

TEMPLE



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

ADAPTED BY THE DIETITIANS GROUP

BIMDG

British Inherited Metabolic Diseases Group



CPT II Deficiency

BASED ON THE ORIGINAL TEMPLE WRITTEN BY
BURGARD AND WENDEL

VERSION 3, APRIL 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).

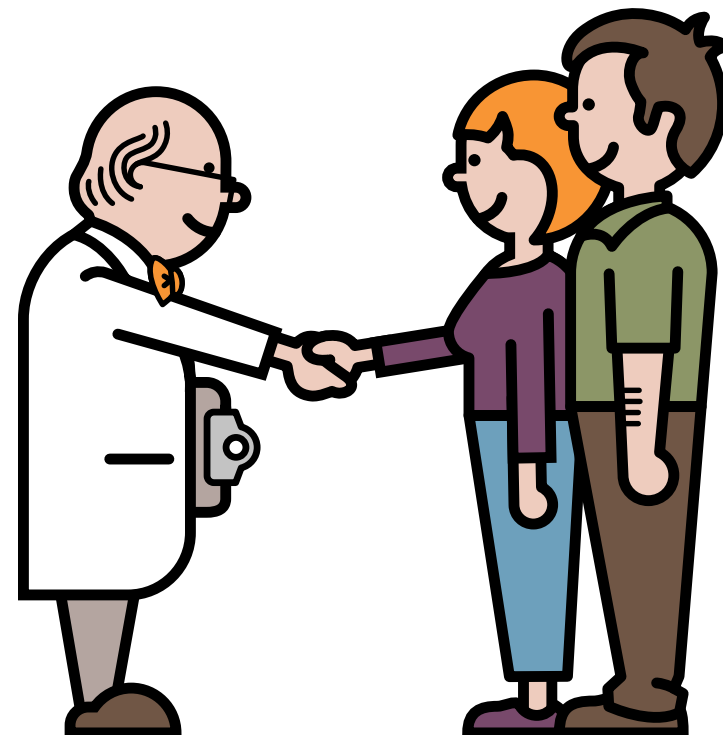
The team are Rachel Skeath, Karen van Wyk, Pat Portnoi and Anita MacDonald. The group is facilitated by Heidi Chan from Nutricia.

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.

Carnitine Palmitoyl Transferase II Deficiency

Information for families following
a new diagnosis



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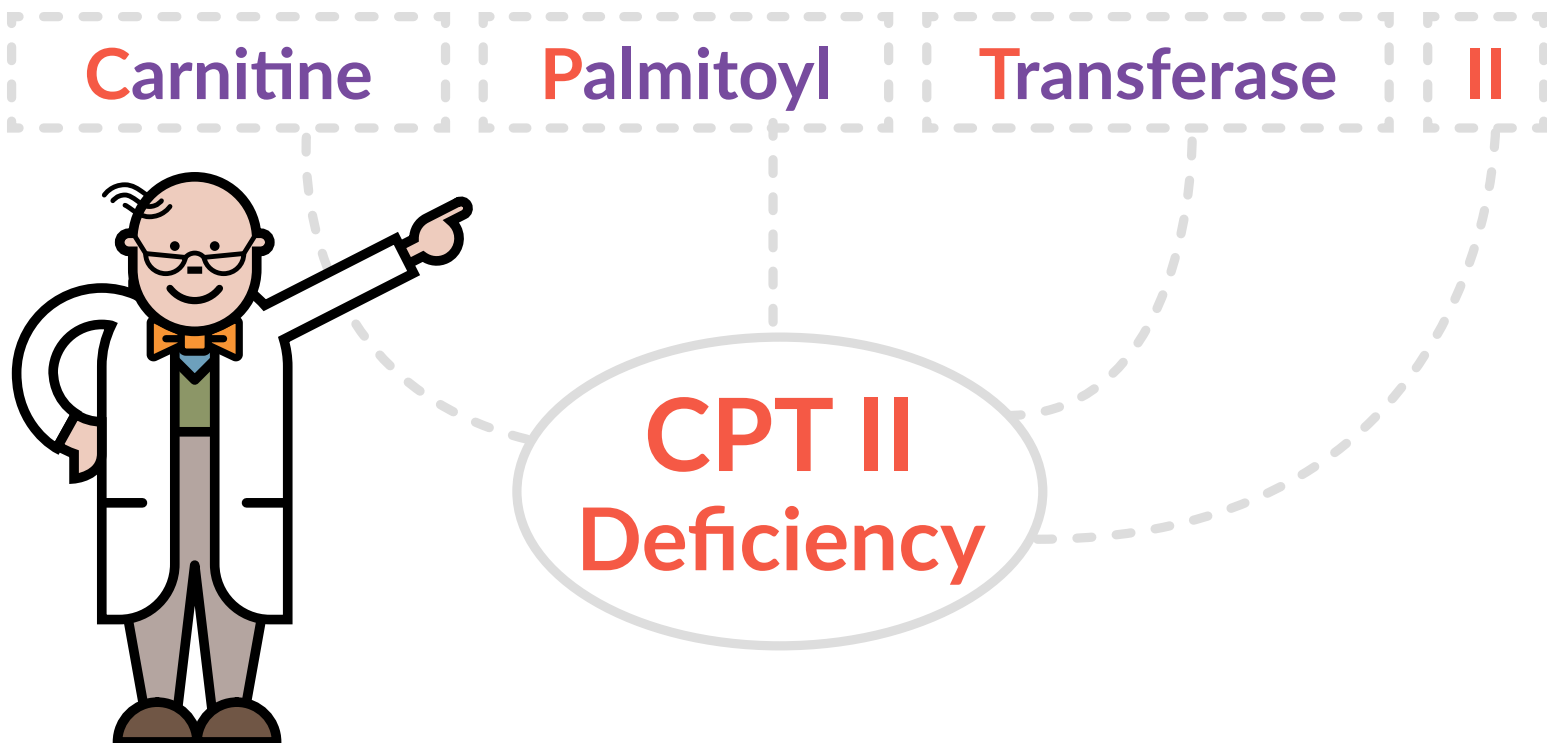
Tools Enabling Metabolic Parents LEarning

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What is CPT II deficiency?

CPT II stands for Carnitine Palmitoyl Transferase II deficiency

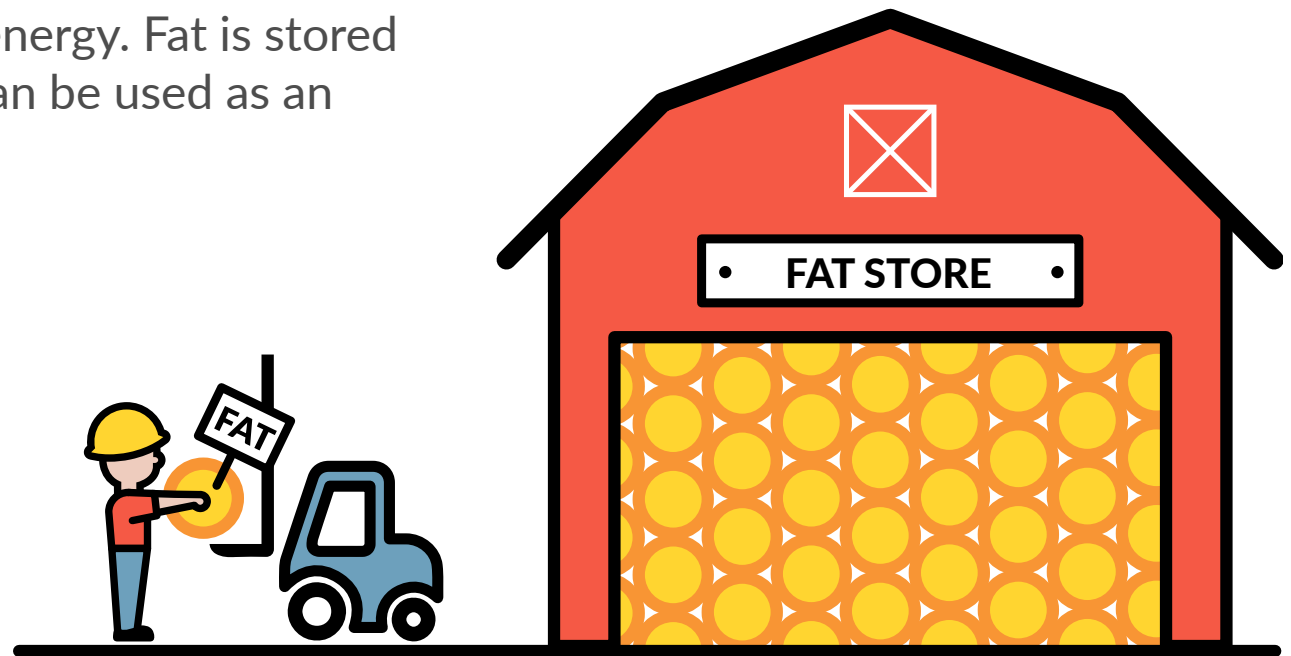
It is an inherited metabolic condition



Which foods supply the body with energy?

