

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 4, JANUARY 2025

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.

This teaching tool is not designed to replace dietary information that may be given by a dietitian in clinic.

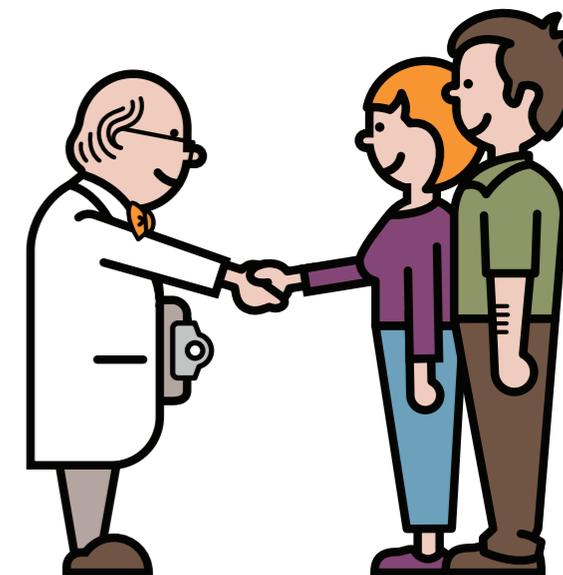
They have been developed by a team of experienced clinical and research metabolic dietitians from the UK who are members of the British Inherited Metabolic Disease Group (BIMDG).

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.

Galactosaemia

Information for families following a new diagnosis



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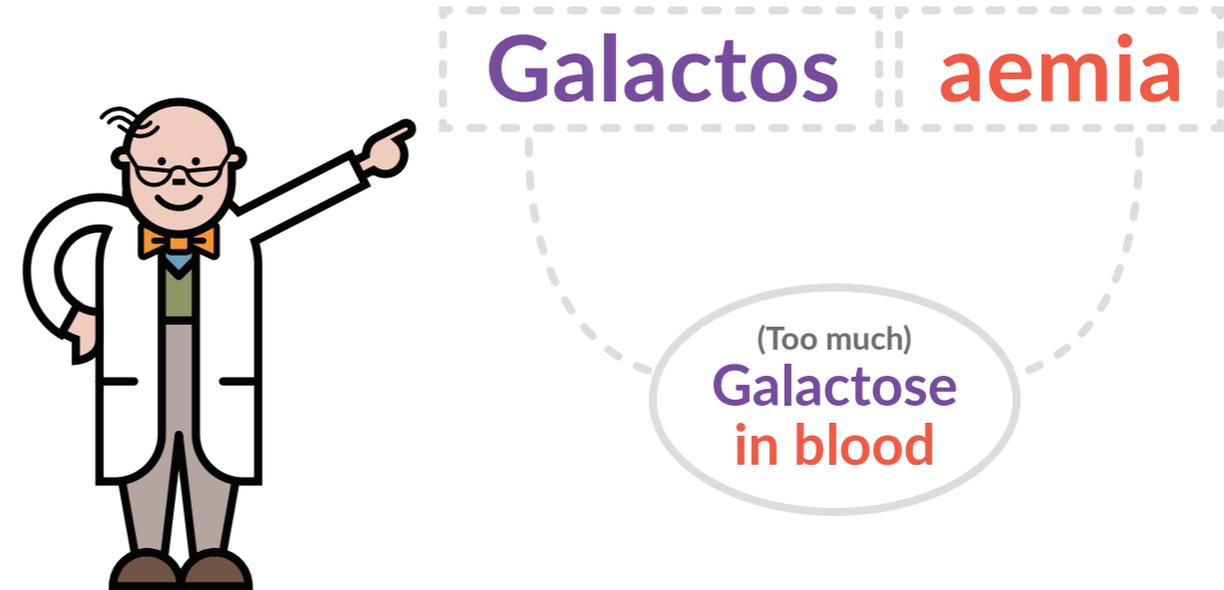


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What is Galactosaemia?

It is an inherited metabolic condition.

