Newborn Screening Programme for
Inborn Errors of Metabolism
Information leaflet series (No. 19)

Medium-chain Acyl-CoA Dehydrogenase Deficiency

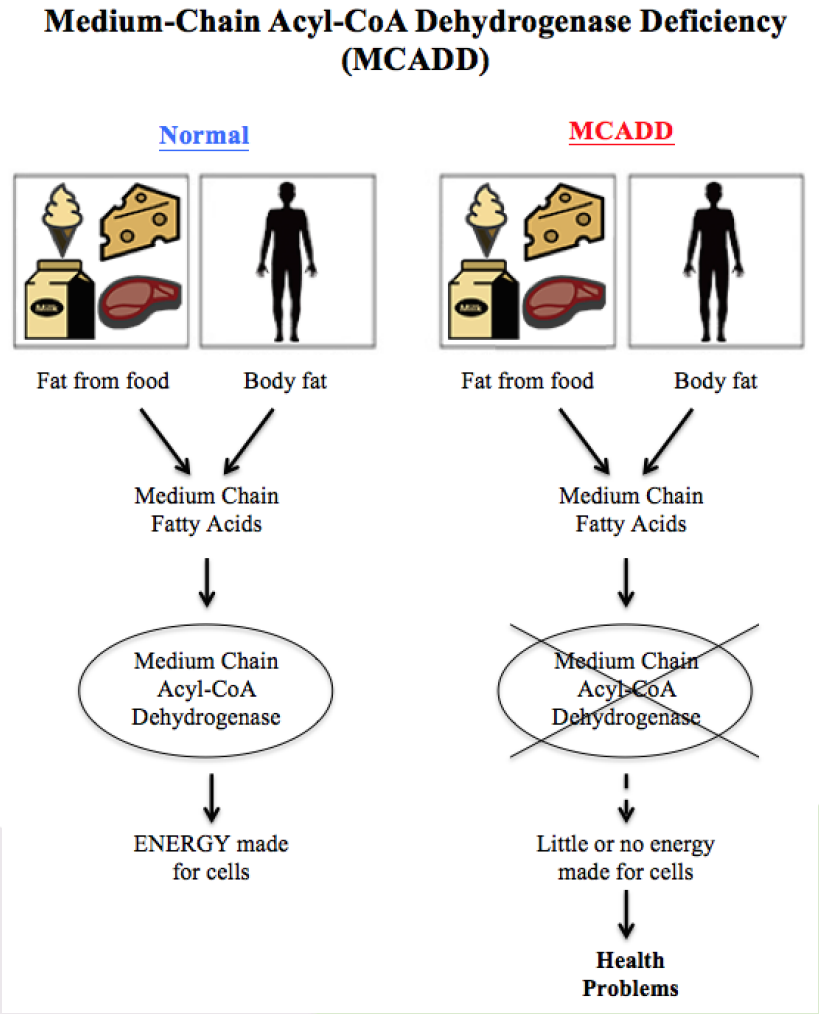
For general queries on Newborn Screening Programme for Inborn Errors of Metabolism, please call: ☎ 5741 4280 (Department of Clinical Genetics, Hospital Authority)

醫院管理局
HOSPITAL AUTHORITY

What is Medium-chain acyl-CoA dehydrogenase deficiency (MCADD)?

MCADD is an inherited fatty acid oxidation disorder caused by deficiency or ineffective functioning of an enzyme called "medium-chain acyl-CoA dehydrogenase" (MCAD). Without this enzyme, certain fats called "medium-chain fatty acids" cannot be metabolized.

