RÉSEAU CANADIEN DES MALADIES RARES

CANADIAN RARE DISEASE NETWORK

~ Rare Lives, Shared Strength ~

Francois Bernier
On behalf of the CRDN Steering Committee

one child every child | UNIVERSITY OF CALGARY

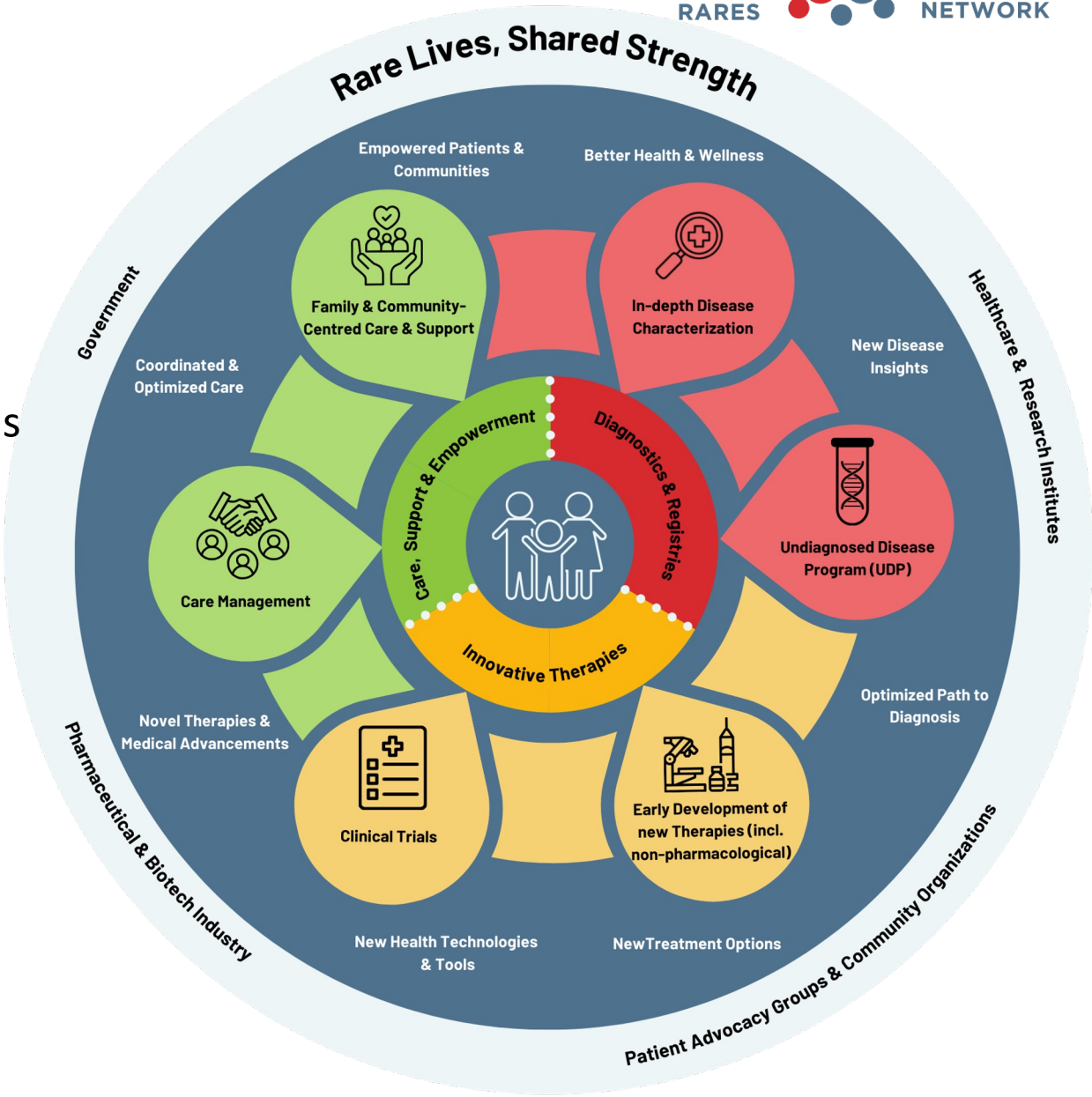
About CRDN

Our Vision: Innovative care and research in Canada so that all patients and families affected by a RD are empowered to live their full potential.