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

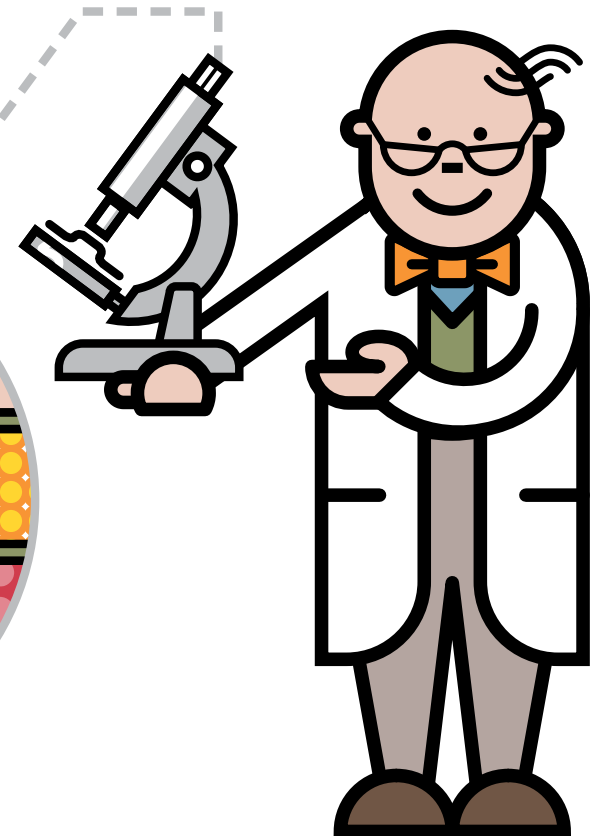
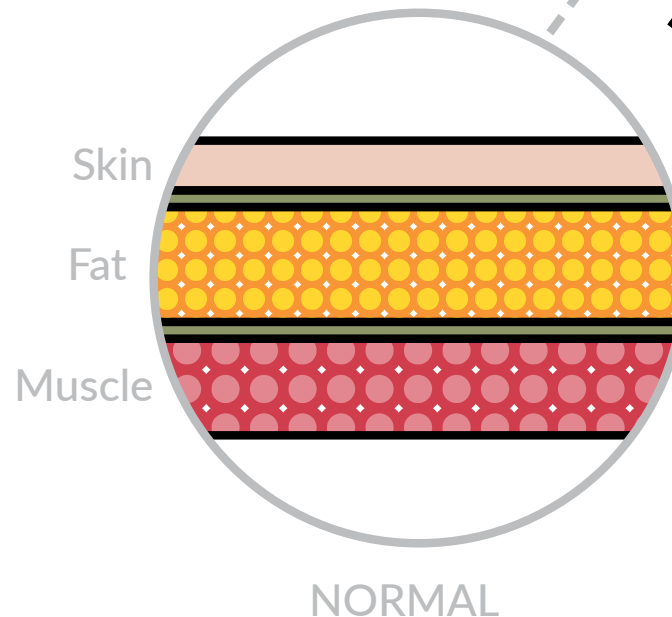
- Carbohydrates (starches and sugars) provide a readily available energy source
- Fats also provide energy. Fat is stored in the body so it can be used as an energy reserve



CPT II deficiency and fat

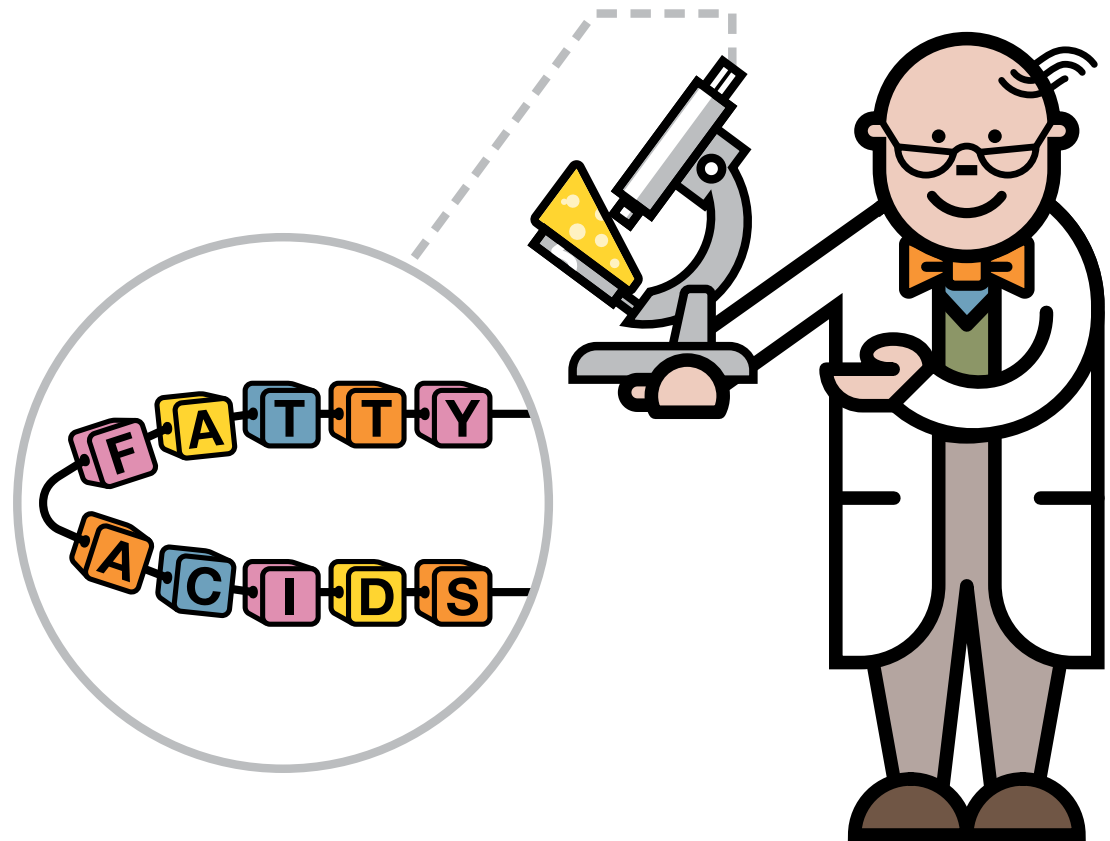
The body uses its own fat stores to provide energy when carbohydrate is depleted.

In CPT II 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.



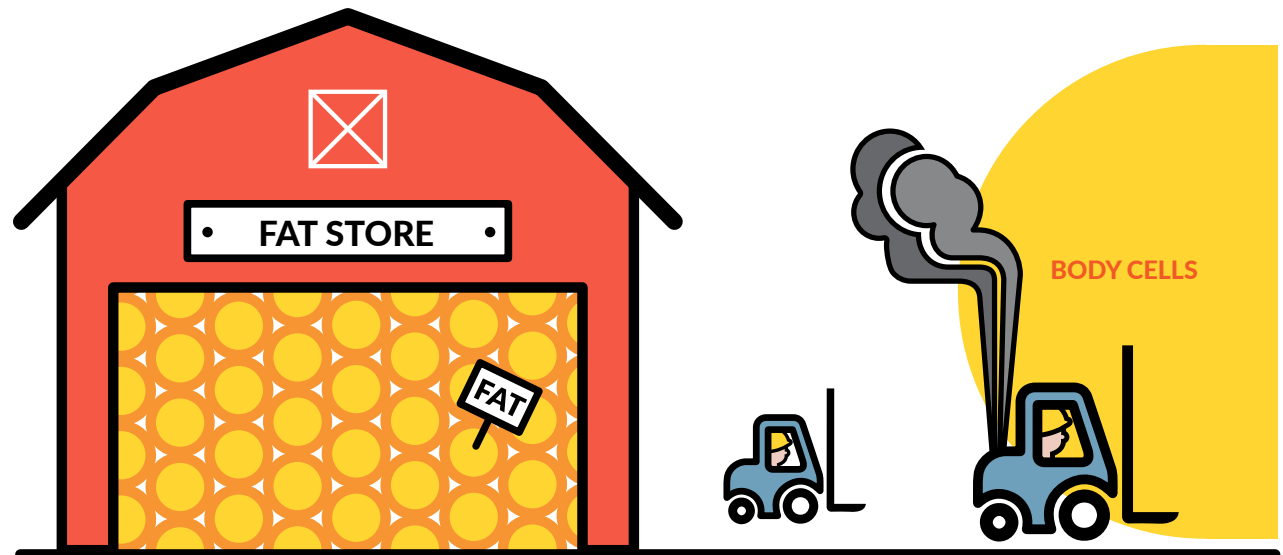
What happens in CPT II deficiency?

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

This **stops** 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.

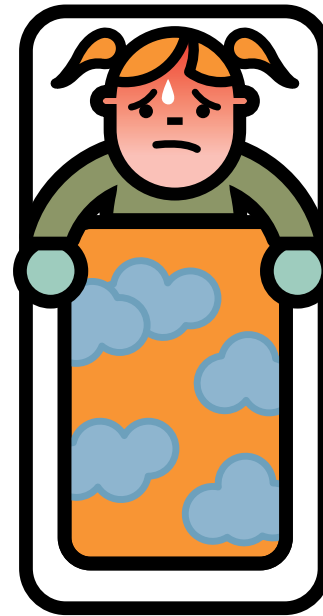
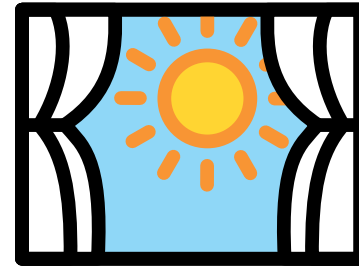


What can go wrong in CPT II deficiency in infants?

