

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

MCADD

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).

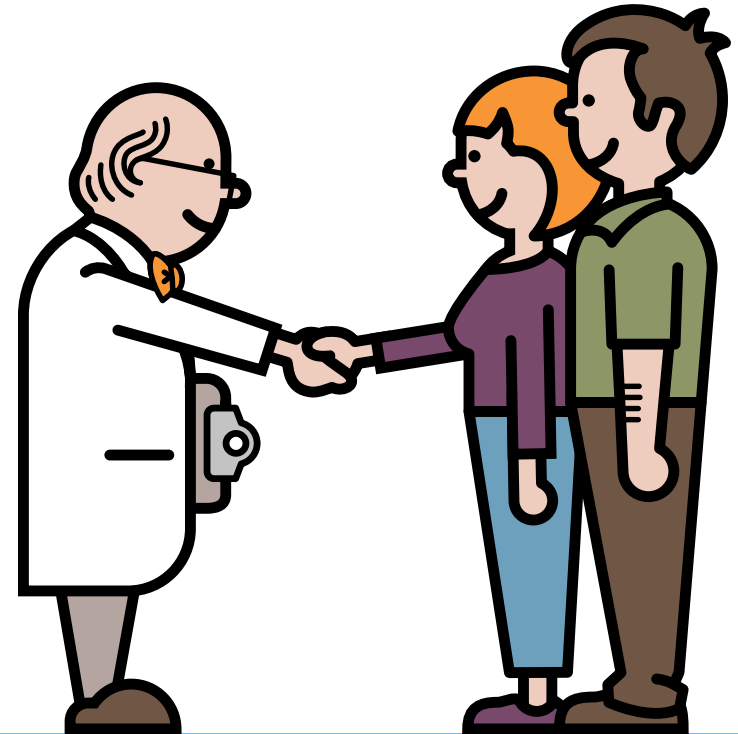
The team are Rachel Skeath, Karen van Wyk, Pat Portnoi and Anita MacDonald. The group is facilitated by Heidi Chan from Nutricia.

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.

MCAD Deficiency

Information for families following
a positive newborn screening



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Tools Enabling Metabolic Parents LEarning

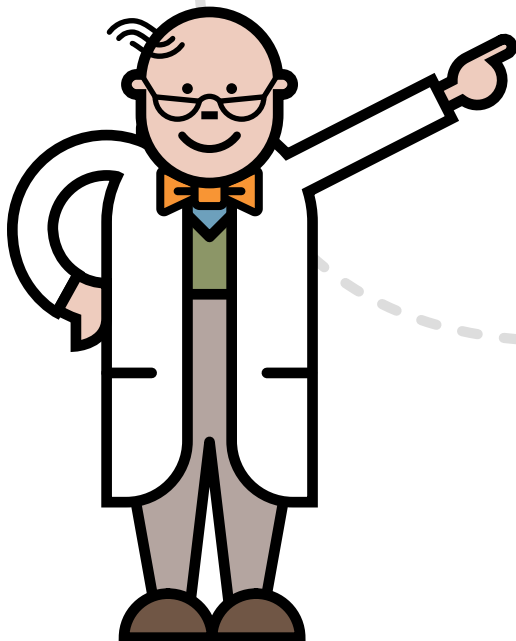
Supported by **NUTRICIA**
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What is MCADD?

MCADD stands for medium chain acyl-CoA dehydrogenase deficiency

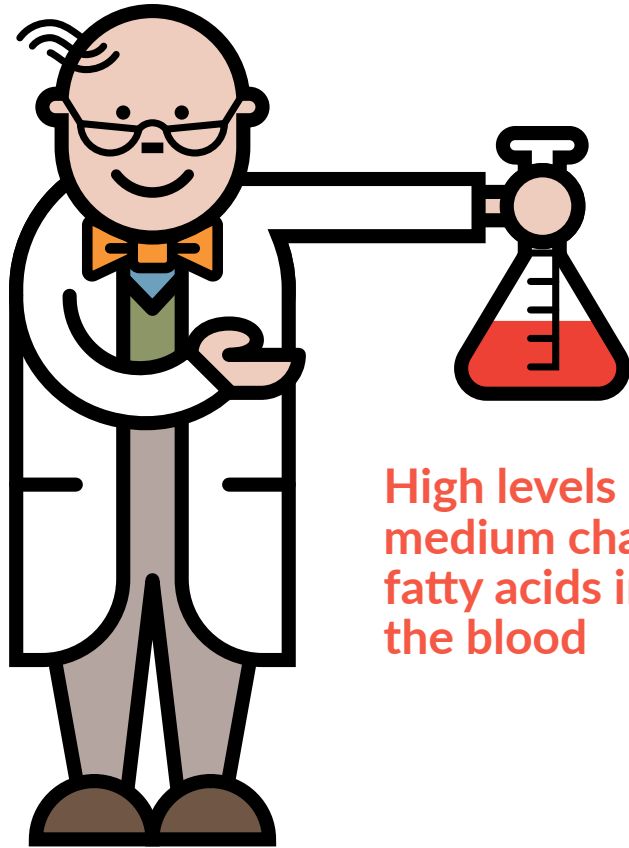
It is an inherited metabolic condition

Medium Chain Acyl-CoA Dehydrogenase

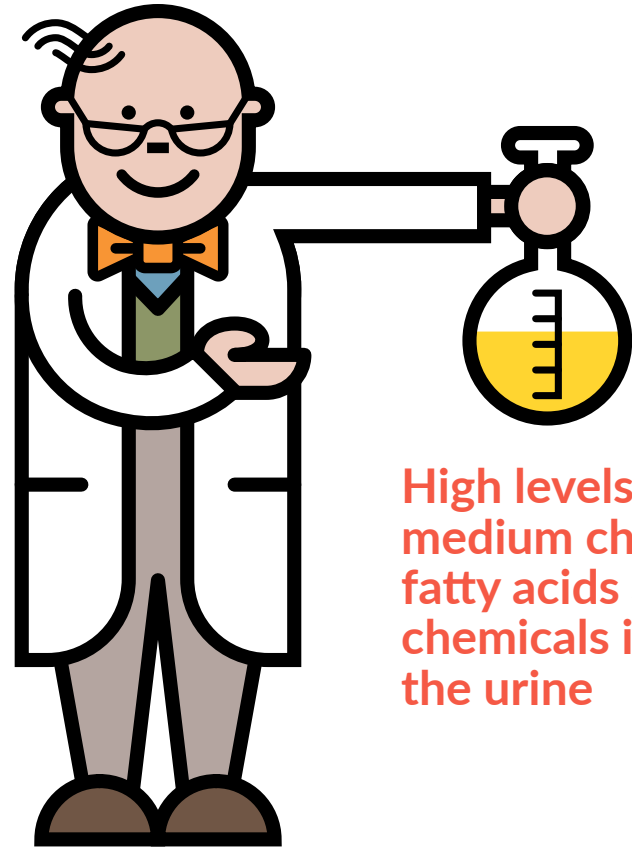


MCAD
Deficiency

What is MCADD?



