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# CIHR Rare Disease Research Initiative Update

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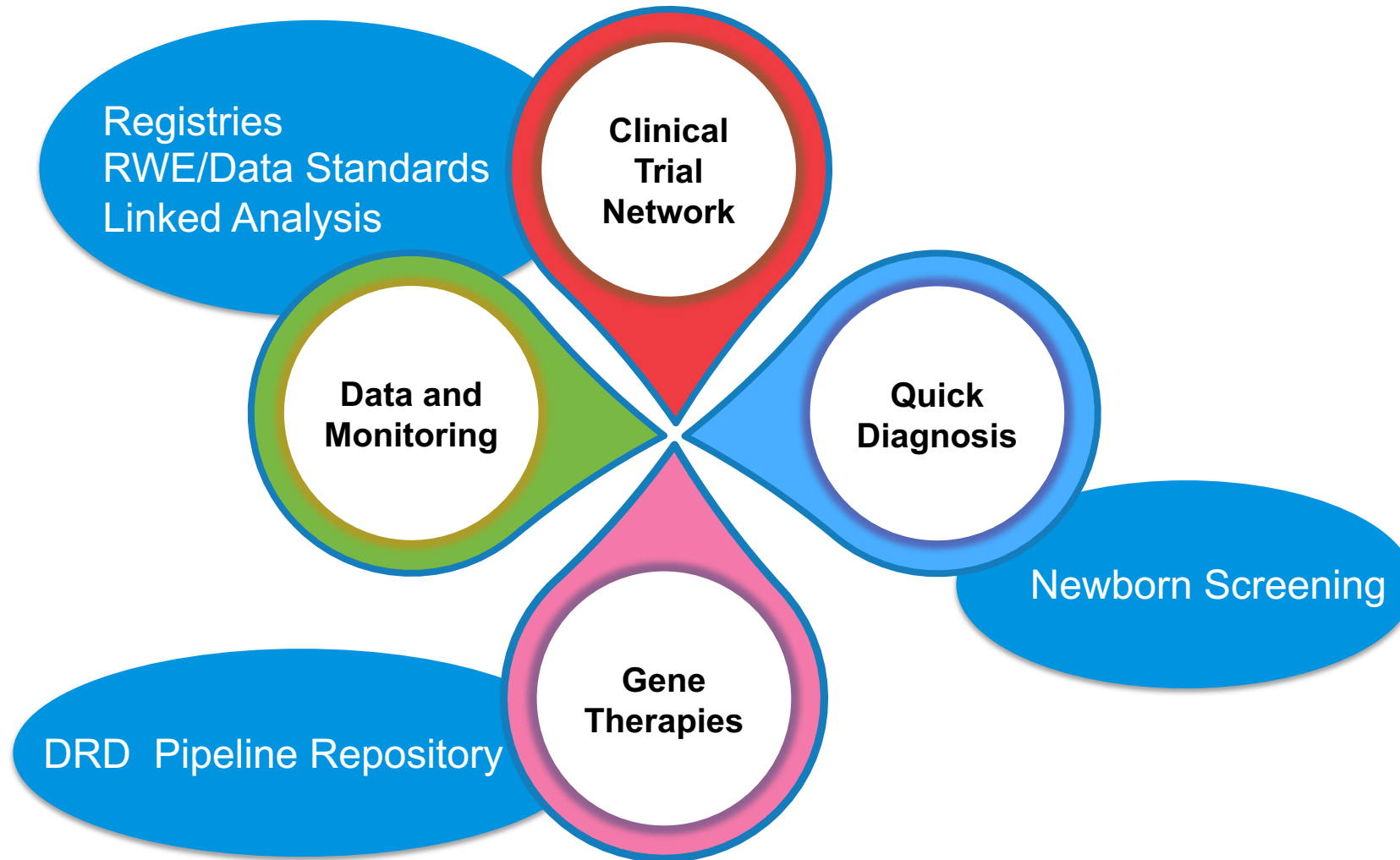
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# CIHR Rare Disease Research Initiative

Part of the National Strategy for Drugs for Rare Diseases



# Improving Diagnosis for Rare Disease Patients

## Purpose

- To determine the **best pathway to diagnose RDs** by moving genomic testing towards the front end of the care pathway.

## Specific Objectives

- Generate **evidence to optimize the RD diagnosis pathway**;
- Evaluate **health outcomes** associated with RDs via the ‘genomics first’ diagnosis pathway;
- Determine the **economic impact** to the health care system;
- Evaluate the **socioeconomic and psychological impacts** on patients and families; and
- **Mobilize knowledge to facilitate the implementation, scale and spread of best practices** for diagnosing RDs in Canada.

## Intersections

- Canadian Rare Disease Network, All for One, Pan Canadian Genomic Library, Silent Genomes Project

# Improving Health and Administrative Data and Monitoring for Rare Diseases

## Purpose

- To support implementation research to **determine the prevalence, direct cost, and impact of RDs** in the Canadian health care system.

## Specific Objectives

- Demonstrate the feasibility of **tracking rare diseases and conducting prospective studies**;
- Increase cross-jurisdictional **collaboration and data sharing**;
- Prospectively **assess the impact** of mortality, co-morbidities, health system usage, and **total direct cost of rare diseases**; and
- Facilitate the **establishment of patient registries**.

## Intersections

- CADTH and CIHI DRD strategy components, CADTH Real World Evidence activities

# Bringing Rare Disease Gene Therapies to Clinical Trial Readiness

## Purpose

- To **bridge the current gap between model organisms/systems and clinical trials for rare disease drugs** in Canada, ultimately increasing the capacity to bring gene therapies to clinic.

## Specific Objectives

- Increase and **advance the development of gene therapies** for rare disease clinical trials in Canada;
- Generate **evidence for first-in-human clinical trials**, in part by working with Canada's **biomanufacturing capacity** (National Research Council of Canada) and **health technology regulator** (Health Canada); and,
- **Increase capacity** across the Canadian landscape to improve readiness of gene therapies for trials.

## Intersections

- Biomanufacturing Strategy, Regulatory Modernization, Clinical Trials Fund

# Pediatric Rare Disease Clinical Trials and Treatment Network

## Purpose

- To create a Pediatric Rare Disease Clinical Network to **streamline RD clinical research across Canada and to facilitate collaboration** among researchers, people with lived and living experience, health care providers, industry, policy and regulatory bodies.

## Specific Objectives

- Develop a platform to **support pediatric rare disease clinical trials** in Canada;
- **Increase the capacity** to perform rare disease clinical trials in Canada;
- **Attract international clinical trials**, increasing investments in Canada and providing earlier access to innovative therapies;
- Foster **diversity and inclusion** in and improve access to clinical trials;
- Implement processes to **acquire data** from clinical trials, real world registries, health economics evaluation, and post-marketing surveillance; and,
- **Increase the number of new rare disease drug submissions** in Canada.

## Intersections

- Canadian Rare Disease Network, ERDERA, National Mirror Group...

# Patient Partners Involvement In the RDRI



# Patient Partnership at CIHR Institute of Genetics

Patient partnership in research is when researchers work together with people with lived experience of a health condition to do research.

The Institute of Genetics now uses the term "patient partnership" instead of "patient engagement" because we believe this term better reflects the value of patient contributions to research.



More Information on our activities!

- Training and resources
- Community of practice
- Patient partner committee
- Funding
- Featured research







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