

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 2, MAY 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).

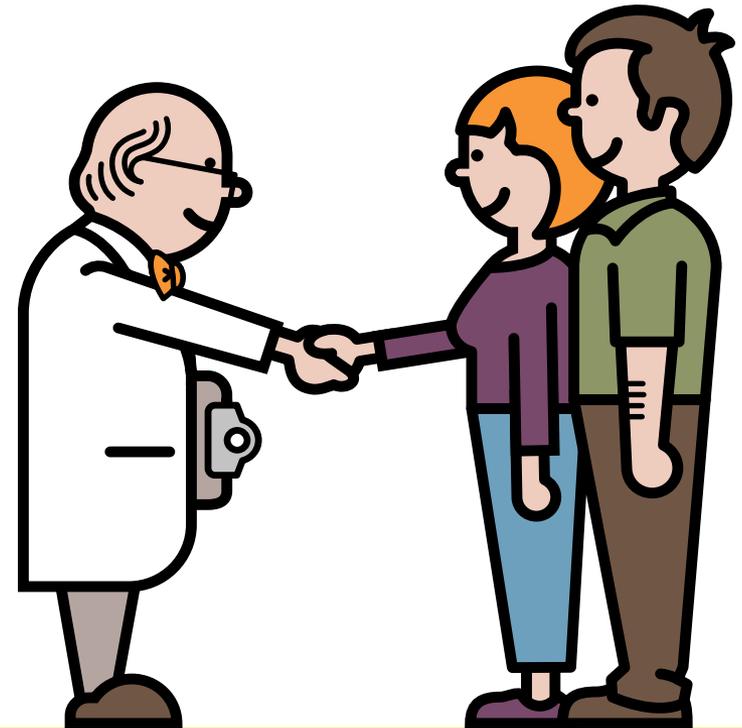
The team are Rachel Skeath, Karen van Wyk, Pat Portnoi and Anita MacDonald. The group is facilitated by Heidi Chan from Nutricia.

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.

Galactosaemia

Information for families following
a new diagnosis



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Tools Enabling Metabolic Parents LEarning

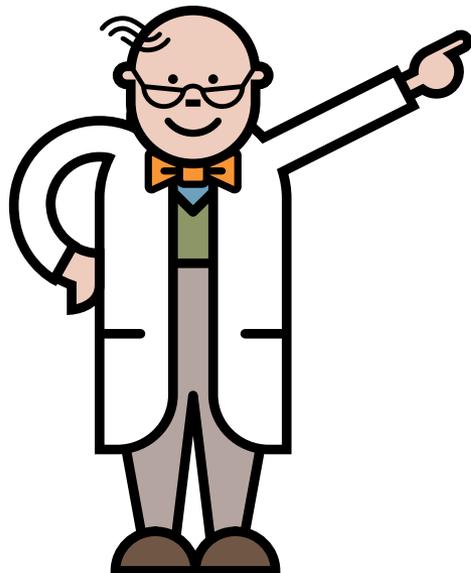
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What is Galactosaemia?

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?

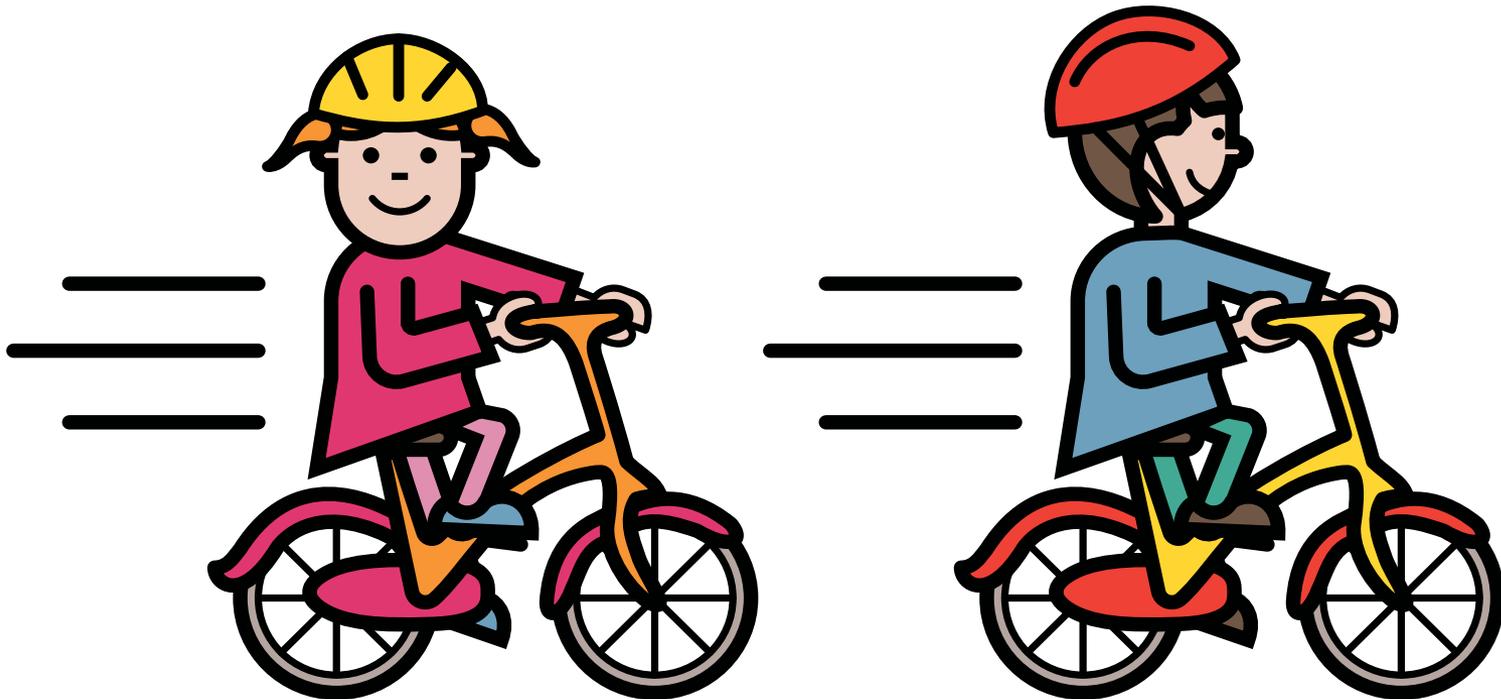
Galactose mainly comes from lactose.

