

# TEMPLE



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

ADAPTED BY THE DIETITIANS GROUP

**BIMDG**

British Inherited Metabolic Diseases Group



# CPT 1 DEFICIENCY

BASED ON THE ORIGINAL TEMPLE WRITTEN BY  
BURGARD AND WENDEL

VERSION 1, DECEMBER 2019

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.**

# Carnitine Palmitoyl Transferase 1 Deficiency

Information for families  
following a new diagnosis



ADAPTED BY THE DIETITIANS GROUP

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British Inherited Metabolic Diseases Group



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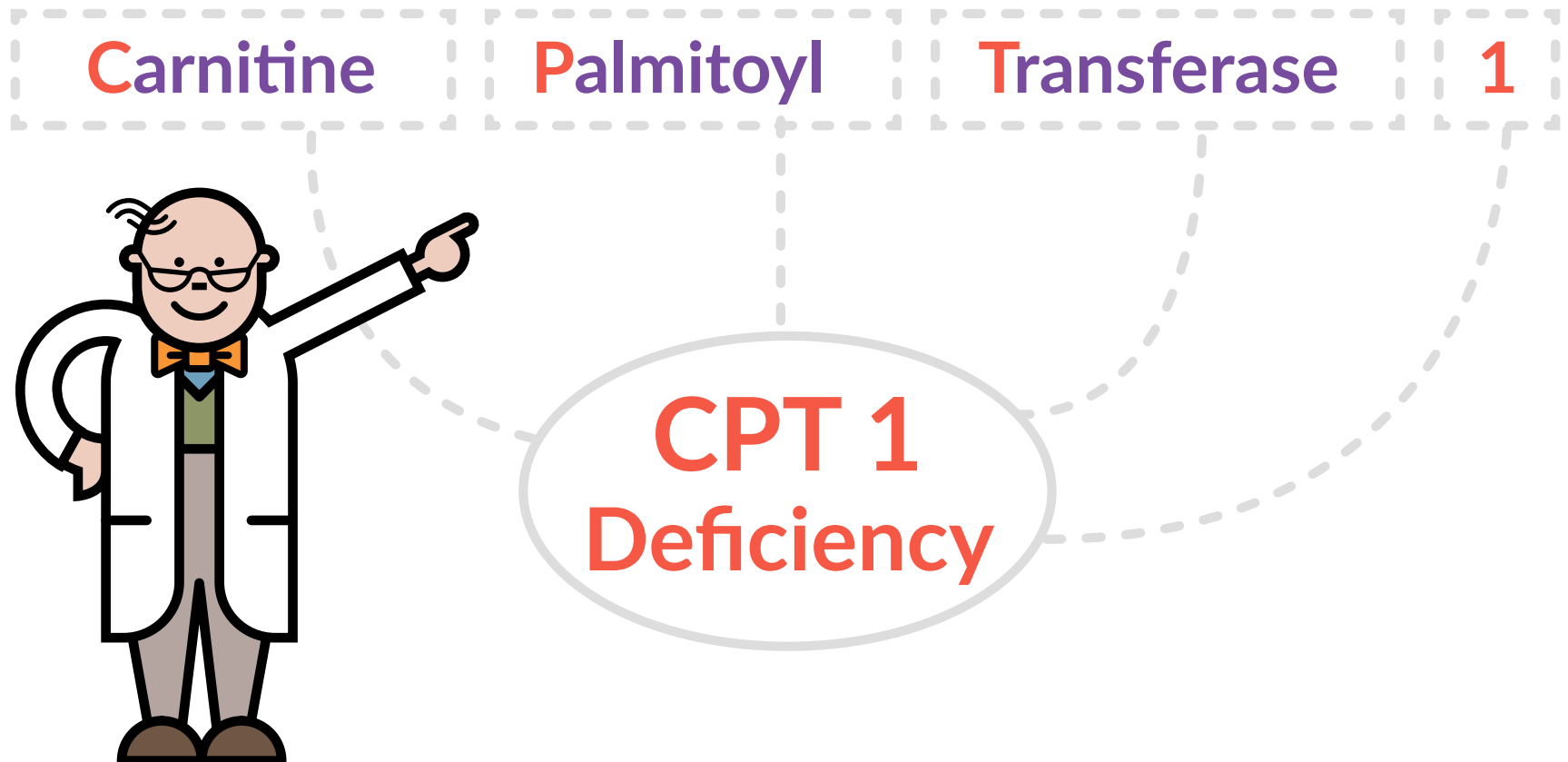
Tools Enabling Metabolic Parents LEarning

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# What is CPT 1 deficiency?

CPT 1 stands for Carnitine Palmitoyl Transferase 1 deficiency

It is an inherited metabolic condition



# Which foods supply the body with energy?

There are two main food groups that supply the body with energy:

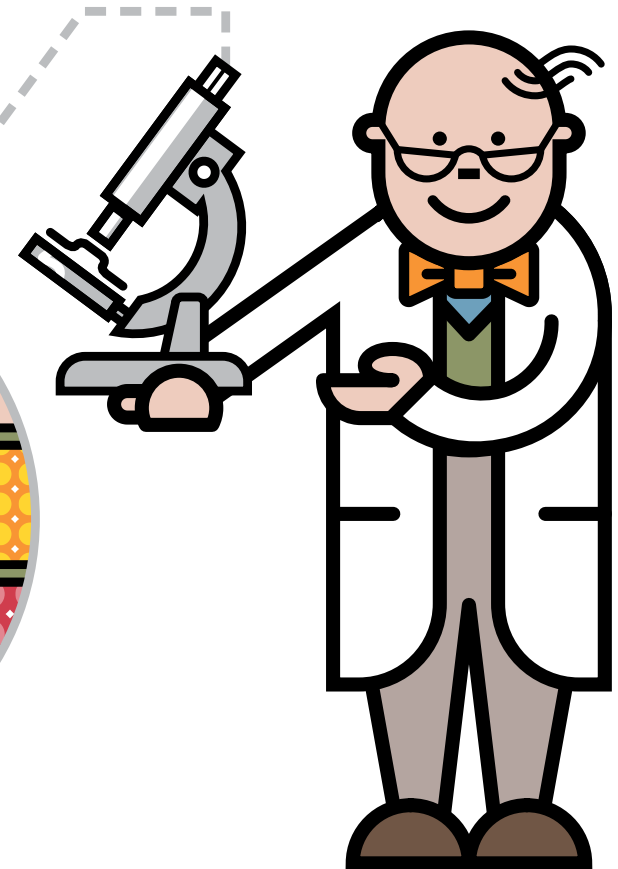
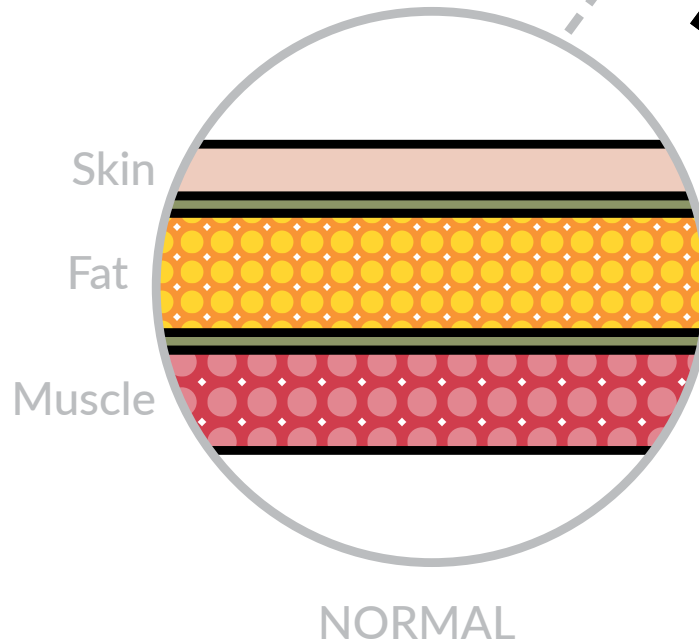
- Carbohydrates (starches and sugars) provide a readily available energy source
- Fats also provide energy. Fat is stored in the body so it can be used as an energy reserve



