

# Nutritional Status and Dietary Intake in Adults with Phenylketonuria

Stefano Mrasouxinkul\*

Department of Pediatric Neurology, Oviedo University, 33006 Oviedo, Spain

## Introduction

Phenylketonuria (PKU) is a rare genetic disorder characterized by a deficiency in the enzyme phenylalanine hydroxylase, which is essential for the metabolism of the amino acid phenylalanine. If left untreated, elevated levels of phenylalanine in the blood can lead to severe neurological impairments and cognitive deficits. To manage PKU effectively, patients must adhere to a lifelong dietary regimen that restricts phenylalanine intake, primarily through a low-protein diet and the use of specialized medical foods and supplements. The dietary restrictions imposed by PKU can significantly impact an individual's nutritional status, potentially leading to deficiencies or imbalances in essential nutrients. For adults with PKU, who are navigating these dietary constraints over a long period, maintaining optimal nutritional status and ensuring adequate intake of all necessary nutrients presents a complex challenge. This paper explores the nutritional status and dietary intake of adults with PKU, focusing on the implications of dietary management on their overall health, and examines strategies to address potential nutritional deficiencies and promote long-term well-being [1].

## Description

Adults with PKU face unique challenges in managing their nutritional status due to the stringent dietary restrictions required to control phenylalanine levels. The standard treatment involves a diet low in phenylalanine, which typically means limiting protein intake from natural sources like meat, dairy, and legumes, while supplementing with specially formulated low-protein products. This restricted diet, while necessary for preventing neurological damage, can lead to inadequate intake of essential nutrients typically obtained from protein-rich foods, such as amino acids, vitamins, and minerals. Studies have shown that individuals with PKU often experience deficiencies in nutrients like vitamin B12, iron, calcium, and zinc, which are crucial for overall health and metabolic function. The reliance on medical foods and supplements, though helpful, may not fully compensate for the nutrient shortfalls resulting from the dietary restrictions [2].

Moreover, maintaining a balanced diet becomes increasingly difficult as adults with PKU age and their dietary needs evolve. The long-term adherence to a low-phenylalanine diet can affect bone health, muscle mass, and cardiovascular health, necessitating regular monitoring and tailored nutritional interventions. For instance, calcium and vitamin D levels need careful management to prevent bone density loss, while protein and amino acid intake must be monitored to support muscle maintenance and metabolic processes.

**\*Address for Correspondence:** Stefano Mrasouxinkul, Department of Pediatric Neurology, Oviedo University, 33006 Oviedo, Spain, E-mail: stefanomrasouxinkul@hotmail.com

**Copyright:** © 2024 Mrasouxinkul S. This is an open-access article distributed under the terms of the creative commons attribution license which permits unrestricted use, distribution and reproduction in any medium, provided the original author and source are credited.

**Received:** 11 June, 2024, Manuscript No. VTE-24-146211; **Editor Assigned:** 13 June, 2024, PreQC No. P-146211; **Reviewed:** 25 June, 2024, QC No. Q-146211; **Revised:** 01 July, 2024, Manuscript No. R-146211; **Published:** 08 July, 2024, DOI: 10.37421/2376-1318.2024.13.318

Research into dietary patterns, nutrient intake, and the effectiveness of supplementation strategies in this population is crucial to address these challenges. Innovative approaches, including personalized nutrition plans and advancements in medical foods, aim to enhance the dietary management of PKU and improve nutritional outcomes [3].

In addition to the challenges associated with nutrient deficiencies, adults with Phenylketonuria (PKU) often face difficulties in managing their dietary intake due to the complexity of their nutritional needs. The restriction of natural protein sources necessitates reliance on specially formulated low-protein products, which can be expensive and may not always provide the complete range of nutrients required for optimal health. For instance, while these products are designed to be phenylalanine-free, they may lack sufficient quantities of essential amino acids and micronutrients, necessitating careful planning to ensure that nutritional requirements are met. Furthermore, the absorption and bioavailability of nutrients from these medical foods can vary, impacting their effectiveness in addressing nutritional deficiencies [4].

Moreover, the long-term adherence to a low-phenylalanine diet can influence metabolic processes in ways that exacerbate nutritional imbalances. For example, restricted intake of protein-rich foods can lead to reduced levels of carnitine, an amino acid important for fatty acid metabolism, potentially affecting energy levels and metabolic efficiency. The potential for imbalances in fatty acids and other nutrients further complicates the management of overall health and metabolic stability. This underscores the importance of ongoing dietary assessment and adjustments to accommodate changes in an individual's health status and lifestyle over time [5].

## Conclusion

The nutritional management of adults with Phenylketonuria presents a complex interplay between dietary restrictions and the need for adequate nutrient intake. While the low-phenylalanine diet is essential for preventing the neurological complications associated with PKU, it also poses significant challenges for maintaining overall nutritional health. Deficiencies in essential nutrients such as vitamins, minerals, and amino acids are common among adults with PKU, highlighting the need for vigilant dietary planning and supplementation. Addressing these nutritional challenges requires a multifaceted approach, including regular monitoring of nutrient status, personalized dietary interventions, and advancements in medical food formulations. Continued research and development in this field are crucial for optimizing nutritional strategies and improving the quality of life for adults with PKU. By enhancing our understanding of the specific nutritional needs and potential deficiencies in this population, healthcare professionals can better support adults with PKU in achieving and maintaining optimal health throughout their lives.

## Acknowledgement

None.

## Conflict of Interest

None.

## References

1. Jaulent, Paul, Sybil Charriere, François Feillet and Claire Douillard, et al. "Neurological manifestations in adults with phenylketonuria: New cases and review of the literature." *J Neural* 267 (2020): 531-542.
2. Hochuli, Michel, Sandra Bollhalder, Carina Thierer and Julie Refardt, et al. "Effects of inadequate amino acid mixture intake on nutrient supply of adult patients with phenylketonuria." *Ann Nutr Metab* 71 (2018): 129-135.
3. Montoya Parra, Gina A., Rani H. Singh, Aysun Cetinyurek-Yavuz and Mirjam Kuhn, et al. "Status of nutrients important in brain function in phenylketonuria: A systematic review and meta-analysis." *Orphanet J Rare Dis* 13 (2018): 1-16.
4. MacDonald, Anita, A. M. J. Van Wegberg, K. Ahring and S. Beblo, et al. "PKU dietary handbook to accompany PKU guidelines." *Orphanet J Rare Dis* 15 (2020): 1-21.
5. Lage, Sergio, María Bueno, Fernando Andrade and José Ángel Prieto, et al. "Fatty acid profile in patients with phenylketonuria and its relationship with bone mineral density." *J Inherit Metab Dis: Official Journal of the Society for the Study of Inborn Errors of Metabolism* 33 (2010): 363-371.

**How to cite this article:** Mrasouxinkul, Stefano. "Nutritional Status and Dietary Intake in Adults with Phenylketonuria." *Vitam Miner* 13 (2024): 318.