Newborn Screening Programme for
Inborn Errors of Metabolism
Information leaflet series (No. 6)

3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency

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

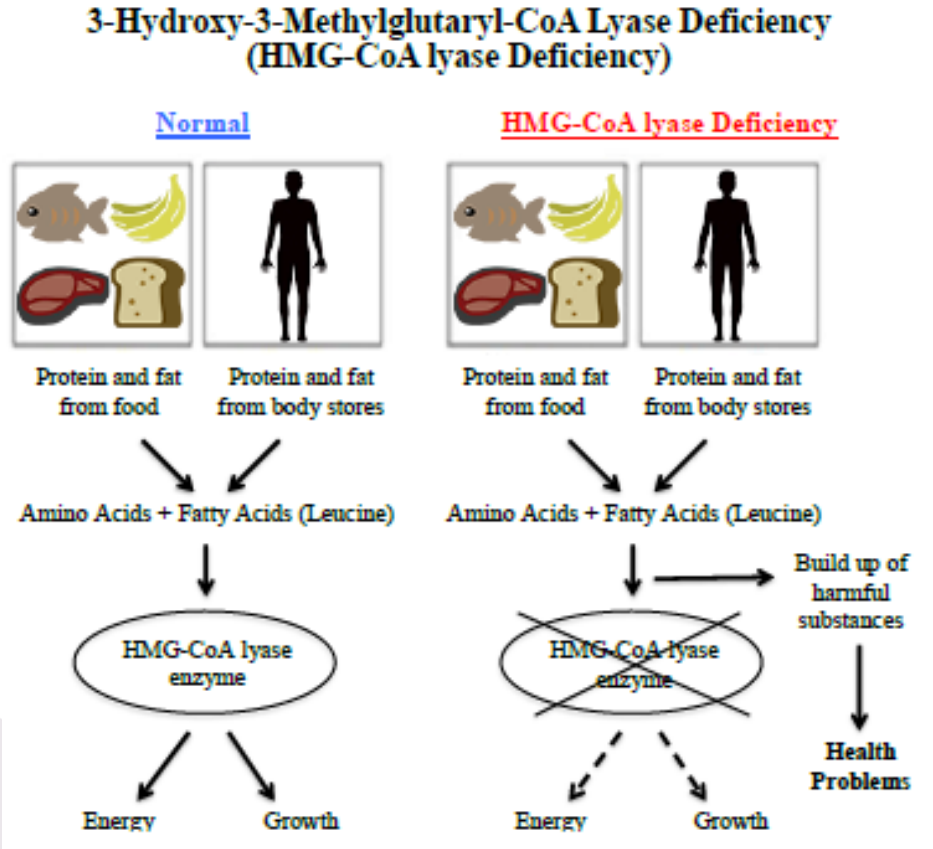
醫院管理局
HOSPITAL AUTHORITY

What is 3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG-CoA lyase deficiency)?

Protein from the food we eat is broken down into building blocks of protein (amino acids) which are then further metabolised by specific enzymes for body growth and energy production.

3-hydroxy-3-methylglutaryl-CoA lyase deficiency (also known as HMG-CoA lyase deficiency) is an inherited organic acid disorder in which the body is unable to process an amino acid called "leucine" due to the deficiency of an enzyme called "HMG-CoA lyase". This enzyme is responsible for breaking down dietary proteins and fats for energy. It also produces ketone during the breakdown of fats. Ketones are the important sources of energy for our body, especially the brain, during periods without food (fasting).