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.

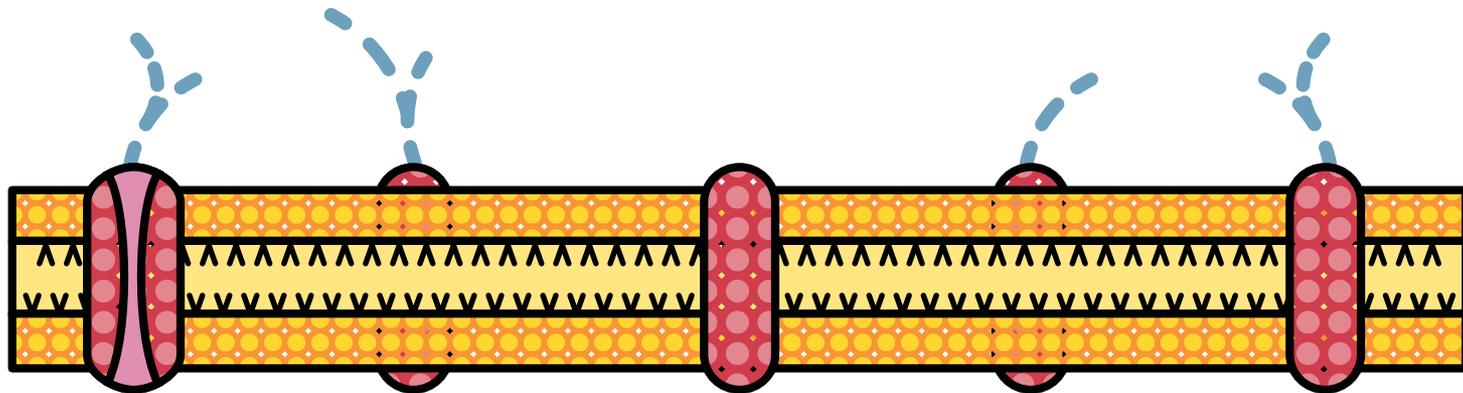


What else does galactose do?

Galactose is a building block of carbohydrate chains.

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

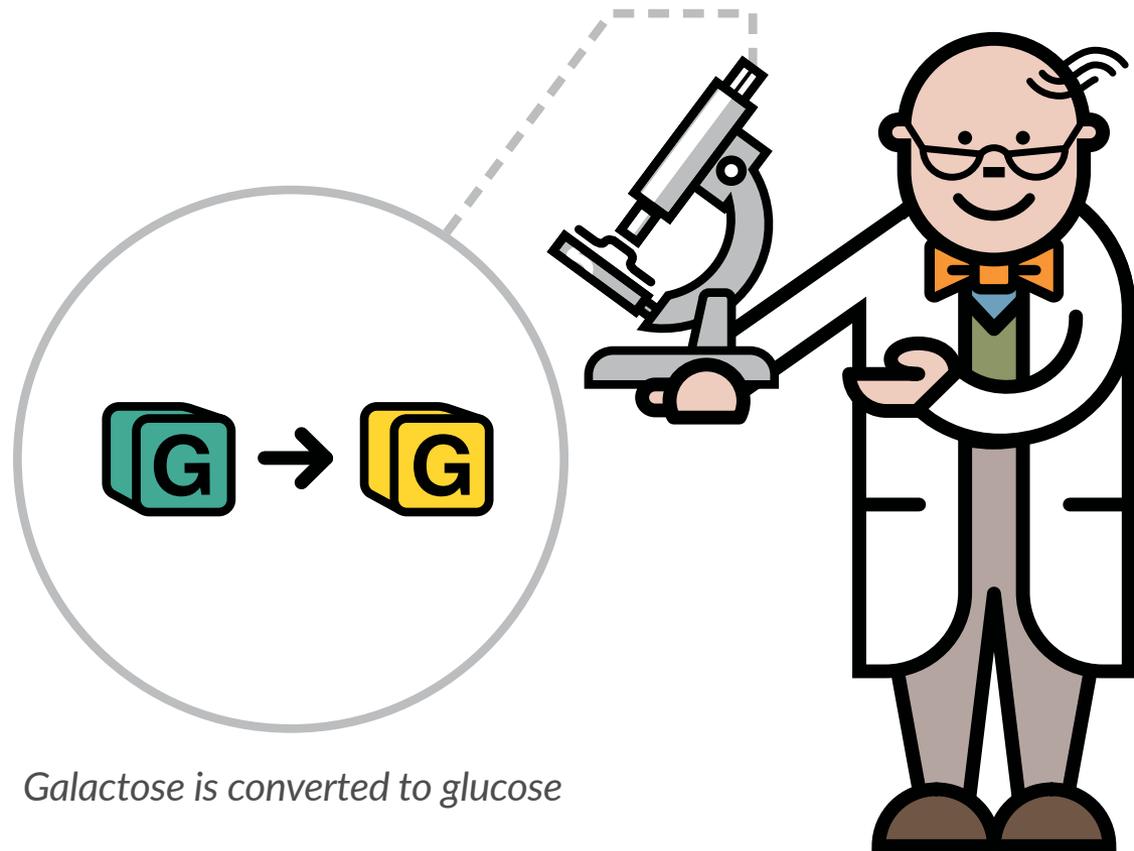
These are important in cell structure.



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).



Galactose is converted to glucose

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**.