High levels of
medium chain
fatty acids in
the blood

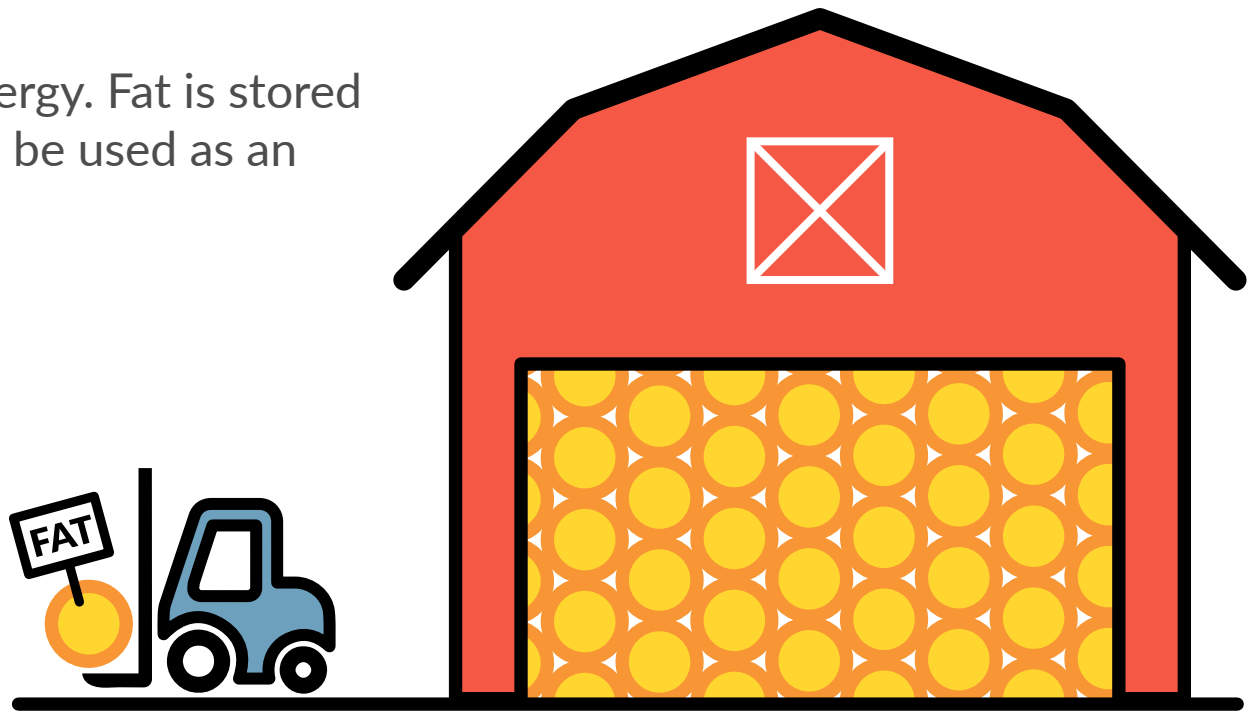


High levels of
medium chain
fatty acids and
chemicals in
the urine

Which foods supply the body with energy?

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

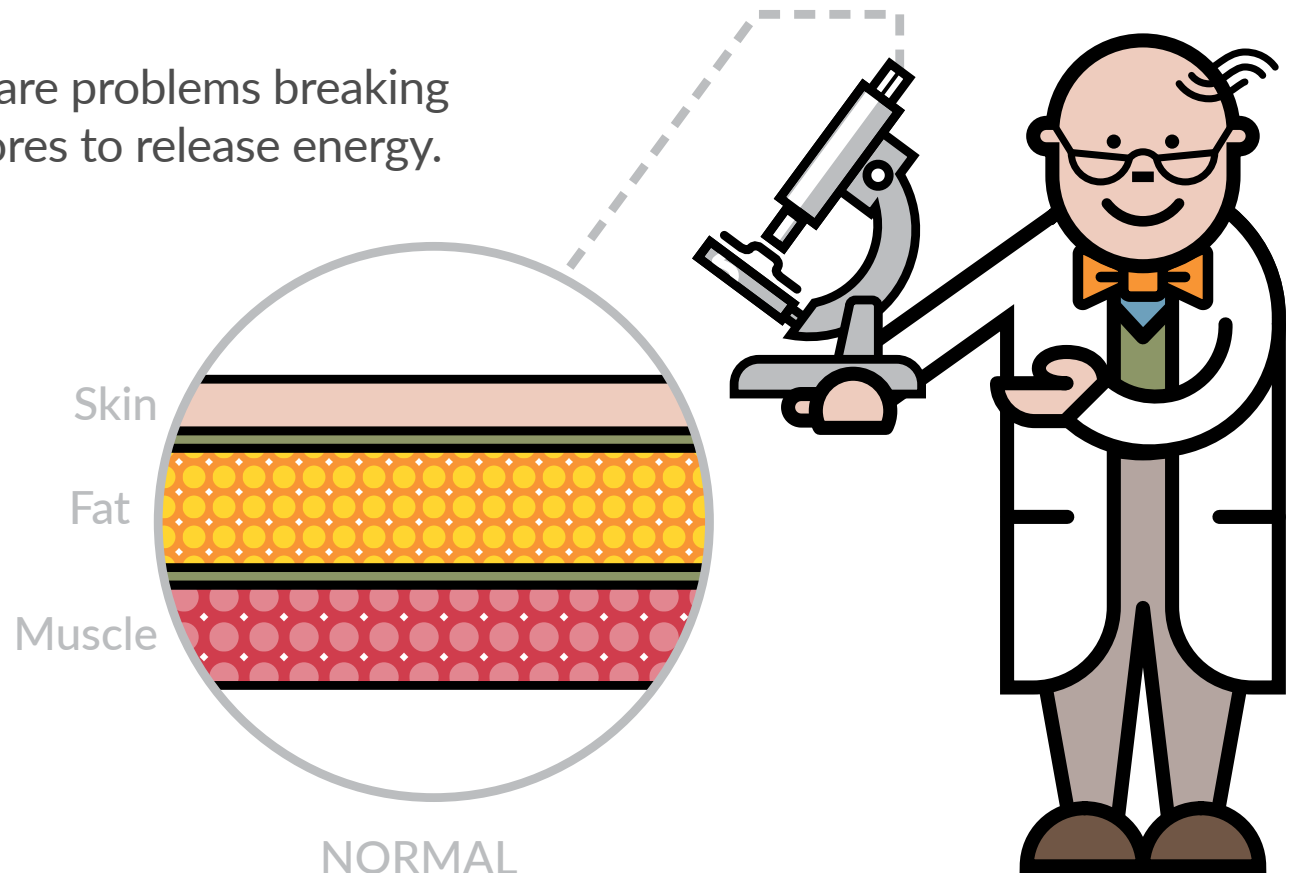
- Carbohydrates (starches and sugars) provide a readily available energy source
- Fats also provide energy. Fat is stored in the body so it can be used as an energy reserve



MCADD and fat

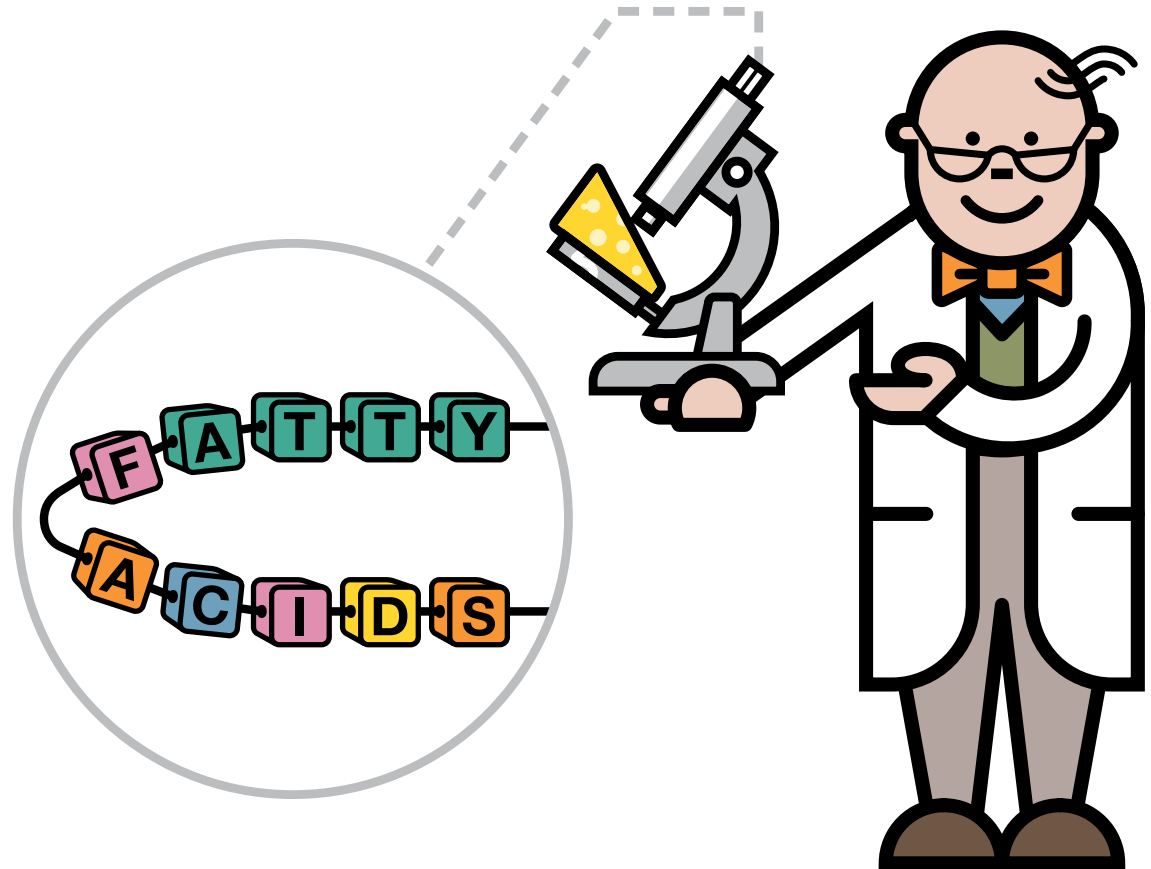
The body uses its own fat stores to provide energy when carbohydrate is depleted.

