

# PKU Pulse Newsletter

May 2025



From everyone here at PTC Therapeutics, we're excited to launch the **PKU Pulse**, where we will bring you the latest updates, community happenings, and personal stories from the PKU community. Whether you're personally living with PKU, caring for a loved one with PKU, an advocate, or provider, we are honored to partner with you on your journey. We look forward to sharing these updates. For more community stories and helpful resources, connect with Reimagine PKU on our new [Facebook](#) and [Instagram](#) pages!

## This Month's Feature: Minding Your Mental Health

Living with PKU can present unique mental health challenges. The intersection of PKU Awareness Month and Mental Health Awareness Month in May highlights the significant impact PKU can have on mental health<sup>1</sup> and underscores the necessity of taking proactive steps to care for both mental and physical well-being.<sup>2</sup>

People with PKU can experience anxiety<sup>1</sup>, depression<sup>1</sup>, and brain fog<sup>3</sup> – daily challenges that can be exacerbated by needing specialized dietary requirements at work, school, or social events.<sup>3,4</sup> This can lead to feelings of isolation and make routine activities, like dining out with friends or traveling, more difficult.<sup>5,6</sup>

The mental health struggles a person with PKU may experience are real; high blood Phe levels can disrupt neurotransmitters like serotonin and dopamine and affect mood.<sup>1,7</sup>

*"It's extremely important to aim for target phenylalanine levels throughout the lifespan of a person living with PKU. **We see higher rates of anxiety and depression in patients that can be exacerbated by higher levels of phenylalanine.**"*



**Erika R. Vucko, MSN, FNP-BC, AGN-BC**

Advanced Practice Provider, Division of Genetics,  
Ann & Robert H. Lurie Children's Hospital of Chicago

## Consider these tips as part of your ongoing mental health check-in:



### Diet Management

Adhering to a Phe-restricted diet could help stabilize blood Phe levels in some patients, which may alleviate some psychological symptoms.<sup>2,8</sup> Planning meals ahead and finding enjoyable and Phe-friendly foods can make social situations less daunting.



### Seek Support

Connecting with others who have PKU can provide a sense of community and understanding. Online forums, support groups, and social media communities can be excellent resources for sharing experiences and advice.



### Expanding Your Care Team

Consulting with mental health professionals can provide strategies tailored to managing the emotional aspects of PKU. Therapy or counseling can offer a safe space to explore feelings of anxiety, depression, or isolation.



### Education and Advocacy

Educating friends, family, and colleagues about PKU can foster understanding and support in social settings. Advocacy can also empower individuals to take control of their health and wellbeing.

**Mental health matters just as much as physical health,** and there are resources available to support those living with PKU on their mental wellness journey.

For more information:

[Visit ReimaginePKU.com](https://www.ReimaginePKU.com)

*"The side effects of PKU and having high Phe levels are far and wide depending on the person. **I've noticed that for me, I sleep poorly or I get frustrated noticeably quicker if I have higher Phe levels.** One of the effects of having a rare disorder like PKU is that you never quite know – is this PKU, or just being a human? Am I frustrated because of my diet, or did I just wake up on the wrong side of the bed? I play this self-assessment game a lot!"*



**Kurt**

Living with PKU and creator of "The Low-Phe Life" web series

## News You Can Use

This year, the American College of Medical Genetics and Genomics (ACMG) published an updated clinical practice [guideline](#) for the diagnosis and management of PKU.

## The key takeaways<sup>2</sup>:

- The guideline strongly recommends maintenance of Phe  $\leq 360$   $\mu\text{mol/L}$  to help support optimal intellectual outcomes.
- The guideline recommends treatment approaches that may include dietary protein restriction, Phe-free protein supplementation with medical foods, and pharmacologic therapies.

## Community Champions The New England Connection for PKU and Allied Disorders (NECPAD)



A volunteer non-profit organization dedicated to supporting PKU and Allied Disorders communities across New England (Maine, Vermont, New Hampshire, Massachusetts, Connecticut, and Rhode Island).

NECPAD's mission is to provide support to all individuals and families with PKU and allied disorders and clinicians, and to encourage research and increase public awareness of the conditions. Visit NECPAD's website to stay updated on many opportunities for connection, resources, and more.

[Visit necpad.org](https://necpad.org)

## Upcoming Events

June 5-6 (Antelope, OR)  
**flok's Family Camp West**



June 6-9 (Spring City, PA)  
**Alliance of PKU Families' Camp Huber**



June 9-14 (Atlanta, GA)  
**Emory University Metabolic Camp**



June 13-15 (Prescott, AZ)  
**ANPAD Camp Knot a Phe**



June 22 (Boston, MA)  
**NECPAD Kimball Farms**



June 27-28 (Toole, UT)  
**IPAD Family Camp**





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