Our Mission: Establish a growing network that builds connections across geographies and disease boundaries to enable timely diagnosis, screening and access to treatment, and facilitate best care, support and empowerment for patients and their families in Canada, ultimately enhancing their quality of life.

Pillars of Our Work:

- Diagnostics & Registries
- Innovative Therapies
- Care, Support & Empowerment
- National & International Collaboration



Our Steering Committee Members

Francois Bernier
Alberta Children's Hospital,
University of Calgary

Durhane Wong-Rieger,
Canadian Organization for Rare Disorders (CORD)

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Sick Kids Hospital,
University of Toronto

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Thierry Lacaze-Masmonteil
University of Calgary;
Maternal, Infant, Child and Youth Research Network (MICYRN)

Deborah Marshall
Alberta Children's Hospital Research Institute (ACHRI),
University of Calgary

How We Work – "It takes a village"

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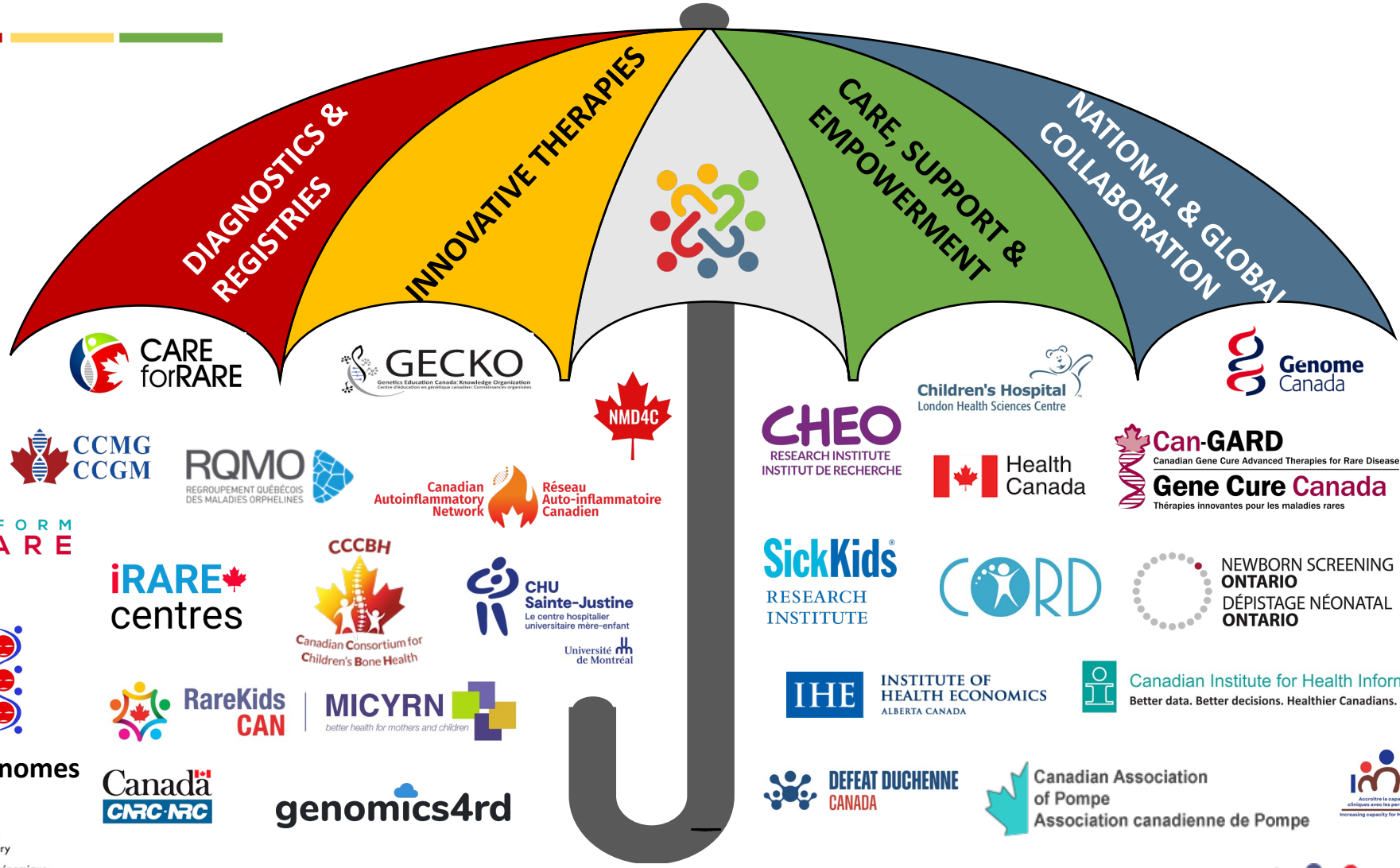


