Overview of Phenylketonuria (PKU)

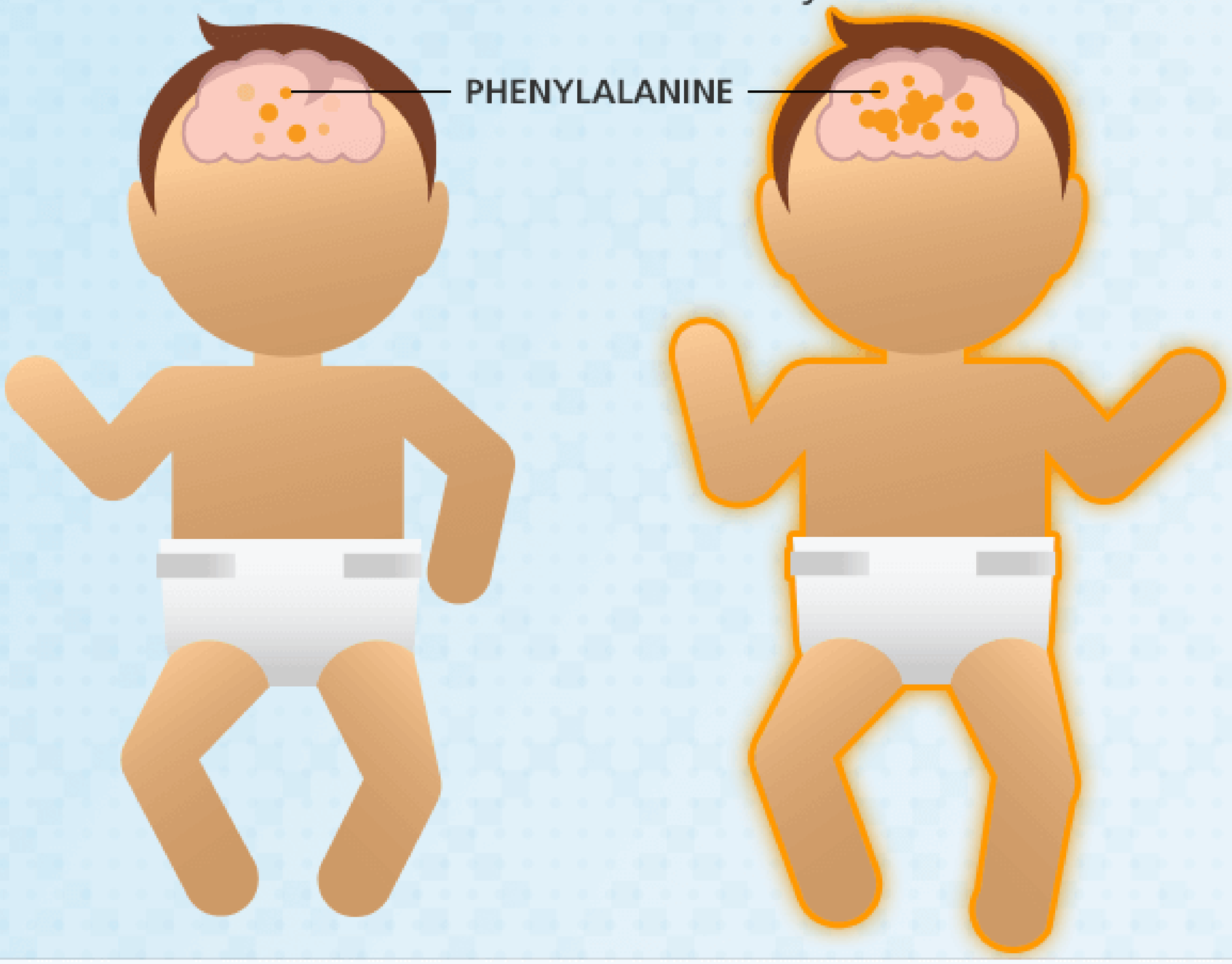
INTRODUCTION: WHAT IS PHENYLKETONURIA?

