

# TEMPLE



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

ADAPTED BY THE DIETITIANS GROUP

**BIMDG**

British Inherited Metabolic Diseases Group



BASED ON THE ORIGINAL TEMPLE WRITTEN BY  
BURGARD AND WENDEL

VERSION 1, DECEMBER 2022

GSD Ia

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).

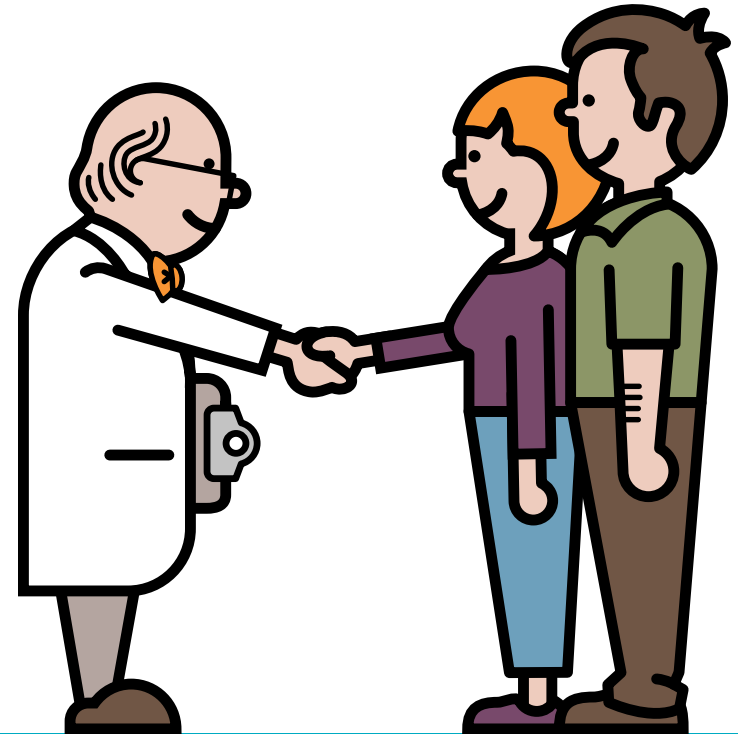
The team are Rachel Skeath, Karen van Wyk, Pat Portnoi and Anita MacDonald. The group is facilitated by Heidi Chan from Nutricia.

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.**

# GSD Ia

Information for families following  
a new diagnosis



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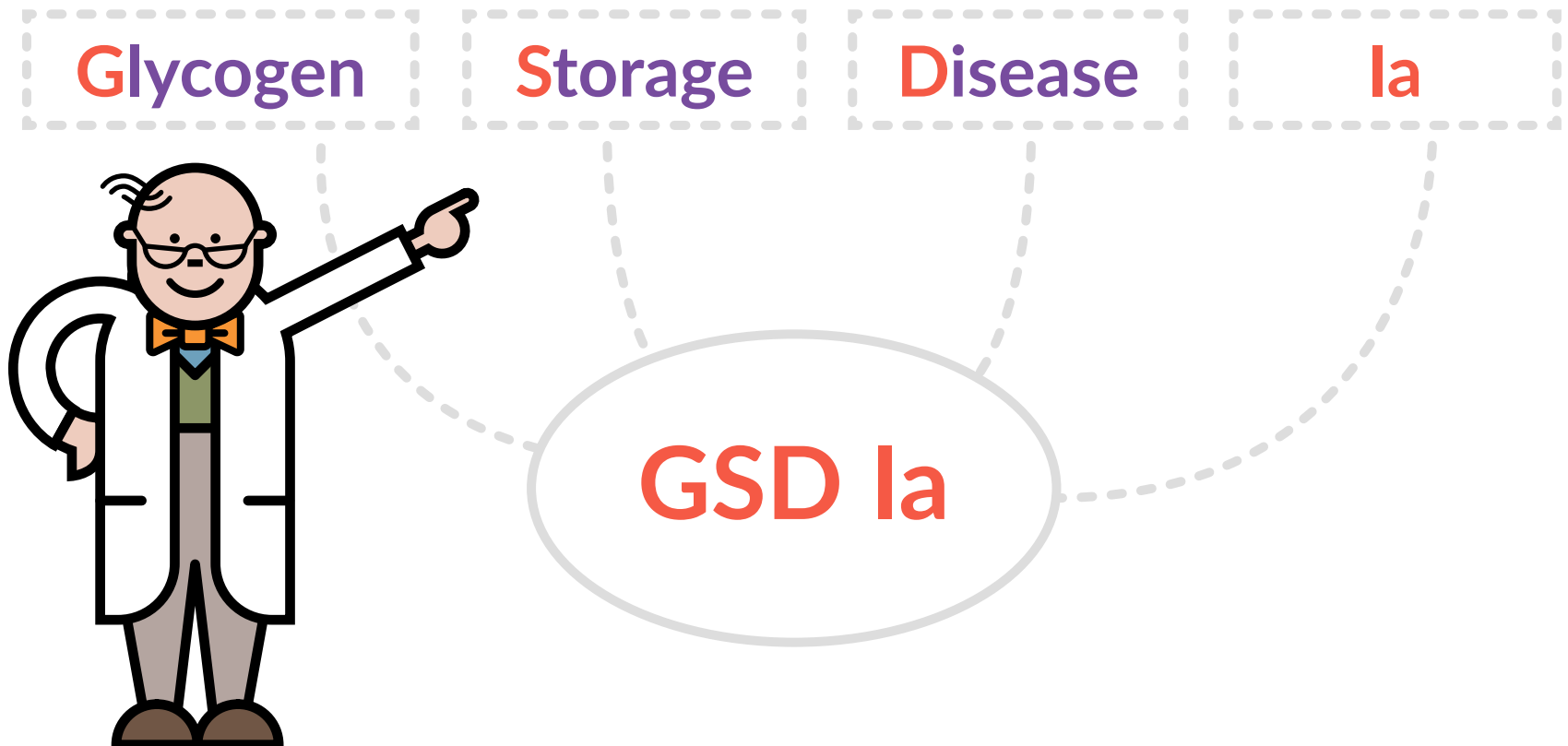


Tools Enabling Metabolic Parents LEarning

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# What is GSD Ia?

GSD Ia stands for Glycogen Storage Disease Ia  
It is an inherited metabolic condition



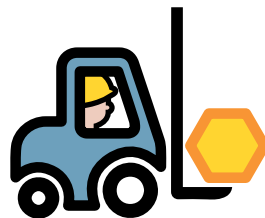
# Glucose and glycogen for energy

We eat food to supply our body with energy.

An important supply of energy is carbohydrate which is broken down into a sugar called glucose.

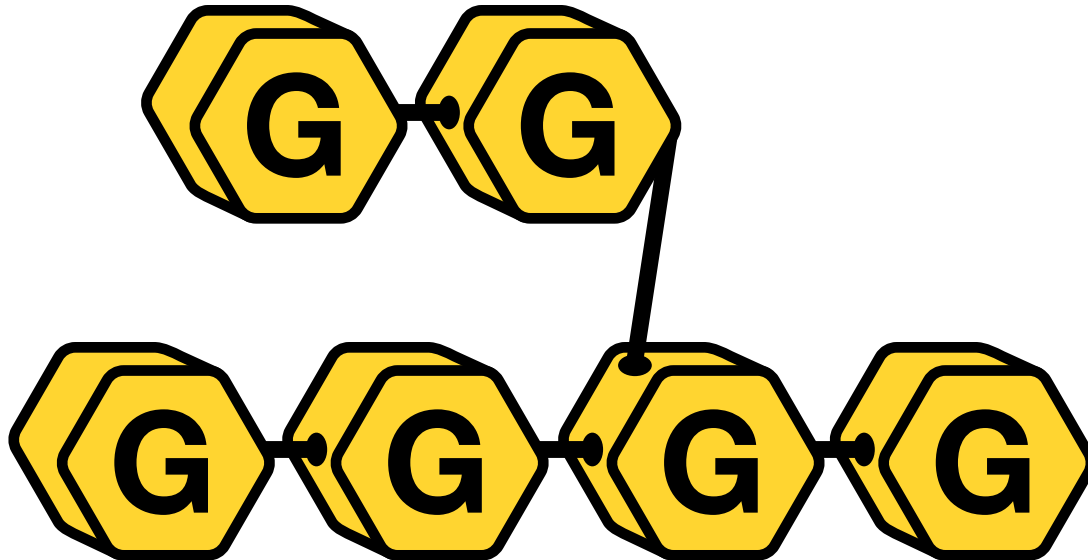
Some of this glucose from food goes into our blood stream and will give a *readily available* supply of energy.