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



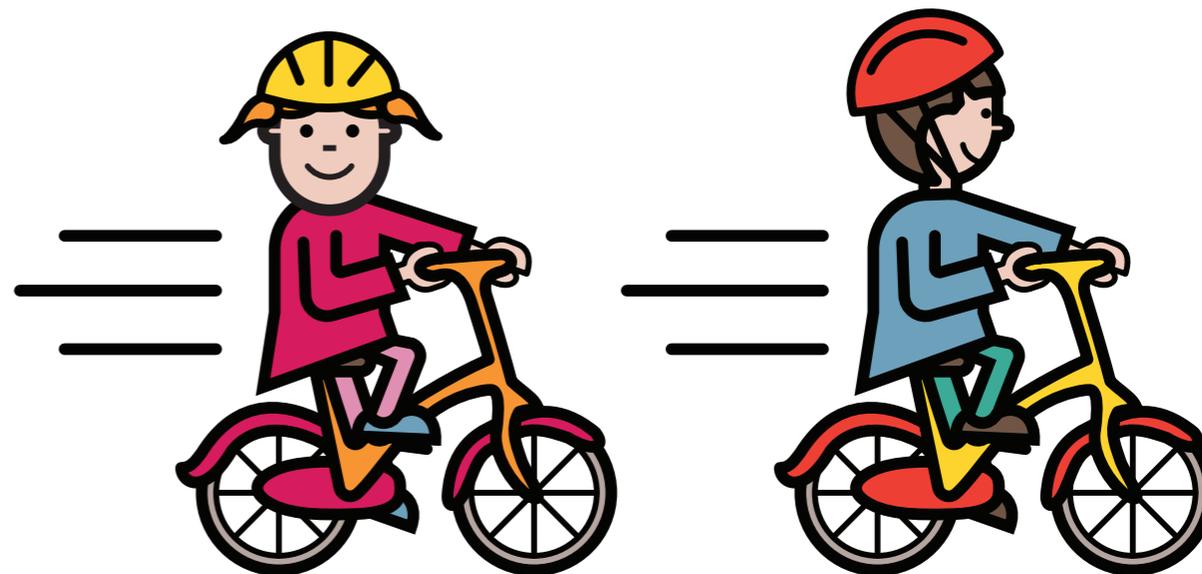
Which foods contain galactose?

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



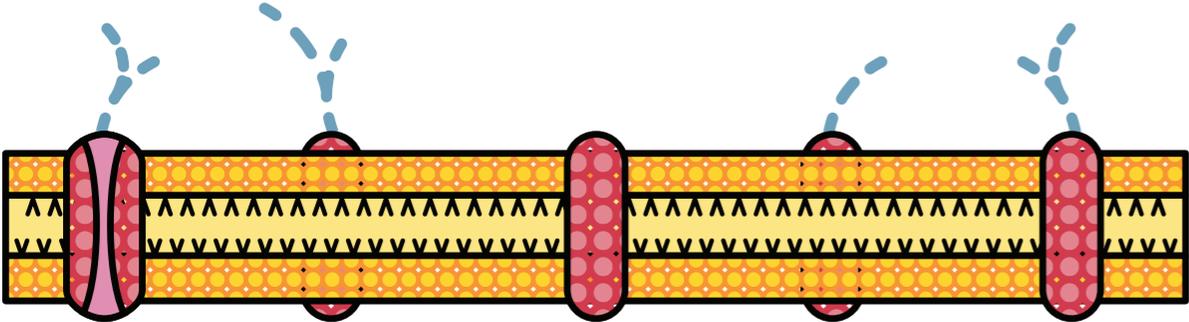
What does galactose do?

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



What else does galactose do?

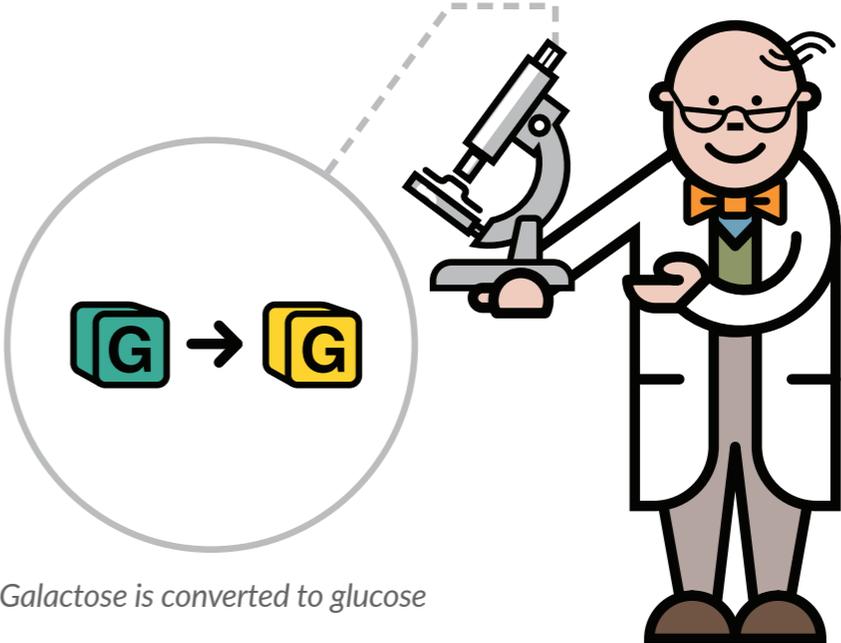
Galactose is a building block of carbohydrate chains.
It joins with **proteins to form glycoproteins** and **fats (lipids) to form glycolipids**.
These are important in cell structure.



