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

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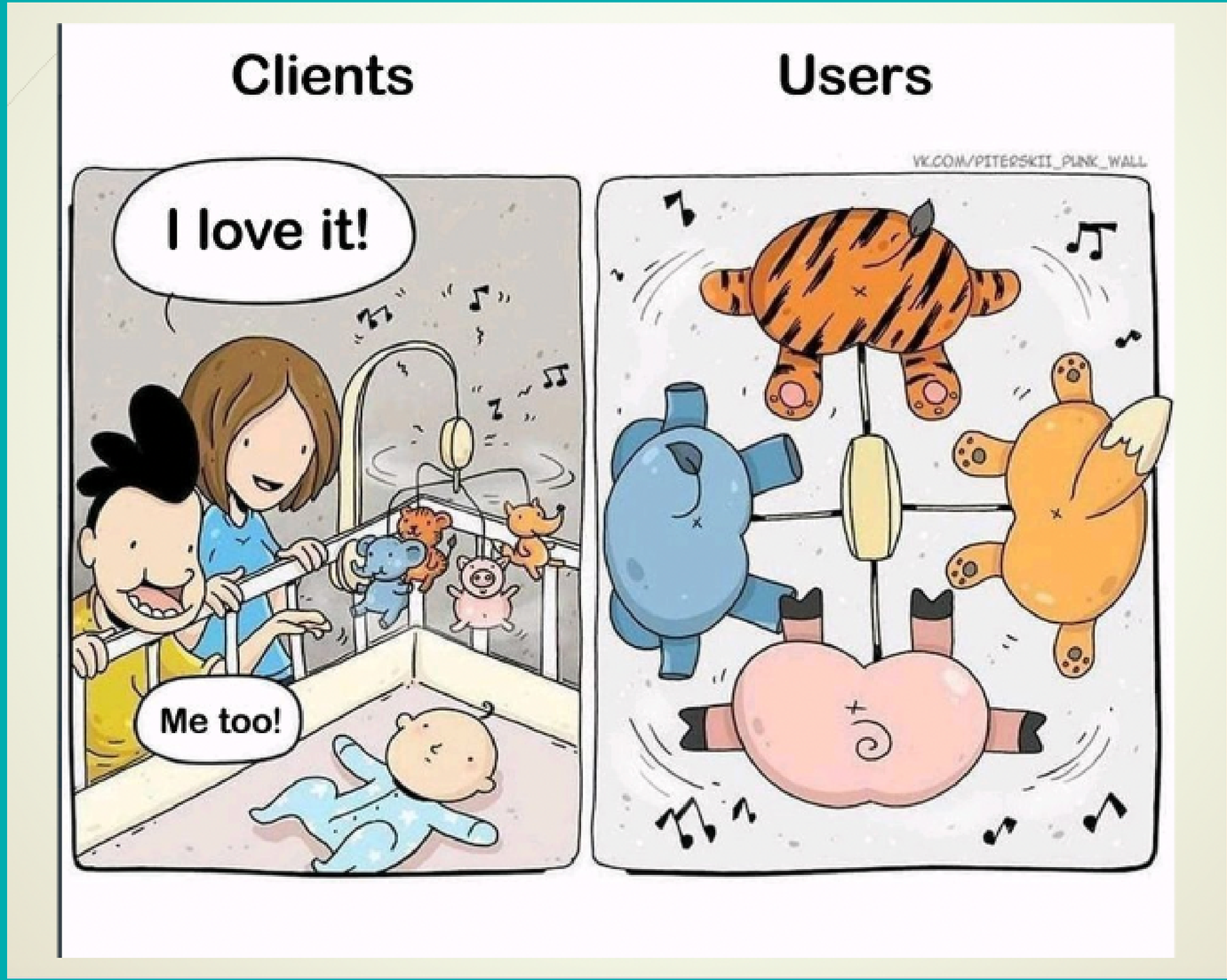
INFORM RARE

Co-lead Patient Engagement

CORD Conference

November 29, 2023

Patients' perspectives



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

Patient and Family Engagement in Clinical Trials: *How, When, and Why*

Resources and references: CTO Participant Experience Toolkit; Bagley et al. 2016; Crocker et al. 2018; Manafo et al. 2018

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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

Funded by: CIHR IRSC Canadian Institutes of Health Research / Instituts de recherche en santé du Canada; SPOR SRAP Putting Patients First / Le patient d'abord

Administered and supported by: INFORM RARE; uOttawa

Why co-develop core outcome sets?

Which outcomes are being measured?

