

Living Well with Phenylketonuria

The Irish PKU Community Perspective

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Introduction

Phenylketonuria (PKU) is a rare metabolic disorder that affects an individual's ability to metabolise the amino acid phenylalanine (PHE) due to the absence or limited source of the hepatic enzyme phenylalanine hydroxylase (PAH). If left untreated PKU can lead to devastating neurological consequences (Blau *et al.* 2010). PKU management consists of adherence to a restrictive low protein dietary therapy for life and frequent blood PHE monitoring. Currently there is no cure for PKU.

Ireland has one of the highest incidences of PKU in the world; 1:4,500 compared with 1:10,000 worldwide. Ireland was one of the first countries to introduce new-born screening to identify patients in 1966 (NCIMD, 2020). The lived experience of the Irish PKU community is not well documented. To date there is limited qualitative data on the quality of life of those living with PKU in the Irish context where the most severe mutation of the PKU gene prevails. Dietary therapy is an effective form of treatment as it prevents serious neurological impairments. However, research now acknowledges adverse physical, cognitive, and behavioural consequences of dietary management (Camp *et al.*, 2014). These consequences impact on the quality of life of people living with PKU. Subsequently, other jurisdictions are currently treating patients with a pharmacological therapy, with proven efficacy, that is not currently available to Irish patients due to resource issues despite drug approval from the National Centre for Pharmacoeconomics.



International PKU Day, Dublin, Ireland. Photo permission granted from the PKU Association of Ireland.

Aims and Objectives

The aim of this study was to identify PKU research priorities from patients and families living with PKU.

- Create a visual representation of their interpretation of living well with PKU.
- List the barriers and facilitators to living well with PKU within the Irish context.
- Recommend research themes to explore the aforementioned barriers and facilitators to enhance the shared experience of those living with PKU.

Participants

Individuals living with PKU in Ireland, which consisted of adults with PKU, their partners, Mother/Father of child or adult with PKU.

Method

A participatory engagement strategy was utilised.

Phase I is called “**Miracle**” the question which was used in solution-focused therapy to encourage participants to focus on possibilities rather than problems. A single open-ended question will be used to capture patients/families' ideas to improve their care: “*If you had a magic wand, what would you change in your healthcare?*”

Phase II is called “**Reflection**” to identify common themes or certain challenges specific to people living with PKU in Ireland.

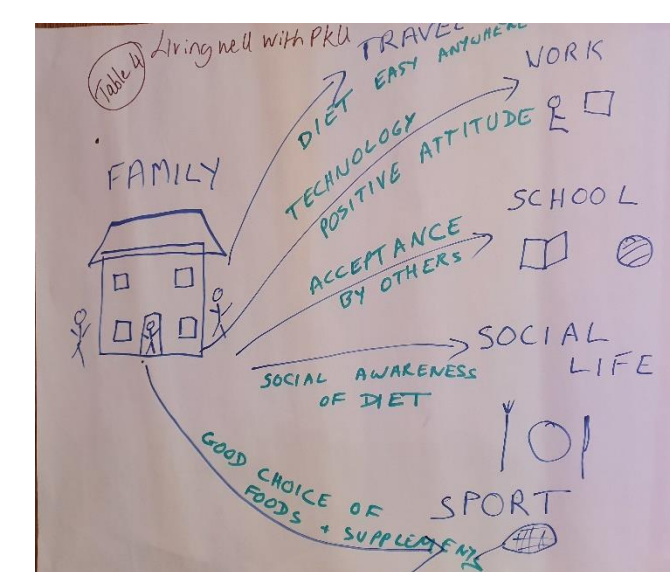
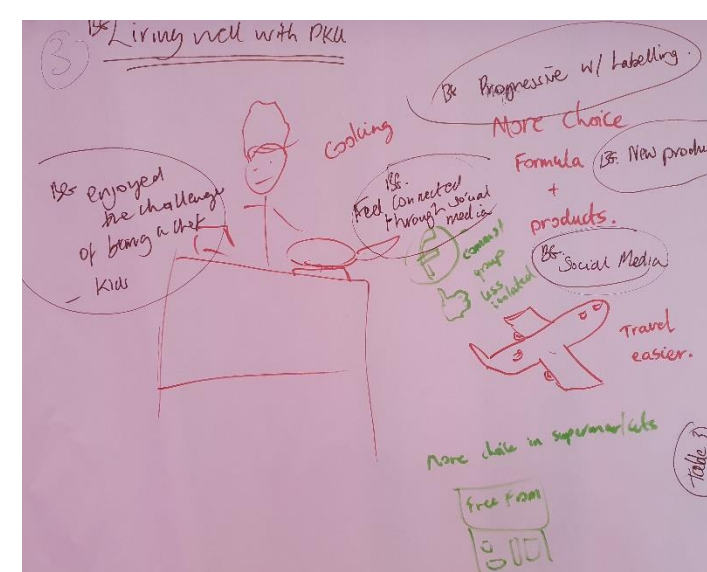
Ethics

