

# 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 3, SEPTEMBER 2020

# LCHADD

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).

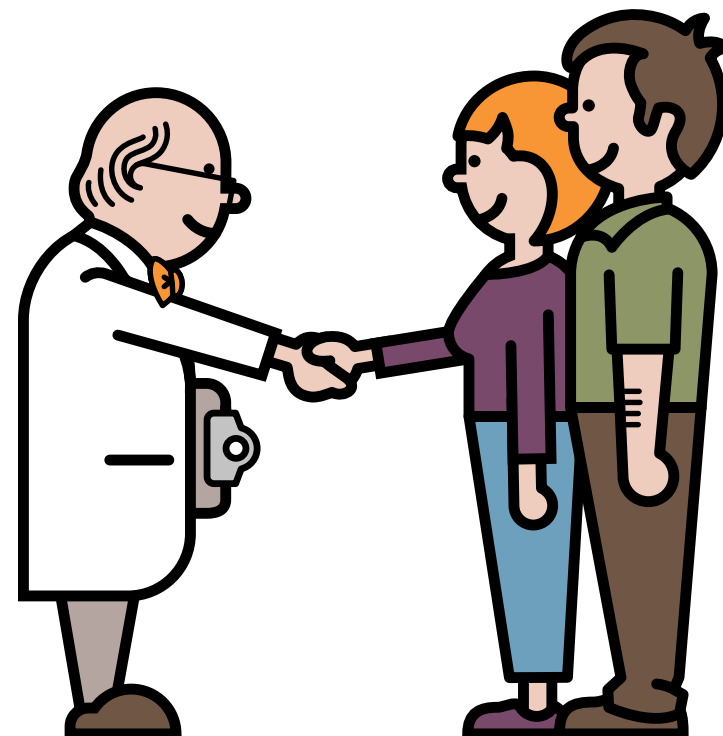
The team are Rachel Skeath, Karen van Wyk, Pat Portnoi and Anita MacDonald. The group is facilitated by Heidi Chan from Nutricia.

Each module produced is reviewed by a consultant clinician who is a member of the BIMDG.

**They are not designed to replace dietary information that may be given by a dietitian in clinic.**

# LCHAD Deficiency

Information for families  
following a new diagnosis



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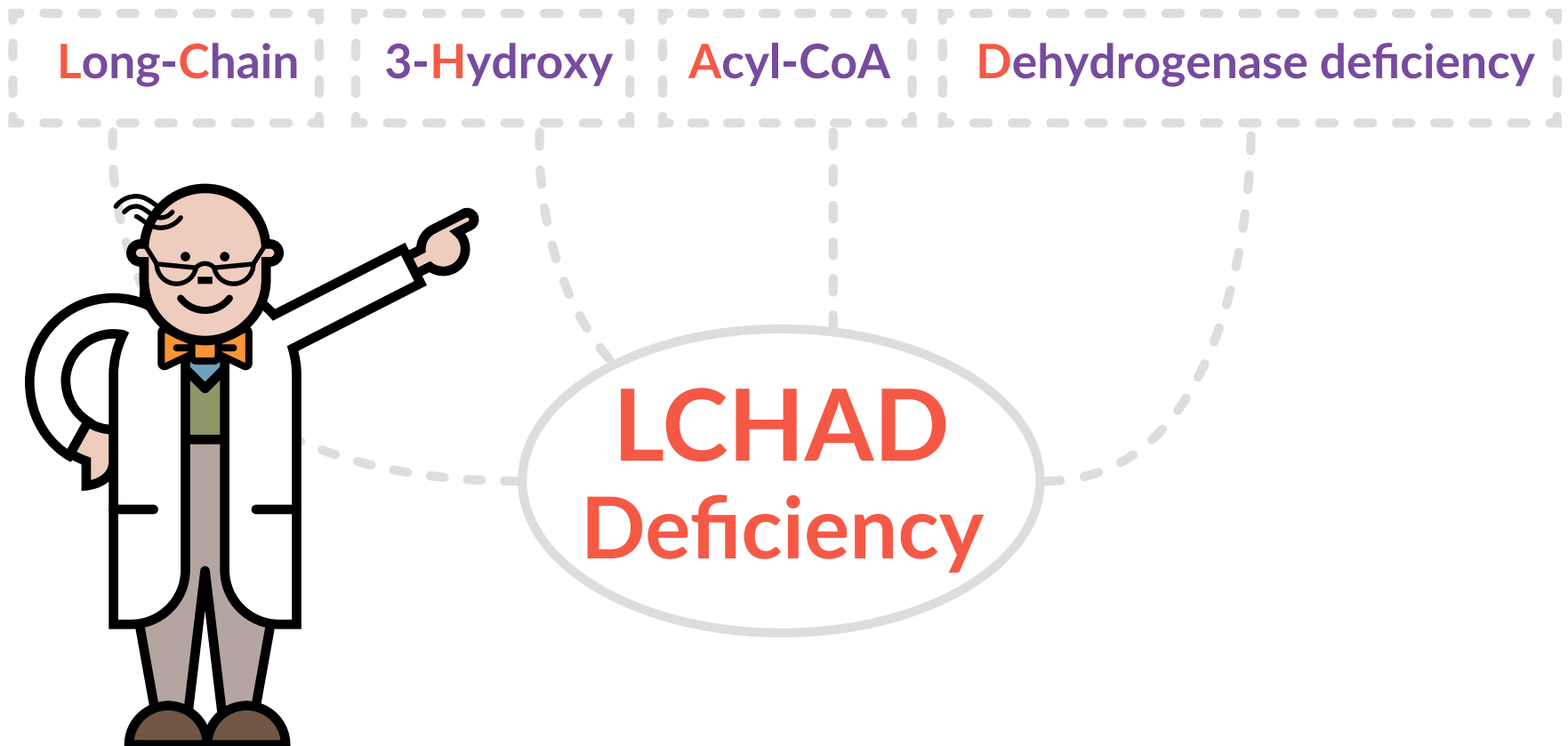
Tools Enabling Metabolic Parents LEarning

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# What is LCHADD?

LCHADD stands for long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency

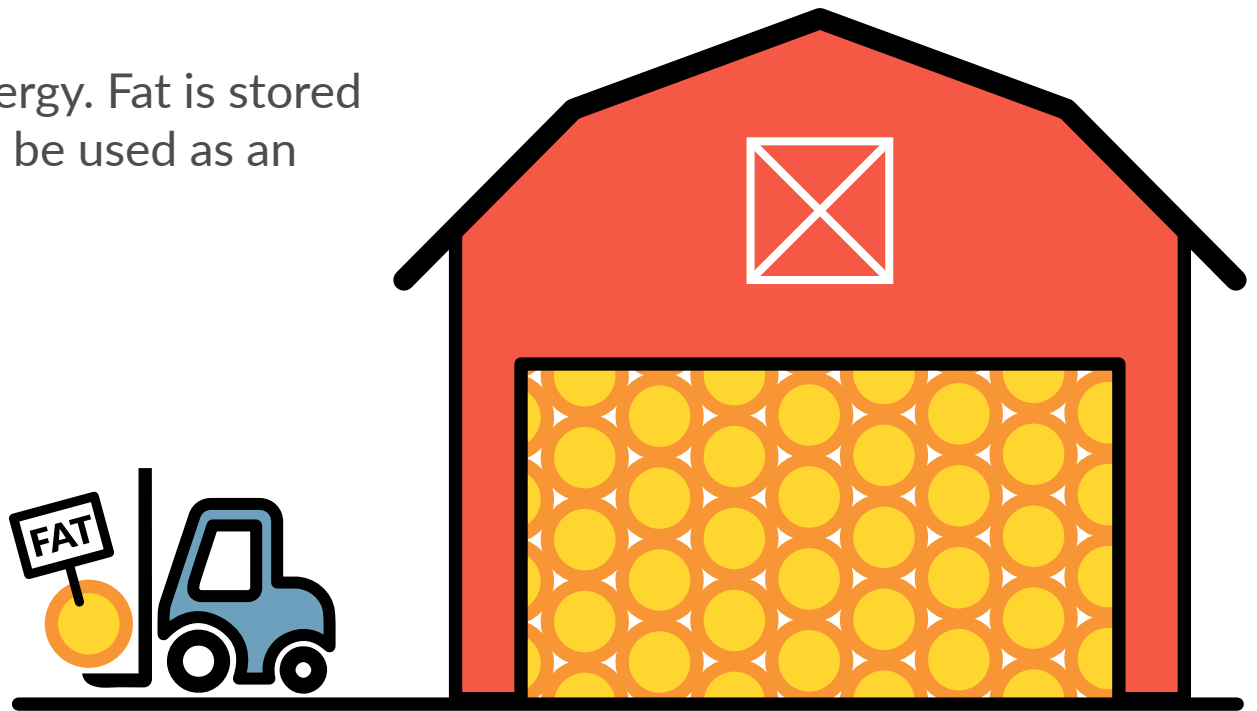
It is an inherited metabolic condition



# Which foods supply the body with energy?

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

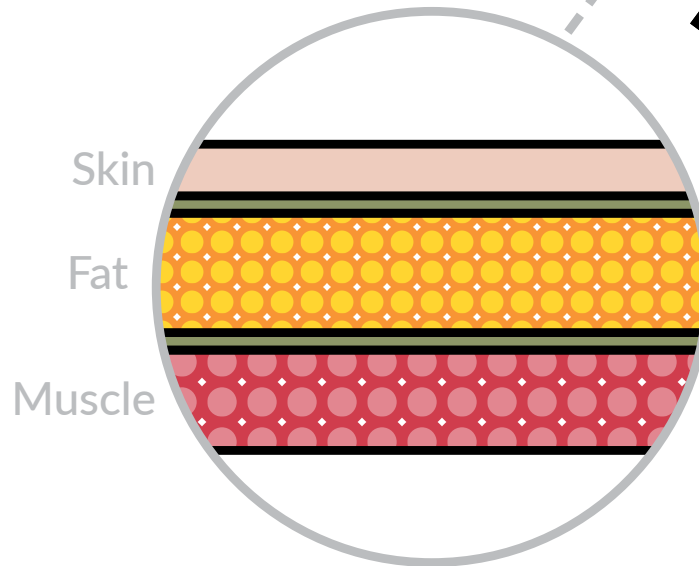
- Carbohydrates (starches and sugars) provide a readily available energy source
- Fats also provide energy. Fat is stored in the body so it can be used as an energy reserve



