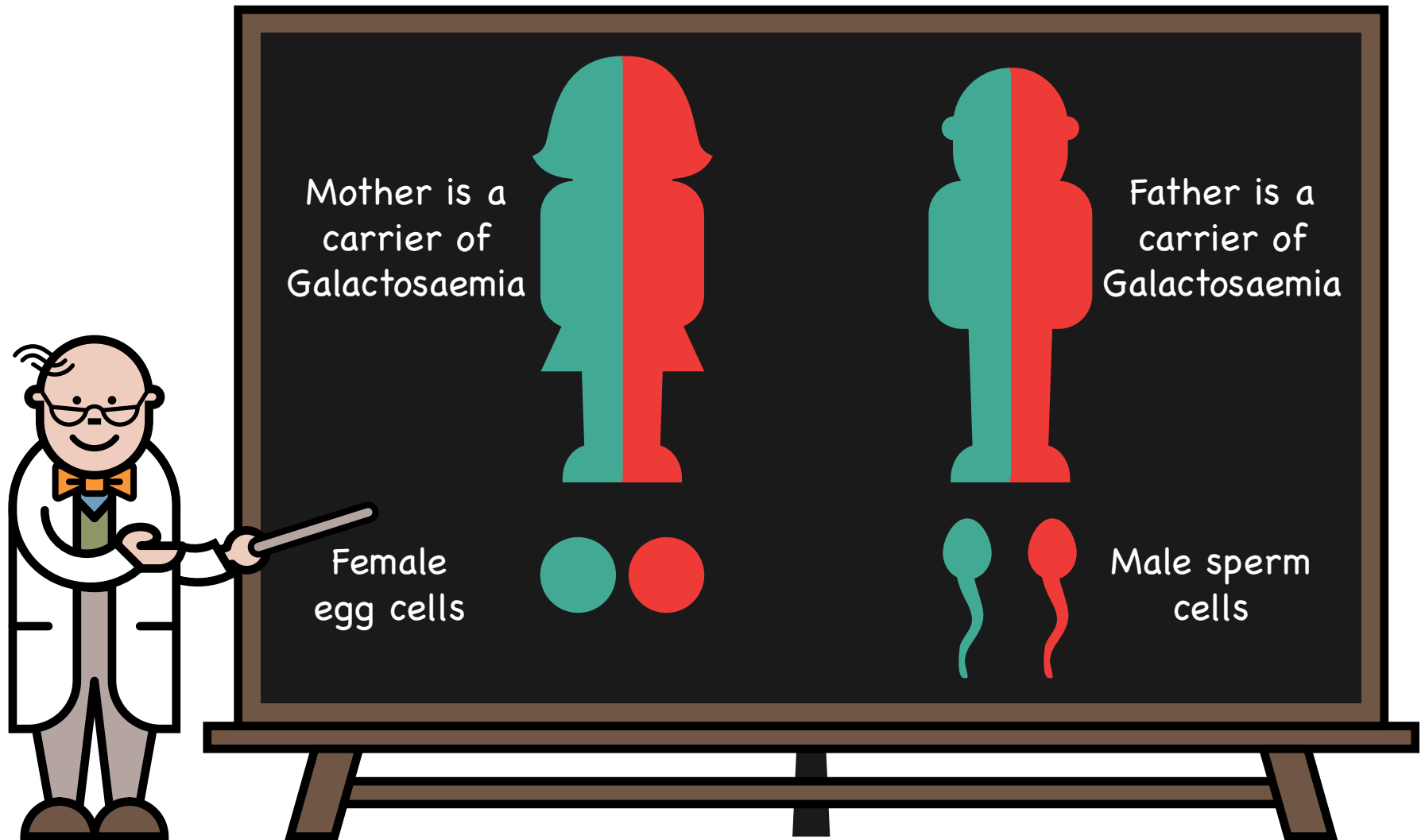
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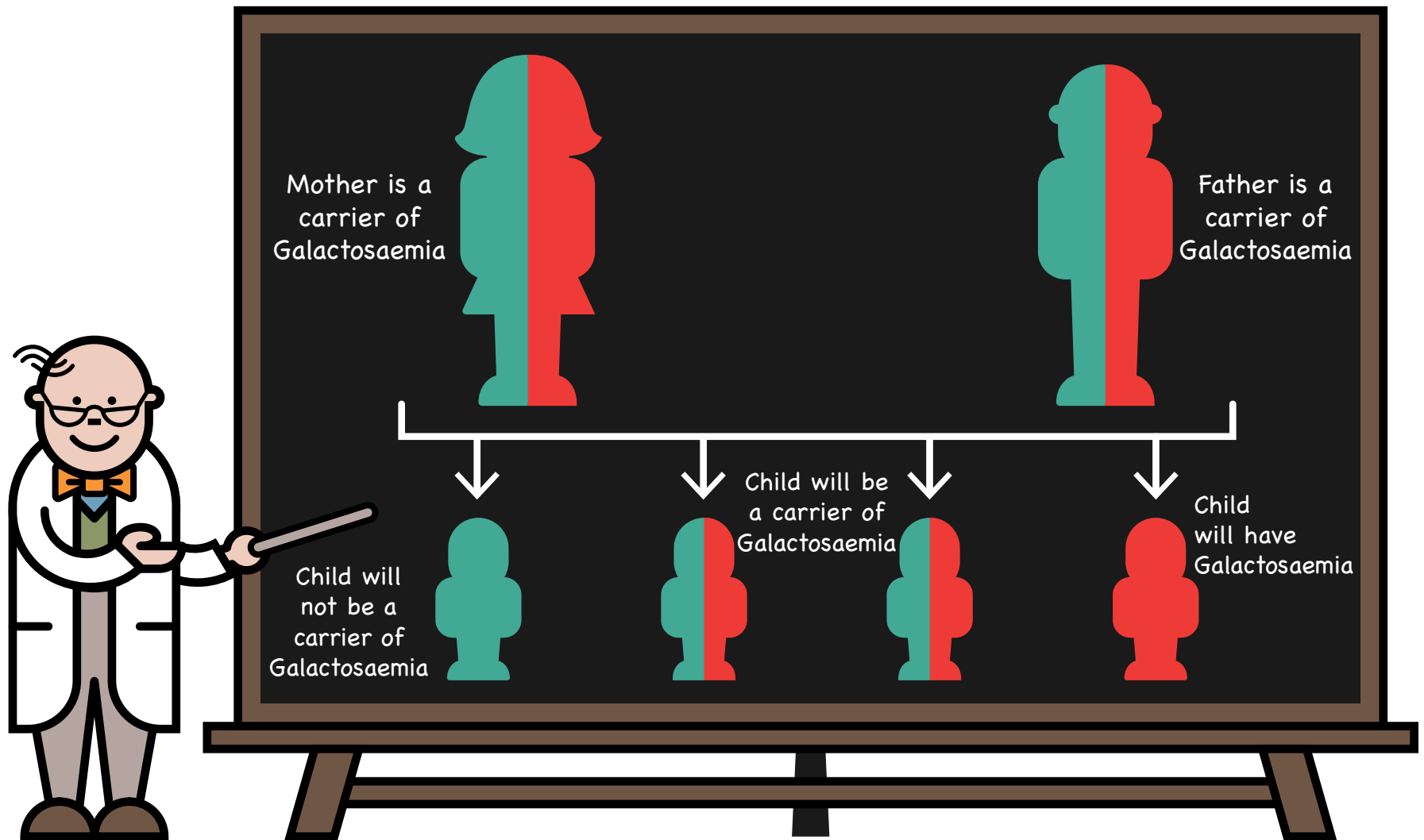