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**.

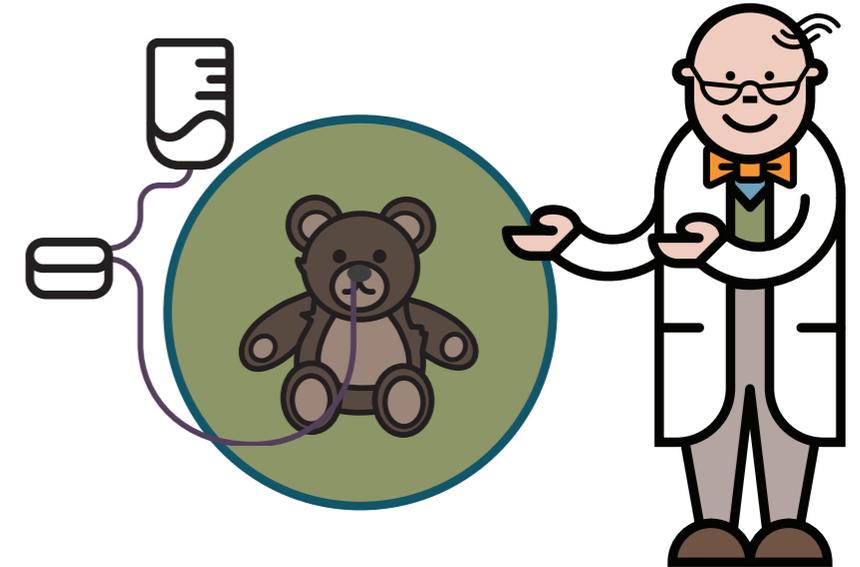
This leads to a build up of galactose and other chemicals leading to symptoms.



What can go wrong in Galactosaemia?

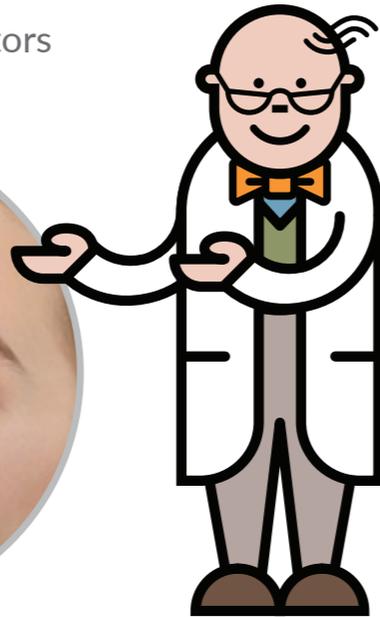
Many babies are very poorly and early symptoms include:

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



What happens with management?

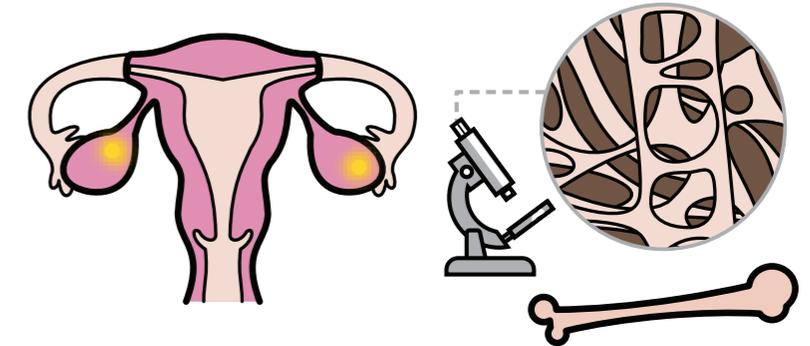
- Once management has been started, babies generally start to improve within a few days
- There should be no long-term liver problems
- Cataracts usually disappear but the doctors will continue to monitor the eyes



...but even with management

some people have:

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



How is Galactosaemia diagnosed?