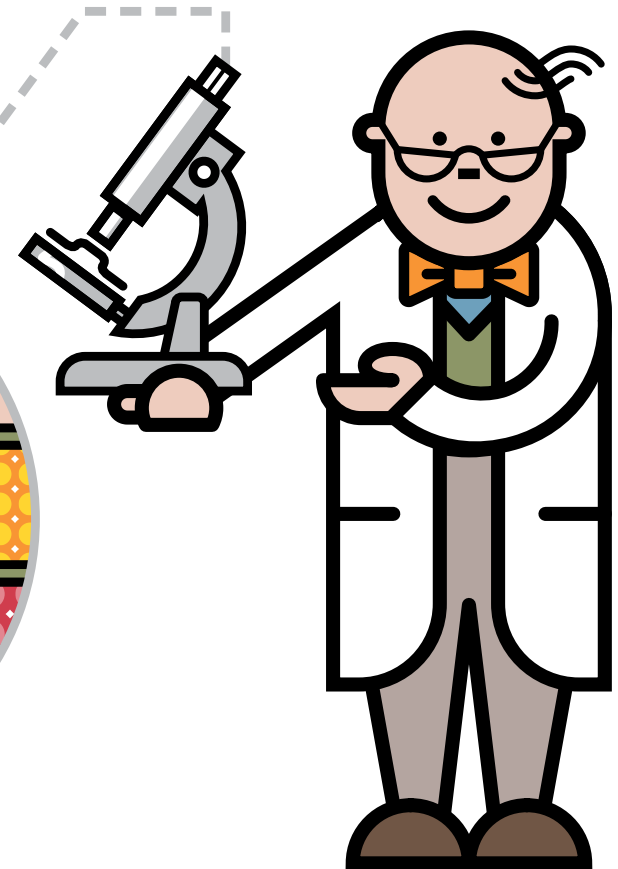
# LCHADD and fat

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

In LCHADD, there are problems breaking down these fat stores to release energy.

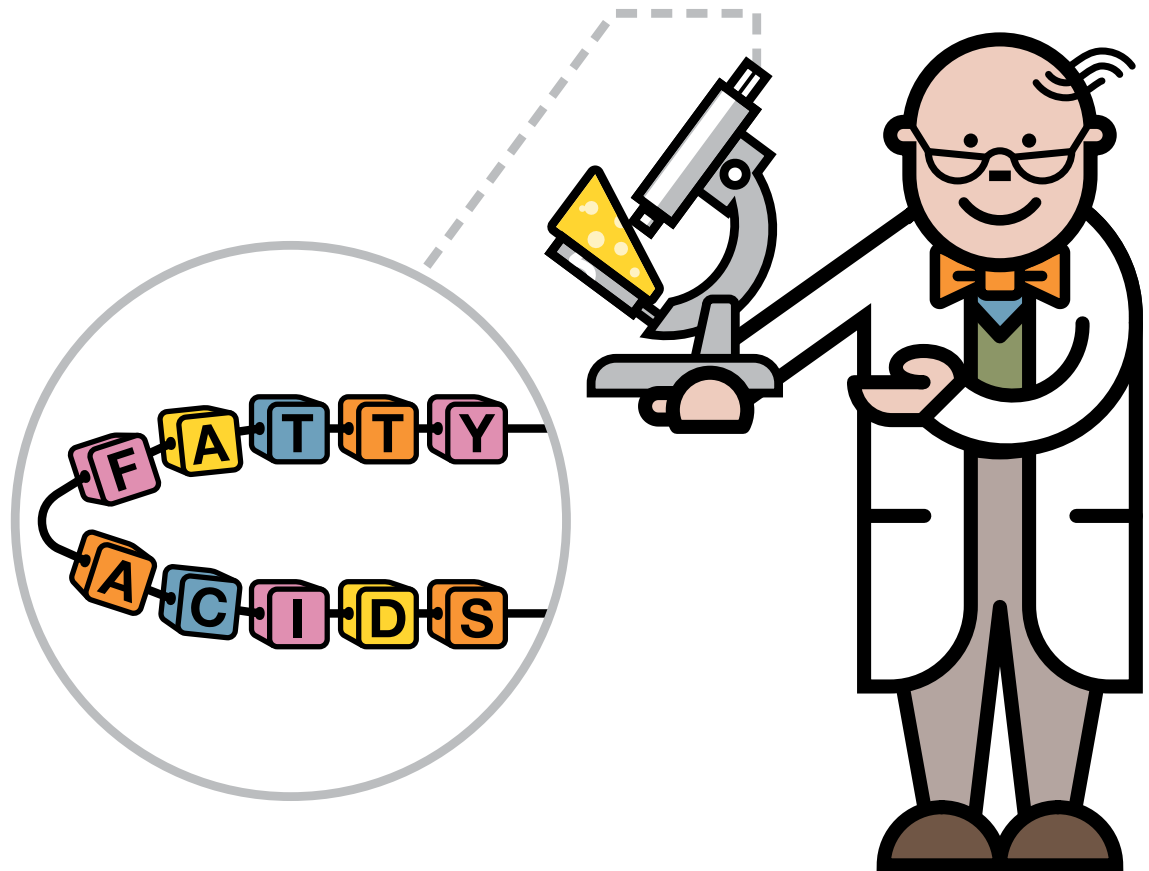


NORMAL



# 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

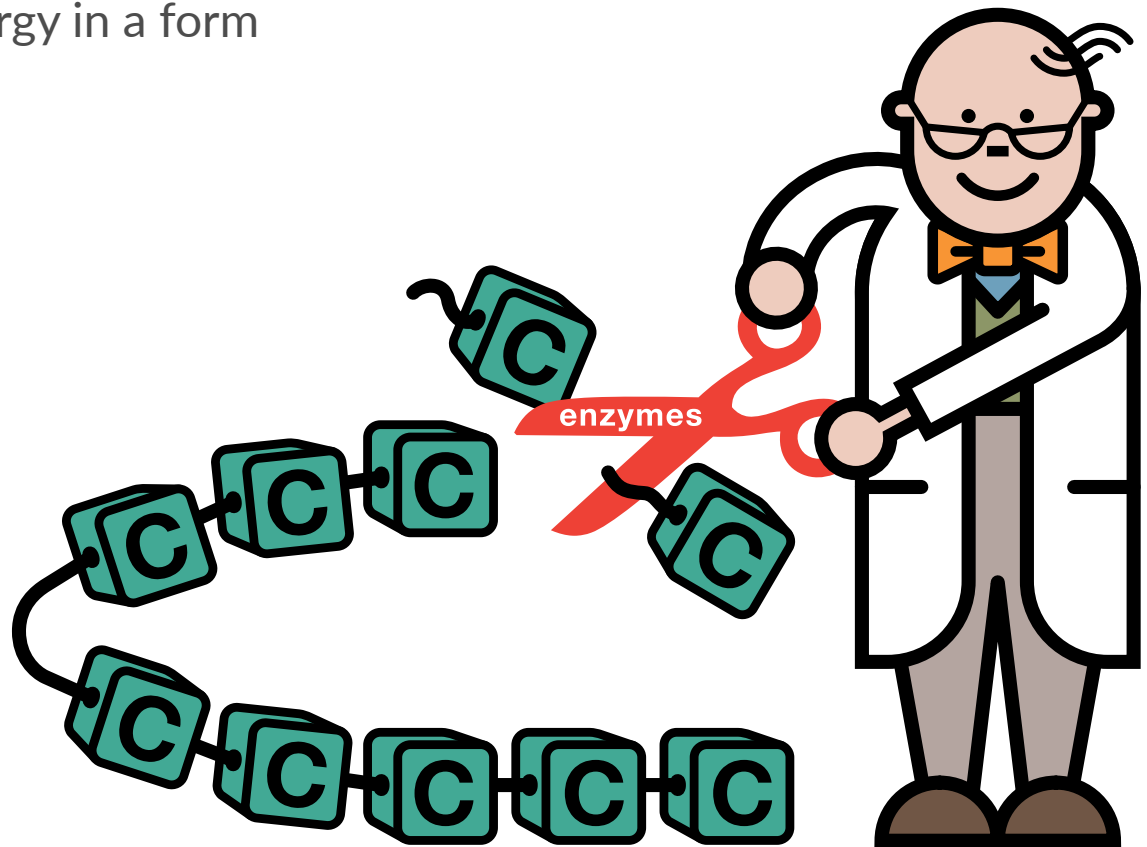


Long chain



# Fatty acids and enzymes

Fatty acid chains are then broken down into smaller units by enzymes (which are like chemical scissors). This enables the body to produce energy in a form which it can use.

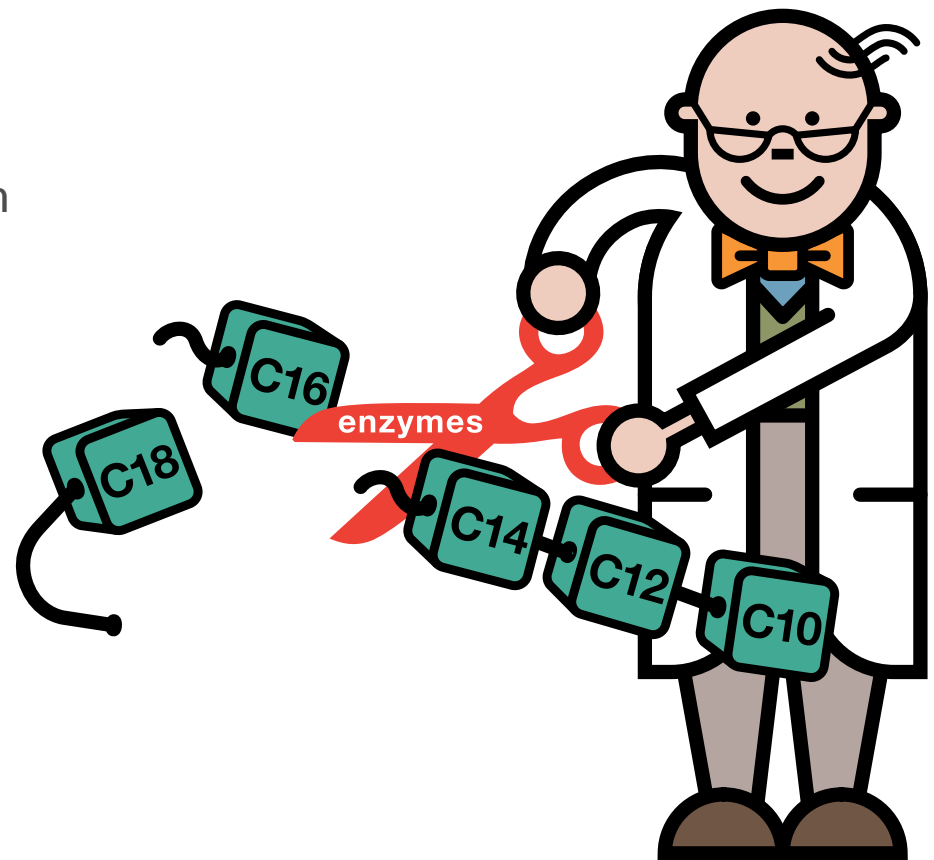


# What happens in LCHADD?

In LCHADD, the body lacks an enzyme that helps convert fat stores into energy.

