Newborn Screening Programme for
Inborn Errors of Metabolism
Information leaflet series (No. 8)

Classic Phenylketonuria

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 phenylketonuria (PKU)?

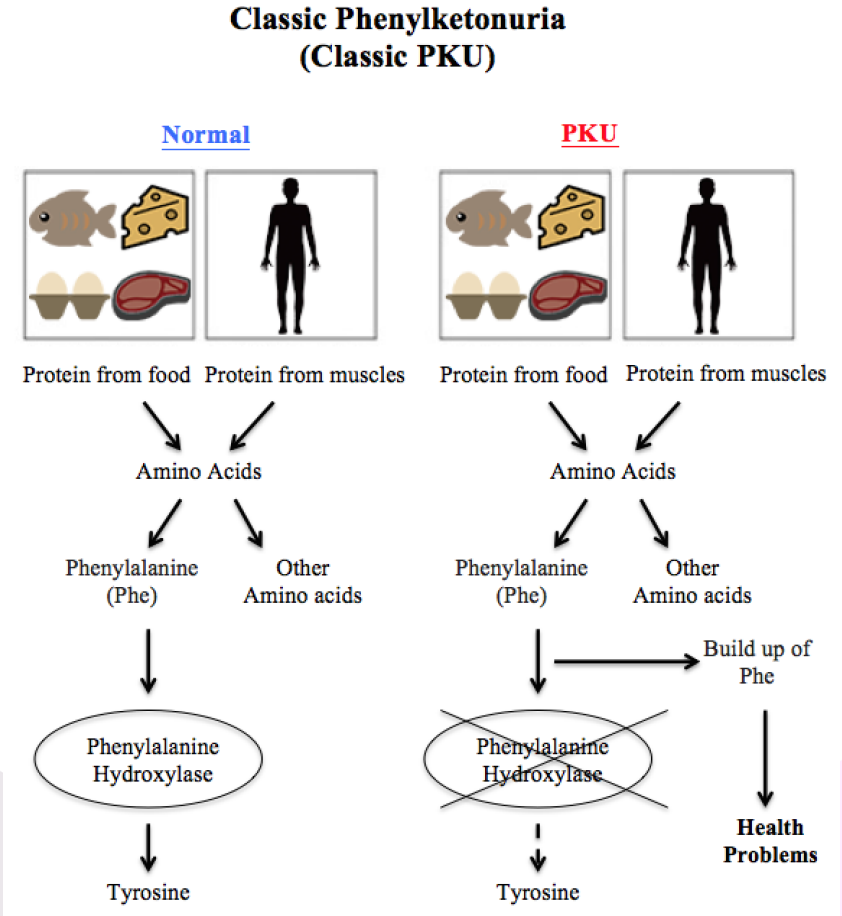
PKU is a rare but treatable amino acid disorder. People with amino acid disorders cannot process amino acids, the building blocks of protein.

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.

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 PKU lack the specific enzyme called “phenylalanine hydroxylase” (PAH) which is responsible to process an amino acid called phenylalanine into another amino acid called tyrosine.

