

# TEMPLE



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

ADAPTED BY THE DIETITIANS GROUP

**BIMDG**

British Inherited Metabolic Diseases Group



## CACT 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).

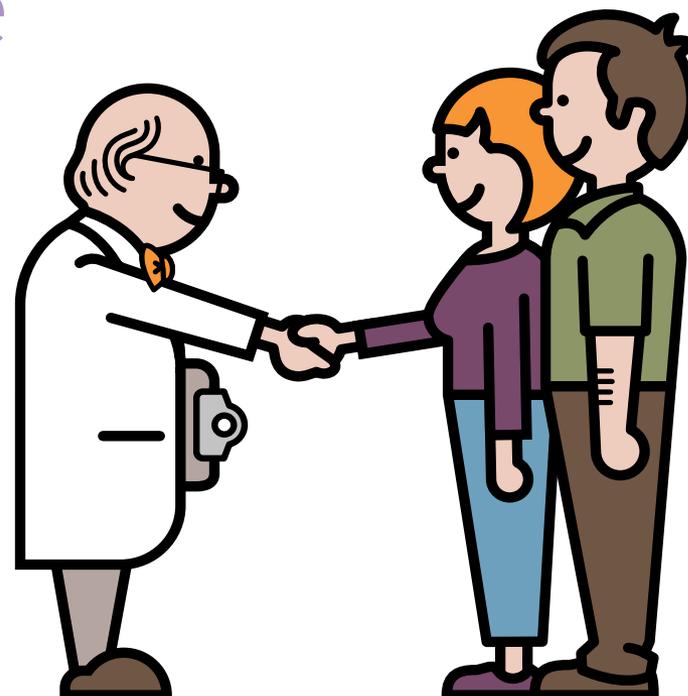
The team are Rachel Skeath, Karen van Wyk, Pat Portnoi and Anita MacDonald. The group is facilitated by Heidi Chan from Nutricia.

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 Acylcarnitine Translocase Deficiency

Information for families following  
a new diagnosis



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**TEMPLE**



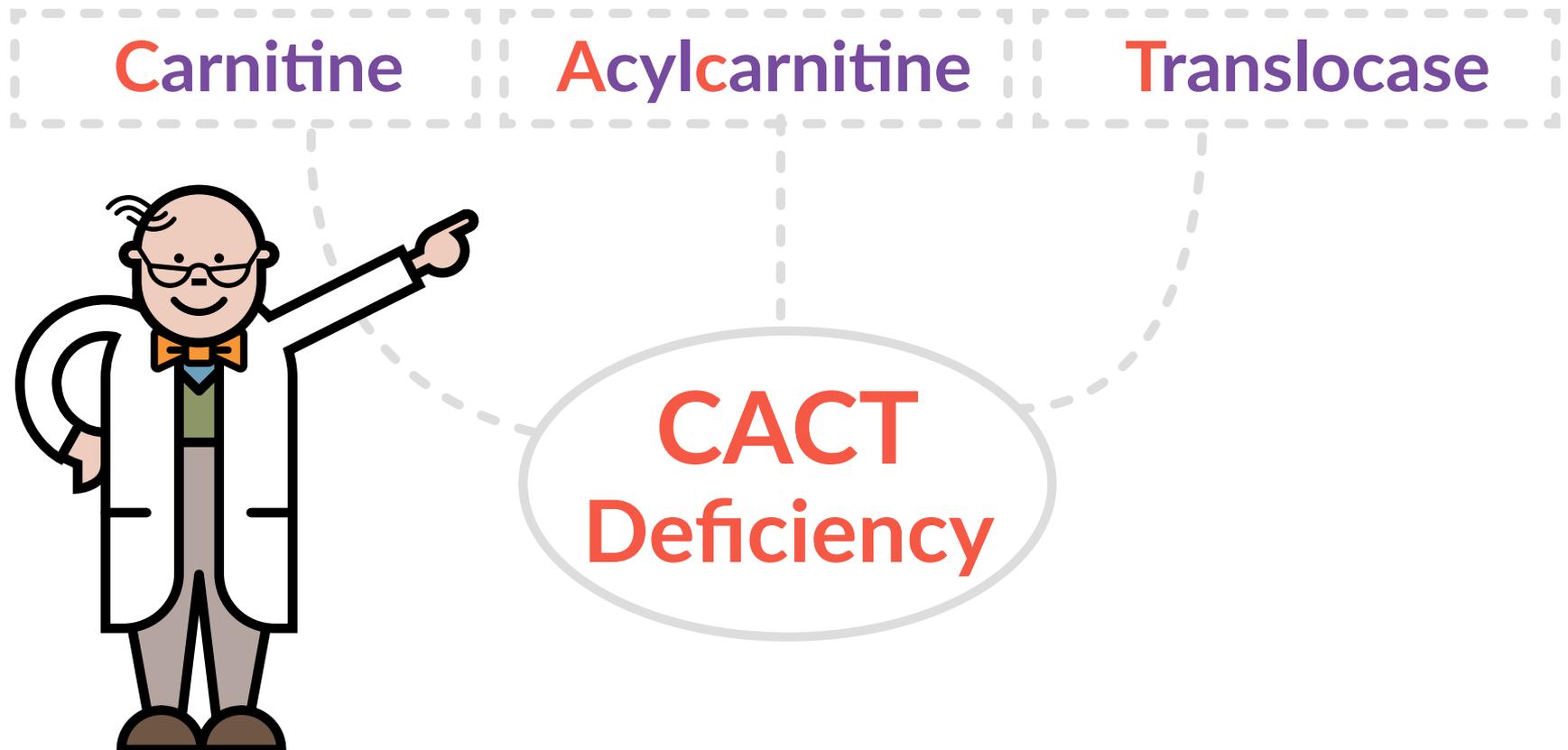
Tools Enabling Metabolic Parents LEarning

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# What is CACT deficiency?