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

When fats need to be broken down quickly, this causes a problem.



# What can go wrong in LCHADD?

There can be a shortage of energy supply and a build up of harmful chemicals during illnesses or fasting.

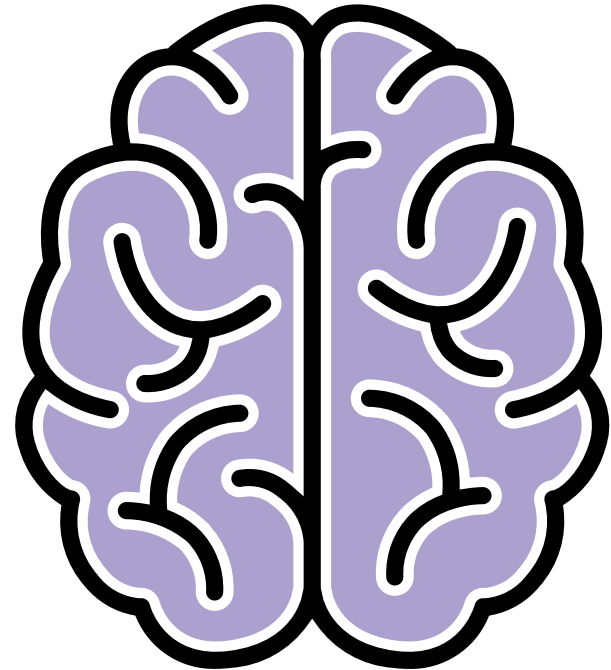
Symptoms include:

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

# What can go wrong in LCHADD?

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.

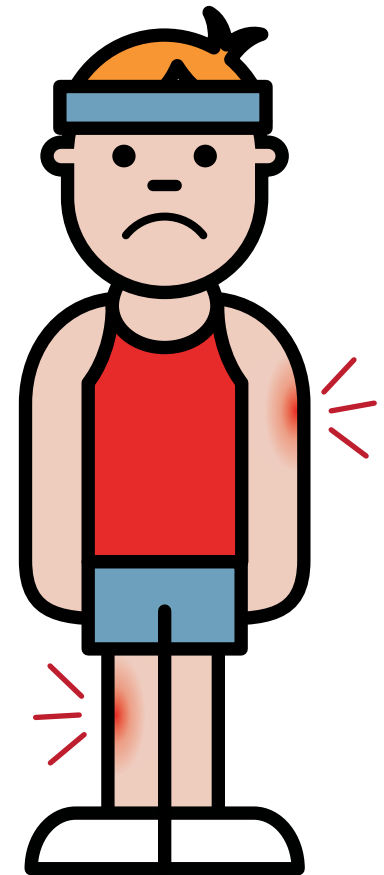
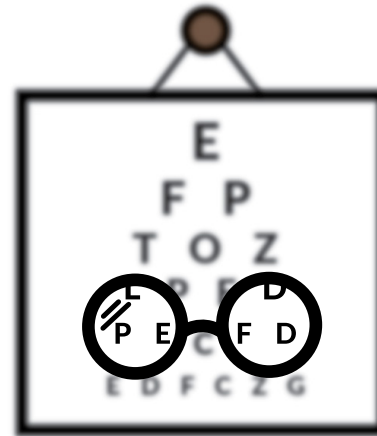
**However, please remember, this can all be prevented with timely and correct management.**



# What else happens in LCHADD

It may also cause

- learning difficulties
- delays in walking
- muscle pain especially with exercise
- vision loss
- nerve problems in arm and legs
- enlarged heart and breathing difficulties



# Metabolic crisis

- A **metabolic crisis** triggers the LCHADD 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 for a long time without food
- Avoidance of a metabolic crisis is essential



# How is LCHADD diagnosed?

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

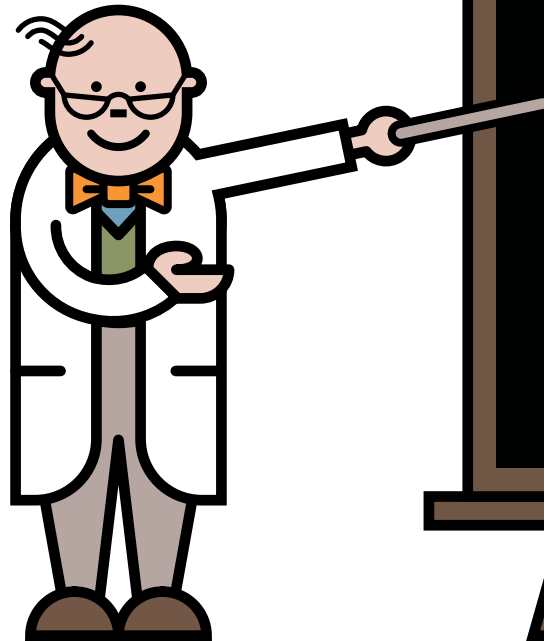
The diagnosis is confirmed by finding mutations in the genes for LCHAD.



# How is LCHADD 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 LCHADD. 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 LCHADD managed day to day?

Special infant feed containing a fat called medium chain triglycerides (MCT)



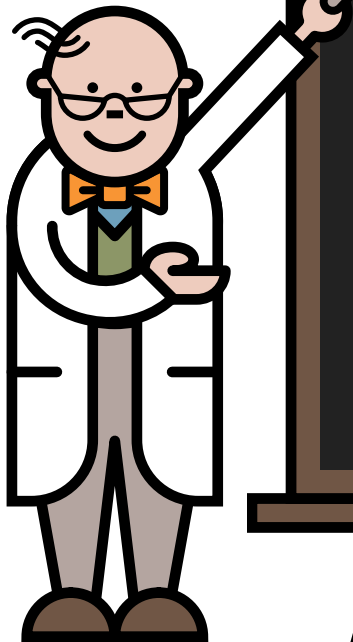
Very low fat diet



MCT oil for cooking or MCT as a supplement



Regular mealtimes as recommended by your metabolic team



# How is LCHADD managed day to day?

Special infant feed containing a fat source as medium chain triglycerides (MCT)



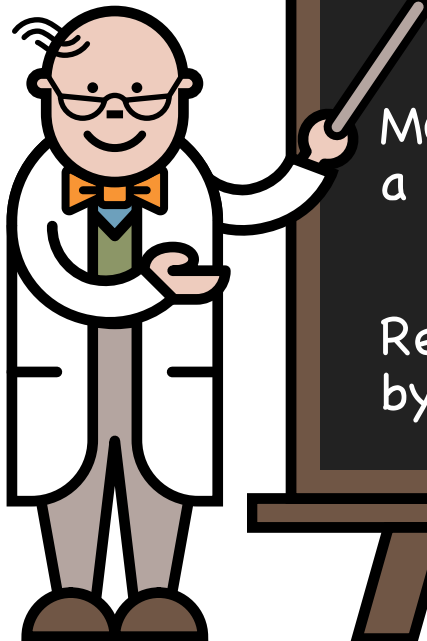
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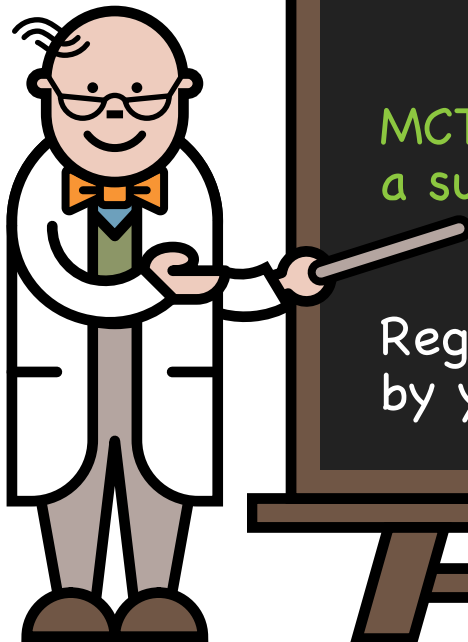
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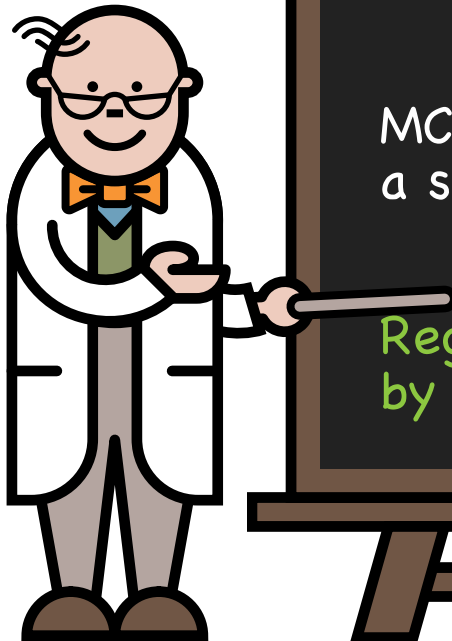
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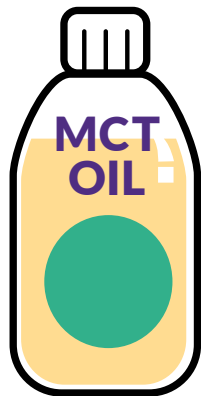


# LCHADD and fat

The diet needs to be very 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.



# Very low fat foods

The diet needs to be very low in fat.