CARE forRARE

GECKO Genetics Education Canada: Knowledge Organization

Alberta Children's Hospital Research Institute

NMD4C

Children's Hospital London Health Sciences Centre

Genome Canada

RARE.Qc

CHEO Research Institute Institut de recherche

Can-GARD Canadian Gene Cure Advanced Therapies for Rare Disease

Gene Cure Canada Thérapies innovantes pour les maladies rares

Health Canada

Rare Disease Foundation

CCMG CCGM

RQMO Regroupement québécois des maladies orphelines

Canadian Autoinflammatory Network

Réseau Auto-inflammatoire Canadien

RARE DISEASES MODELS and MECHANISMS NETWORK

INFORM RARE

iRARE centres

CCCBH Canadian Consortium for Children's Bone Health

CHU Sainte-Justine Le centre hospitalier universitaire mère-enfant

Université de Montréal

SickKids Research Institute

CORD

NEWBORN SCREENING ONTARIO DÉPISTAGE NÉONATAL ONTARIO

CANPKU+ www.CANPKU.org

CIHR IRSC Canadian Institutes of Health Research Instituts de recherche en santé du Canada Institute of Genetics Institut de génétique

BC Children's Hospital Research Institute

IHE Institute of Health Economics Alberta Canada

Canadian Institute for Health Information Better data. Better decisions. Healthier Canadians.

CanChild

RareKids CAN

MICYRN better health for mothers and children

AllforOne Canada's Precision Health Partnership

Silent Genomes

Canada CNRC-NRC

genomics4rd

DEFEAT DUCHENNE CANADA

Canadian Association of Pompe Association canadienne de Pompe

IMPaCT Accroître la capacité à conduire des essais cliniques avec les personnes enceintes et les enfants Increasing capacity for Maternal and Paediatric Clinical Trials

CDA AMC

Pan-Canadian Genome Library Bibliothèque génomique pancanadienne

MUSCULAI DYSTROPH CANADA DYSTROPHIE MUSCULAIRE CANADA

neuro

CNDR ORG Canadian Neuromuscular Disease Registry

Centre universitaire de santé McGill Institut de recherche

McGill University Health Centre Research Institute

The Ottawa Hospital Research Institute

L'Hôpital d'Ottawa Institut de recherche

D2R DNA TO RNA DE L'ADN À L'ARN

Co-Development of Our Strategic Plan

RÉSEAU CANADIEN DES MALADIES RARES
CANADIAN RARE DISEASE NETWORK

STRATEGIC ENGAGEMENT

May – August 2024

Targeted engagement sessions (n=12) with a select but diverse group **of 34 experts**, resulting in **>900 minutes** of meaningful dialogue

VIRTUAL TOWNHALL

31 October 2024

Overview of draft strategic plan presented to broader community **(>160 attendees)** and launch of community feedback survey

