RÉSEAU
CANADIEN
DES MALADIES
RARES

CANADIAN
RARE
DISEASE
NETWORK

~ Rare Lives, Shared Strength ~

SCAN ME

Francois Bernier, Leanne Ward, and Ian Stedman
On behalf of the CRDN Steering Committee

one child every child

Why a National Network?

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- **Lack of Comprehensive, National Approach:** Rare Diseases Drug Strategy addressing access to treatment, but not broader spectrum of challenges faced by patients
- **Fragmentation of Efforts**: Efforts at local and national levels but there is fragmentation and lack of coordination

National leadership and coordination needed to catalyze and maximize rare disease (RD) efforts across Canada

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CANADIAN RARE DISEASE NETWORK

Our Vision

Innovative care and research in Canada so that all patients and families affected by a rare disease are empowered to live their full potential.

Our Mission

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

About CRDN

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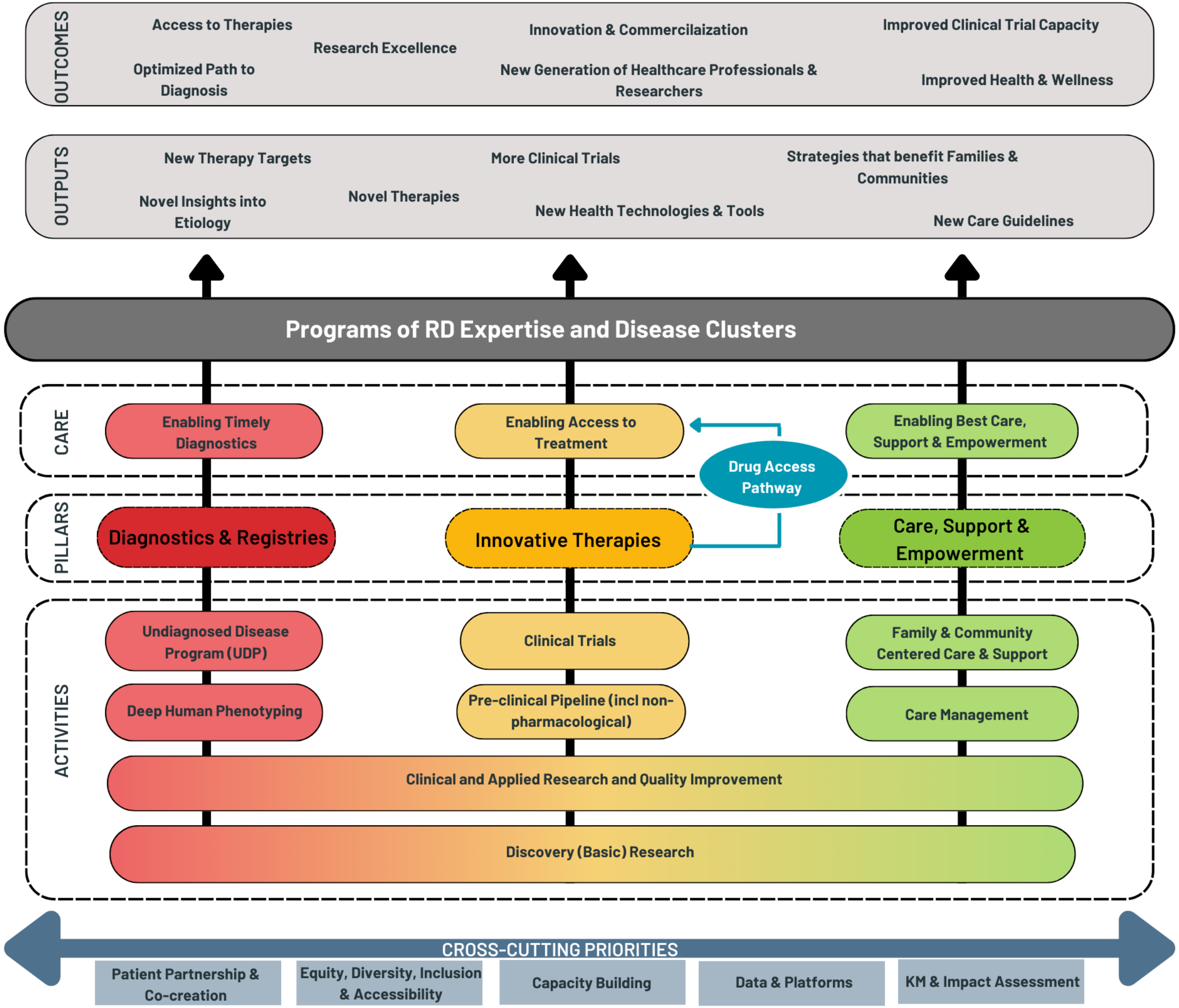
Pillars of our Work