CACT stands for Carnitine Acylcarnitine Translocase deficiency

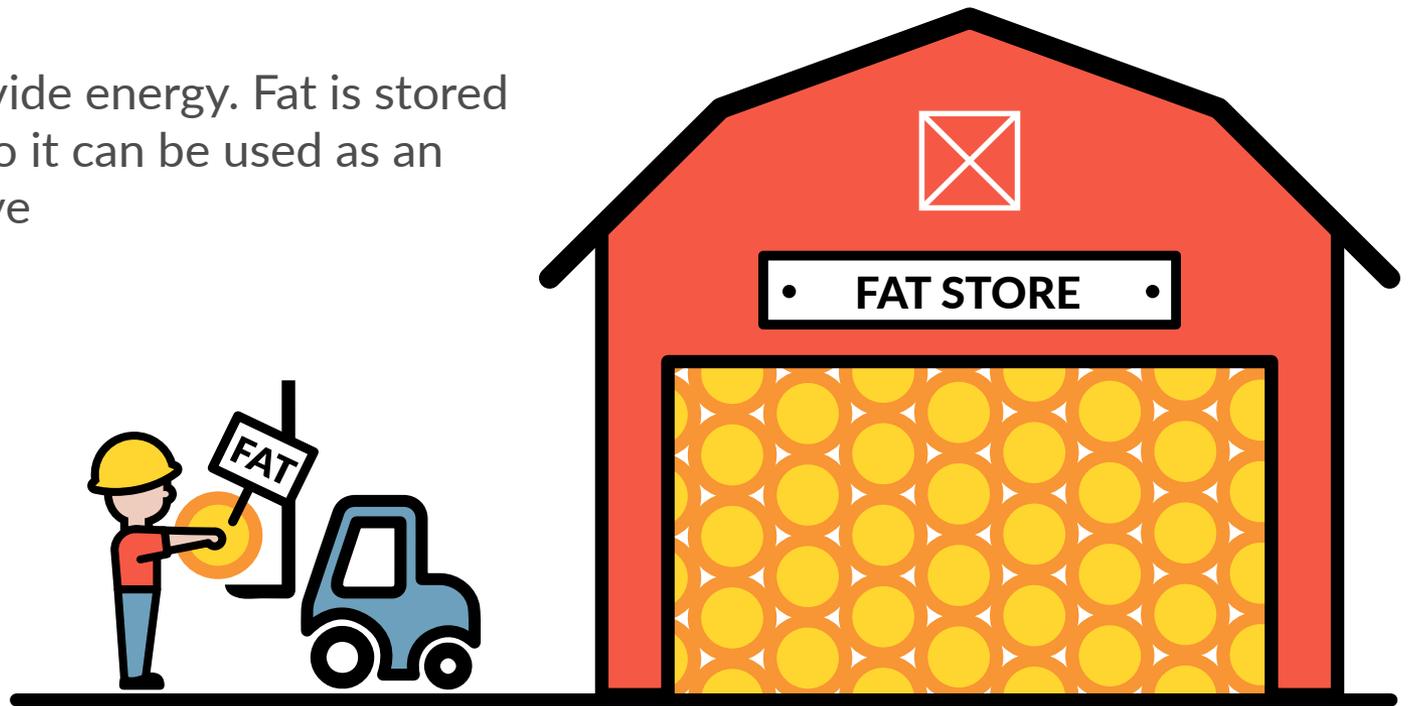
It is an inherited metabolic condition



# Which foods supply the body with energy?

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

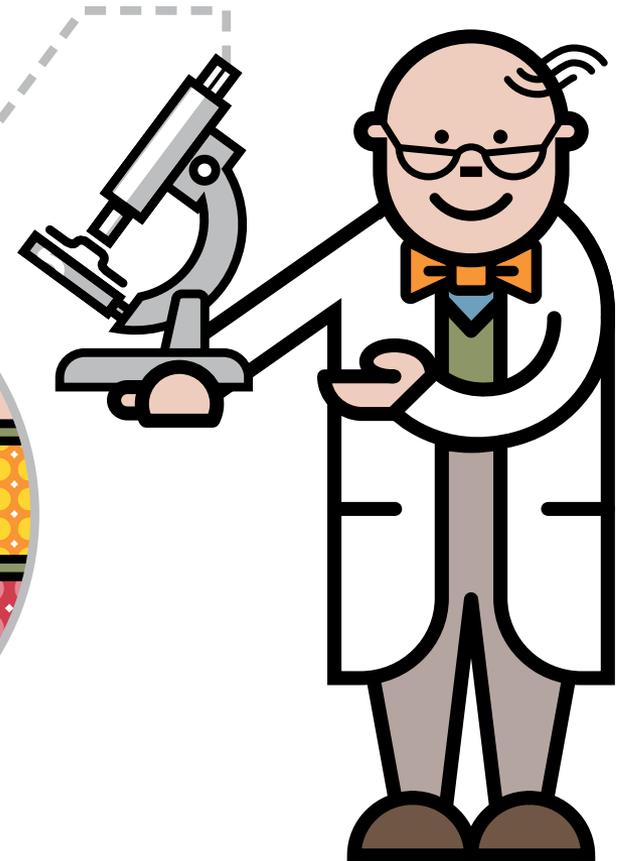
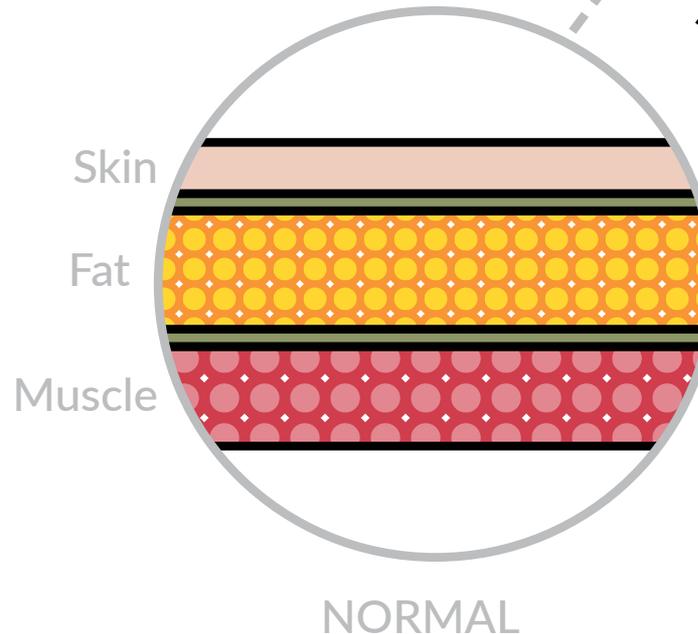
- Carbohydrates (starches and sugars) provide a readily available energy source
- Fats also provide energy. Fat is stored in the body so it can be used as an energy reserve



# CACT deficiency and fat

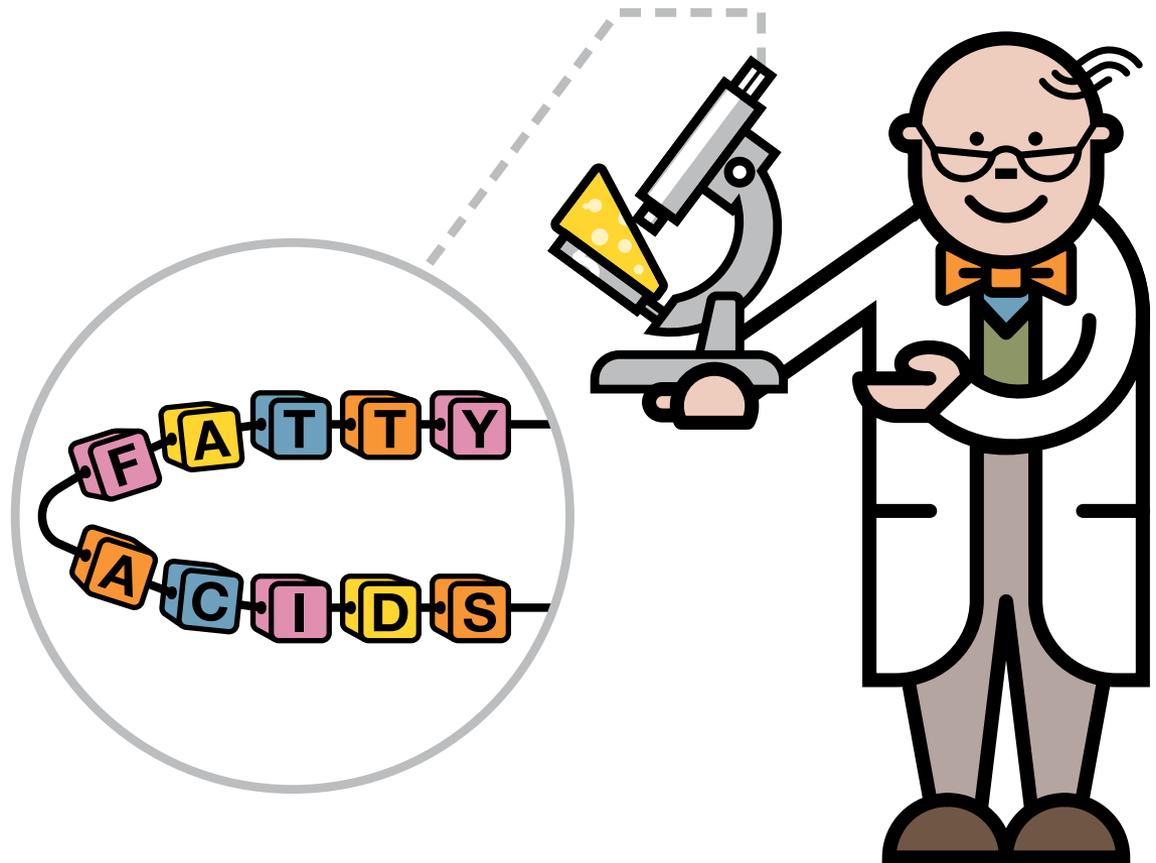
The body uses its own fat stores to provide energy when carbohydrate is depleted.

In CACT 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.



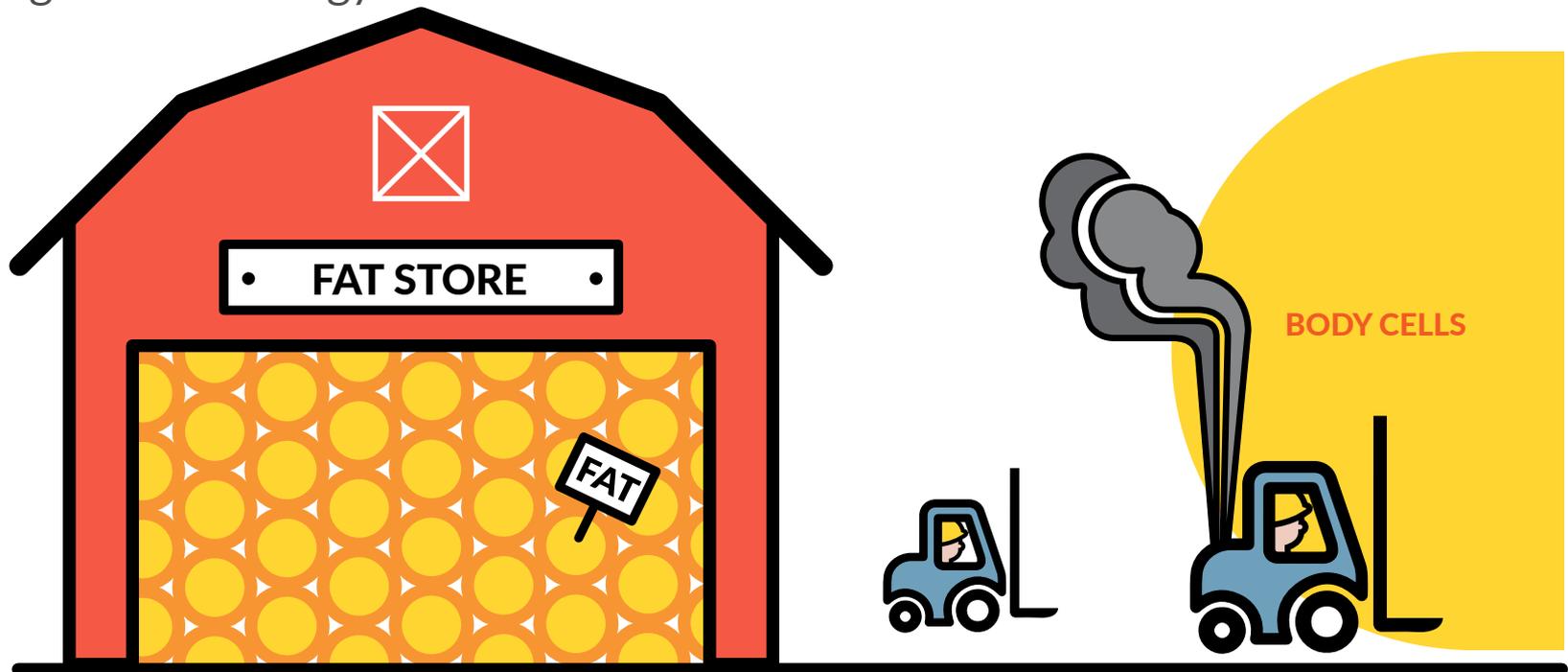
# What happens in CACT deficiency?

