

# A Whirlwind Tour of the Wild, Wacky, Wonderful World of Patient Partnered Research



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INFORM RARE

Co-lead Patient Engagement

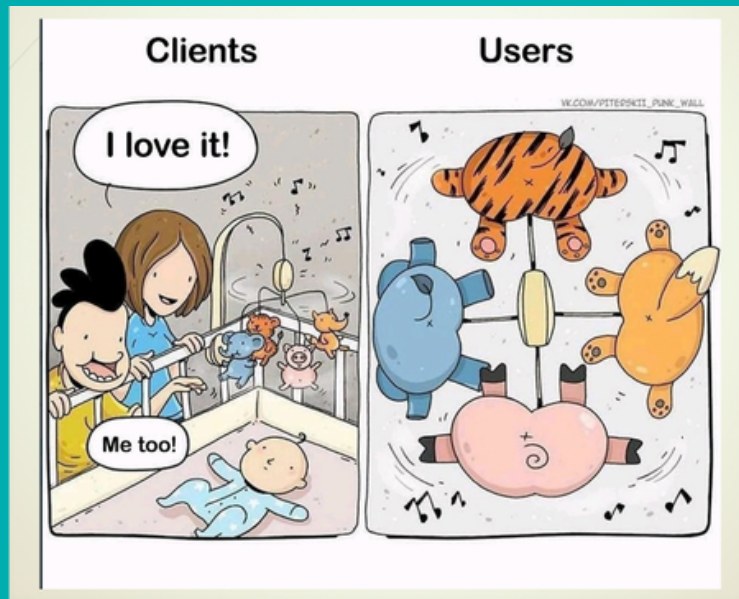
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10 minutes to highlight some of the amazing and groundbreaking work that is the result of patient and patient group engagement in research

I'll present a few examples from the work conducted by the INFORM RARE Research Network, one of many research teams who partner with patients, including youth, and caregivers to co-design research that results in outcomes that matter to patients and caregivers

# Patients' perspectives



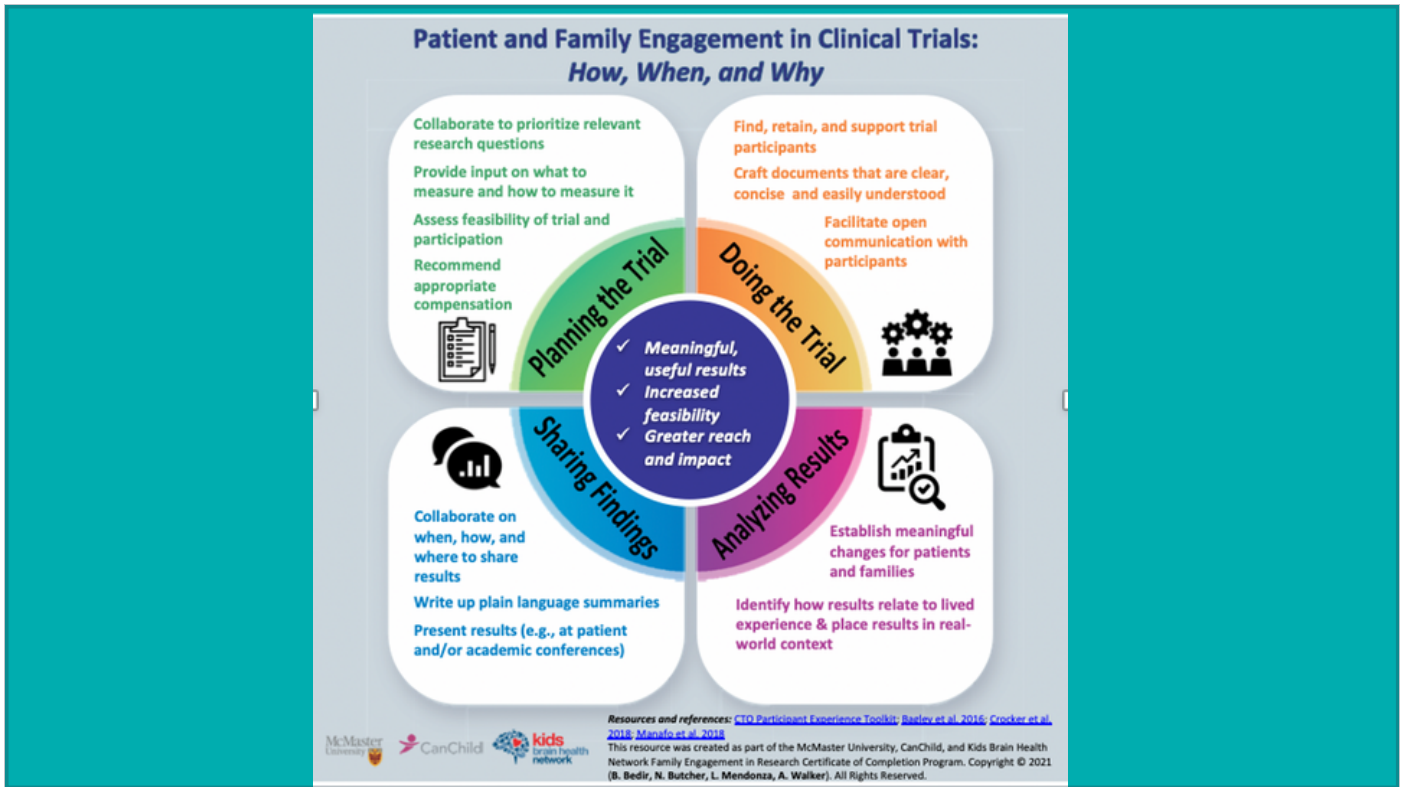
If you are looking for a reason to engage patients, this says it all.

