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 newborn 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 for 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.

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 of 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

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
Acknowledgment: The authors are incredibly thankful to individuals who generously shared their experience with us. Also, UCD PPI Ignite for supporting this project via seed funding.