Phenylketonuria (also called PKU) is a rare condition that makes it hard for your baby to breakdown a substance called phenylalanine, or Phe for short. If the body cannot break down this substance, it can build up in the body and cause damage to the brain.

Phe levels are tested as part of newborn screening. While your child was found to have a high blood Phe, there's a lot you can do to prevent or minimize any risks.

Usually, the body is able to break down phenylalanine

If it cannot be broken down, phenylalanine builds up in the body



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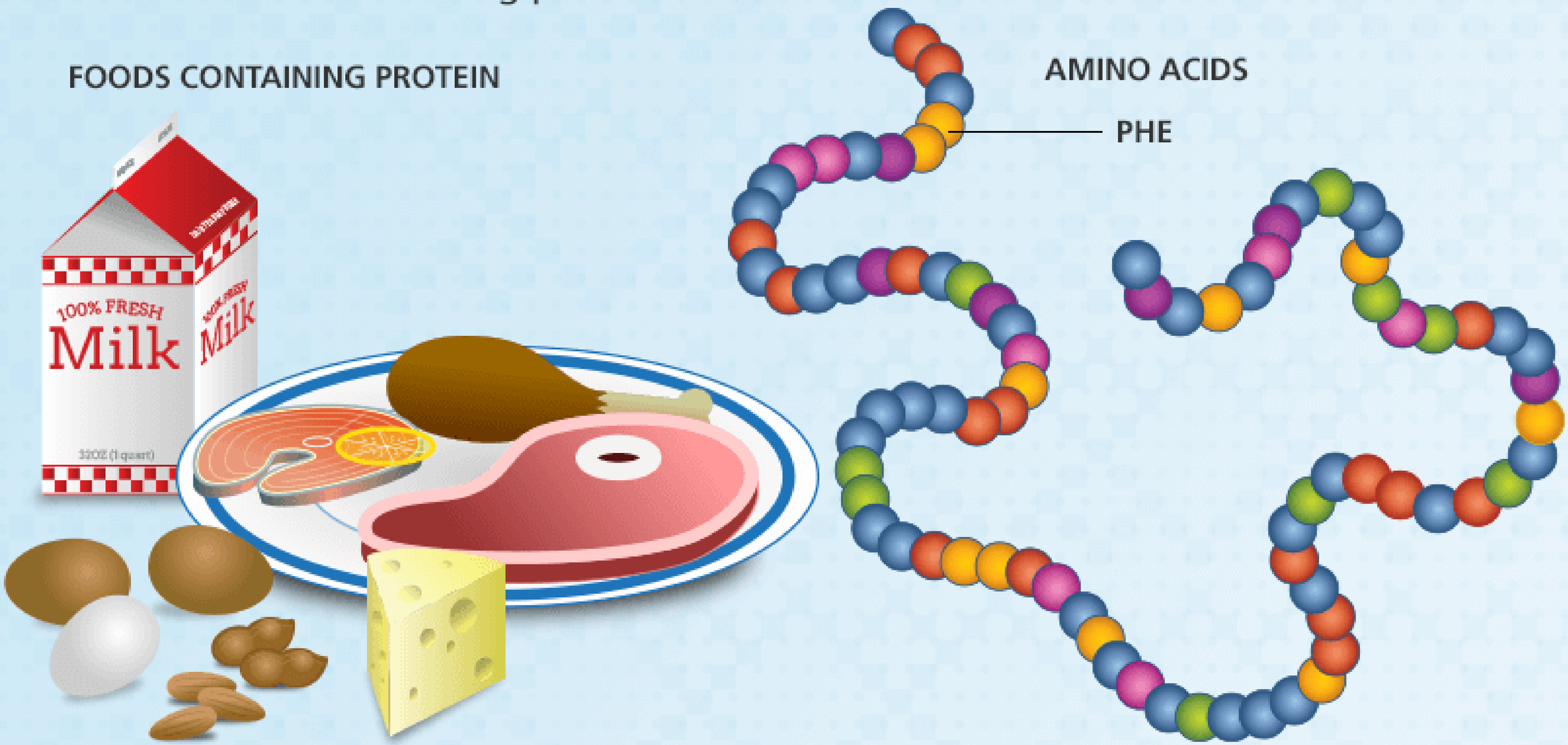
WHAT IS PHE?

