Newborn Screening Programme for
Inborn Errors of Metabolism
Information leaflet series (No. 16)

Carnitine Uptake 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 Uptake Deficiency (CUD)?

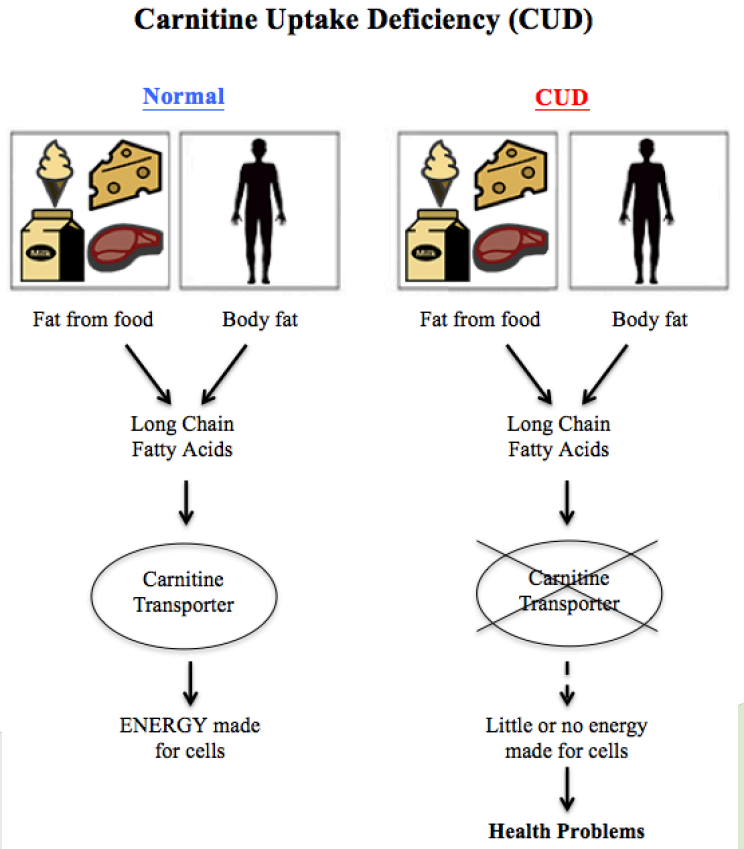
CUD is an inherited fatty acid oxidation disorder due to inadequate or dysfunction of "carnitine transporter" resulting in failure of carnitine reabsorption from the kidney leading to the lack of carnitine inside the cells.

Carnitine, a natural substance acquired mostly through the diet, is used by the cells to process fats and produce energy.

If there is inadequate carnitine intracellularly, long chain fatty acids, predominantly oxidized in the mitochondria of the cells, are unable to be completely broken down resulting in failure of energy production leading to muscle weakness and low blood glucose level (hypoglycemia). Other toxic metabolites may also build up inside the cells and damage multiple organs including the liver, the heart and the muscles.

Other names for carnitine uptake deficiency (CUD) include:

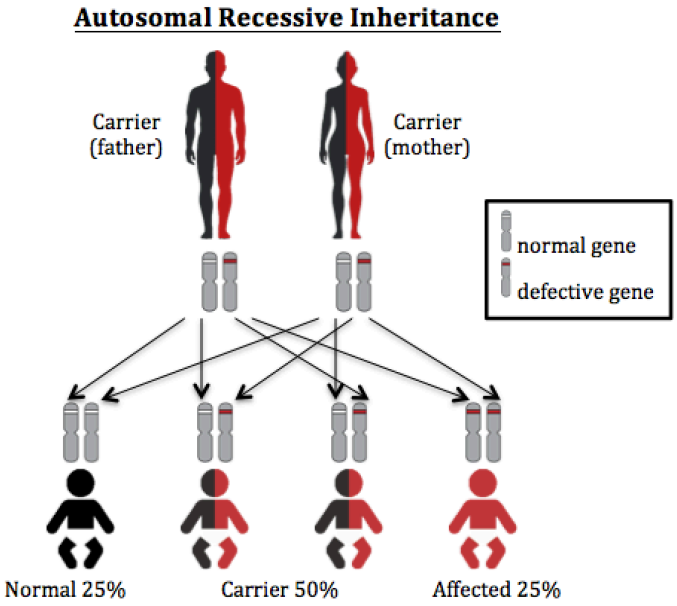
- Carnitine transporter deficiency
- Systemic primary carnitine deficiency



How is CUD inherited?

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

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



What may happen if your baby has CUD?

With prompt and careful treatment nowadays, children with CUD can have healthy lives with normal growth and development. Many CUD babies diagnosed and treated early through newborn screening may never develop signs or symptoms of the disease. However, if CUD 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 CUD

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

Other problems that some CUD 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 CUD?

Metabolic paediatricians and dieticians will work together to give expert advice and care to your baby. When necessary, treatment is usually needed throughout the life.

The goal of treatment is to prevent accumulation of toxic metabolites and metabolic crisis. It is important that babies with CUD be fed regularly and do not go for long periods without eating.

Specific treatment for this condition includes L-carnitine which is a safe and natural substance that helps the body cells to make energy and get rid of harmful wastes. It is usually a lifelong treatment for most CUD patients.

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