Newborn Screening Programme for
Inborn Errors of Metabolism
Information leaflet series (No. 17)

Carnitine-Acylcarnitine Translocase Deficiency

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

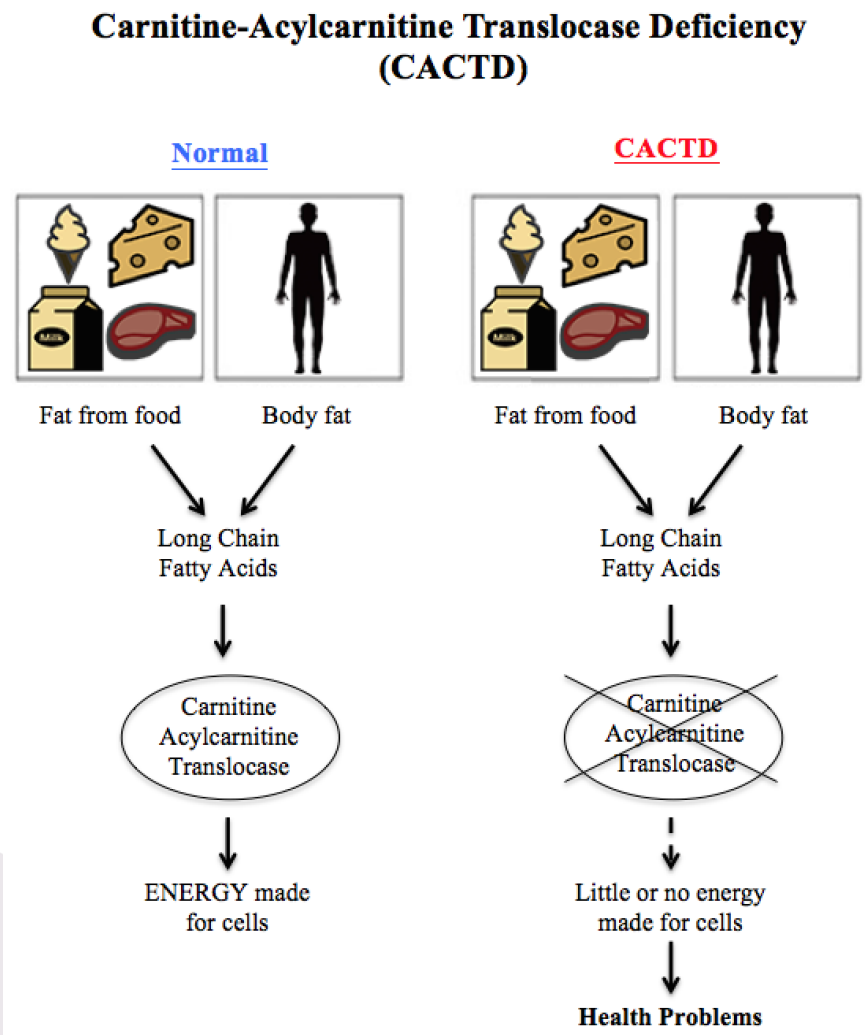
醫院管理局
HOSPITAL AUTHORITY

What is Carnitine-acylcarnitine translocase deficiency (CACTD)?

CACTD is an inherited fatty acid oxidation disorder in which the body cannot effectively process certain fats called “long chain fatty acids” due to inadequate or ineffective functioning of the “carnitine-acylcarnitine translocase” (CACT) enzyme.

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. Normally, long chain fatty acids must attach to carnitine to enter into the mitochondria inside the cells for energy production.

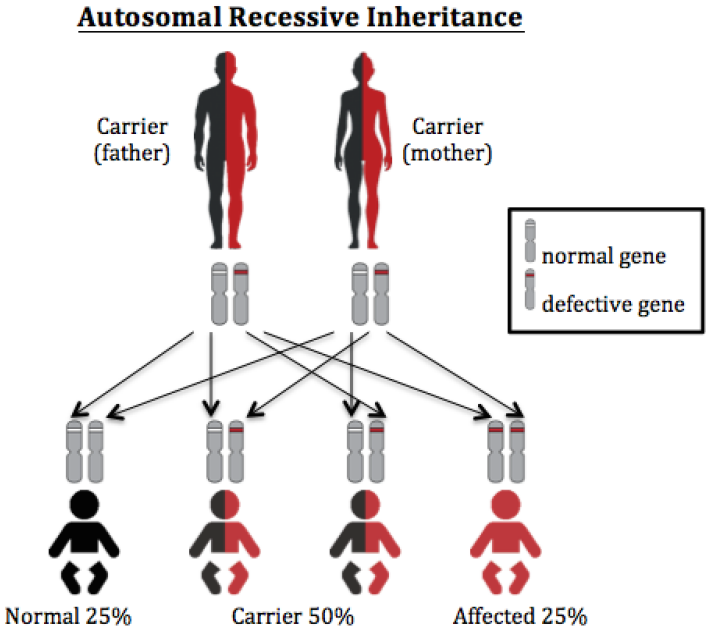
When CACT is insufficient or not functioning well, there will be problems in breaking down long chain fatty acids. This will result in problems with converting them into energy resulting in low blood sugar level (hypoglycaemia) as well as other problems arising from the build-up of toxic substances in the blood.



How is CACTD inherited?

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

CACTD is an autosomal recessive disease. Only when babies inherit two faulty copies of the gene for CACTD from parents, the enzyme made does not work properly or is not even made at all.



What may happen if your baby has CACTD?

Babies with CACTD often show signs and symptoms within the first week of life. CACTD can cause episodic metabolic crisis.

Possible Signs and Symptoms of CACTD

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

Other problems that CACTD patients may have include:

- high levels of ammonia in the blood which may affect brain development
- low muscle tone (floppy muscles and joints) and muscle weakness
- enlarged liver
- heart problems including an enlarged heart and rhythm problem
- breathing problems

What is the treatment for CACTD?

Metabolic paediatricians and dieticians will work together to give expert advice and care to your baby. When necessary, treatment is usually needed throughout life. The goal of treatment is to prevent accumulation of toxic metabolites and metabolic crisis.

It is important that babies with CACTD be fed regularly and do not go for long periods without eating.

Treatment of CACTD may include:

- a special diet with low-fat & high carbohydrate
- L-carnitine which is a safe and natural substance that helps body cells to make energy. It also helps the body to get rid of harmful wastes.

Also very important is to 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.

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 CACTD, including this pamphlet, to the hospital with you.