Protein is made up of 20 different amino acids that are linked together. **Phe** is one of the amino acids in protein. The body needs **Phe** but it cannot make it itself. It comes from foods containing protein.



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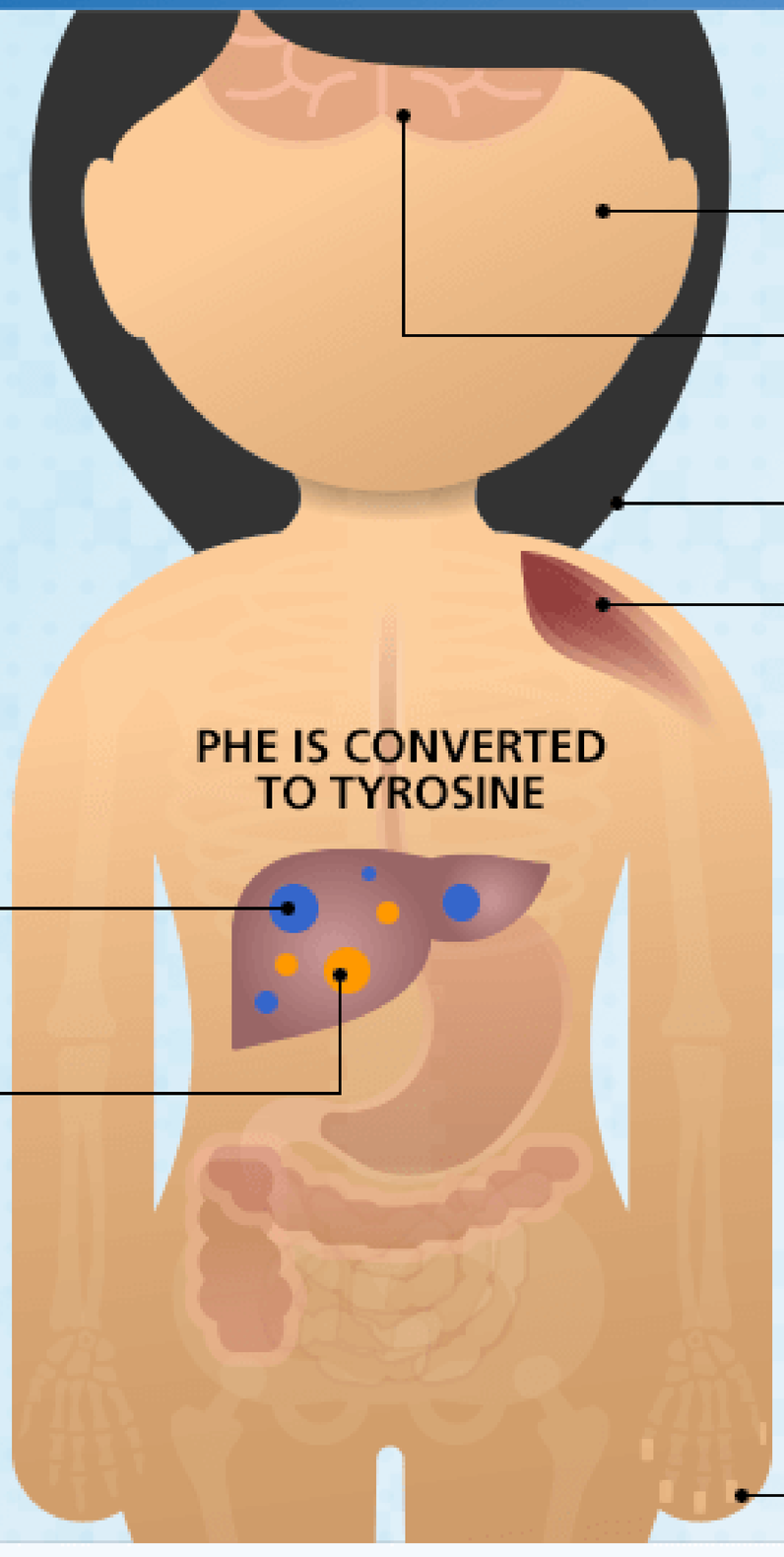
WHAT HAPPENS TO PHE IN THE BODY?