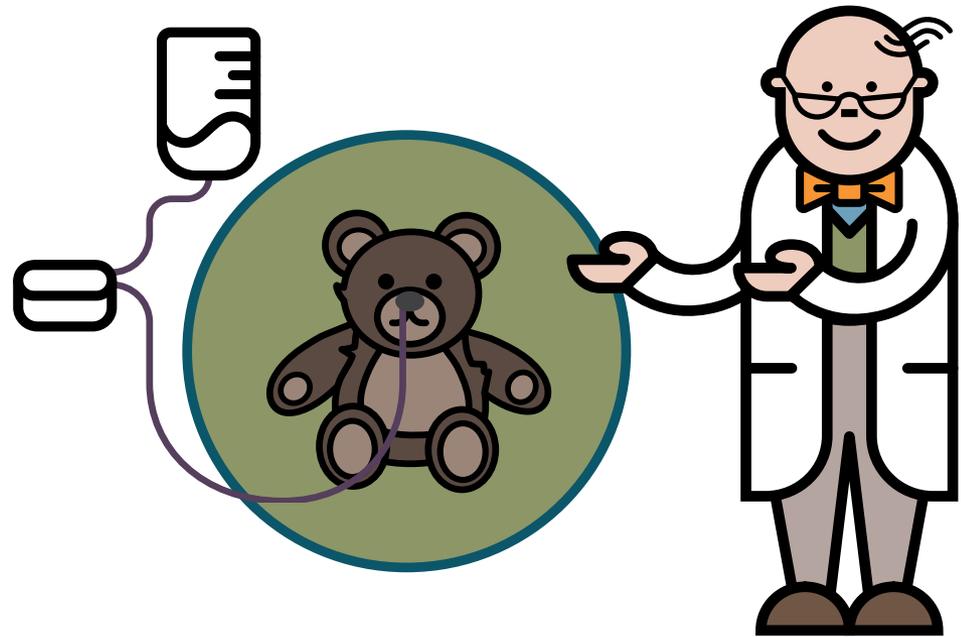
This leads to a build up of galactose and other chemicals leading to symptoms.



What can go wrong in Galactosaemia?

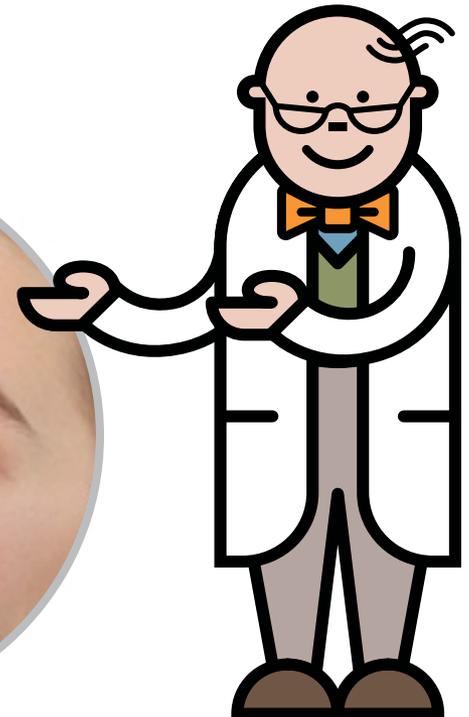
Many babies are very poorly and early symptoms include:

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



What happens with management?

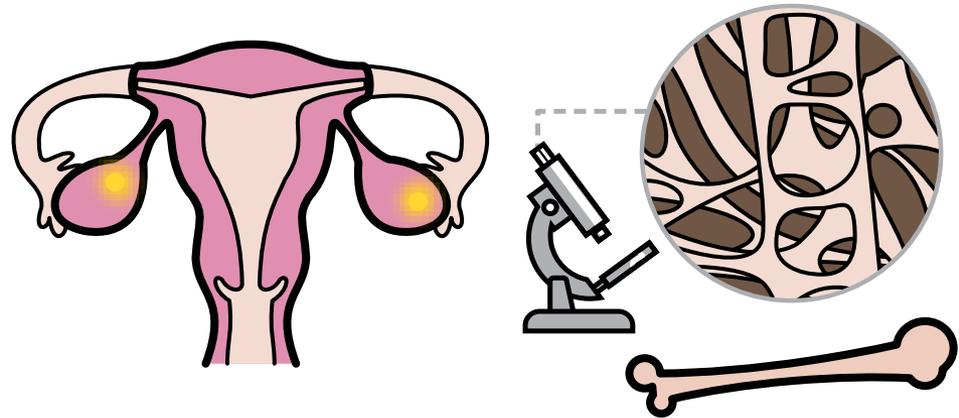
- Once management has been started, babies generally start to improve within a few days
- There should be no long-term liver problems
- Cataracts usually disappear but the doctors will continue to monitor the eyes



...but even with management

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 managed day to day?

Galactose is found in breast milk, infant feed 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



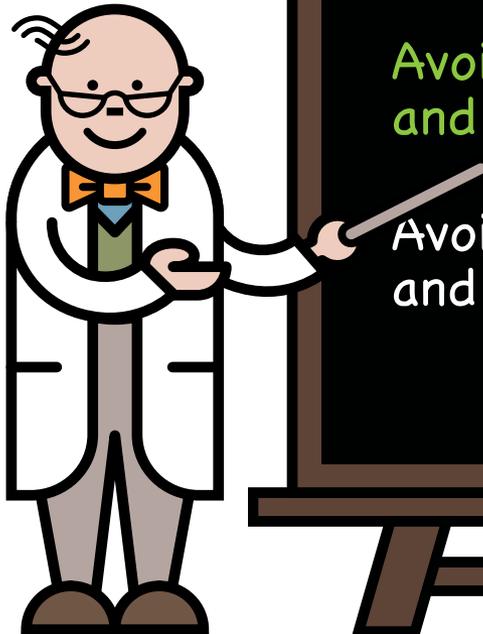
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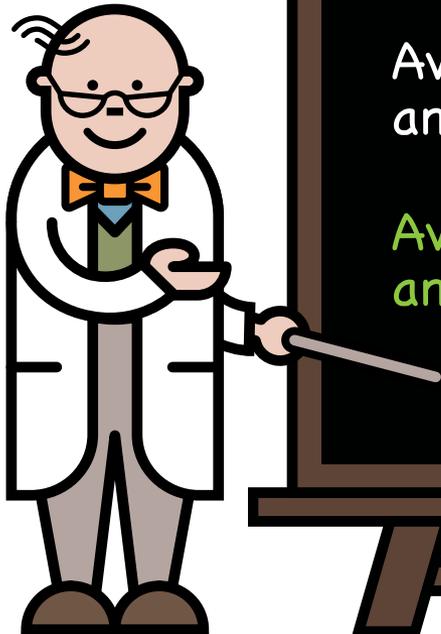
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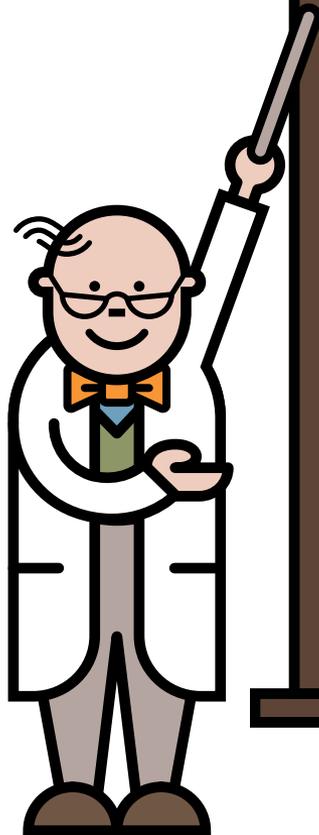
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How is Galactosaemia managed day to day?