The study has been approved by the Research and Ethics Committee at the University College Dublin.

Findings

Participants (n=21, 9 males/12 females) were recruited by the PKU Association of Ireland via email and various social media platforms to partake in a PKU Research Prioritisation workshop. Adults with PKU (>18yr), their partners, parents of children/adults with PKU identified facilitators and barriers to living well with PKU. Findings were grouped under the overarching themes of *facilitators to living well with PKU* and *barriers to living well with PKU*. Participants also generated a number of ideas for improvements in their experience of PKU when asked the open-ended question “*If you had a magic wand, what would you change about your experience of PKU?*”

Sample Participatory workshop output



Facilitators of Living Well with PKU

Patient and Family

- Being optimistic for the future of PKU
- Seeking support from other patients, parents/carers
- Feeling less isolated by connecting with the PKU community through PKUAI, social media and other platforms
- Increased understanding and support from immediate and extended family
- A positive attitude in the workplace
- Acceptance from others in a school environment
- Awareness of dietary therapy in a social environment

Healthcare

- Adequate service provision
- Positive engagement with healthcare staff particularly for getting back on diet
- Advancements in the quality of medical products
- Good choice of medical food and synthetic protein supplements for participation in physical activity
- Psychological support when available
- Cooking support as part of paediatric care

Medical food company

- Applications and Technology
- Cooking demonstrations
- Website recipes
- Nutritional management booklets

Barriers of Living Well with PKU

Patient and Family

- Feeling a burden to others
- Daily planning and organisation
- Difficulty explaining PKU
- Difficulty accessing information on medical developments
- Lack of clinical trial opportunities
- Social engagement between adults with PKU being discouraged

Healthcare

- “Blood results too late, too slow” (adult care)
- Lack of satiety with dietary therapy
- Lack of access to newly approved medication
- More lived experience informed research
- Use of inappropriate language “*I am her father, not her carer, don't take that away from me*”
- Inconsistencies in dietary management practice
- Lack of awareness of PKU and the importance of dietary therapy amongst pharmacy staff and peripheral hospitals
- Dublin only based clinics
- Frayed relationships with clinic staff “*Dismissive parental attitude*”
- Access to psychological support (adult care)
- Physiological support

Food Industry

- Inaccurate protein content labelling.
- Lack of awareness on the importance of dietary therapy when eating out.

Conclusion

In this study, we engaged with people living with PKU in Ireland through a participatory workshop to create a visual representation of their interpretation of living well with PKU. A common theme to emerge from the findings was the detrimental impact that lack of awareness of PKU and dietary therapy has on this community particularly from the food industry, pharmacies, peripheral hospitals, in education and workplace settings. Further research on the perspective of Irish PKU healthcare service providers should also be explored. Despite early intervention and considerable advances in PKU care and management practices in Ireland, there are many issues that continue to warrant attention to support the Irish PKU community.

References available upon request

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