In MCADD, 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



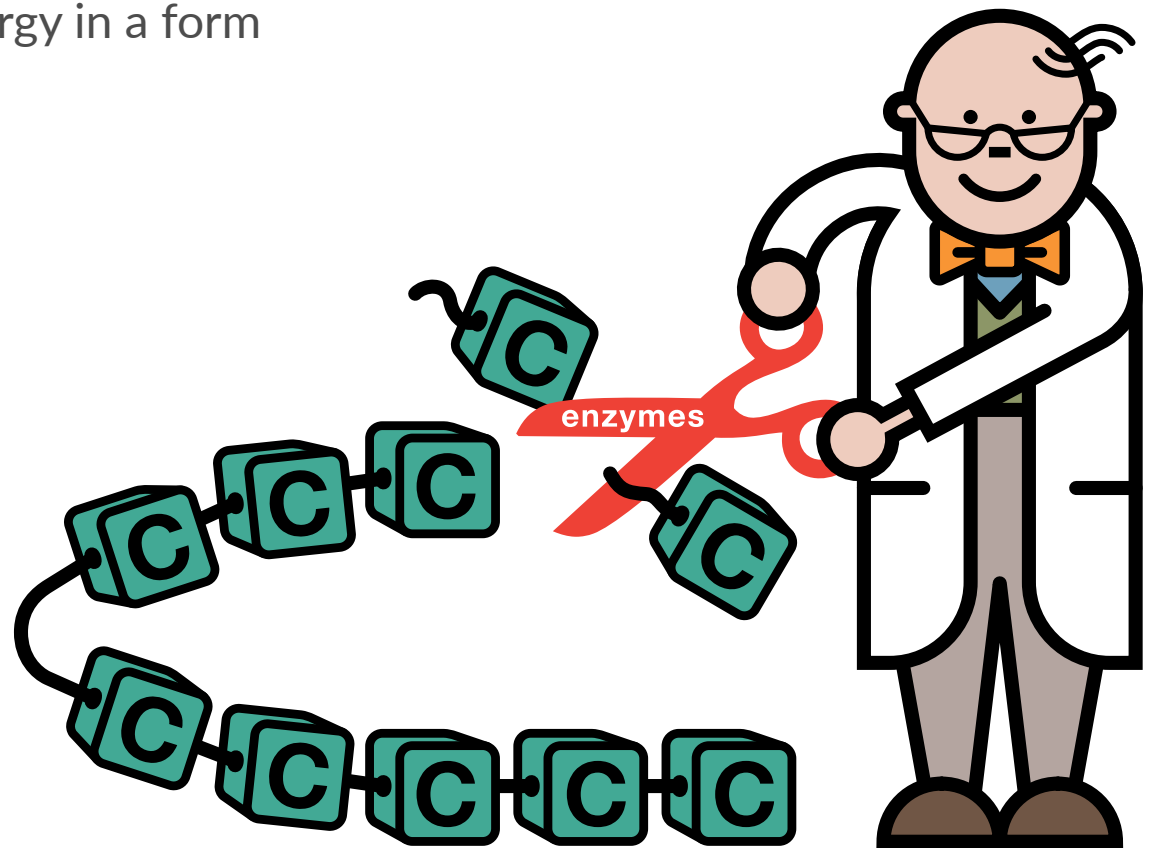
Medium chain



Long chain

Fatty 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.



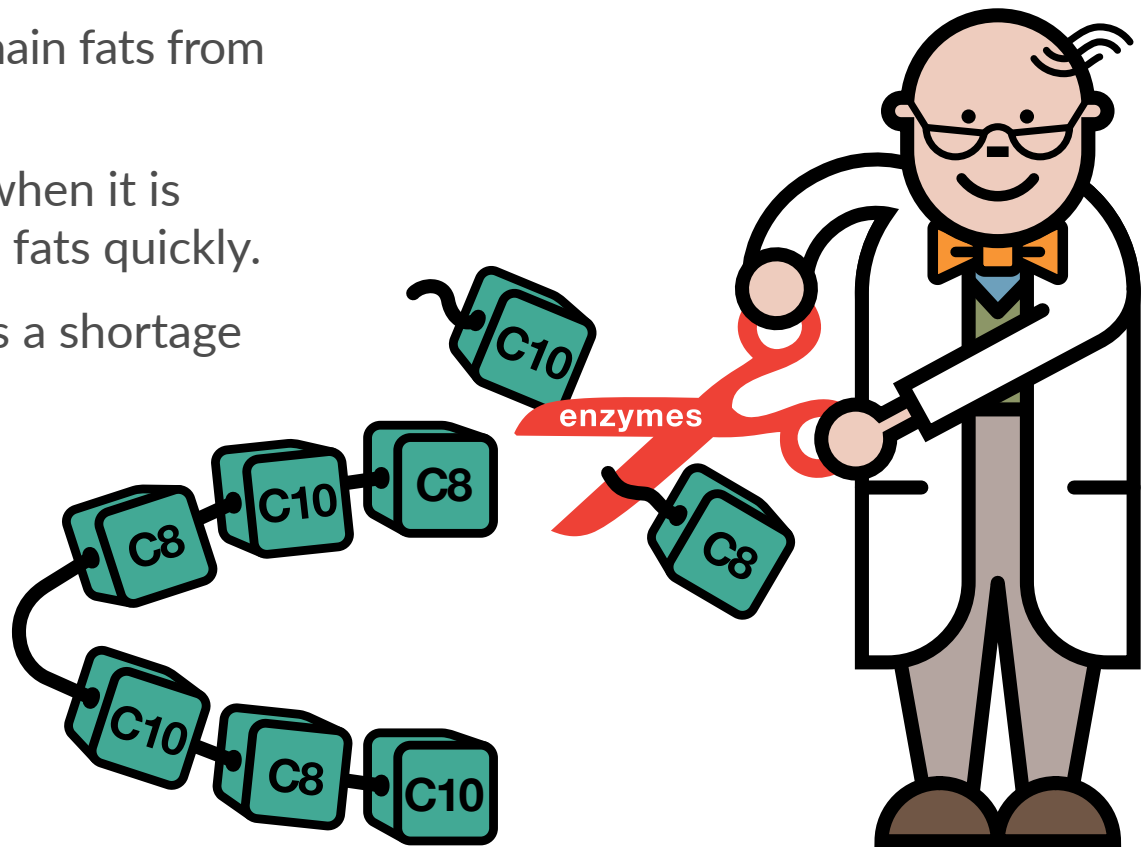
What happens in MCADD?

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

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

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

This occurs when there is a shortage of energy supply.



What can go wrong in MCADD?

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

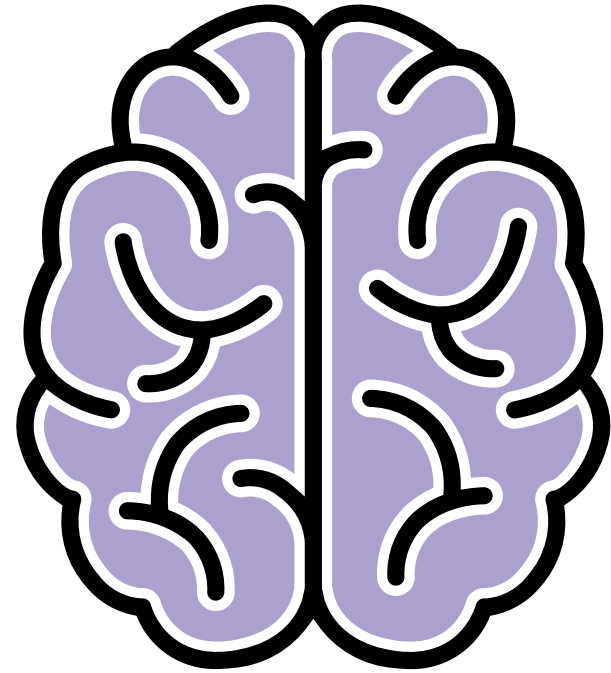
Symptoms include:

- poor feeding
- excessively sleepy
- rapid breathing
- seizures
- low blood sugar

What can go wrong in MCADD?

If shortage of energy supply is left unmanaged, 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.



Metabolic crisis

- A **metabolic crisis** triggers the MCADD symptoms
- This leads to a lack of energy supply and build up of toxic chemicals
- It is usually precipitated by childhood infections or viruses causing high temperatures, vomiting, and diarrhoea
- Avoidance of a metabolic crisis is essential



How is MCADD diagnosed?

MCADD is diagnosed by newborn screening. High levels of medium chain fatty acids and other chemicals are found in the blood.

