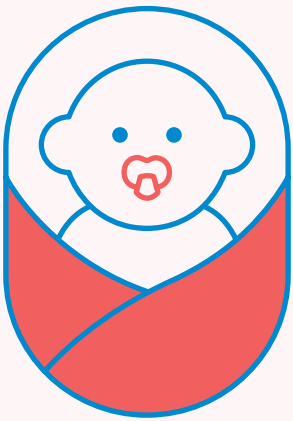
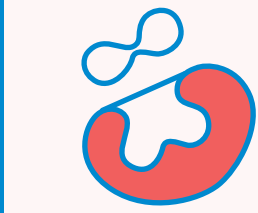
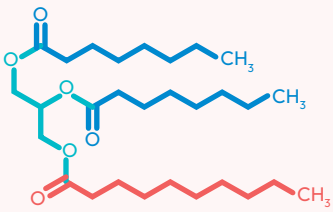
SingHealth DukeNUS
ACADEMIC MEDICAL CENTRE

Genomic
Medicine Centre

Medium-Chain Acyl-CoA Dehydrogenase Deficiency







SingHealth

What is medium-chain acyl-CoA dehydrogenase deficiency?

Medium-chain acyl-CoA dehydrogenase deficiency (MCADD) is an inherited metabolic disorder in which the body is unable to break down certain forms of fat to produce energy.

Enzymes help to break down food that we eat, and certain enzymes break down fats into their building blocks – fatty acids.

Fatty acids are built like chains and come in many lengths such as short, medium, long or very long. The enzyme called medium-chain acyl-CoA dehydrogenase is in charge of breaking down medium-length fatty acids.

In individuals with MCADD, this enzyme is either missing or not working properly. Hence, they are unable to turn medium-length fatty acids into energy.

This is harmful as fat is needed for energy when their body runs out of sugar, such as during prolonged periods without food (fasting) or illness. In such instances, as the body is unable to utilise fat as a source of energy, the individual is at risk of becoming very sick.

How common is MCADD?

MCADD is one of the most common fatty acid oxidation disorders. It is estimated that **one in 5,000 to 12,000 infants** are born with MCADD.

What are the symptoms of MCADD?

Most people with MCADD do not show any symptoms unless they are sick, vomiting or not eating well.

Symptoms or signs that may be seen during illness or times of fasting:

Lethargy – tired and sleepy

Hypoglycaemia (low blood sugar)

Vomiting

Shakiness

Irritability

Left untreated, this can progress to:

Heart problems

Enlarged liver (hepatomegaly)

Seizures

Breathing problems

Coma

Death

The health outcome for most individuals with MCADD is excellent. Growth, development and general health are expected to be normal if the diagnosis is made early and emergency treatment is given during periods of illness or poor feeding.

How is MCADD managed?

The management of MCADD includes:

- Avoidance of fasting
- Use of emergency treatment protocol
- Use of emergency regimen (provided by dietitian)
- Carnitine supplementation in some cases (if carnitine level is low)
- Intravenous glucose drip in the hospital if the child vomits or refuses to eat
- Avoidance of concentrated sources of medium-chain triglycerides (MCT), like MCT oil and specialised products with high MCT oil content

Is there any monitoring required?

As MCADD is a lifelong condition, there will be regular monitoring of growth and development, and assessment of diet and nutrition.

Most people with MCADD, with appropriate care and management, go on to live long, healthy lives.

What is an emergency letter and when do I need it?

An emergency letter is a letter provided by the metabolic clinic. If you have not received a letter, please contact your metabolic clinic and ask for one.

Bring the letter to the emergency department in the event of an illness or poor eating.

When should I bring my child to seek medical attention?

Seek medical attention if your child:

Is vomiting

Is hard to wake up

Has diarrhoea

Refuses to eat due to nausea or sore throat

Has an infection

Is scheduled for medical or dental procedures or prior to any surgery

Has a fever

What causes MCADD?

