Newborn Screening Programme for
Inborn Errors of Metabolism
Information leaflet series (No. 3)

Methylmalonic Acidaemia (Methylmalonyl-CoA mutase 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

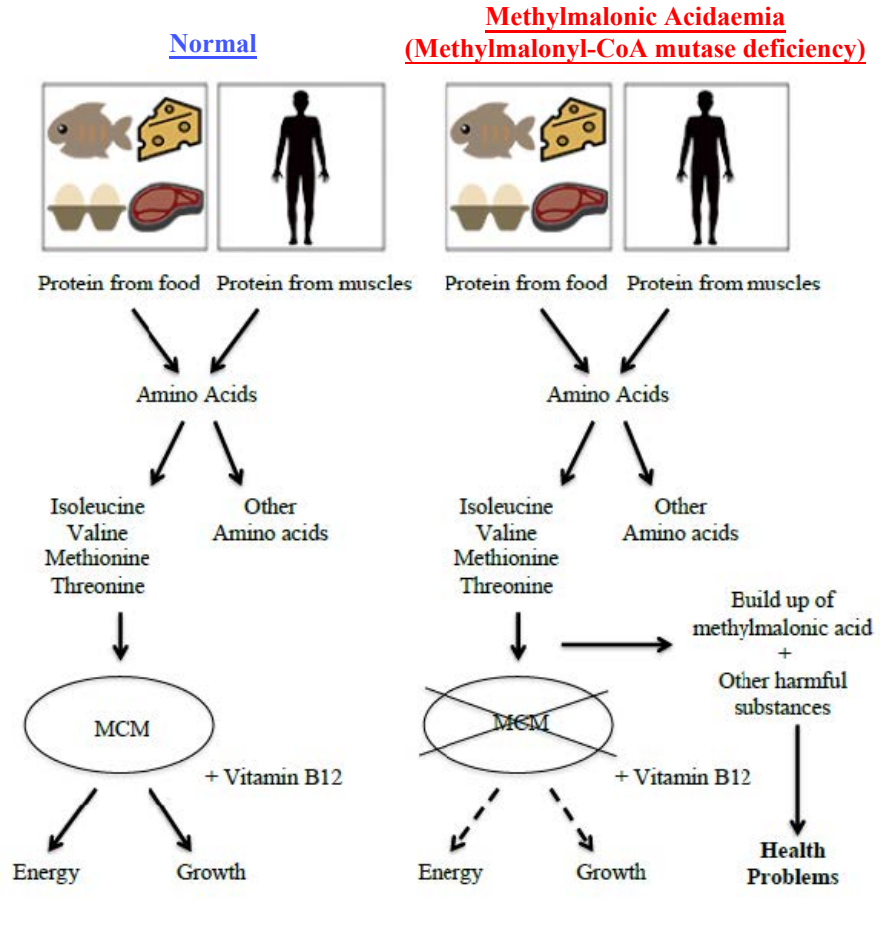
What is Methylmalonic Acidaemia (Methylmalonyl-CoA mutase deficiency)?

Methylmalonic Acidaemia due to methylmalonyl-CoA mutase (MCM) deficiency is a rare but treatable organic acid disorder. People with organic acid disorders cannot process amino acids, the building blocks of protein.

Our body breaks down protein in food into amino acids when we eat, and breaks down protein in our muscle into amino acids during prolonged fasting and stress. Amino acids are then processed by special chemicals called enzymes in order for the body to use them. Different enzymes target specifically at different amino acids.

The metabolic process of the four amino acids (Isoleucine, Valine, Methionine and Threonine) cannot be catalysed if babies lack the essential enzyme MCM. As a result, these amino acids cannot be utilized and harmful substances build up in the body, causing health problems.

