MCAD Deficiency
Information for families following a positive newborn screening

ADAPTED BY THE DIETITIANS GROUP

BIMDG
British Inherited Metabolic Diseases Group

BASED ON THE ORIGINAL TEMPLE WRITTEN BY BURGARD AND WENDEL
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Supported by as a service to metabolic medicine
What is MCADD?

MCADD stands for medium chain acyl-CoA dehydrogenase deficiency

It is an inherited metabolic condition
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 untreated, 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 treated?

In babies, it is treated 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.
How is MCADD managed day to day?

Breast milk or standard infant formula

A normal, healthy family diet for older babies and children

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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.
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Do not use sugar-free drinks during 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

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Phone your metabolic team
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
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Height and weight

Developmental checks

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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
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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.
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Inheritance – Autosomal recessive (carriers of MCADD)

Mother is a carrier of MCADD

Father is a carrier of MCADD

Female egg cells

Male sperm cells
Inheritance – Autosomal recessive – possible combinations

Mother is a carrier of MCADD

Father is a carrier of MCADD

Child will not be a carrier of MCADD

Child will be a carrier of MCADD

Child will have MCADD
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
MCADD is a serious inherited metabolic condition that causes life threatening symptoms if left untreated.

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 treated and people should lead normal, healthy and active lives.
MCADD is a serious inherited, metabolic disorder that may cause life threatening symptoms if left untreated.

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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
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Who’s who

• My dietitians
• My nurses
• My doctors
  – Contact details, address, photos