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
- heart problems, irregular heartbeat
- liver problems



What can go wrong in CPT II 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.

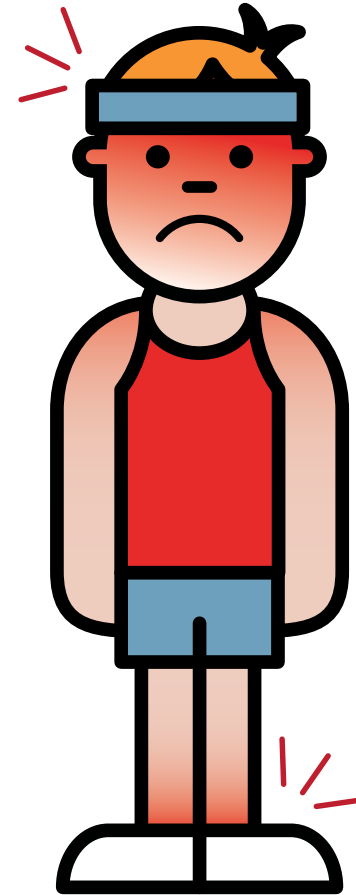
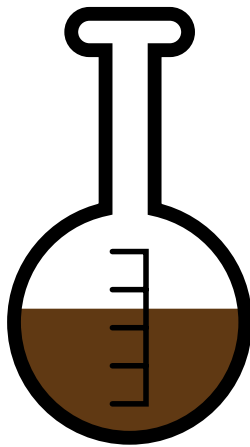
However, please remember, this can all be prevented with timely and correct management.



Some children may be diagnosed in early or later childhood

Signs and symptoms:

- Low blood sugars
- Muscle pain especially with exercise
- Reddish-brown coloured urine



Metabolic crisis

- A metabolic crisis triggers the CPT II 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 II deficiency diagnosed?

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

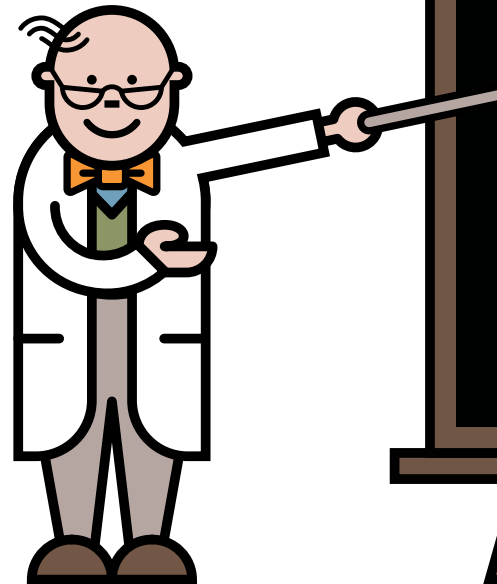
The diagnosis is confirmed by finding mutations in the CPT II gene.



How is CPT II 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**.

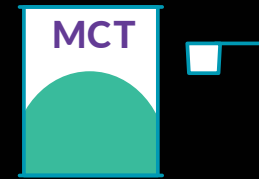


SAFE FASTING TIMES

- The safe fasting time varies for each baby with CPT II 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 II deficiency managed day to day?

A low fat infant feed with medium chain triglycerides (MCT) may be recommended by your metabolic team



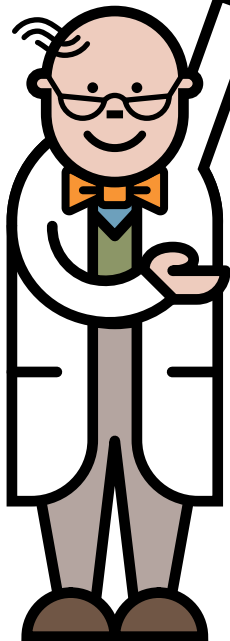
Low fat diet



MCT oil for cooking may be used

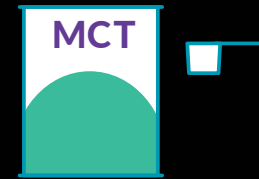


Regular mealtimes as recommended by your metabolic team



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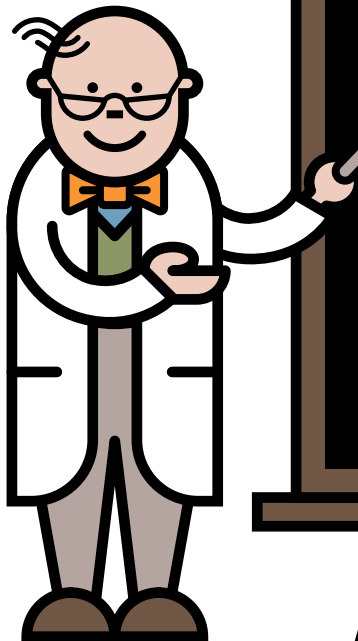
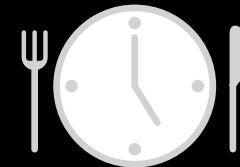
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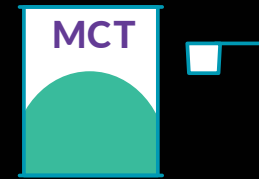


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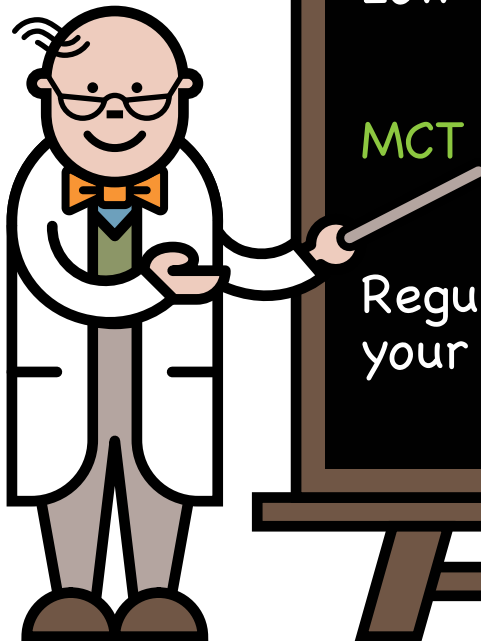
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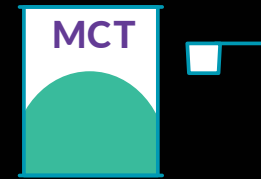


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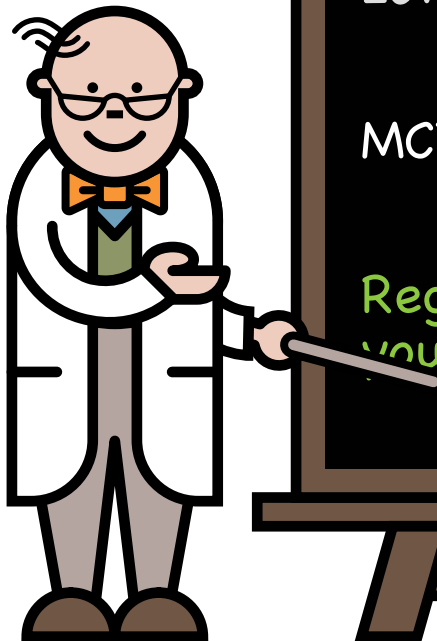
Low fat diet



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Regular mealtimes as recommended by your metabolic team



CPT II deficiency and fat

The diet needs to be low in fat.

Foods high in fat are avoided.

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

The diet needs to be low in fat.

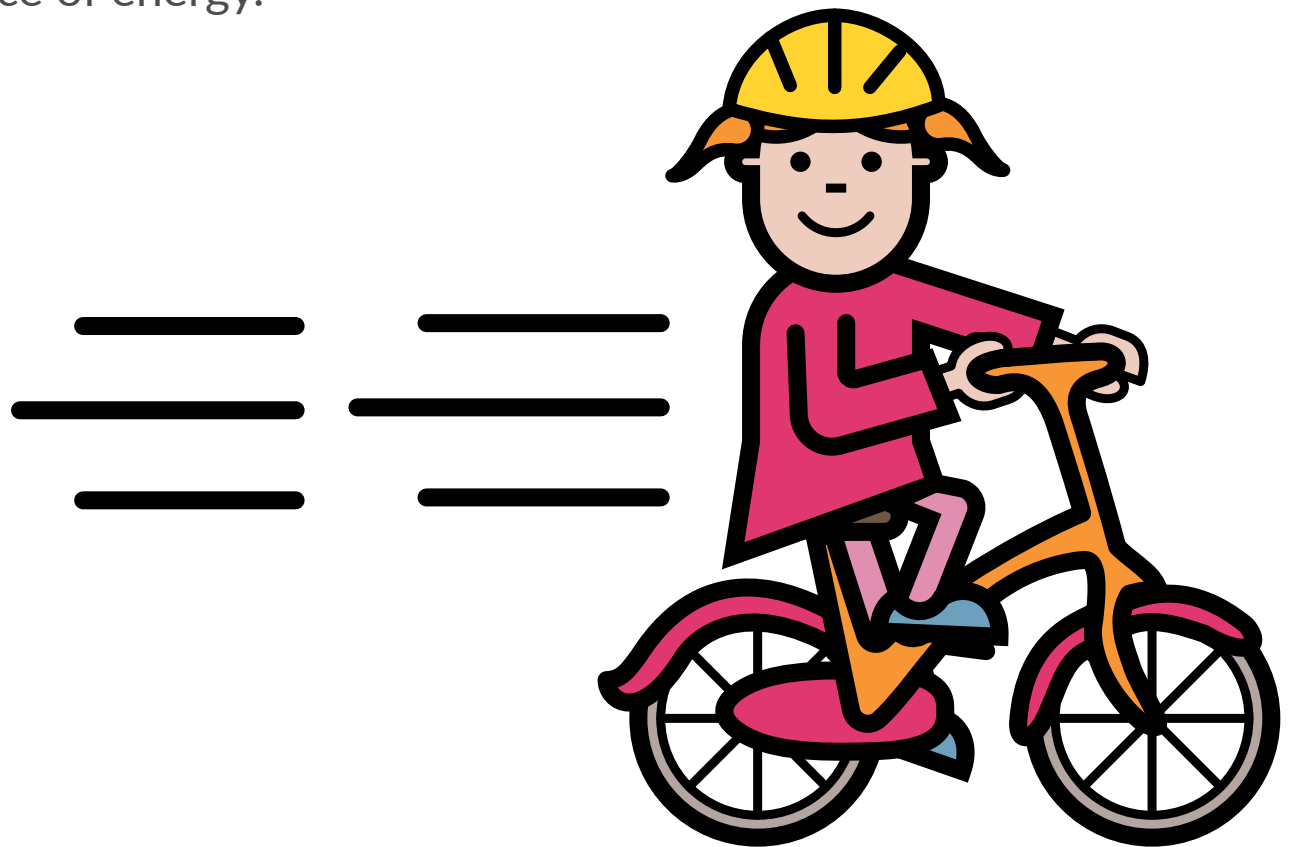
Low fat foods are permitted.

