TEMPLE

Tools **E**nabling **M**etabolic **P**arents **LE**arning

Galactosaemia

ADAPTED BY THE DIETITIANS GROUP

BIMDG

British Inherited Metabolic Diseases Group

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.

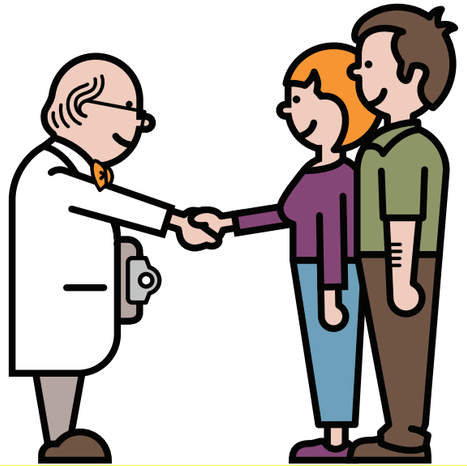
They have been developed by a team of experienced clinical and research metabolic dietitians from the UK who are members of the British Inherited Metabolic Disease Group (BIMDG).

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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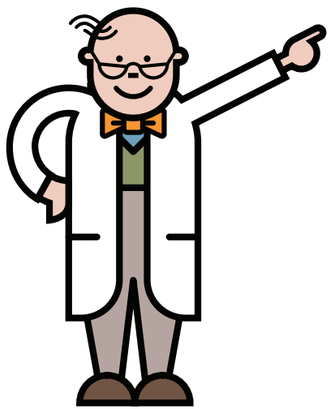
What is Galactosaemia?

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

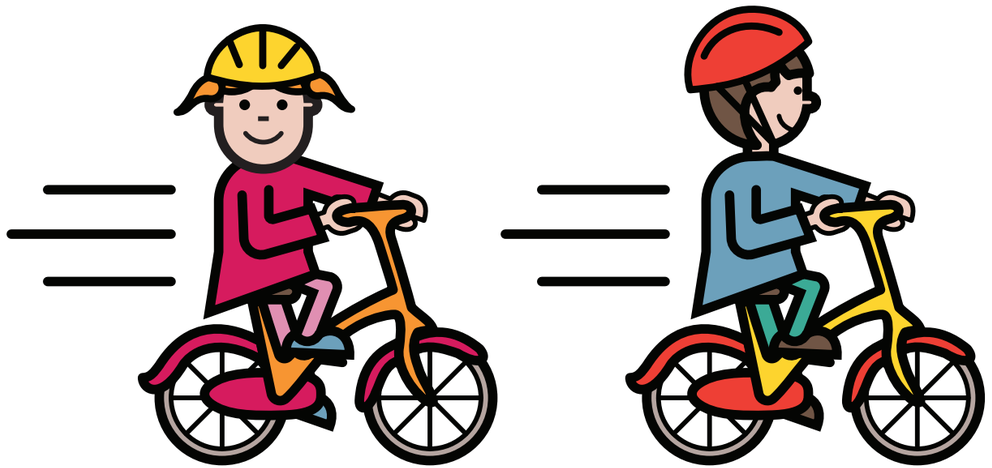
Which foods contain galactose?

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.



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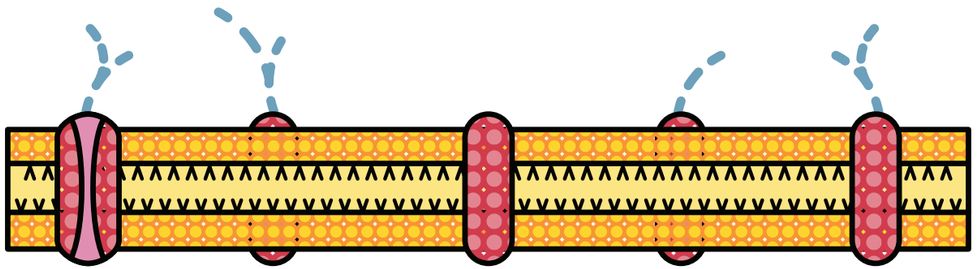
What else does galactose do?

Galactose is a building block of carbohydrate chains.