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

A Research-To-Care Transformation Framework



OUTCOMES

Access to Therapies

Research Excellence

Innovation & Commercilaization

Improved Clinical Trial Capacity

Optimized Path to Diagnosis

New Generation of Healthcare Professionals & Researchers

Improved Health & Wellness

OUTPUTS

New Therapy Targets

More Clinical Trials

Strategies that benefit Families & Communities

Novel Insights into Etiology

Novel Therapies

New Health Technologies & Tools

New Care Guidelines

Programs of RD Expertise and Disease Clusters

CARE

Enabling Timely Diagnostics

Enabling Access to Treatment

Drug Access Pathway

Enabling Best Care, Support & Empowerment

PILLARS

Diagnostics & Registries

Innovative Therapies

Care, Support & Empowerment

ACTIVITIES

Undiagnosed Disease Program (UDP)

Clinical Trials

Family & Community Centered Care & Support

Deep Human Phenotyping

Pre-clinical Pipeline (incl non-pharmacological)

Care Management

Clinical and Applied Research and Quality Improvement

Discovery (Basic) Research

CROSS-CUTTING PRIORITIES

Patient Partnership & Co-creation

Equity, Diversity, Inclusion & Accessibility

Capacity Building

Data & Platforms

KM & Impact Assessment

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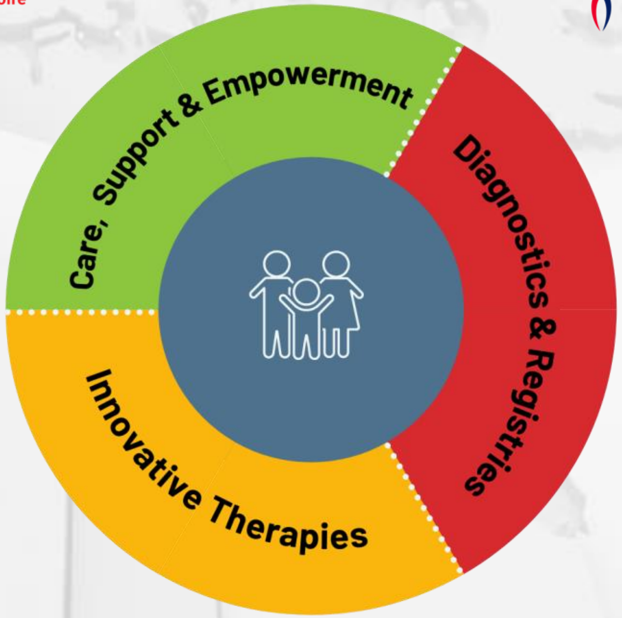
Collaborative Effort to Drive Impact



NEWBORN SCREENING ONTARIO
DÉPISTAGE NÉONATAL ONTARIO

MUSCULAR DYSTROPHY CANADA
DYSTROPHIE MUSCULAIRE CANADA

CHU Sainte-Justine
Le centre hospitalier universitaire mère-enfant
Université de Montréal

Centre universitaire de santé McGill
Institut de recherche
McGill University Health Centre
Research Institute

neuro

RARE DISEASES MODELS and MECHANISMS NETWORK

Canadian Autoinflammatory Network
Réseau Auto-inflammatoire Canadien

AllforOne
Canada's Precision Health Partnership

Children's Hospital
London Health Sciences Centre

Canadian VHL Alliance
Connecting a Community of Warriors

YORK U

genomics4rd

iRARE centres

Can-GARD
Canadian Gene Cure Advanced Therapies for Rare Disease
Gene Cure Canada
Thérapies innovantes pour les maladies rares