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

Pain

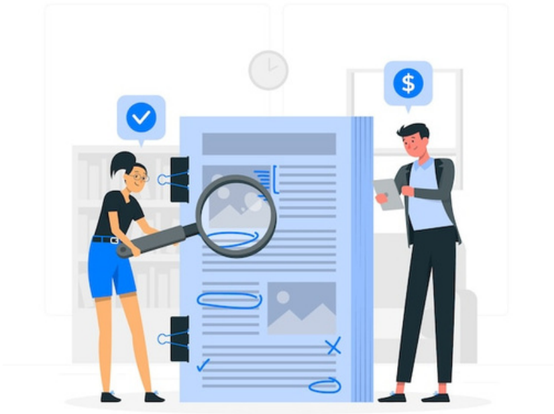
Mobility

Enzyme activity

Cognitive development

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

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**

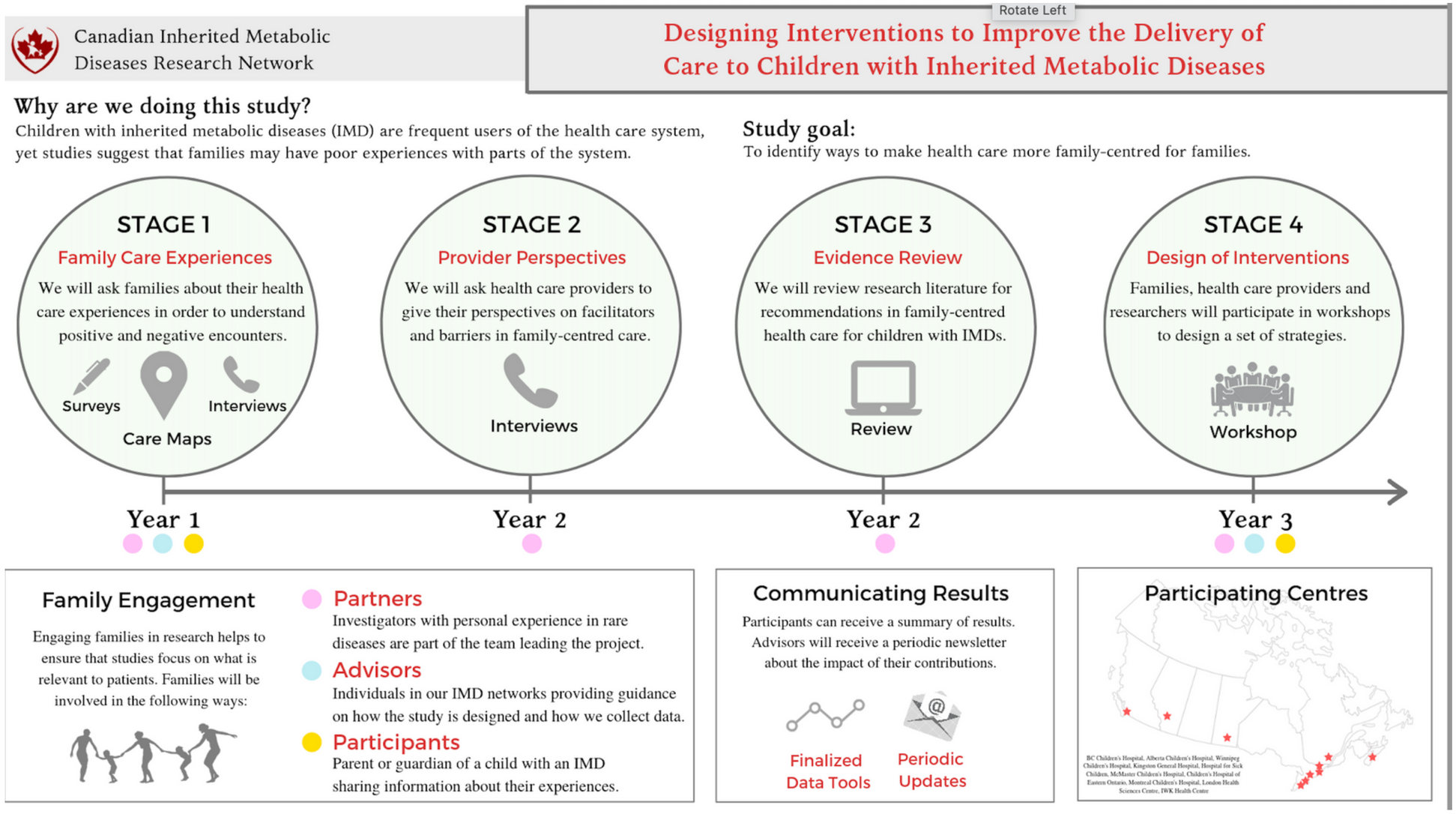
Pediatric rare disease core outcome sets

Phenylketonuria (PKU)

Medium Chain Acyl CoA Dehydrogenase Deficiency (MCADD)

Mucopolysaccharidosis

Creatine Transporter Deficiency &
Guanidinoacetate Methyltransferase Deficiency



Improving health care delivery for children diagnosed with rare metabolic diseases by learning from families and providers: Protocol for Phase I, a prospective, mixed methods cohort study of families' health care experiences

Andrea J. Chow, Pranesh Chakraborty, Isabel Jordan, Nicole Pallone, Maureen Smith, Alvi Rahman, Laure Tessier, Jamie Brehaut, Eyal Cohen, Sarah Dyack, Jane Gillis, Cheryl R. Greenberg, Jeremy Grimshaw, Robin Hayeems, Ann Jolly, Sara Khangura, Jennifer MacKenzie, Nathalie Major, John Mitchell, Stuart Nicholls, Andreas Schulze, Rebecca Sparkes, Kathy Speechley, Sylvia Stockler, Mari Teitelbaum, Yannis Trakadis, Clara Van Karnebeek, Jagdeep Walia, Brenda Wilson, Kumanan Wilson, Beth K. Potter

Patient Registries

Designing patient registries for children with rare metabolic diseases

The Canadian MPS Society, Canadian PKU and Alllied 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) IAMRARE® 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.

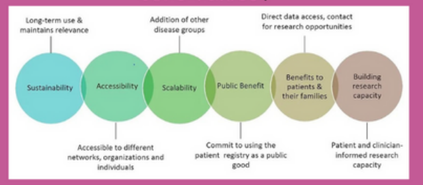
Why are patient registries important?

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.

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 anticipate expanding the registry to include adults in the future.

Launch date: Winter 2024

To learn more about the patient registries, visit:

- mpsregistry.ca
- pkuregistry.ca

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?

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

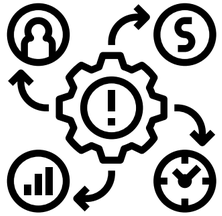
Key Opportunities for patient engagment in research

A Favourable Landscape

Urgency

Timing

Support



Impact

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