When we eat a food containing protein, it is broken down into amino acids (including Phe) and then further processed (or metabolized) in the liver. In the liver, Phe is usually changed to another amino acid called tyrosine.



TYROSINE IS USED TO MAKE:

MELANIN, THE PIGMENT THAT GIVES SKIN ITS COLOR

CHEMICAL MESSENGERS IN THE BRAIN (NEUROTRANSMITTERS)

PHE IS USED TO:

BUILD BODY PROTEIN MUSCLE, SKIN, HAIR AND NAILS

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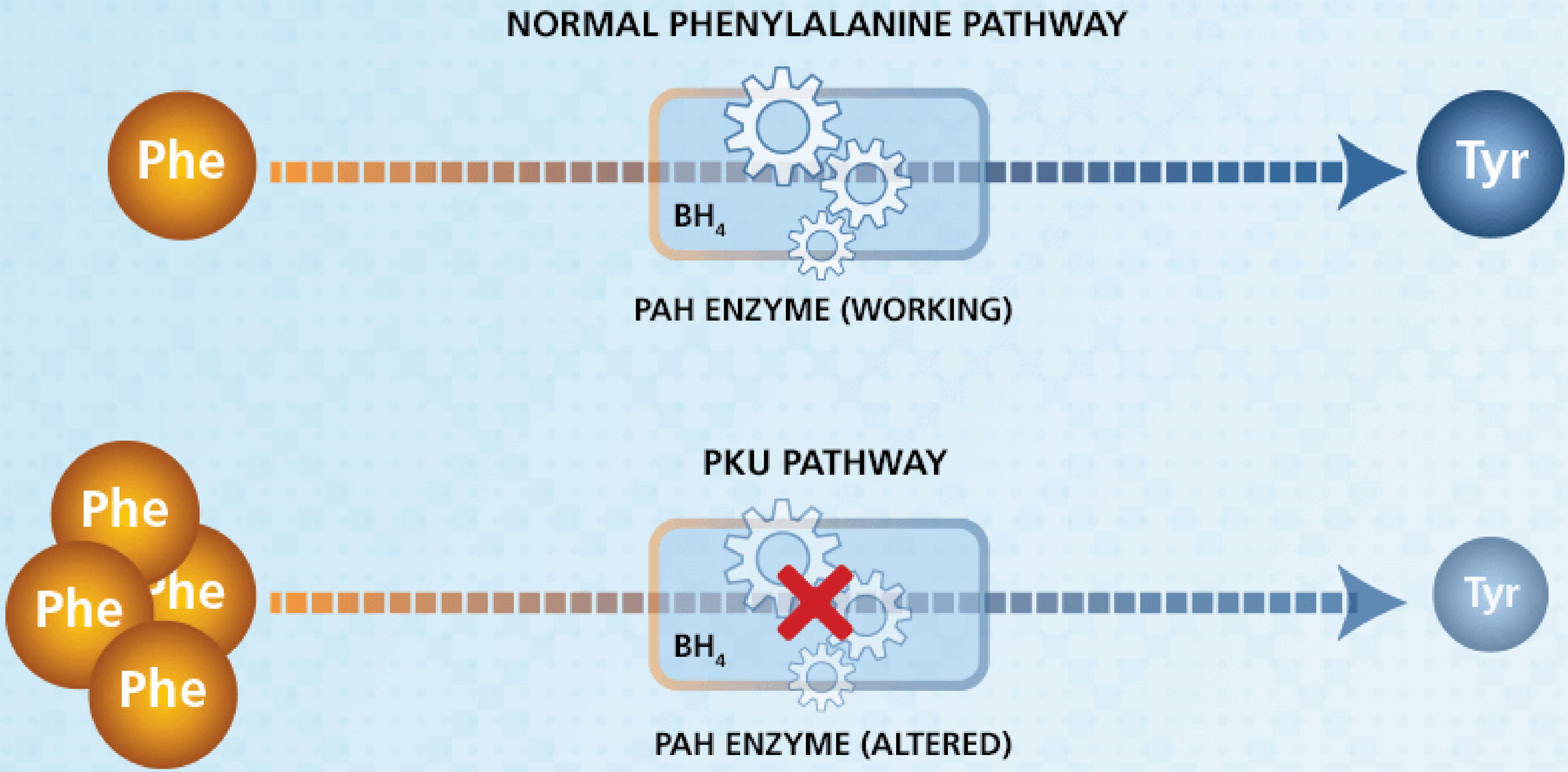
WHAT HAPPENS IF PHE CANNOT BE CONVERTED TO TYROSINE?