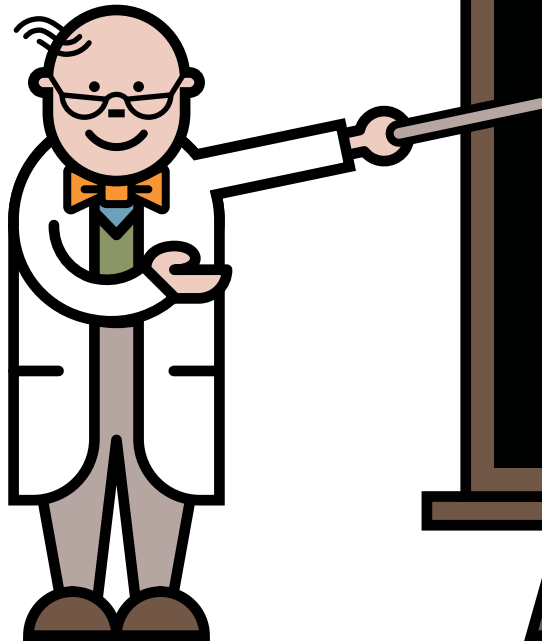


How is MCADD managed?

In babies, it is managed 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**.

The **safe fasting time** increases with age and it should be normal in older children when well.



SAFE FASTING TIMES

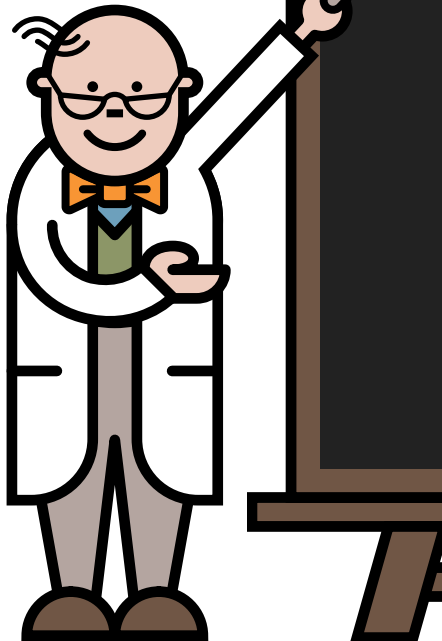
<u>Age</u>	<u>Time</u>
0–4 months	6 hours
4–8 months	8 hours
8–12 months	10 hours
Over 1 year	12 hours

How is MCADD managed day to day?

Breast milk or standard infant feed

A normal, healthy family diet for older babies and children

Regular meals

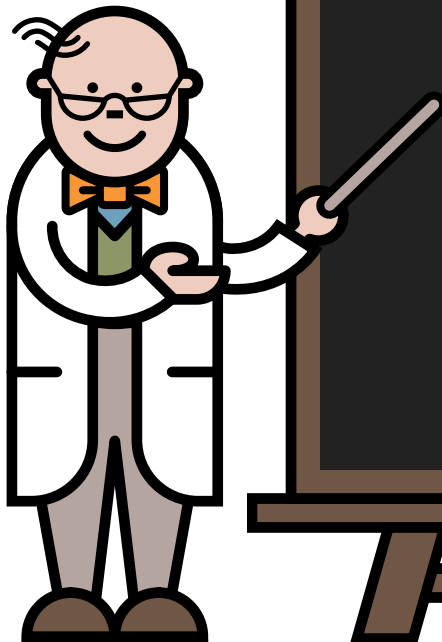


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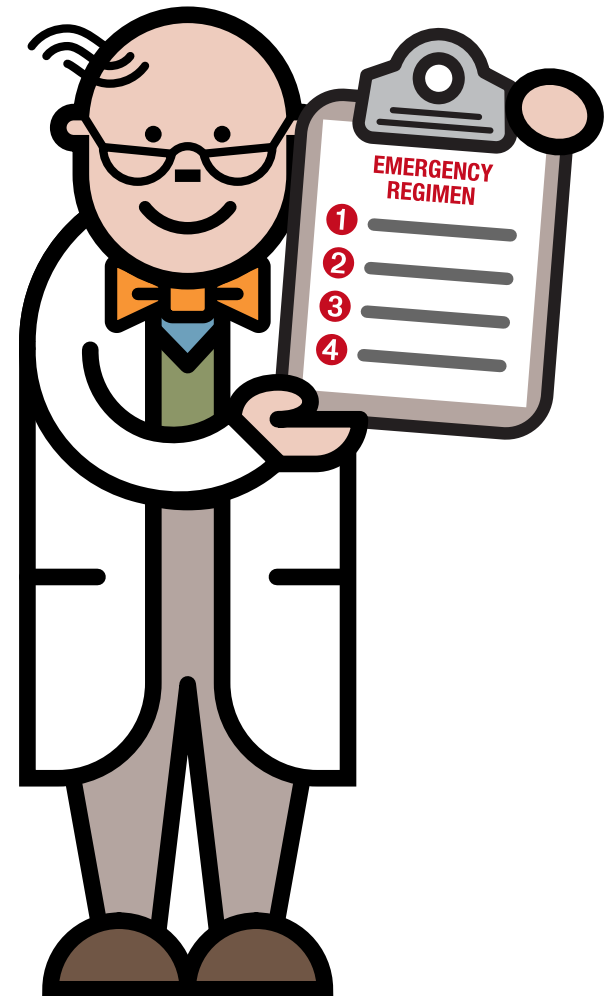
A normal, healthy family diet for older babies and children

Regular meals



How is MCADD 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 MCADD managed during illness?

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



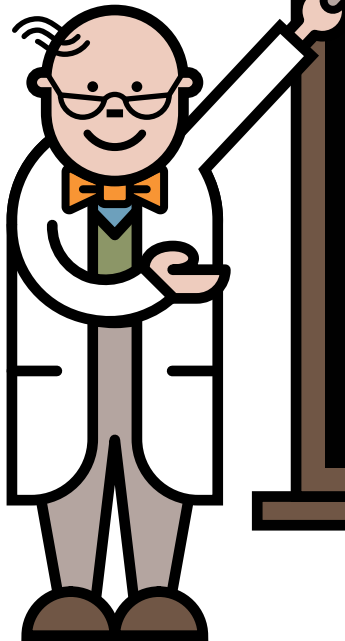
This must be given regularly
day and night



Contact your metabolic team
for help if your child is unwell



Do not use sugar-free drinks during illness



How is MCADD managed during illness?

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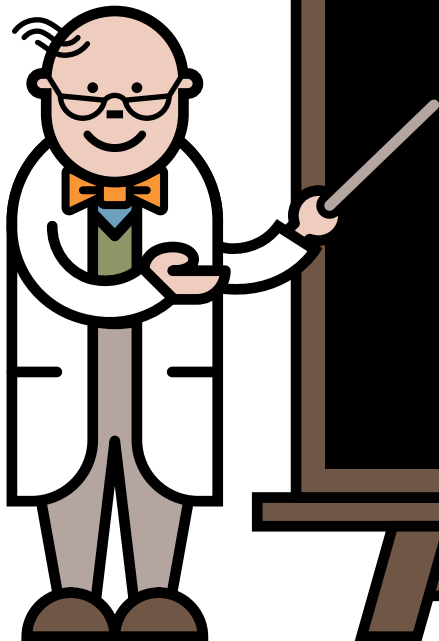
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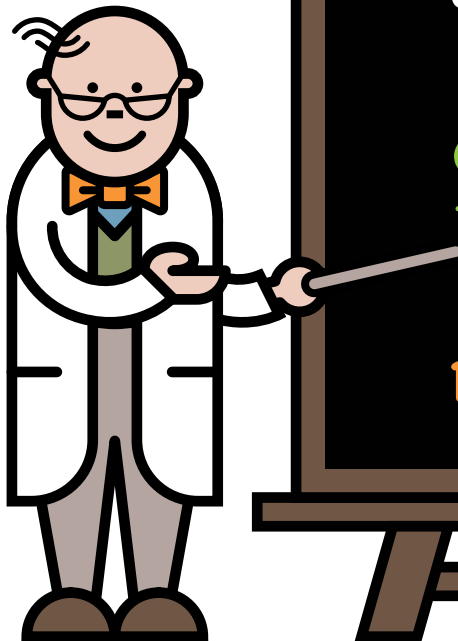
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Checklist for illness

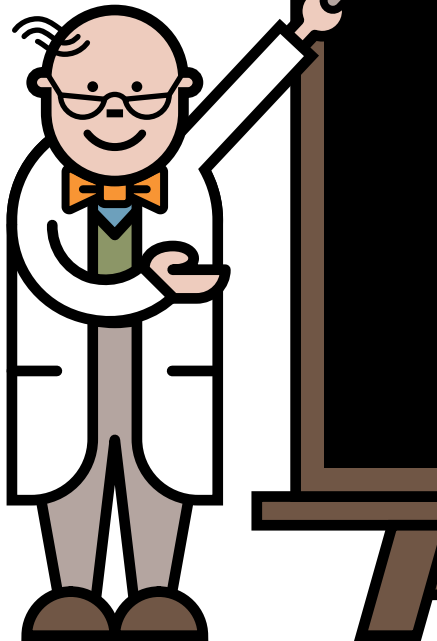
Always take full amounts of emergency feeds as prescribed