# CPT 1 deficiency and fat

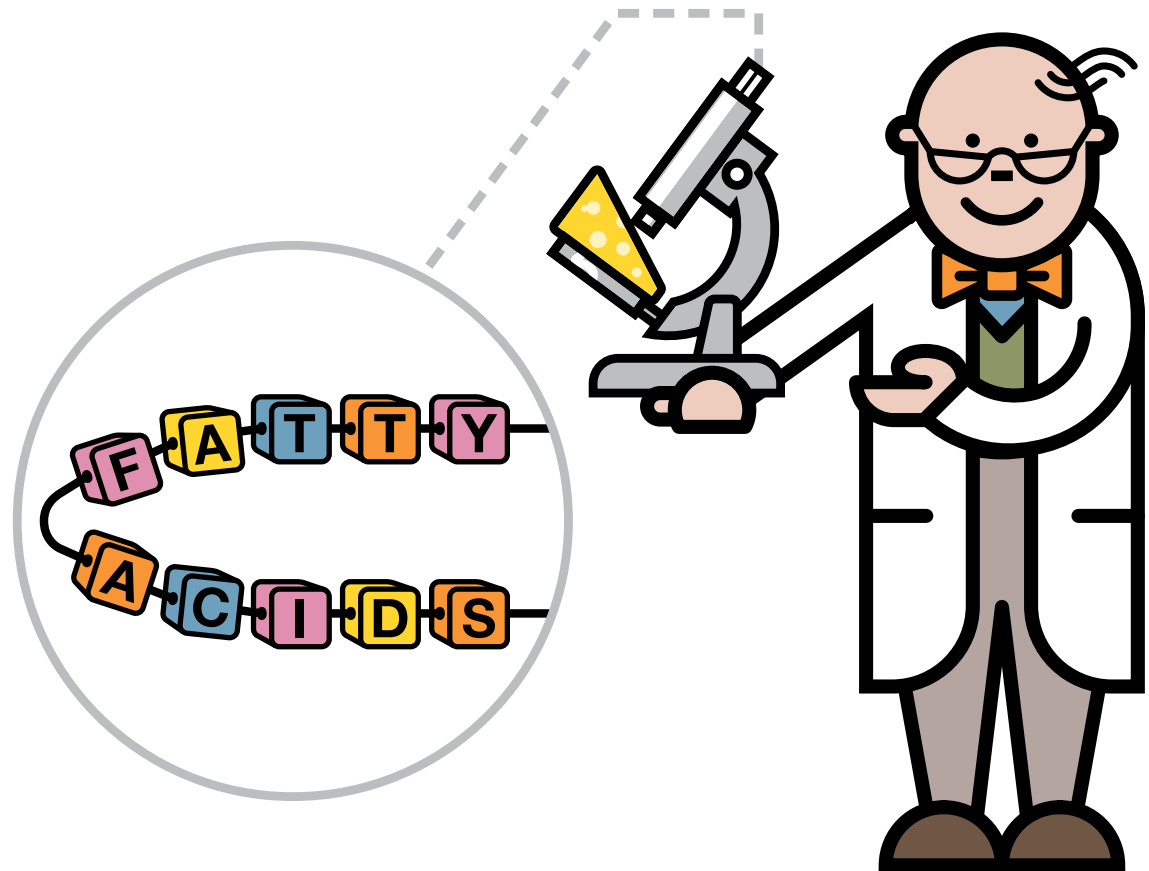
The body uses its own fat stores to provide energy when carbohydrate is depleted.

In CPT 1 deficiency, there are problems breaking down these fat stores to release energy.



# Breaking down fat stores for energy

Body fat stores are broken down into fatty acids.



# What are fatty acids?

Fatty acids are made up of carbon atoms joined together to form chains of many different lengths.



Short chain



Medium chain



Long chain



# Fatty acid transport

Fatty acid chains need to be transported into the cells of the body. This enables the body to produce energy in a form which it can use.



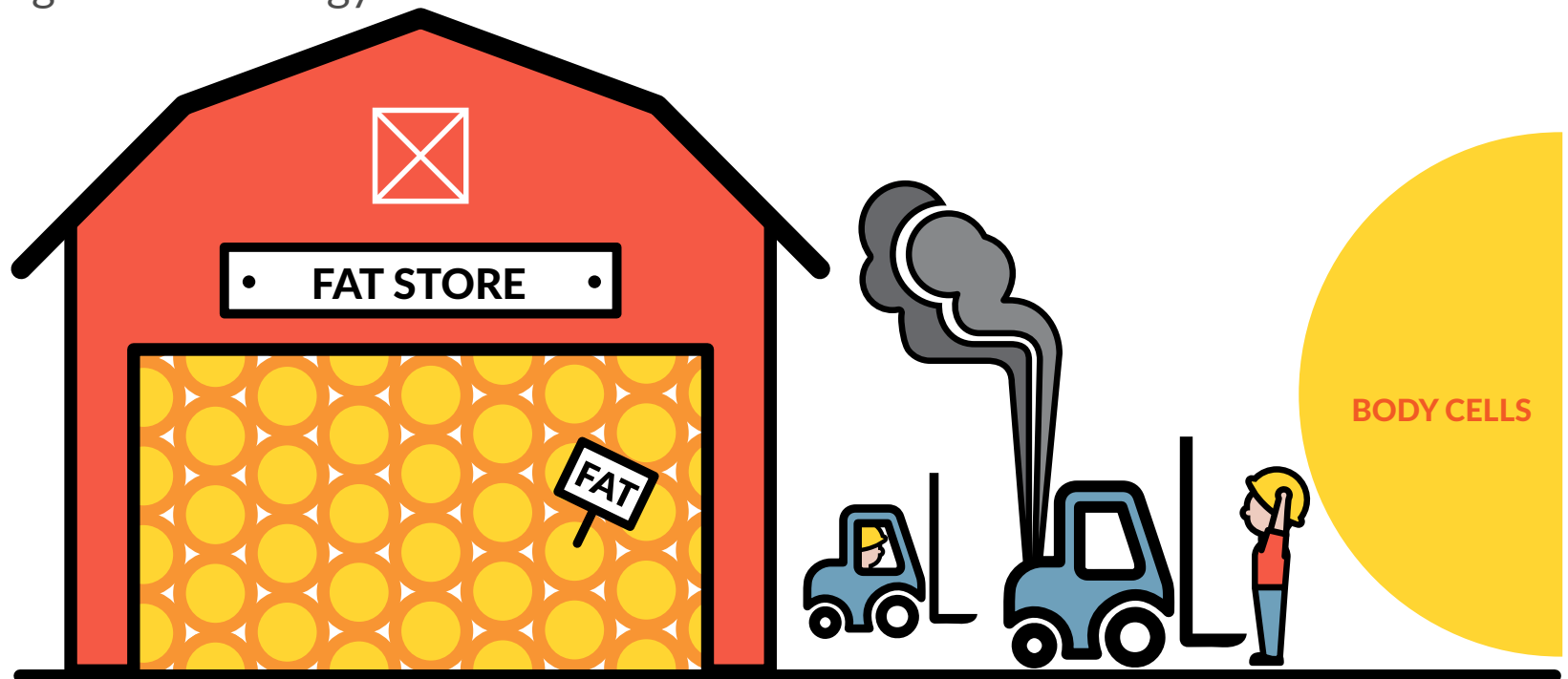
# What happens in CPT 1 deficiency?

In CPT 1 deficiency, the body lacks a chemical (enzyme) that helps convert fat stores into energy.

This **prevents** the long chain fats from being used for energy.

This is a problem when it is necessary to break down fats quickly.

This causes a shortage of energy supply.

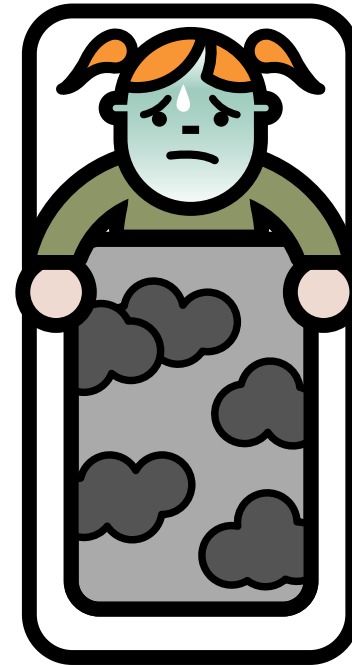
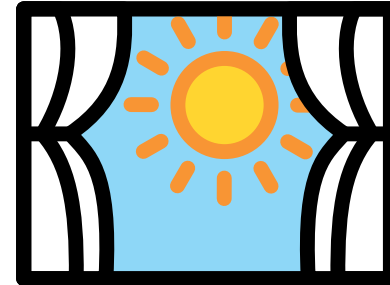


# What can go wrong in CPT 1 deficiency in infants?

There can be a shortage of energy supply and a build up of harmful chemicals with illness or lack of food.

Symptoms include:

- poor feeding
- excessive sleepiness
- rapid breathing
- seizures
- low blood sugar
- floppiness
- liver problems
- heart problems



# What can go wrong in CPT 1 deficiency?

If there is a shortage of energy and this is not corrected, it can lead to coma and brain damage and it may be life threatening.