The process of changing Phe into tyrosine requires help from an enzyme called phenylalanine hydroxylase (PAH). When PAH does not work properly, there is too much Phe and too little tyrosine. This causes the problems seen in untreated PKU.



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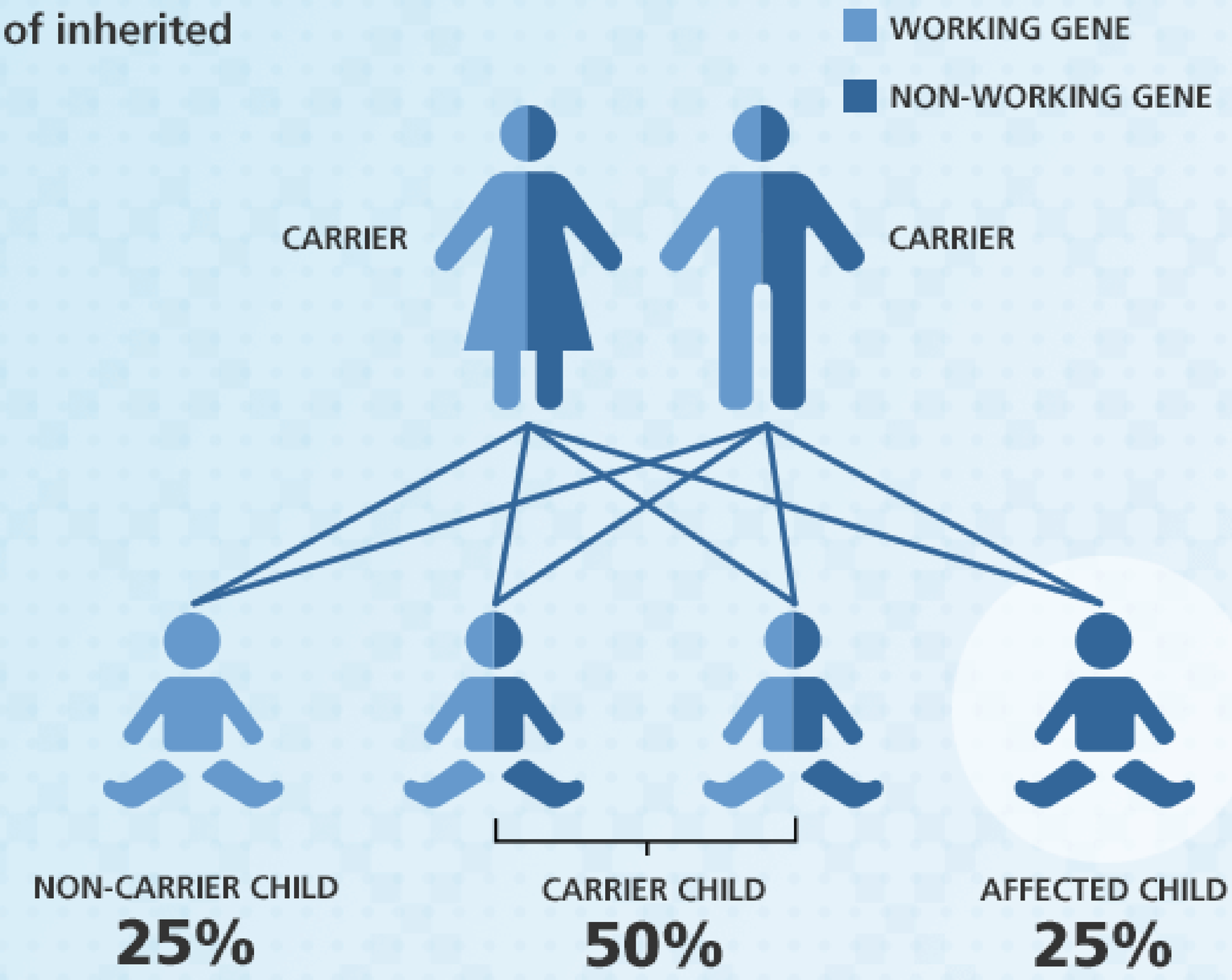
HOW DOES A BABY GET PKU?

