

## Background

Phenylketonuria (PKU) is a rare metabolic condition that disrupts the normal processing of foods containing phenylalanine (PHE). Without treatment from birth, PKU can lead to permanent intellectual impairment. There are several ways to treat PKU including following the PKU diet, using PKU formula, or using PKU medications. While each of these treatments work, there is no evidence about which option works best for which patients.

## Purpose

The purpose of the Inborn Errors of Metabolism Collaborative Stakeholder Network (IBEMCSN) is to build a network of patient partners and stakeholders (providers, public health professionals, scientists) focused on developing patient centered research to inform treatment decisions for PKU. A strong emphasis will be put on building partnerships and fostering co-learning among all members of the network.

## Participation

Members of the IBEMCSN will participate actively in monthly webinars lasting one hour each, and attend two in person meetings (1.5 days each). Webinars will be a combination of education, skill building, and facilitated discussions. Some webinars will be targeted to specific members (**Table 1**); however, all members of the IBEMCSN will be invited and encouraged to attend all webinars.

**Table 1: Webinar Topics and Expected Participants.**

Topic	# of Webinars	Required Participants
Introduction to PCORI and the IBEMCSN	1	patient partners and stakeholders
Self-advocacy	5	patient partners
Working with patient-partners	2	stakeholders
What is CER?	1	patient partners and stakeholders
Introduction to research	3	patient partners, optional for stakeholders
Making sense of statistics	1	patient partners, optional for stakeholders
Introduction to patient-reported outcomes (PROM)	1	patient partners
Selecting or developing the right patient-reported outcome	2	patient partners and stakeholders
Discussion of how to best recruit patients for IBEMCSN-proposed CER studies	1	patient partners and stakeholders
Preparation for second annual summit – discussion of ongoing roles for IBEMCSN members, preparation for CER discussions, preparation for identifying additional stakeholders, and preparation for dissemination of results	2	patient partners and stakeholders

## Roles and Responsibilities

**Project Lead:** An individual who oversees the day-to-day business of the IBEMCSN.

### Responsibilities

- Participate in all decision making for the IBEMCSN
- Assure compliance with all administrative requirements
- Assure project costs stay within budget
- Provide oversight and facilitate conflict resolution

## Roles and Responsibilities (continued)

**Lead Patient Partner:** An individual who represents the voice of patients on the leadership team.

### Responsibilities

- Participate in all decision making for the IBEMCSN
- Provide mentorship and advocacy training to patient partners
- Assure that education, training, and facilitated discussions are inclusive and patient friendly
- Participate in the development of grant proposals for patient centered outcomes research

**Lead Clinician:** An individual who represents the voice of clinicians on the leadership team.

### Responsibilities

- Participate in all decision making for the IBEMCSN
- Assure that education, training, and facilitated discussions are clinically relevant and accurate
- Participate in the development of grant proposals for patient centered outcomes research

**Lead Researcher:** An individual who is motivated to conduct patient centered outcomes research

### Responsibilities

- Participate in all decision making for the IBEMCSN
- Assure that education, training, and facilitated discussions are scientifically relevant and accurate
- Coordinate the development of grant proposals for patient centered outcomes research

**Patient Partner:** An individual with PKU, or an individual who cares for someone with PKU

### Responsibilities

- Actively participate in webinars and in-person meetings
- Help identify gaps in current knowledge regarding the management of PKU
- Identify outcomes important to patients and caregivers
- Suggest and help develop ideas for patient centered outcomes research studies.
- Participate in the development of grant proposals for patient centered outcomes research

**PKU Provider Stakeholder:** An individual who provides care for persons with PKU such as a physician or a nutritionist

### Responsibilities

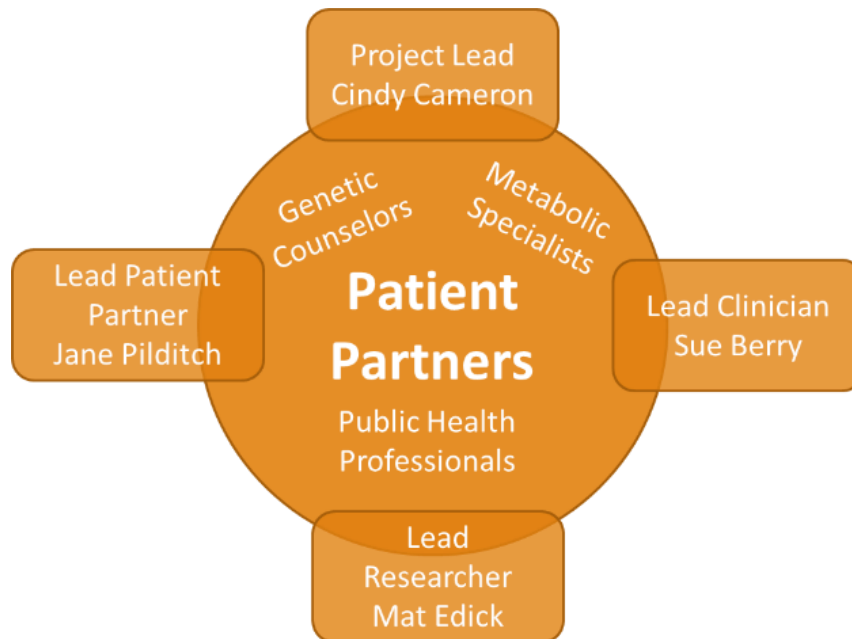
- Actively participate in webinars and in-person meeting
- Help identify gaps in current knowledge concerning management of PKU
- Identify important clinical outcomes
- Participate in the development of grant proposals for patient centered outcomes research

**Public Health Stakeholder:** An individual who works in public health and has an interest in improving outcomes for PKU

### Responsibilities

- Actively participate in webinars and in-person meeting
- Help identify gaps in current knowledge concerning management of PKU
- Identify important public health outcomes
- Participate in the development of grant proposals for patient centered outcomes research

## Organizational Structure



**Organizational structure of the IBEMCSN.** Patient partners and other stakeholders reside at the center of the IBEMCSN. All work accomplished by the IBEMCSN will be guided by patient partners and other stakeholders. The leadership team provides critical direction and fosters co-learning among patient partners and other stakeholders.

## Decision Making

Decisions about administrative activities, such as scheduling webinars and meetings, will be made by project leadership with the input of patient partner and other stakeholder members. Every effort will be made to accommodate the schedules of all partners. Opportunities to listen to webinars and confer with project staff will be made available to IBEMCSN members unable to attend learning opportunities at the scheduled times.

Decisions surrounding all other aspects of the project including recommendations of research topics, study designs, important outcomes, and dissemination plans will be made in collaboration with all partners.

## Changes in Governance

The IBEMCSN will operate as a living entity. As the group matures, it is possible that modifications to the governance document will be required. At any time during the project, any member of the IBEMCSN may point out a need or desire for a change in the governance document. At such time, the leadership team will arrange a group discussion around the proposed changes and whether or not they should be adopted.