Babies are given a milk-free infant feed



They are given milk-free weaning foods



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



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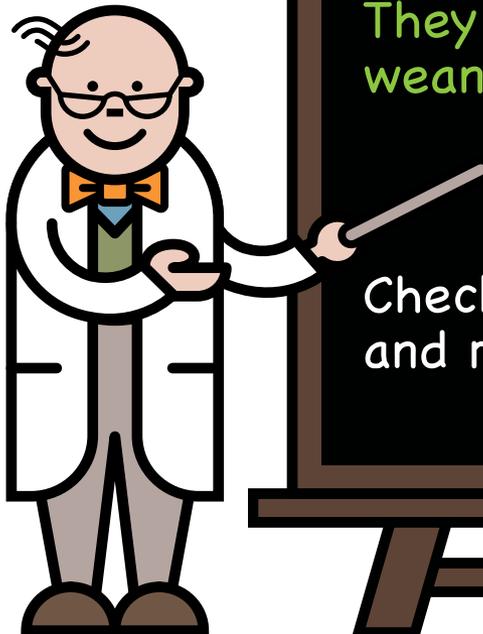
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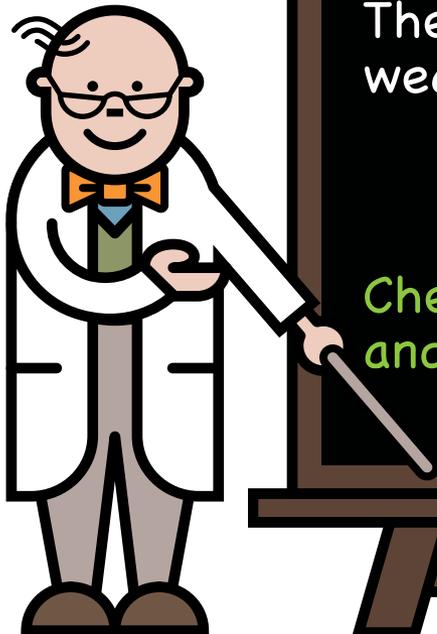
Babies are given a milk-free infant feed



They are given milk-free weaning foods



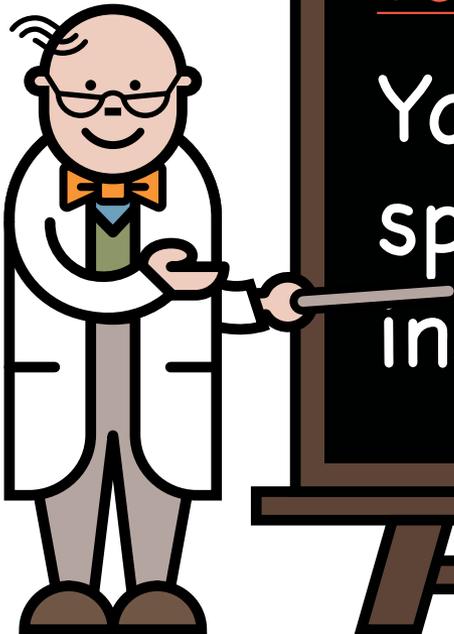
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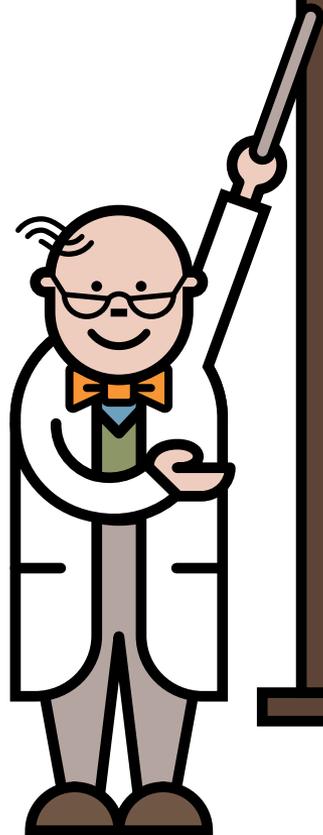
Key message

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

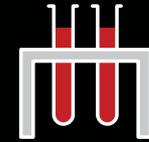
Your baby will be given a special milk-free formula instead.



How is Galactosaemia monitored?



Blood tests



Height and weight



Child development
and eye checks



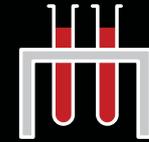
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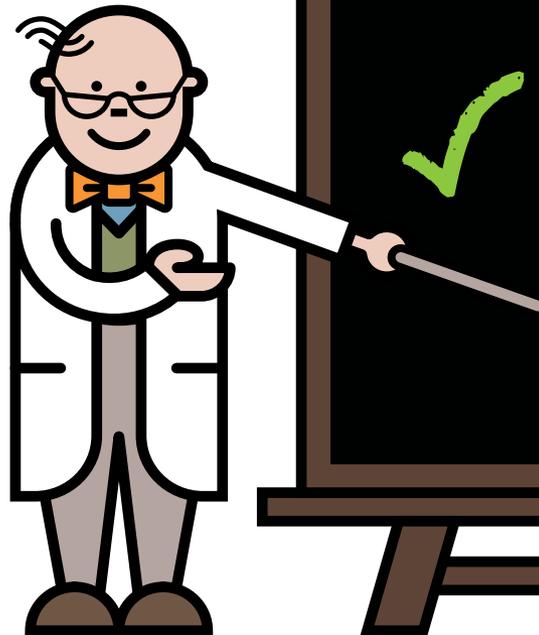
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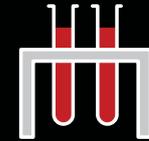
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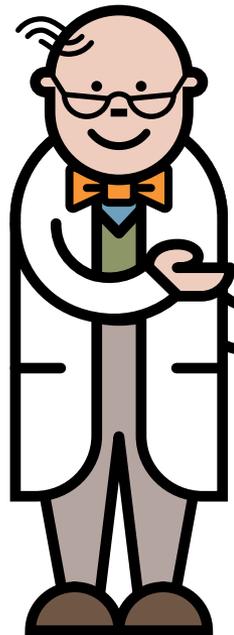
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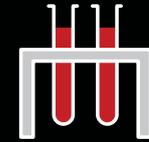
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Chromosomes, genes, mutations