In CACT 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.

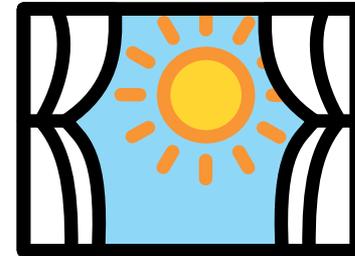


# What can go wrong in CACT deficiency in infants?

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

Symptoms include:

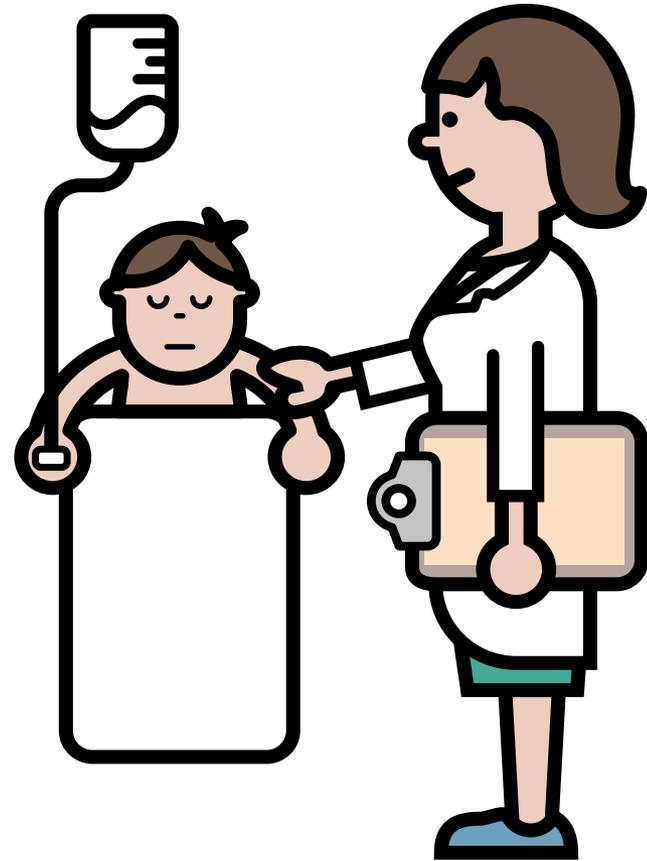
- low blood sugar
- floppiness
- seizures
- heart problems
- irregular heartbeat
- muscle weakness
- liver problems



# What can go wrong in CACT 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.

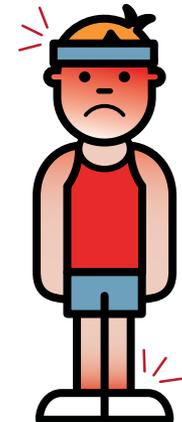
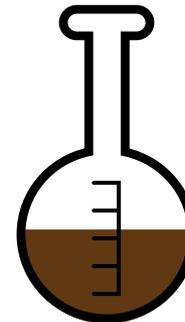
**However, please remember, this can all be prevented with timely and correct management.**



# Some children may be diagnosed in early or later childhood

Signs and symptoms:

- Low blood sugars
- It may affect development and learning
- Exercise may cause muscle pain which may lead to reddish brown coloured urine
- Reddish-brown coloured urine



# Metabolic crisis

- A **metabolic crisis** triggers the CACT 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 CACT deficiency diagnosed?

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

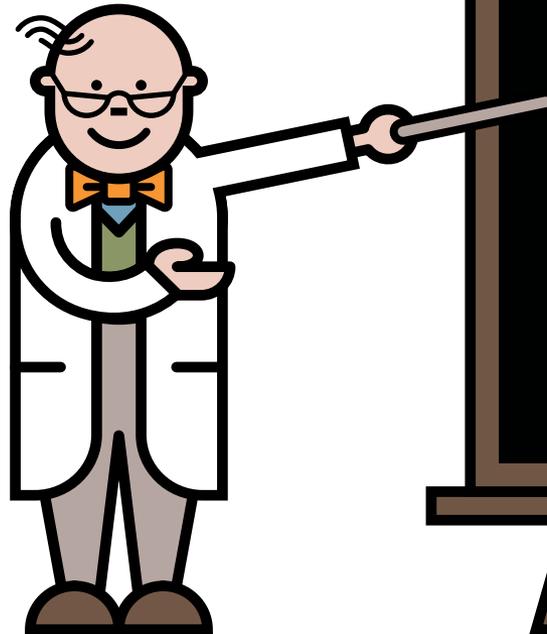
The diagnosis is confirmed by finding mutations in the CACT gene.



# How is CACT 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 CACT 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 CACT deficiency managed day to day?

A low fat infant feed with medium chain triglycerides (MCT)



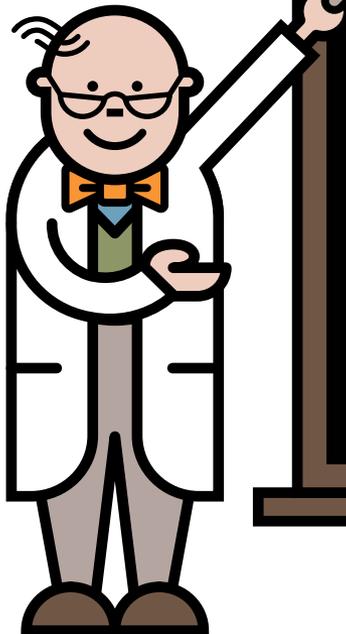
Low fat diet



MCT oil for cooking may be used

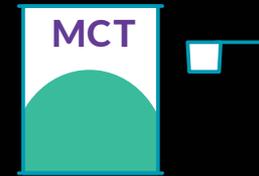


Regular mealtimes as recommended by your metabolic team



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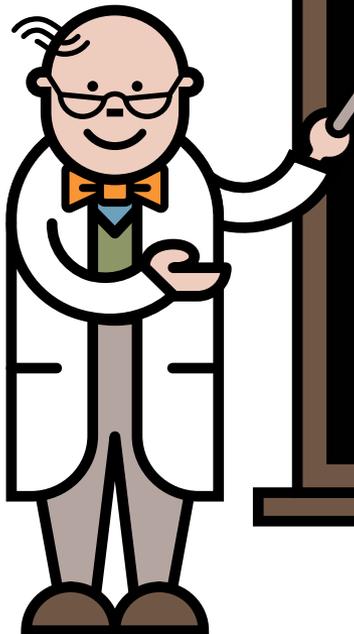
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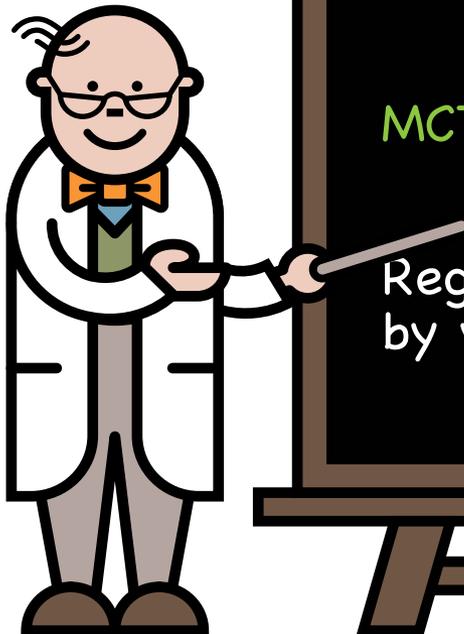
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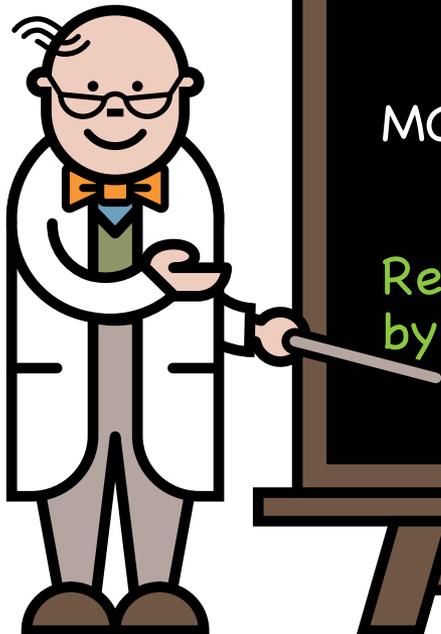
Low fat diet



