# Impact of PKU on daily life: Insights from a patient survey of 46 adults in Germany

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## Background & Objective

- Although experiences of medical professionals with phenylketonuria (PKU) management are well described in the literature, publications on perspectives from individuals with PKU remain limited[1]
- While high blood phenylalanine (Phe) levels in individuals with PKU are associated with neurocognitive deficits, impaired executive function, and reduced quality of life, lifelong adherence to a restrictive low-Phe diet is challenging, and few individuals of any age maintain recommended blood Phe levels with dietary restriction alone[2,3]
- Insights from individuals with PKU can provide valuable information on the management of PKU, the potential challenges of living with PKU, and possible gaps in PKU care. Herein, we present the outcomes of a patient survey, including verbatim quotes from individuals with PKU

## Methods

- A questionnaire to assess the impact of PKU on adult's personal and professional lives was developed to explore a wide variety of important topics such as the impact on personal and professional life, family planning and experiences with clinical care
- Individuals with PKU were recruited through the German patient advocacy group DIG-PKU (Deutsche Interessengemeinschaft Phenylketonurie) and metabolic centers specializing in PKU treatment
- The questionnaire was completed by adults (≥18 years) with PKU following registration via QR code. They provided informed consent prior to participation, and received compensation for their time
- Participants rated items on a 0-10 scale, with higher scores indicating greater perceived difficulty or impact
- Optional free-text fields allowed participants to share their experiences

## Results

- 46/71 registered individuals with PKU (52% female) completed the questionnaire. The respondents had a mean age of 33 years. Mean blood Phe level was 627 µmol/L (30-1,507 µmol/L, based on the last measured value)
- As the survey focused on perceived difficulties related to PKU management and recruited participants through clinics and patient networks, the findings may overrepresent negative aspects of the participant experience while underrepresenting individuals lost to follow-up or care. Results should be interpreted within this context

### Impact of PKU and dietary restriction on daily life

- Strict dietary adherence remains challenging for adults with PKU, with cost, limited motivation, time demands, and poor taste of Phe-free foods identified as key barriers **(Figure 1)**

> "As a working adult, it's very difficult to stick to the diet. If you have a family, it's even more difficult, as you have to cook two different meals. Often there is no time or energy left to prepare something low in protein. You can't just spontaneously buy something at a bakery or somewhere else either."

> "Sometimes feeling like I'm not a full person. Not being able to participate in social events [eating] like everyone else. Rarely, but still stressful, stupid comments from others. You feel ashamed for eating differently and then comments like 'Yuck. What are you eating? What does that look like?'"

**Figure 1. Participant-perceived biggest challenges of a low-Phe diet**

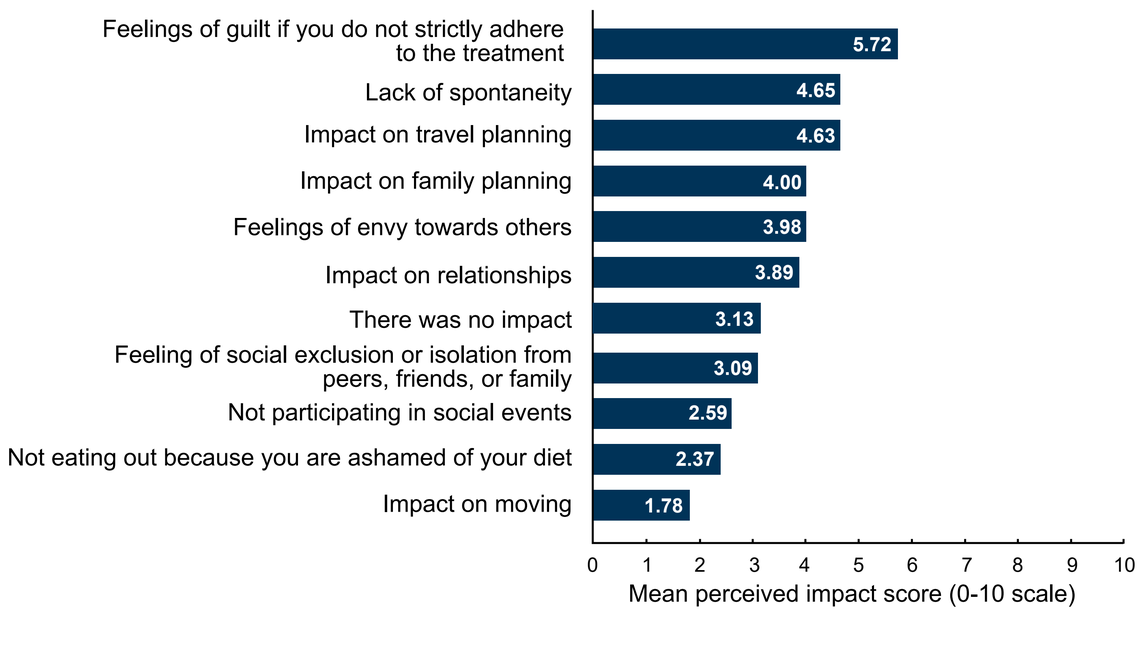
| | Mean perceived impact score (0-10 scale) |
|---|---|
| Cost of Phe-free foods | 6.67 |
| Limited motivation to stick to the diet | 6.50 |
| Time constraints associated with adhering to the diet | 6.09 |
| Poor taste of Phe-free foods | 5.24 |
| Limited variety of Phe-free foods | 4.74 |
| Feeling hungry | 4.37 |
| Lack of awareness of the effects of the diet on your well-being | 4.35 |
| Feeling socially isolated when eating with friends | 4.26 |
| Access to prohibited foods at school (eg, trading with others) | 3.80 |
| Lack of support from those around you in sticking to the diet | 3.74 |
| Problems with the availability of Phe-free foods | 3.59 |