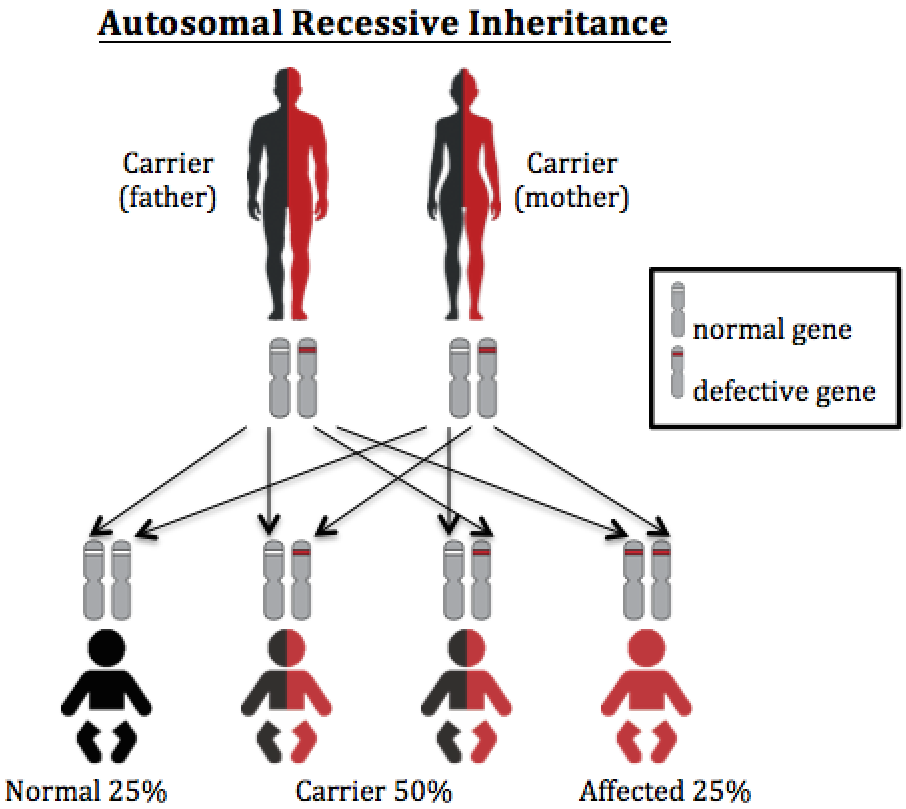
Fatty acids are a major source of energy for the heart and muscles. During periods of fasting, fatty acids are also an important energy source for the liver and other tissues. People with MCADD rely on glucose for main source of energy. When glucose has been used up, for example after skipping a meal or no eating for a long time, the body will try to use fat for energy but without success. This can cause low blood sugar level (hypoglycaemia). At the same time, medium-chain fatty acids or other partially metabolized fatty acids may build up in tissues and cause damage to the body.



How is MCADD inherited?

Everybody has two copies of genes, one from each parent, which tell the body how to make specific enzymes.

Medium-chain acyl-CoA dehydrogenase deficiency is an autosomal recessive disease. Only when babies inherit two faulty copies of the gene for medium-chain acyl-CoA dehydrogenase deficiency from parents, the enzyme made does not work properly or is not even made at all.



What may happen if your baby has MCADD?

With prompt and careful treatment nowadays, children with MCADD usually have healthy lives with normal growth and development.

Most MCADD babies diagnosed and treated early through newborn screening may never develop signs or symptoms of the disease.

However, if MCADD is not diagnosed early, patients may develop problems in early infancy or childhood with episodic bouts of illness called metabolic crisis.

Possible Signs and Symptoms of MCADD

- extreme sleepiness
- behavioral changes/irritability
- poor appetite
- nausea
- vomiting
- diarrhea
- muscle weakness
- low blood sugar level (hypoglycaemia)

What is the treatment for MCADD?

Metabolic paediatricians and dieticians will work together to give expert advice and care to the babies with MCADD. The goal of treatment is to prevent metabolic crisis and accumulation of toxic metabolites.

Babies with MCADD can usually eat a normal diet. However, it is very important to observe the following:

- avoid long periods without eating during the newborn and infancy period even when baby is well. The length of time MCADD patients can go without eating varies depending on their age; generally speaking the younger they are, the more frequent they need to be fed.
- discuss and design a care plan with your doctor and dietician beforehand -to let you know how to care for and provide extra sugary foods during illness or when your baby is not feeding well to prevent a metabolic crisis. This includes common illnesses such as fever, diarrhea, or vomiting. If the sugary foods do not help or your child refuses to take them, he or she may need to be treated in hospital.

When should I seek immediate help? What should I do?

If you are worried that your baby is ill, it is important to follow medical advice. Bring your baby to your local accident and emergency department immediately. Take any information that you have been given about MCADD, including this pamphlet, to the hospital with you.