Newborn Screening Programme for
Inborn Errors of Metabolism
Information leaflet series (No. 20)

Very Long-chain Acyl-CoA Dehydrogenase Deficiency

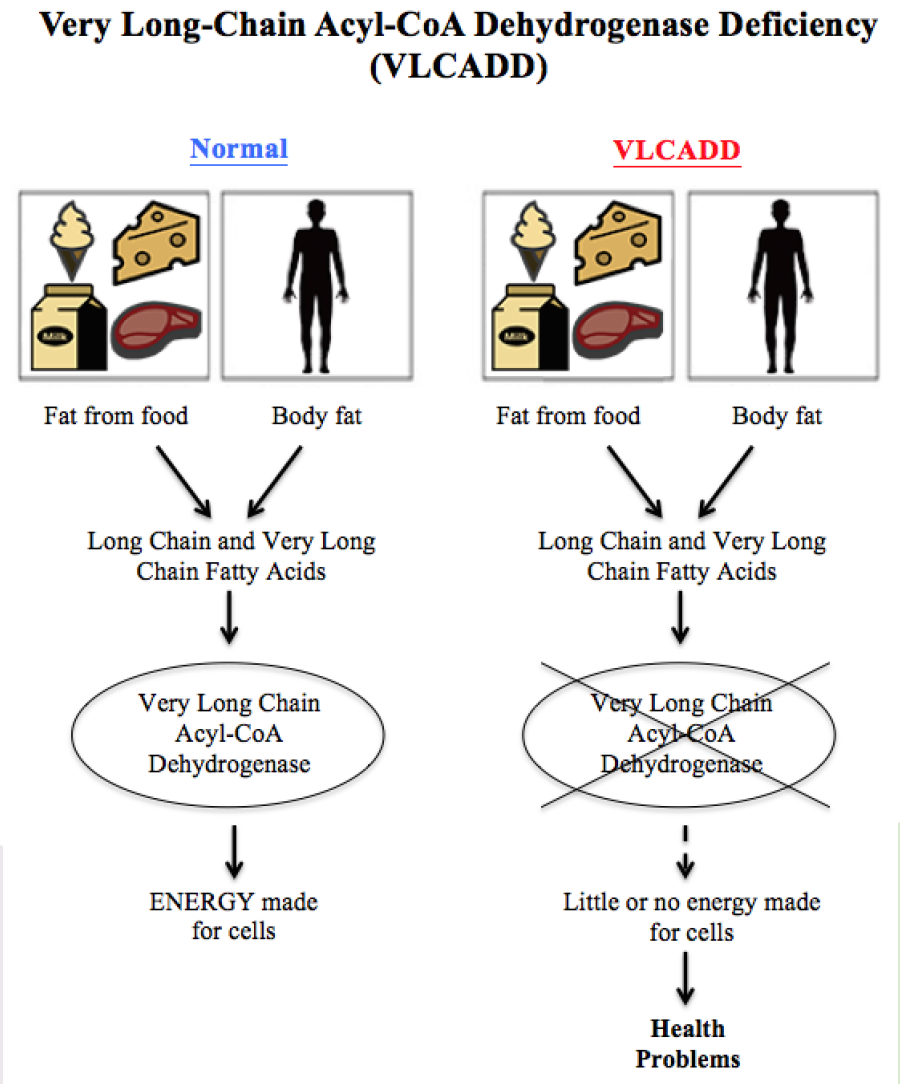
For general queries on Newborn Screening Programme for Inborn Errors of Metabolism, please call: ☎ 5741 4280 (Department of Clinical Genetics, Hospital Authority)

醫院管理局
HOSPITAL AUTHORITY

What is Very long-chain acyl-CoA dehydrogenase deficiency (VLCADD)?

VLCADD is an inherited fatty acid oxidation disorder caused by deficiency or ineffective functioning of an enzyme called “very long-chain acyl-CoA dehydrogenase” (VLCAD). Without this enzyme, certain fats called “very long-chain fatty acids” cannot be broken down properly.

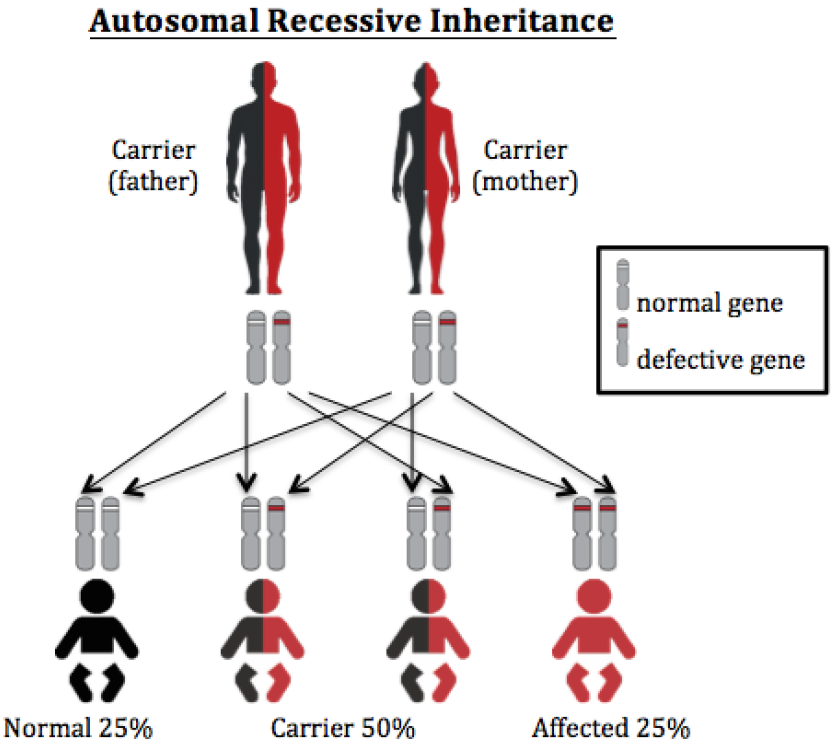
Fatty acids are a major source of energy for the heart and muscles. During periods of fasting, fatty acids are also an important energy source for the liver and other tissues. When very long-chain fatty acids are not metabolized properly, they cannot be converted to energy resulting in low blood sugar level (hypoglycaemia) and lethargy. At the same time, other toxic metabolites may also build up inside cells and damage multiple organs including the liver, heart and muscles.



How is VLCADD inherited?

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

Very long-chain acyl-CoA dehydrogenase deficiency is an autosomal recessive disease. Only when babies inherit two faulty copies of the gene for very long-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 VLCADD?

VLCADD can be very variable causing mild effects in some patients and more serious problems in others. Symptoms may start in infancy or later in adulthood.

Some VLCADD babies diagnosed and treated early through newborn screening may not develop signs or symptoms of the disease.

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

Possible Signs and Symptoms of VLCADD

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

Other problems that some VLCADD patients may have include:

- enlarged liver and other liver problems
- heart problems including an enlarged heart and rhythm problem
- breathing problems
- muscle cramps, weakness, sometimes muscle breakdown after vigorous exercise.

What is the treatment for VLCADD?

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

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

Treatment for VLCADD can be varied in different patients. Certain treatment such as a special diet or medication like L-carnitine may be advised for some patients but not the others. When indicated, L-carnitine is a safe and natural substance that helps body cells make energy. It also helps the body 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 VLCADD, including this pamphlet, to the hospital with you.