Newborn Screening Programme for
Inborn Errors of Metabolism
Information leaflet series (No. 10)

Argininosuccinic Acidaemia

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

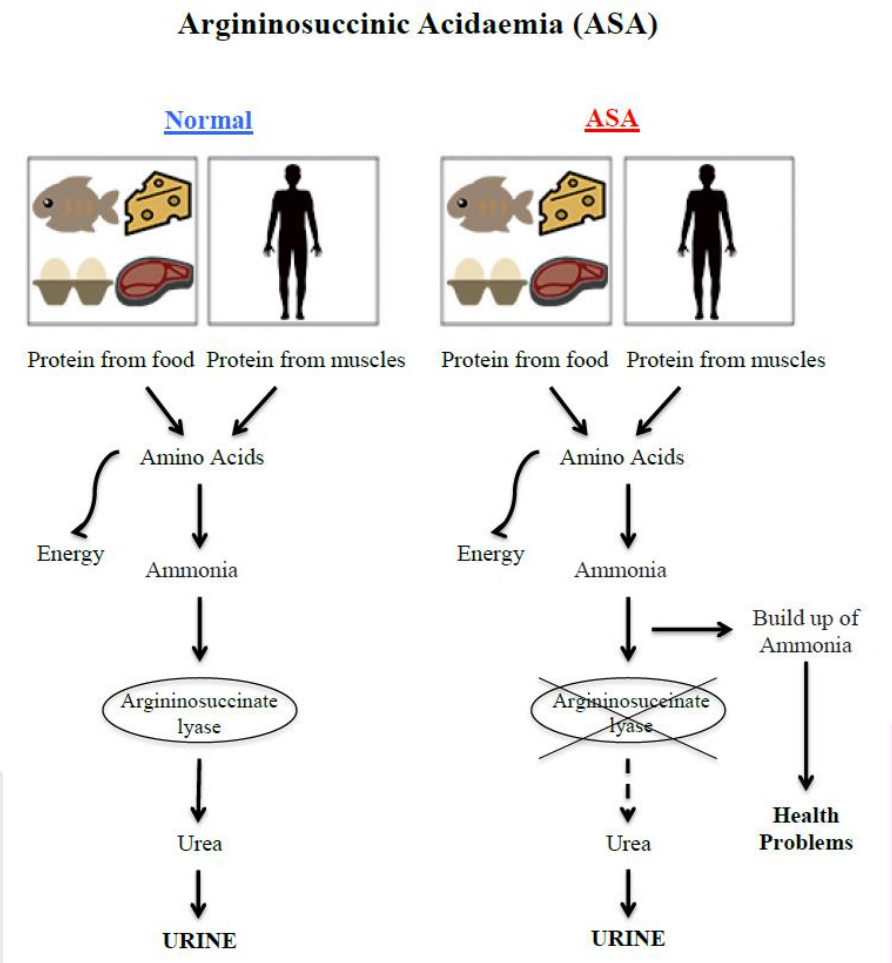
醫院管理局
HOSPITAL AUTHORITY

What is Argininosuccinic acidaemia (ASA)?

ASA is a rare but treatable amino acid disorder. People with amino 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. Excessive amino acids are converted to a toxic waste product called ammonia. Ammonia has to be processed immediately into urea before excretion. This process is mediated by six special chemicals called enzymes, which form a chain of reactions called the urea cycle. The urea cycle is also responsible to produce an amino acid called arginine.

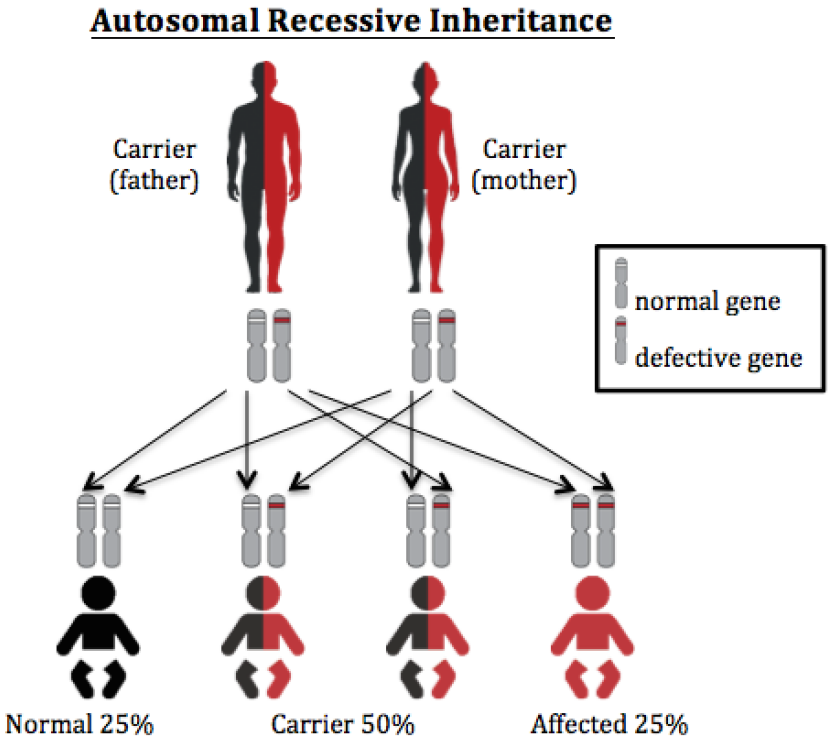
Babies with ASA lack the specific enzyme called “argininosuccinate lyase”, one of the enzymes in the urea cycle, and have an ineffective urea cycle. As a result, toxic metabolites including ammonia build up in the body. Arginine, which is important for the health of blood vessels, becomes insufficient. This causes long term health problems including brain damage, high blood pressure and liver disease.



How is ASA inherited?

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

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



What may happen if your baby has ASA?

Babies with ASA 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.

If right treatment is started early, babies with ASA are well and can have healthy and active lives.

Possible signs and symptoms of ASA

- Poor feeding; nausea & vomiting; poor growth
- Coldness; breathing difficulties; fast breathing
- Irritability or sleepiness; weakness; floppiness or spasm; stroke; seizures; coma
- Enlarged liver; dry and brittle hair

Some children have very mild or no symptom, and do not develop symptoms of metabolic crisis until they are older. Some children still develop health problems including liver fibrosis, high blood pressure, learning disability and epilepsy, even if they have never had a metabolic crisis.

What is the treatment for ASA?

Babies with ASA can be treated with special low protein diet and arginine supplement. It is important to feed regularly and not to go for long periods without eating. Medications may also be given.

Babies with ASA 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 to let you know how to care for your baby during illness or other times when baby is not feeding well in order 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 the prescribed medication, special infant formula and any information that you have been given about ASA, including this pamphlet, to the hospital with you.