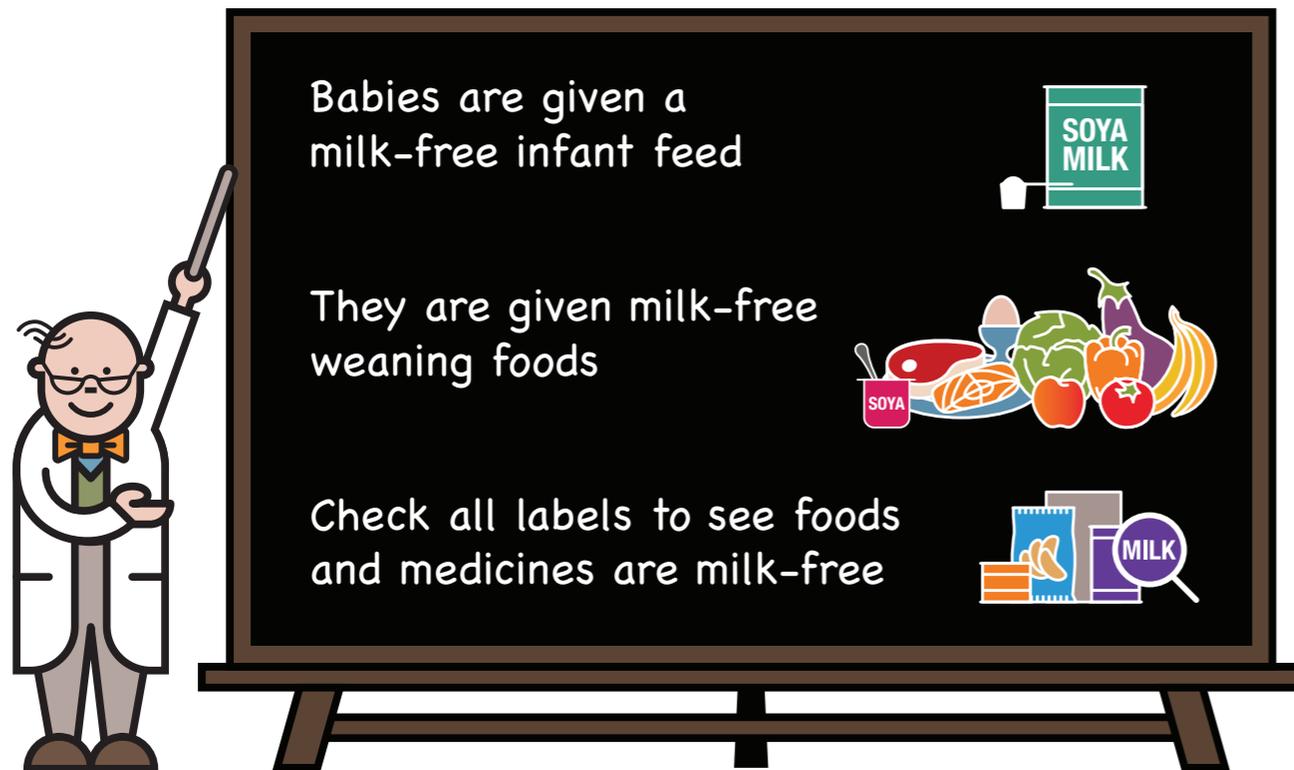
Galactosaemia is usually diagnosed by looking at enzyme levels in the blood and at the body's genes.



How is Galactosaemia managed day to day?

A cartoon illustration of a doctor with glasses and a bow tie, pointing with a stick to a blackboard. The blackboard contains text and icons. The text on the blackboard reads: "Galactose is found in breast milk, infant feed and many foods. It is therefore necessary to:" followed by three bullet points: "Avoid all animal milk (including breast milk)", "Avoid yoghurt, most cheese and milk products", and "Avoid milk in packet and processed foods". To the right of the text are three icons with red diagonal lines through them, indicating avoidance: a breast, a carton of milk with a bowl of yogurt, and a packet of milk powder with a bowl of processed food. A magnifying glass is over the word "MILK" on the packet icon.

How is Galactosaemia managed day to day?