> "Eat at home as often as possible, check menus in advance if necessary, and choose restaurants accordingly. In an emergency, eat at home beforehand and only have a small snack with friends, eg, a salad."

> "The care of patients with PKU who want to have children is inadequate in Germany. No time is taken and there are no resources in the clinics for staff or information events."

- Responses indicated that PKU interferes with daily life, with guilt related to dietary non-adherence, reduced spontaneity, and impacts on travel, family planning (mainly the decision to have children) and social life **(Figure 2)**

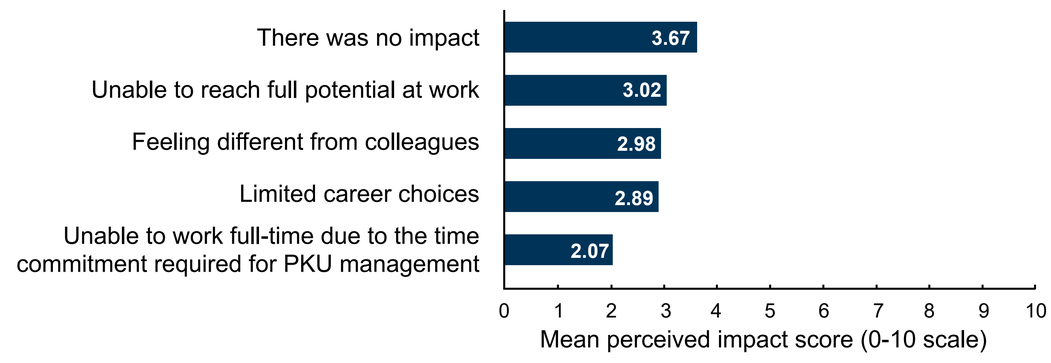
**Figure 2. Participant-perceived impact of PKU on personal life**




- Overall, the perceived impact on the respondents' professional life was lower, but there were struggles with being unable to reach full potential at work, feeling different from colleagues and limited career choices **(Figure 3)**

> "Due to the high prices of dietary foods, which are not covered by health insurance, I feel very limited in my future career choices. I therefore prioritize earnings over interest (eg, public service rather than research or journalism). Interest can therefore only be pursued on the side and not completely in line with my career."

**Figure 3. Participant-perceived impact of PKU on professional life**



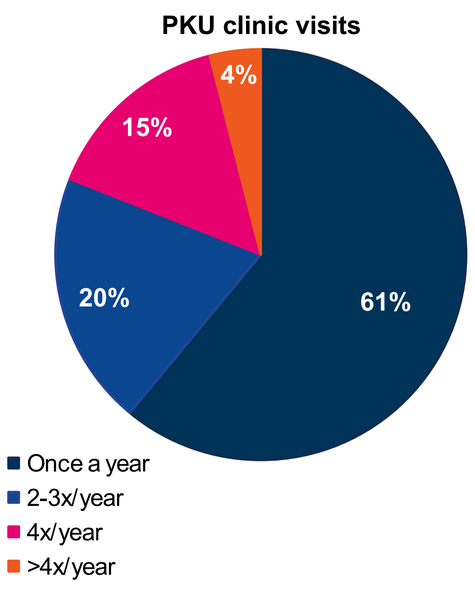

### Challenges in PKU clinical care and follow-up

- Most participants (61%) reported seeing their treatment team only once per year **(Figure 4)**, and 71% did not transfer to an adult clinic after turning 18. Of those transitioning to adult care, 42% reported challenges during the process

> "There was no real transition. We were 'kicked out' of one clinic and had to find the next point of contact for adults with PKU ourselves. There was no coordination or information about where we should go."