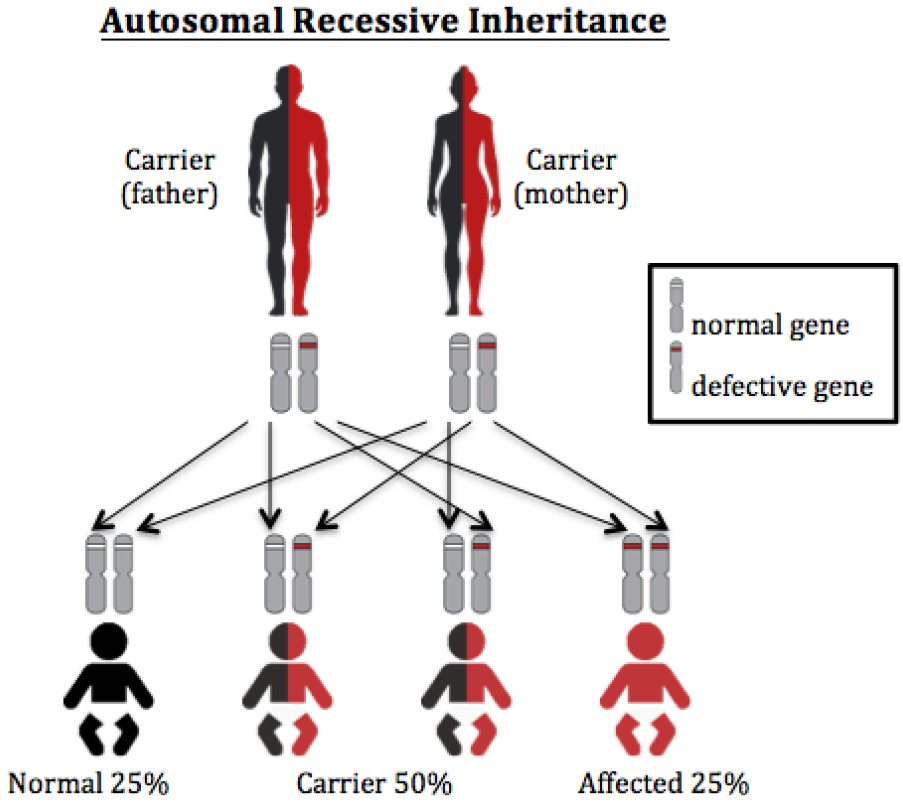
Methylmalonic Acidaemia
(Methylmalonyl-CoA mutase deficiency)



How is Methylmalonic Acidaemia (Methylmalonyl-CoA mutase deficiency) inherited?

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

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



What may happen if your baby has Methylmalonic Acidaemia (Methylmalonyl-CoA mutase deficiency)?

Babies with Methylmalonic Acidaemia (Methylmalonyl-CoA mutase deficiency) are usually healthy at birth but they can develop metabolic crisis within the first few days of life due to the build-up of toxic substances in the body.

Metabolic crisis is a period of time when a metabolic disorder makes the baby seriously ill. Babies tend to develop metabolic crisis when they do not have food for long periods of time, or when they have infection, fever or stomach upset. Left untreated, they deteriorate with seizure and coma which can be life threatening.

Signs and Symptoms of Methylmalonic Acidaemia (Methylmalonyl-CoA mutase deficiency)

- Poor feeding; nausea & vomiting; poor growth
- Irritability or sleepiness; floppiness and weakness
- Muscle spasm; abnormal posture or movements
- Coldness; breathing difficulties; fast breathing
- Stroke; seizures; coma

Symptoms vary from person to person. Some children have very mild or no symptom, and do not develop symptoms of metabolic crisis until they are older. Some develop health problems even if they have never had a metabolic crisis. They may have brain damage and intellectual disabilities, poor growth, frequent infections, pancreatitis, heart, kidney and liver dysfunction, and visual impairment.

What is the treatment for Methylmalonic Acidaemia (Methylmalonyl-CoA mutase deficiency) ?

Babies with Methylmalonic Acidaemia (Methylmalonyl-CoA mutase deficiency) benefit significantly from early treatment and can have healthy and active lives.

Some patients with Methylmalonic Acidaemia (Methylmalonyl-CoA mutase deficiency) can be treated with regular vitamin B12 injections. Other patients with Methylmalonic Acidaemia (Methylmalonyl-CoA mutase deficiency) who do not respond to vitamin B12. It is important to feed regularly and not to go for long periods without eating. Medications may also be given.

Babies with Methylmalonic Acidaemia (Methylmalonyl-CoA mutase deficiency) need to see their specialist metabolic team regularly even when they do not have symptom. It is important to discuss and design a possible care plan with your doctor and dietician beforehand, in order to provide extra sugary foods during illness or other times when baby is not feeding well to prevent 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 Methylmalonic Acidaemia (Methylmalonyl-CoA mutase deficiency), including this pamphlet, to the hospital with you.