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Regular mealtimes as recommended by your metabolic team



# CACT 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.

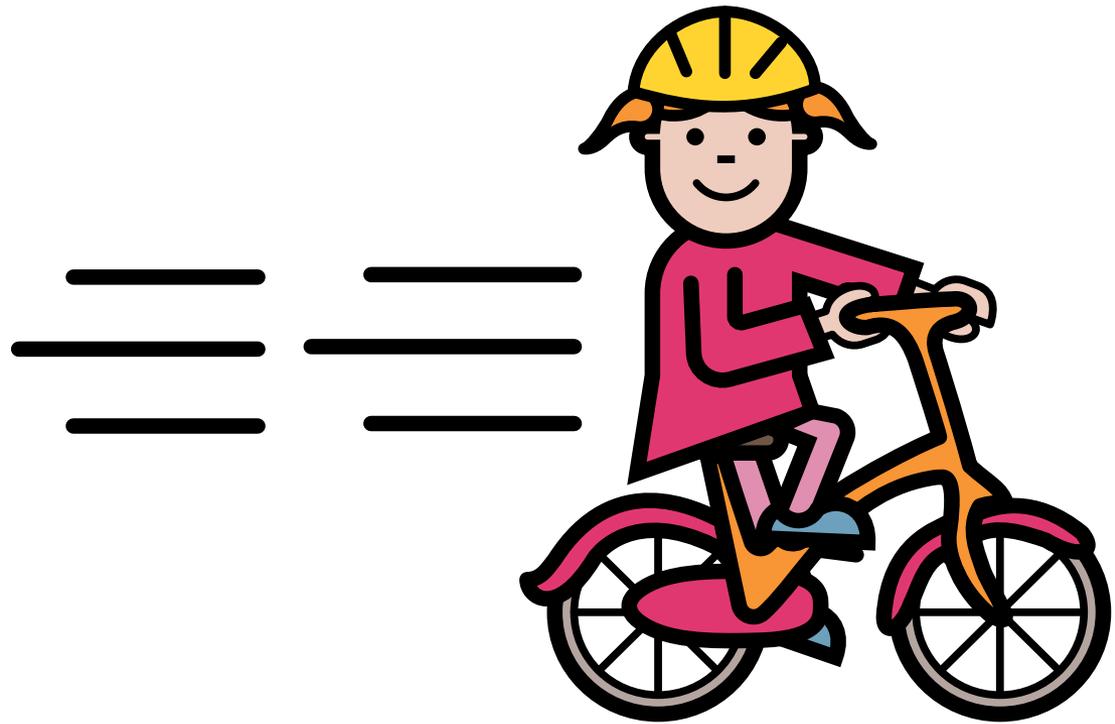
**Low fat foods are permitted.**

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



# CACT deficiency and exercise

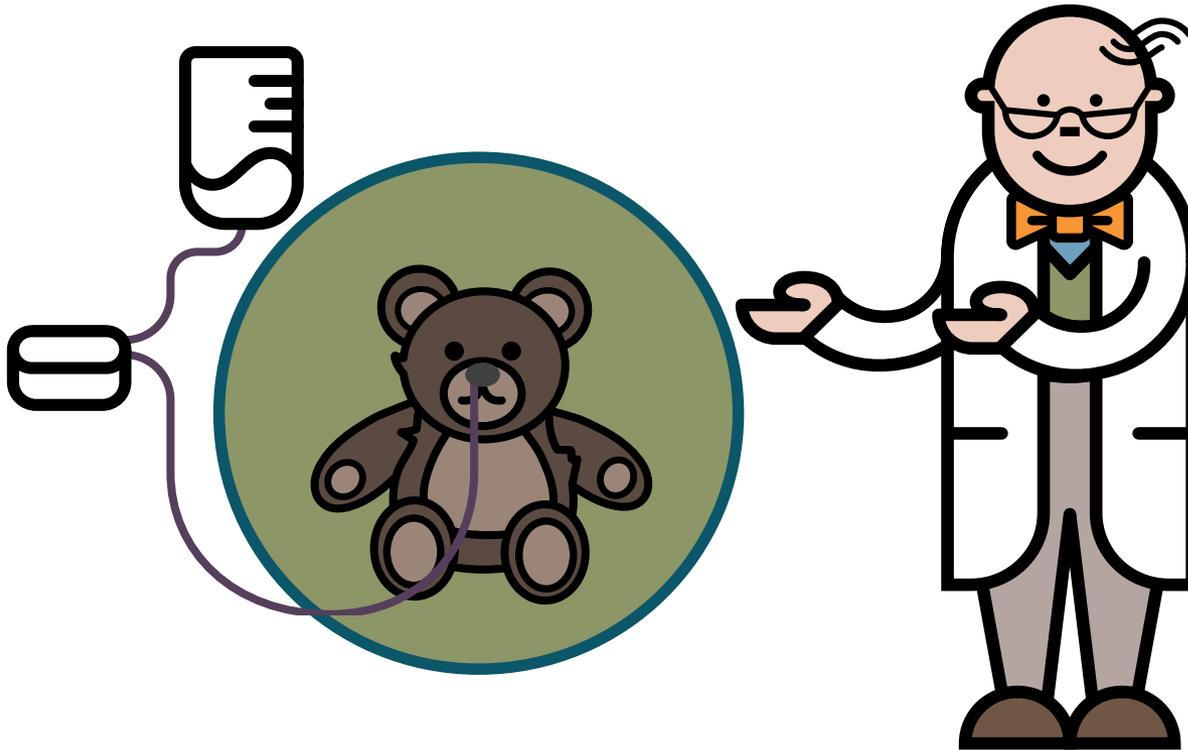
It is advisable to take a high sugar snack or drink pre exercise.  
This will supply an extra source of energy.



# Is tube feeding needed?

In babies with the most severe forms of CACT 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 CACT 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 CACT deficiency managed during illness?

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



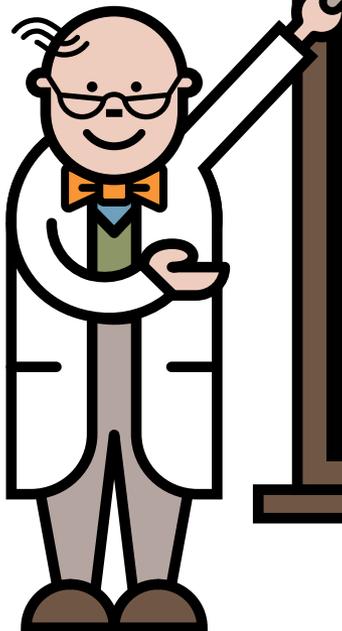
This must be given regularly  
day and night



Phone your metabolic team  
for help if your child is unwell



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



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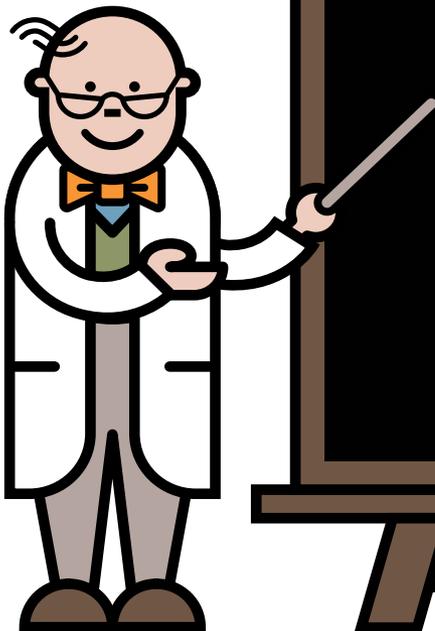
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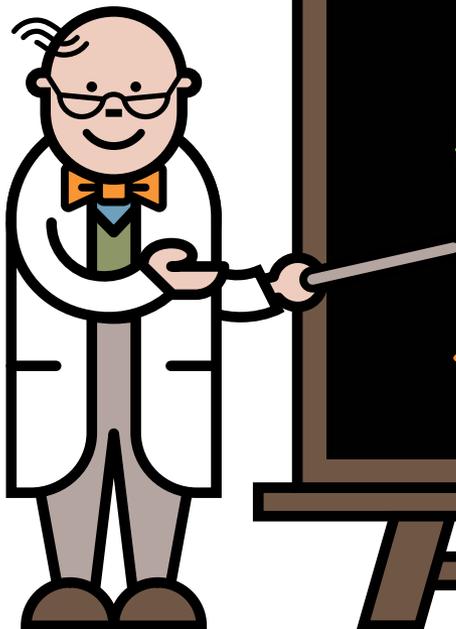
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# Checklist for illness