If symptoms continue and/or you are worried, go immediately to the hospital



Phone your metabolic team



Checklist for illness

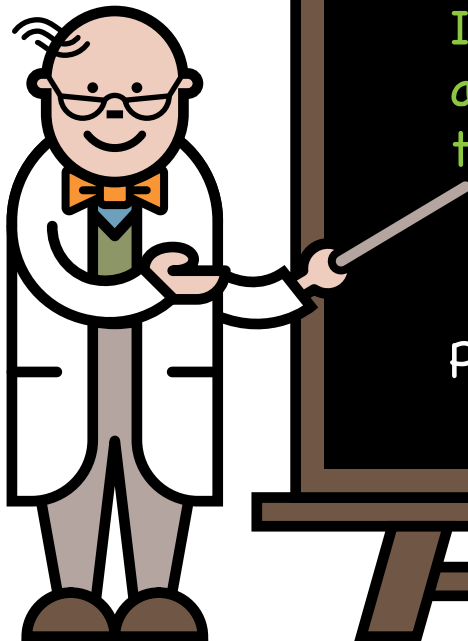
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Checklist for illness

Always take full amounts of emergency feeds as prescribed



If symptoms continue and/or you are worried, go immediately to the hospital



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Key message

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



How is MCADD monitored?

Blood tests to check chemical levels

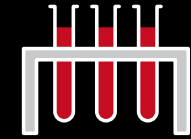
Height and weight

Developmental checks

Emergency feeds are adjusted with age

How is MCADD monitored?

Blood tests to check
chemical levels



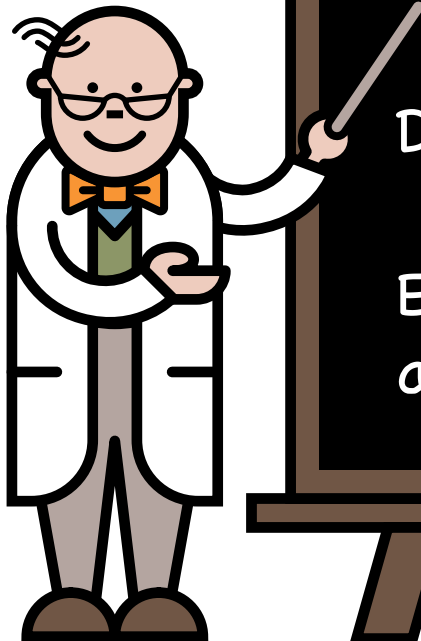
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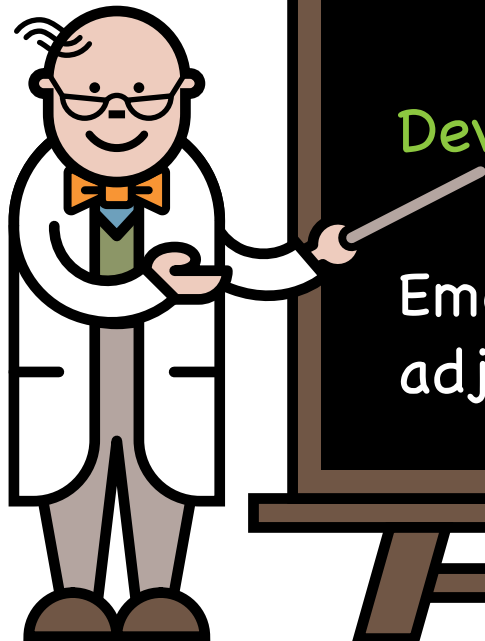
Developmental checks



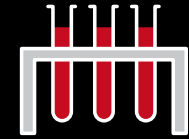
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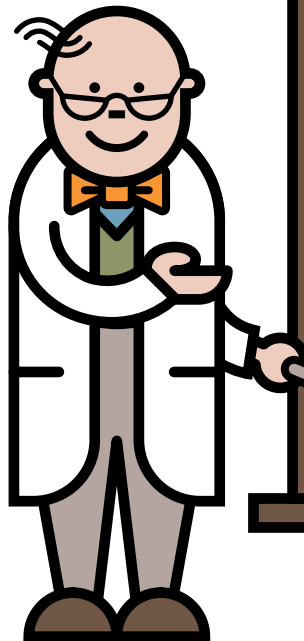
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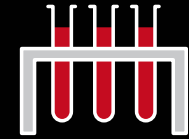
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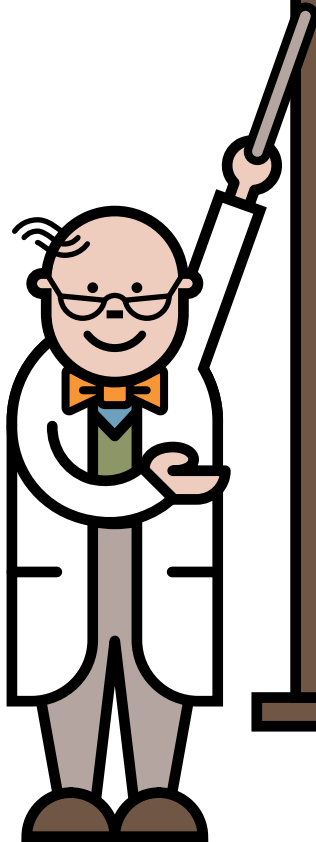
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Chromosomes, genes, mutations



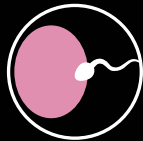
Humans have chromosomes composed of DNA



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



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

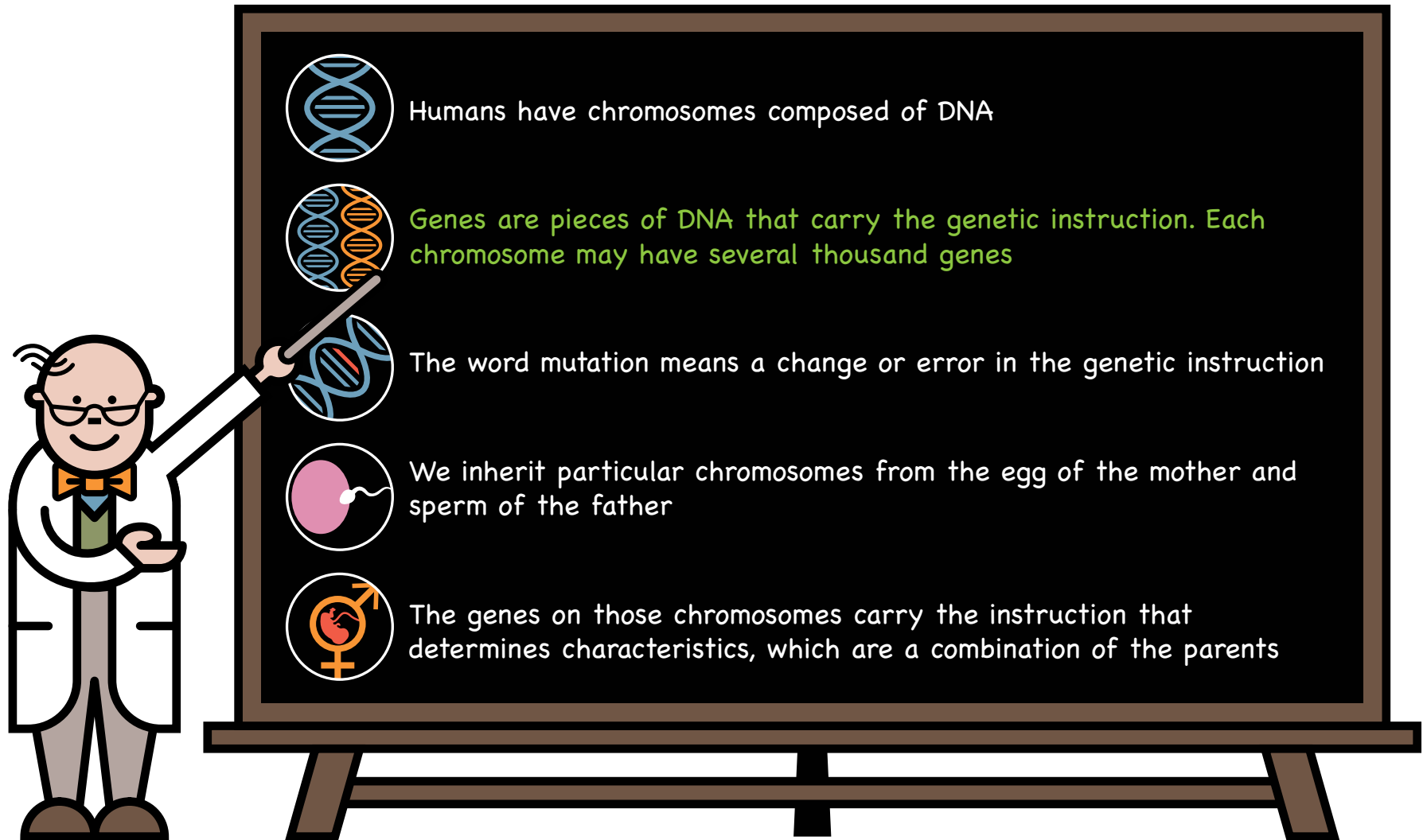



We inherit particular chromosomes from the egg of the mother and sperm of the father