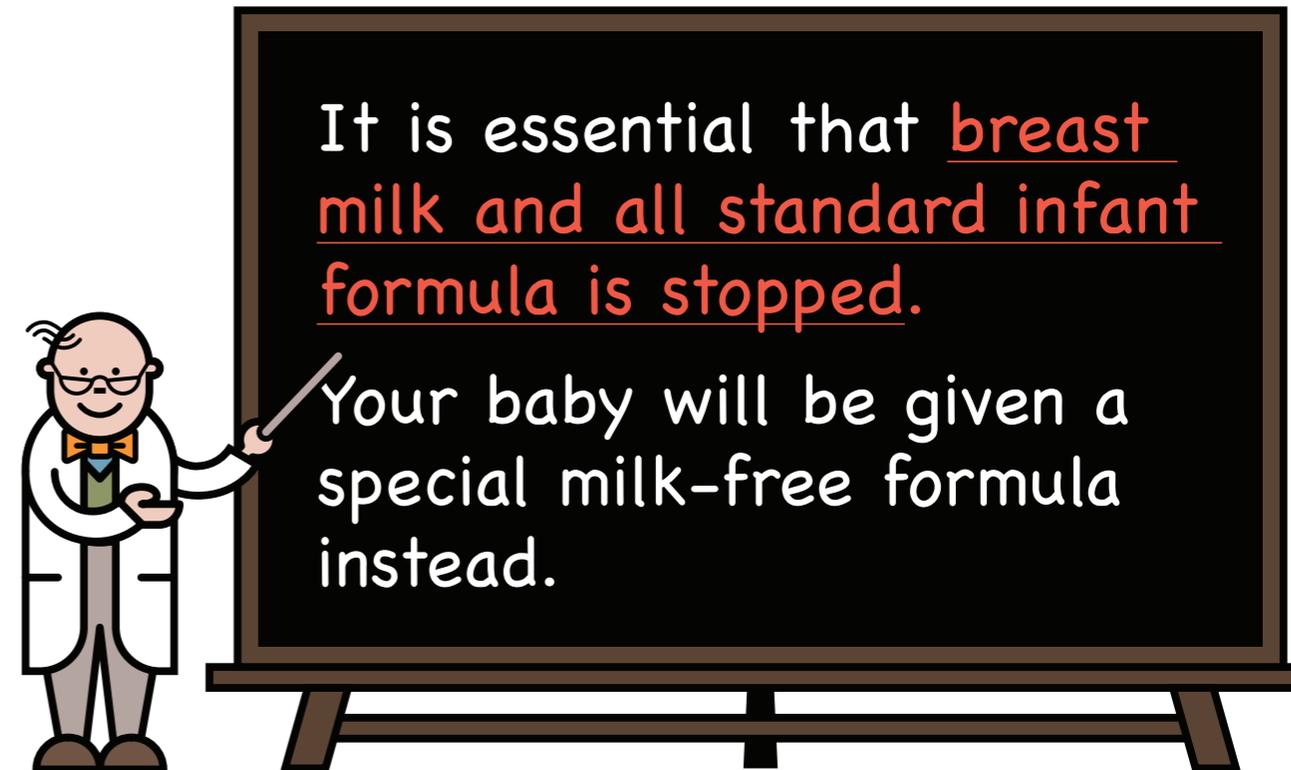
Babies are given a milk-free infant feed

They are given milk-free weaning foods

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



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.

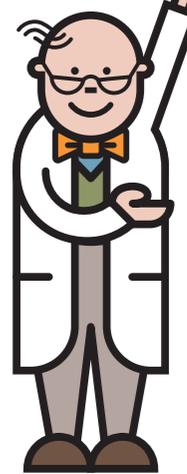
How is Galactosaemia monitored?

- ✓ Blood tests
- ✓ Height and weight
- ✓ Child development and eye checks
- ✓ Regular diet checks by the dietitian

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

Inheritance



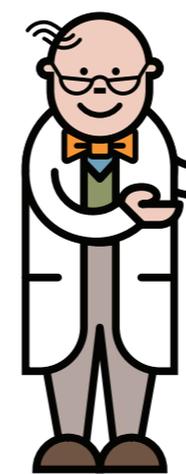
✓ Galactosaemia is an inherited condition. There is nothing that could have been done to prevent your baby from having galactosaemia

✓ 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 – Autosomal-recessive (carriers of Galactosaemia)