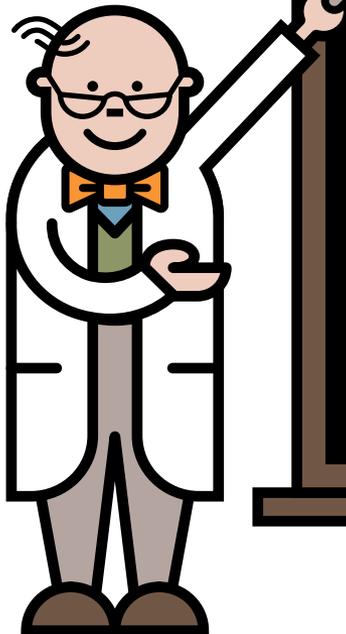
Always take full amounts of emergency feeds as prescribed



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



Phone your metabolic team



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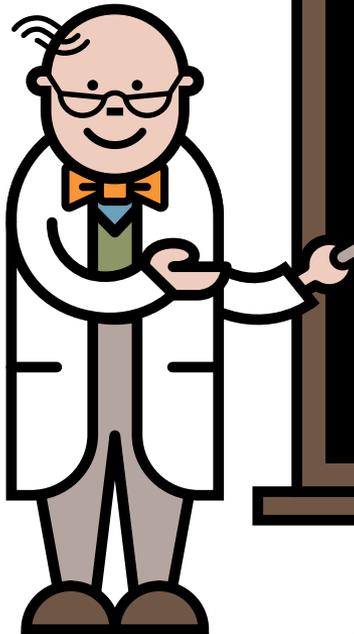
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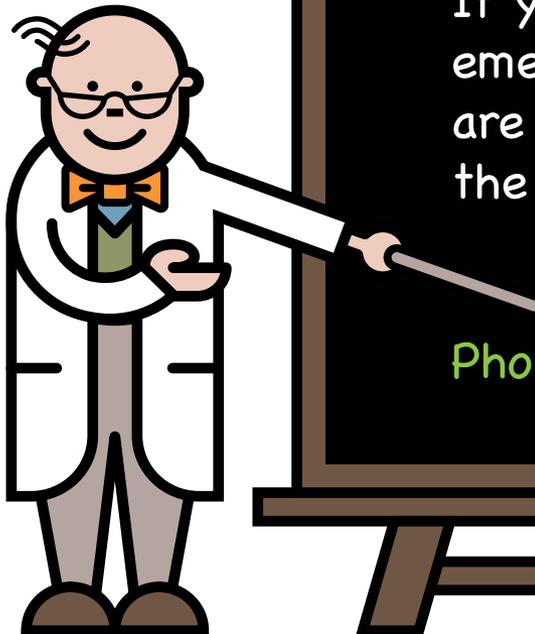
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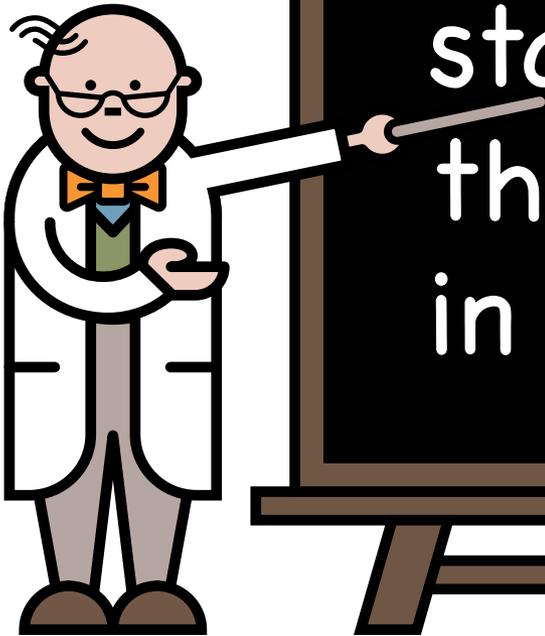


Phone your metabolic team



# Key message

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



# How is CACT 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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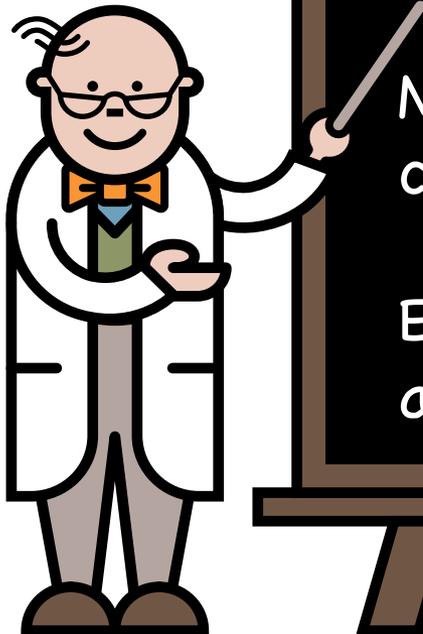
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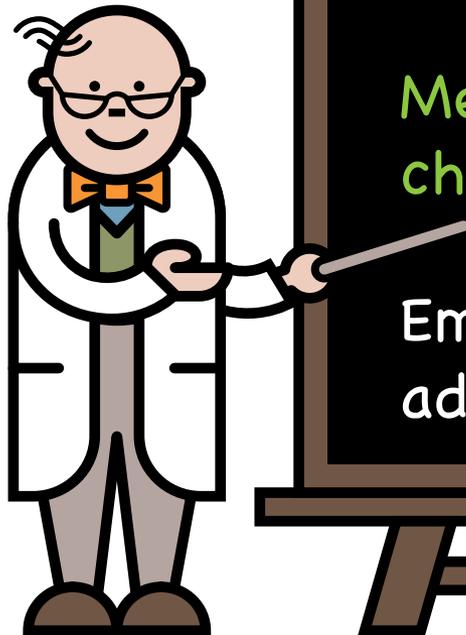
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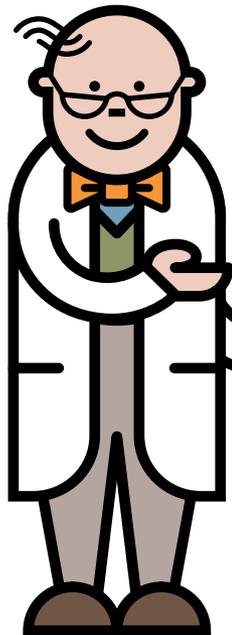
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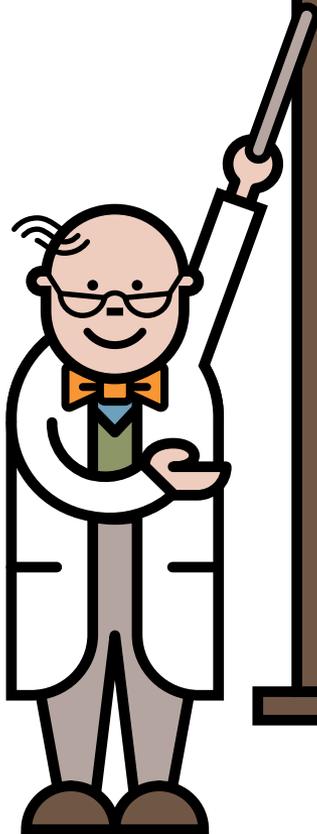
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# Chromosomes, genes, mutations