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



CPT II deficiency and exercise

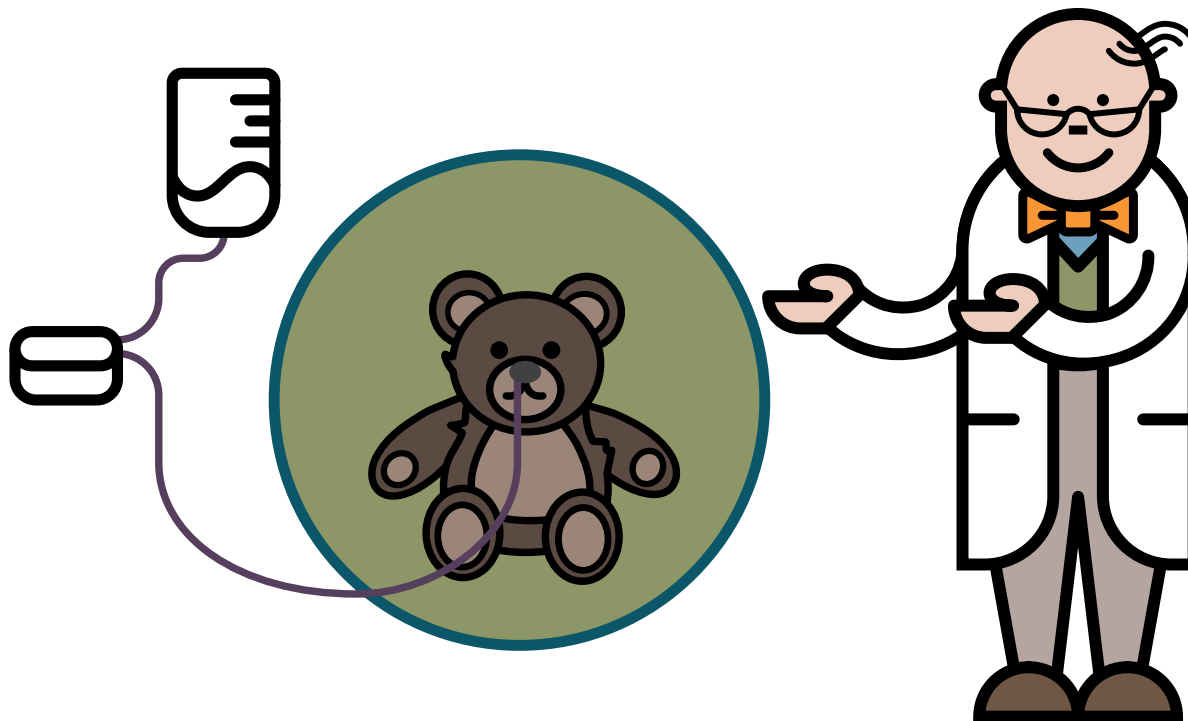
It is advisable to take a high sugar snack or drink pre exercise. This will supply an extra source of energy.



Is tube feeding needed?

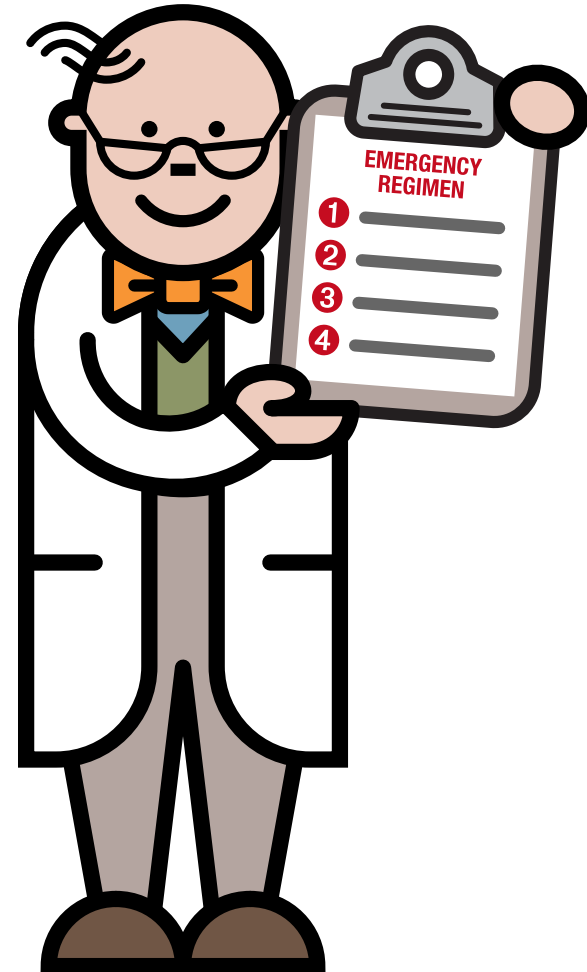
In babies with the most severe forms of CPT II deficiency, tube feeding may be necessary. This will ensure energy, nutrient and fluid needs are met.

In children not on tube feeds, a late night snack containing carbohydrate may be necessary. This is to limit fasting time.



How is CPT II 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 II deficiency managed during illness?

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



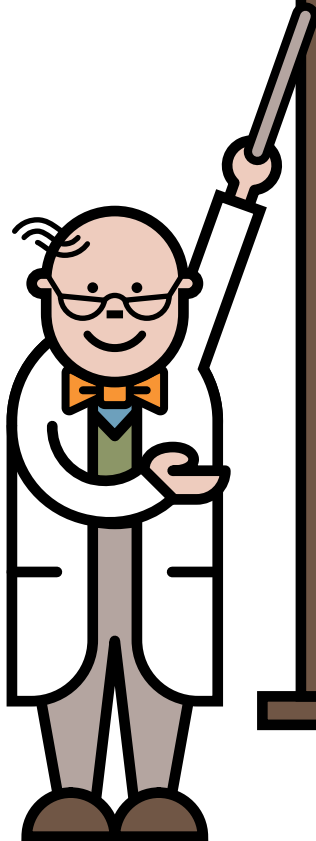
This must be given regularly
day and night



Phone your metabolic team
for help if your child is unwell



Do not use sugar-free drinks during illness



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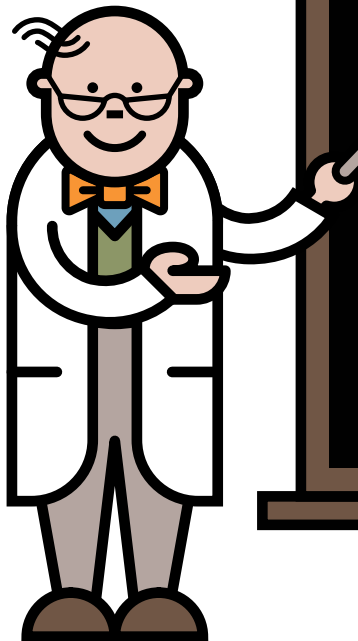
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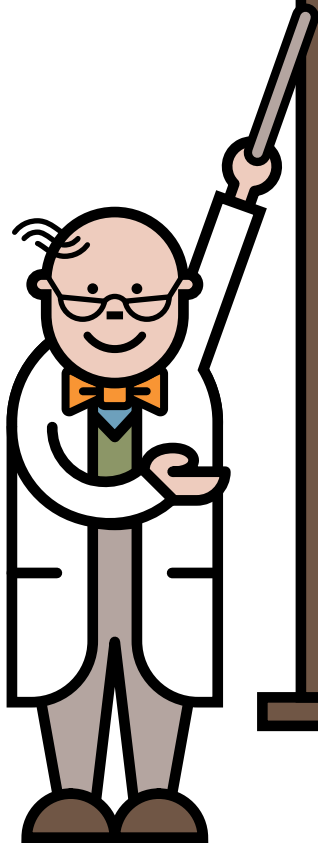
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Do not use sugar-free drinks during illness



