Newborn Screening Programme for
Inborn Errors of Metabolism
Information leaflet series (No. 7)

Beta-Ketothiolase Deficiency

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

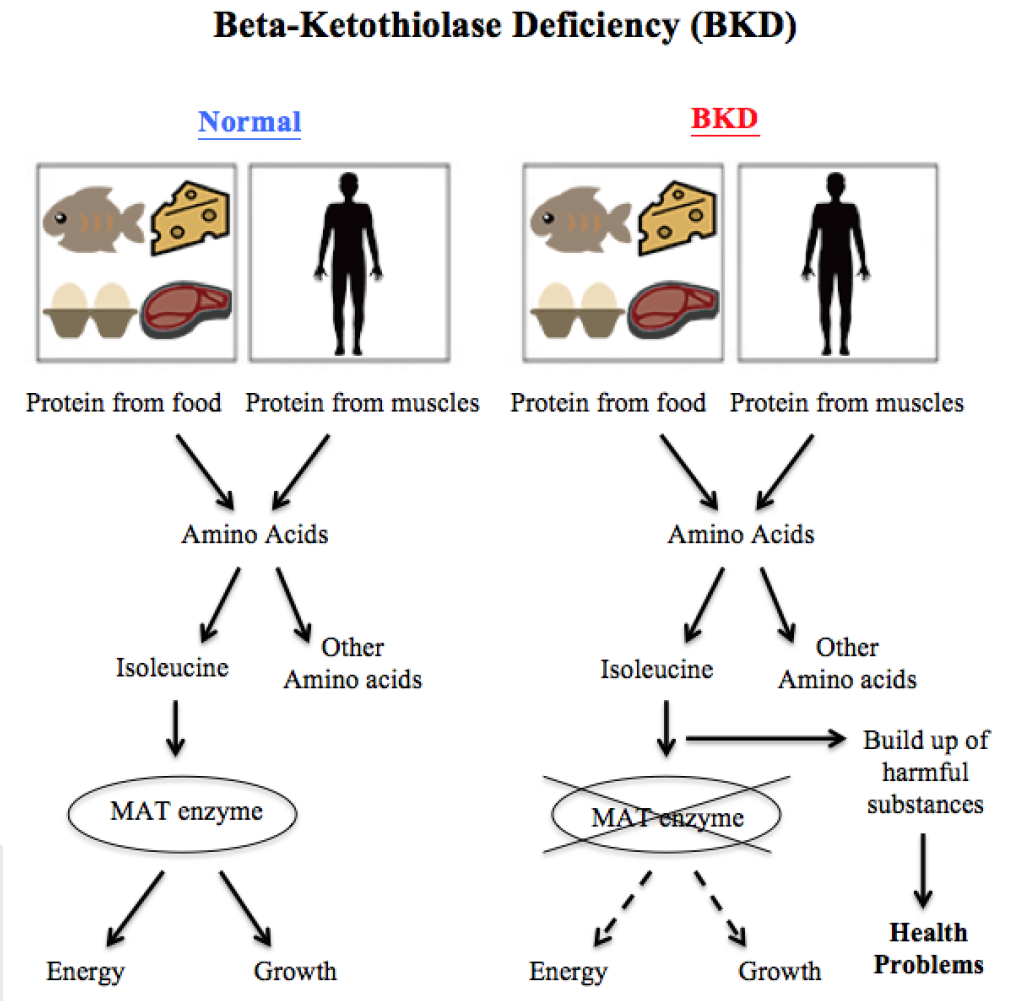
醫院管理局
HOSPITAL AUTHORITY

What is Beta-ketothiolase Deficiency (BKD)?

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.

Beta-ketothiolase deficiency (BKD) is an inherited organic acid disorder due to deficiency of an enzyme called "mitochondrial acetoacetyl-CoA thiolase" (MAT).

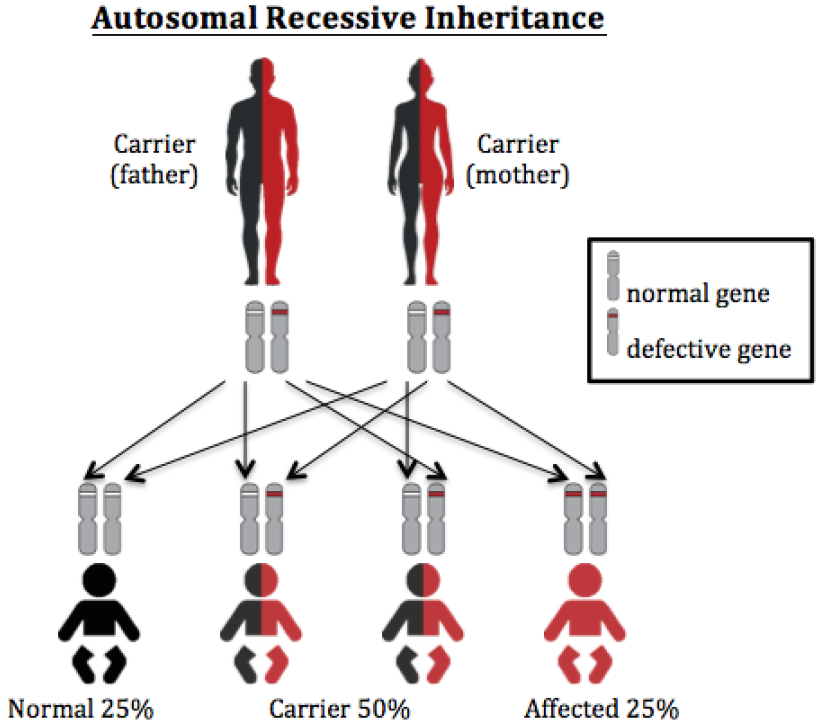
Without this enzyme, the body cannot effectively process an amino acid called "isoleucine". The body's ability to process "ketones", which are the molecules produced during breakdown of fat, is also impaired. Harmful substances called organic acid will accumulate and damage the body's tissues and organs, particularly the nervous system. Consequently, metabolic crisis and serious health problem may occur.



How is BKD inherited?

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

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



What may happen if your baby has BKD?

The signs and symptoms of BKD are usually not obvious at early stage. Each baby with BKD may experience slightly different symptoms.

Metabolic crisis, which is a period of time when a metabolic disorder makes the baby seriously ill, may develop. Such episode, which is called ketoacidotic attack, typically appears between the age of 6 and 24 months and is frequently triggered by infection, prolonged period without food or increased intake of protein-rich food.

With early treatment, babies with BKD can have healthy growth and development. There are cases where children with BKD show signs of the condition even with treatment. However, these signs usually decrease with age.

Signs and Symptoms of BKD during metabolic crisis

- Poor appetite / vomiting and diarrhea
- Excessive sleepiness or tiredness
- Breathing problem
- Convulsion
- Coma
- Ketones in urine (metabolites produced during the breakdown of fat)

What is the treatment for BKD?

The treatment is usually needed throughout life and the goal of treatment is to prevent metabolic crisis and accumulation of toxic metabolites.

The treatment for BKD may include:

- A restricted diet to avoid too many harmful proteins (i.e. low-protein diet)
- Fat-rich diet that induce ketone production should also be avoided
- 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 BKD 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 BKD, including this pamphlet, to the hospital with you.