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

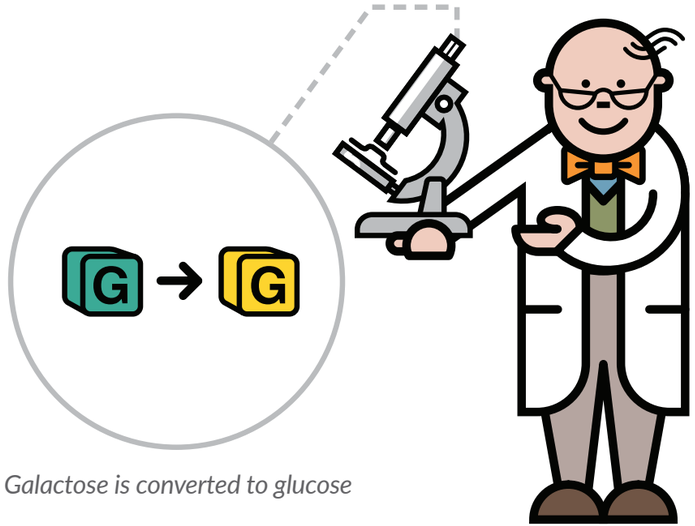
These are important in cell structure.



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.

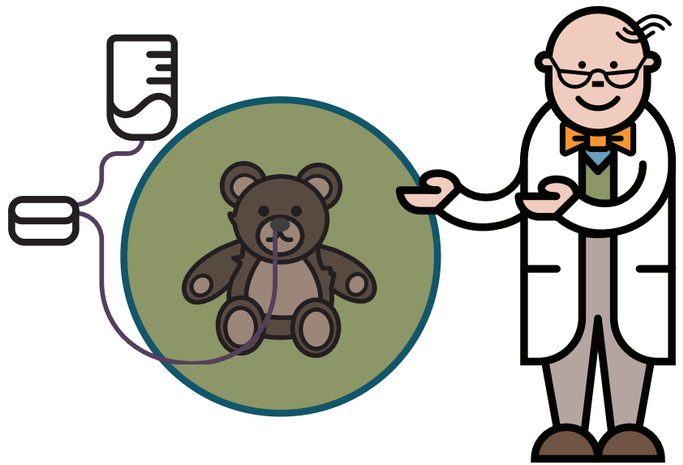
The enzyme is called **galactose-1-phosphate uridyl transferase**.

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

What can go wrong in Galactosaemia?

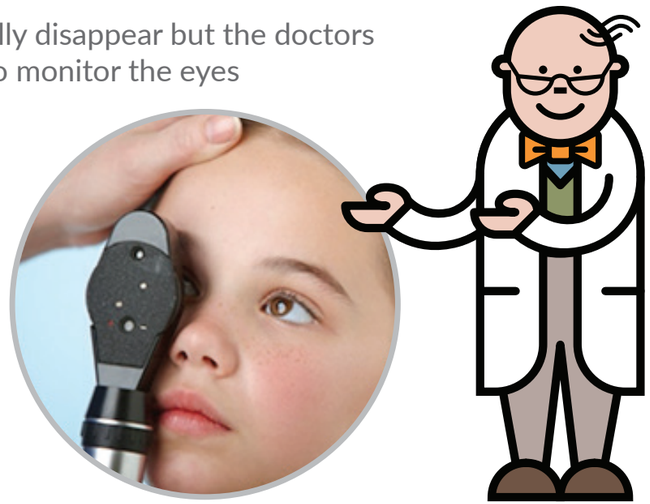
Many babies are very poorly and early symptoms include:

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



What happens with management?