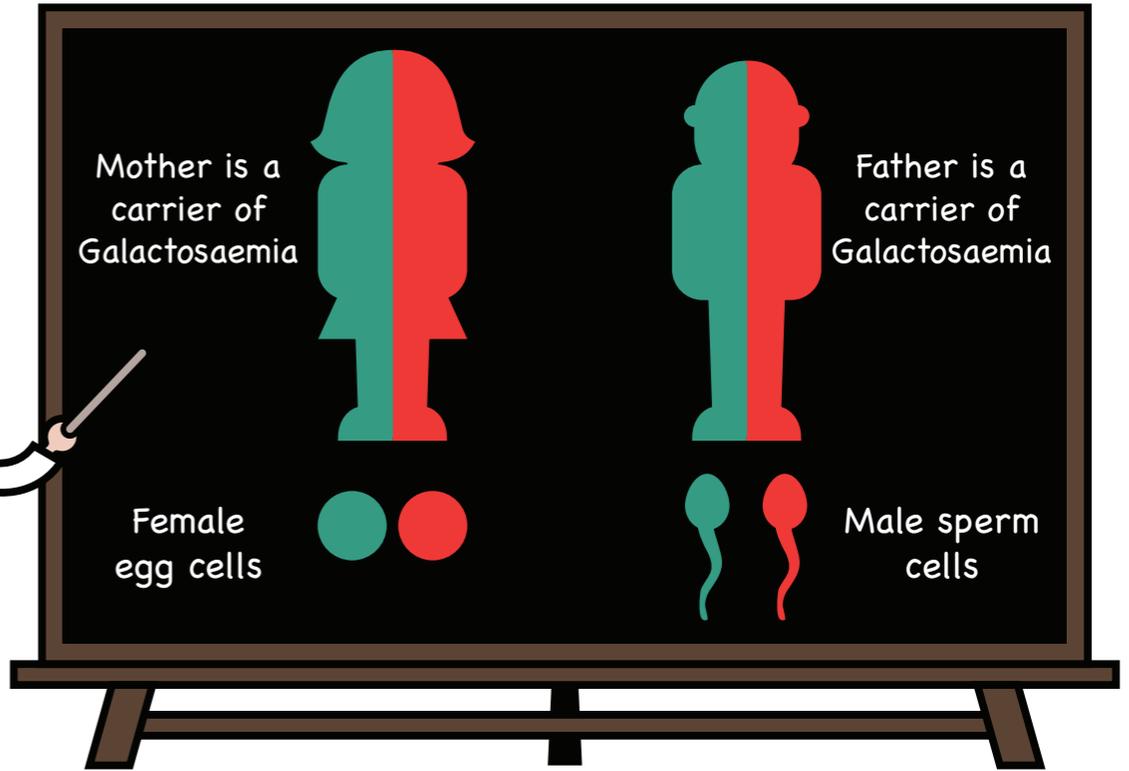


Mother is a carrier of Galactosaemia

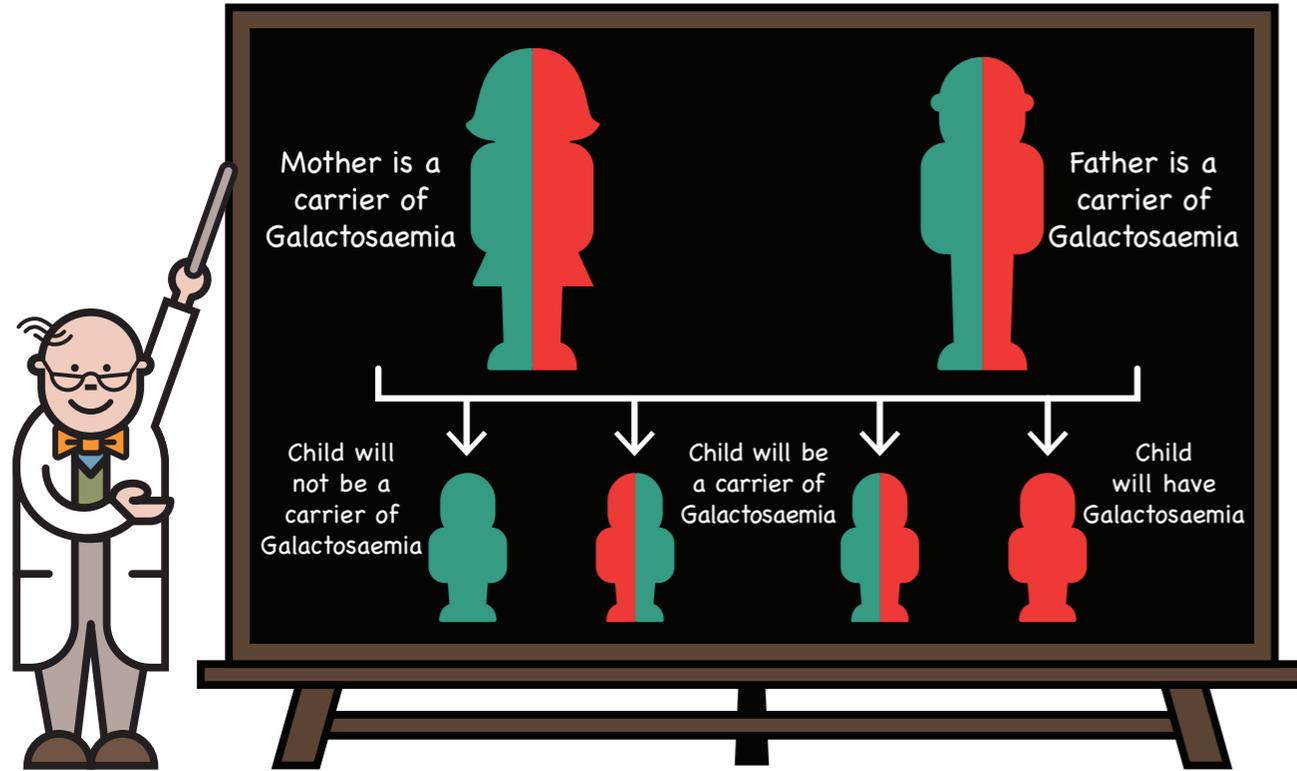
Father is a carrier of Galactosaemia

Female egg cells

