Newborn Screening Programme for
Inborn Errors of Metabolism
Information leaflet series (No. 5)

Isovaleric Acidaemia

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

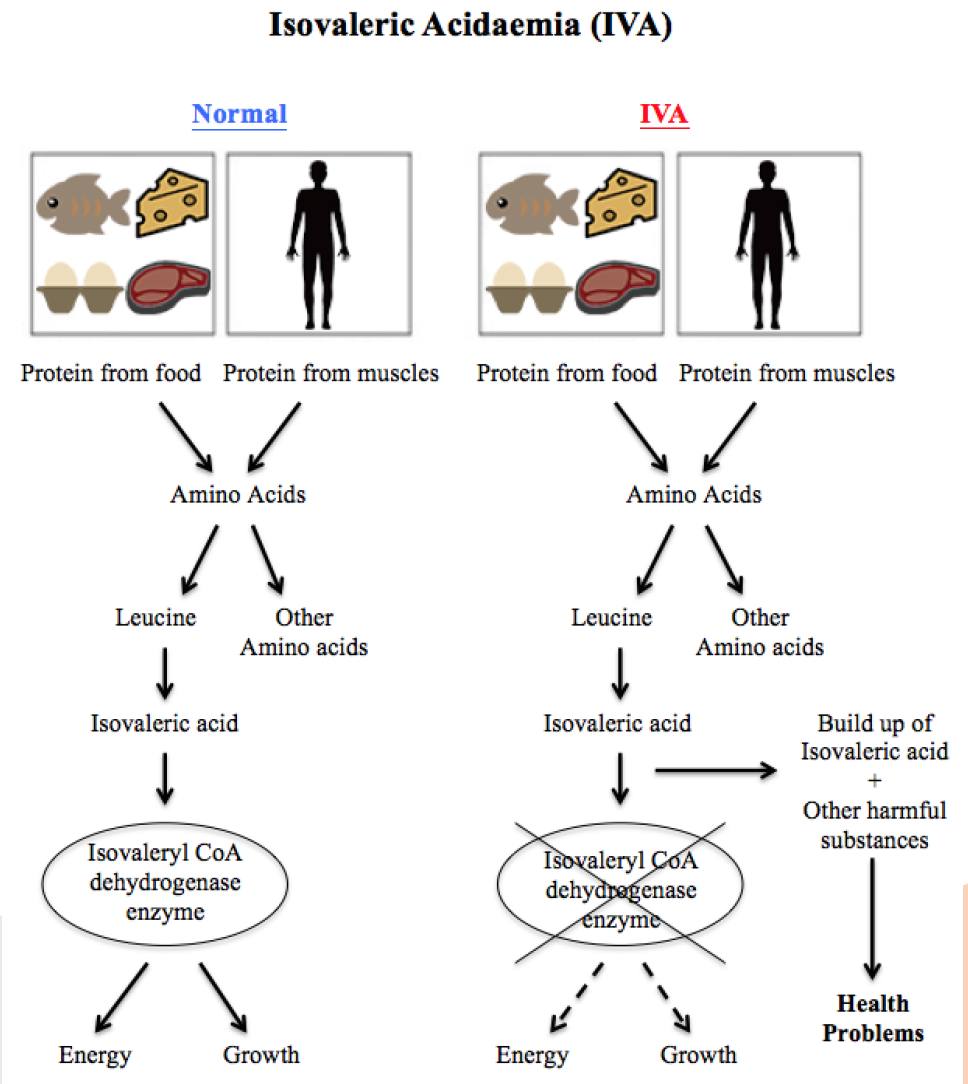
醫院管理局
HOSPITAL AUTHORITY

What is Isovaleric acidaemia (IVA)?

IVA 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 so that the body can use them. Different enzymes target specifically at different amino acids.

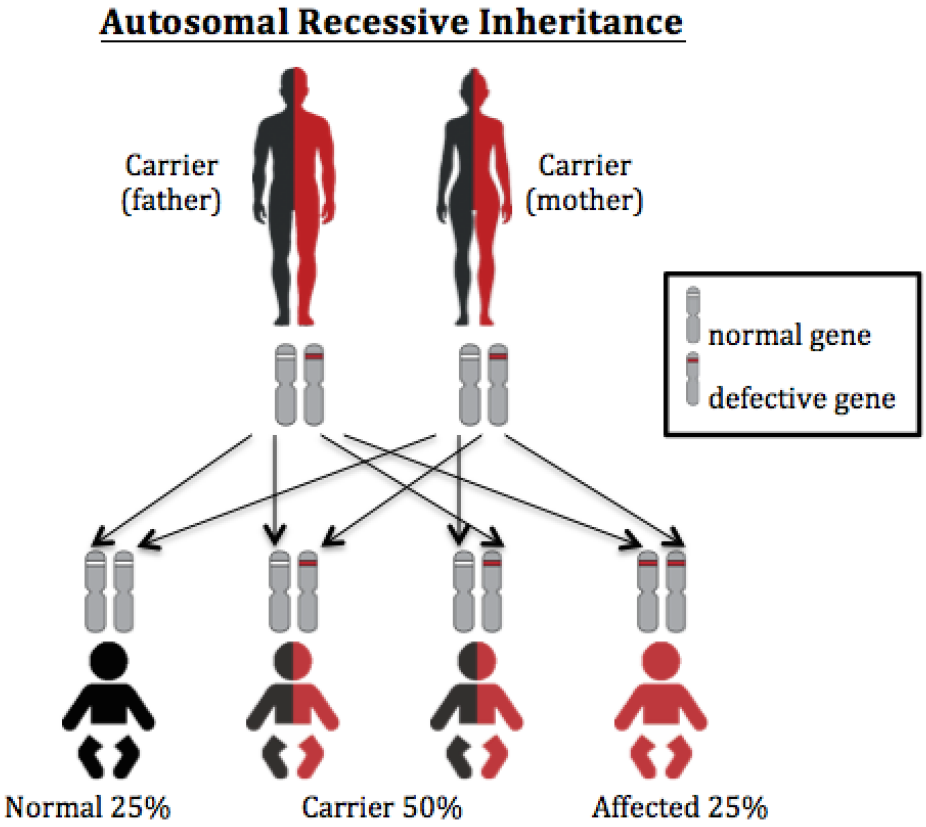
Babies with IVA lack the specific enzyme called “Isovaleryl CoA dehydrogenase”. This enzyme is responsible to process an amino acid called leucine. Whilst leucine cannot be utilized for body’s use, harmful substances including isovaleric acid also build up in the body, causing health problems.



How is IVA inherited?

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

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



What may happen if your baby has IVA?

Babies with IVA 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 IVA

- Poor feeding; nausea & vomiting; poor growth
- Irritability or sleepiness; floppiness and weakness
- Muscle spasm; abnormal posture or movements
- Coldness; breathing difficulties; fast breathing
- Seizures; coma
- Smell of sweaty feet

Symptoms vary from person to person. Some children with IVA have very mild or no symptom, and do not develop symptoms of a metabolic crisis until they are older**.**

What is the treatment for IVA?

Babies with IVA benefit significantly from early treatment and can have healthy and active lives.

IVA can be treated with special diet and supplements. Some babies with mild IVA only need treatment when they are unwell. It is important to feed regularly and not to go for long periods without eating. Medication may also be given.

Babies with IVA need to see their specialist metabolic team regularly even when they do not have symptom. It is important to discuss and design a possible 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 IVA, including this pamphlet, to the hospital with you.