COMMUNITY FEEDBACK

October – November 2024

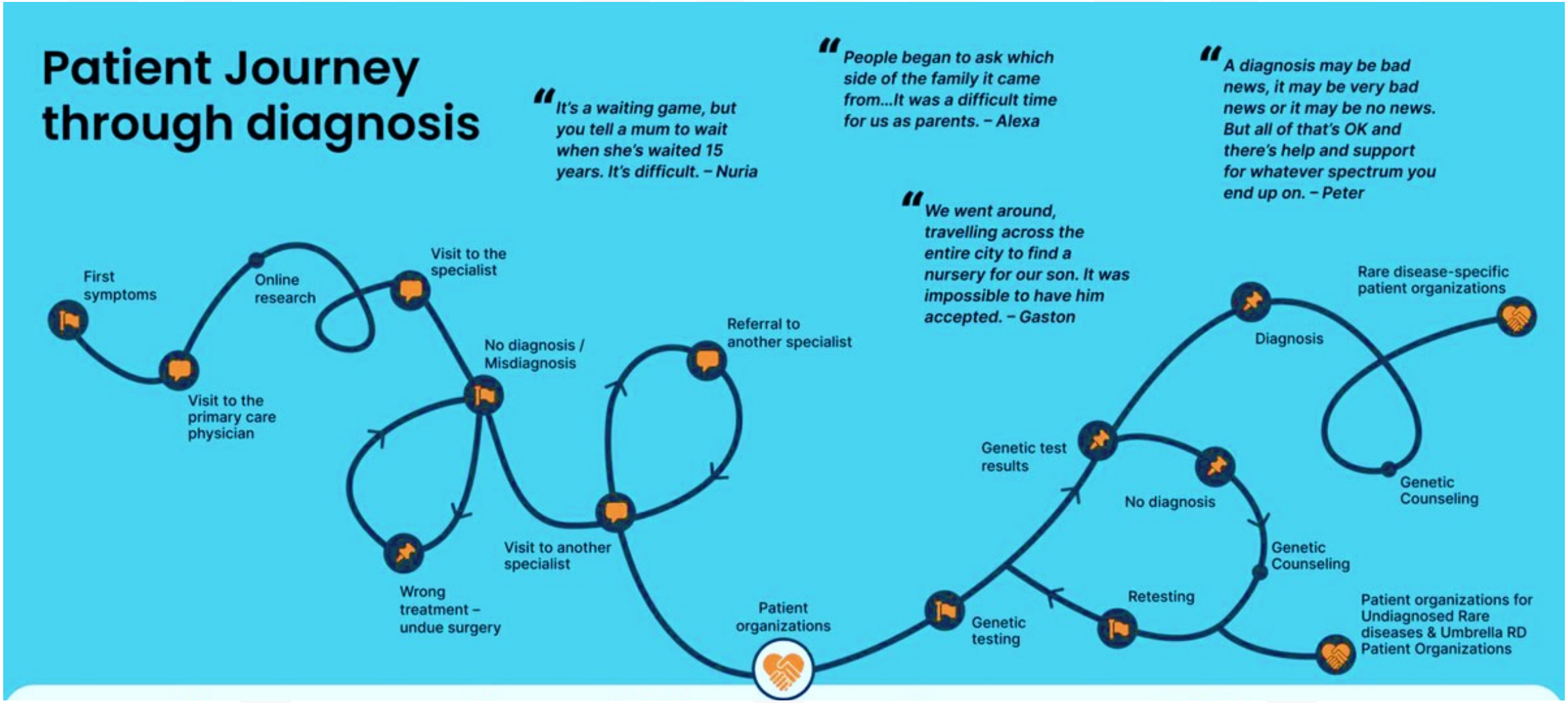
Widely distributed public survey gathered extensive feedback from **115 community members from 10 out of Canada's 13 provinces and territories**

REVIEW & APPROVAL

December – March 2025

Review and approval by CRDN Steering Committee, broad dissemination, and moving into implementation

Pillar 1 – Diagnostics & Registries



Pillar 1 – What We Hope to Achieve

01 Diagnostics & Registries

Goal 1.1: All RD patients will receive the right diagnostic test at the right time regardless of where they live in Canada.

Goal 1.2: Genetic diagnostic laboratories across Canada will integrate resources and best practice guidelines to ensure high-quality GWS for patients.

Goal 1.3: All families with RD will have access to relevant registries for secondary research and re-contact.

Goal 1.4: RD diagnostics and research will be a political priority and sustainably funded.

Goal 1.5: Canada will be a world leader in RD mechanism discovery and translation of new technologies into the clinic.