PKU is an inherited or genetic condition. Genes come in pairs, one from each parent.

There are 3 possible combinations of inherited genes for each pregnancy:

- The baby may inherit 2 non-working genes (1 from each parent) and develop PKU
- The baby may inherit just 1 non-working gene—and then he or she is considered to be a carrier for PKU, but will not have the condition
- The baby may inherit 2 working genes and then the baby will not have PKU and will not be a carrier

There are many different types of non-working genes for PAH, some that code for milder forms and some that code for more severe forms of PKU. This is referred to as genotype.



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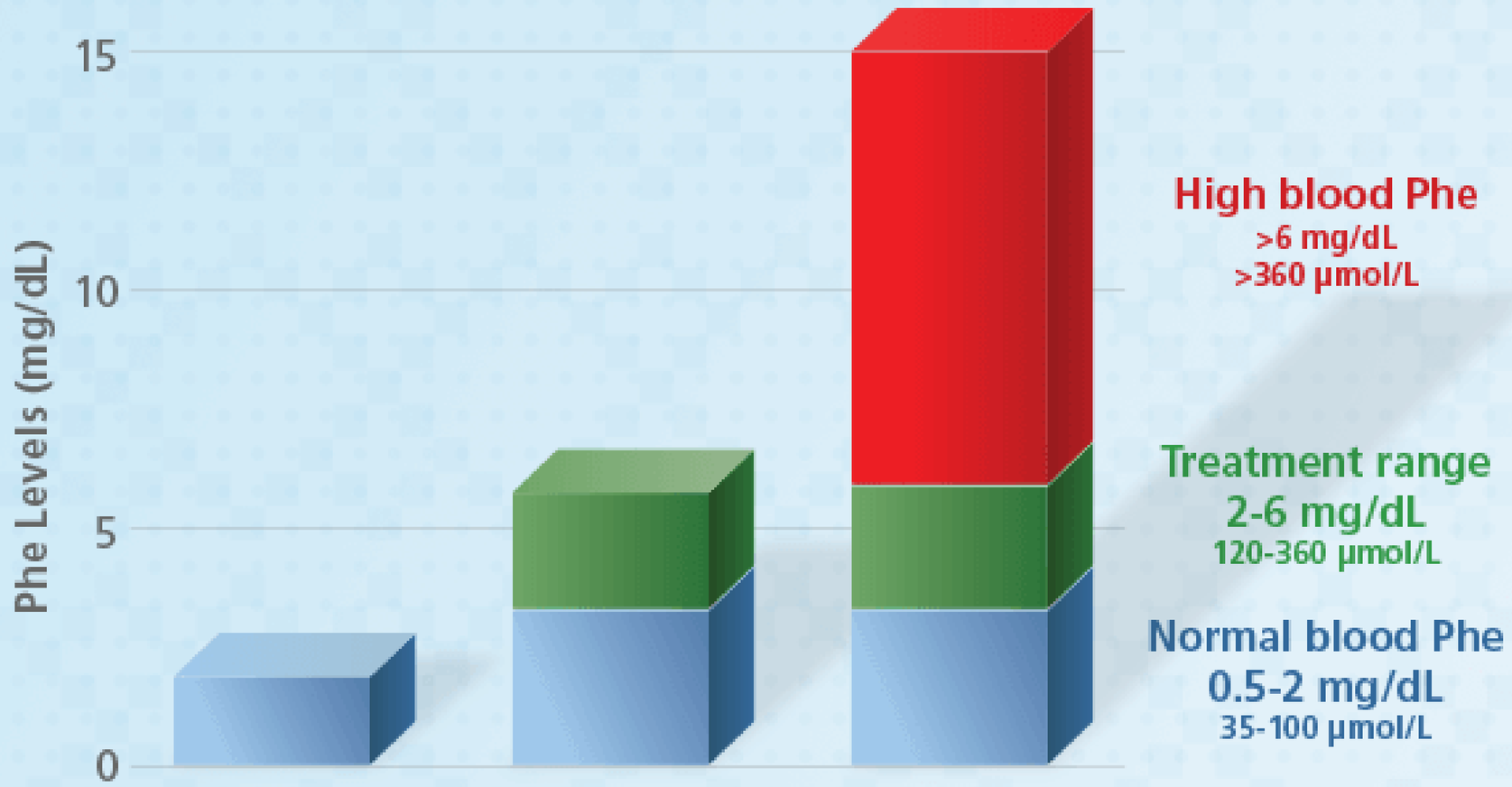
WHAT IS CONSIDERED A HIGH BLOOD PHE?

Blood phe higher than 6 mg/dL (>360 µmol/L) requires treatment.