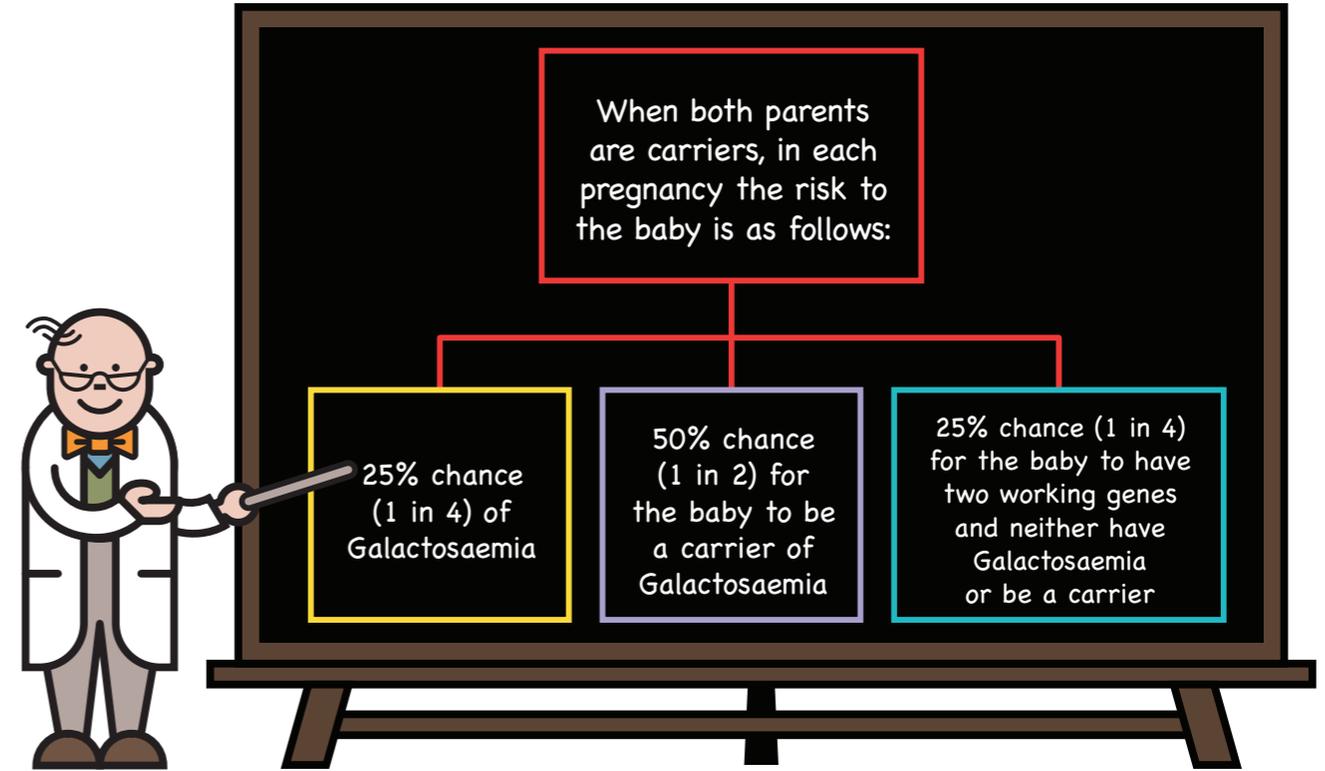
Male sperm cells



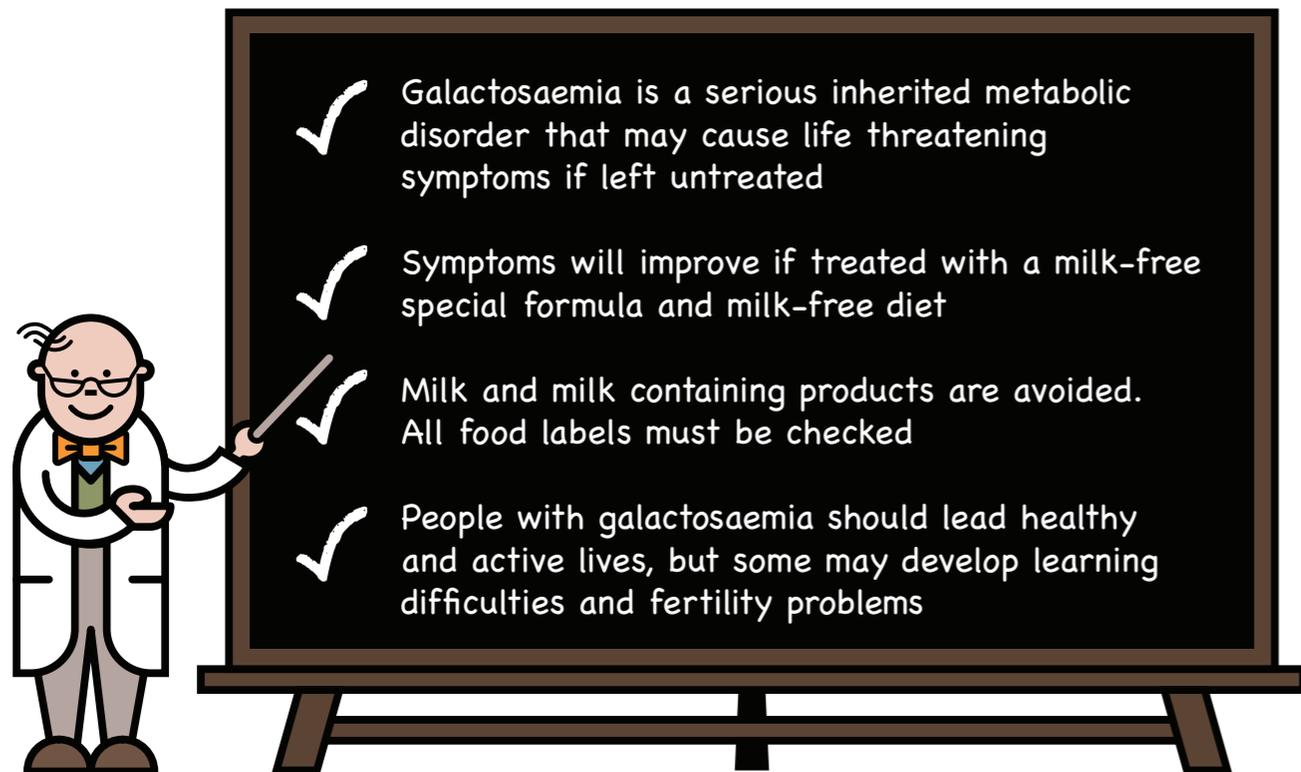
Inheritance — Autosomal recessive – possible combinations



Future pregnancies



Take home messages



Helpful hints



Visit www.nutricia.co.uk/patients-carers/living-with/low-protein-diet.html 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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