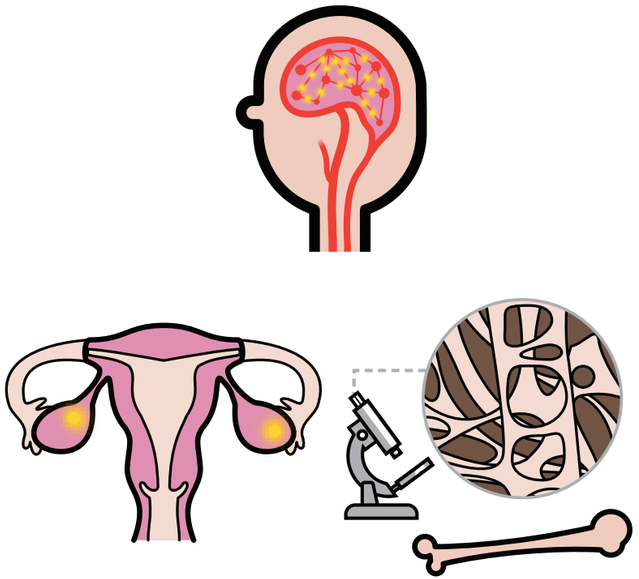
- Once management has been started, babies generally start to improve within a few days
- There should be no long-term liver problems
- Cataracts usually disappear but the doctors will continue to monitor the eyes



...but even with management

some people have:

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



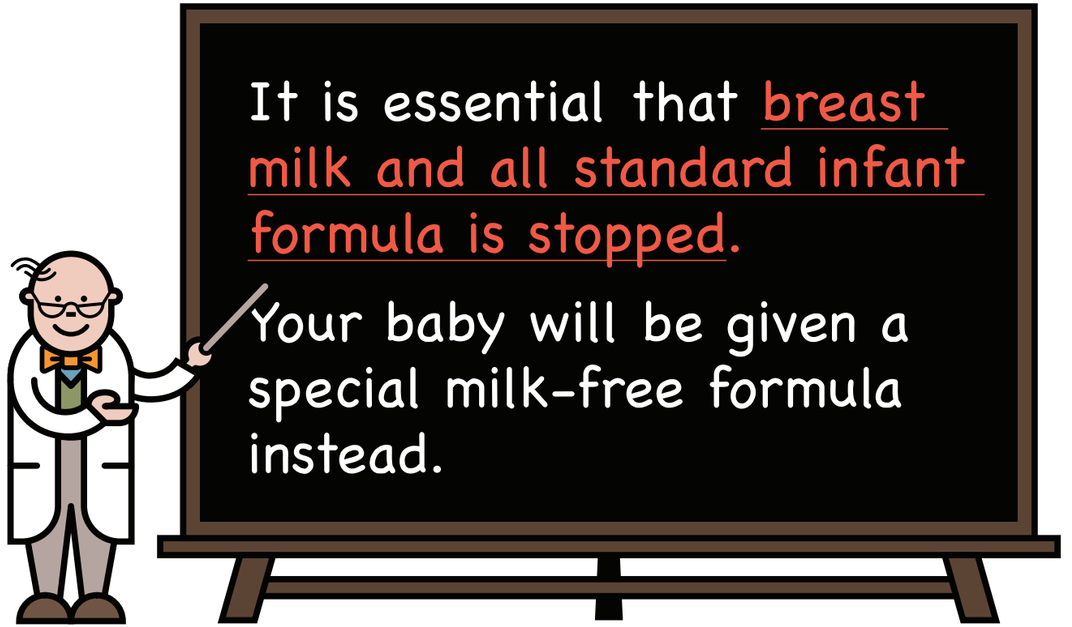
How is Galactosaemia diagnosed?

