

# NUCDF Partner Network: Building Capacity for the UCD Community to Engage in Patient-Centered Outcomes Research

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## Background

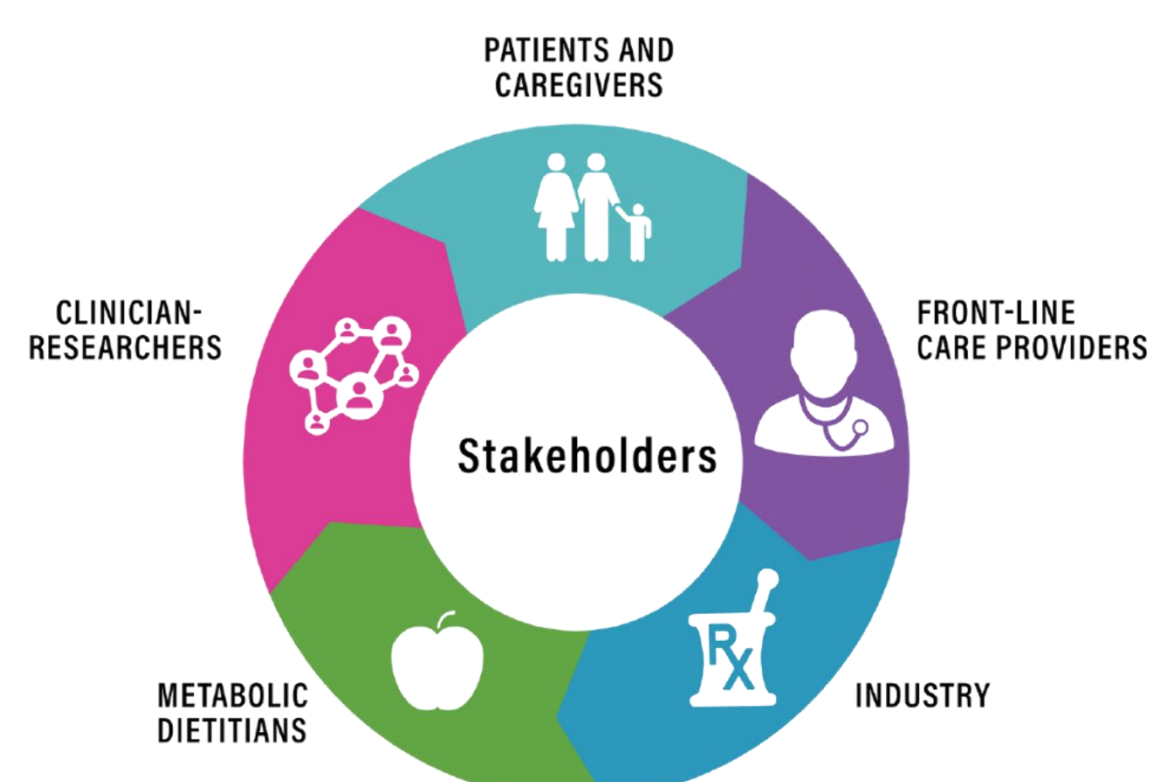
Urea cycle disorders (UCDs) are rare genetic diseases that often result in coma, brain damage and death. Investigator-driven research has improved survival rates and neurological outcomes; new treatments including gene therapies and mRNA medicines are under development. However, there has been no formal assessment of patient needs or meaningful patient engagement in ongoing research.

As a result, significant unmet patient needs remain and data to guide treatment choices is lacking. Our experiences at the National Urea Cycle Disorders Foundation (NUCDF) underscore this gap, as we often field urgent patient questions on topics like liver transplant v. mRNA/gene therapy, how best to transition care to adulthood, and choices of medications.

There is therefore a critical need to engage patients and other stakeholders as research partners and to develop a sustainable infrastructure for patient-centered comparative clinical effectiveness research (CER) that prioritizes the UCD community's needs.