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


Chromosomes, genes, mutations




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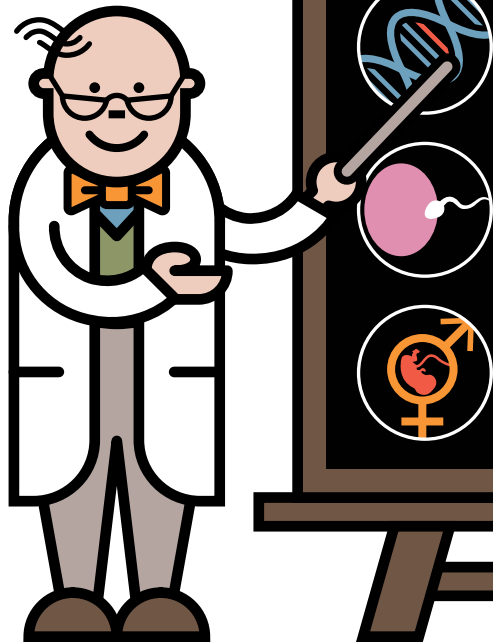
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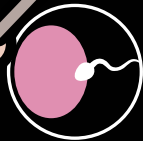
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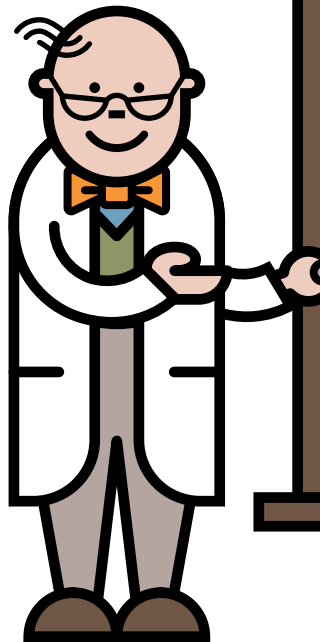


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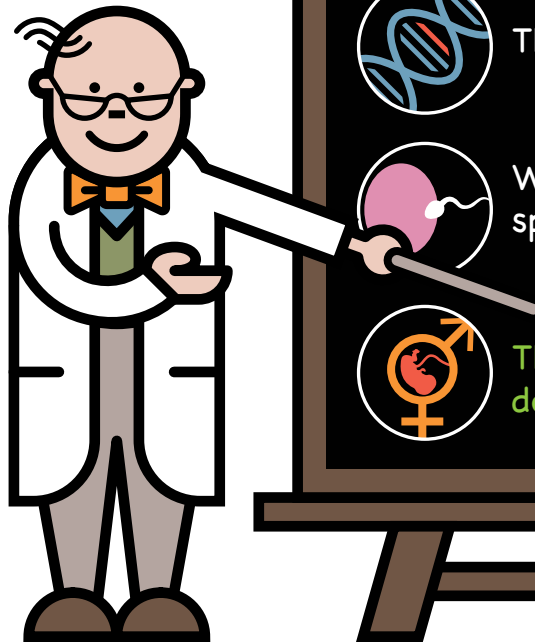


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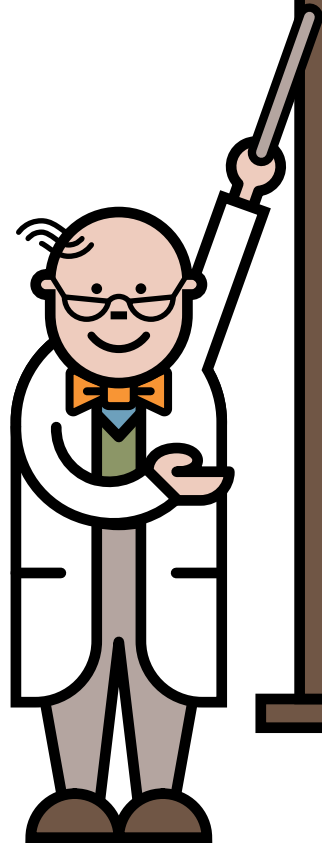


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Inheritance



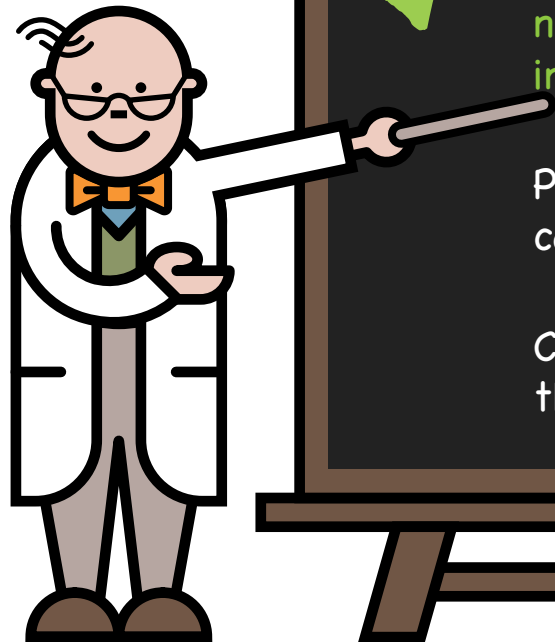
MCADD is an inherited condition. There is nothing that could have been done to prevent your baby from having MCADD

Everyone has a pair of genes that make the medium chain acyl-CoA dehydrogenase enzyme. In children with MCADD, neither of these genes works correctly. These children inherit one non-working MCADD gene from each parent

Parents of children with MCADD are carriers of the condition

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

Inheritance



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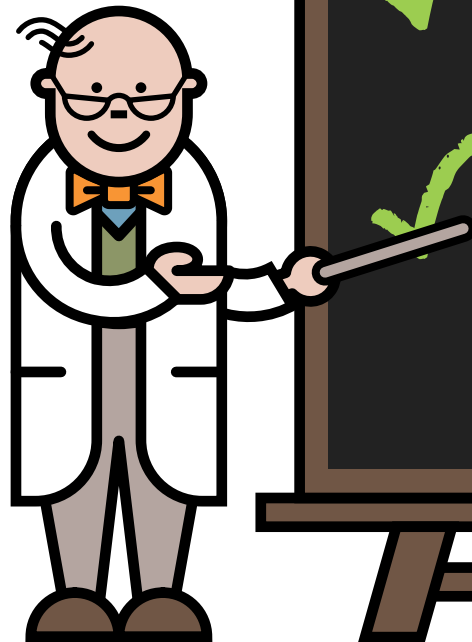


