# Phenylketonuria

## What is phenylketonuria?

Phenylketonuria (commonly known as PKU) is an inherited disorder that increases the levels of a substance called phenylalanine in the blood. Phenylalanine is a building block of proteins (an amino acid) that is obtained through the diet. It is found in all proteins and in some artificial sweeteners. If PKU is not treated, phenylalanine can build up to harmful levels in the body, causing intellectual disability and other serious health problems.

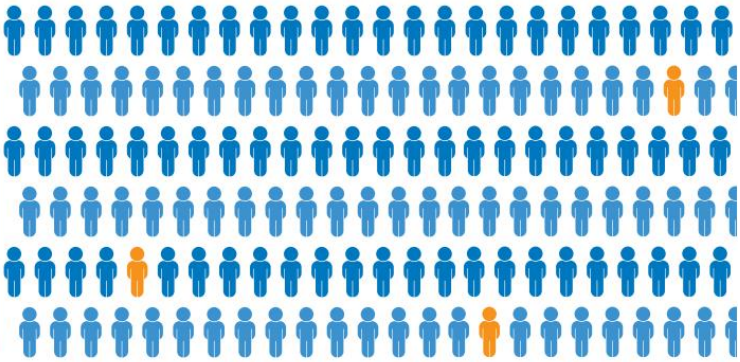
The signs and symptoms of PKU vary from mild to severe. The most severe form of this disorder is known as classic PKU. Infants with classic PKU appear normal until they are a few months old. Without treatment, these children develop permanent intellectual disability. Seizures, delayed development, behavioral problems, and psychiatric disorders are also common. Untreated individuals may have a musty or mouse-like odor as a side effect of excess phenylalanine in the body. Children with classic PKU tend to have lighter skin and hair than unaffected family members and are also likely to have skin disorders such as eczema.

Less severe forms of this condition, sometimes called variant PKU and non-PKU hyperphenylalaninemia, have a smaller risk of brain damage. People with very mild cases may not require treatment with a low-phenylalanine diet.

Babies born to mothers with PKU and uncontrolled phenylalanine levels (women who no longer follow a low-phenylalanine diet) have a significant risk of intellectual disability because they are exposed to very high levels of phenylalanine before birth. These infants may also have a low birth weight and grow more slowly than other children. Other characteristic medical problems include heart defects or other heart problems, an abnormally small head size (microcephaly), and behavioral problems. Women with PKU and uncontrolled phenylalanine levels also have an increased risk of pregnancy loss.

# PHENYLKETONURIA (PKU) AT A GLANCE

It is a **rare genetical condition caused by the metabolism of proteins which affects about one person out of ten thousand.** It determines a **toxic concentration of phenylalanine** in the body and a severe **deficit of tyrosine.**



## Does a therapy exist?

### DIET

- ✓ Preferring types of food with little proteins (fruit and vegetables)
- ✓ Eating special low protein food

### AMINO ACIDS

- ✓ Food for special medical purposes available in different formats
- ✓ essential to balance the lack of important amino acids which cannot be ingested through the diet.

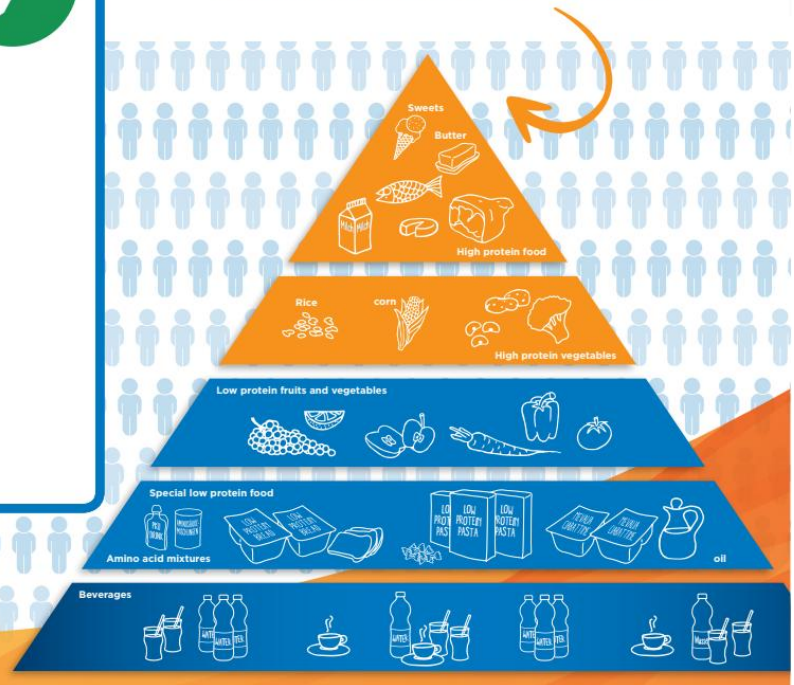
## THE RESULTS OF A NON TREATED PKU

- Severe psychological and physical developmental disorder
- Neurological disorders
- Reduced life expectancy

## THE BENEFITS OF A PROPERLY TREATED PKU

- Normal development and growth of the child
- Absence of neurological and psychiatric deficits
- Normal socialisation and integration into society

## THE PKU FOOD PYRAMID

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