**Hypoglycemia, lactic acidosis, and ketonuria can be prevented with timely and correct treatment.**



# Metabolic crisis

- A **ketone body** triggers the CPT 1 deficiency symptoms
- This leads to a lack of energy and build up of toxic chemicals
- It is usually triggered by childhood infections causing high temperatures, vomiting, and diarrhoea
- It can also be caused by going a very long time without food
- Avoidance of a metabolic crisis is essential



# How is CPT 1 deficiency diagnosed?

CPT 1 deficiency is suspected because of the pattern of chemicals (acylcarnitines and organic acids) found in the blood and urine.

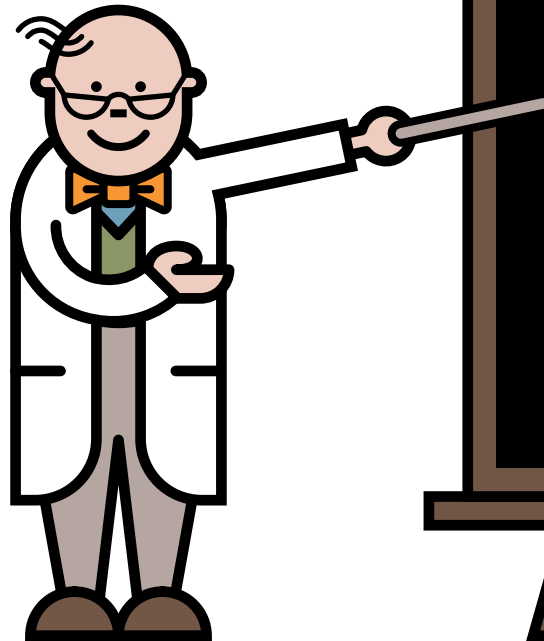
The diagnosis is confirmed by finding mutations in the CPT 1 gene.



# How is CPT 1 deficiency managed?

In babies, it is managed day to day by **avoiding** long periods without feeding, even when well.

The length of time babies can go without feeds is known as **the safe fasting time**.



## SAFE FASTING TIMES

- The safe fasting time varies for each baby with CPT 1 deficiency. Your metabolic team will advise.
- It is important they receive regular feeds during the day and at night.
- They should not miss scheduled feeds.

# How is CPT 1 deficiency managed day to day?

Special formula containing a fat called medium chain triglycerides (MCT)



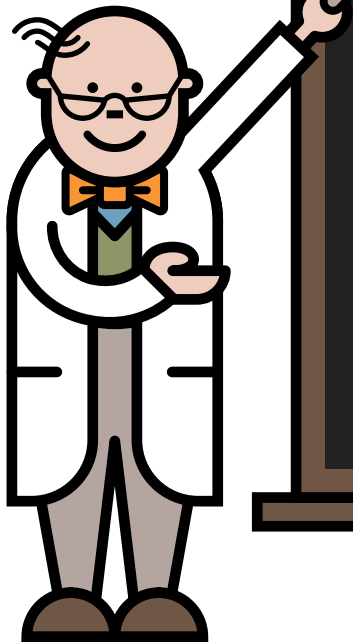
Low fat diet



MCT oil for cooking or MCT as a supplement may be used



Regular mealtimes as recommended by your metabolic team





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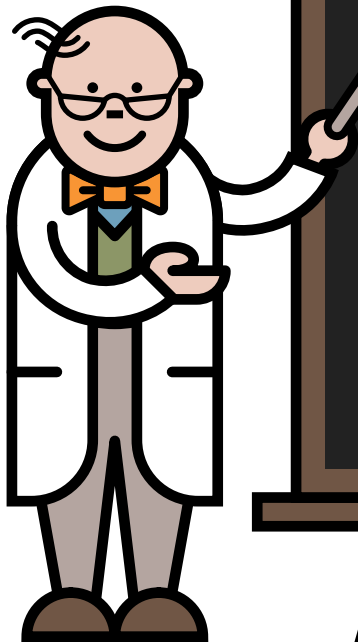
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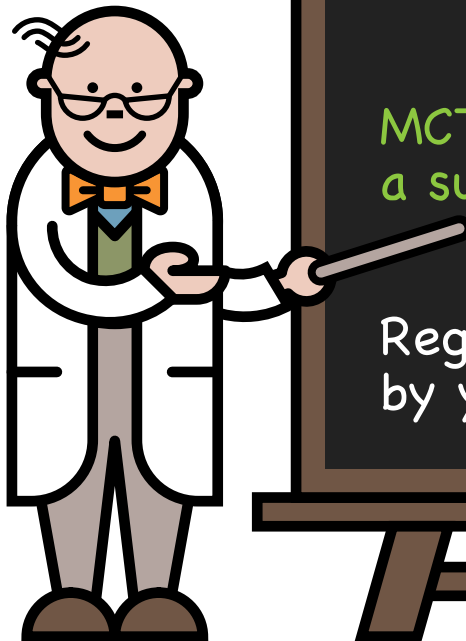
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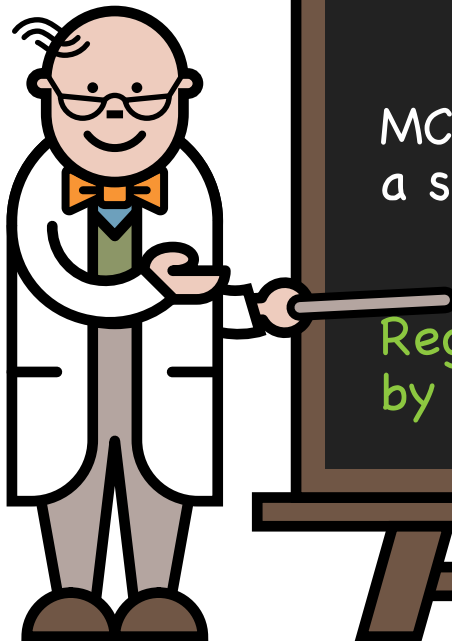
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# CPT 1 deficiency and fat

The diet needs to be low in fat.

**F** Many foods are high in fat e.g. full fat

Many foods are high in fat e.g. full fat milk, full fat cheese, fatty meat, eggs, ice cream, chips, crisps and chocolate.