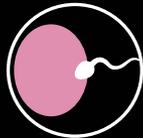
Humans have chromosomes composed of DNA



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

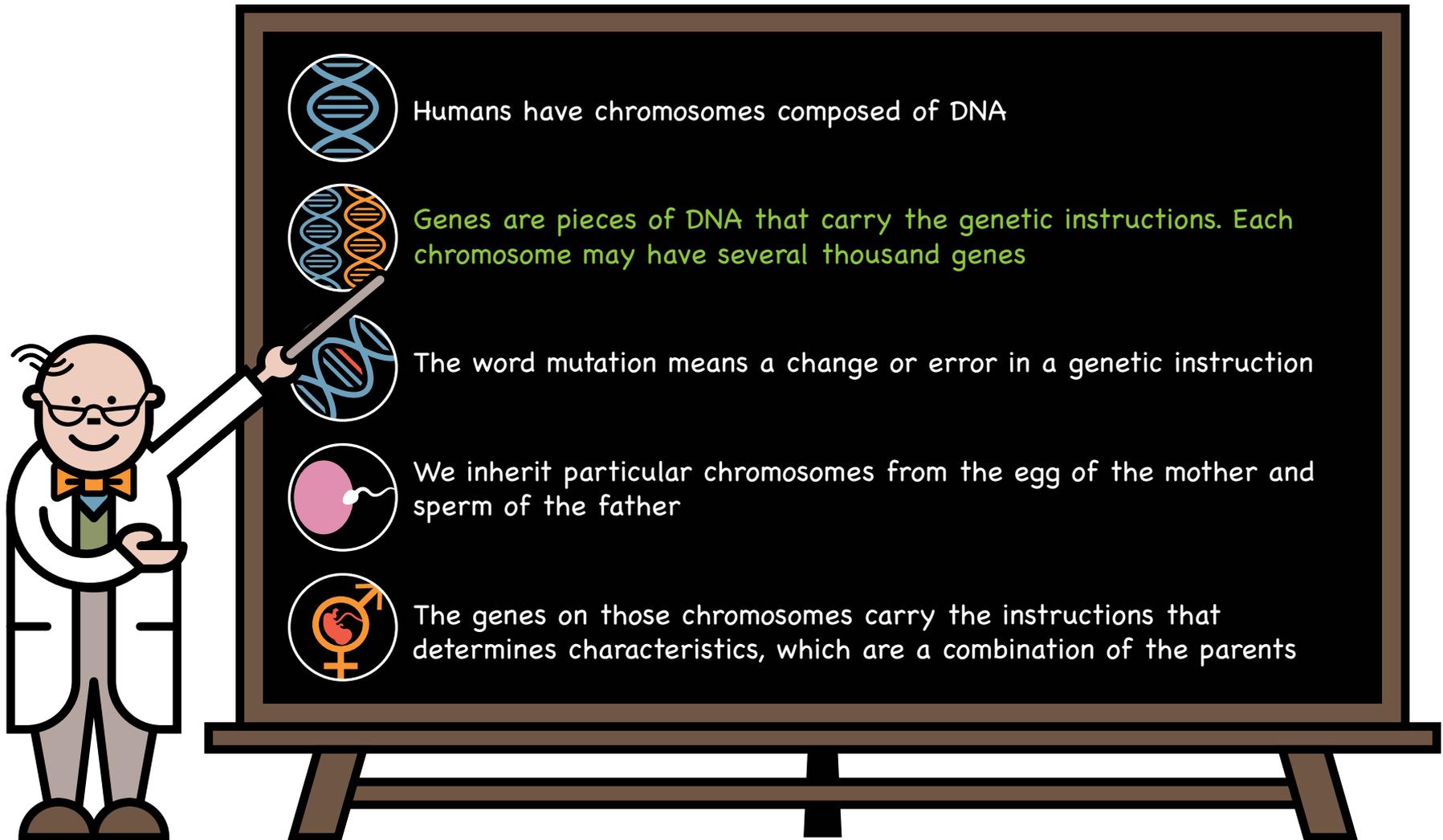


We inherit particular chromosomes from the egg of the mother and sperm of the father



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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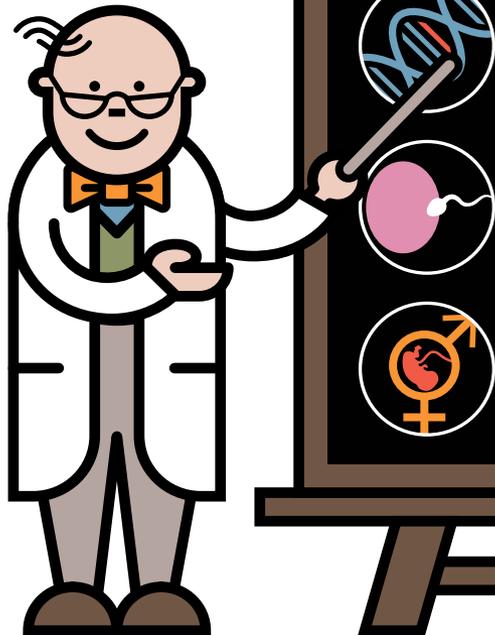
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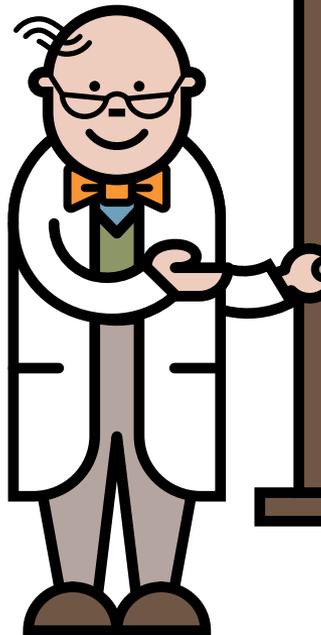


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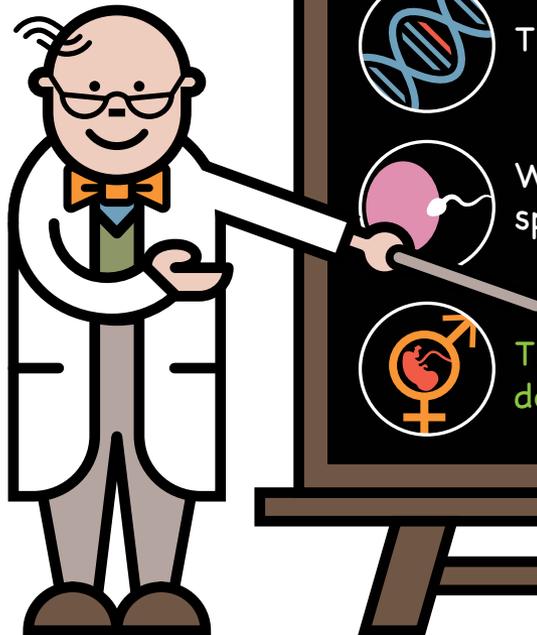


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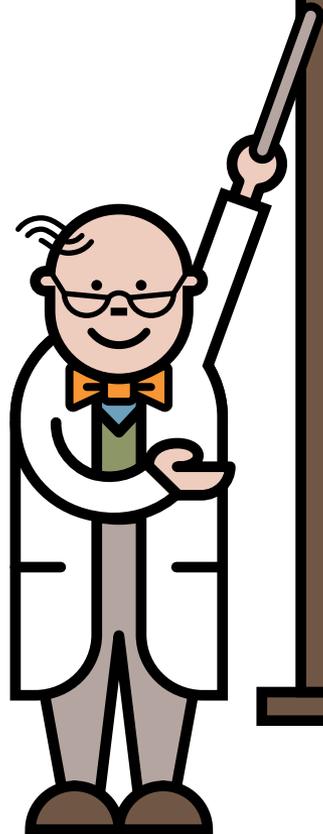


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# Inheritance



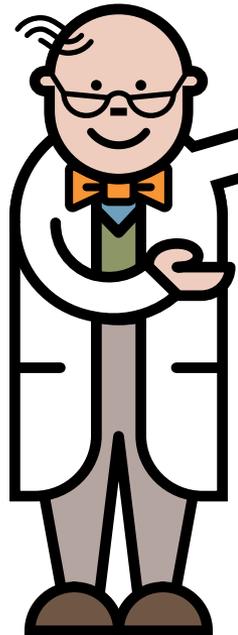
CACT deficiency is an inherited condition. There is nothing that could have been done to prevent your baby from having CACT deficiency

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

Parents of children with CACT deficiency are carriers of the condition

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

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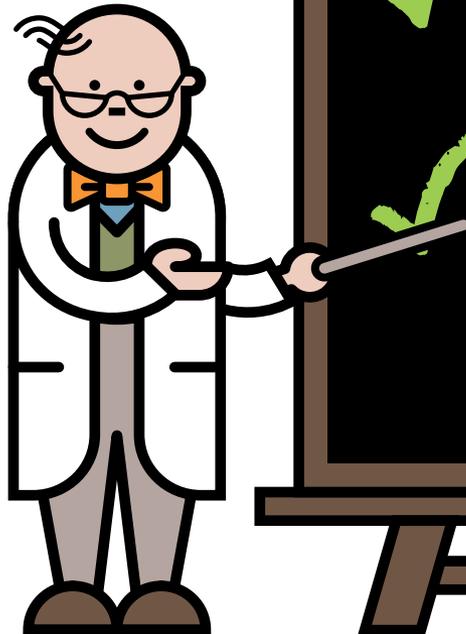