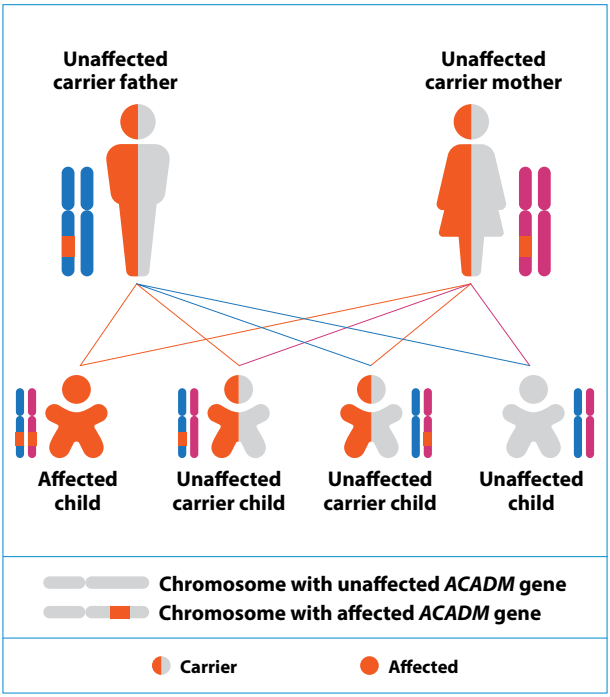
MCADD is caused by a mutation in the *ACADM* gene. This gene gives the body instructions for making an enzyme called medium-chain acyl-CoA dehydrogenase, needed to break down fats into energy.

How is MCADD diagnosed?

In the past, individuals with MCADD were diagnosed when they presented with symptoms. However, with newborn screening, babies can now be screened for MCADD after 24 hours of life, when they are asymptomatic and well.

If the newborn screening indicates suspected MCADD, additional testing via blood and urine will be needed to confirm if the child has MCADD.

How is MCADD inherited?



Everyone has two copies of each gene in their body's cells, one copy from each parent. MCADD follows a **recessive inheritance pattern**. This means that a patient with MCADD must have two faulty copies of *ACADM* to cause features of MCADD.

An individual with one faulty copy of *ACADM* is known as a carrier. Typically, carriers of an autosomal recessive condition do not have any signs or symptoms.

How likely will I have another child with MCADD?

Parents who are both carriers of a disease-causing variant in the *ACADM* gene have a 25% chance of having an offspring with MCADD.

MCADD is a lifelong condition. Should you require financial assistance or emotional support, please approach your doctor for referral to a medical social worker.

Support Group

FOD (Fatty Oxidation Disorders) Family Support Group
FOD Family Support Group is an international organisation that supports and connects FOD families and professionals around the world.
www.fodsupport.org

Acknowledgements

Genetics Service, KK Women's and Children's Hospital

Nutrition & Dietetics Department,
KK Women's and Children's Hospital

Information contributed by:

SingHealth DukeNUS ACADEMIC MEDICAL CENTRE

Genomic Medicine Centre

KK Women's and Children's Hospital
6294 4050
www.kkh.com.sg

Singapore General Hospital
6321 4377
www.sgh.com.sg

Changi General Hospital
6850 3333
www.cgh.com.sg

Sengkang General Hospital
6930 6000
www.skh.com.sg

National Cancer Centre Singapore
6436 8088
www.nccs.com.sg

National Heart Centre Singapore
6704 2000
www.nhcs.com.sg

National Neuroscience Institute
6321 4377 (SGH Campus)
6330 6363 (TTSH Campus)
www.nni.com.sg

Singapore National Eye Centre
6227 7266
www.snec.com.sg

Find out more about the Centre at:
www.singhealth.com.sg/genomic-medicine-centre

SingHealth
www.singhealth.com.sg

SingHealth Hospitals

Singapore General Hospital | Changi General Hospital | Sengkang General Hospital | KK Women's and Children's Hospital

National Specialty Centres

National Cancer Centre Singapore | National Dental Centre Singapore | National Heart Centre Singapore | National Neuroscience Institute | Singapore National Eye Centre

SingHealth Community and Primary Care

Polyclinics SingHealth | SingHealth Community Hospitals

Reg. No.: 200002698Z
Information correct as at February 2023

Disclaimer: *The information provided does not replace information from your healthcare professional. Please consult your healthcare professional for more information.*

Scan for a digital copy