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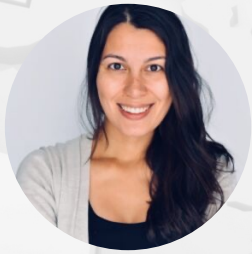
Pillar Lead:

Kim M Boycott
Children's Hospital of Eastern Ontario (CHEO), University of Ottawa

Pillar Members:

Gregory Costain
Sick Kids Hospital, University of Toronto



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BC Children's Hospital, University of British Columbia

Jodi Warman Chardon
Ottawa Hospital, University of Ottawa

Reducing the time it takes to identify rare diseases

Pillar 2 – Innovative Therapies

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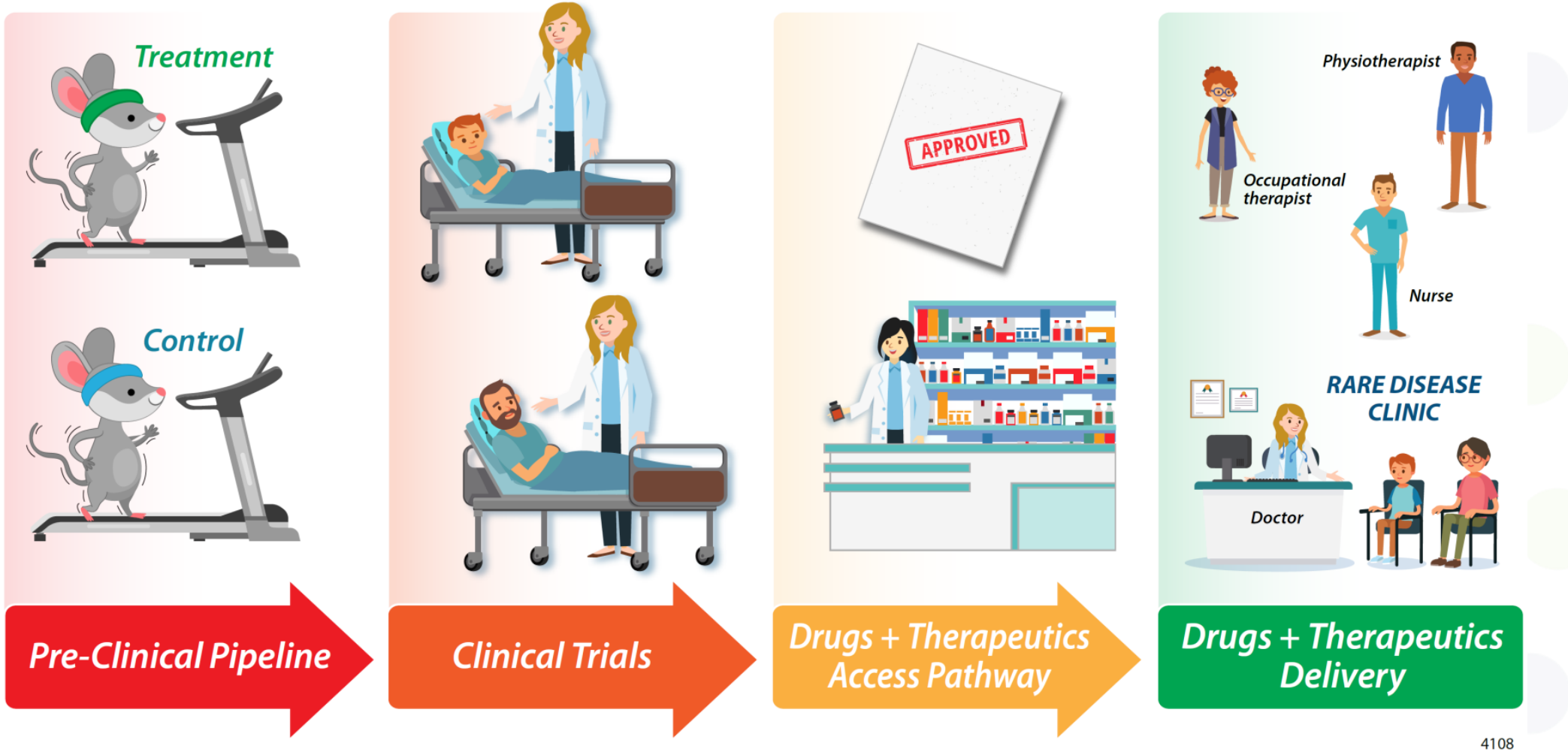
CANADIAN RARE DISEASE NETWORK

Effective, Innovative Therapies



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Pillar 2 – What We Hope to Achieve

02 Innovative Therapies

Goal 2.1: Canada will lead in the discovery and validation of novel therapeutic targets and treatments for RD patients.

Goal 2.2: All RD patients, regardless of their age, location, or social context, will have equitable access to clinical trials and innovative therapies.