Your child's newborn screening blood Phe was:

Your child's blood Phe today is:

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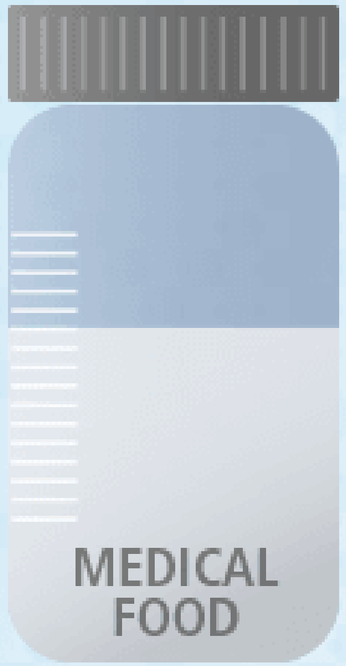
HOW IS PKU TREATED?

The primary treatment for PKU is a low-Phe diet. Since Phe is a part of protein, this will mean limiting how much protein and Phe your child gets from food and including a special kind of protein to his or her diet.

As an infant, your baby will also have restricted amounts of breast milk or regular infant formula.



There are special PKU formulas (called medical foods) that are taken throughout life. These supply the protein your child needs without the Phe.

As your child grows and you introduce solid foods, the diet will contain low Phe foods such as fruits, vegetables, and low protein foods.