RQMO
REGROUPEMENT QUÉBÉCOIS DES MALADIES ORPHELINES

Canada
CNRC·NRC

CHEO
RESEARCH INSTITUTE
INSTITUT DE RECHERCHE

CARE forRARE

Canadian Association of Pompe
Association canadienne de Pompe

NMD4C

INFORM RARE

BC Children's Hospital
Research Institute

The Ottawa Hospital
Research Institute
L'Hôpital d'Ottawa
Institut de recherche

SickKids
RESEARCH INSTITUTE

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

Alberta Children's HOSPITAL
RESEARCH INSTITUTE

CORD

RareKids CAN

MICYRN
better health for mothers and children

CNDR.ORG
Canadian Neuromuscular Disease Registry

DEFEAT DUCHENNE CANADA

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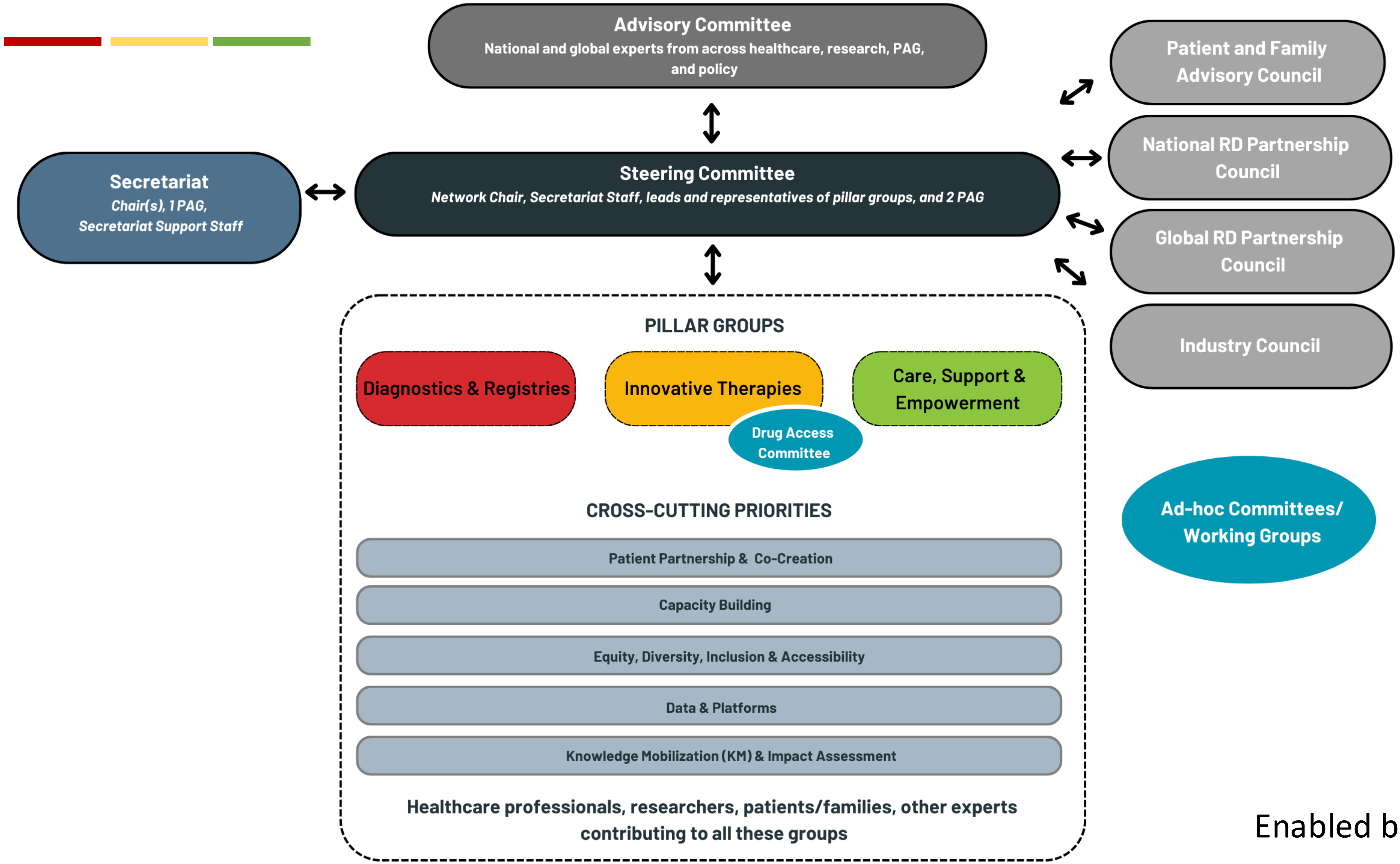
Governance



Advisory Committee
National and global experts from across healthcare, research, PAG, and policy

Secretariat
Chair(s), 1 PAG,
Secretariat Support Staff

Steering Committee
Network Chair, Secretariat Staff, leads and representatives of pillar groups, and 2 PAG

Patient and Family Advisory Council

National RD Partnership Council

Global RD Partnership Council

Industry Council

Ad-hoc Committees/ Working Groups

PILLAR GROUPS

- Diagnostics & Registries
- Innovative Therapies
  - Drug Access Committee
- Care, Support & Empowerment