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

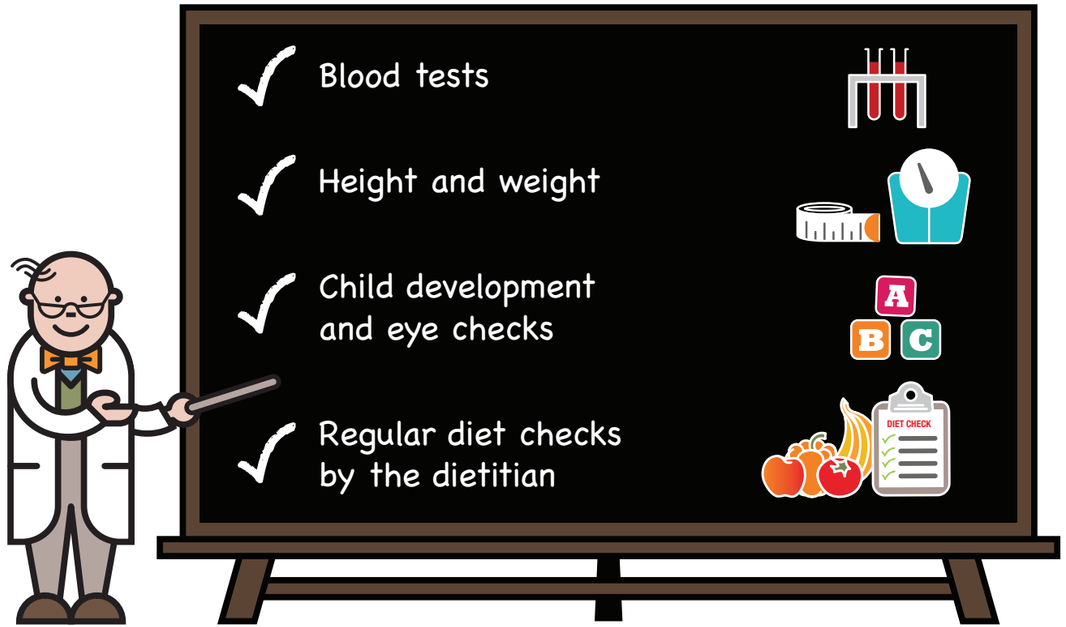
How is Galactosaemia managed day to day?

How is Galactosaemia managed day to day?