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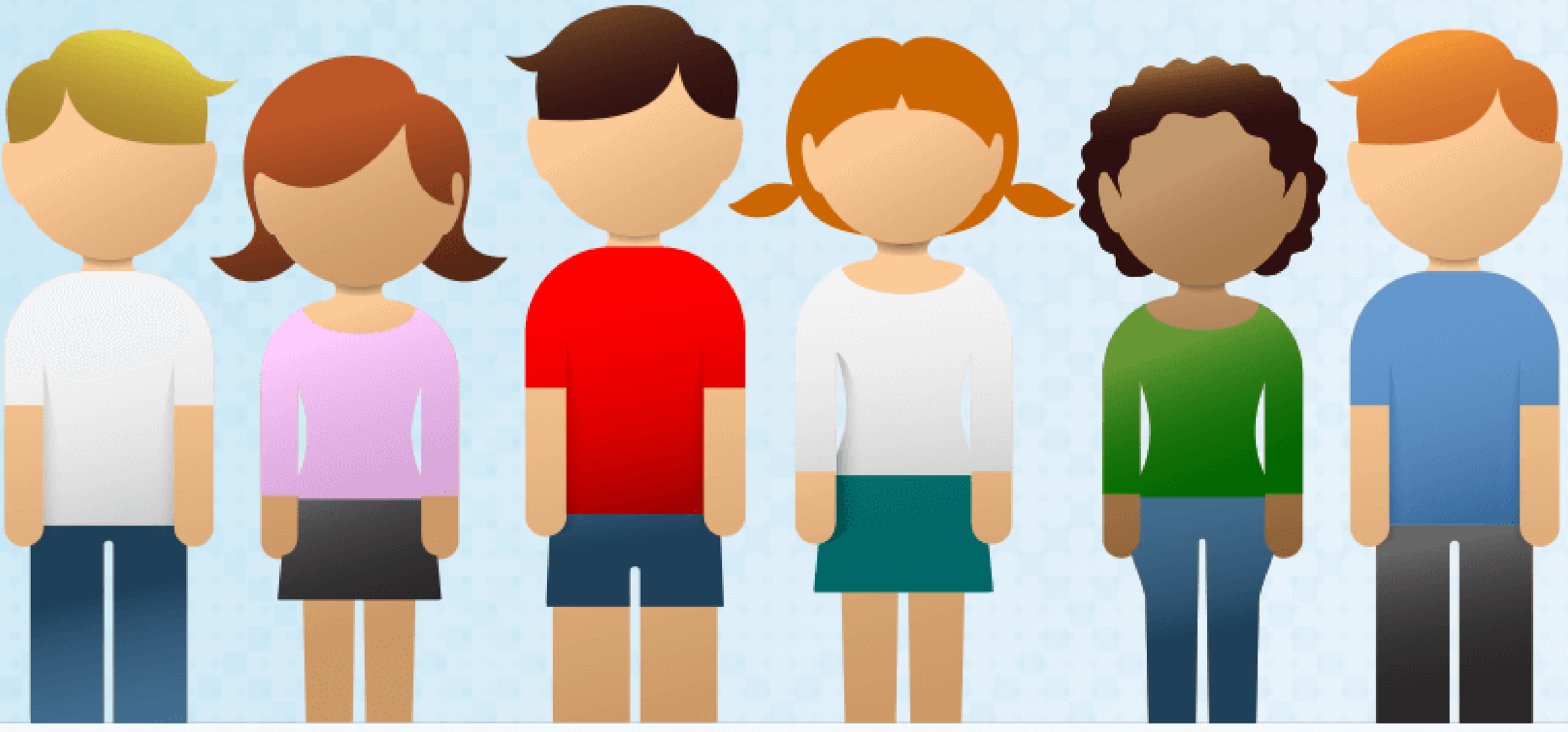
WHAT IS THE OUTLOOK FOR MY CHILD?

If treatment is started early and blood Phe kept in the treatment range, your child is expected to grow and develop normally.

You will need to work with your dietitian to plan a diet that meets your child's needs.



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FOR MORE INFORMATION

National PKU Alliance
(family support group):
www.npkua.org/

Fact sheet on PKU:
http://www.newbornscreening.info/Parents/aminoaciddisorders/PKU.html

CDC website on PKU with links to scientific articles:
http://www.cdc.gov/genomics/public/features/PKU.htm

DISCLAIMER

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