Goal 2.3: Innovative therapies will be readily integrated into clinical practice to improve patient care and outcomes.

Goal 2.4: Canada will be recognized globally for its RD clinical trials potential and as an attractive hub for investment and partnerships.

Expanding treatment possibilities

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Pillar Lead:

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Children's Hospital of Eastern Ontario (CHEO), University of Ottawa

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Risini Weeratna
National Research Council (NRC)

Durhane Wong-Rieger
Canadian Organization for Rare Disorders (CORD)

Pillar 3 – Care, Support, & Empowerment

Pillar 3– What We Hope to Achieve

03 Care, Support & Empowerment

Goal 3.1: All RD patients, along with their families and caregivers, will be aware of and have equitable access to the resources and supports they need.

Goal 3.2: All individuals affected by RDs will have the opportunity to be empowered and engaged in meaningful opportunities in research and beyond.

Goal 3.3: All RD patients and their families will receive the mental health and wellbeing support they need regardless of their location or social context.

Goal 3.4: Canada will have a unified RD community that strengthens comprehensive care and support systems for RD patients and their families.

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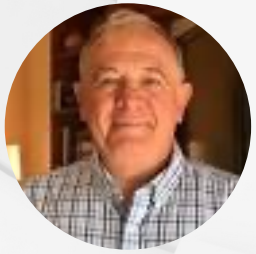
Pillar Lead:

Ian Stedman
York University

Pillar Members:



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Canadian Association of Pompe

Deborah Marshall
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Stephen Parrott
Kidney Cancer Canada Board

Jonathan Pratt
Regroupement Québécois des maladies orphelines (RQMO)

Nicola Worsfold
World Duchenne Organization

Supporting patients and their families

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Goal 3.4: Canada will have a unified RD community that strengthens comprehensive care and support systems for RD patients and their families

04 National & Global Collaboration

Goal 4.1: Canada will have a unified national approach to RD that drives innovation and improves care for all affected by RDs.

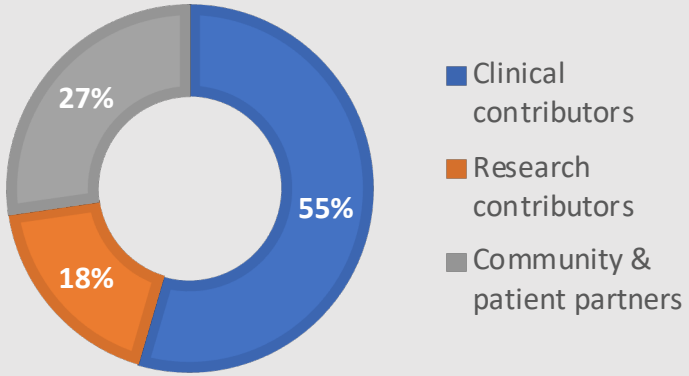
Goal 4.2: Canada will be recognized as a key global player in RD research, innovation, and knowledge exchange, benefiting patients worldwide

Our Growth By Numbers

RÉSEAU CANADIEN DES MALADIES RARES
CANADIAN RARE DISEASE NETWORK

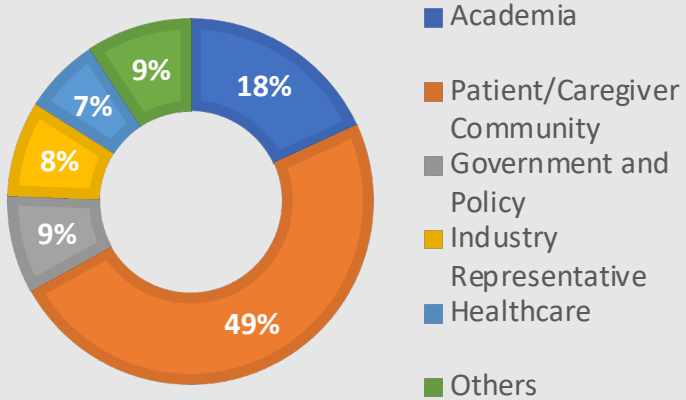
Network Growth

- **1 Steering Committee** and **3 Pillar Committees** set up
- **22 committee meetings** held
- **49 contributors** from **5 provinces** and across sectors



