Newborn Screening Programme for
Inborn Errors of Metabolism
Information leaflet series (No. 18)

Carnitine Palmitoyltransferase II Deficiency

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

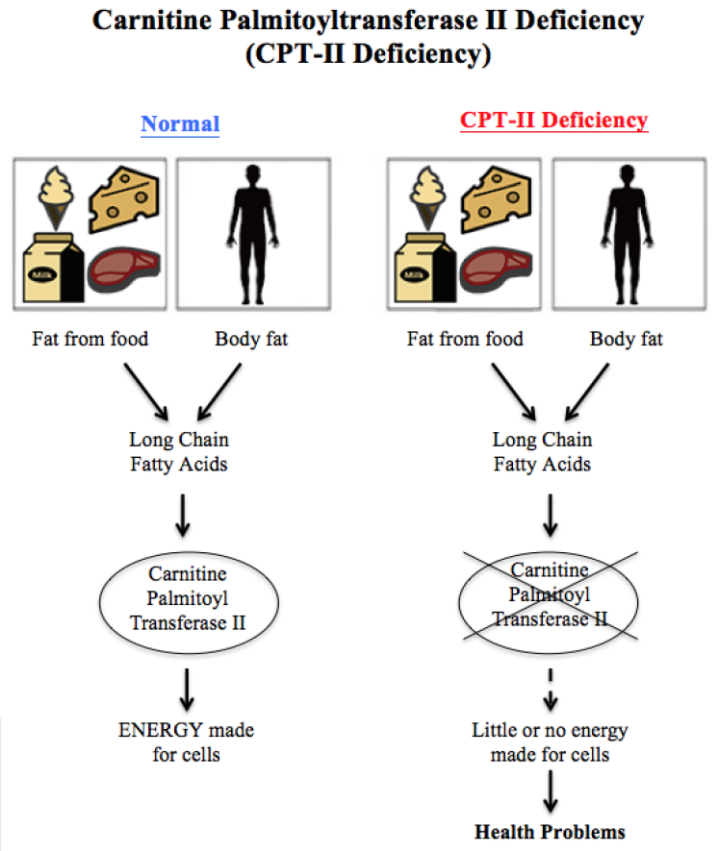
醫院管理局
HOSPITAL AUTHORITY

What is Carnitine palmitoyltransferase II deficiency (CPT-II deficiency)?

CPT-II deficiency is an inherited fatty acid oxidation disorder. People with fatty acid oxidation disorders cannot break down fatty acid, the building blocks of fat, into energy.

Fatty acids are major source of energy for the heart and muscles. During periods of fasting, fatty acids from body store are also an important energy source for the liver and other tissues. Fatty acids are processed by special chemicals called enzymes so that the body can use them. Carnitine palmitoyltransferase II (CPT-II) enzyme is responsible for processing a specific type of fatty acids called “long chain fatty acids”.

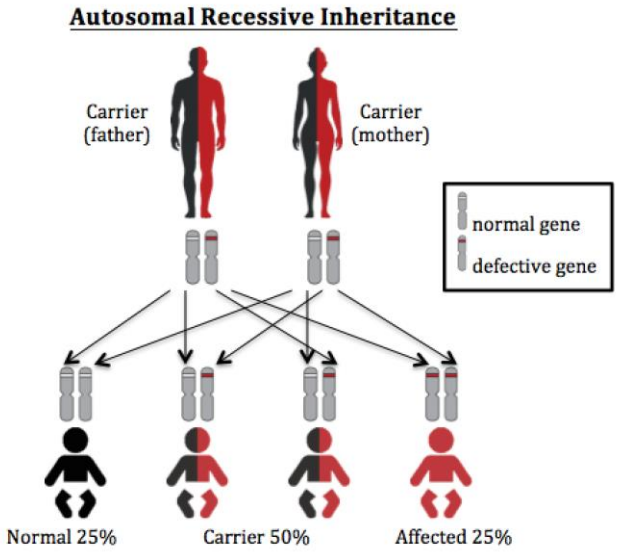
When CPT-II is insufficient or not functioning well, there will be problems in breaking down long chain fatty acids and converting them into energy. Body runs out of energy as a result. Body develops low blood sugar (hypoglycaemia), when the other energy source, sugar, is also used up. When body tries to breakdown fatty acids without success, toxic substances build up in the body, causing health problems.



How is CPT-II deficiency inherited?

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

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



What may happen if your baby has CPT-II deficiency?

Most babies with CPT-II deficiency do not have symptoms until adolescence or early adulthood. Only a few severely affected have symptoms soon after birth or during infancy.

CPT-II deficiency may cause episodic metabolic crisis in severely affected babies. Metabolic crisis is a period of time when a metabolic disorder makes the baby seriously ill.

Signs & symptoms of metabolic crisis in CPT-II deficiency

- extreme sleepiness / irritability
- poor appetite
- nausea / vomiting / diarrhea
- muscle weakness
- low blood sugar level (hypoglycaemia)
- breathing difficulties; seizures; coma

More commonly, patients with CPT-II deficiency start to have episodic muscle weakness and cramps in their teenage or young adulthood, usually after prolonged heavy exercise, cold exposure, fasting, or infection. In severe cases, muscles break down, causing red color urine and kidney failure.

Some problems may also be seen in more severely affected patients:

- enlarged heart and rhythm problem
- liver dysfunction and enlargement
- low muscle tone (floppy muscles and joints)
- congenital kidney, eye or brain defects

What is the treatment for CPT-II deficiency?

Metabolic paediatricians and dieticians will work together to give expert advice and care to your baby. The goal of treatment is to keep energy supply to the body, prevent accumulation of toxic metabolites and metabolic crisis.

It is important that babies with CPT-II deficiency be fed regularly and do not go for long periods without eating.

It is also very important 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 necessary, lifelong treatment may be needed:

- a special diet with low fat and high carbohydrate.
- L-carnitine, a safe and natural substance that helps body cells to make energy and remove harmful wastes.
- MCT (medium-chain triglyceride) oil, a special oil that contains medium-chain fatty acids, which can serves as an alternative energy source for the body.

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 CPT-II deficiency, including this pamphlet, to the hospital with you.