> "I am still being treated at a children's hospital, but they now take adult PKU very seriously, provide good support, and reliably issue prescriptions. The biggest problems tend to be referrals from my family doctor."

**Figure 4. Frequency of follow-up with the treatment team**

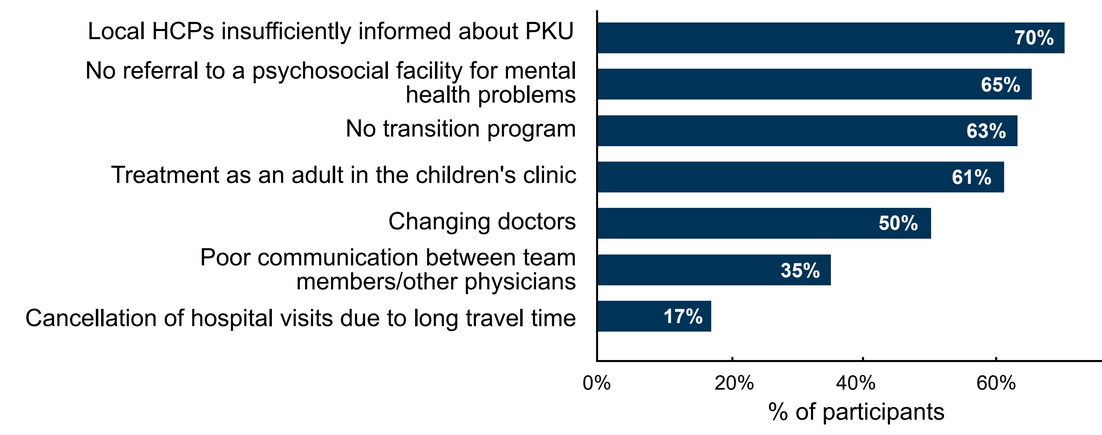



- Most reported challenges with the treatment team included lack of informed local healthcare professionals (HCPs), psychosocial support and transition programs **(Figure 5)**

> "The constant change of doctors is really a burden. I also expect doctors to keep me informed about new developments […]. I have to gather all the information myself, sometimes from English-speaking countries. You don't feel cared for, but rather rushed through, so I regularly ask myself what the point of the appointment was. I have also often mentioned that I have problems with motivation. I have NEVER received any (psychological) help in this regard. Of course, I know that it would be better for my health to stick to the diet. But I just don't FEEL it enough to take on all the hardships in my already stressful everyday life and constantly have to make sacrifices (enjoyment, taste, finances...)."

> "It's really striking how the approaches of different doctors are. […] I was even asked why I wanted to go on the diet at all as an adult, because it doesn't work and is just exhausting, and I wouldn't be able to stick to it anyway. As a result, I was only prescribed amino acids for one month."

**Figure 5. Participant-perceived gaps in PKU clinical care**




## Conclusions

- Even with the improved taste and variability of Phe-free foods, strict dietary adherence remains challenging and often leads to interruptions in diet. This is associated with feelings of guilt and may lead to social isolation from fear of being different and living with a chronic illness
- The lack of specialized metabolic centers for adults with PKU also remains a challenge in Germany (Approximately 10 metabolic units per est. 6,000 adults)
- Future research should focus on a broader population, including individuals with PKU of all ages and their caregivers, and focus on experiences during childhood and adolescence

**References**

**1.** Lange K et al. Mol Genet Metab Rep 2025;43:101201; **2.** van Spronsen FJ et al. Nat Rev Dis Primers 2021;7:36; **3.** Yagundina R et al. Nutrients 2024;16:3119

**Acknowledgements**

Medical writing assistance was provided by Ismar Healthcare

**Disclosures**

ACM received research funding and fees from BioMarin Pharmaceuticals, Jnana Therapeutics, Maze Therapeutics, Otsuka Pharmaceutical, Pluvia Biotech, and PTC Therapeutics.
SS received honoraria for scientific presentations and consulting services from BioMarin, PTC Therapeutics, Sanofi, and Immedica, and received conference attendance support from BioMarin and PTC Therapeutics.
TSH is an employee of DIG-PKU. TSH received speaker honoraria and compensations for consulting services from BioMarin Pharmaceutical International Ltd., Danone Trading Medical BV, Dierks & Company Consulting GmbH, Münster University Hospital, and RPP Deutschland GmbH.
MM and CH are employees of BioMarin Pharmaceutical Inc.

**Abbreviations**

**BMI:** body mass index; **DIG-PKU:** Deutsche Interessengemeinschaft Phenylketonurie; **HCP:** healthcare professional; **Phe:** phenylalanine; **PKU:** phenylketonuria

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Presented at 40th E.S.PKU Conference 2026: 22-25 October, 2026; Copenhagen, Denmark

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