## What is patient partnered research?

- CIHR definition: “patient partner” describes when patients contribute to the research process and research-related activities, different from the traditional, more passive role, as research participant
- Involved in conducting research activities, **at all stages** of the research process, including supporting grant applications, assisting with participant recruitment and performing research dissemination activities



Just to make sure that we are all on the same page, I'm referring to patient partnered research. In Canada, you'll often hear the term "patient partner"



And here are the stages in clinical trials where patient engagement should occur

# INFORM RARE Research Network

- A pan-Canadian pediatric rare disease network that aims to support decision making about rare disease therapies for children by:

## 1. Conducting registry-based randomized trials for three “exemplar” diseases:

- Mucopolysaccharidosis: effect of a biological therapy on bodily pain
- Phenylketonuria: comparing medical formulas’ impact on metabolic control and acceptability
- Spinal muscular atrophy: effect of home-based exergame on motor function

## 2. Developing capacity in patient-oriented research and innovative clinical trials for pediatric rare diseases.

[www.informrare.ca](http://www.informrare.ca)



Administered and supported by:



I'll tell you very briefly about INFORM RARE and how we embed patient partnered research

# Why co-develop core outcome sets?

Which outcomes are being measured?

When each research team decides which outcomes to measure in their study...

**Pain**



**Mobility**



**Enzyme  
activity**



**Cognitive  
development**



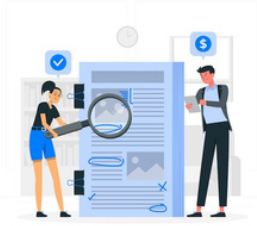
... differences in outcome measures make it difficult to compare and combine the overall evidence to inform policy and practice

To make best use of resources, we need research that includes high quality study designs - especially clinical trials

And this research needs to address questions that matter to patients, caregivers, and healthcare professionals who make treatment decisions

Researchers measure 'outcomes' in their trials to determine if a treatment is working; these outcomes should be meaningful to patients and to other decision-makers

# Why is this an issue?



**Evidence cannot be combined**, making it hard to make treatment decisions



Data collection and analysis of many outcomes is **costly**



Outcomes may not align with **patients' priorities**

# Pediatric rare disease core outcome sets



Phenylketonuria (PKU)

Medium Chain Acyl CoA Dehydrogenase  
Deficiency (MCADD)



Mucopolysaccharidosis



Creatine Transporter Deficiency &  
Guanidinoacetate Methyltransferase Deficiency





# Patient Registries



**Designing patient registries for children with rare metabolic diseases**

The Canadian MPS Society, Canadian PKU and Allied Disorders Inc. and the INFORM RARE research network partnered to co-design two new patient registries!

