

Assessing the Digital Care Pathways Unmet Needs for Rare Brain Diseases. The Case of Phenylketonuria

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Background

Digital Care Pathways for Rare Brain Diseases research project (2023-2024) is coordinated by the European Brain Council. The study is looking at the unmet needs while using digital care pathways and aiming to assess the benefits of health digital tools from the patient' perspective based on the COVID-19 pandemic experience, presenting the phenylketonuria (PKU) case study. PKU is a rare neurometabolic disorder. In Europe, PKU prevalence is about 10:100,000 newborns. The Covid-19 pandemic accelerated the healthcare sector's digital transformation agenda. The delivery of telemedicine services instead of many face-to-face procedures has been expanded and many healthcare services shifted online remotely. The study is looking at the unmet needs of patients related to information, education and communication in the management and monitoring of Phenylketonuria (PKU) along the digital care pathways.

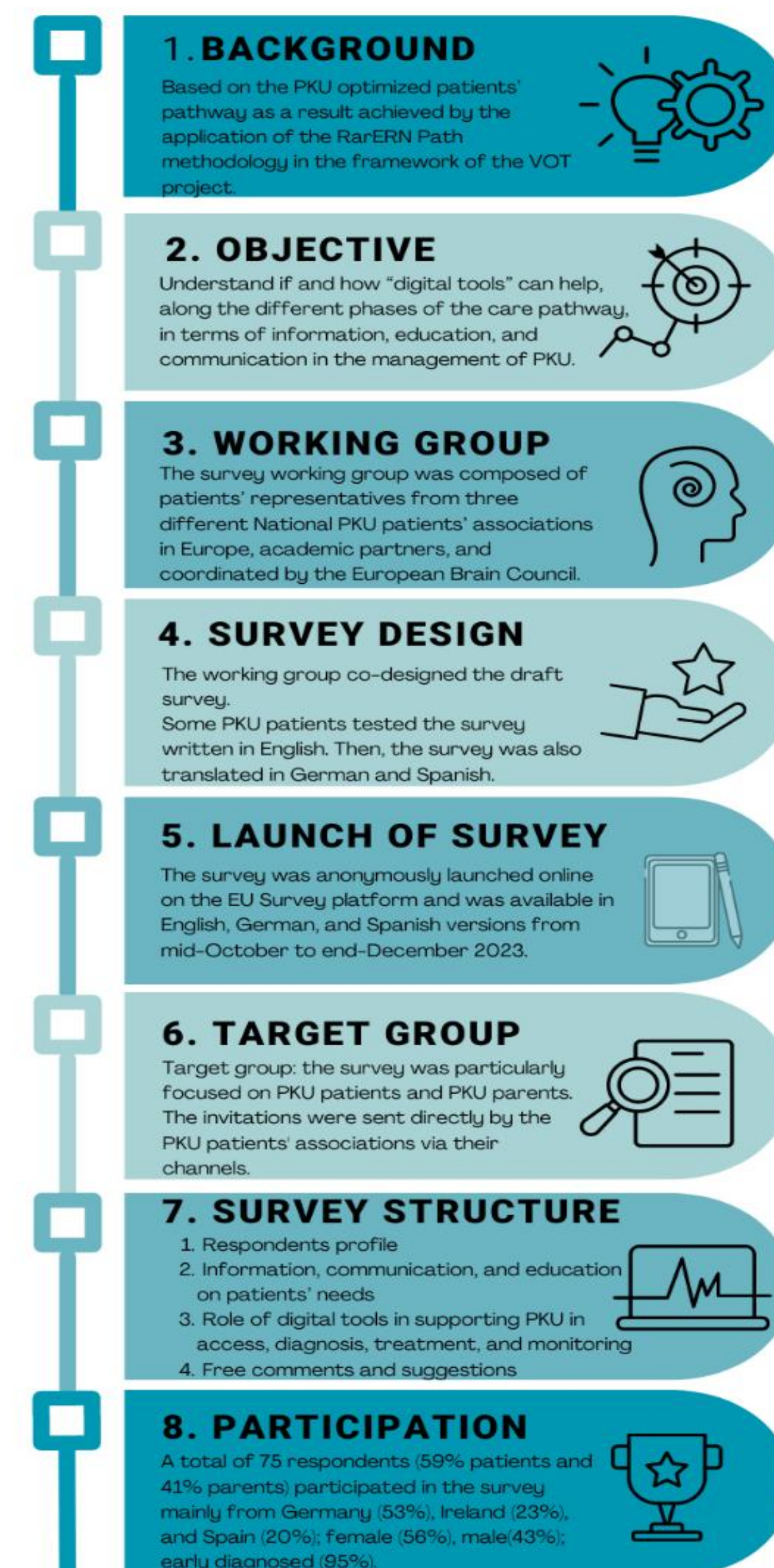
Methods

A survey (see fig. 1) was co-designed with PKU patients' representatives, translated in German and Spanish, and anonymously launched in the EU Survey platform. The survey contained questions on demographic profile of respondents, 13 questions about patient unmet needs on information, communication and education, 8 questions regarding the patients experience on access to the care pathways during the COVID-19, and 13 questions on the role of digital tools in supporting PKU management.

Fig. 1: Patient survey

"WHERE AND HOW CAN DIGITAL TOOLS SUPPORT PKU DIAGNOSIS, TREATMENT AND FOLLOW-UP"?