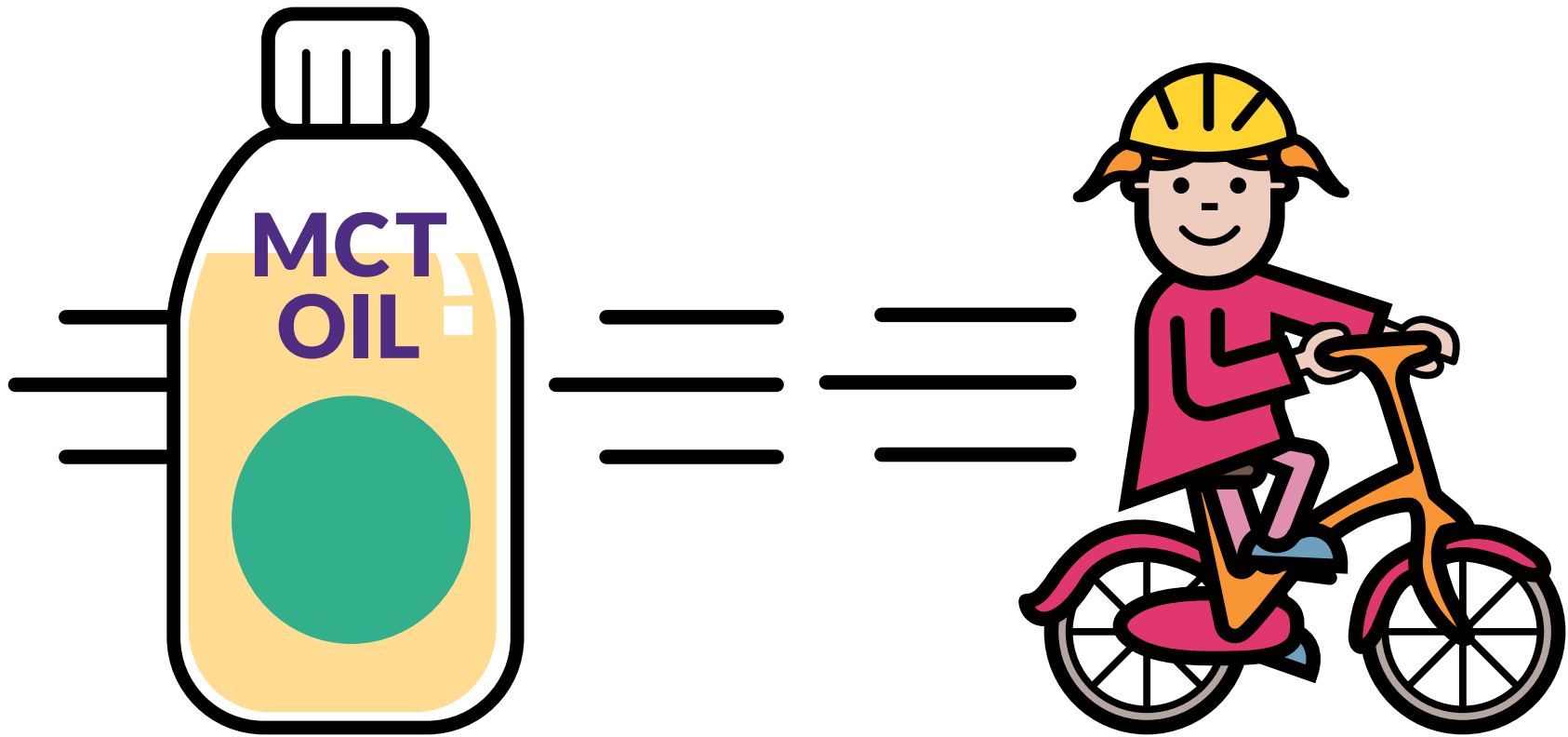
**Very low fat foods are permitted.**

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



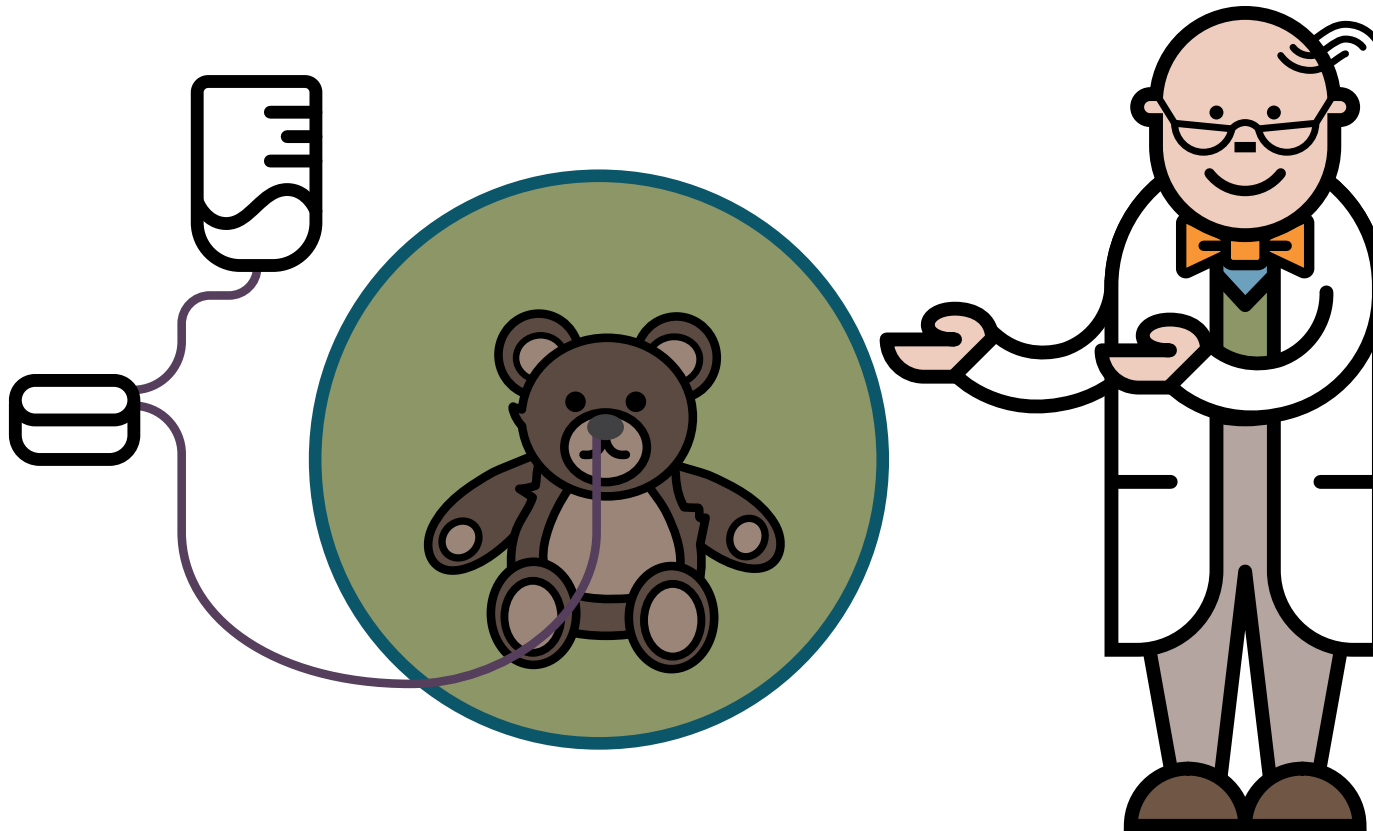
# LCHADD and exercise

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



# Is tube feeding needed?

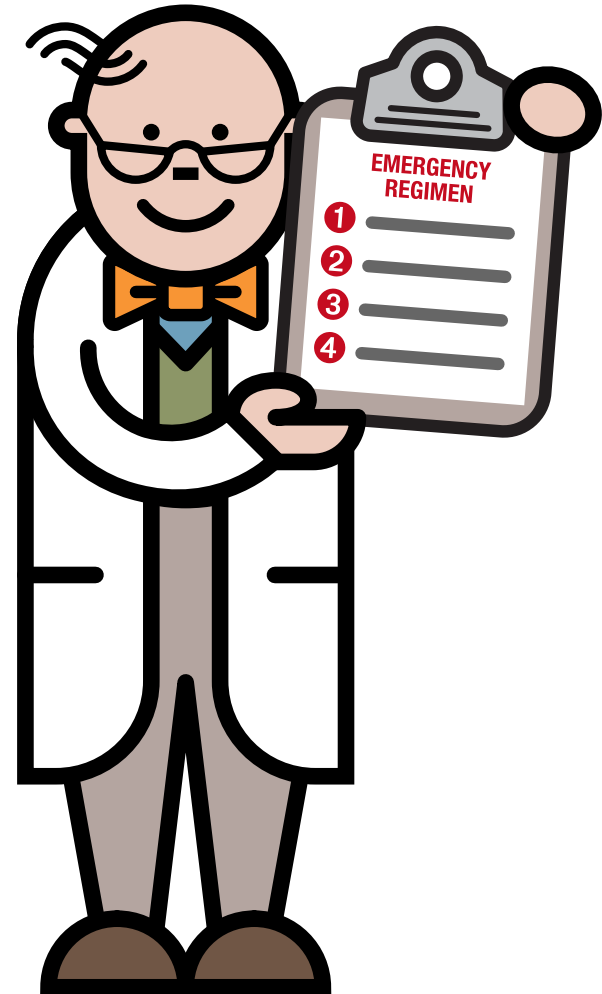
In babies with the most severe forms of LCHADD, tube feeding may be necessary. This will ensure energy, nutrient and fluid needs are met.