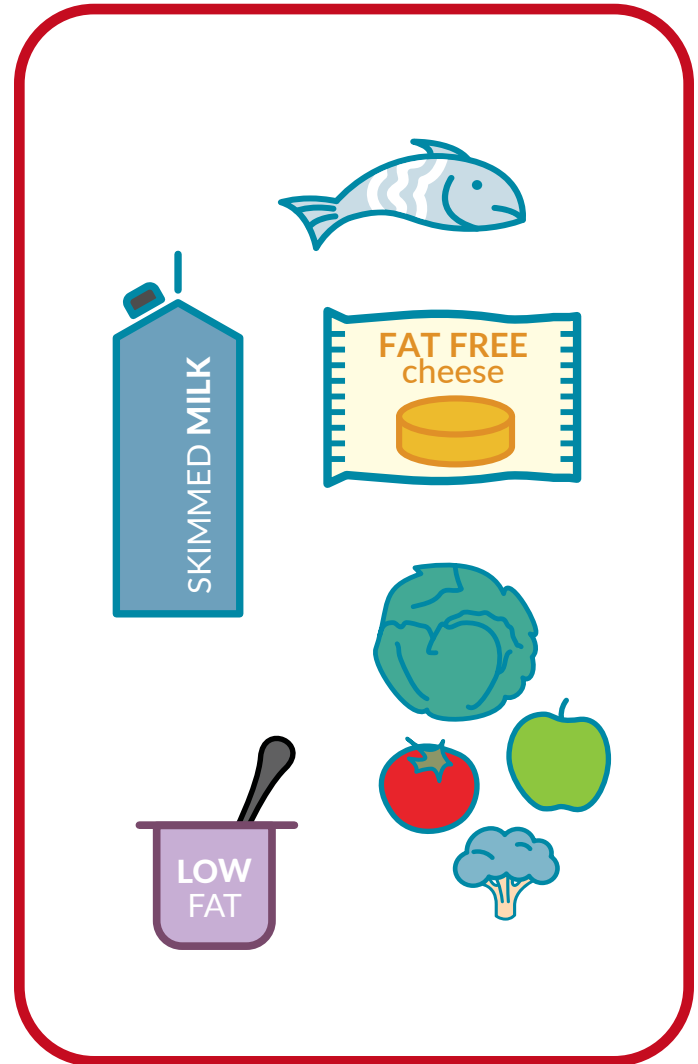


# Low fat foods

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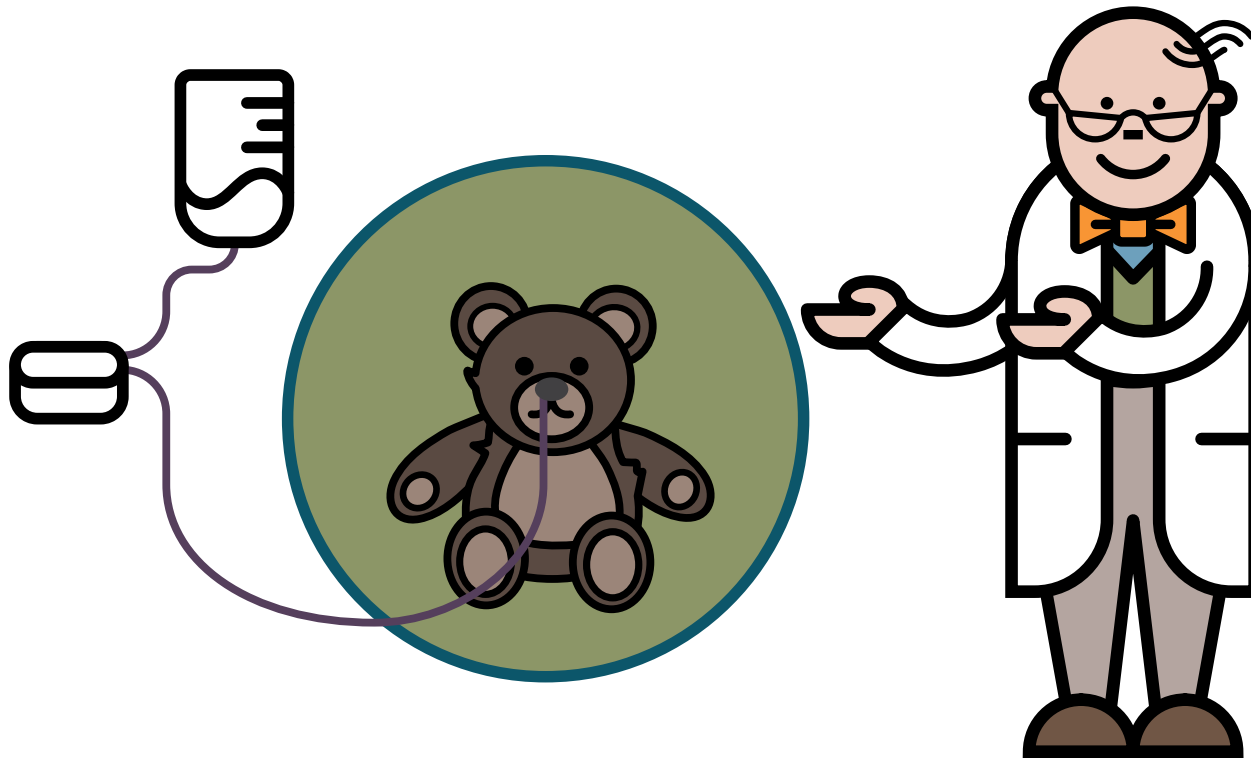
## Low fat foods are permitted.

There is a wide range of low fat foods available e.g. skimmed milk, low fat yoghurt/cheese, white fish, fruit and vegetables.



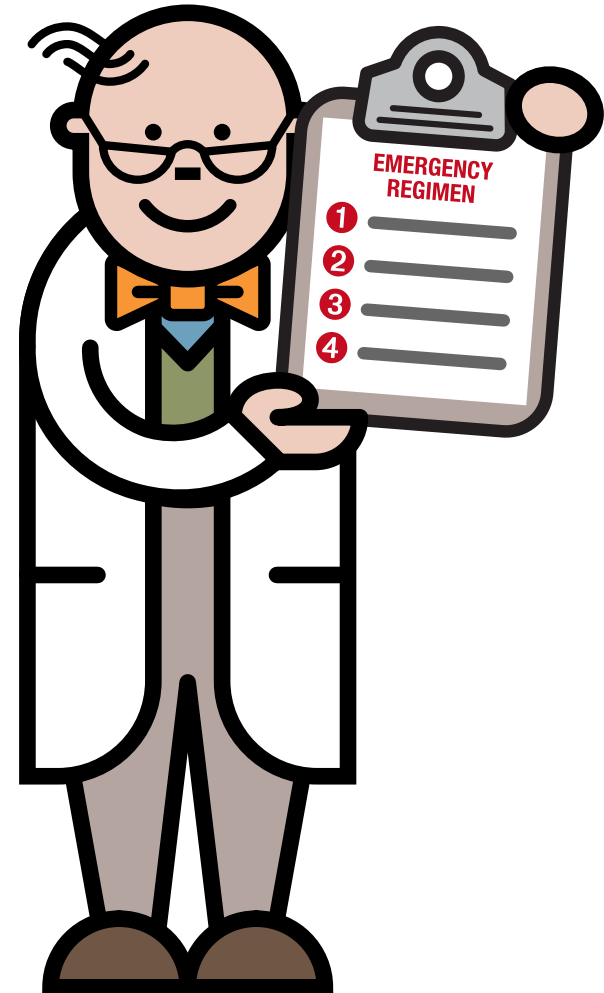
# Is tube feeding needed?

In babies with the most severe forms of CPT 1 deficiency, tube feeding may be necessary. This will ensure energy, nutrient and fluid needs are met.



# How is CPT 1 deficiency managed during illness?

- During any childhood illness, an emergency regimen is given
- This provides energy and prevents build up of harmful chemicals that cause a metabolic crisis



# How is CPT 1 deficiency managed during illness?

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



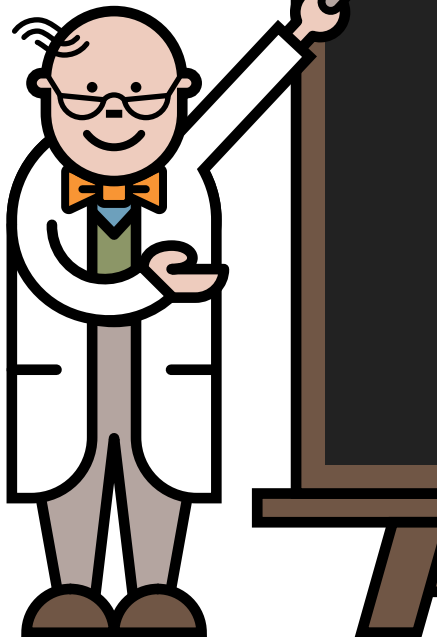
This must be given regularly  
day and night



Phone your metabolic team  
for help if your child is unwell



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





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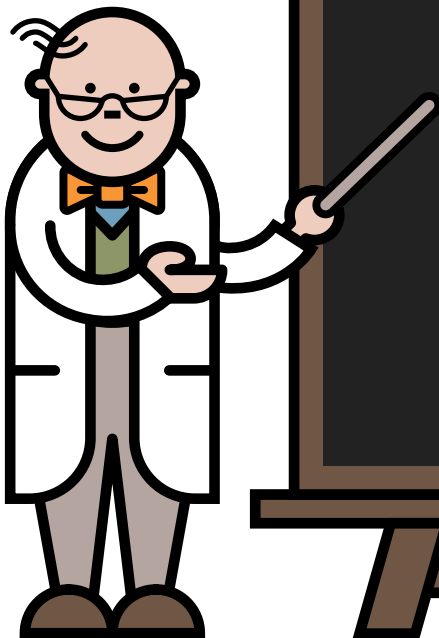
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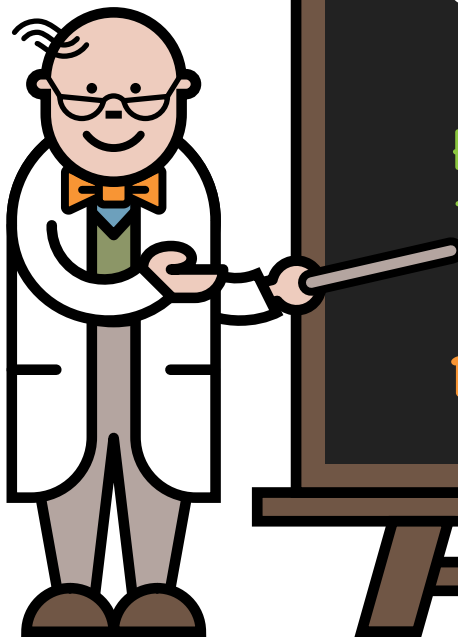
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# Checklist for illness