We also put some glucose into storage as an energy reserve. **This is converted into glycogen in the liver and muscle.**



# What is glycogen?

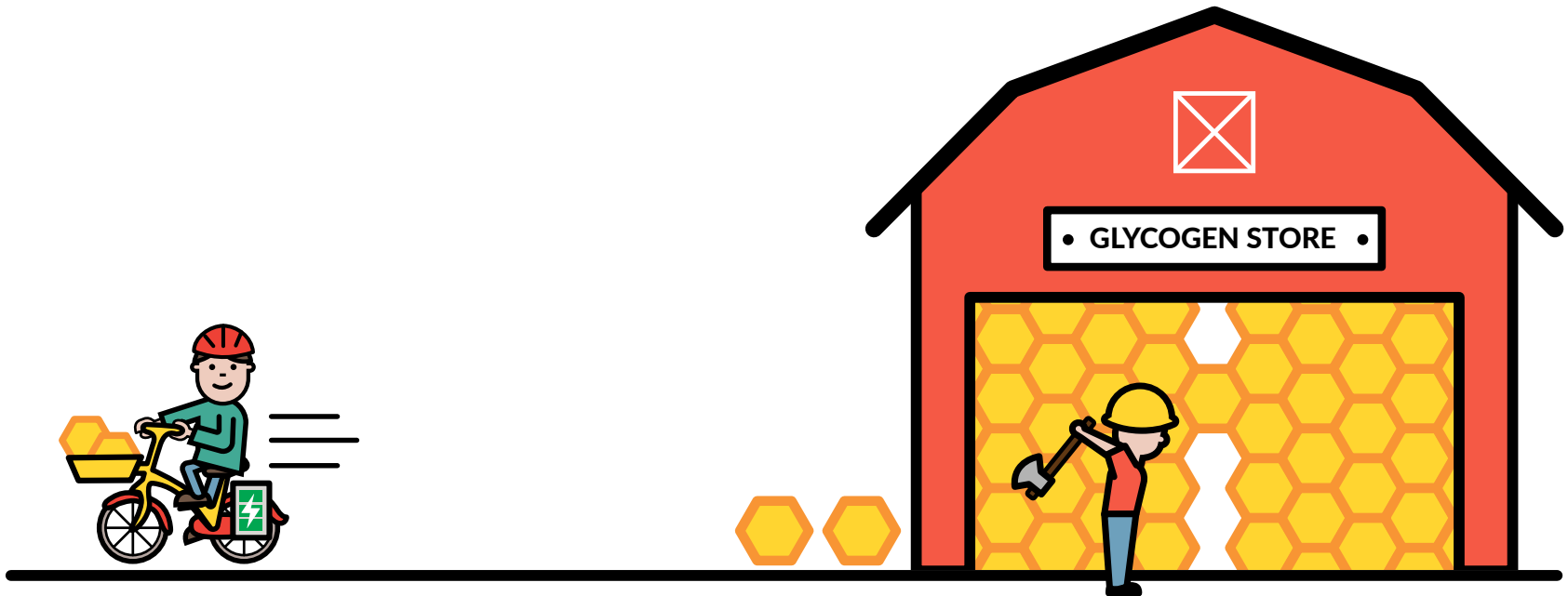
Glycogen is made up of units of glucose joined together.



# Glucose and glycogen for energy

When energy is needed, the body uses the *readily available* glucose as its energy supply.

Once glucose is used up, the body then breaks down the stored glycogen to make more glucose.



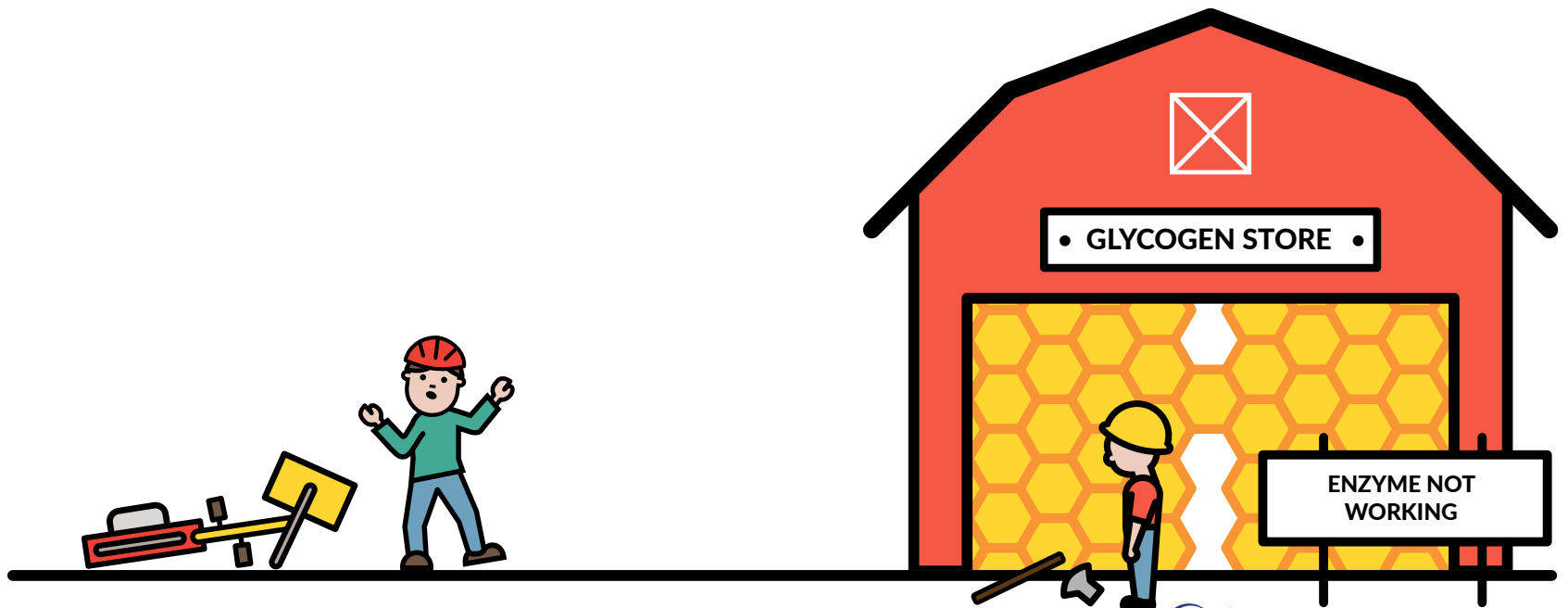
# What happens in GSD Ia?

In GSD Ia there are problems breaking down the glycogen stores to release glucose.

The body lacks an enzyme called glucose-6-phosphatase that helps break down glycogen into glucose.

This stops the glycogen being used for energy.

It causes a shortage of energy supply and a low blood glucose.





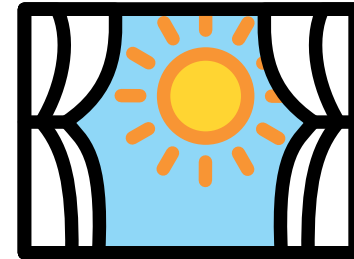
# What can go wrong with GSD 1a?

Some babies become ill in the first few months of life.

Symptoms include:

- low blood glucose
- floppiness
- seizures
- large liver
- high levels of lactate and fats in the blood
- poor growth

**The effects of low blood glucose is very harmful if unmanaged.**



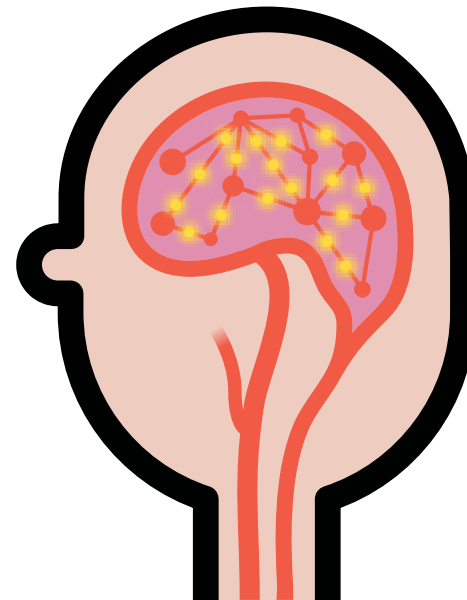
# What can go wrong with GSD 1a?

If there is a low blood glucose and this is not corrected, it can lead to coma and brain damage. It may be life threatening.

**However, please remember, this can be prevented with timely and correct management.**

Other issues include:

- Thin bones
- Some children may not be as tall as expected



# How is GSD Ia diagnosed?

GSD Ia is suspected because of the pattern of chemicals found in the blood.