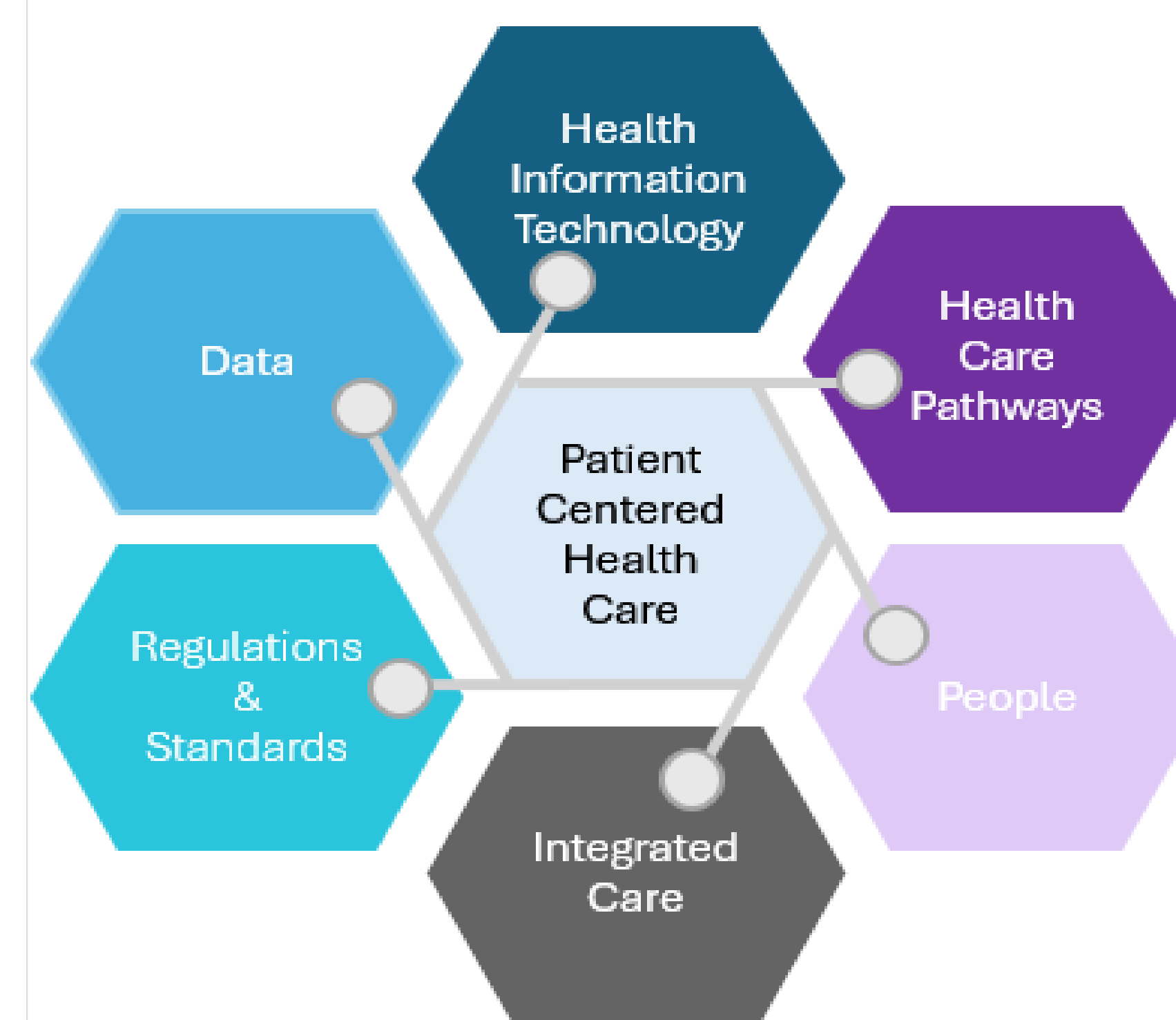
PKU PATIENTS PERSPECTIVE



Results

75 respondents (59% patients, 41% parents) participated to the survey. Information about PKU and communication between patient and healthcare professionals are extremely important (44% and 60%), and education on digital platform for the management of PKU is very important (47%). Digital tools (e-mail, mobile Apps, WhatsApp messaging, virtual calls) are needed for sharing information and for communicating before the clinic visit (75%) and enhance understanding the information provided for treatment or monitoring (90%). On day-to-day PKU management, Apps, wearables, e-mail, telephone call could help for understanding information received from the center and for communication (90%).

Fig. 2: Connected Health



Source: EBC The Value of Education.Digital Care Pathways for Rare Brain Diseases. The case of Phenylketonuria, 2024

Patient's statements

Using the digital tools before the clinic visit in sharing the information and for communicating could help in making the experience of the medical appointment more effective. For a more understandable and effective diagnosis phase, a more extensive/better use of digital tools could enhance understanding of what healthcare professionals tell the patients about PKU and afford the opportunity to ask questions and clarifications.

Digital tools could help in terms of education as a part of the management of PKU. Educational activities and programs tailored to different target users could help the patients, the parents, and the caregivers to be better prepared during the clinical visits.

Education on dietary management could be facilitated by adoption of digital tools.

Conclusion

Overall, treatment management and care delivered to PKU patients demonstrated the benefits and interest for more use of digital tools in the management and follow-up. Results provided valuable insight into understanding the needs of patients with PKU and defining the best channels to engage and communicate with them. While looking at the patient perspective, the clinician perspective is equally important.

Acknowledgements

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