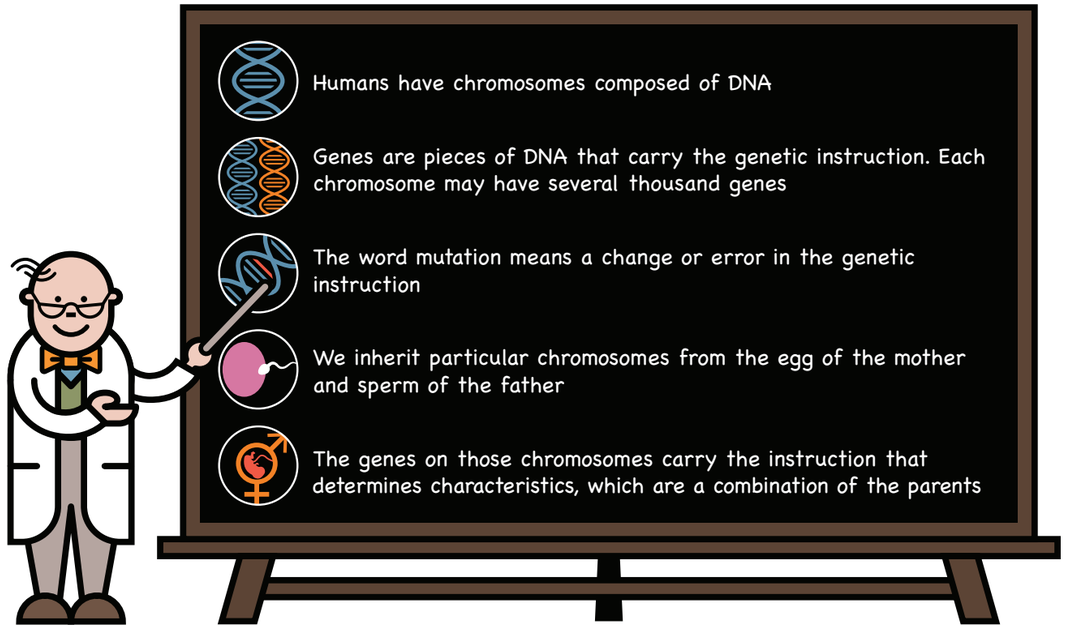
Key message



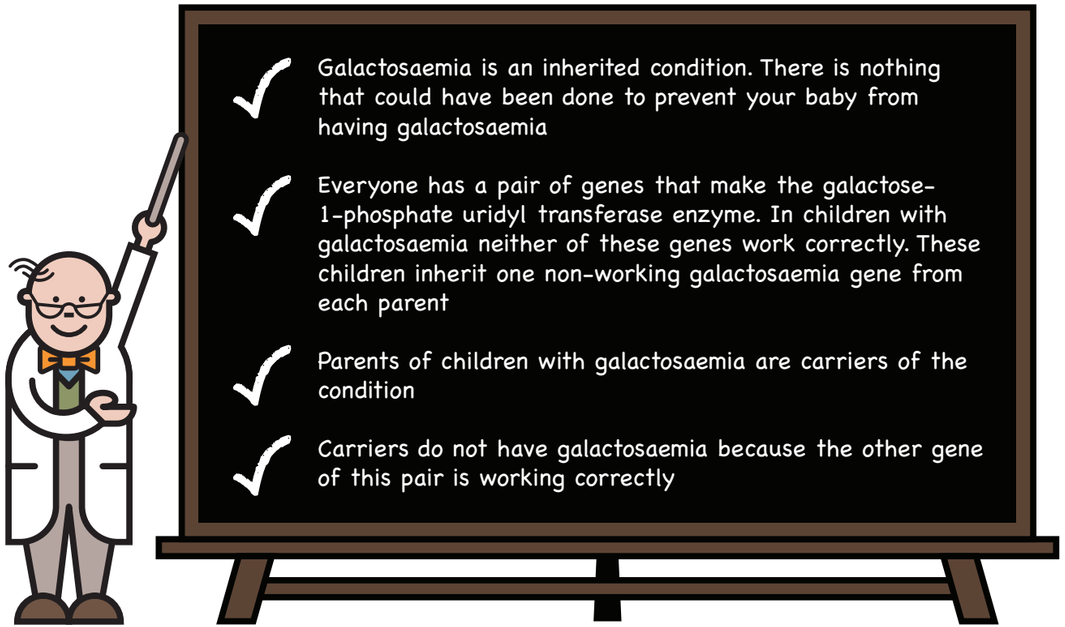
How is Galactosaemia monitored?