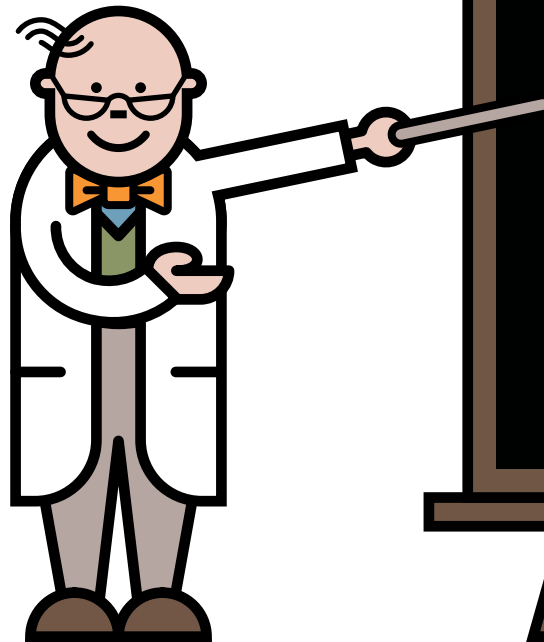
The diagnosis is confirmed by looking for mutations in the GSD Ia gene.



# How is GSD Ia managed?

It is managed by **avoiding** long periods without feeding.

The length of time that someone can go without feeds or eating is known as the **safe fasting time**.



## SAFE FASTING TIMES

- The safe fasting time varies for each person with GSD Ia. Your metabolic team will advise
- It is important that regular feeds are given during the day and at night
- Scheduled feeds or meals should not be missed or delayed

# How is GSD Ia managed day to day?

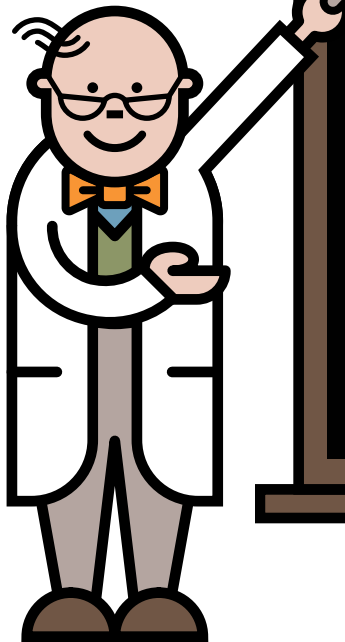
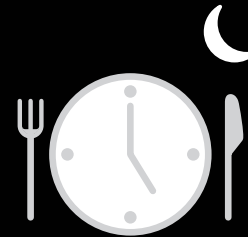
Regular meals and snacks based on carbohydrate



Healthy balanced diet

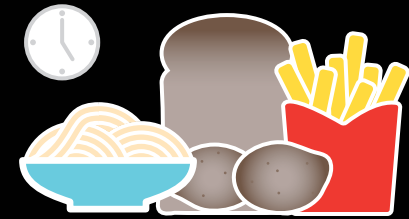


Overnight feeds or regular feeds/  
drinks overnight



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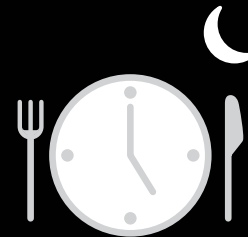
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# How is GSD Ia managed day to day?

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Overnight feeds or regular feeds/  
drinks overnight



# Which foods contain carbohydrate?

Many foods contain carbohydrate.

Good sources of carbohydrate include breast milk, bread, rice, potato, pasta and cereals.





# Uncooked cornstarch

- Uncooked cornstarch is a source of carbohydrate that is slowly broken down into glucose
- Sometimes a modified cornstarch is used instead
- These are commonly used in GSD 1a to extend safe fasting times
- Your dietitian will talk to you about this

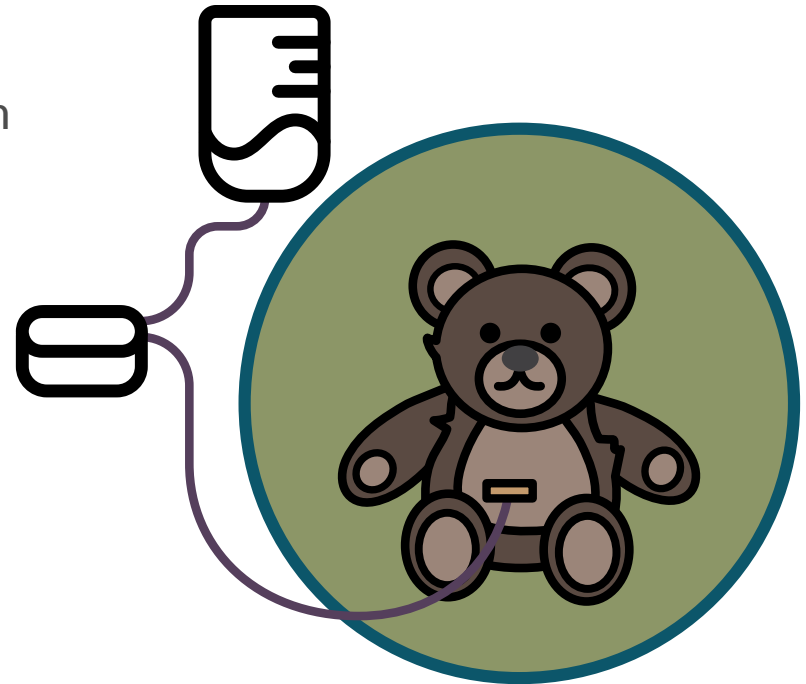


# Overnight feeding

Babies and children with GSD Ia cannot safely fast overnight without their blood glucose dropping too low.

They will need to have regular drinks or feeds during the night.

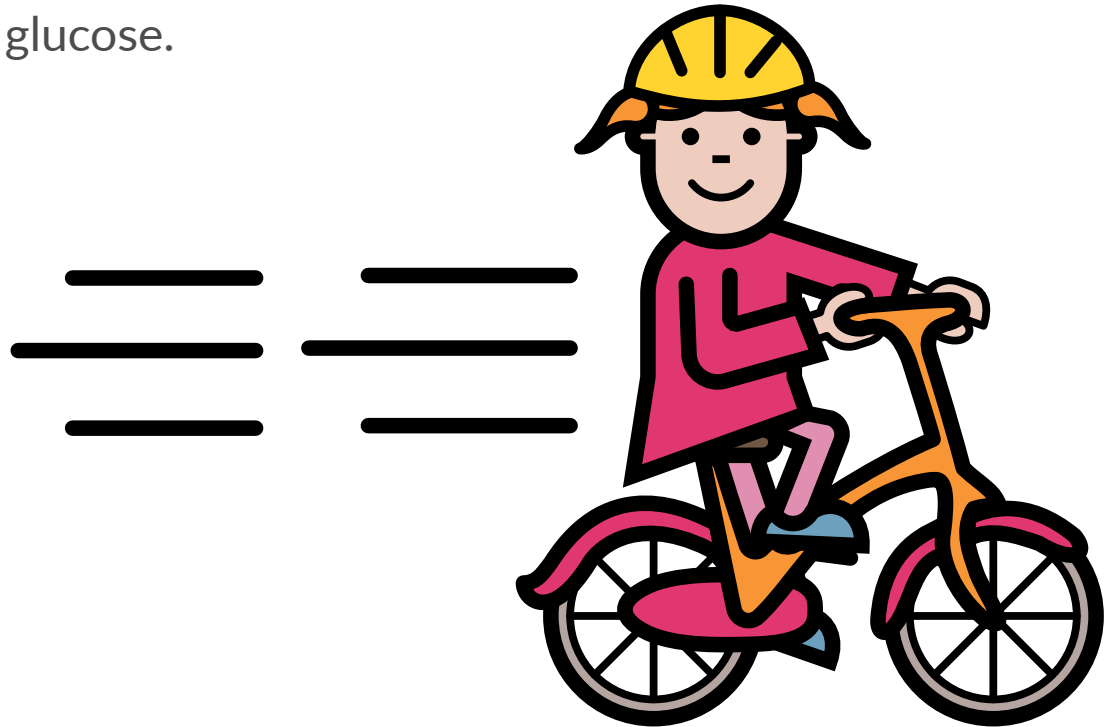
Many babies and children are fed through a tube overnight so that they can sleep without having to wake up.