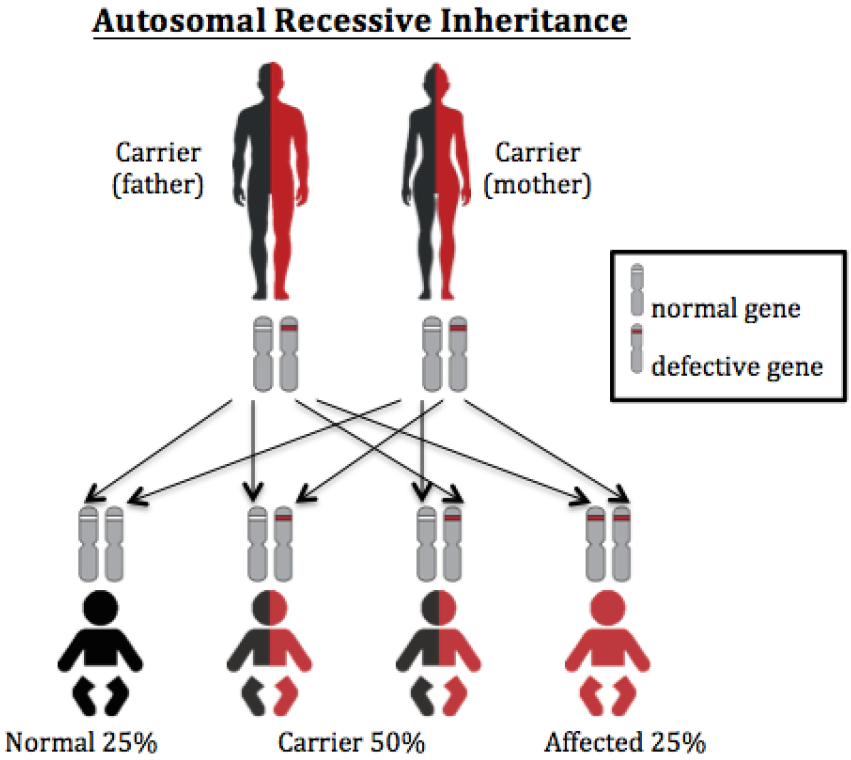
In this condition when the body cannot process leucine normally, buildup of organic acid can cause the blood to become too acidic (metabolic acidosis). In addition, a shortage of ketone leads to low sugar level (hypoglycaemia). Both metabolic acidosis and low blood sugar can result in cell damage, particularly in the brain.



How is HMG-CoA lyase deficiency inherited?

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

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



What may happen if your baby has HMG-CoA lyase deficiency?

Babies with HMG-CoA lyase deficiency may experience slightly different signs and symptoms. Most affected children start to show signs at 3 months to 2 years of age though few may show signs of the condition just a few days after birth. Metabolic crisis, which is a period of time when a metabolic disorder makes the baby seriously ill, may develop and such episode is frequently triggered by infection, prolonged period without food or increased intake of protein-rich food.

Signs and Symptoms of* HMG-CoA lyase deficiency *during metabolic crisis

- Poor appetite
- Vomiting and diarrhoea
- Irritability or excessive sleepiness/ tiredness, Floppiness and weakness
- Behavioural changes
- Low blood sugar (hypoglycaemia)
- Breathing difficulty
- Coma

With early and careful treatment, babies with HMG-CoA lyase deficiency can have healthy growth and development. It is possible, even with treatment, for babies with HMG-CoA lyase deficiency to have low blood sugar and other signs of HMG-CoA lyase deficiency.

Early screening and treatment of HMG-CoA lyase deficiency are very important and babies who do not receive treatment usually die or develop permanent brain damage.

What is the treatment for HMG-CoA lyase deficiency?

The treatment for HMG-CoA lyase deficiency may include:

- Low-leucine diet with limited amounts of fat and protein intake to prevent metabolic crises
- L-carnitine supplementation that can help to remove toxic substances from the body
- Discuss and design a possible plan with your doctor and dietician beforehand, in order to restrict protein and provide extra starchy or sugary foods during illness or other times when your child loses appetite in order to prevent a metabolic crisis

It is important that babies with HMG-CoA lyase deficiency need to be fed regularly and should not go for long periods without eating (i.e. fasting). They also need to see their metabolic doctors regularly.

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 HMG-CoA lyase deficiency, including this pamphlet, to the hospital with you.