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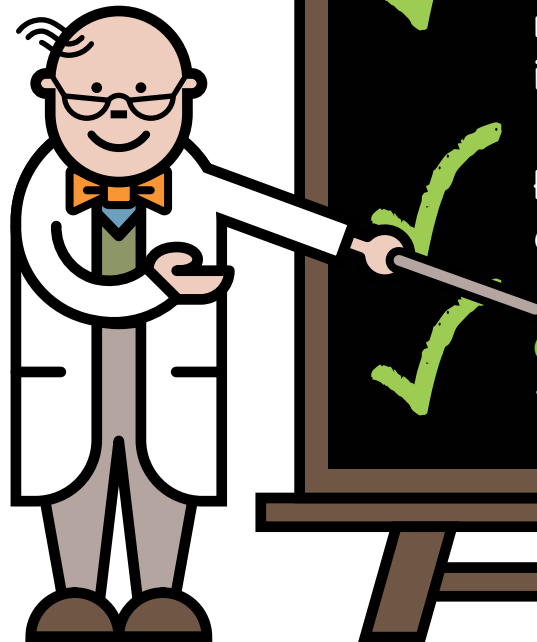
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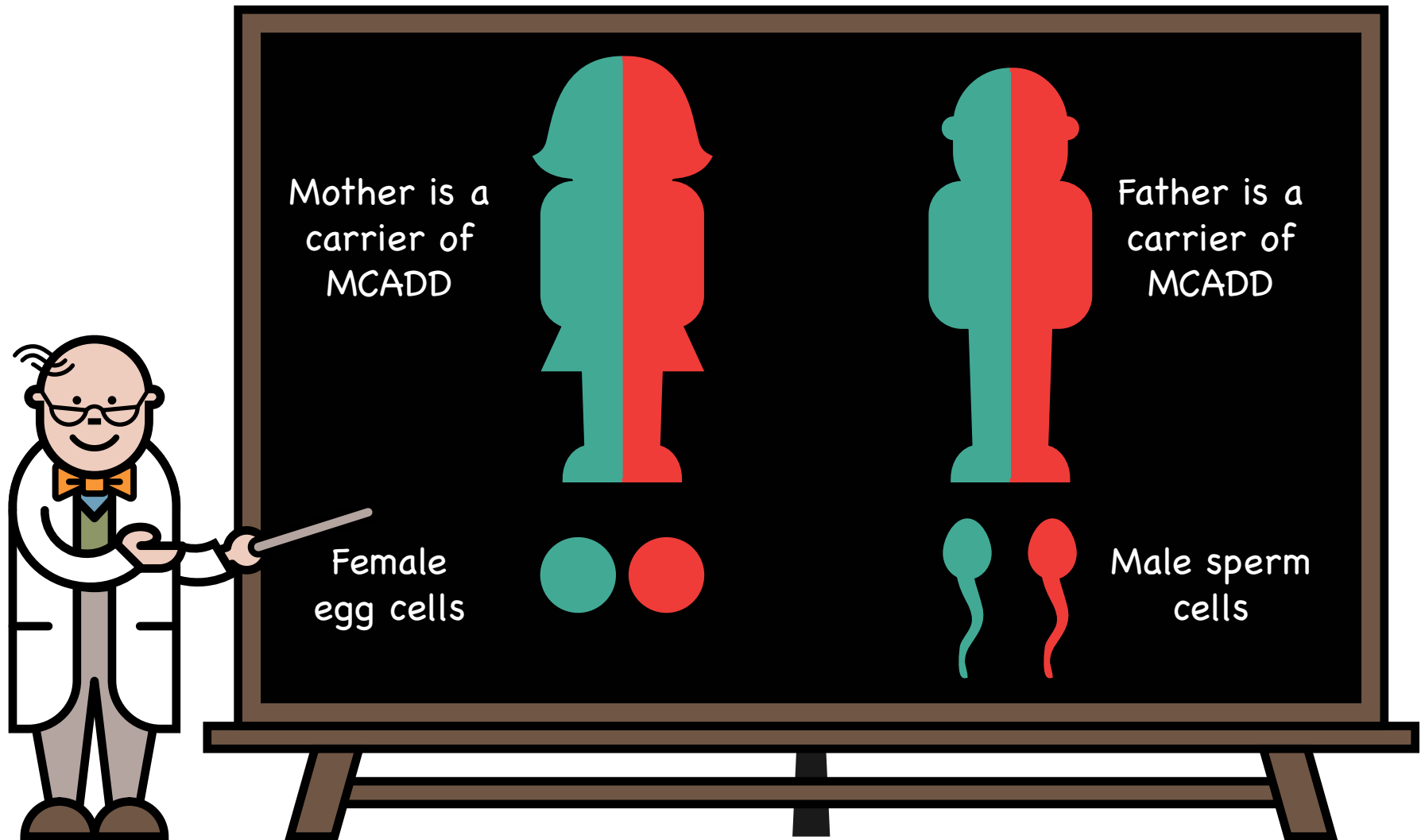
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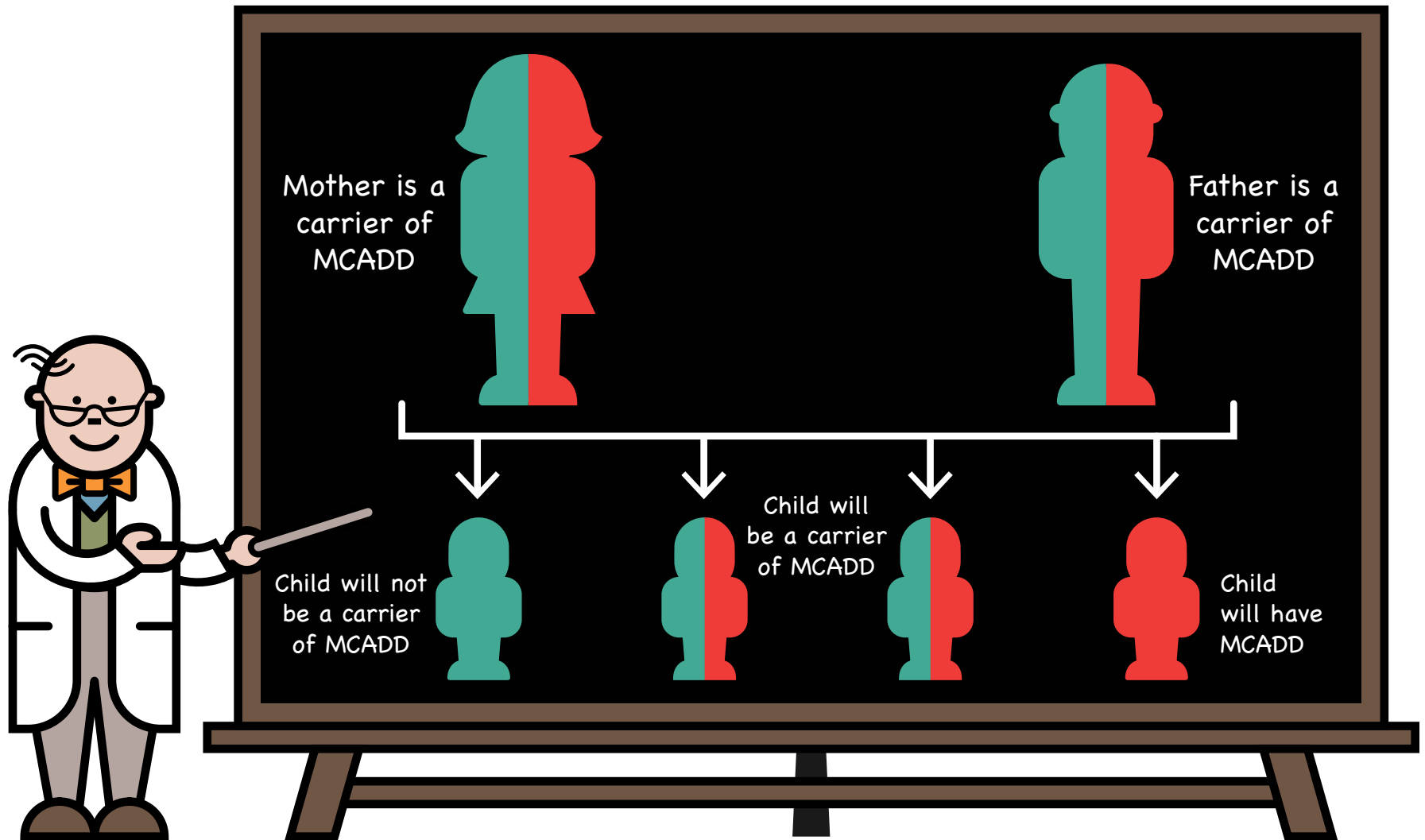
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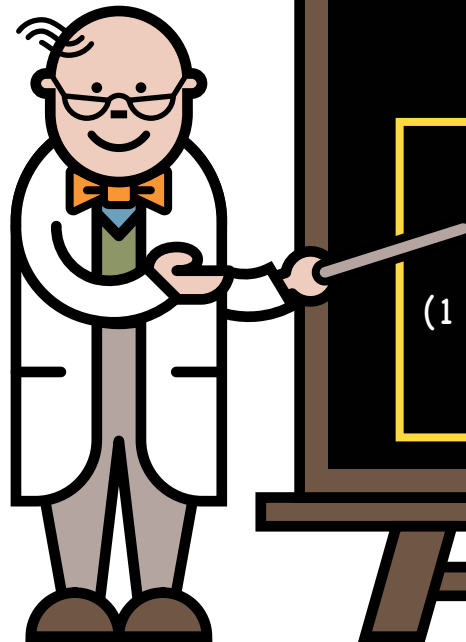
Inheritance – Autosomal recessive (carriers of MCADD)