# GSD 1a and exercise

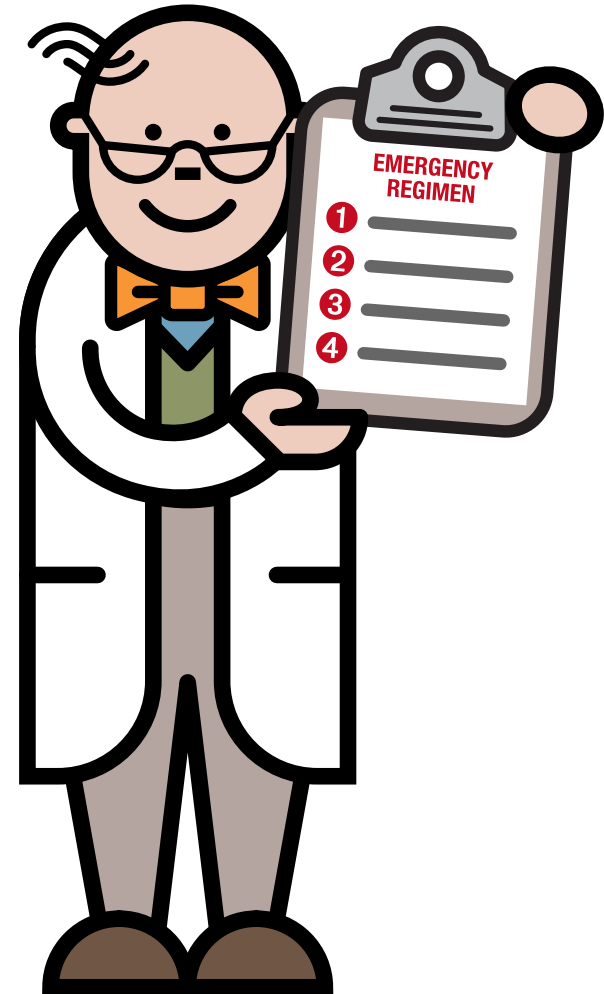
Exercise and activity helps keep children with GSD 1a healthy.

It might be advised that a snack or drink is taken before exercise. This will supply an extra source of glucose.



# How is GSD 1a managed during illness?

- During illness your child may not be able to follow their usual feeding plan
- You will be given an emergency regimen to use during this time
- This provides energy and prevents low blood glucose



# How is GSD Ia managed during illness?

Start the emergency regimen.  
This is made up of glucose polymer



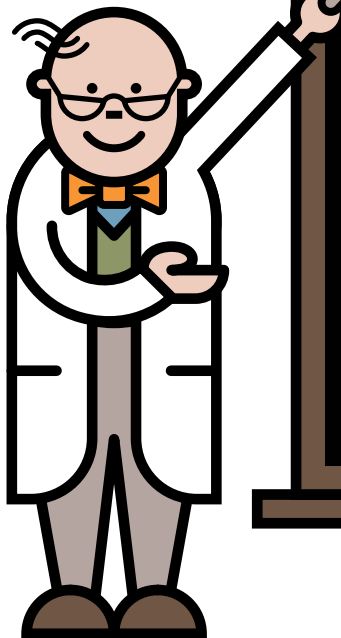
It must be given day and night



Phone your metabolic team  
for help if your child is unwell



**Do not use sugar-free drinks during illness**



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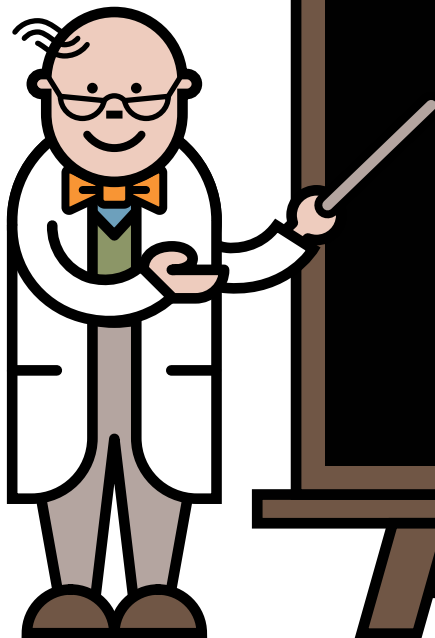
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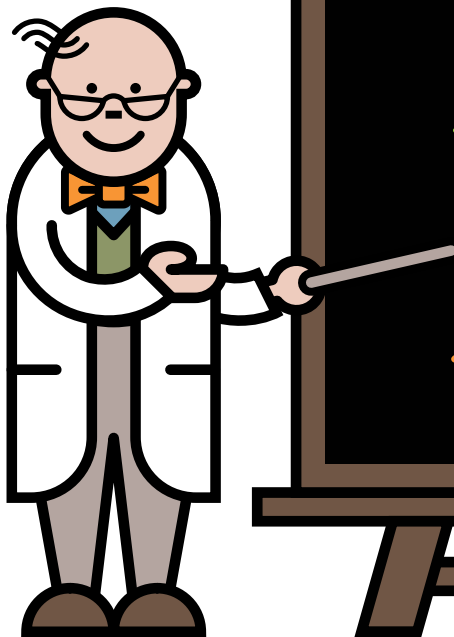
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# Checklist for illness

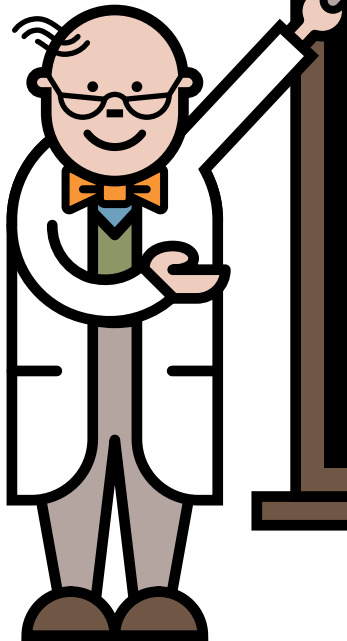
Always take full amounts of emergency feeds as prescribed



If your child vomits or refuses the emergency feeds and/or you are worried, go immediately to the hospital



Phone your metabolic team





# Checklist for illness

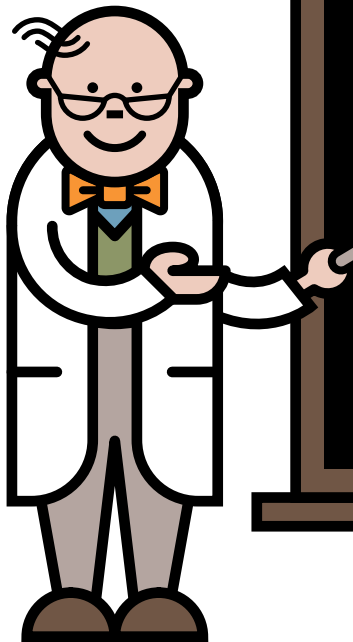
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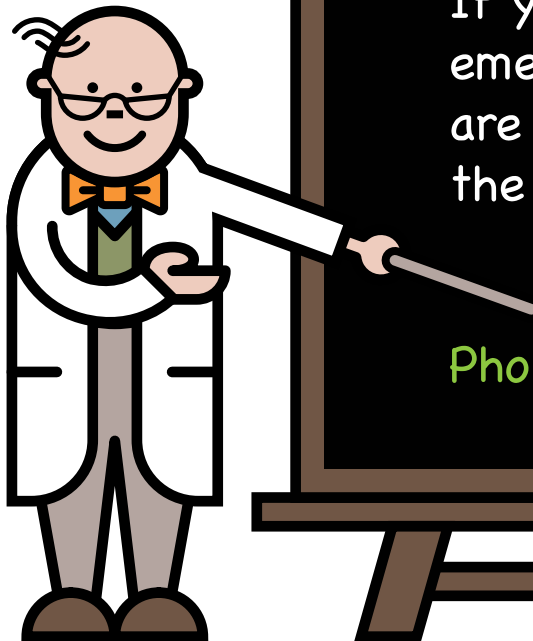
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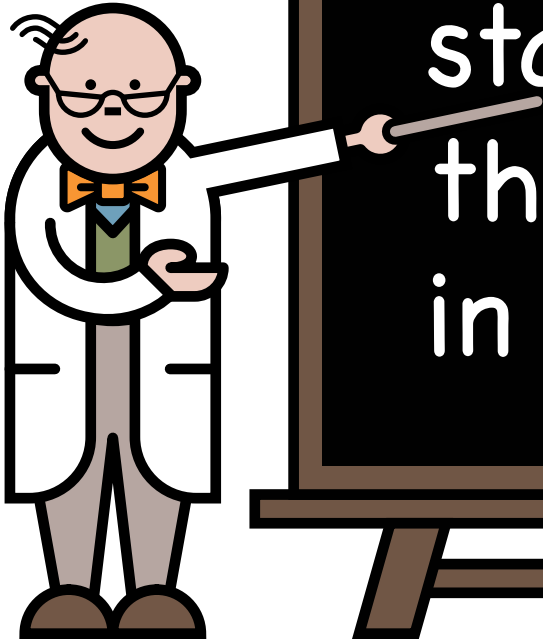


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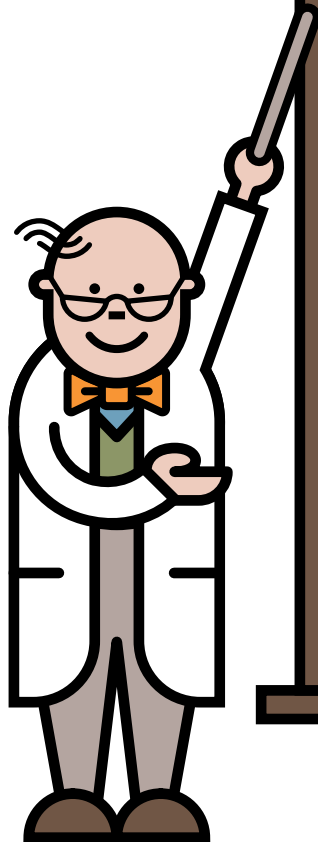


# Key message

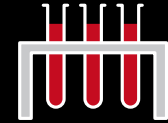
It is imperative that emergency feeds are started **promptly** and there are **no delays** in management.