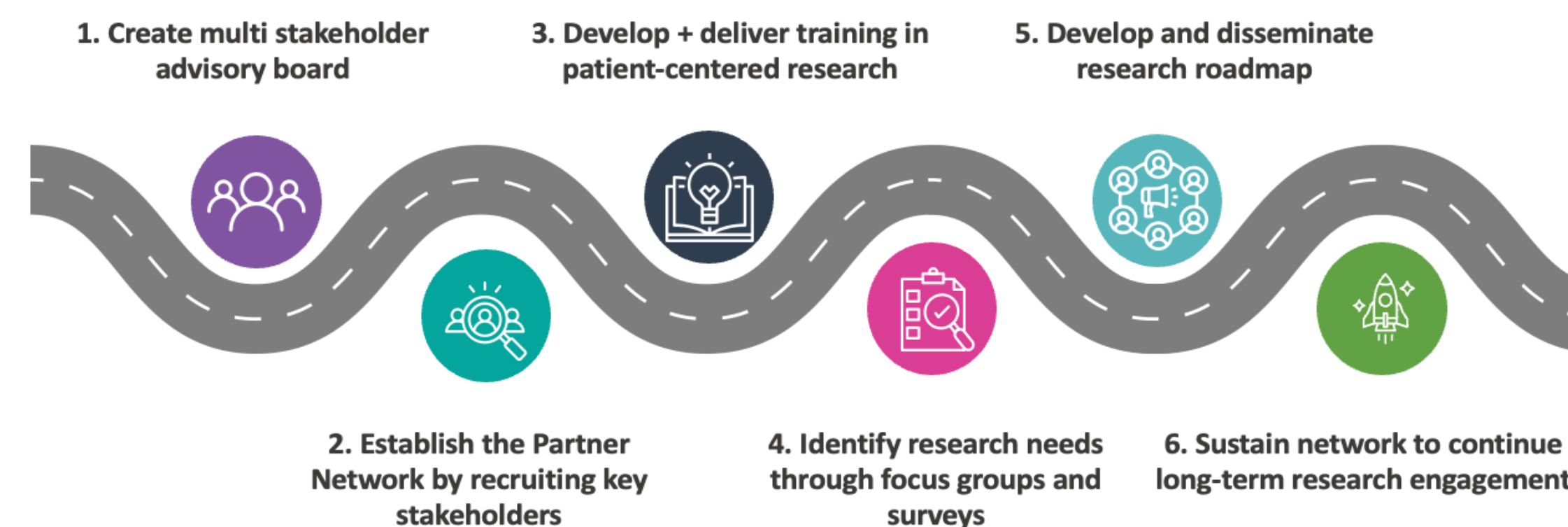
## Objective

Our objectives are to (a) establish the NUCDF Partner Network to build the UCD community's capacity to engage in CER and (b) to develop a roadmap for patient-centered research that can support future CER. The Partner Network will comprise a diverse group of stakeholders including patients and caregivers, clinician-researchers, front-line healthcare providers, metabolic dietitians, and industry representatives.



## Methods

Our two-year, PCORI-funded project consists of five main steps plus sustainability:



We have recently established a 17-member, multi-stakeholder Advisory Board to guide the formation of the Partner Network.

In fall 2025, we will recruit a diverse group (n=100) of patients, caregivers, bereaved parents, front-line healthcare providers, clinicians, researchers, industry, and metabolic dietitians to join the NUCDF Partner Network.



**Patients and caregivers:** People with lived experience of UCDs



**Clinician-researchers:** Experts in both UCD research and clinical care



**Metabolic dietitians:** The front-line experts who manage patients' low-protein diets.



**Front-line providers:** Those who first diagnose UCD patients and often coordinate their care



**Industry:** Lead commercialization of novel UCD therapies and essential partners in research

We are working to develop UCD-specific training materials and we will train the NUCDF Partner Network in patient-centered outcomes research. Next, to identify and prioritize the research needs of the UCD community, we will employ a modified Delphi technique that begins with virtual focus groups to elicit the research needs of different stakeholders. These topics will then be prioritized via a survey distributed to Partner Network members.

Finally, we will develop a roadmap based on these results with input from the Advisory Board. All NUCDF Partner Network members will have an opportunity to provide feedback before it is finalized.

We will disseminate our results via a scientific journal publication as well as in plain language for the broader UCD Community.

## Discussion

By the end of the project period, we will have created the NUCDF Partner Network, a diverse group of stakeholders trained in patient-centered research; developed a patient-prioritized research agenda and stakeholder-informed roadmap to guide our journey toward CER; and shared our findings with the research and patient communities.

From 0-2 years post-project, we will have a sustainable infrastructure for patient-centered CER with NUCDF as its hub. The NUCDF Partner Network will be meaningfully engaged in launching research projects that reflect the priorities of the patient community. From 3+ years post-project, our Partner Network will be increasingly engaged in defining research questions and participating in studies. Ideal long-term outcomes will be research projects that meaningfully address patient questions and improve health outcomes.

To learn more about the project or assess your eligibility to join the NUCDF Partner Network, scan the QR code below.



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