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

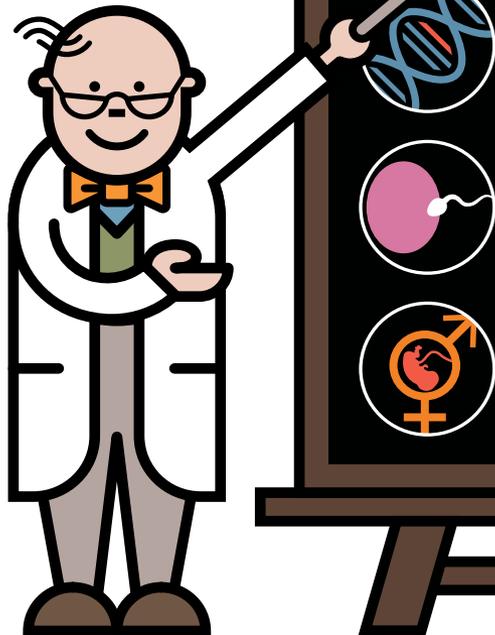


We inherit particular chromosomes from the egg of the mother and sperm of the father



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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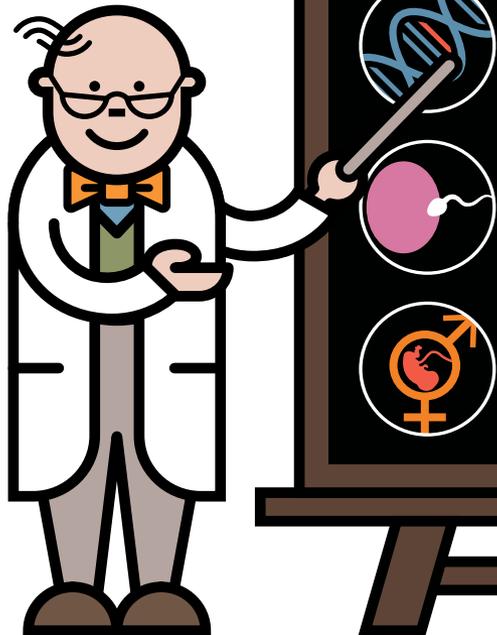


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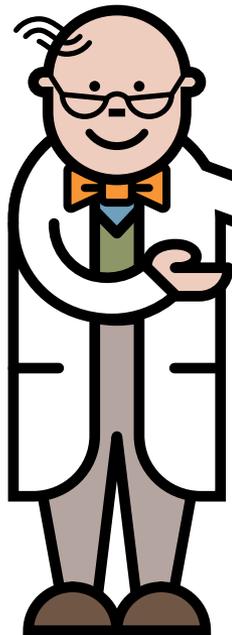


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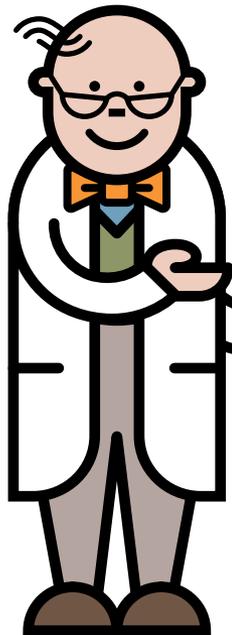


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Inheritance

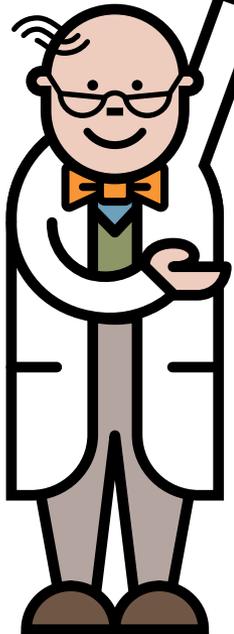


Galactosaemia is an inherited condition. There is nothing that could have been done to prevent your baby from having galactosaemia

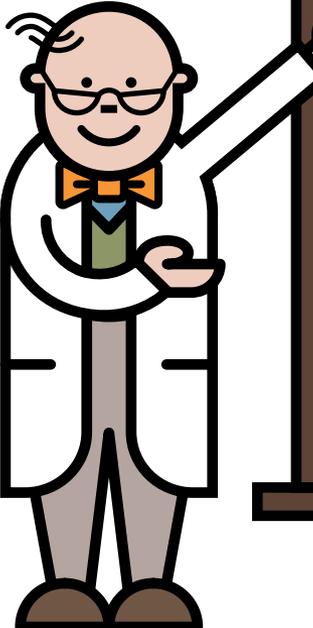
Everyone has a pair of genes that make the galactose-1-phosphate uridyl transferase enzyme. In children with galactosaemia neither of these genes work correctly. These children inherit one non-working galactosaemia gene from each parent