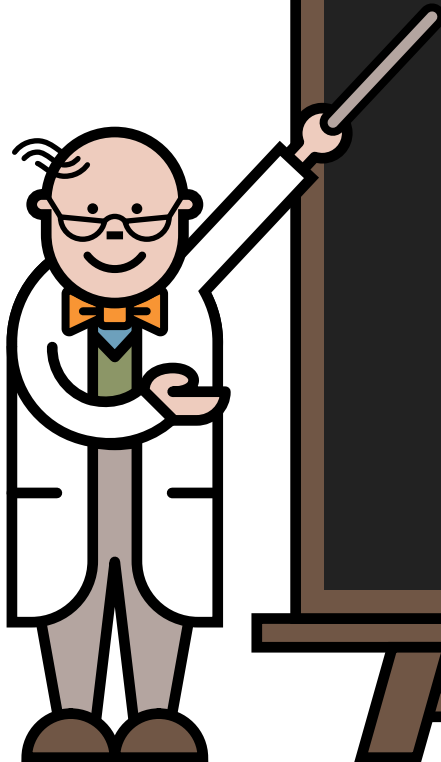
Always take full amounts of emergency feeds as prescribed



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



Phone your metabolic team



# Checklist for illness

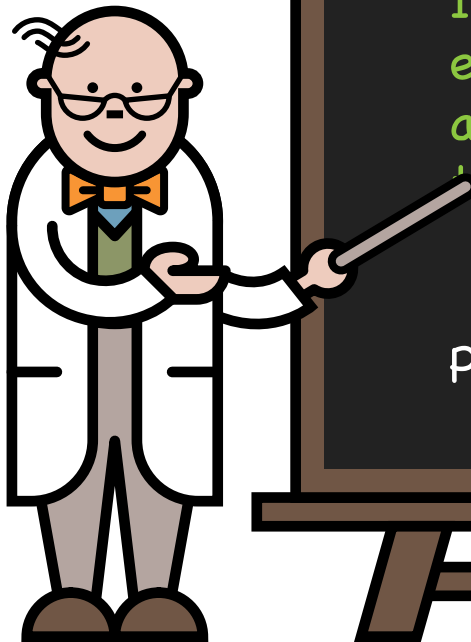
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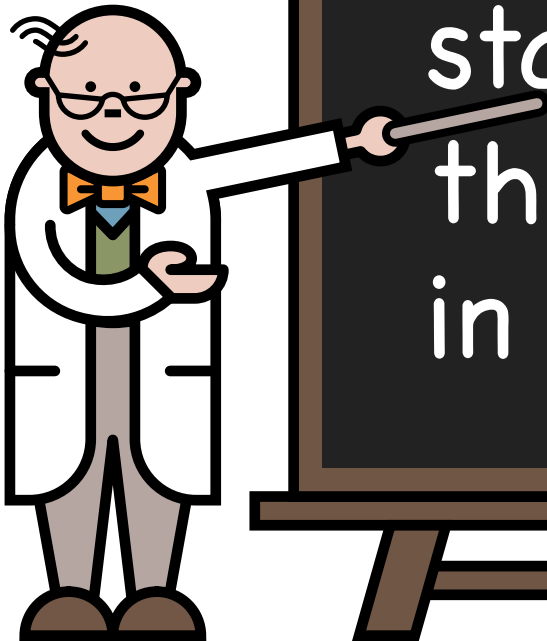


Phone your metabolic team



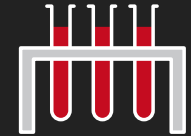
## Key message

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



# How is CPT 1 deficiency monitored?

Blood tests to check  
nutrient levels



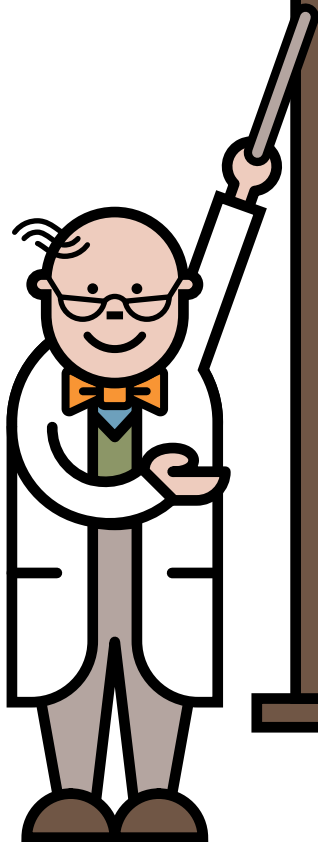
Height and weight



Developmental checks  
& other medical checks

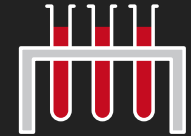


Emergency feeds are  
adjusted with age



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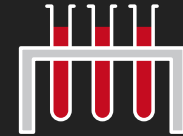
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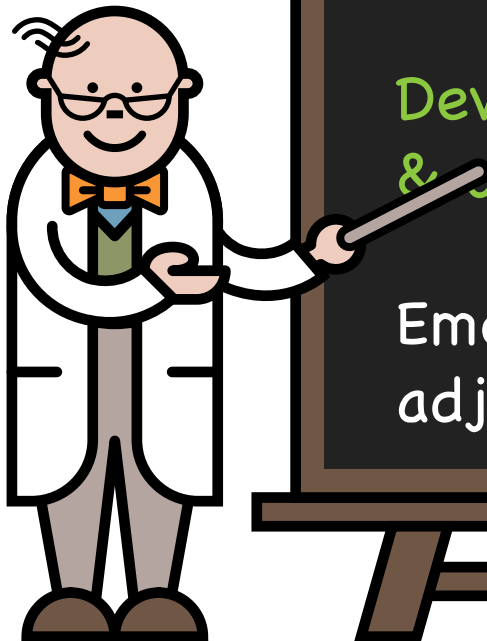
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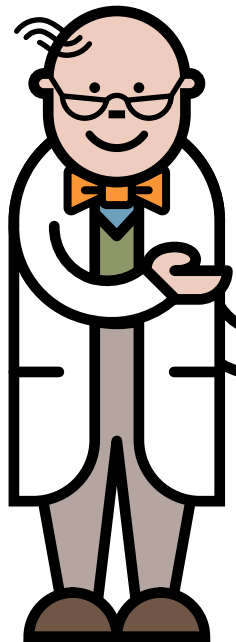
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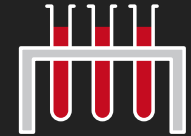
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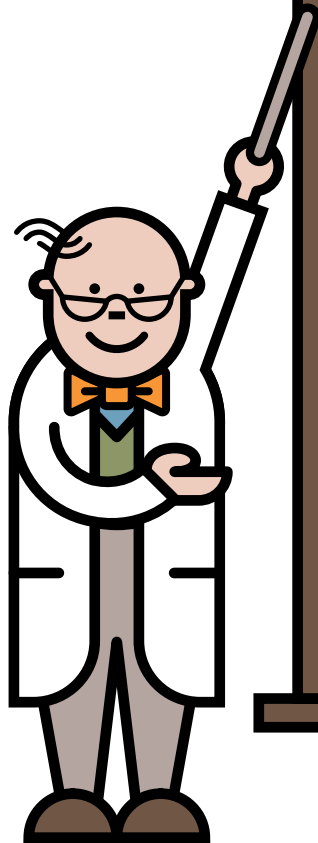
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# Chromosomes, genes, mutations