Checklist for illness



Always take full amounts of emergency feeds as prescribed



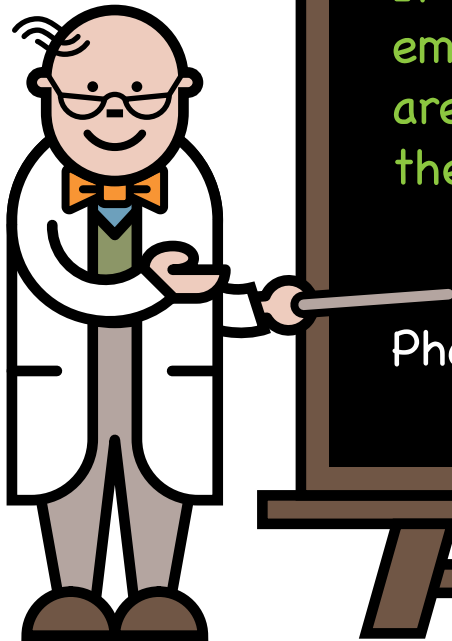
If your child vomits or refuses the emergency drinks and/or you are worried, go immediately to the hospital



Phone your metabolic team



Checklist for illness



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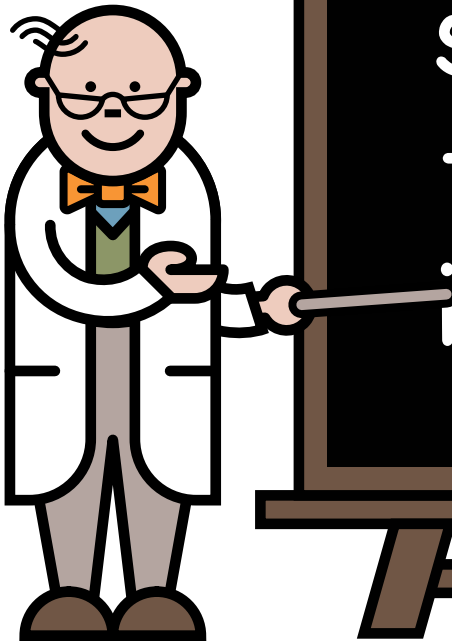


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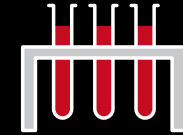
Key message

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



How is CPT II deficiency monitored?

Blood tests to check
nutrient levels



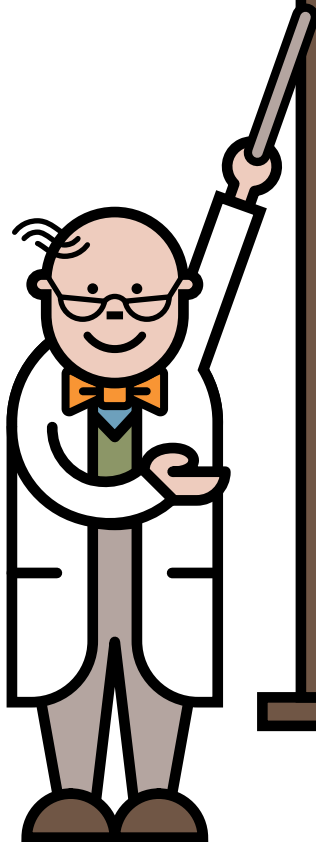
Height and weight



Medical & development checks

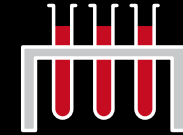


Emergency feeds are
adjusted with age



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Blood tests to check
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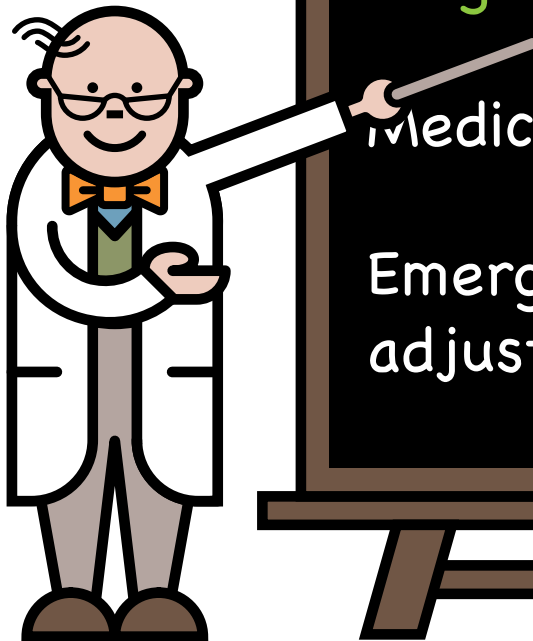
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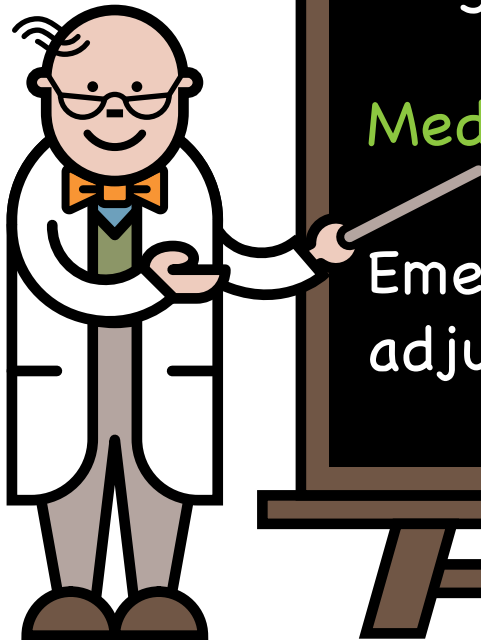
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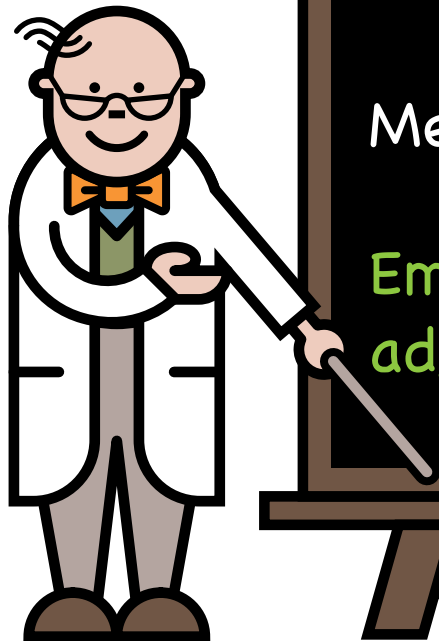
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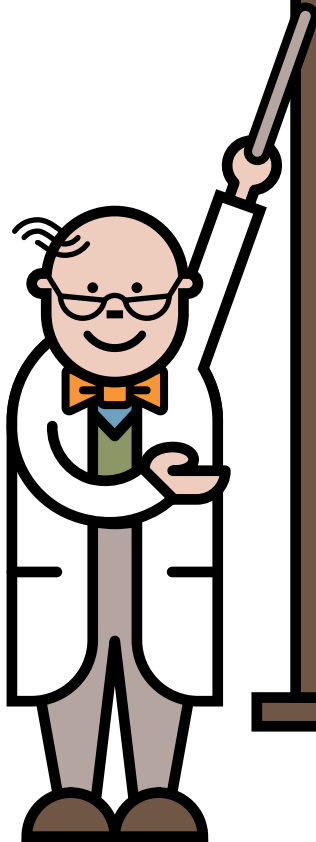
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Chromosomes, genes, mutations



