Newborn Screening Programme for
Inborn Errors of Metabolism
Information leaflet series (No. 24)

Classic Galactosaemia

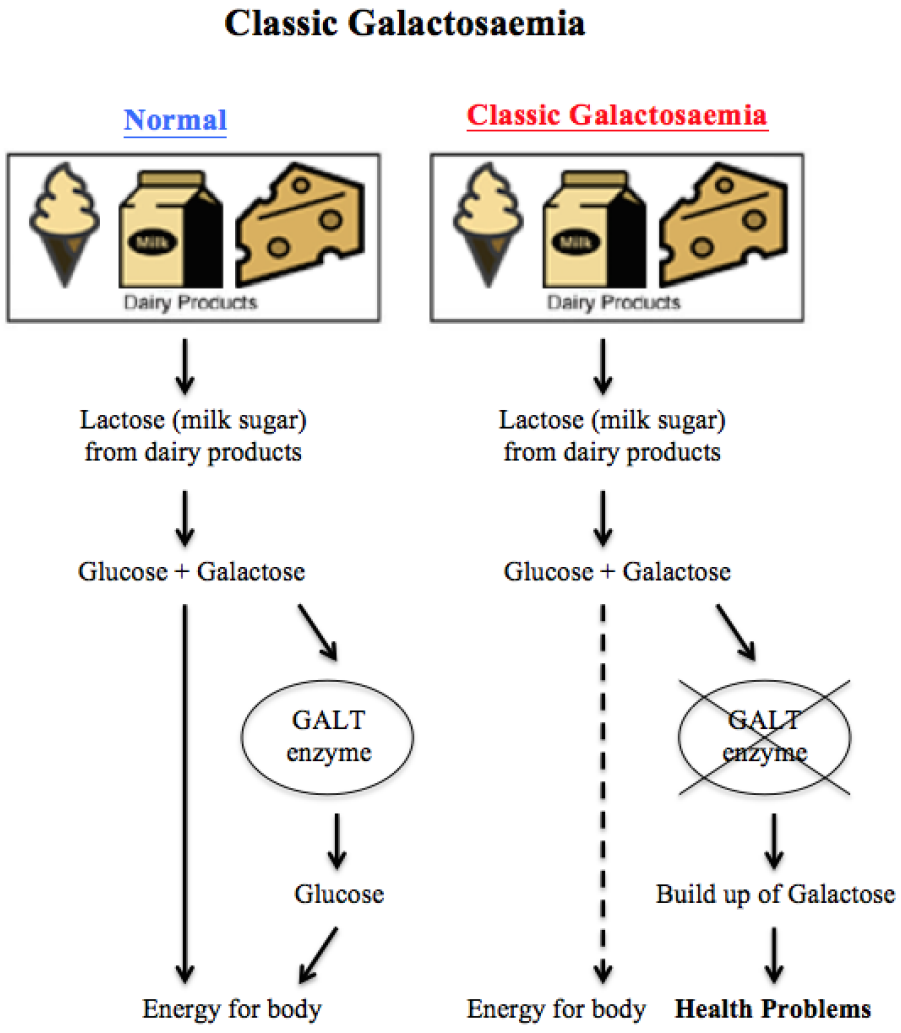
For general queries on Newborn Screening Programme for Inborn Errors of Metabolism, please call: ☎ 5741 4280 (Department of Clinical Genetics, Hospital Authority)

醫院管理局
HOSPITAL AUTHORITY

What is Classic Galactosaemia(GALT)?

Classic galactosaemia is a condition that affects how the body breaks down a sugar called galactose.

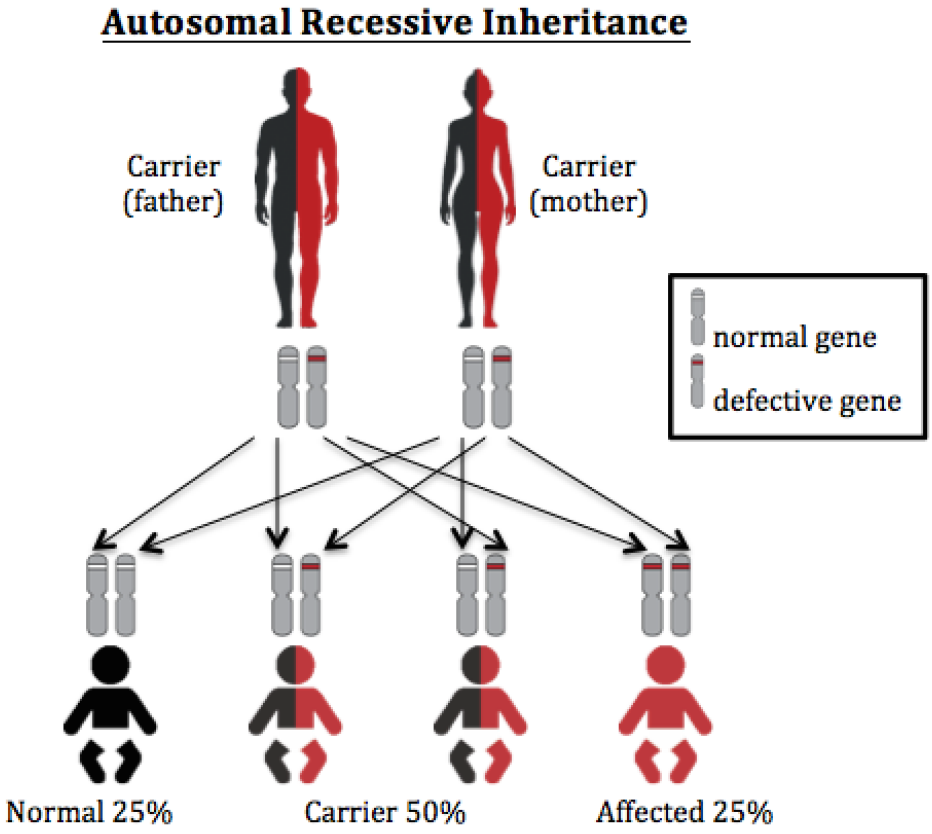
Galactose is part of a larger sugar called lactose, and is found in all foods that contain milk. Children with classic galactosaemia lack an enzyme called 'galactose-1-phosphate uridyl transferase' (GALT). Without the GALT enzyme, galactose cannot be broken down into glucose for the body to utilise for energy, and excess galactose builds up in parts of the body such as the brain, eyes, liver and kidneys.



How is Classic Galactosaemia inherited?

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

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



What may happen if your baby has Classic Galactosaemia?

Babies with classic galactosaemia are usually healthy at birth. The signs and symptoms of classic galactosaemia can appear within a few days after birth.

Possible Signs and Symptoms of Classic Galactosaemia

- Poor feeding and sucking
- Failure to gain weight and grow as expected
- Vomiting
- Diarrhoea
- Low blood sugar level
- Excessive sleepiness
- Liver damage and bleeding
- Overwhelming bacterial infections

Symptoms may vary from person to person. Affected children are at increased risk for early cataracts (clouding of the lens of the eye), intellectual disability and future reproductive problems.

What is the treatment for Classic Galactosaemia?

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

Doctors and dieticians will work together to give expert advice and care to the babies with classic galactosaemia. The goal of treatment is to prevent accumulation of undigested sugars in the blood.

The treatment for classic galactosaemia may include:

- Avoidance of regular milk and dairy products
- A special milk formula that contains low levels of galactose. Dieticians will advise on a galactose-restricted diet

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 classic galactosaemia, including this pamphlet, to the hospital with you.