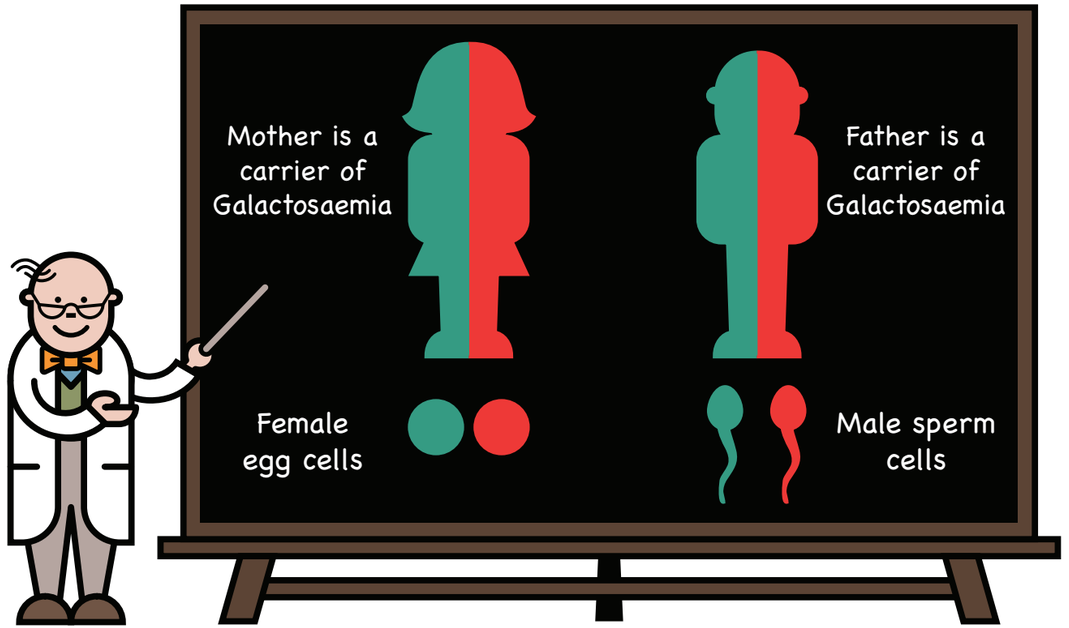
Chromosomes, genes, mutations



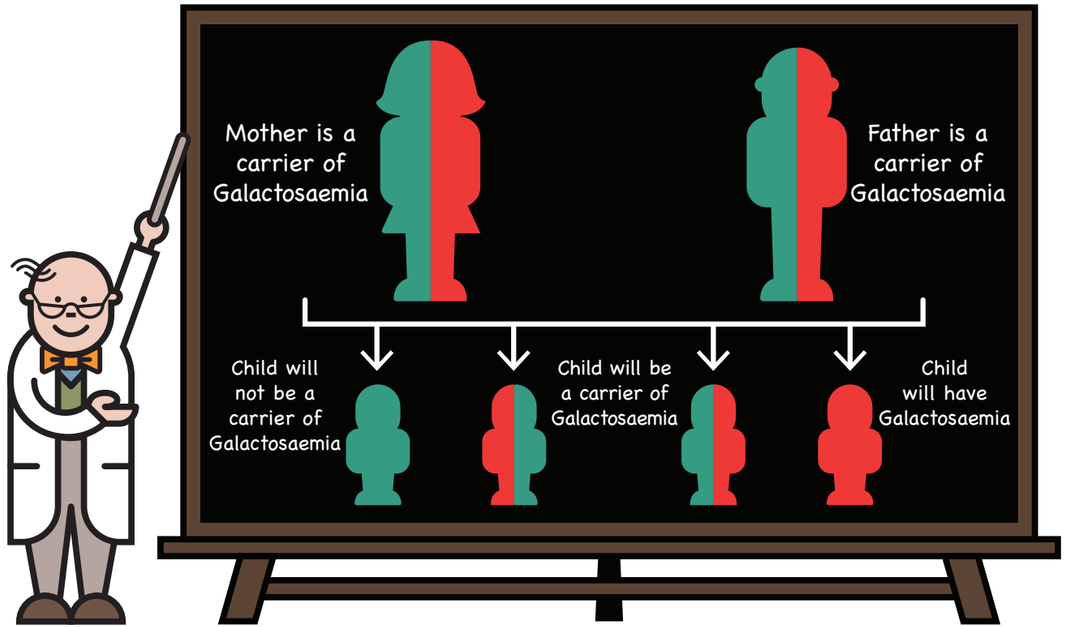
Inheritance



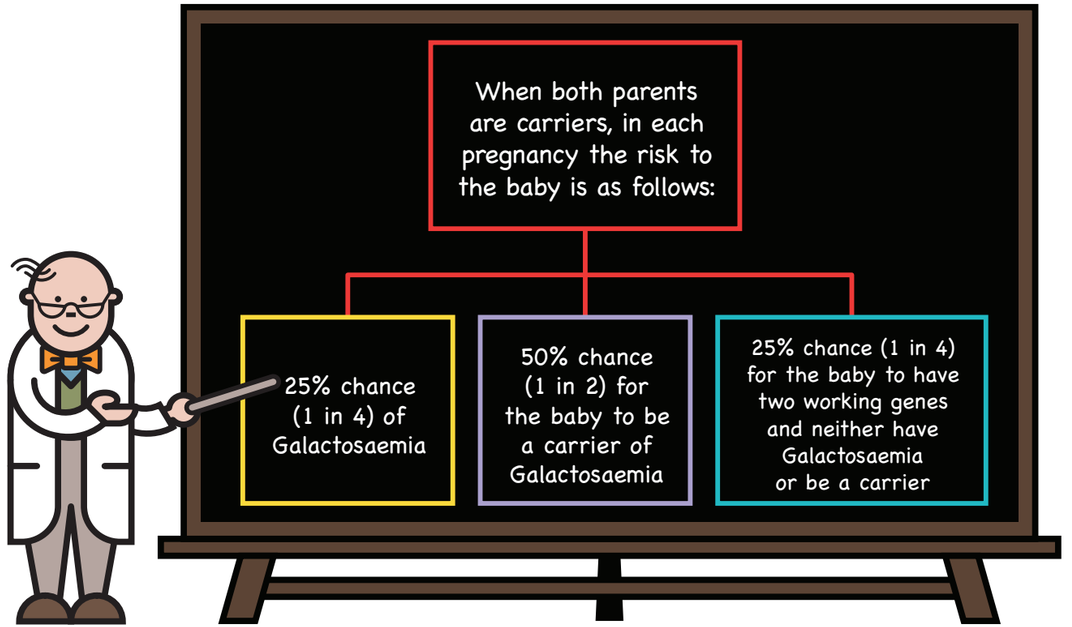
Inheritance – Autosomal-recessive (carriers of Galactosaemia)