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

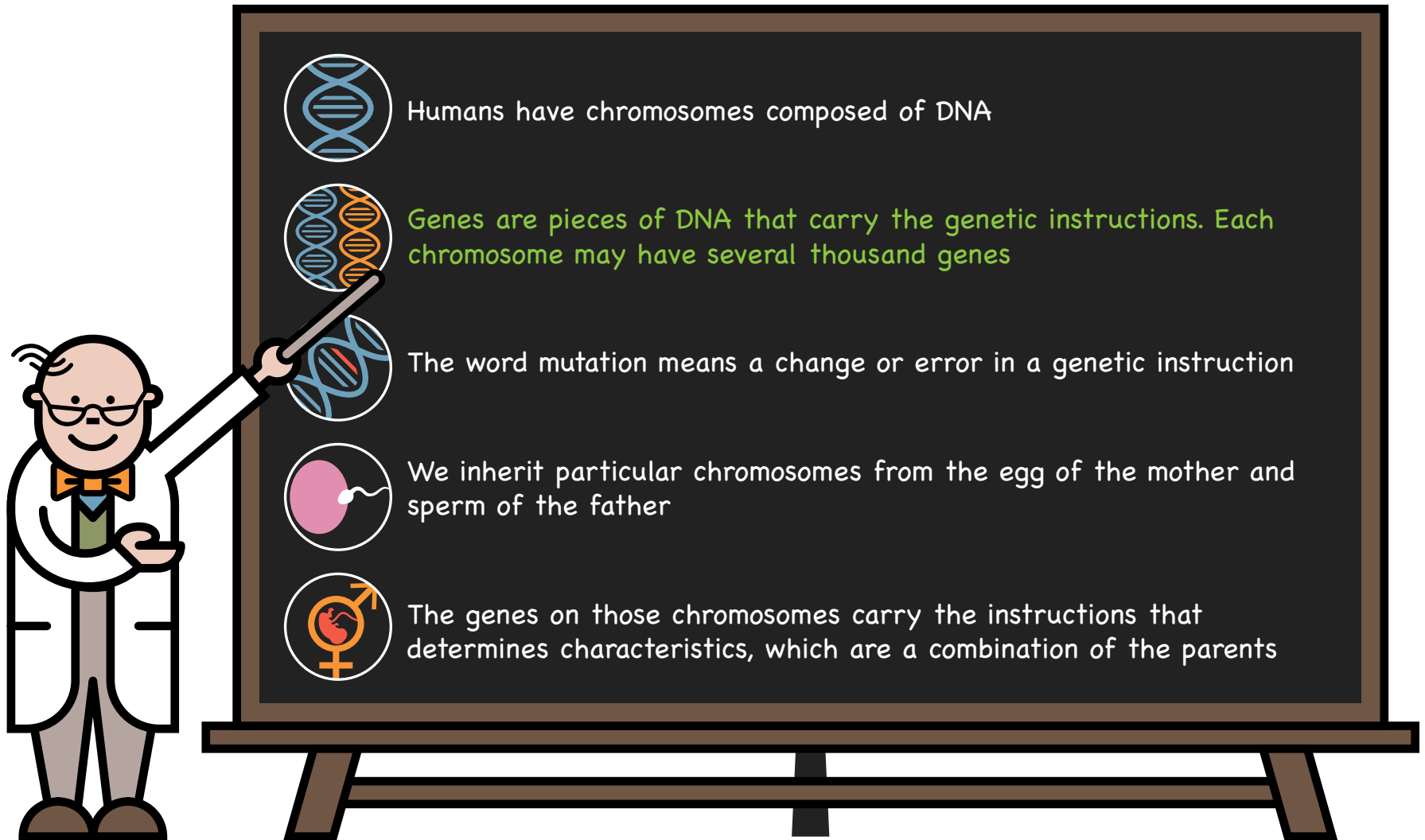


We inherit particular chromosomes from the egg of the mother and sperm of the father



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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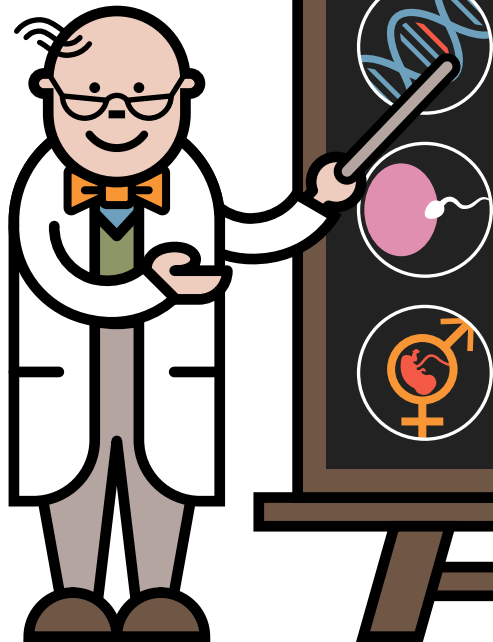
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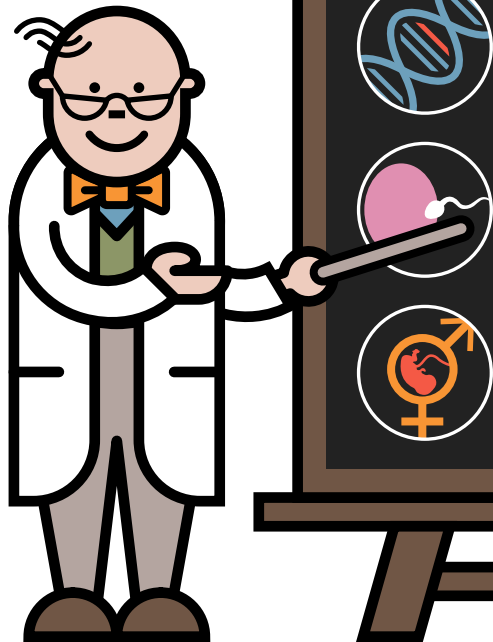


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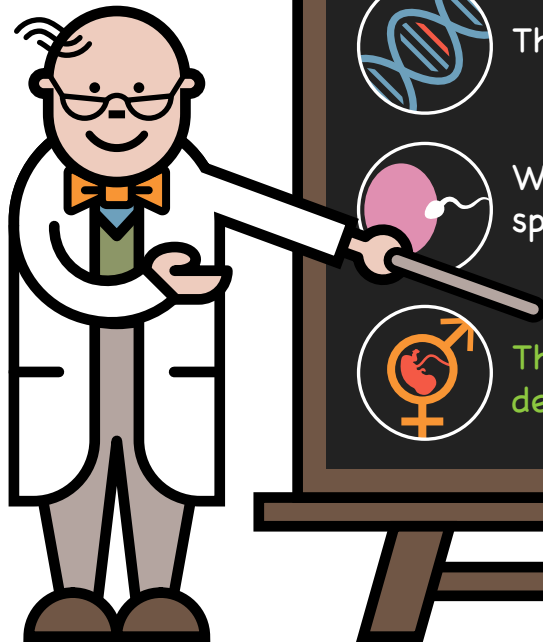


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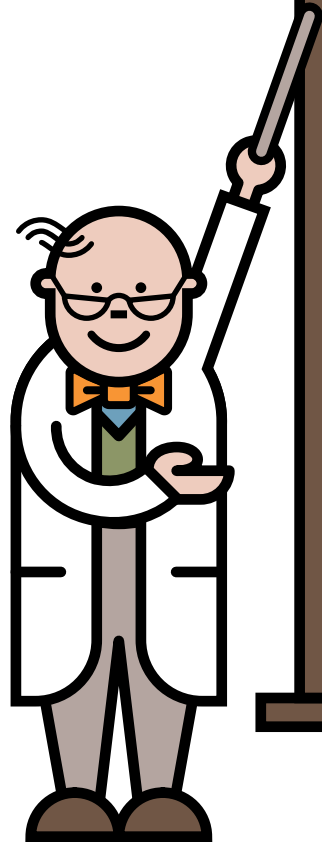


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Inheritance



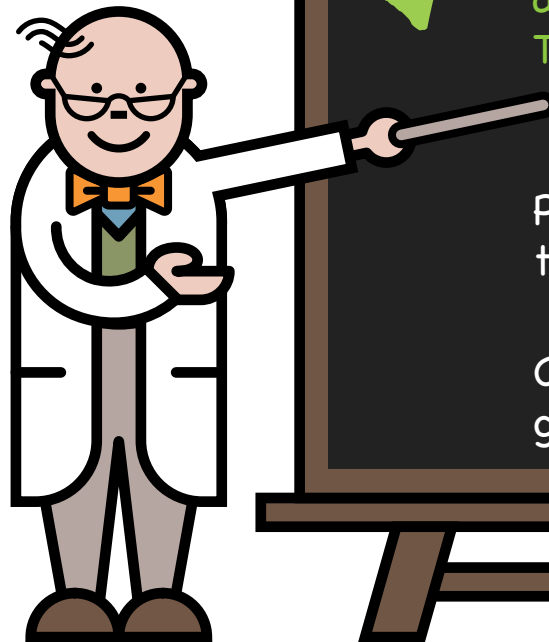
CPT II deficiency is an inherited condition. There is nothing that could have been done to prevent your baby from having CPT II deficiency