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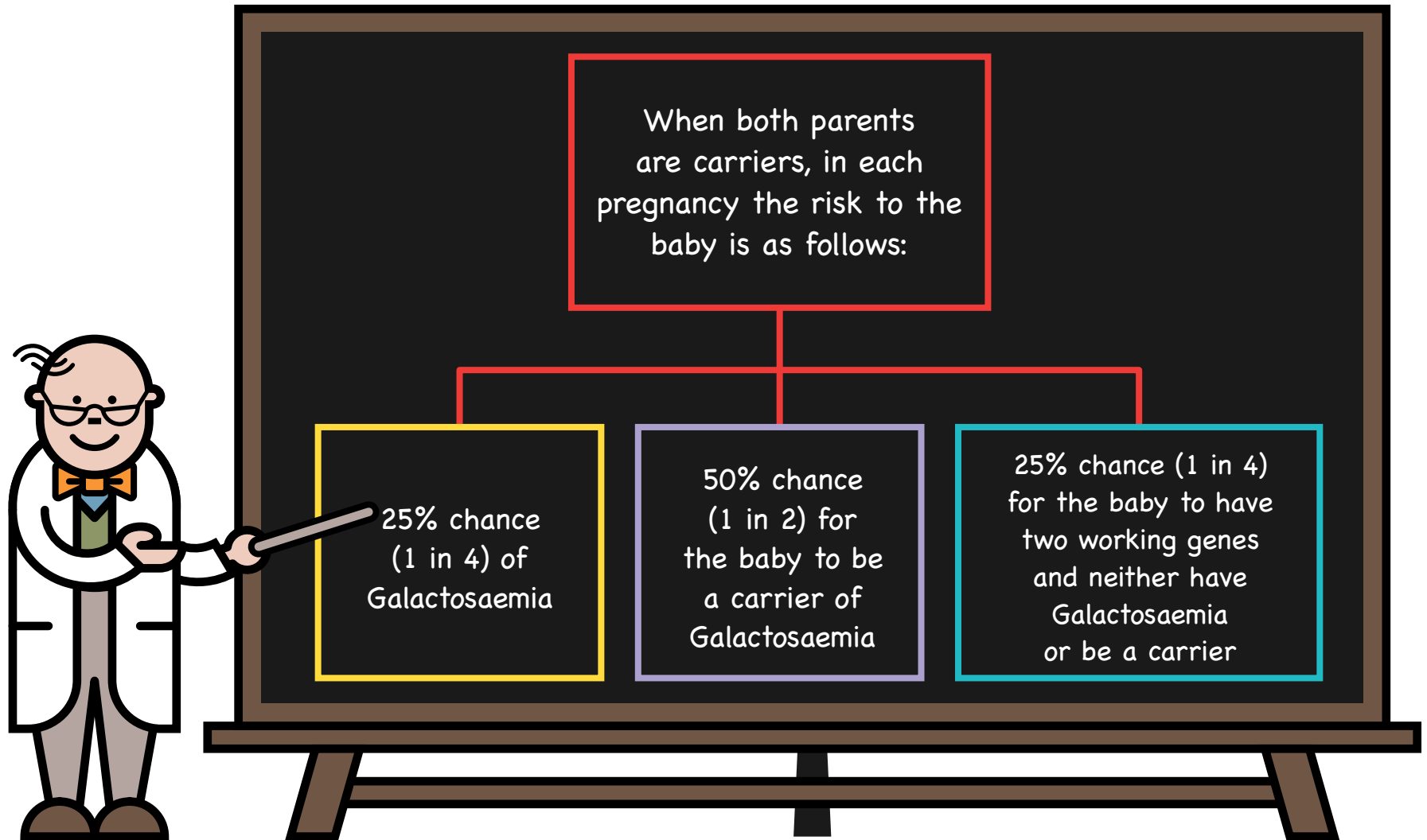
Inheritance – Autosomal-recessive (carriers of Galactosaemia)