# How is LCHADD 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 LCHADD managed during illness?

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



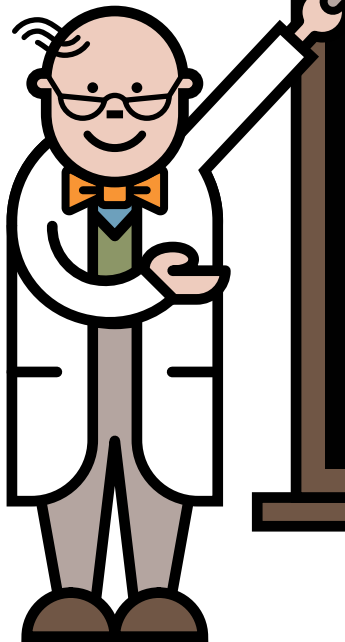
This must be given regularly  
day and night



Phone your metabolic team  
for help if your child is unwell



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



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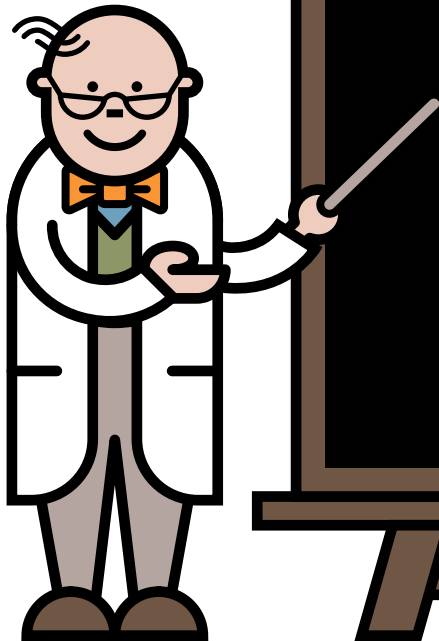
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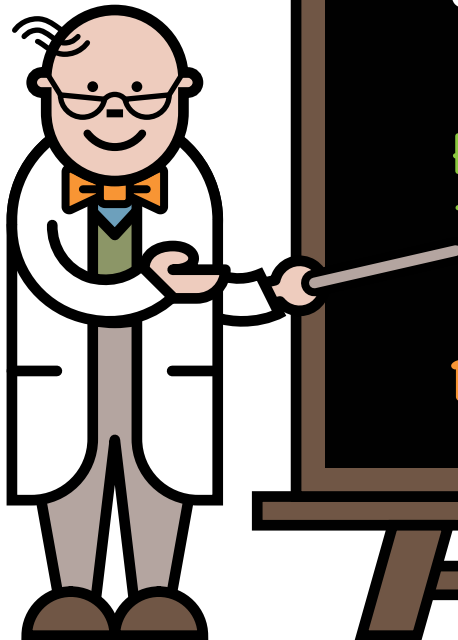
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# Checklist for illness

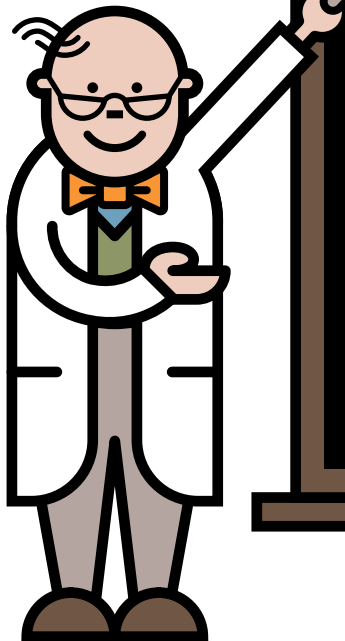
Always take full amounts of emergency feeds as prescribed



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

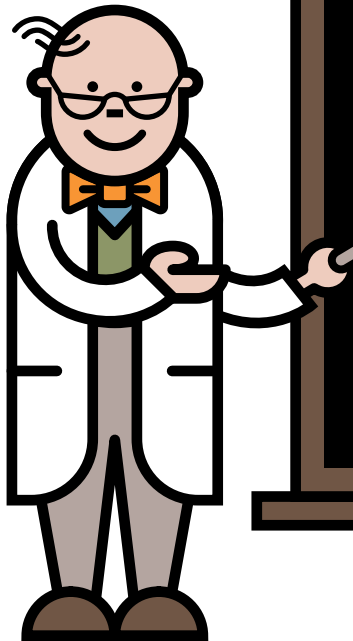
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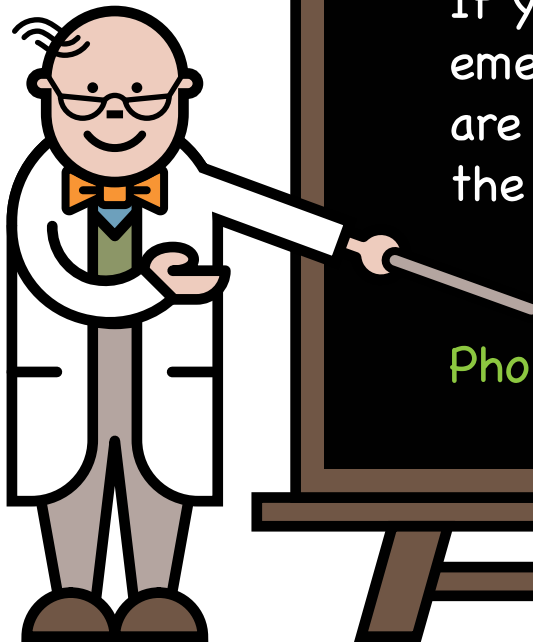
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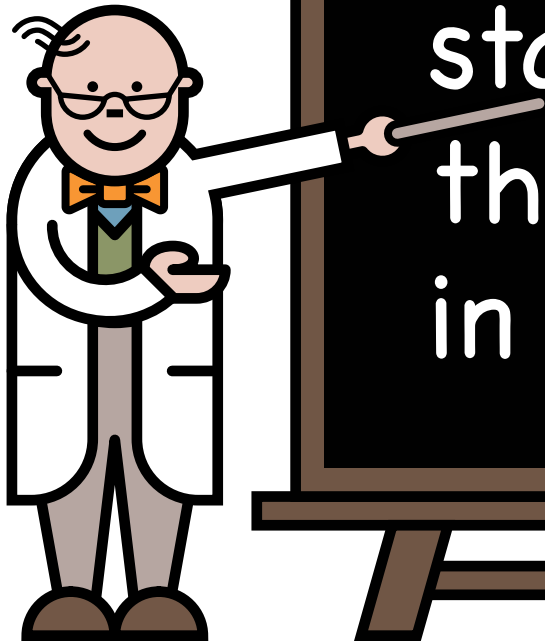


Phone your metabolic team



# Key message

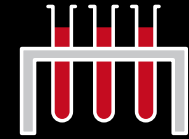
It is imperative that emergency feeds are started **promptly** and there are **no delays** in management.





# How is LCHADD monitored?

Blood tests to check  
nutrient levels



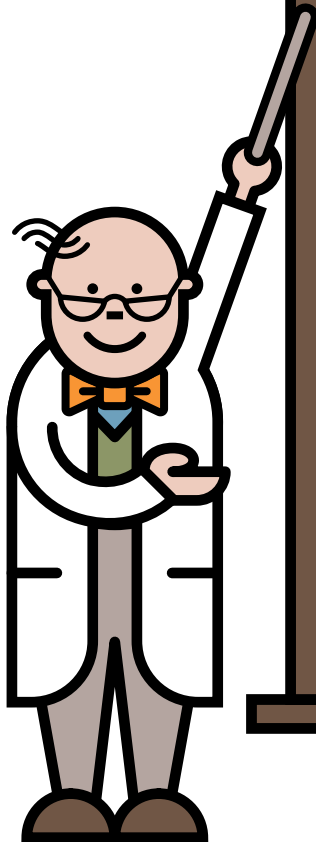
Height and weight



Developmental checks  
& other medical checks

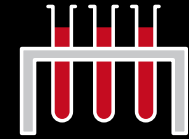


Emergency feeds are  
adjusted with age



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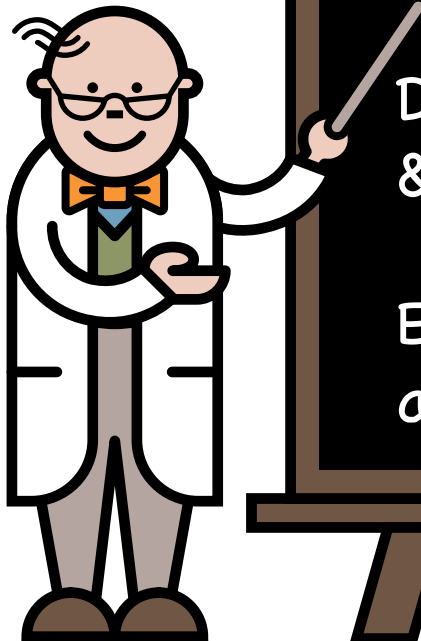
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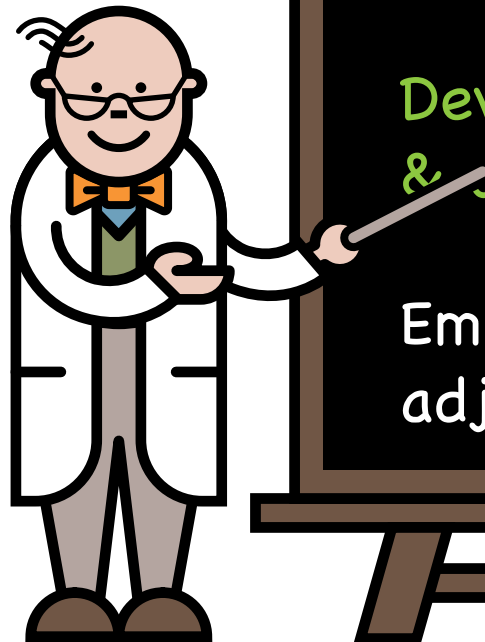
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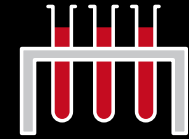
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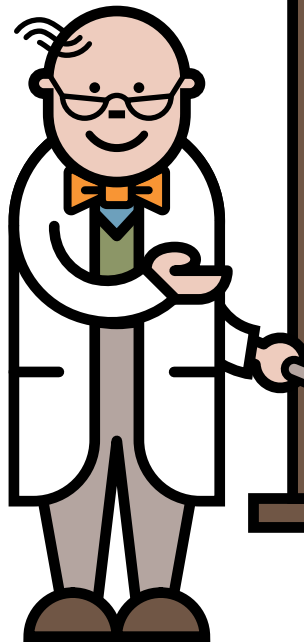
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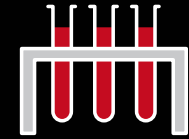
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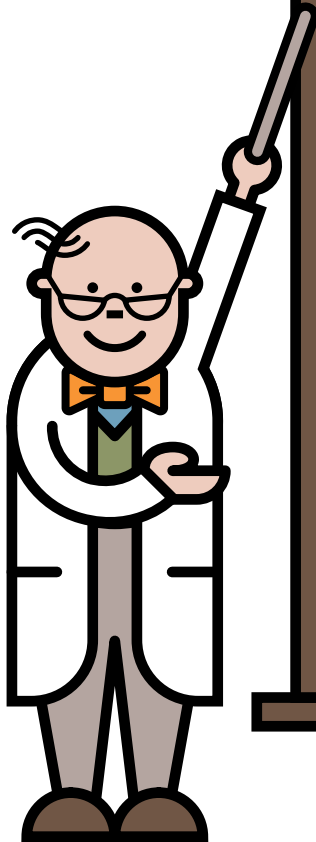
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# Chromosomes, genes, mutations