CROSS-CUTTING PRIORITIES

- Patient Partnership & Co-Creation
- Capacity Building
- Equity, Diversity, Inclusion & Accessibility
- Data & Platforms
- Knowledge Mobilization (KM) & Impact Assessment

Healthcare professionals, researchers, patients/families, other experts contributing to all these groups

Enabled by: one child every child

Our Steering Committee Members

Francois Bernier, MD
Alberta Children's Hospital,
University of Calgary

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

Jim Dowling, MD, PhD
Sick Kids Hospital,
University of Toronto

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

Jacques L. Michaud
Centre de recherche du CHU Sainte Justine,
Université de Montréal

Gail Ouellette, PhD
iRARE Centre, RQMO

Lawrence Korngut, MD
Hotchkiss Brain Institute,
University of Calgary

Angela Genge, MD
Montreal Neurological Institute – Hospital,
McGill University

Jonathan Pratt
Regroupement Québécois des maladies orphelines (RQMO)

Craig Campbell
Children's Hospital LHSC,
Western University

Leanne Ward, MD
Children's Hospital of Eastern Ontario (CHEO) Research Institute,
University of Ottawa

Ian Stedman, PhD
York University

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

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

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Our Story Thus Far

Feb 2024

Public & Website Launch

Public launch of CRDN on RD Day

CORD
Rare Disease Day

March 2024

Global Positioning

CRDN gathering global attention

RARE DISEASES INTERNATIONAL
IRDiRC INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM
orpha News

May 2024

ERDERA
RareKids CAN | MICYRN better health for mothers and children

ERDERA NMG Co-Leadership

ERDERA NMG co-leadership with RareKids-CAN

Jun-Aug 2024

Strategic Plan Engagement

Targeted engagement sessions (n=12) with 33 experts to co-develop strategic plan

Sep 2024

Partnership Explorations

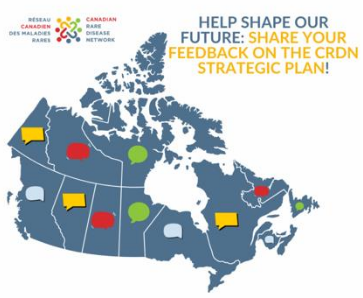
Meetings and exploration of collaborations

Health Canada

Oct 2024

Townhall & Public Consultation

Overview of strategic plan presented to over 150 attendees and launched feedback survey



Co-Development of Our Strategic Plan

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STEP 01

STRATEGIC ENGAGEMENT

May – August 2024

Targeted engagement sessions (n=12) with a select but diverse group of 33 experts, resulting in >900 minutes of meaningful dialogue

STEP 02

VIRTUAL TOWNHALL

31 October 2024

Overview of draft strategic plan presented to broader community and launch of community feedback survey

STEP 03

COMMUNITY FEEDBACK

October – November 2024

Widely distributed public survey to gather extensive feedback from the broader community over a 1-month period

STEP 04

REVIEW & APPROVAL

December – January 2025

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

Note: Interested parties will have an opportunity to provide feedback on the proposed strategic plan between October 31 to November 30, 2024.

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 diagnosed and undiagnosed RDs 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

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 determinants of health, 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 and market potential, thereby attracting investment and accelerating access to life-changing therapies

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 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 determinants of health