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



Inheritance



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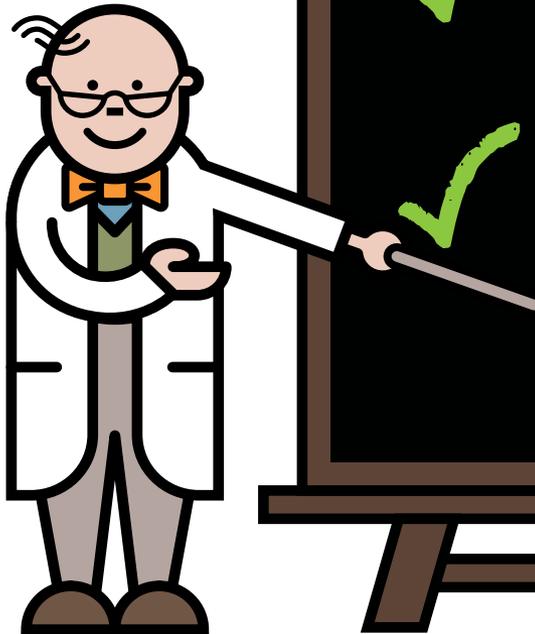
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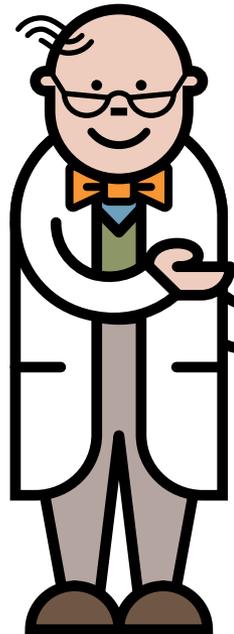
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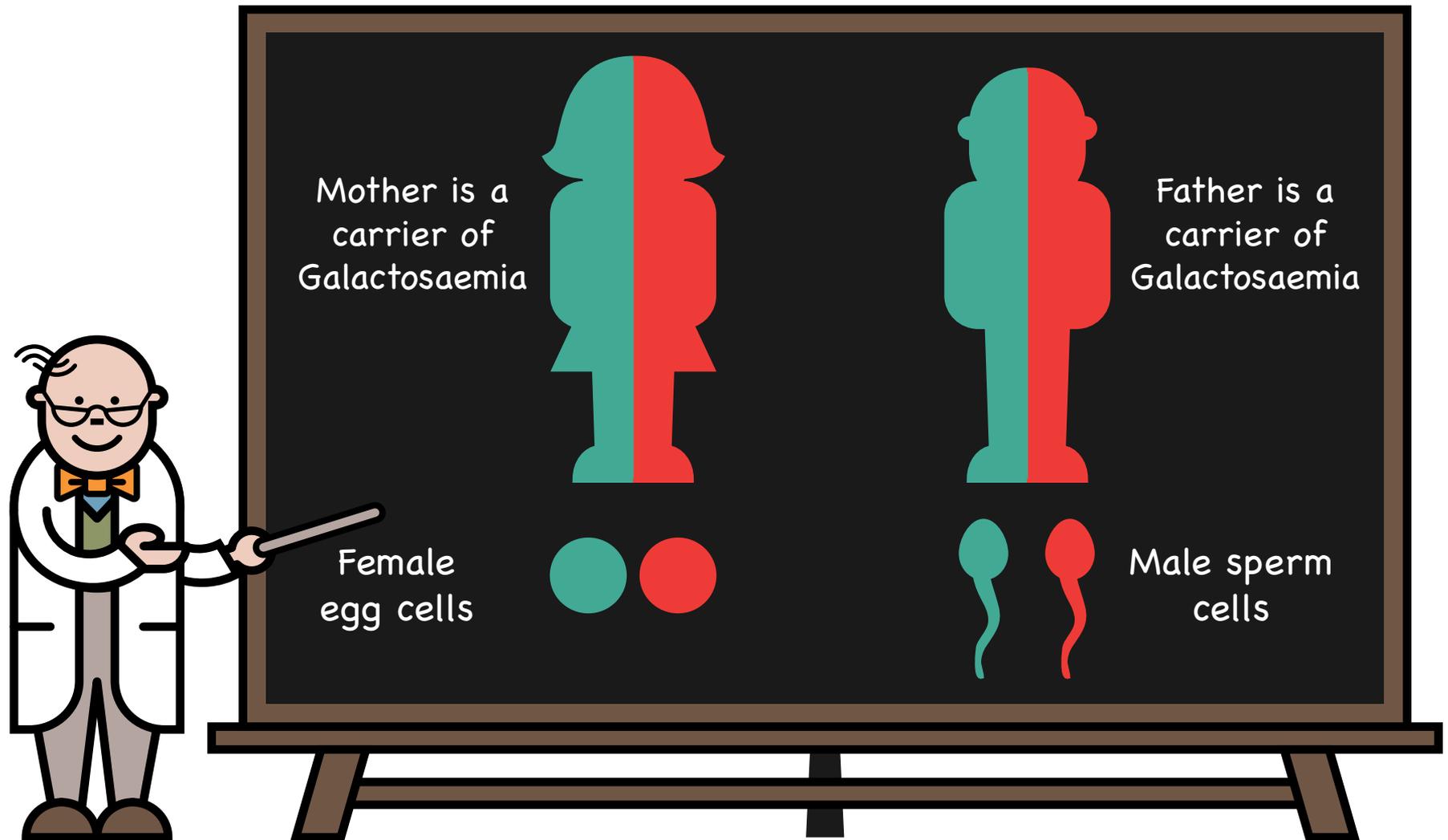


