PKU
Phenylketonuria

Guide for daycare services and school

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2014

What is PKU?

PKU is for Phenylketonuria

Phenyl**Ke**ton**U**ria

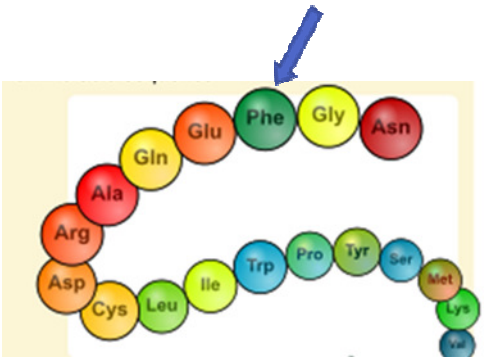
Phenyl = phenylalanine
Ketone = toxique product
Uria = urine

New term: Phenylalanine hydroxylase (PAH) Deficiency

PKU is a genetic disorder that prevents the individual to metabolize (digest) an amino acid found in proteins. This amino acid is called **Phenylalanine (Phe)**. People with PKU lack the enzyme needed to metabolize phenylalanine.

Without the enzyme, Phe accumulates in the blood and becomes toxic. High Phe concentrations can cause **permanent brain damage**. Untreated patients generally have severe neurological damage.

Today, PKU is diagnosed at birth and treatment is started as soon as possible. When the treatment is followed, these individuals usually develop normally and lead normal lives.



Symptoms of <u>untreated</u> PKU

- Severe intellectual and neurological deficiencies
- Difficulty with motor / coordination / reflexes
- Atypical behavior / autistic traits
- Convulsions
- Eczema or "rash"
- Musty or mousy odor

The main role of the missing enzyme is to convert Phe into **tyrosine.** Patients with PKU receive a tyrosine supplement because their body is unable to produce it. Tyrosine is essential for maintaining brain function and plays a role in:

- Cognitive functions
- Motivation
- Behaviour regulation
- Attention
- Memory
- The ability to learn

TREATMENT

The only effective treatment is a restricted phenylalanine diet that must be followed for life. The goal of the diet is to maintain Phe levels below 360 umol /L. Since phenylalanine is found in proteins, the individual must limit the amount of <u>all sources</u> of protein in his/her diet.

Protein is important for growth, repairing cells, fighting bacteria, etc. The PKU child can consume a very small amount of natural protein. This amount would not be sufficient to meet nutritional requirements. The child must drink a **special formula** that contains amino acids (proteins) and vitamins but that does not contain any phenylalanine. The formula is the most important food in the child's diet and we encourage that it be taken 3 - 4 times a day (such as meals).

Parents must calculate all foods that are consumed to ensure that the child receives the right amount of natural protein and formula every day. In addition to the Phe-free formula, the PKU Clinic offers a range of special low protein products like pasta, crackers and bread.

Some foods are **never allowed** because they are too high in phenylalanine:

- Meat, fish, seafood
- Eggs, nuts, legumes (soy beans)
- Dairy products (yogurt, cheese, butter)
- Grain products (flour, pasta, bread)

In general, most fruits and vegetables are unlimited.

PKU is not an allergy! If the child eats a high protein food, he or she will not have any discomfort or life-threatening reaction. In this case, it is still very important to notify the parent of the food that was consumed and its amount. The parent will be able to adjust the diet for the rest of the day and try to keep Phe levels in the optimal range. However, even if consuming a high protein food does not cause any immediate damage, these foods are to be avoided at all costs.

Diet compliance can be very difficult to maintain. Both children and adults may tend to "cheat" because they want to "be like others", don't find the time to prepare their food / formula, or have developed a taste for these non-permitted foods. It is important that the child does not develop a taste for these foods and must only consume the foods that are allowed.

When Phe levels are higher, the following symptoms may occur:

- Tiredness
- Difficulty sleeping
- Difficulty concentrating
- Behaviour problems
- Depression, anxiety

Many say they feel "in a haze".

MONITORING PHE LEVELS

Parents check the Phe levels about twice a month by collecting a blood sample at home. Blood is drawn from the fingertip and absorbed onto filter paper. This paper is sent by mail to the laboratory and the result is received about a week later. The optimal zone for Phe levels ranges from 120 to 360 umol/ L. However, it happens that the levels fluctuate outside this area.

Phe levels are influenced by:

- the quantity of ingested protein
- illness (flu, fever, surgery)
- insufficient intake of calories / protein in the diet

Regular followups with the PKU Clinic are necessary to adjust the diet as needed and to ensure optimal treatment.

WATCH FOR EXECUTIVE FUNCTION DIFFICULTIES

Executive function difficulties are some of the most common difficulties associated with PKU. This is sometimes true even for persons for whom PKU has been well controlled from an early age.

What are Executive Functions?

A set of skills that help you plan and execute activities or tasks effectively. They can be classified into two categories of skills which are related and overlap:

Cognitive skills needed to identify and achieve goals and to solve problems	*Skills needed to manage and regulate behaviour and emotions*
• Planning • Organization of material • Time management • Working memory • Metacognition	• Inhibition • Flexibility • Emotional control • Initiation and persistence • Sustained attention

The higher the Phe levels or the more they fluctuate, the more the child may have problems with executive functioning, behaviour, mood, etc. For some children, executive dysfunctions are present more or less permanently, but the severity of difficulties may still vary with the Phe level.

The psychometrist / psychologist at the PKU Clinic can conduct a psycho-educational assessment as needed. She is also available to offer advice and to develop and implement intervention plans in collaboration with the family and the school in order to help optimize the child's learning abilities in, functioning and well-being.

ROLE OF CARETAKER / TEACHER

Caretakers and teachers have an important role in how the child will perceive their condition and follow the treatment. Children with PKU are just like other children, except that they must follow a special nutritional treatment.

What you can do to help:

- The child should only eat food he brought from home or that was permitted by parents in a specific amount.

- Return all uneaten food in the lunch box.
- Notify parents in advance if there is a special class event that includes food (birthday, special occasion) so that parents can prepare a special low protein snack.
- Remember that children with PKU cannot consume nuts, dairy or eggs. Their snacks are often homemade baking goods because low protein "ready to serve" foods and snacks are limited and not always available. These foods/ snacks are not a risk for others with allergies. For example, a low-protein product that resembles “peanut butter” is available, but it does not contain any nuts, dairy products or eggs.
- Encourage the child to drink his/her formula at school.
- Inform parents if the child has or may have consumed a non-permitted food.
- Organize classroom activities that show that everyone is unique and that being different is fun!