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

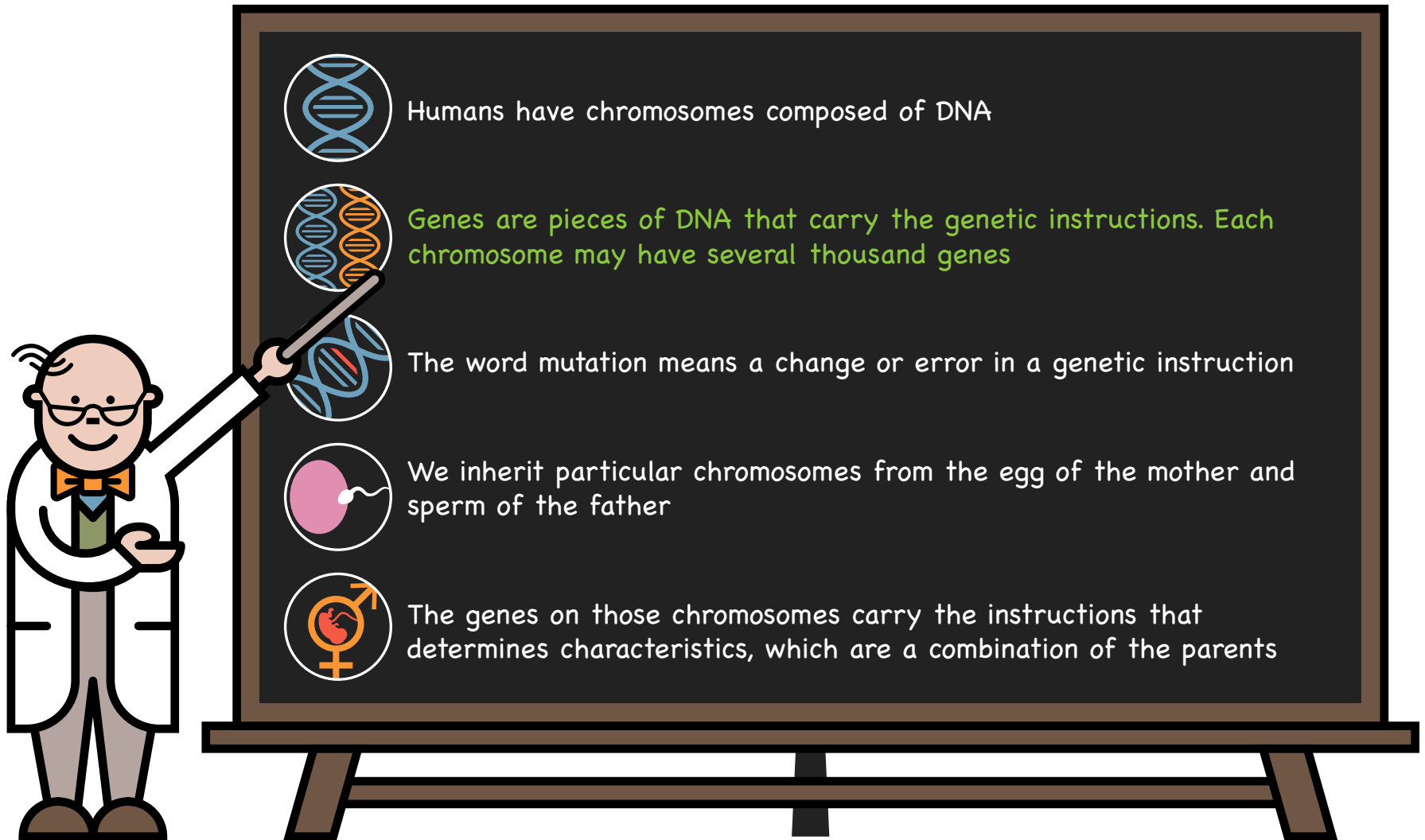



We inherit particular chromosomes from the egg of the mother and sperm of the father





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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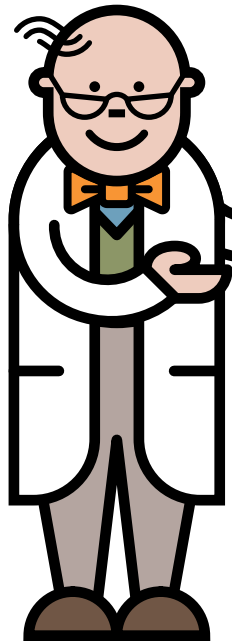
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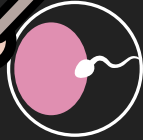
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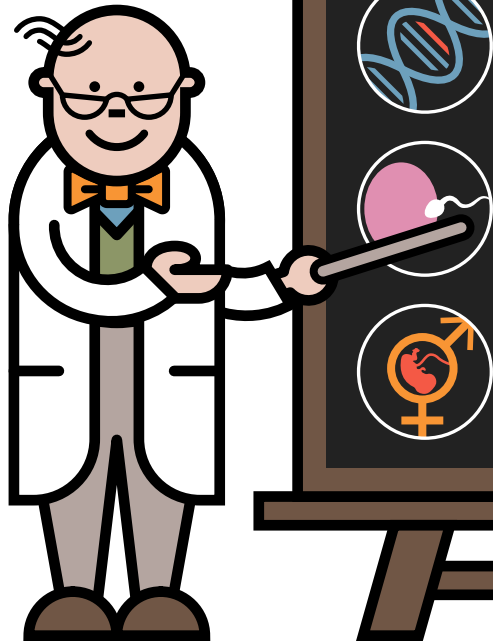


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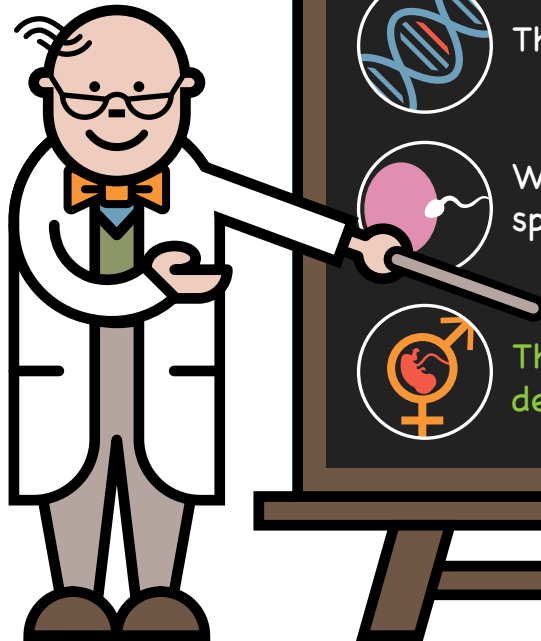


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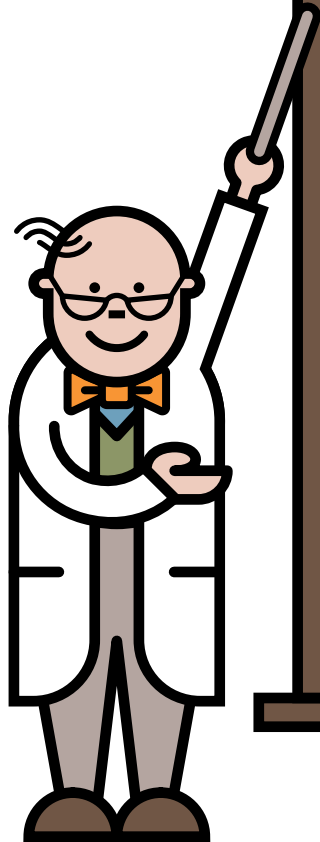


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# Inheritance



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

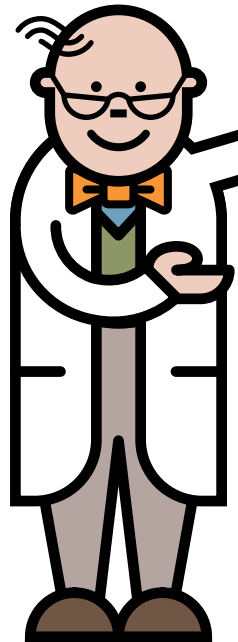
Everyone has a pair of genes that make the Carnitine Palmitoyl Transferase 1 enzyme. In children with CPT 1 deficiency, neither of these genes works correctly. These children inherit one non-working CPT 1 gene from each parent

Parents of children with CPT 1 deficiency are carriers of the condition

Carriers do not have CPT 1 deficiency because the other gene of this pair is working correctly