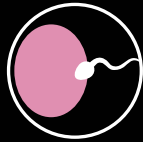
Humans have chromosomes composed of DNA



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

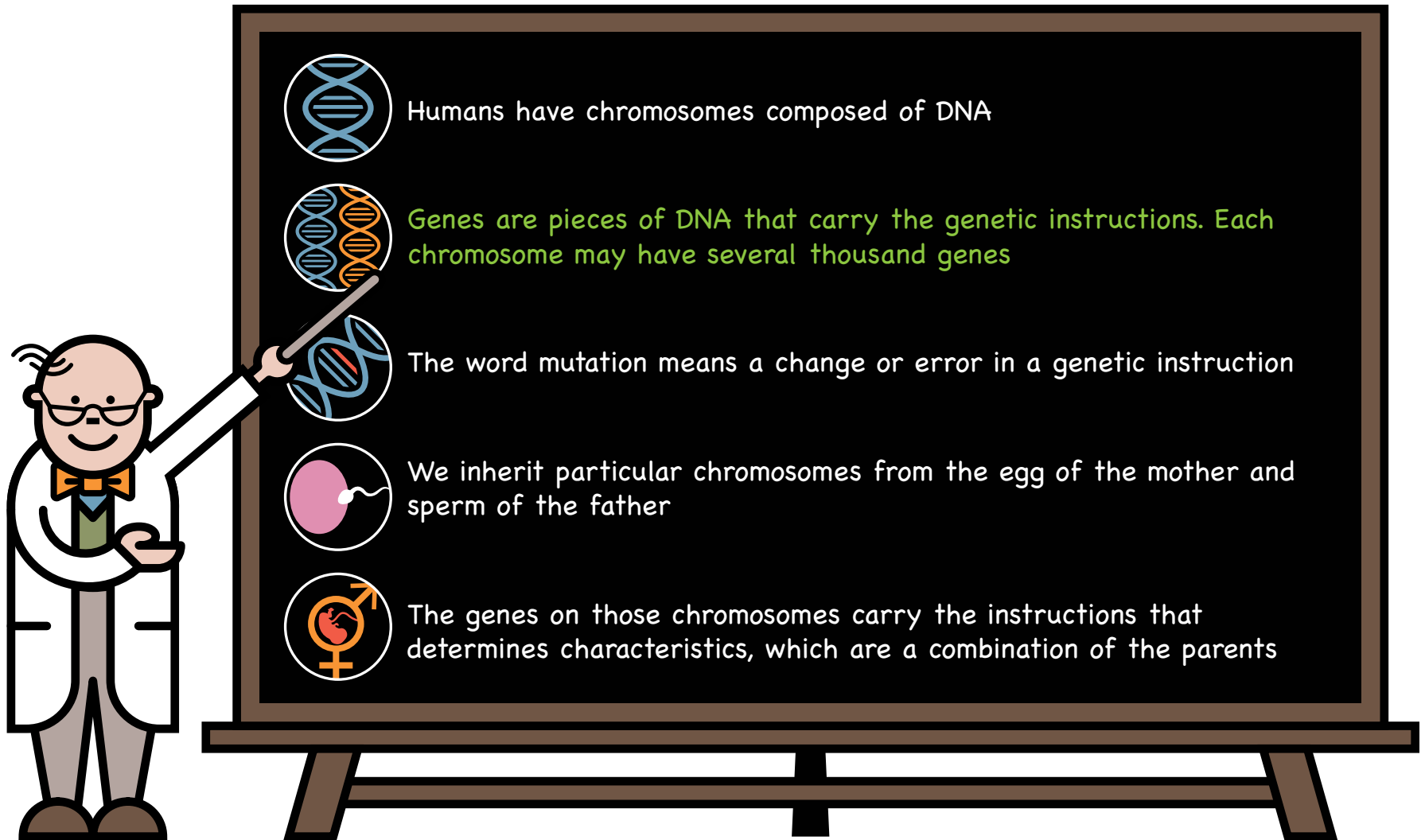



We inherit particular chromosomes from the egg of the mother and sperm of the father





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


# Chromosomes, genes, mutations




 Humans have chromosomes composed of DNA

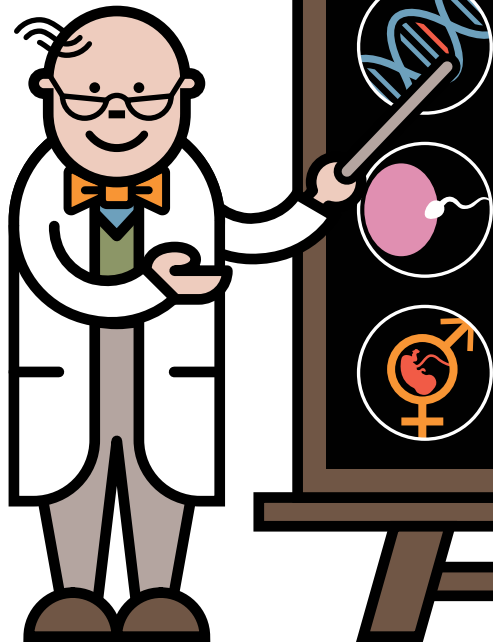
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# Chromosomes, genes, mutations



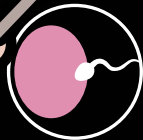
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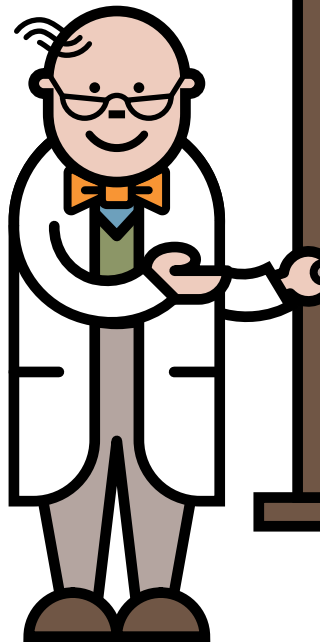


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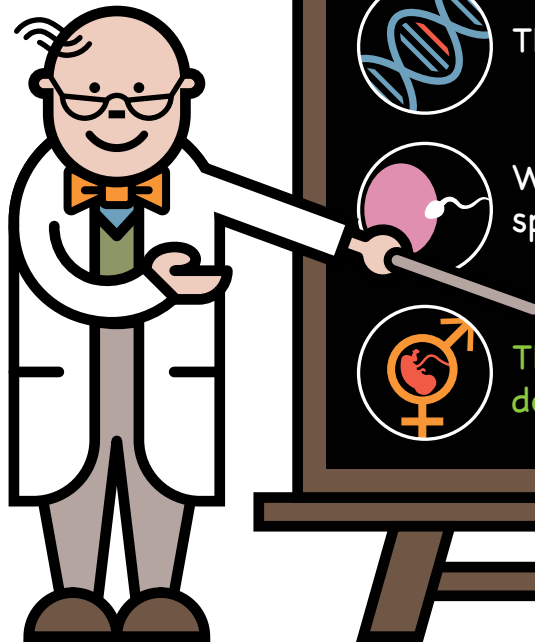
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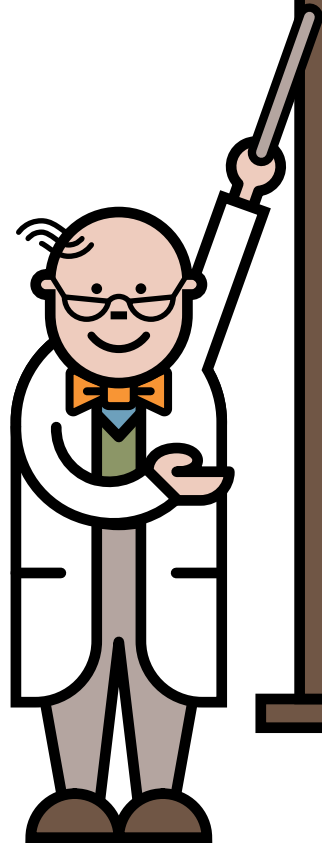


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# Inheritance



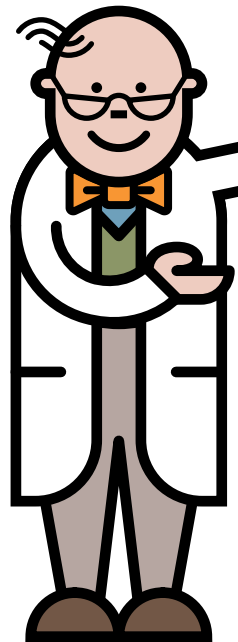
LCHADD is an inherited condition. There is nothing that could have been done to prevent your baby from having LCHADD

Everyone has a pair of genes that make the long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency enzyme. In children with LCHADD, neither of these genes works correctly. These children inherit one non-working LCHAD gene from each parent

Parents of children with LCHADD are carriers of the condition

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

# Inheritance



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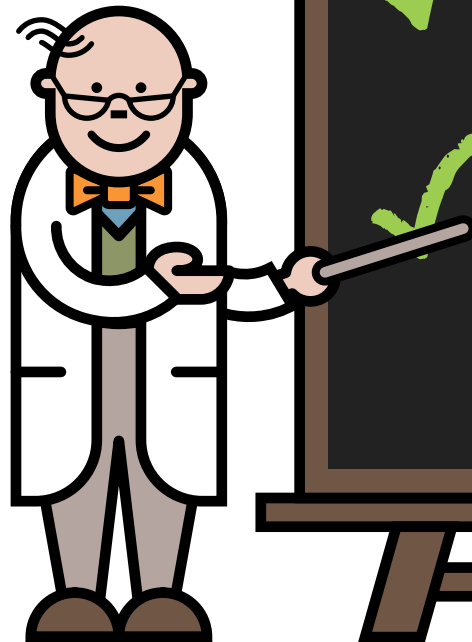