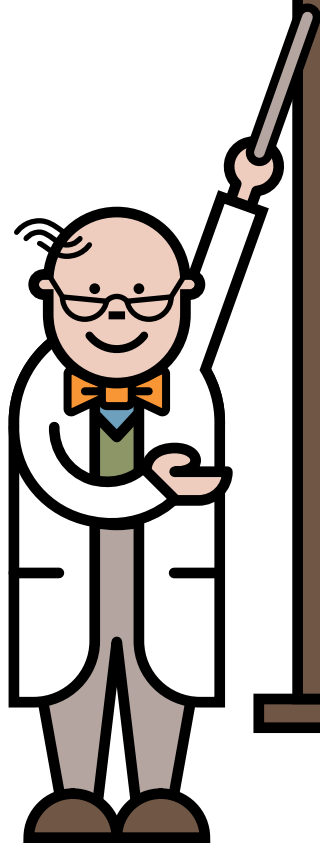
Inheritance – Autosomal recessive – possible combinations



Future pregnancies



Take home messages



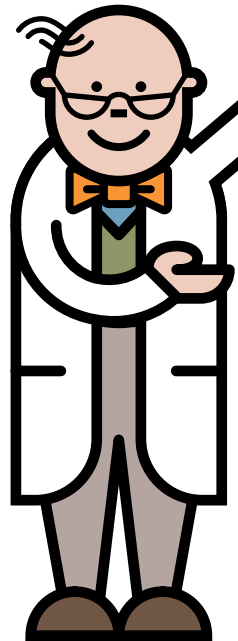
Galactosaemia is a serious inherited metabolic disorder that may cause life threatening symptoms if left untreated.

Symptoms will improve if treated with a milk-free special formula and milk-free diet.

Milk and milk containing products are avoided. All food labels must be checked.

People with galactosaemia should lead healthy and active lives, but some may develop learning difficulties and fertility problems.