# How is GSD Ia monitored?



Blood tests to check to check metabolic control



Safe fasting times are tested by monitoring blood glucose levels and other chemicals in the blood



Height and weight



Medical and development checks

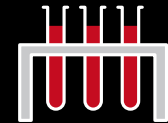


Emergency feeds are adjusted with age and weight



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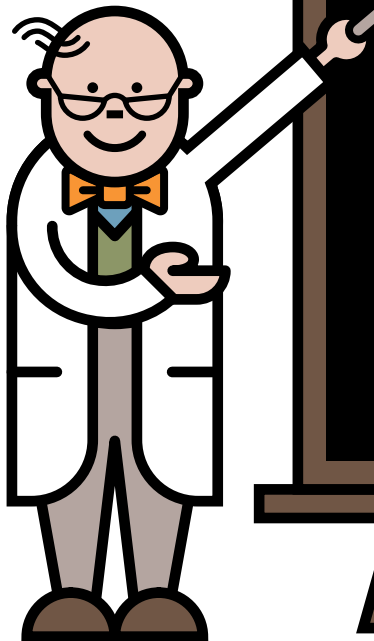
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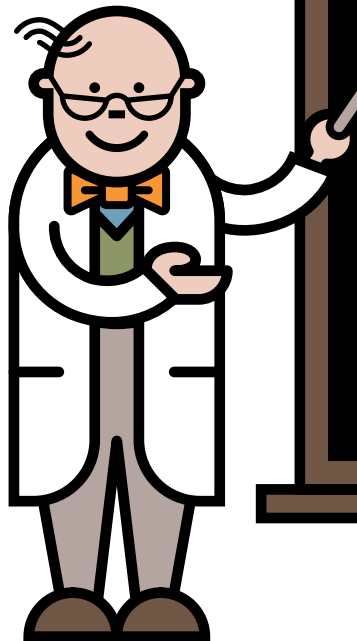
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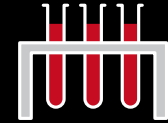
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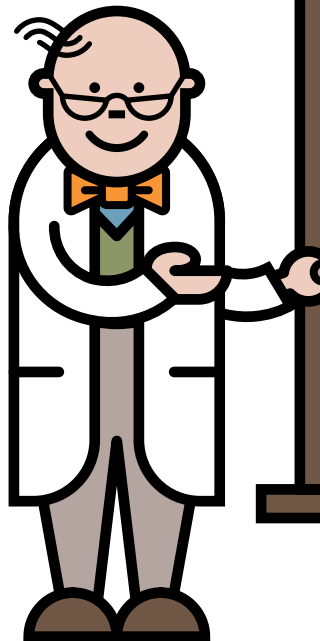
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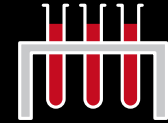
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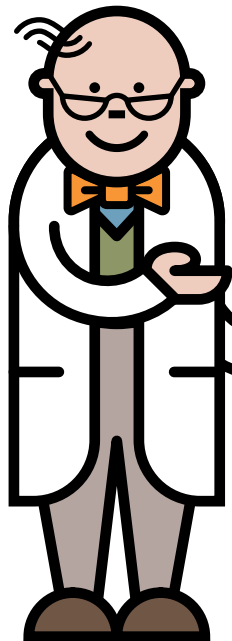
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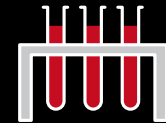
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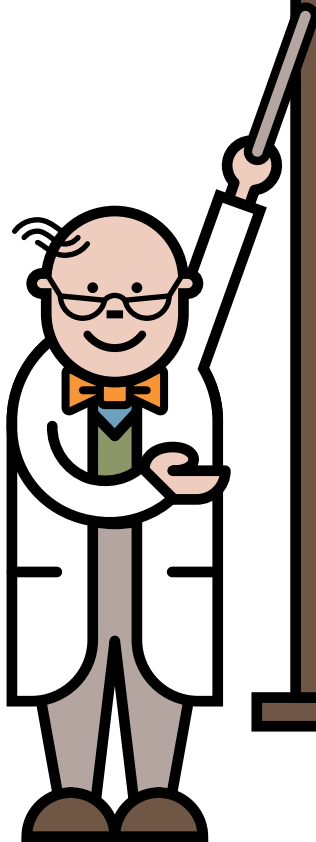


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# Chromosomes, genes, mutations



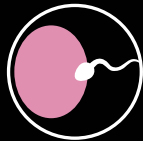
Humans have chromosomes composed of DNA



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



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

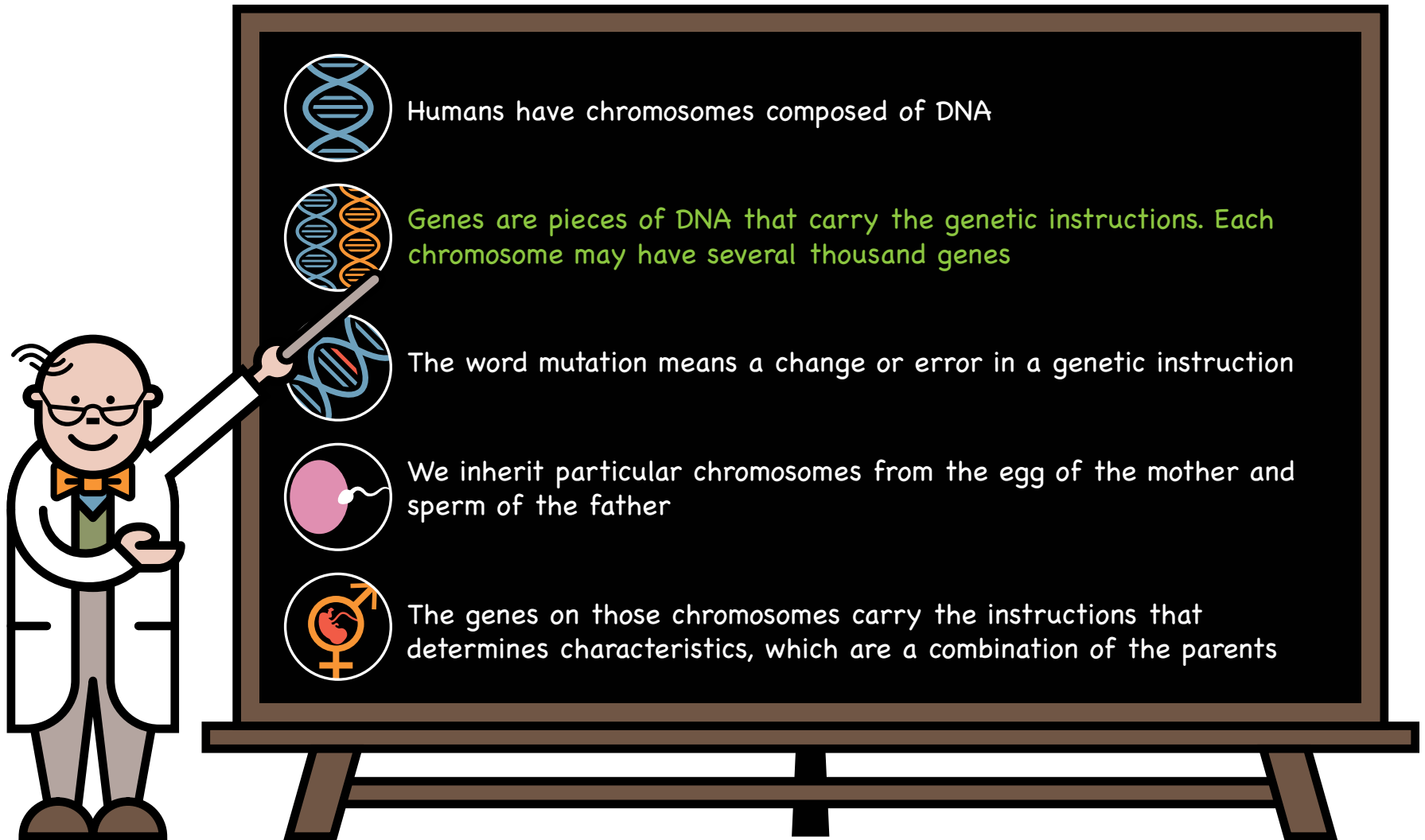



We inherit particular chromosomes from the egg of the mother and sperm of the father