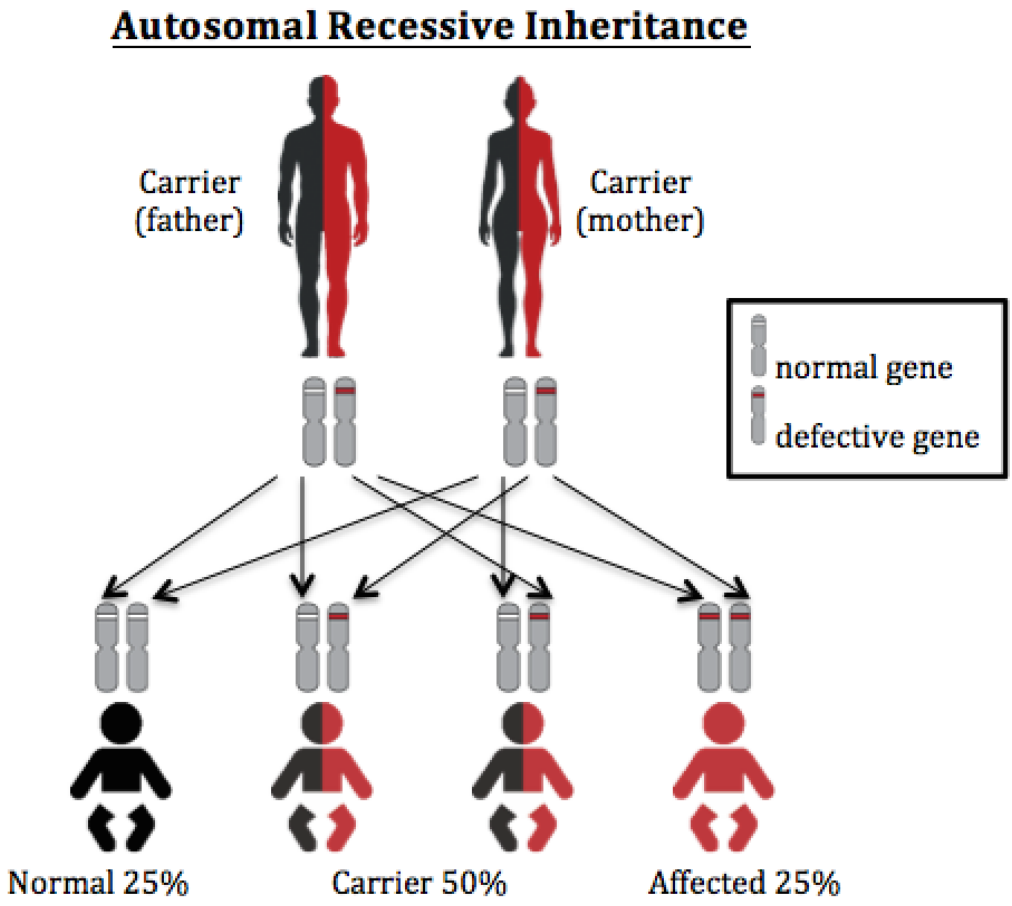
When phenylalanine hydroxylase is missing or malfunctioning, phenylalanine accumulates to a harmful level. Tyrosine, which is essential for proper brain functioning, may be insufficient. This causes long term health problems including learning difficulties.



How is PKU inherited?

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

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



What may happen if your baby has PKU?

Babies with PKU are usually healthy at birth. They gradually develop symptoms during infancy as a result of the accumulation of phenylalanine. This includes progressive and irreversible brain damage, leading to neurological disorders, learning difficulties and behavioural problems.

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

Possible signs and symptoms of PKU

- Developmental delay and floppiness in infants
- Intellectual disabilities, slow thinking in older patients
- Behavioral problems, hyperactivity, attention deficits
- Seizures
- Poor growth, fair skin and hair, eczema
- A “musty” or “mousy” body odour

Uncommonly, some babies have the condition called “mild hyperphenylalaninaemia”, with only slightly increased phenylalanine levels in the blood. They do not have classic PKU. They might not need any treatment and only need regular monitoring of phenylalanine levels in the blood.

What is the treatment for PKU?

PKU can be treated with special low phenylalanine diet and supplements which reduce the build-up of phenylalanine in the body and prevent irreversible brain damage. Dietary treatment should start as early as possible and continue throughout the life.

Babies with PKU need to take a special nutritionally balanced phenylalanine-free infant formula. High protein foods including milk (both breast milk and normal infant formula) have to be limited in order to provide small amount of phenylalanine just right for normal growth, development and health.

Patients with PKU need to see their specialist metabolic team regularly even when they do not have the symptoms. They need to have regular monitoring of phenylalanine levels in the blood. The metabolic team will adjust the diet accordingly.

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. When you are going to the hospital, take the special infant formula and any information that you have been given about PKU, including this pamphlet, with you.