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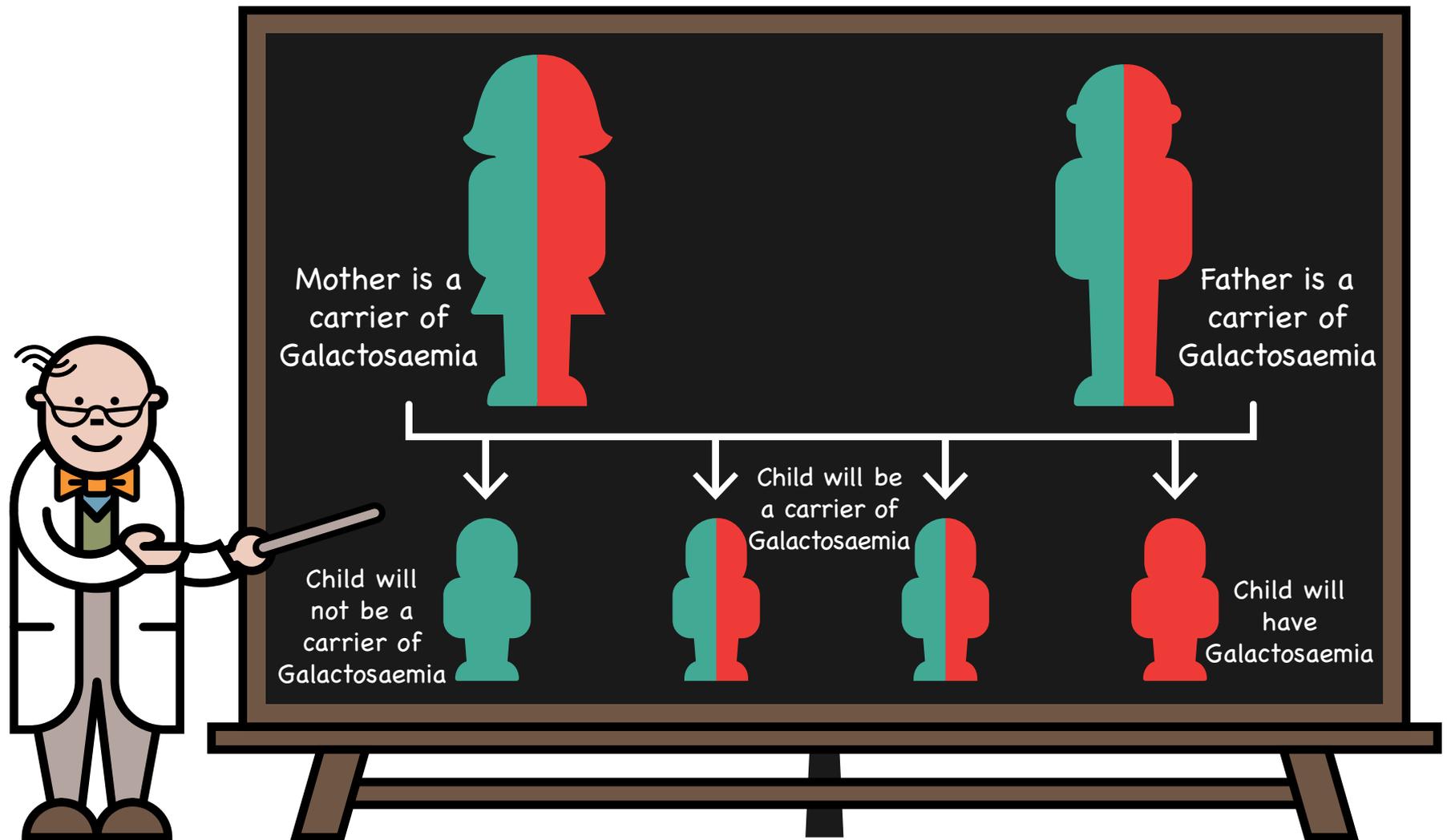


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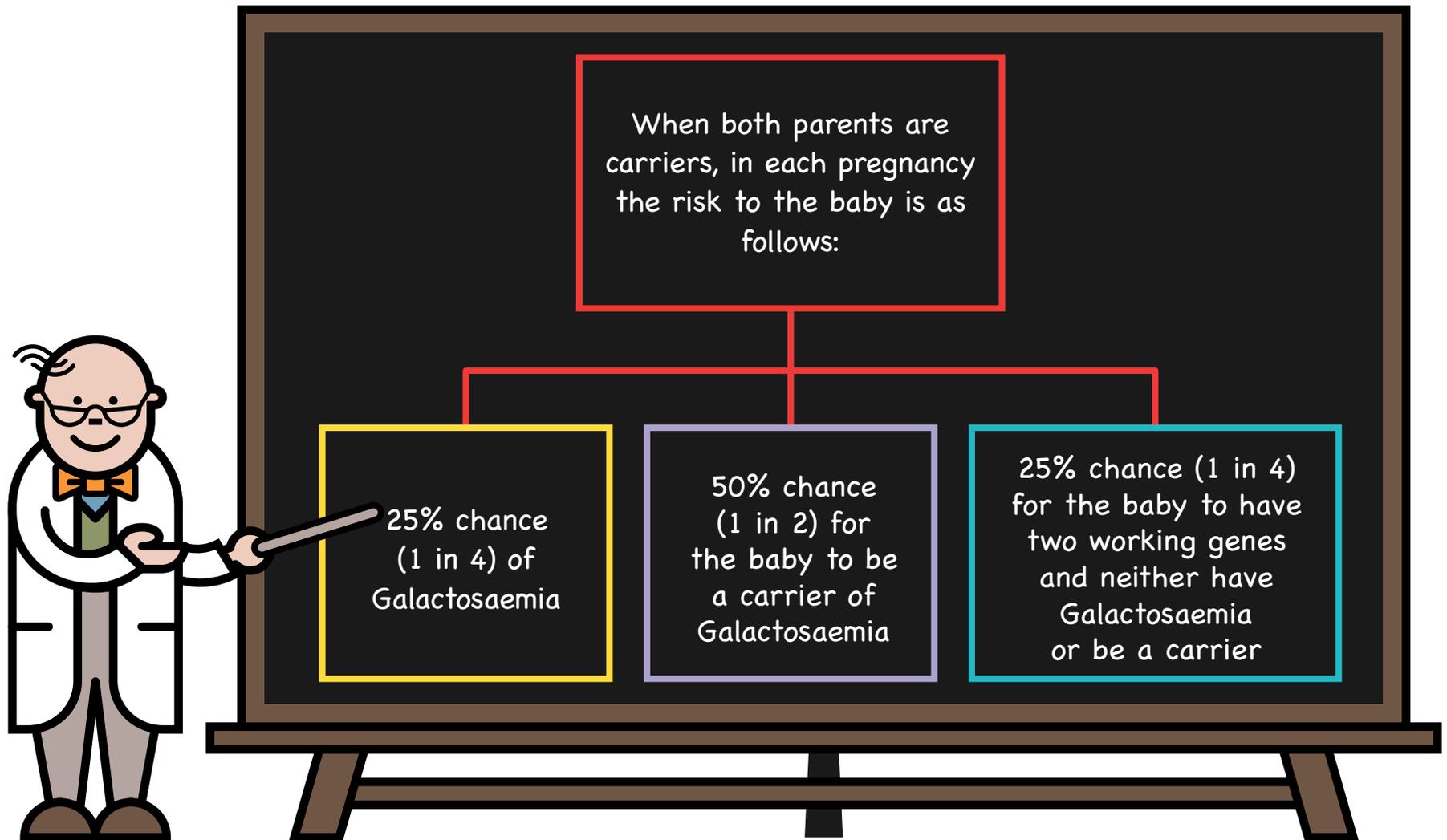
Inheritance – Autosomal-recessive (carriers of Galactosaemia)