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


# Chromosomes, genes, mutations




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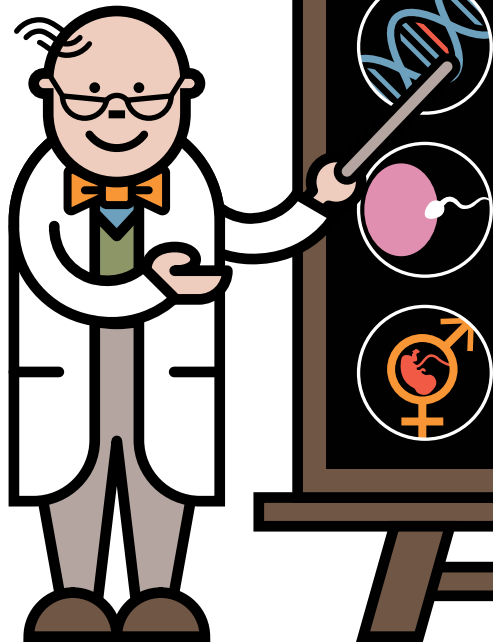
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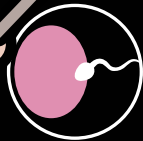
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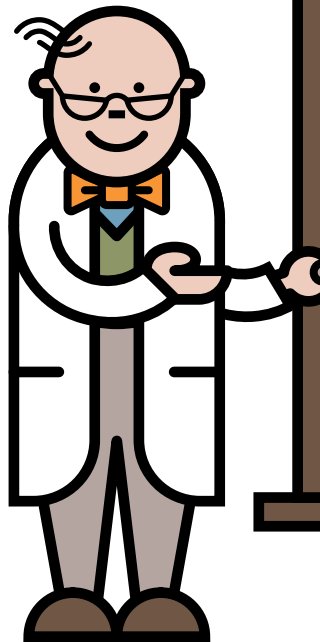


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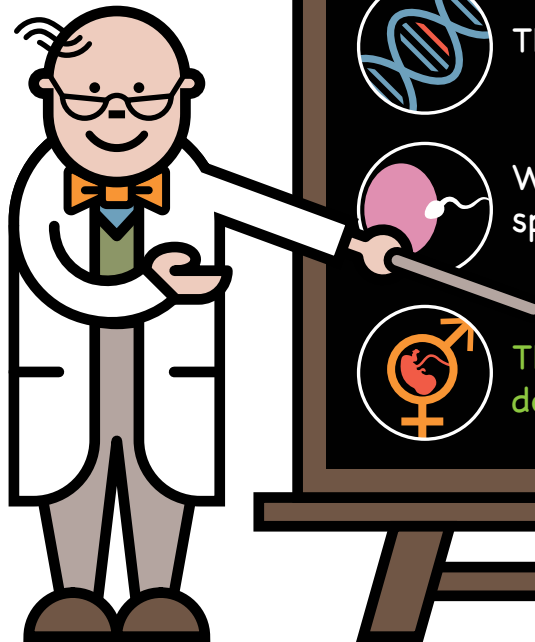


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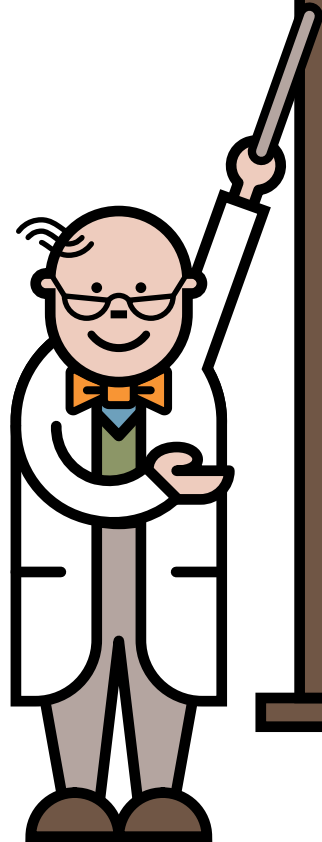


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# Inheritance



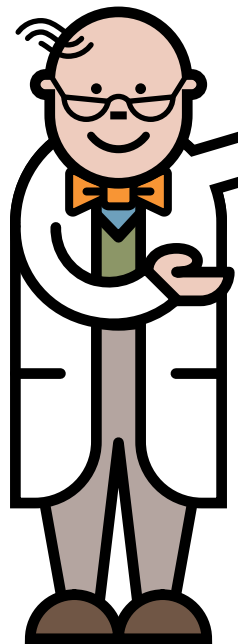
GSD 1a is an inherited condition. There is nothing that could have been done to prevent your baby from having GSD 1a

Everyone has a pair of genes that makes glucose-6-phosphatase enzyme. In children with GSD 1a, neither of these genes work correctly. These children inherit one non-working glucose-6-phosphatase gene from each parent

Parents of children with GSD 1a are carriers of the condition

Carriers do not have GSD 1a because the other gene of this pair is working correctly

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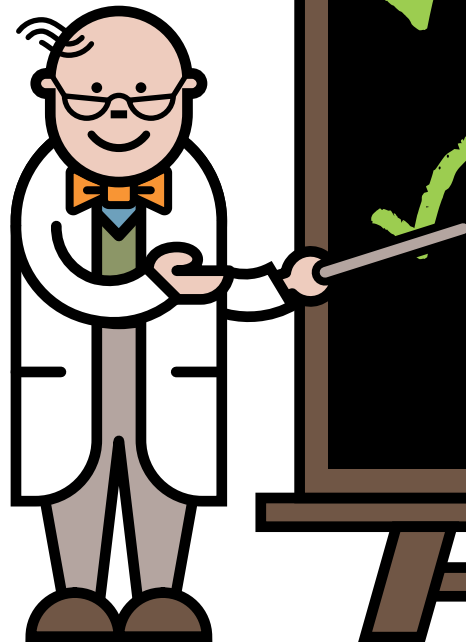


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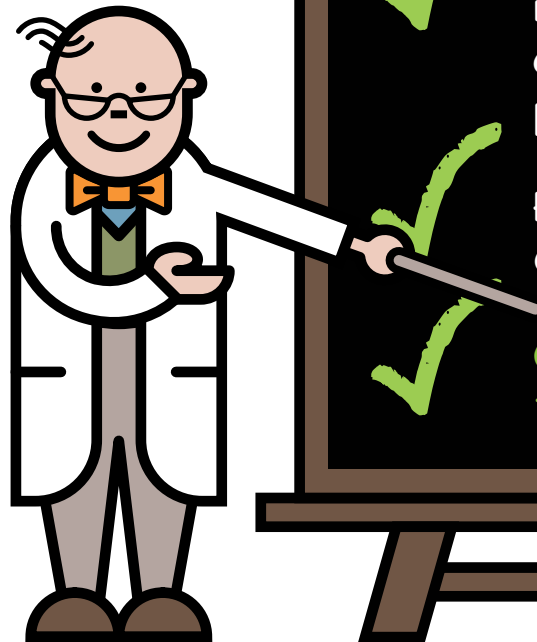