**What is a patient registry?**  
A special database that contains information about people diagnosed with a specific type of disease.  
• The Canadian Mucopolysaccharidoses (MPS) Registry and The Canadian Phenylketonuria (PKU) Registry were built on the National Organization for Rare Diseases (NORD) iMARE® platform.  
• Registry data will be stored securely on servers located in Canada, and subject to Canadian laws governing access to and protection of personal health data.  
Spinal muscular atrophy already has a registry: the Canadian Neuromuscular Disease Registry.

**What are the patient registries' core values?**

**Who is in charge of the patient registries?**  
The patient registries are governed by a Steering Committee, composed of patients, patient organization representatives, health care providers, methodologists, and ethicists. This governance structure was co-developed by patient organizations and INFORM RARE.

**Who is eligible to participate?**  
Individuals aged 18 years and younger, with a diagnosis of MPS or PKU, and receiving disease-specific care in Canada. We participate expanding the registry to include adults in the future.

**Launch date: Winter 2024**  
To learn more about the patient registries, visit:  
• [mpsregistry.ca](https://mpsregistry.ca)  
• [pkuregistry.ca](https://pkuregistry.ca)

**Why are patient registries important?**  
For rare diseases, high-quality patient registries that collect meaningful patient-reported longitudinal data\* have strong potential to be valuable to:  
• Enable patient organizations and their partners to promote and support patient-oriented research  
• Further our understanding of the natural history of a disease, thereby addressing a common gap in evidence for rare diseases  
• Evaluate intervention effectiveness (e.g., through registry-based randomized trials)  
• Use as a clinical contact database for academic- and industry-sponsored research, which is particularly important for small and geographically dispersed patient populations  
\*Longitudinal data: tracking the same type of information on the same people at multiple points in time.

**Why is youth and family engagement essential?**  
Your input will enable us to co-develop a registry that responds to the needs of youth and their families and is easy to navigate. Some things we may seek your advice and feedback on:  
• Is the process to enroll in the registry clear?  
• Are the consent and assent processes clear? Do they answer your questions?  
• The registry will collect patient-reported data. We'd like your feedback on several aspects of the surveys that are used for this purpose.  
• How do you feel about incentives (such as prize draws) for participation?

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Many patient registries being co-developed with patient groups and patients. These two registries are the result of an incredible collaboration with two patient groups: the Canadian PKU and Allied Disorders and the Canadian MPS Society.

And we now have a PhD candidate, Catherine Stratton, here with us in Calgary, who is devoting her PhD thesis to co-designing a framework for rare disease patient registries

# New Frontiers

**Youth Engagement**

**More focus /better understanding of Equity, Diversity & Inclusion and Indigenous Ways of Knowing**

**Co-Development of Core Outcome Sets**

**Patient Engagement in Reporting Guidelines**

**Governance of Research Networks**

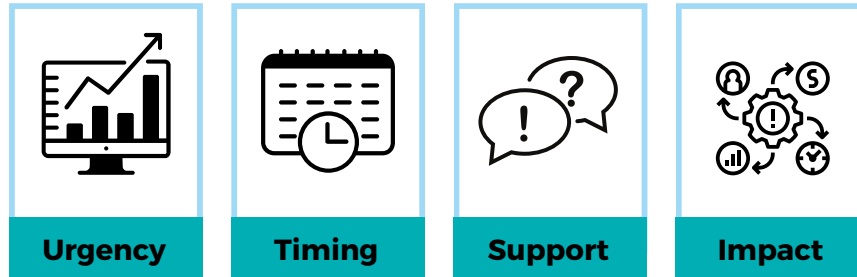


To sum it all up, I've barely touched on the new frontiers in patient partnered research.

# Key Opportunities for patient engagement in research



A Favourable Landscape



**Urgency:** Always the case in rare diseases as people wait for access to therapies and the work needed to make this a reality from coast to coast to coast

**Timing:** Opportunities to be engaged in new research networks, in funding promises, in the direction of future research

**Support:** Amazing pan-Canadian and international community who have always worked together

**Impact:** many efforts to evaluate the impacts of patient engagement in research - listening to both the patient partners and the researchers to create the most impactful partnerships



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**Youth Advisors**

**Alison Howie - Research Associate**



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