Inheritance – Autosomal recessive – possible combinations



Future pregnancies



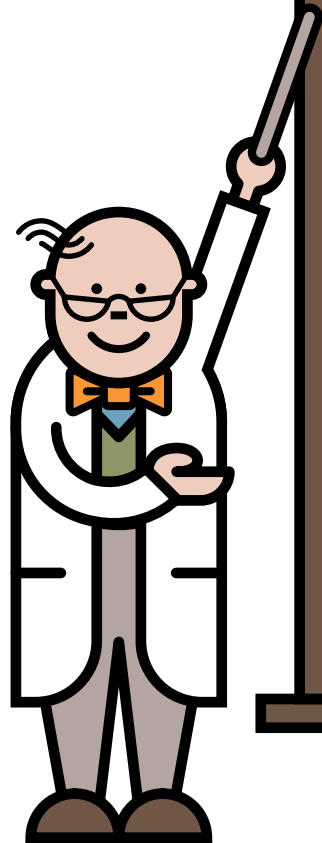
When both parents are carriers, in each pregnancy the risk to the baby is as follows:

25% chance
(1 in 4) of MCADD

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

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

Take home messages



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

Damage can be prevented with timely and appropriate use of emergency drinks during illness and avoiding long periods without eating

When babies and children are well, no special diet is needed

MCADD is easily managed and people should lead normal, healthy and active lives

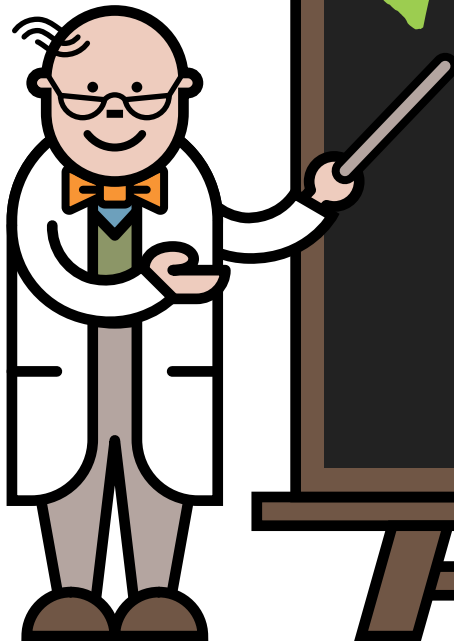
Take home messages

✓ MCADD is a serious inherited, metabolic disorder that may cause life threatening symptoms if left unmanaged

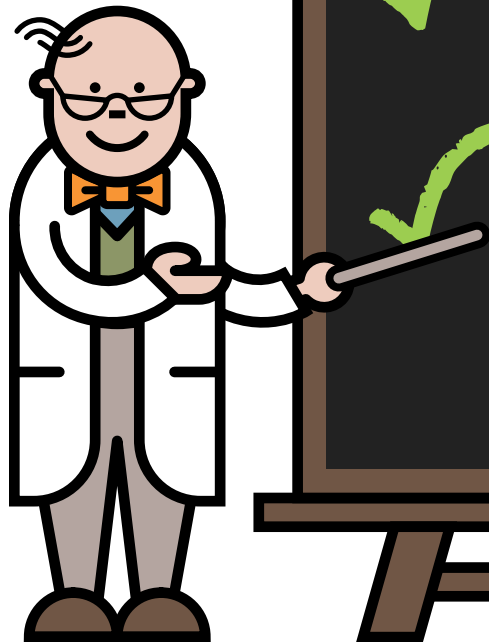
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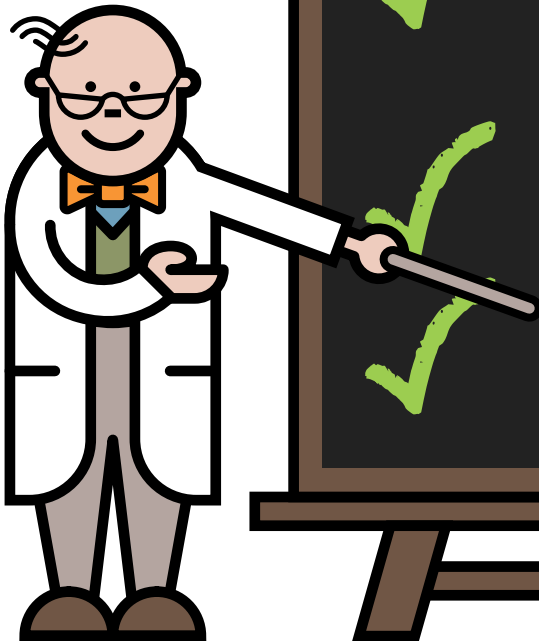
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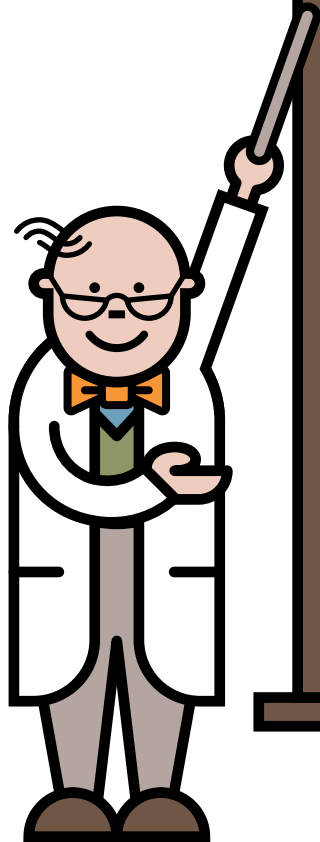
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Helpful hints



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

Your glucose polymer powder is prescribed by your GP and you obtain it 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



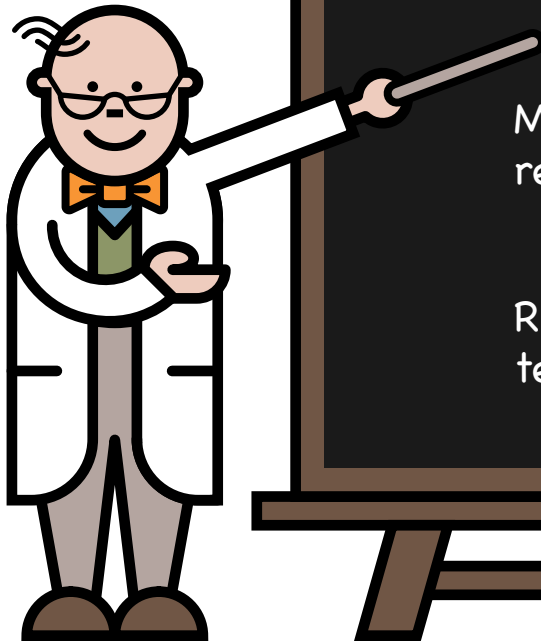
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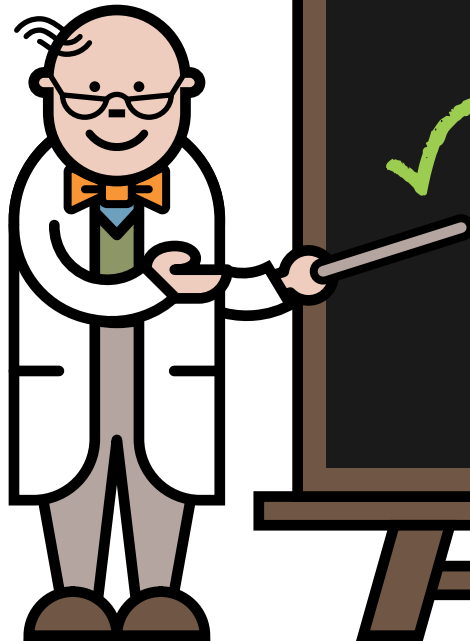
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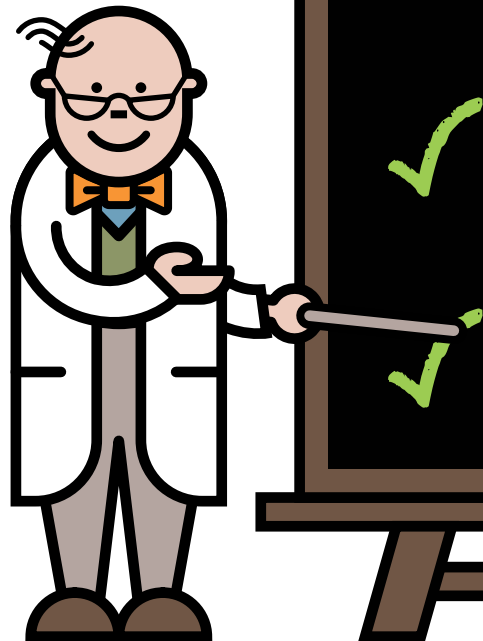
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Who's who

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

BIMDG

British Inherited Metabolic Diseases Group



www.bimdg.org.uk

The Nutricia logo features a blue arc above the word "NUTRICIA" in a bold, blue, sans-serif font. Below it, the tagline "LIFE-TRANSFORMING NUTRITION" is written in a smaller, blue, sans-serif font.

NUTRICIA
LIFE-TRANSFORMING NUTRITION

www.nutricia.co.uk

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