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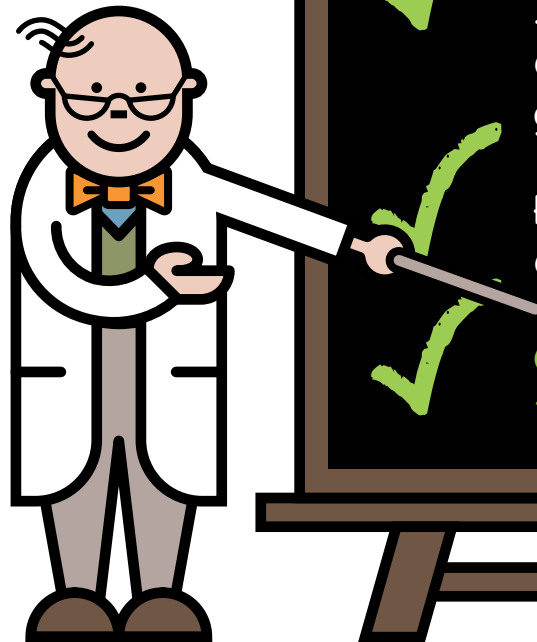
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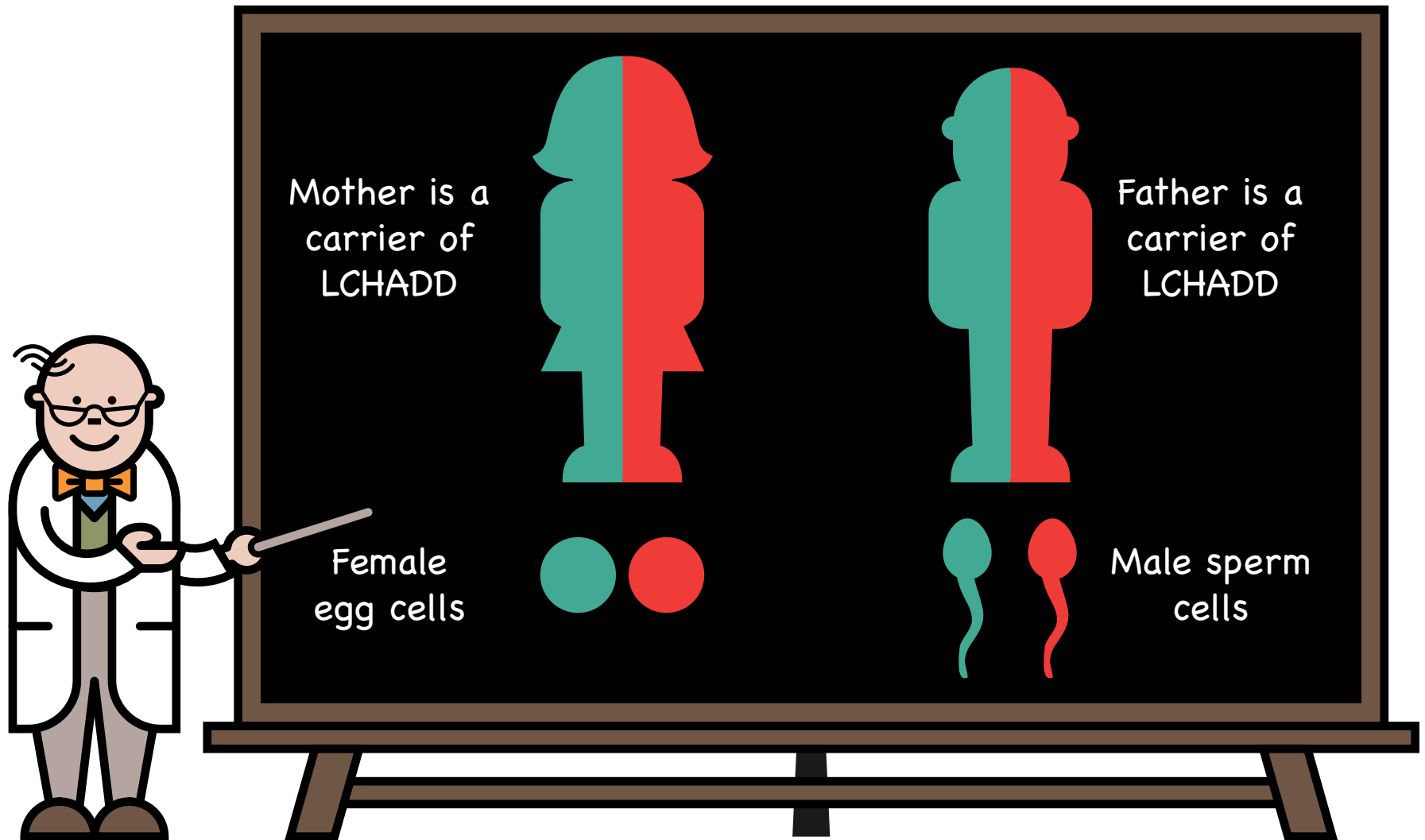
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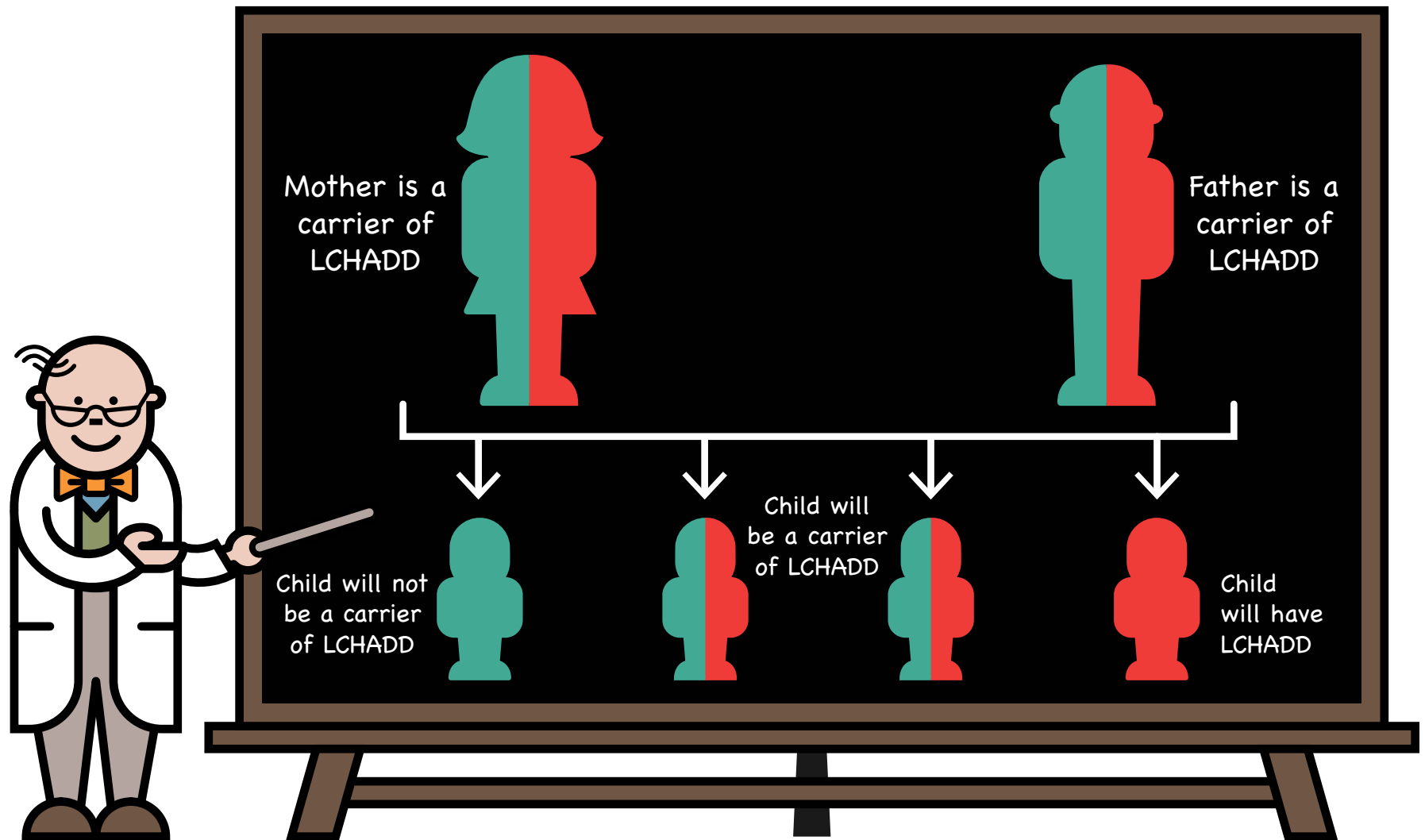
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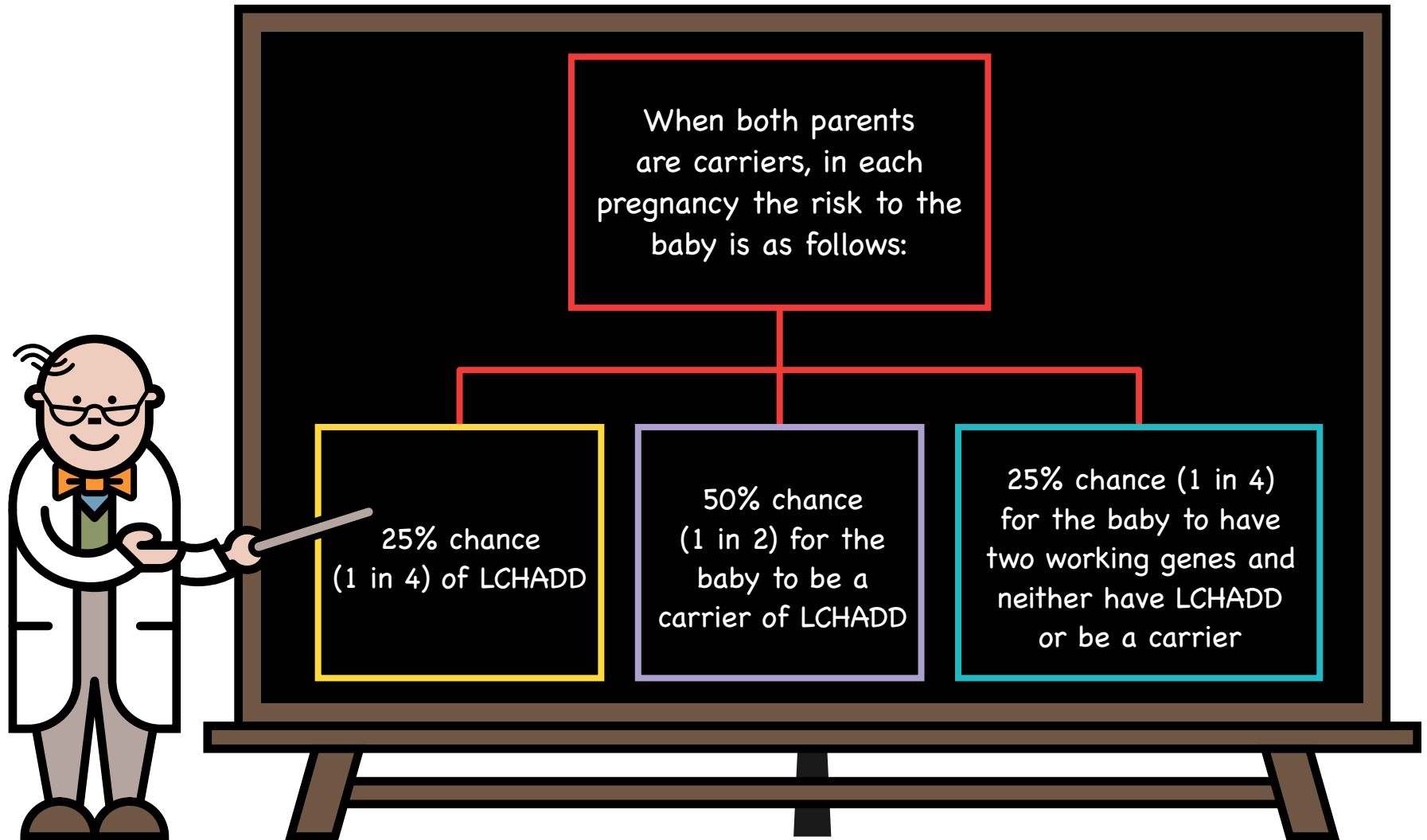
# Inheritance – Autosomal recessive (carriers of LCHADD)