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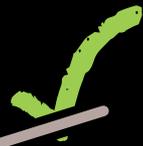
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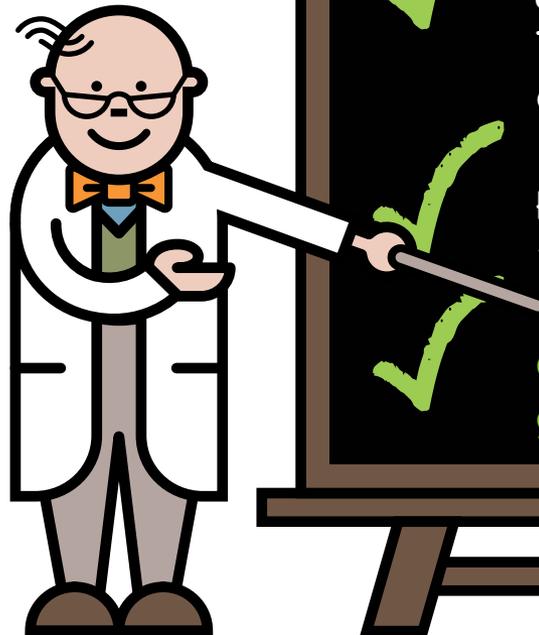
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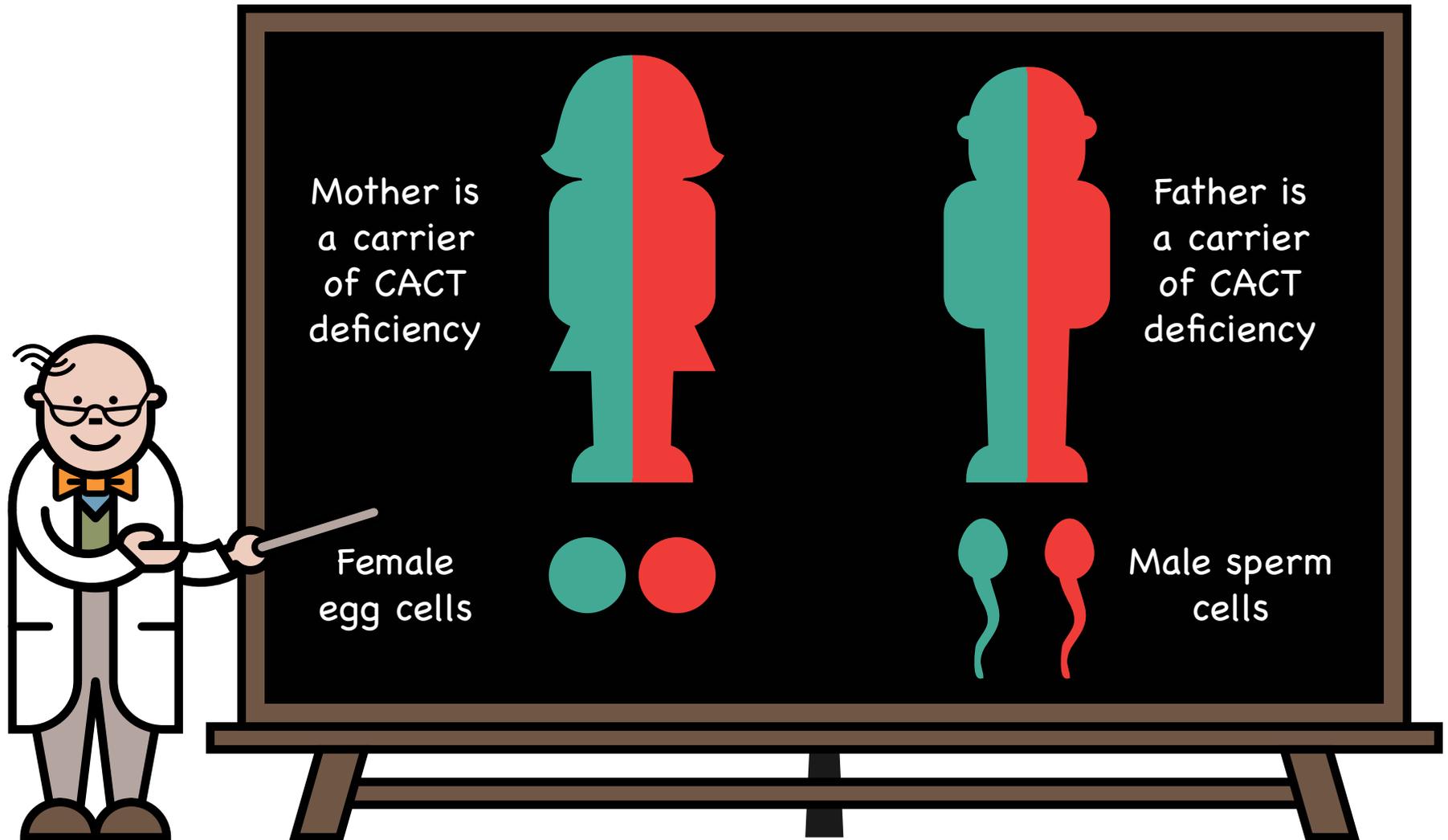
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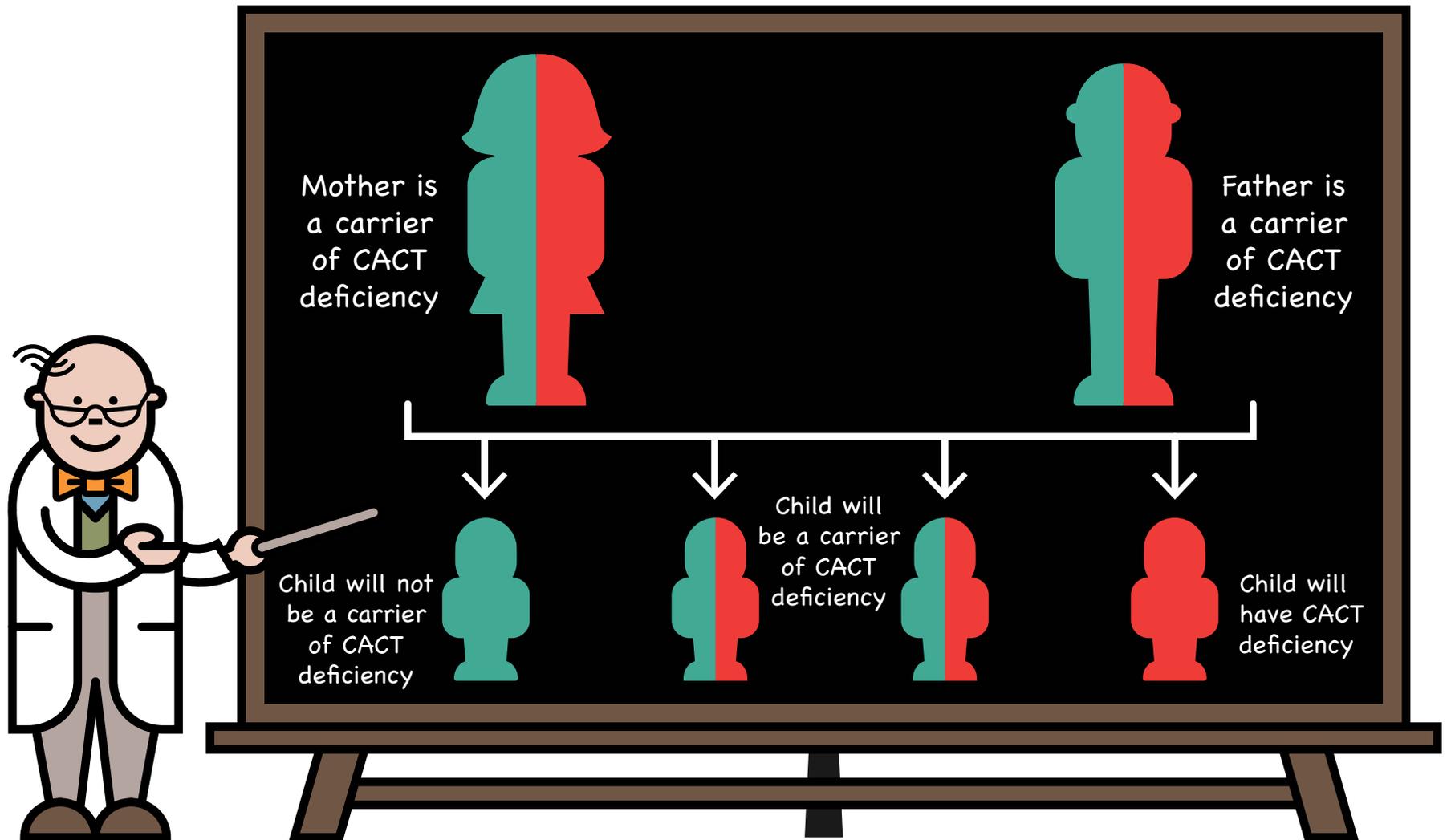
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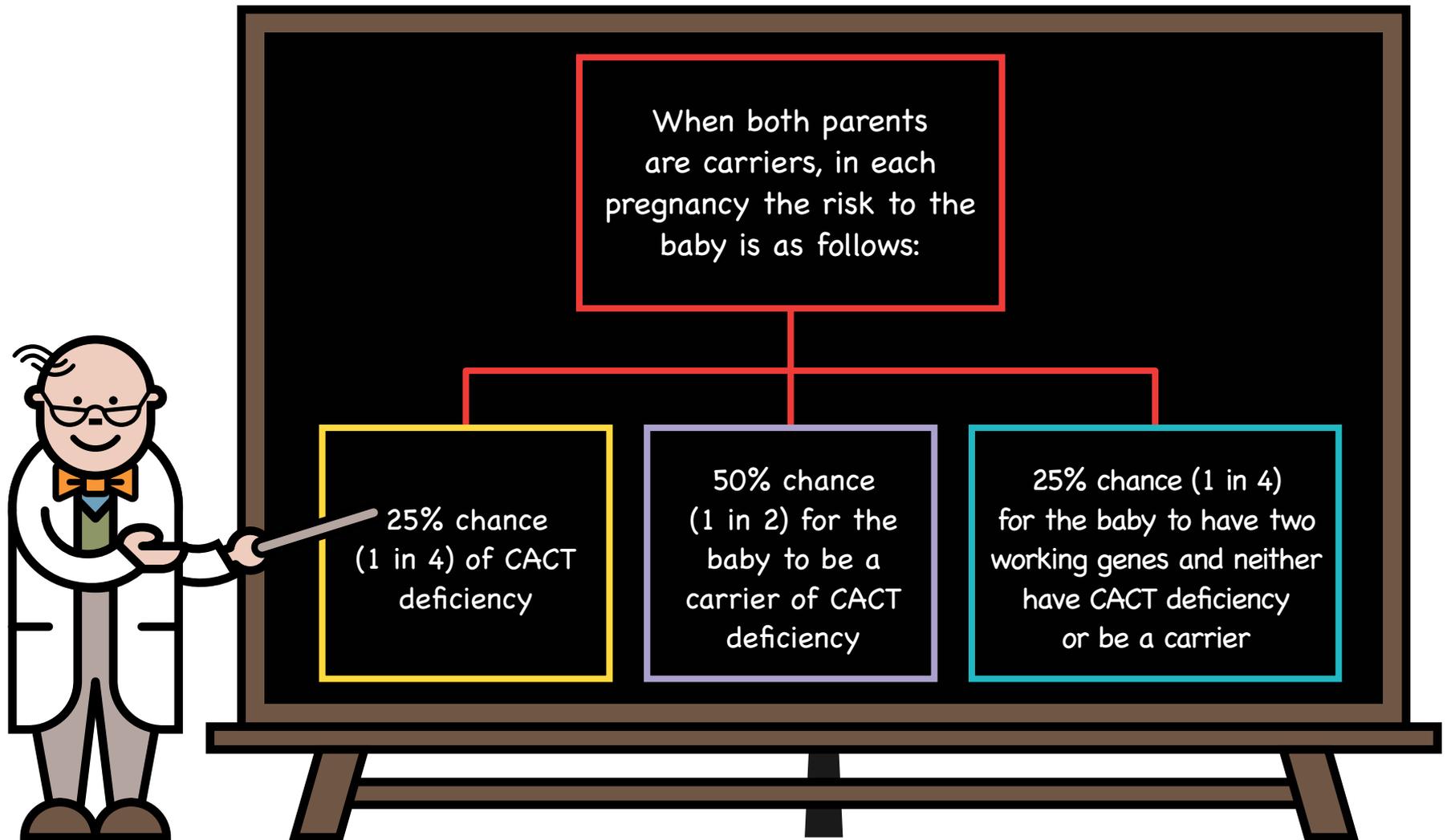
# Inheritance – Autosomal recessive (carriers of CACT deficiency)