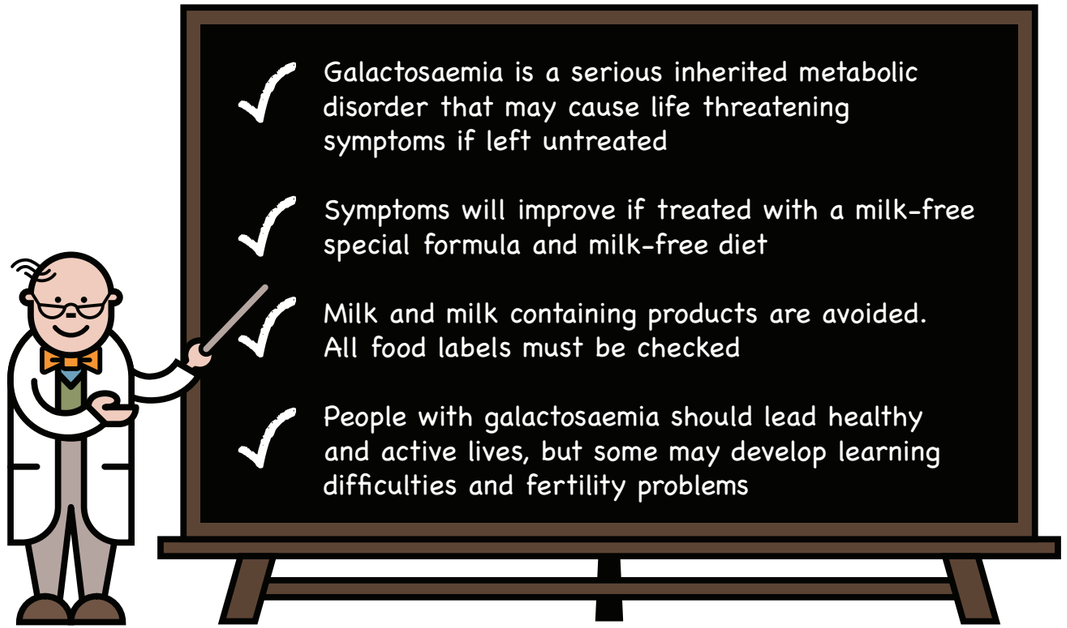
Inheritance – Autosomal recessive – possible combinations



Future pregnancies



Take home messages



Helpful hints

Notes

Notes

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.

Low Protein Connect

@LowProConnect
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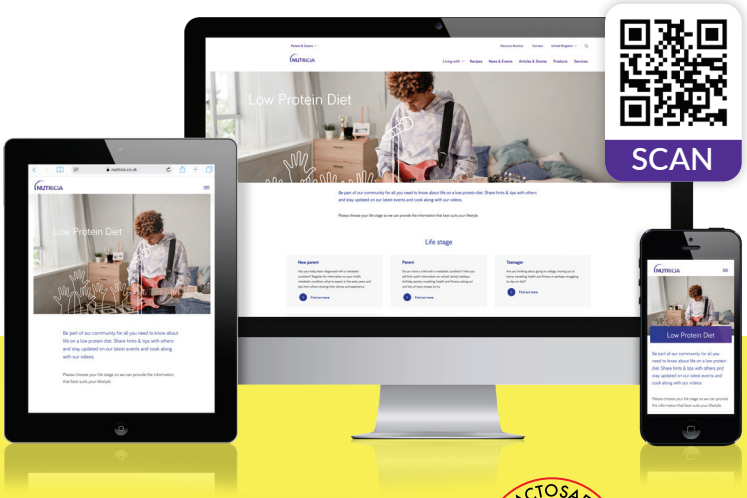

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www.galactosaemia.org