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

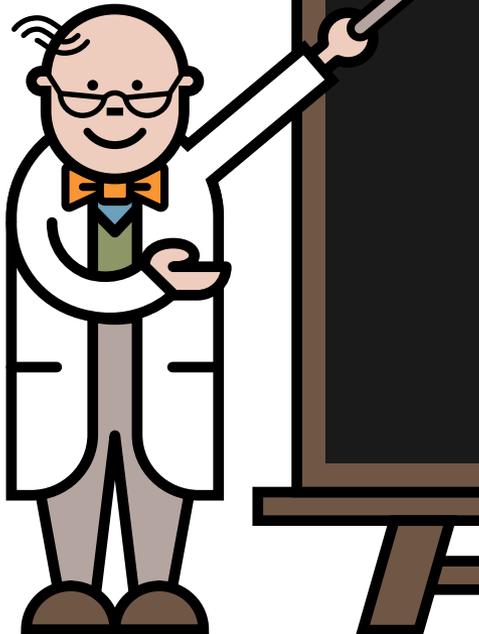
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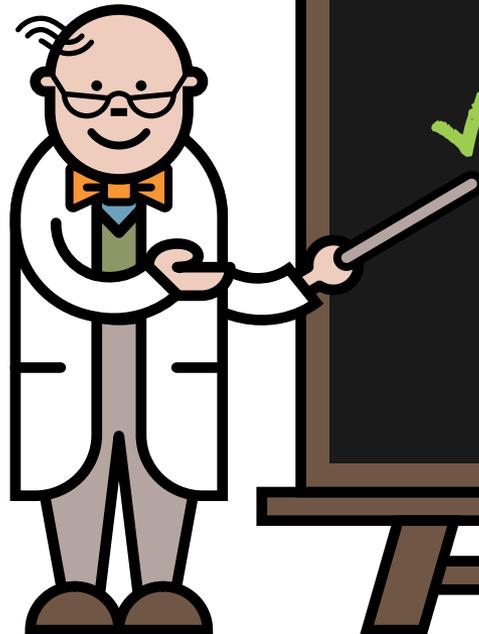
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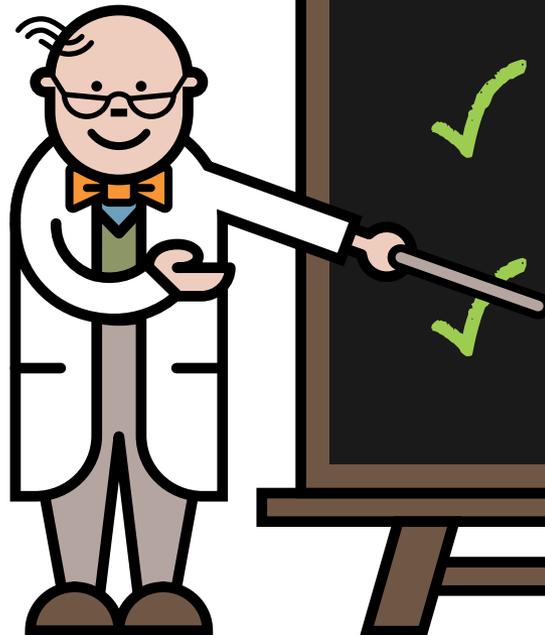
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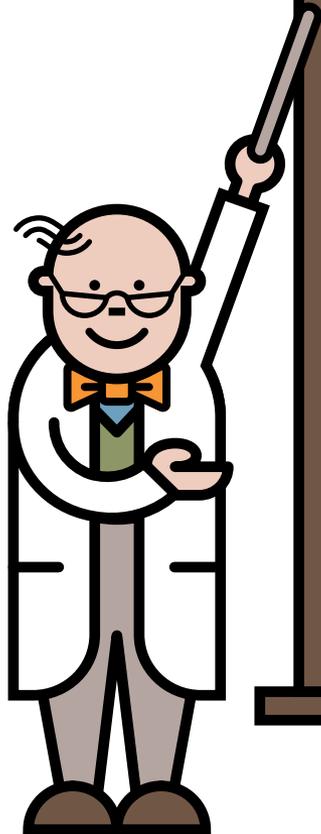
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Helpful hints



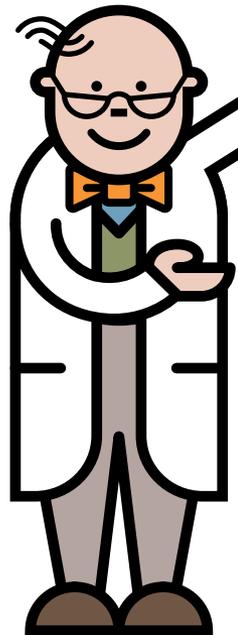
✓ Always ensure you have a good supply of your milk-free formula and it is in date

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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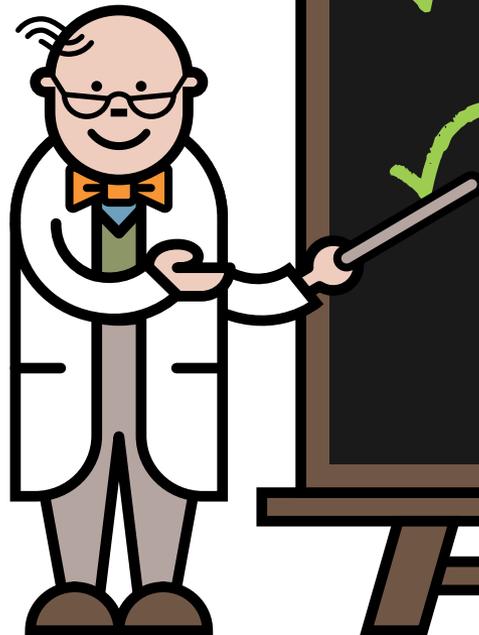


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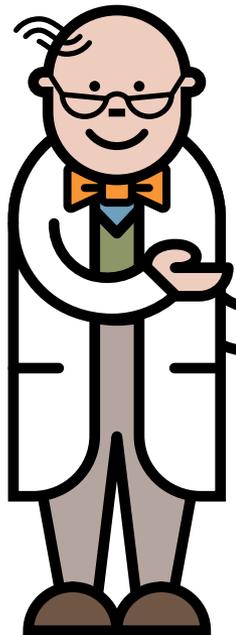
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Who's who

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

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www.bimdg.org.uk

NUTRICIA

www.nutricia.co.uk



www.galactosaemia.org