Newborn Screening Programme for
Inborn Errors of Metabolism
Information leaflet series (No. 9)

6-Pyruvoyl-Tetrahydropterin Synthase 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

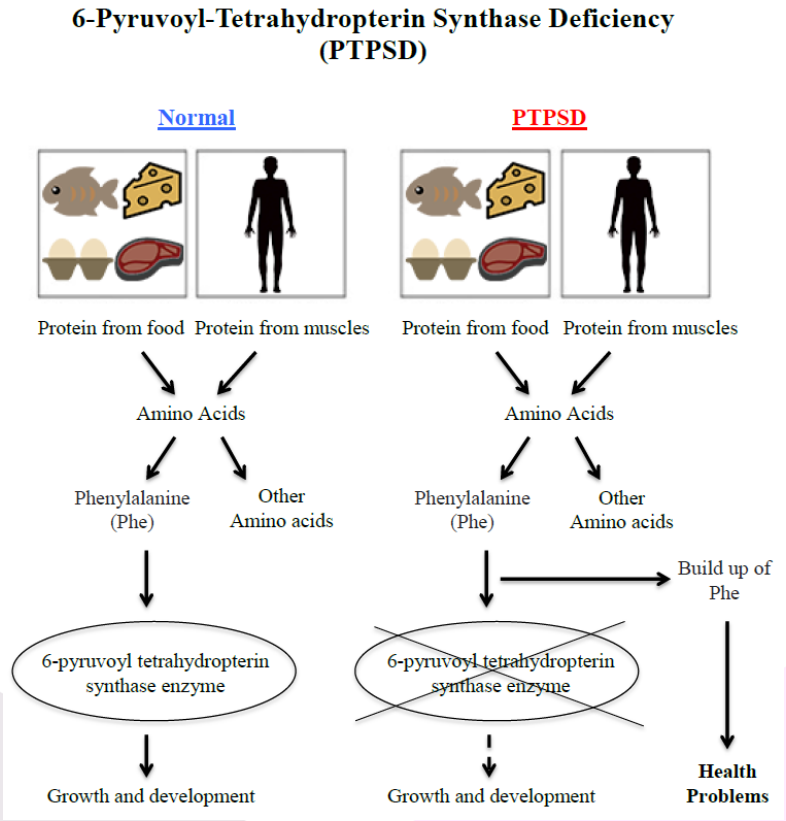
What is 6-pyruvoyl-tetrahydropterin synthase deficiency (PTPSD)?

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

After digestion, our body breaks down food into small molecules for further use. For example, protein is broken down into its building blocks, which is called amino acids. 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. Some enzymes require chemicals called co-enzymes for proper functioning.

Babies with PTPSD lack the enzyme called "6-pyruvoyl-tetrahydropterin synthase", an enzyme needed for production of a chemical called "tetrahydropterin" (BH4).

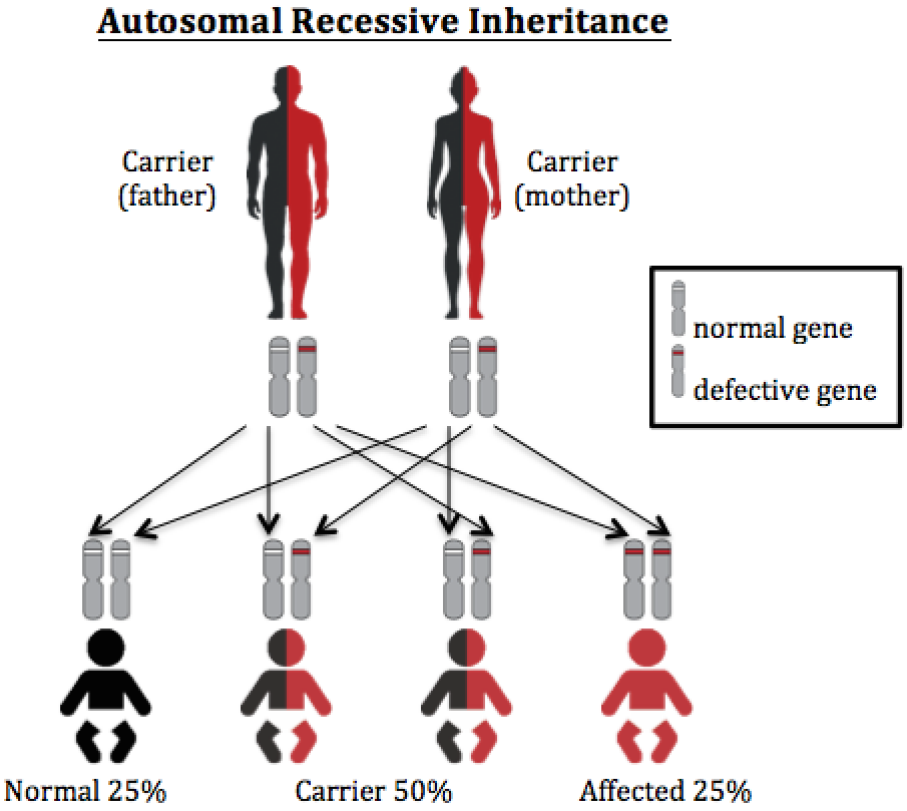
BH4 is an essential co-enzyme for several important enzymes in the body. Inadequate BH4 will affect the conversion of amino acid phenylalanine into another amino acid called tyrosine by the other enzyme. Tyrosine, which is vital for the brain functioning, may be insufficient. The function of neurotransmitters of which transmitting signals through nerve cells will also be affected. Moreover, the accumulation of phenylalanine causes long term brain damage.



How is PTPSD inherited?

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

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



What may happen if your baby has PTPSD?

Babies with PTPSD are usually healthy at birth. They gradually develop progressive and irreversible brain damage, leading to neurological disorders, learning difficulties and behavioural problems.

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

Possible signs and symptoms of PTPSD

- Developmental delay and floppiness in infants
- Intellectual disabilities, slow thinking in older patients
- Behavioural problems, hyperactivity, attention deficits
- Seizures
- Poor growth, fair skin and hair, eczema

What is the treatment for PTPSD?

PTPSD can be treated with BH4 and medicines that help in restoring the normal function of signal transmission through the nerve cells.

Patients with PTPSD need to see their specialist metabolic team and have blood tests regularly even when they do not have symptom.

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 hospital, bring the prescribed medicines, any information that you have been given about PTPSD, including this pamphlet, with you.