# Inheritance – Autosomal recessive – possible combinations

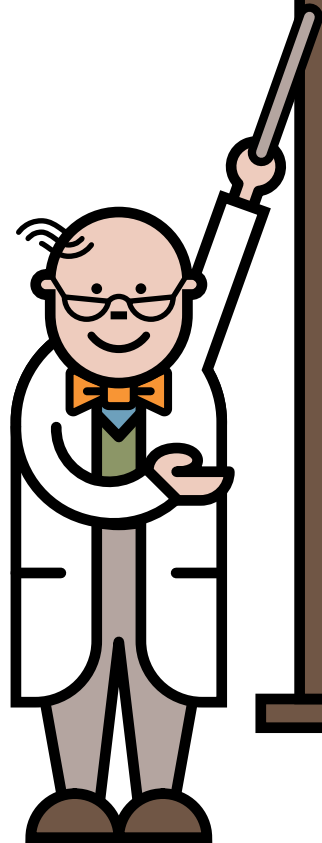


# Future pregnancies





# Take home messages



LCHADD 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

Children with LCHADD are managed with a low fat diet, MCT oil and regular meals

People with LCHADD should lead normal, healthy and active lives

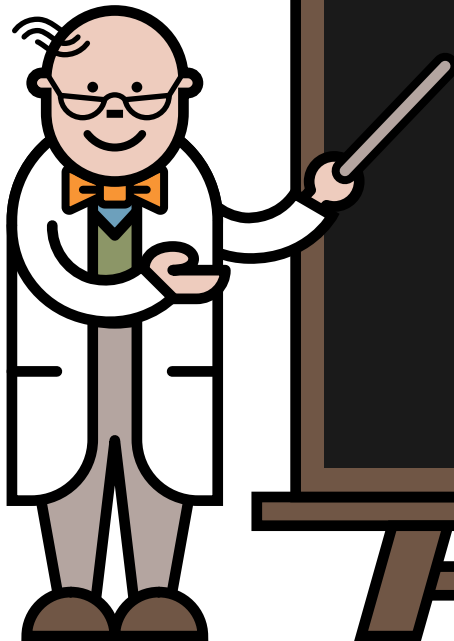
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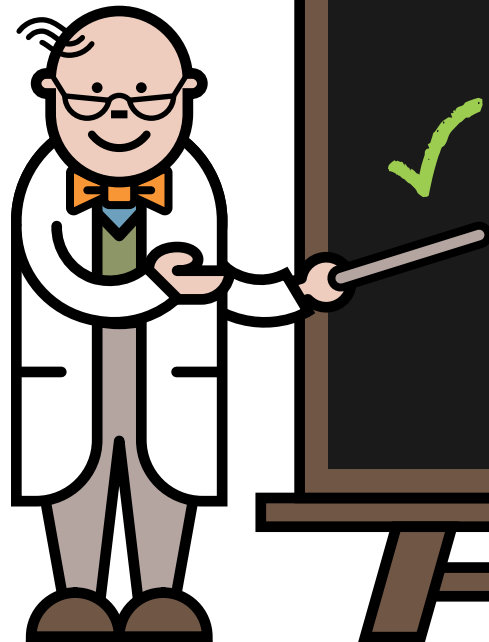
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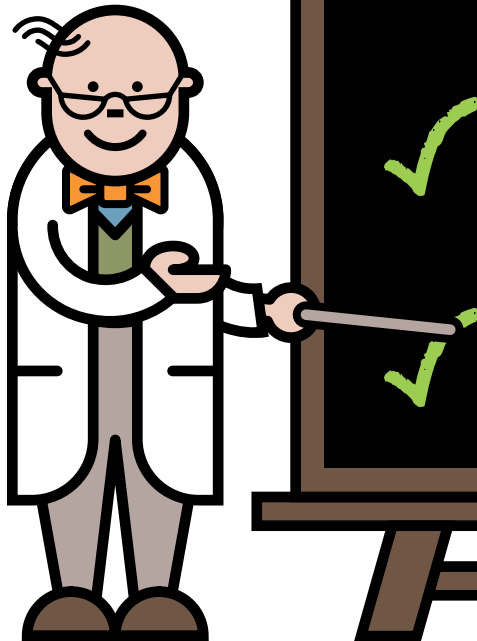
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LCHADD is a serious inherited metabolic condition that causes life threatening symptoms if left unmanaged



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

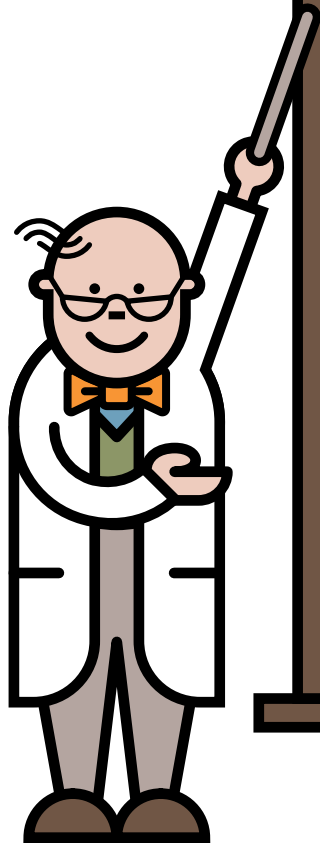


Children with LCHADD are managed with a low fat diet, MCT oil and regular meals



People with LCHADD should lead normal, healthy and active lives

# 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

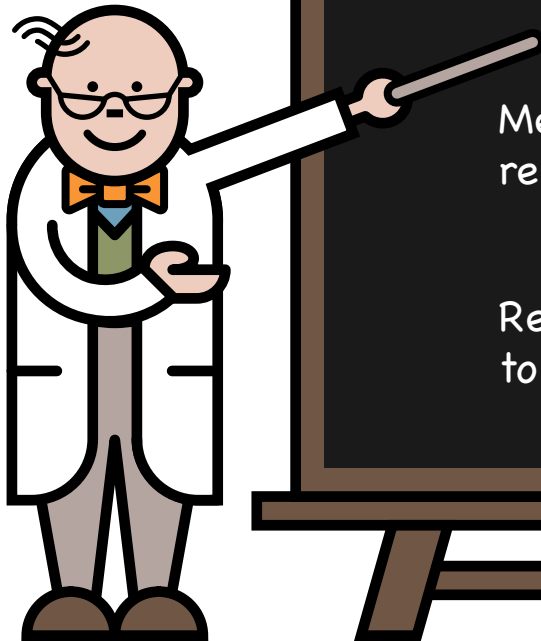
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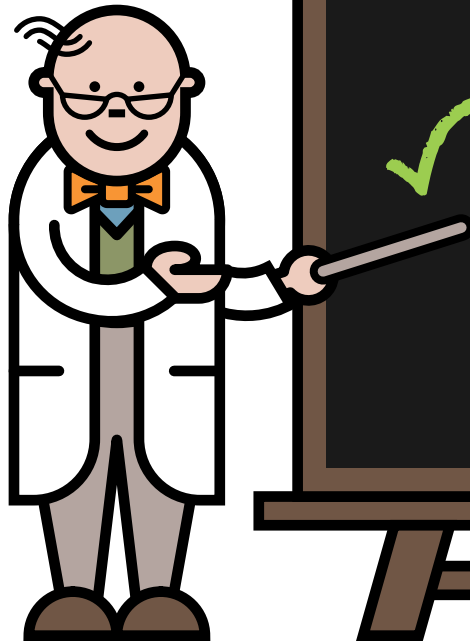
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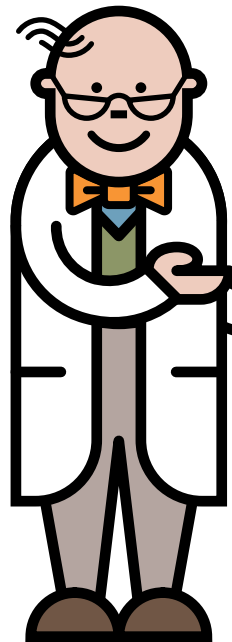
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# Who's who

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

**BIMDG**

British Inherited Metabolic Diseases Group



**NUTRICIA**  
LIFE-TRANSFORMING NUTRITION

**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)

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