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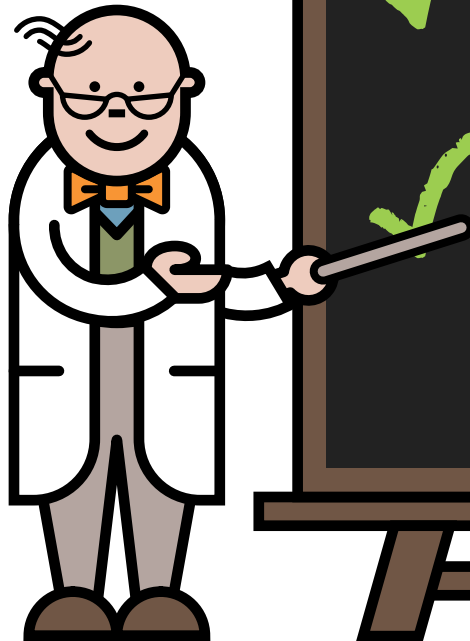
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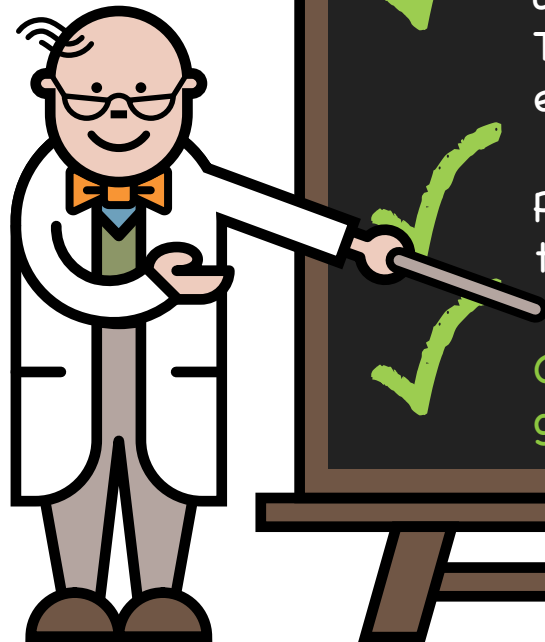
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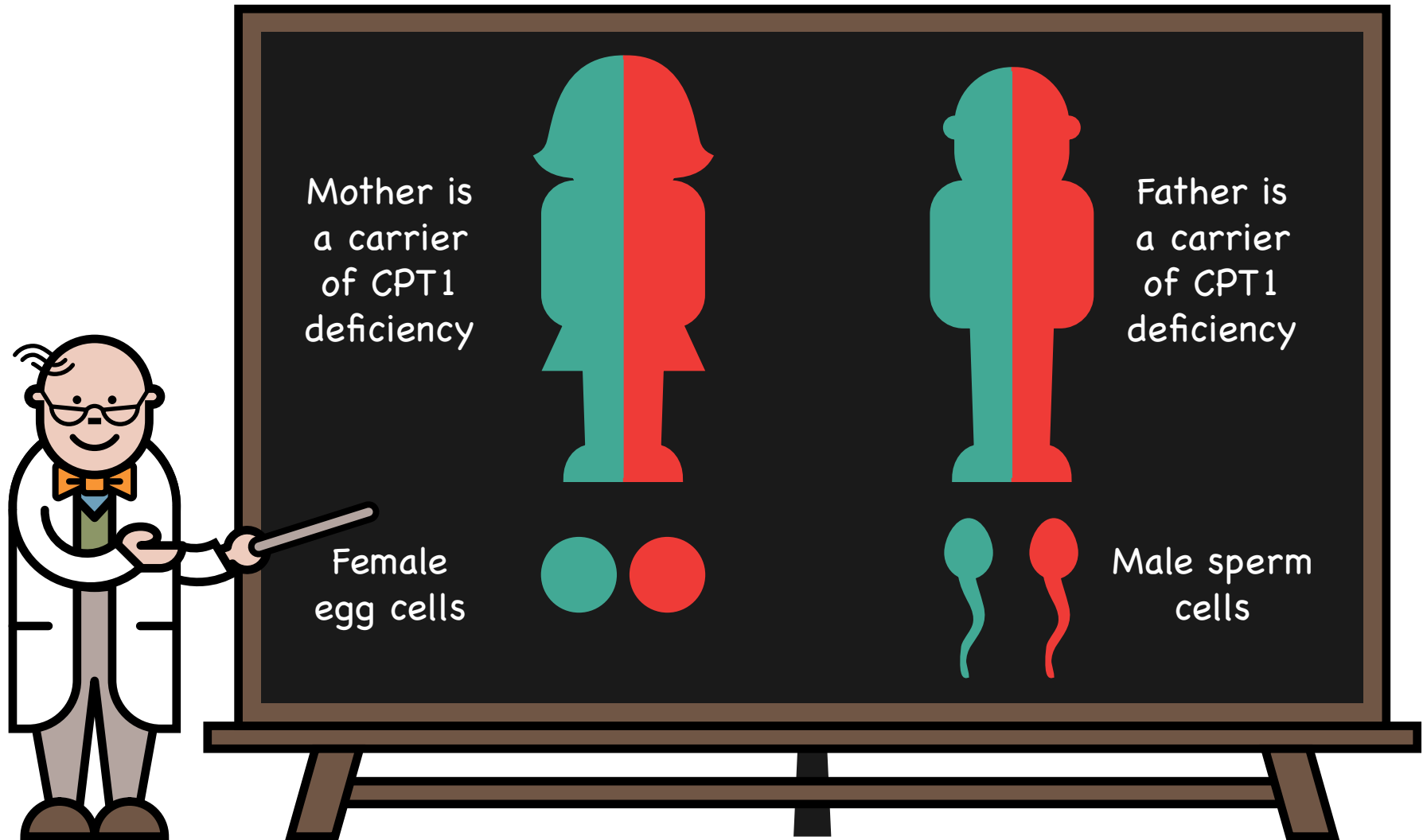
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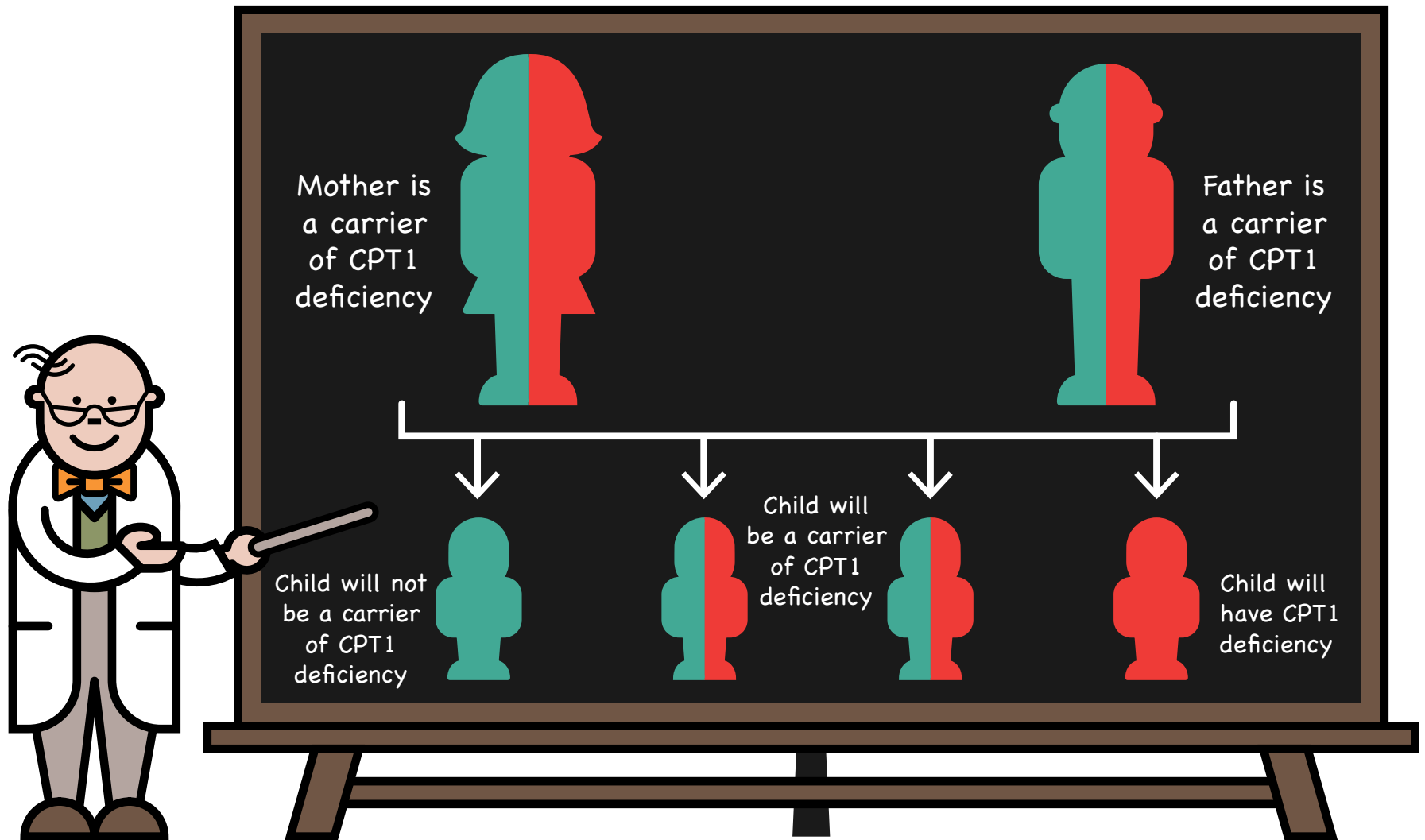
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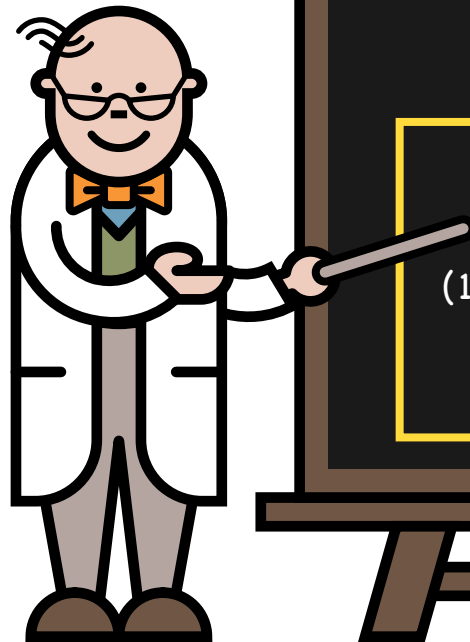
# Inheritance – Autosomal recessive (carriers of CPT1 deficiency)