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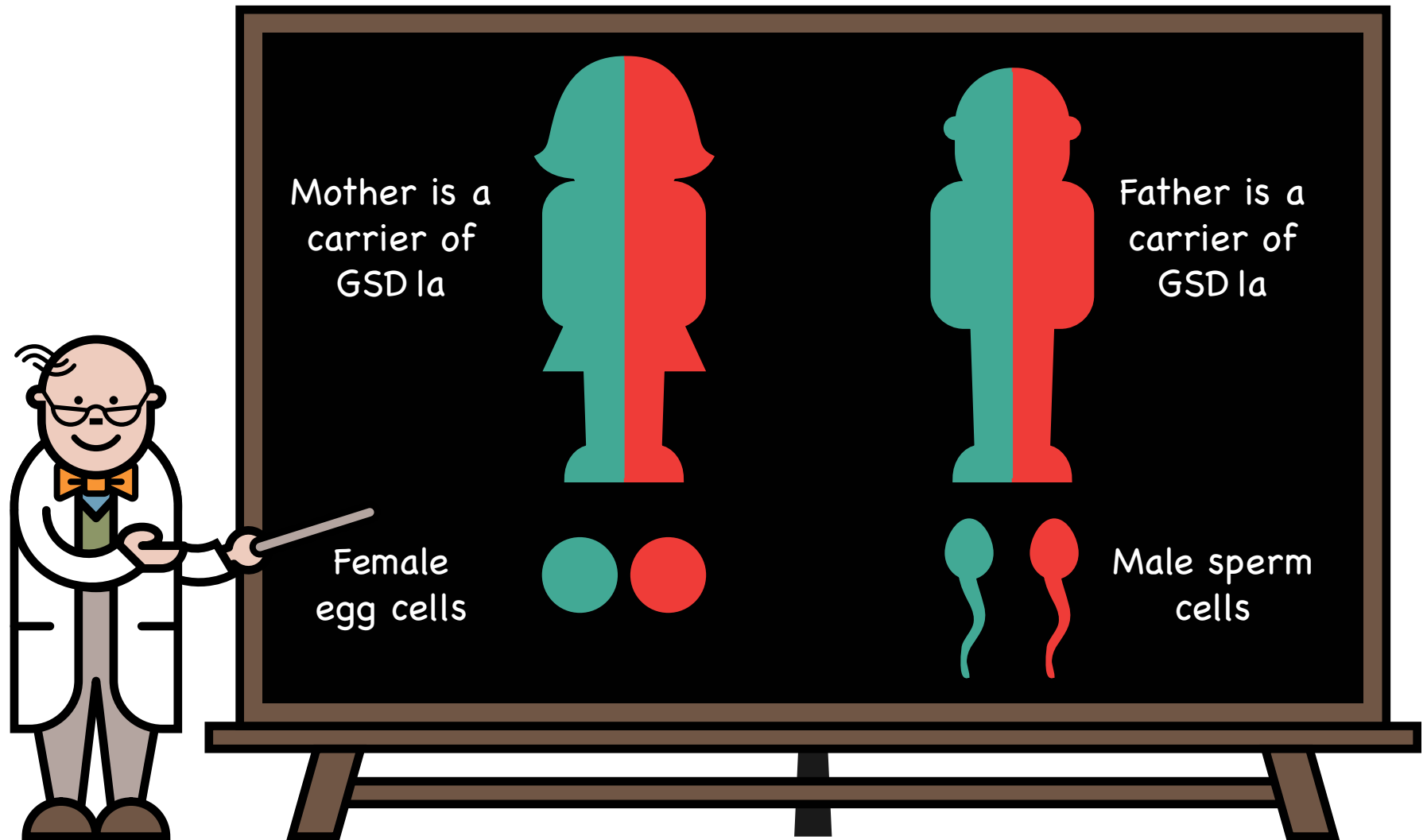
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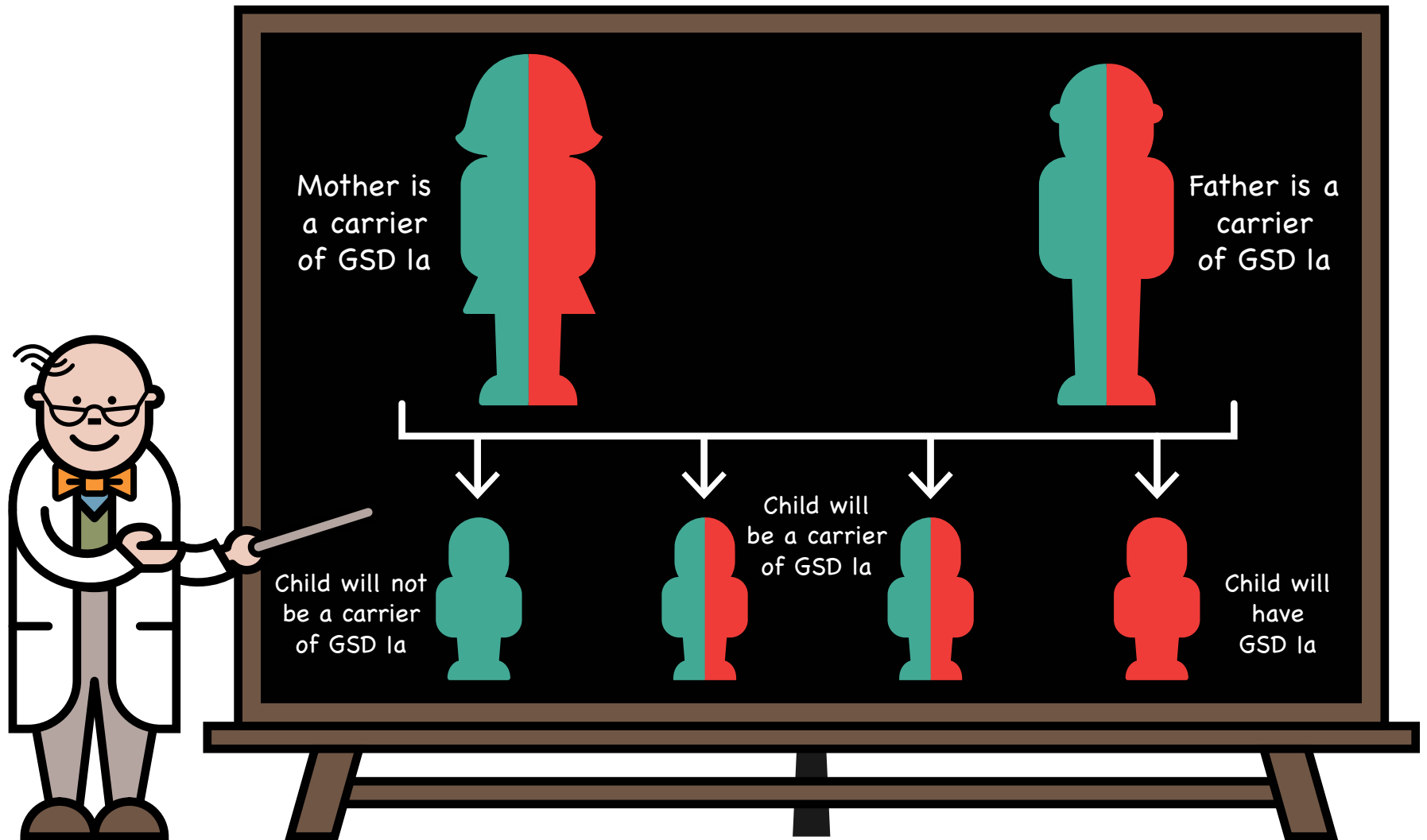
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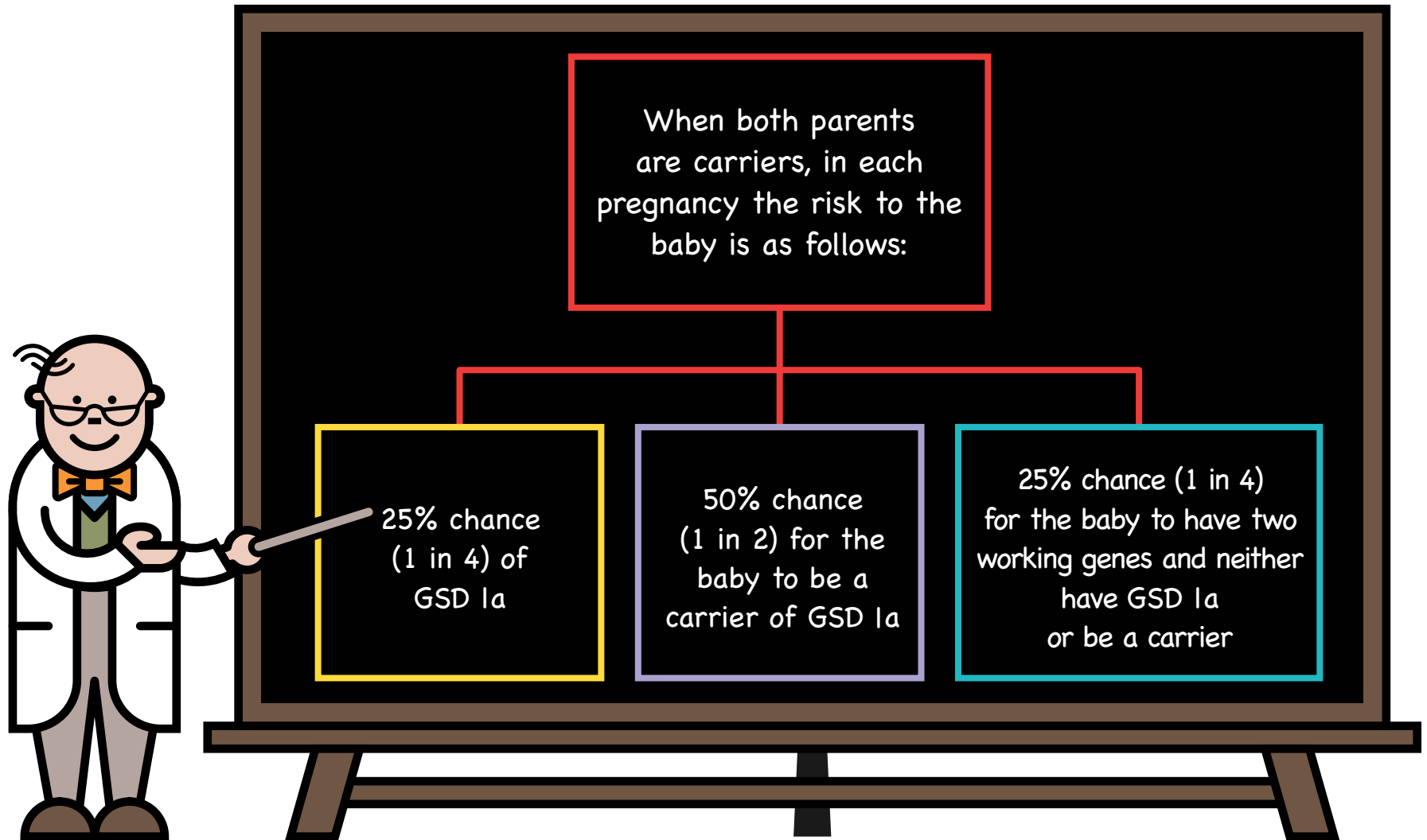
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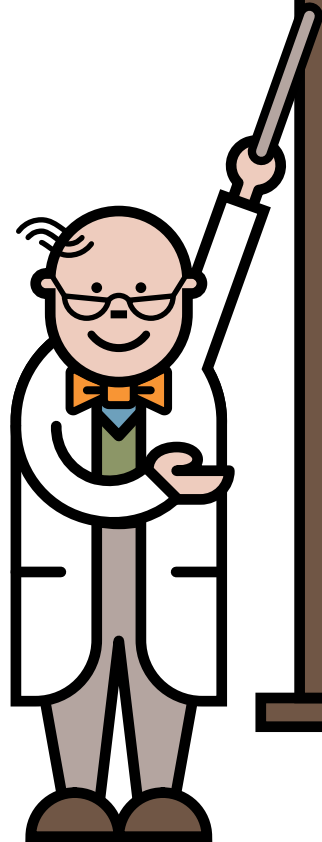
# Inheritance