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

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

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

Inheritance



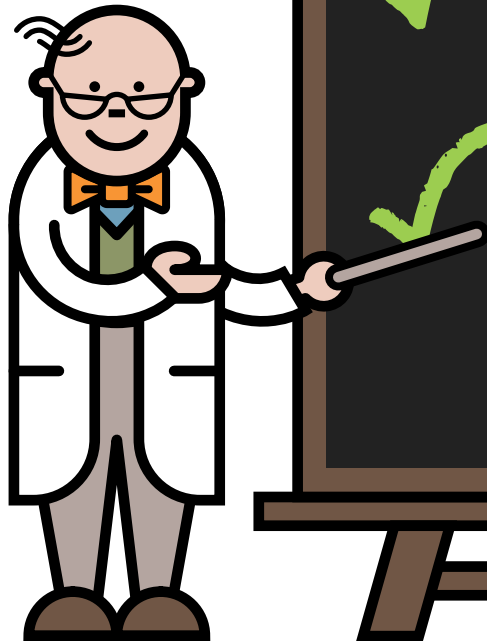
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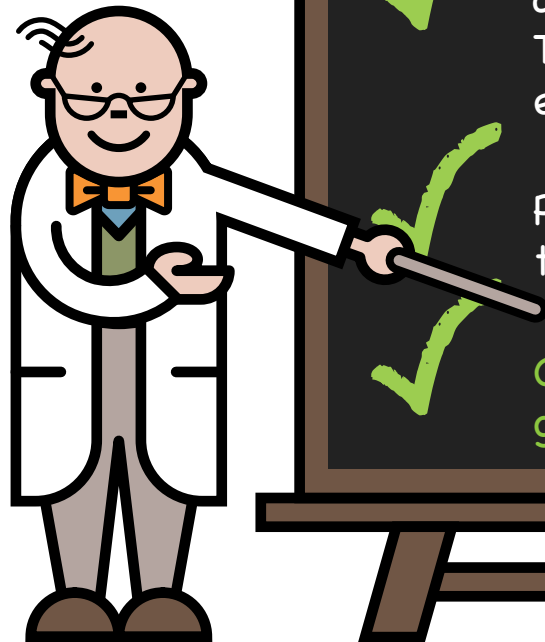
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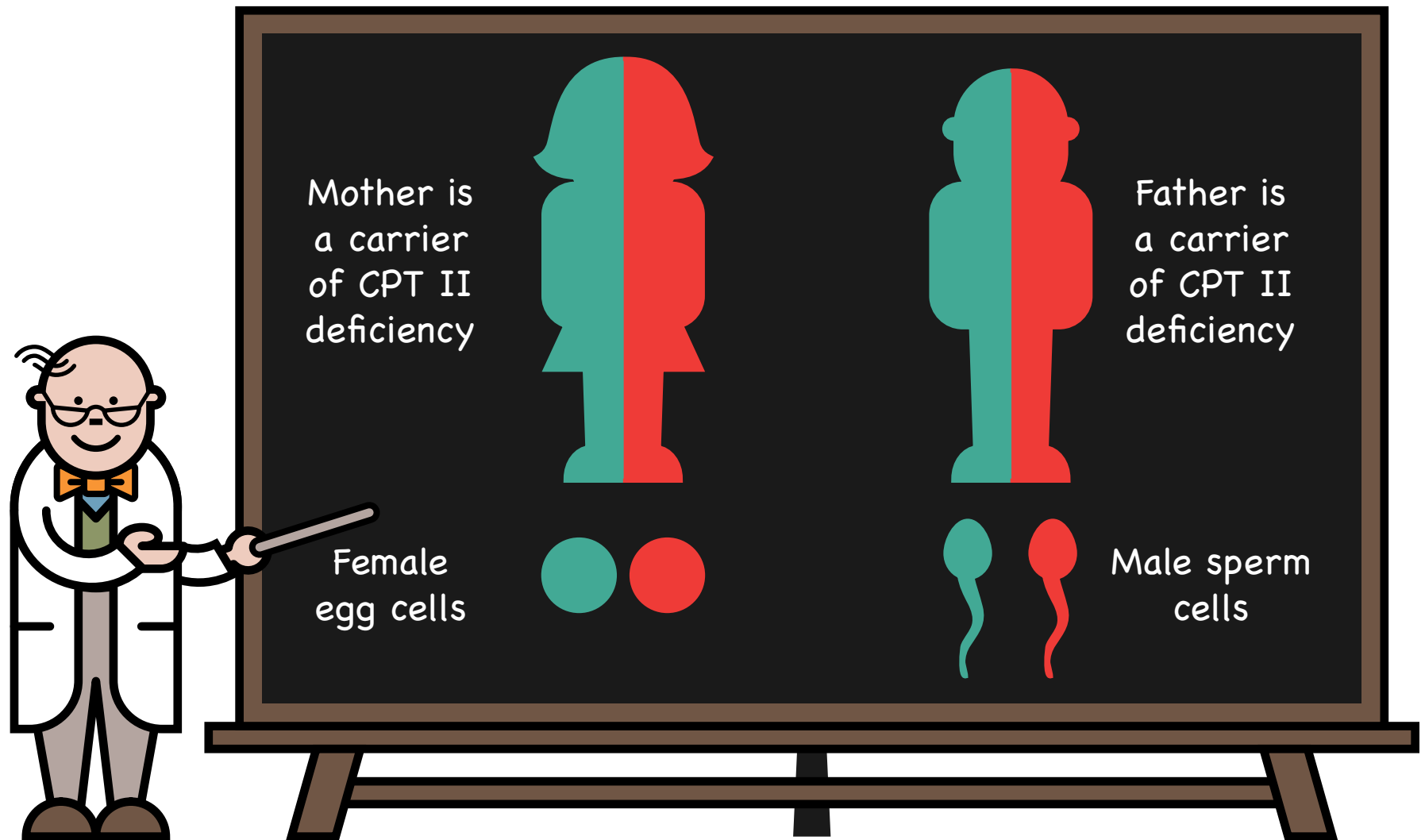
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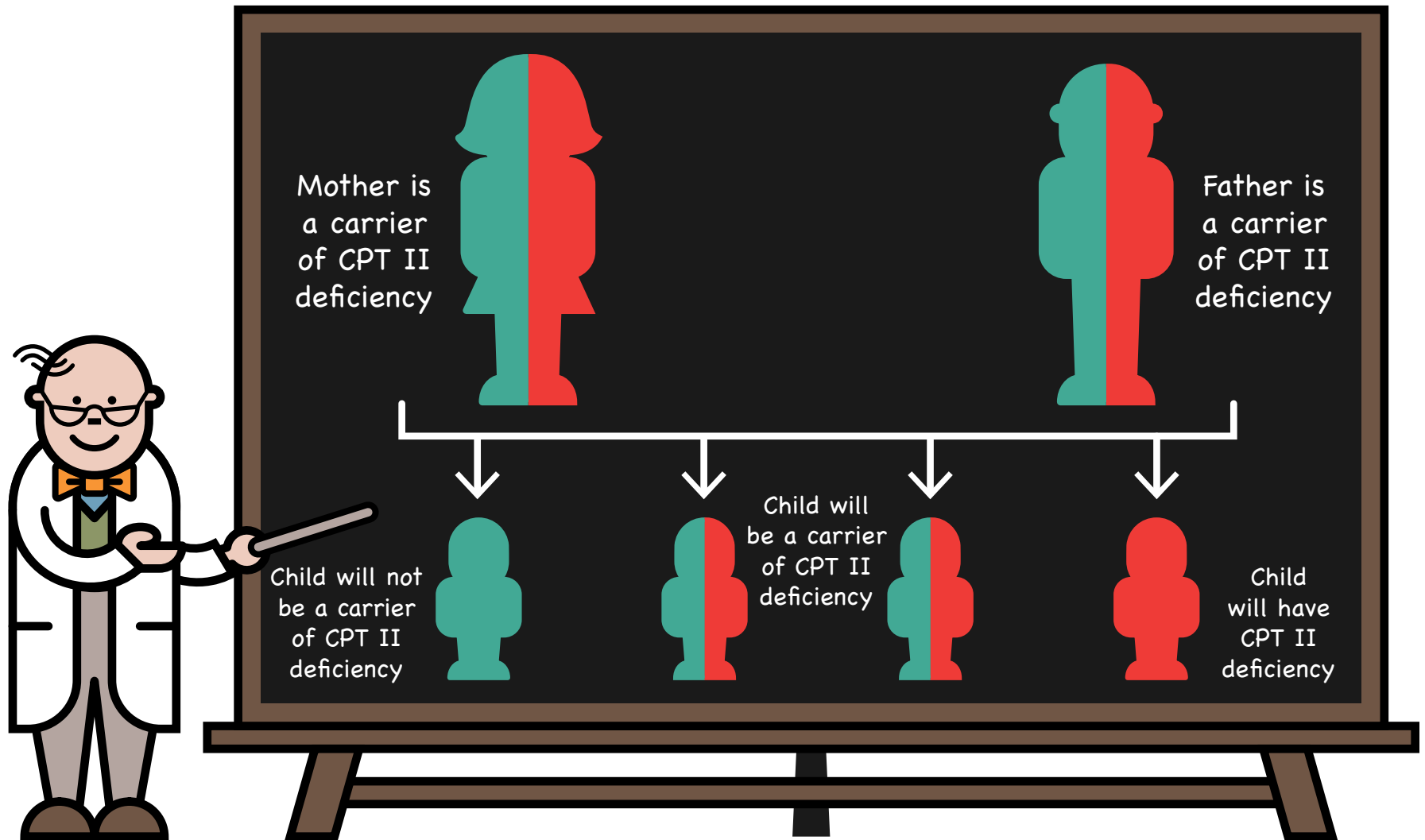
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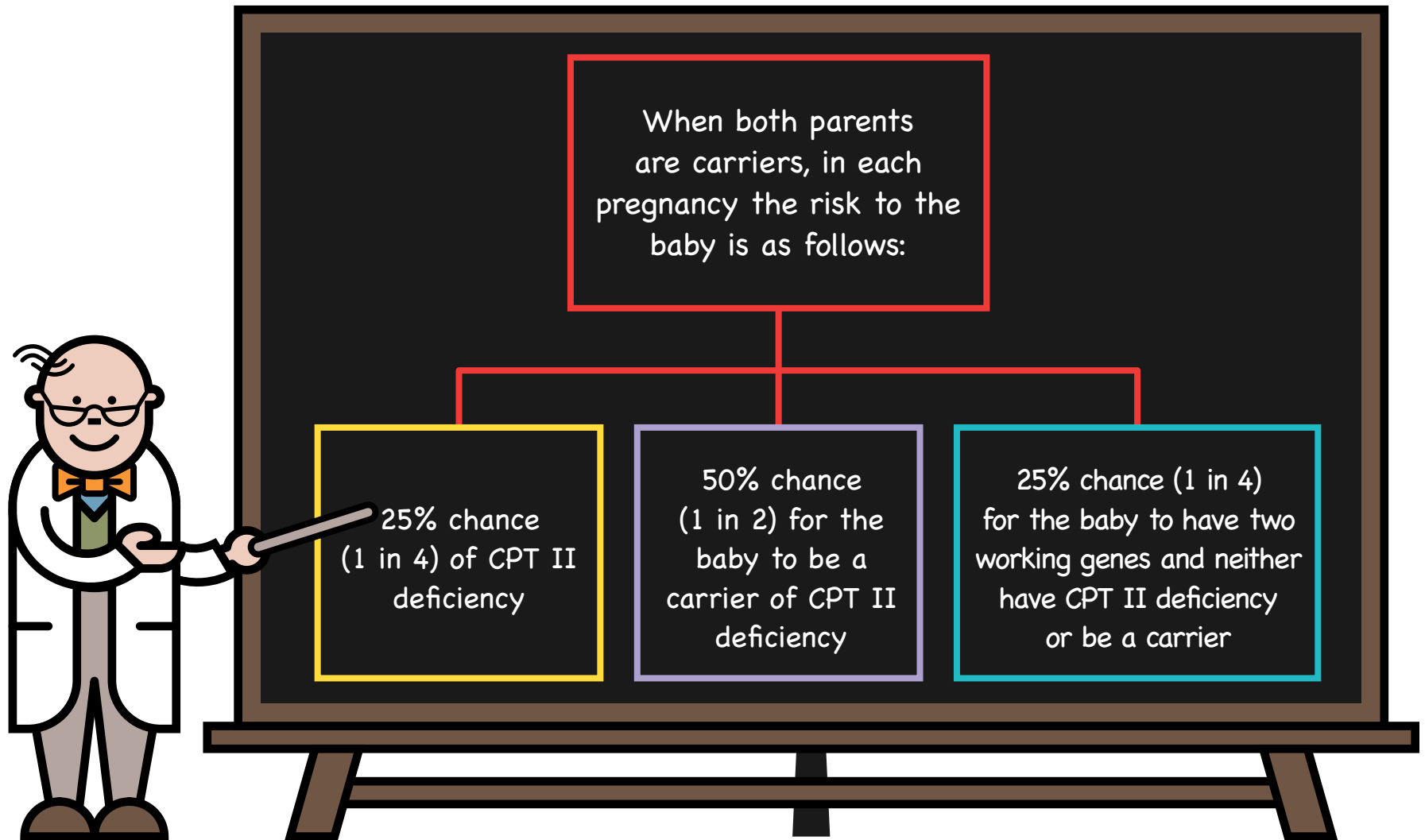
Inheritance – Autosomal recessive (carriers of CPT II deficiency)



