PHENYLKETONURIA (PKU)
CONSUMER SUMMARY

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

Nutrition Guidelines for Individuals with PKU [1,2,3] December 2018

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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