# Future pregnancies



# Take home messages

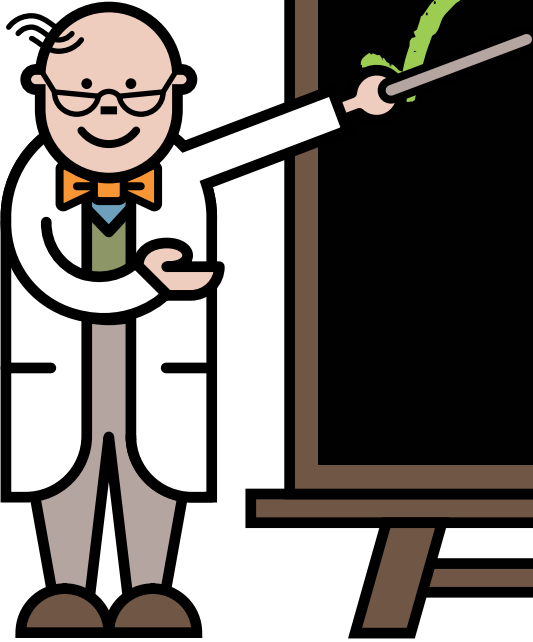


GSD 1a is a serious inherited metabolic condition that causes life threatening symptoms if left unmanaged

Children with GSD 1a are managed with regular meals and snacks based on carbohydrate, a healthy balanced diet and overnight feeding or regular feeds/drinks overnight

Remember, during illness, it is imperative that emergency feeds are started promptly, followed strictly and there are no delays

# Take home messages



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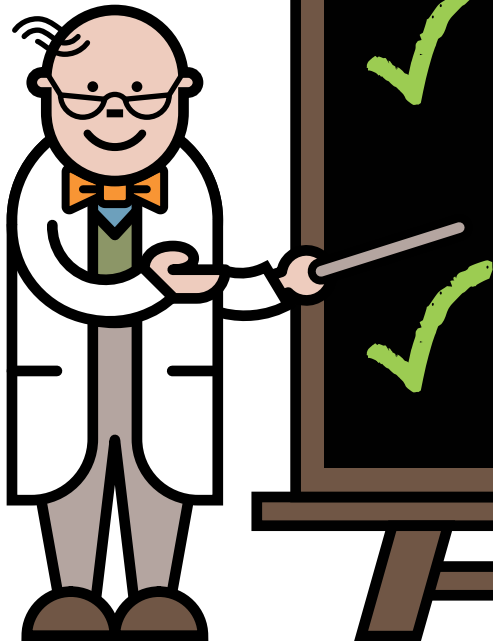
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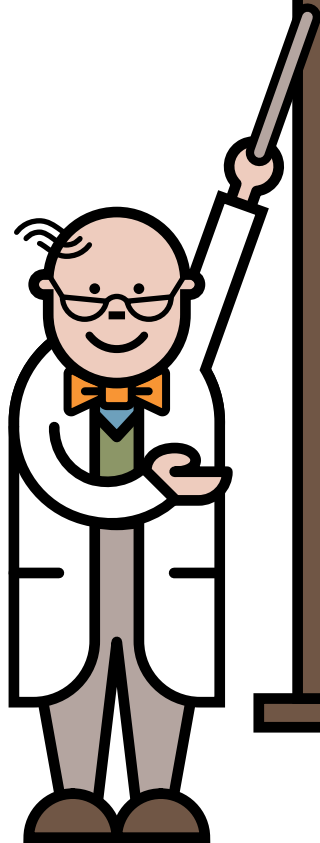
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# Helpful hints



Always ensure you have a good supply of your dietary products and glucose polymer powder and it is in date

Special feeds and dietary products are prescribed by your GP and you obtain them from your pharmacy or home delivery

Medications to control fever should be given as normally recommended – always keep supplies available

Remember to keep contact numbers of your metabolic team to hand

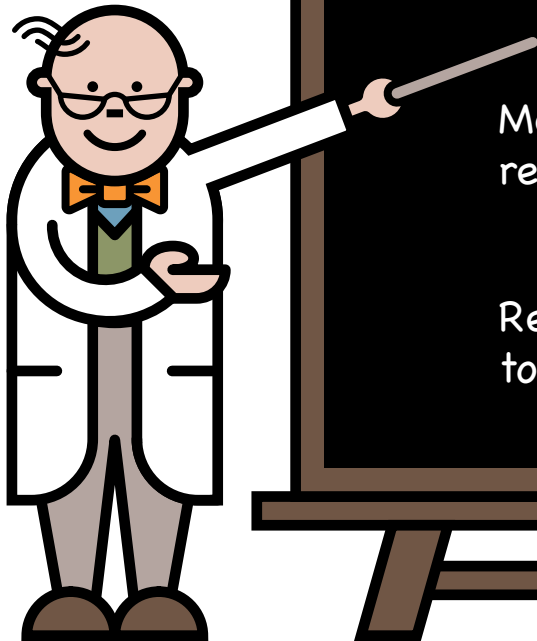


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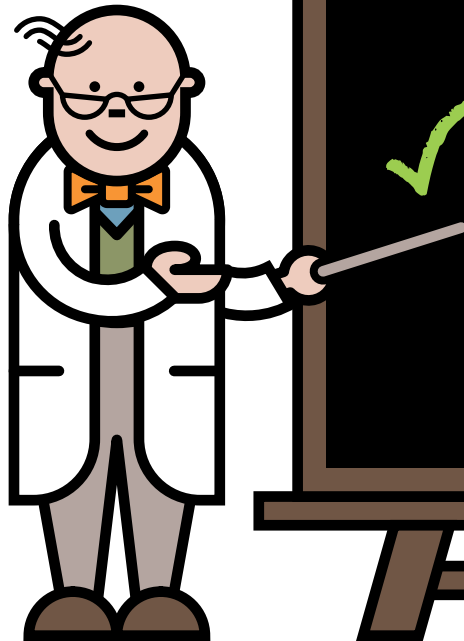
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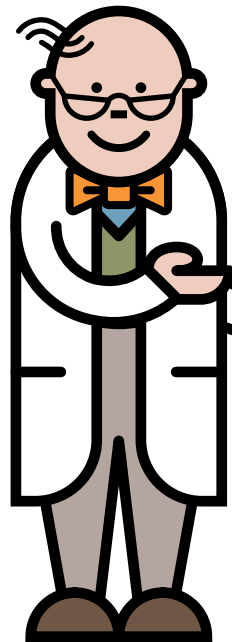
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# Who's who

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

**BIMDG**

British Inherited Metabolic Diseases Group



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