Take home messages



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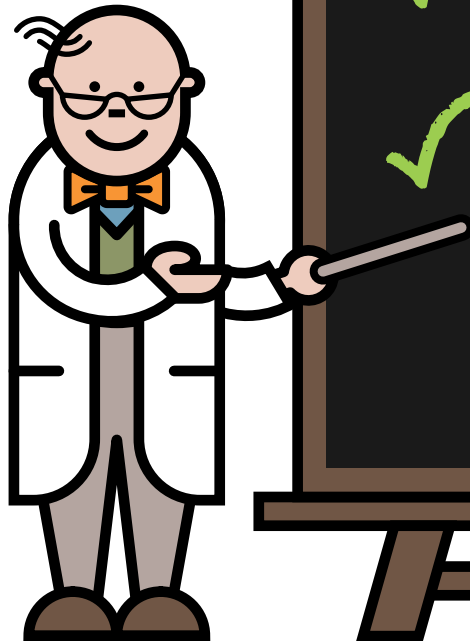


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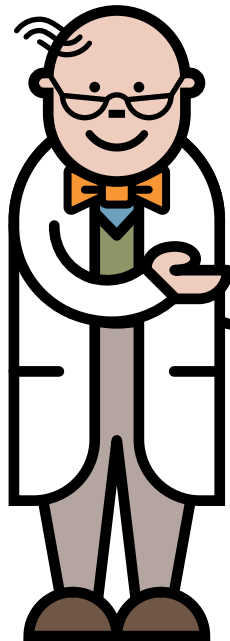
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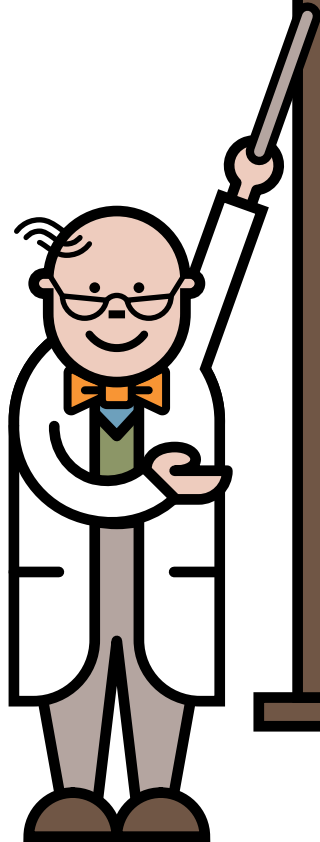
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Helpful hints



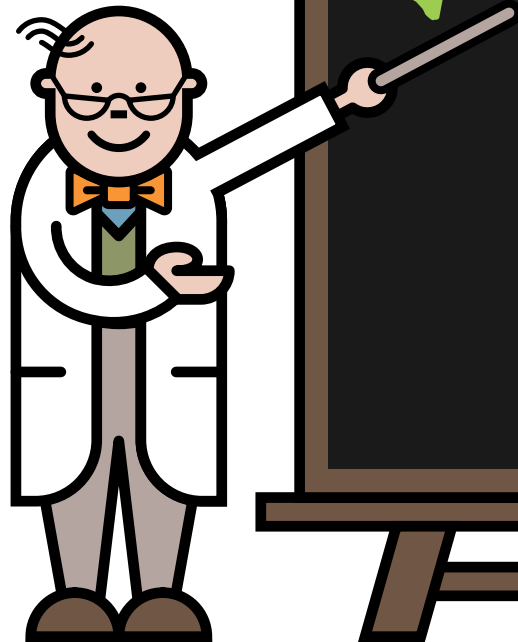
Always ensure you have a good supply of your milk-free formula and it is in date.

Your special milk-free formula is prescribed by your GP. This is obtained via a pharmacy.

Check all medications for lactose or galactose on the ingredient list.

Remember to keep contact numbers of your metabolic team to hand.

Helpful hints



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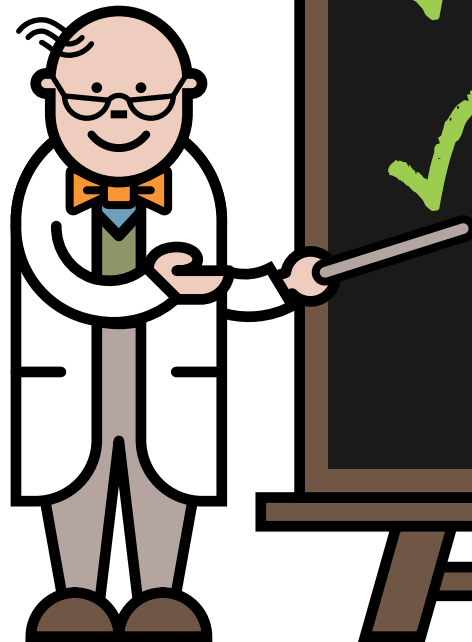


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Who's who

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

BIMDG

British Inherited Metabolic Diseases Group



www.bimdg.org.uk



www.nutricia.co.uk



www.galactosaemia.org