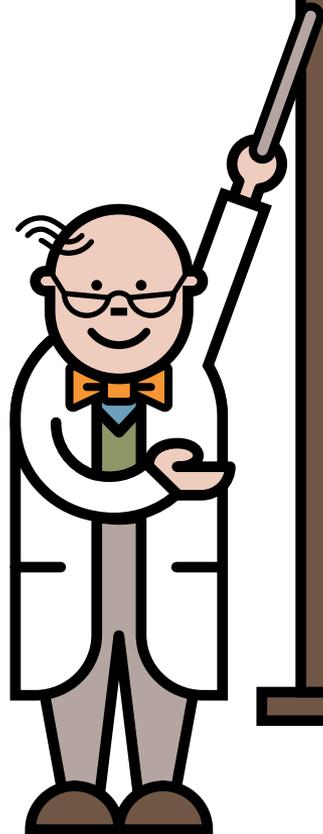
# Inheritance – Autosomal recessive – possible combinations



# Future pregnancies



# Take home messages



CACT deficiency is a serious inherited metabolic condition that causes life threatening symptoms if left untreated

Children with severe CACT deficiency are managed with MCT feed, a low fat diet and regular meals

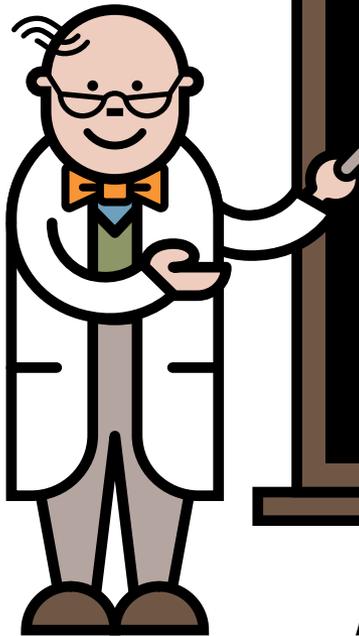
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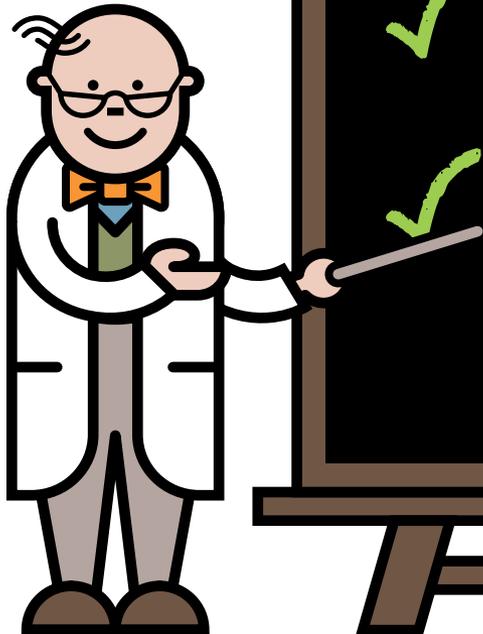


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# Helpful hints



✓ 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

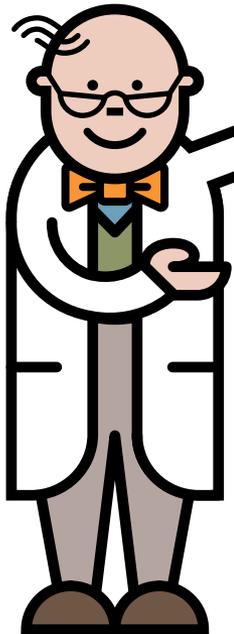
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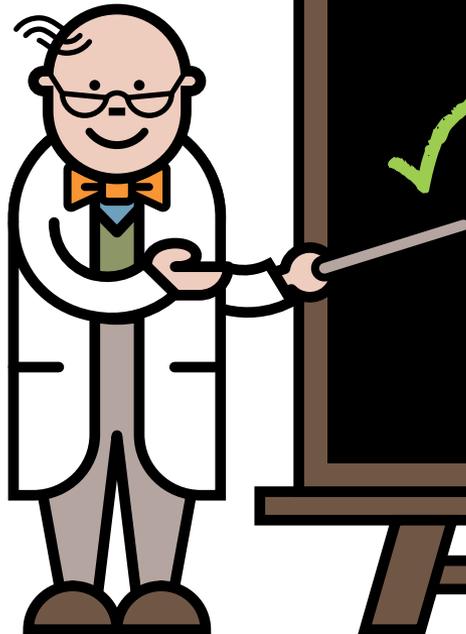
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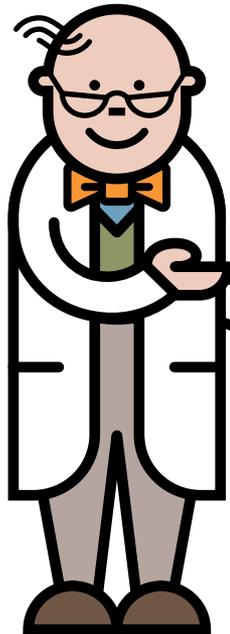
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# Who's who

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

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

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