PHENYLKETONURIA (PKU)
CONSUMER SUMMARY

SERN/GMDI Nutrition Management Guidelines **Second Edition**

Nutrition Guidelines for Individuals with PKU [1,2,3] July 2022

This information is intended for individuals with Phenylketonuria (PKU) and/or their caregivers (hereafter referred to as "you/ your"). This summary includes current management recommendations and suggests topics that you can discuss with your metabolic team to help identify a plan that is best for you.

- Follow your metabolic clinic's instructions to meet your dietary needs for phenylalanine (PHE), protein, calories, and other nutrients. The amounts prescribed are based on your age, weight and height, activity level, blood PHE control, and lab results. Ask your metabolic clinic if you should receive any additional testing or supplements.
- Your metabolic clinic will recommend a PHE and/or dietary protein restriction needed to keep your blood PHE levels within a safe treatment range (usually 120-360 µmol/L). Your clinic will also prescribe the best medical food (formula) to maintain adequate growth and good nutrition. Together, your restricted diet and your medical food will support your PKU control and help to keep you healthy.
- Medications are now available for PKU treatment (Kuvan® and Palinziq™). Talk with your metabolic clinic about whether one of these may be appropriate for you, what you need to do to determine if one may help you with managing your PKU, and if it works for you how the medication could change for your diet and medical food treatment recommendations.
- Follow your metabolic clinic's recommendations for how often to take a blood sample, and what tests are needed. They may recommend testing for nutritional status as well as PKU control. They will also recommend how often to attend metabolic clinic appointments. At each clinic visit, your weight and height, and (head circumference in young children) may be measured to decide if your treatment should be modified.
- Follow your clinic's recommendations for keeping track of the PHE and/or protein you get from food and how much medical food you take. Keep a three day food diary prior to your blood test and bring it with you to clinic to help you follow your diet.
- Your metabolic clinic can help you with suggestions for following your diet while at school or work, resources for special needs, more information about PKU, and support groups that are available.
- If you are a female, your metabolic clinic will discuss the importance of blood PHE control (usually 120-360 µmol/L) **before becoming pregnant**. Blood PHE >360 µmol/L any time during pregnancy is known to cause significant birth defects. They may also offer advice on appropriate birth control.
- **If you are already pregnant**, this is considered a **medical emergency** and you should contact your metabolic clinic immediately.

This document is not meant to substitute for the medical advice provided by your doctor.

[1] *For the child, teenager, or adult living with PKU and their caregivers.*

[2] *Based on the 2015 Nutrition Management Guidelines for Phenylketonuria (PKU) by GMDI/SERN: https://southeastgenetics.org/ngp/guidelines_pku.php*

[3] *The Management Guidelines Advisory Committee used the nationally standardized condition abbreviation of PKU; curated by the US National Library of Medicine for this and related guideline products: https://newbornscreeningcodes.nlm.nih.gov/*

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Considering Pegvaliase Therapy? What You Need to Know

Eligibility:

Your clinic may consider offering you pegvaliase therapy if you are:

- over 18 years of age, and
- have a blood PHE >600 µmol/L, and
- if you are female, and are not planning a pregnancy in the next year

Some clinics may also consider offering you pegvaliase therapy if you are:

- currently on sapropterin, or
- have a blood PHE <600 µmol/L but want to achieve a normalized diet, or
- have a neurocognitive deficit (including late-treated individuals)

Some clinics may also consider pegvalise therapy for individuals as young as 16 years of age.

Considerations

Before beginning pegvaliase therapy, you should discuss the following with your clinic:

- possible side effects and how they may be treated,
- medication your clinic team may recommend when starting pegvaliase to help with side effects,
- what your clinic expects from you for clinic visits, blood tests, good communication, and reasons for stopping pegvaliase, and
- what you can expect after starting pegvaliase (time to response, dose/number of injections, and diet adjustments).

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What to expect

Your pegvaliase dose will be adjusted based on your blood PHE response and any side effects that you may experience, with a goal of a blood PHE concentration below 360 µmol/L and above 30 µmol/L while eating a healthy amount of protein.

If you are taking sapropterin, you may be asked to stop taking it once your blood PHE concentration is in your clinic's treatment range.

If your blood PHE falls too low, you may be asked to increase the amount of foods containing protein in your diet and/or decrease your pegvaliase dose.

If you are taking medical food, you may be asked to decrease or discontinue your medical food as the amount of protein from food in your diet increases.

If your blood TYR concentrations repeatedly fall below 30 µmol/L, your clinic may recommend that you increase the amount of protein in your diet and/or take a TYR supplement.

Women of childbearing age who are on pegvaliase therapy should use birth control to prevent unexpected pregnancies. If you are planning a pregnancy, or become pregnant while taking pegvaliase, discuss with your clinic your options for either returning to diet or continuing to take pegvaliase.

If you choose to return to a PHE-restricted diet for pregnancy, you should tell your clinic, and stop taking pegvaliase at least 4 weeks prior to a planned pregnancy.

If you choose to take pegvaliase while you are breastfeeding, your clinic will discuss options with you, including the need for frequent monitoring of your blood PHE concentration.

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