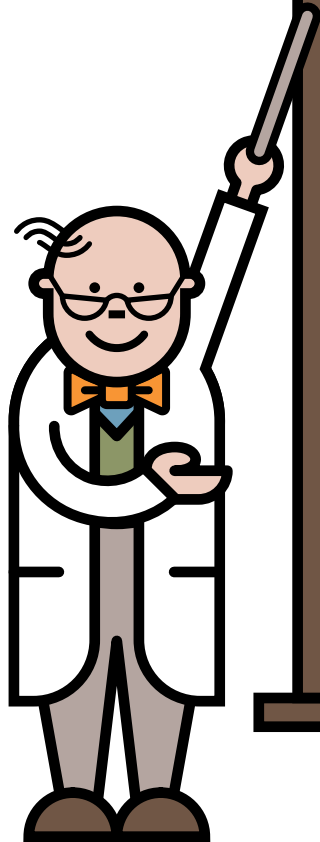
Inheritance – Autosomal recessive – possible combinations



Future pregnancies



Take home messages



✓ CPT II deficiency is a serious inherited metabolic condition that causes life threatening symptoms if left untreated

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

Children with severe CPT II deficiency are managed with a low fat diet and regular meals

People with CPT II deficiency should lead normal, healthy and active lives

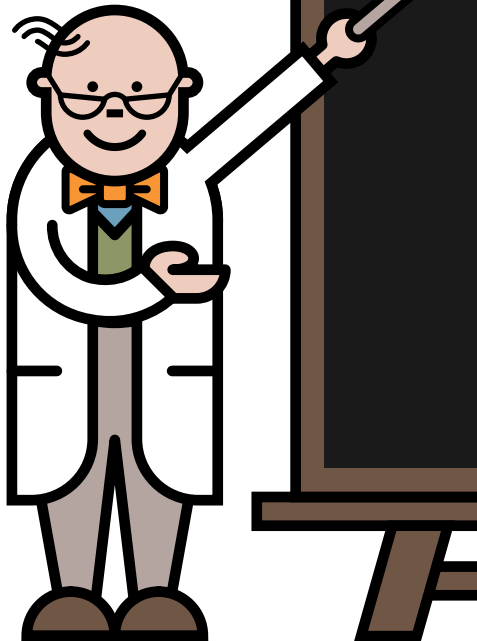
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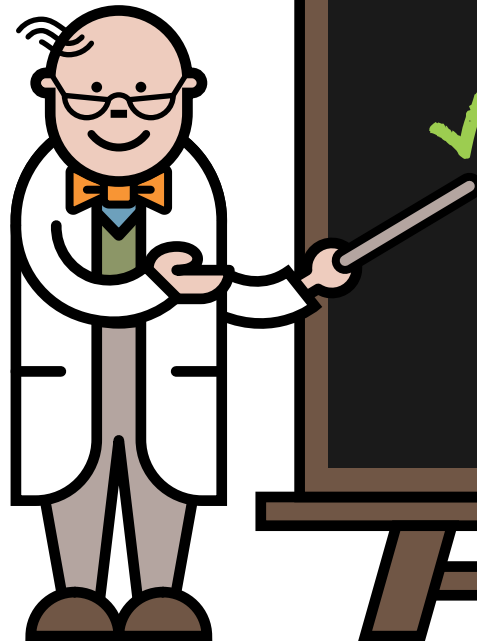
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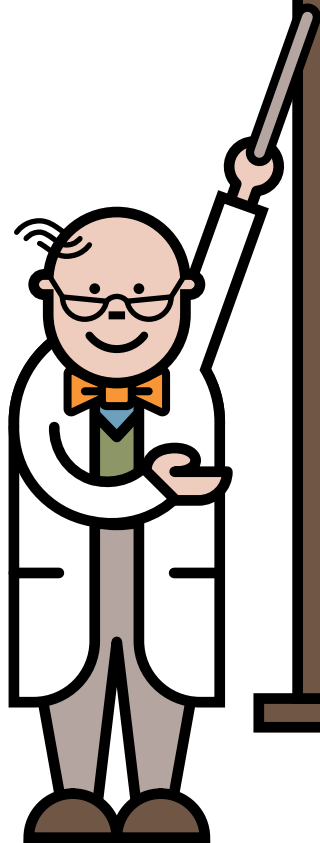
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Helpful hints



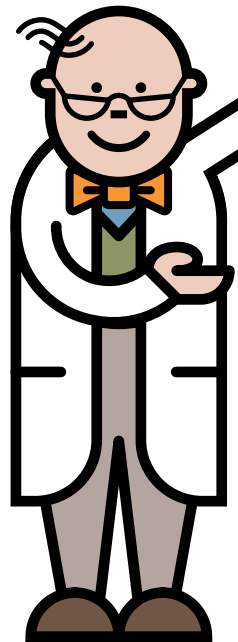
Always ensure you have a good supply of your emergency glucose polymer powder and it is in date

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

Helpful hints



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

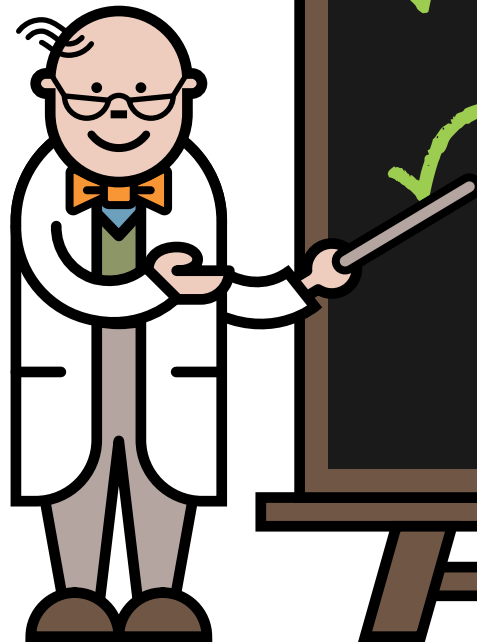


Special feeds and dietary products are prescribed by your GP and you obtain them from your pharmacy or home delivery

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

Helpful hints



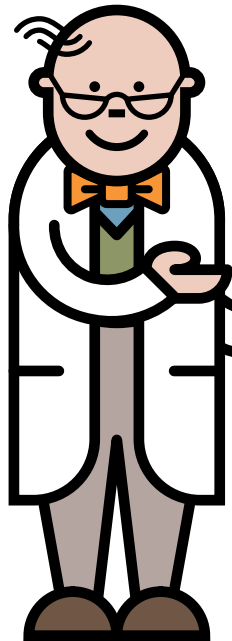
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Who's who

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

BIMDG

British Inherited Metabolic Diseases Group



**NUTRICIA**

**METABOLIC
SUPPORT UK**

Your rare condition.
Our common fight.

www.bimdg.org.uk

www.nutricia.co.uk

www.metabolicsupportuk.org