Communication Reach

- **>2,350 Social media followers**
- **>25K website views**
- **>370 newsletter subscribers**



- **Global reach** (e.g., US, Europe, UK, Australia, India) and featured in **OrphaNews**, **IRDiRD**, **HTAi**, **RDI Mapping Rare**

Strategy, Events, & Influence

- **1 strategic plan** co-developed with the community
- **5 Hosted/Co-hosted events**, including 2 in-person workshops, 1 virtual townhall, and 1 hybrid event
- Total of **>640** attendees
- **13 external event presentations**, including 2 international (UK, Doha)
- Participated in **2 policy consultations/roundtables**

Partnerships & Progress Highlights

RÉSEAU CANADIEN DES MALADIES RARES | CANADIAN RARE DISEASE NETWORK

#1 Joining Global Momentum in Rare Disease Research

Co-developing and leading the Canadian National Mirror Group (NMG) of the European Rare Diseases Research Alliance (ERDERA) with RareKids-CAN/MICYRN to:

- Foster national coordination among RD bodies/groups
- Align national efforts with European research priorities
- Serve as gateway to global opportunities

#2 Informing on Canada's Genomic Diagnostics Ecosystem

Collaborative process to identify gaps and opportunities to strengthen Canada's genomic and health data ecosystem.

The outcome is a **report to Health Canada with 14 recommendations** to enhance national coordination and support equitable access to genomics-informed diagnostic care. The report will be shared with P/Ts.

#3 Exploring Pre-Clinical Therapy Development

Co-hosted a short workshop to initiate conversations on how to strengthen national "home-grown" pre-clinical therapy development and lay the groundwork for identifying gaps, opportunities, strategic collaborations and paths forward.

Working towards a "What We Heard" report and actionable next steps.

Highlight- #1

Joining Global Rare Disease Research

CRDN is co-leading Canada's National Mirror Group (NMG) with RareKids-CAN and MICYRN, supporting Canada's engagement in the European Rare Diseases Research Alliance (ERDERA).

What We Are Doing

Bringing together key interest holders, including representatives of the National Strategy for Drugs for Rare Diseases, federal/provincial funding bodies, research networks, academic and healthcare institutions, and patient organizations.

Organizing Canadian NMG meetings to foster knowledge exchange, shape the structure and priorities of the Canadian NMG, and attending ERDERA meetings, workshops, and events to bring best practices and opportunities to Canada.

Why It Matters

Aligns Canada's rare disease strategies with global efforts

Fosters coordination across Canadian rare disease bodies/groups

Opens doors to international research, funding, and training opportunities

Promotes Canadian participation in global initiatives

Highlight – #2

Informing on Canada's Genomic Diagnostic Ecosystem

Health Canada

To support the implementation of Canada's first *National Strategy for Drugs for Rare Diseases*, Health Canada sought expert advice to help improve early, equitable diagnosis.

What We Did

Asset Mapping of existing genomic initiatives, data assets, and infrastructure across Canada

2-Day Genomic Interest Holder Workshop to share insights and co-develop priorities

Targeted Consultations to gather input from additional interest holders and provincial/territorial partners

Total of 41 interest holders consulted

Outcome/ Impact

Report to Health Canada with 3 identified priority areas and 14 actionable recommendations that will be shared with PTs and can guide federal-provincial-territorial collaboration aimed at improving access to rare disease diagnostics.

Priority Areas Identified
1. Timely Access to screening and diagnostic care
2. High-Quality Genetic Testing across jurisdictions
3. Learning Healthcare System powered by genomic data

Recommendations (14 total) grouped under:
- **Infrastructure & IT** (e.g., national health data ecosystem, AI tools)
- **Human Resources** (e.g., training, awareness, capacity building)
- **System Improvements** (e.g., standards, life-course screening, interprovincial care)

Highlight – #3

Exploring Pre-Clinical Therapy Development

Challenges & Opportunities

Fragmented efforts and lack of coordination across institutions and sectors

Limited infrastructure and support

High unmet need for therapies with low commercial interest

Regulatory and operational barriers

Existing assets (e.g., research expertise, GMP facilities, etc.)

Workshop to initiate national conversation on strengthening Canada's pre-clinical therapy development

Path forward?

RareKids CAN | MICYRN better health for mothers and children

Join us!

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SCAN ME

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