# Inheritance – Autosomal recessive – possible combinations



# Future pregnancies



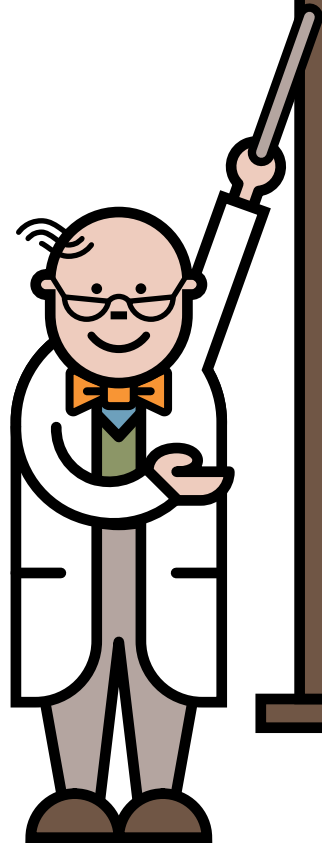
When both parents are carriers, in each pregnancy the risk to the baby is as follows:

25% chance  
(1 in 4) of CPT1  
deficiency

50% chance  
(1 in 2) for the  
baby to be a  
carrier of CPT1  
deficiency

25% chance (1 in 4)  
for the baby to have two  
working genes and neither  
have CPT1 deficiency  
or be a carrier

# Take home messages



CPT1 deficiency is a serious inherited metabolic condition that causes life threatening symptoms if left unmanaged

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

Infants with CPT1 deficiency are managed with MCT formula. Older children need regular meals and some may also require a low fat diet and MCT oil

People with CPT1 deficiency should lead normal, healthy and active lives

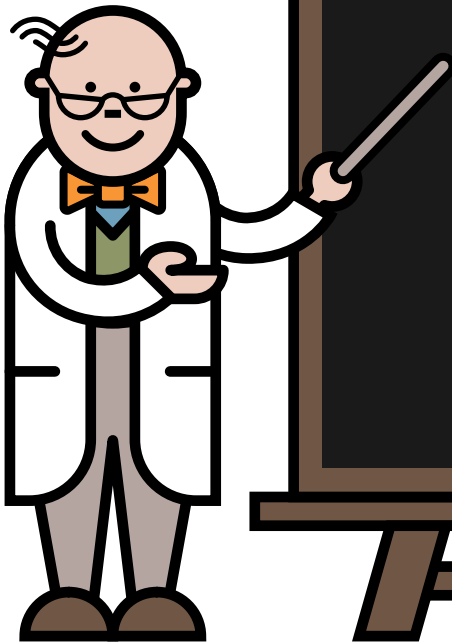
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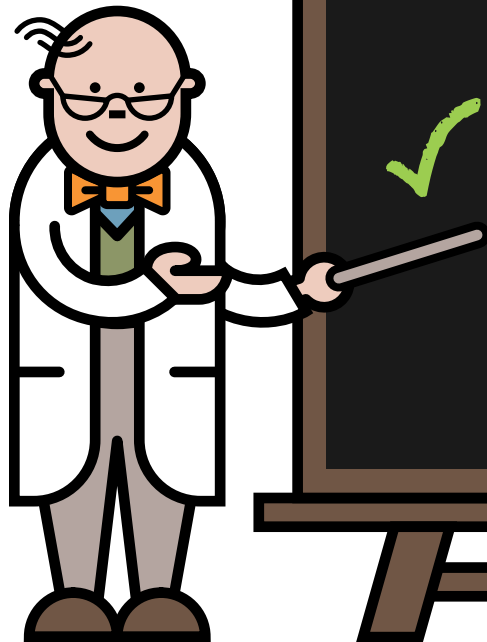
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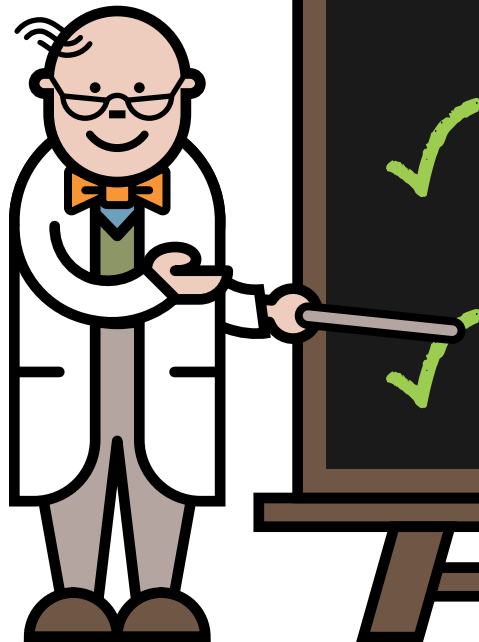
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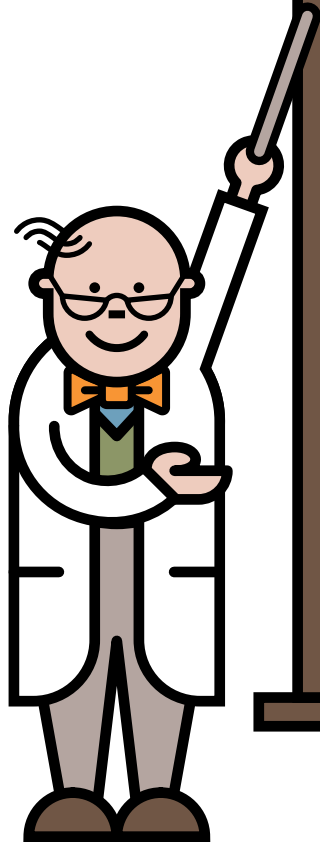


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# Helpful hints



Always ensure you have a good supply of your emergency 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

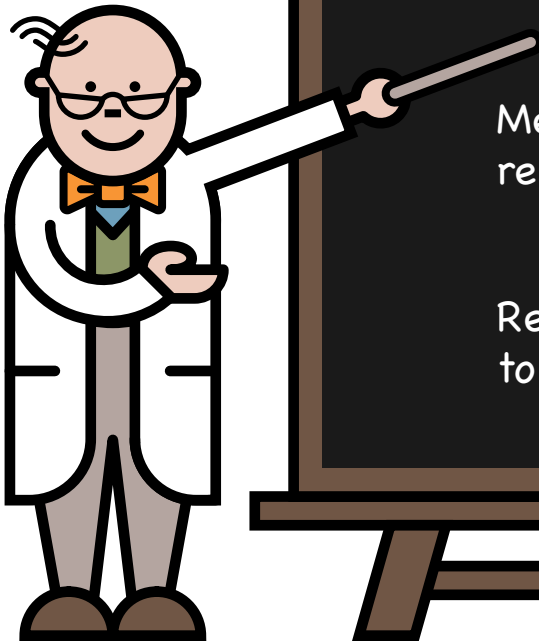
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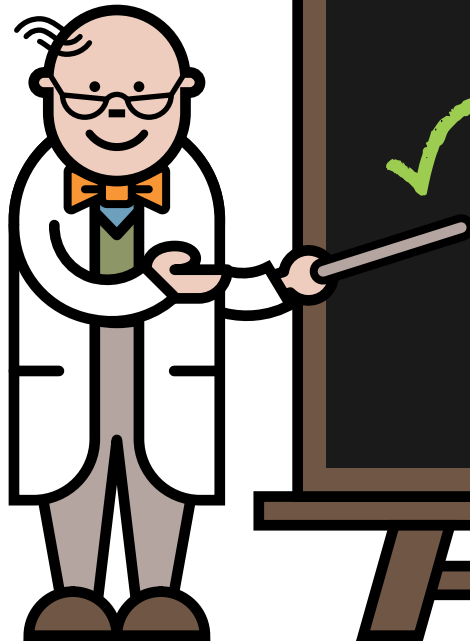
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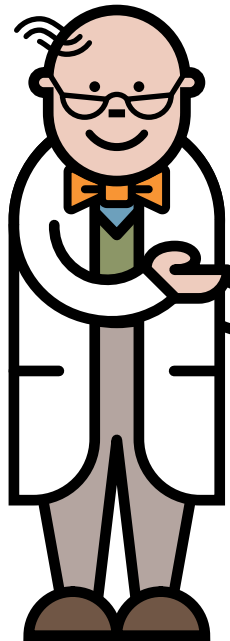
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✓ Special feeds and dietary products are prescribed by your GP and you obtain them from your pharmacy or home delivery

✓ 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

# Helpful hints



- ✓ Always ensure you have a good supply of your emergency 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

# Who's who

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

**BIMDG**

British Inherited Metabolic Diseases Group



**METABOLIC  
SUPPORT UK**

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[www.bimdg.org.uk](http://www.bimdg.org.uk)

[www.nutricia.co.uk](http://www.nutricia.co.uk)

[www.metabolicsupportuk.org](http://www.metabolicsupportuk.org)