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

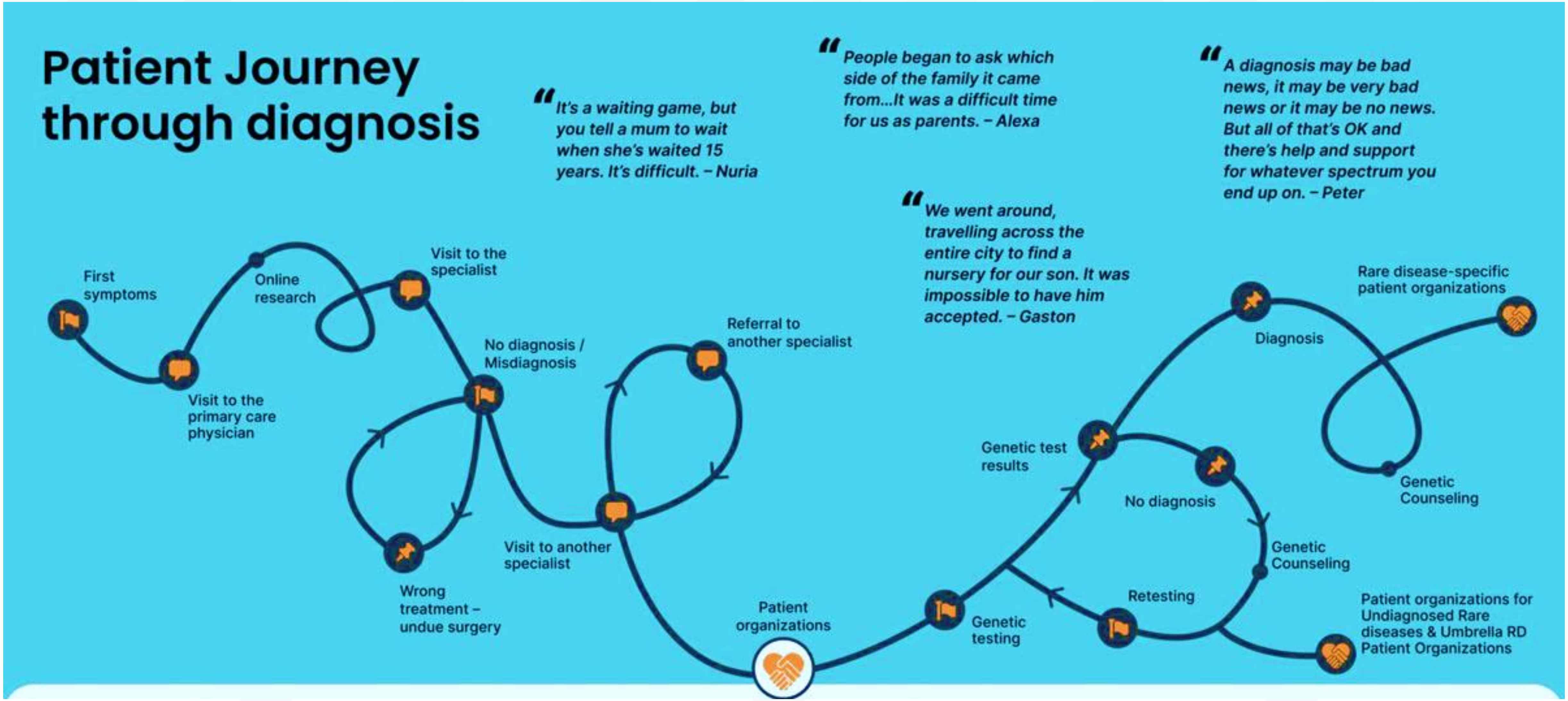
04 National & Global Collaboration

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

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

Diagnostics & Registries

Pillar 1 – Diagnostics & Registries



Patient Journey through diagnosis

> *It's a waiting game, but you tell a mum to wait when she's waited 15 years. It's difficult. – Nuria*

> *People began to ask which side of the family it came from...It was a difficult time for us as parents. – Alexa*

> *A diagnosis may be bad news, it may be very bad news or it may be no news. But all of that's OK and there's help and support for whatever spectrum you end up on. – Peter*

> *We went around, travelling across the entire city to find a nursery for our son. It was impossible to have him accepted. – Gaston*

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Pillar 1 - Members

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

Taila Hartley
Care4Rare

Lawrence Korngut
Hotchkiss Brain Institute,
University of Calgary

Bhavi Modi
BC Children’s Hospital,
University of British Columbia

Jillian Parboosingh,
University of Calgary

Beth Potter
University of Ottawa

Myriam Srour
Montreal Children’s Hospital
McGill University

Stuart Turvey
BC Children’s Hospital,
University of British
Columbia

Hilary Vallance
BC Children’s Hospital,
University of British
Columbia

Jodi Warman Chardon
Ottawa Hospital,
University of Ottawa

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Pillar 1 – What We Hope to Achieve

Reducing the time it takes to identify rare diseases

All rare disease patients will receive the right diagnostic test at the right time regardless of where they live in Canada

Genetic diagnostic laboratories across Canada will integrate resources and best practice guidelines to ensure high-quality genome-wide sequencing (GWS) for patients

All families with diagnosed and undiagnosed rare diseases will have access to relevant registries for secondary research and re-contact

Rare disease diagnostics and research will be a political priority and sustainably funded

Canada will be a world leader in rare disease mechanism discovery and translation of new technologies into the clinic

Benefits:

- Improved healthcare provider knowledge **and** wider use of high-quality genetic testing
- Faster, more accurate diagnoses **and** access to relevant registries

Innovative Therapies

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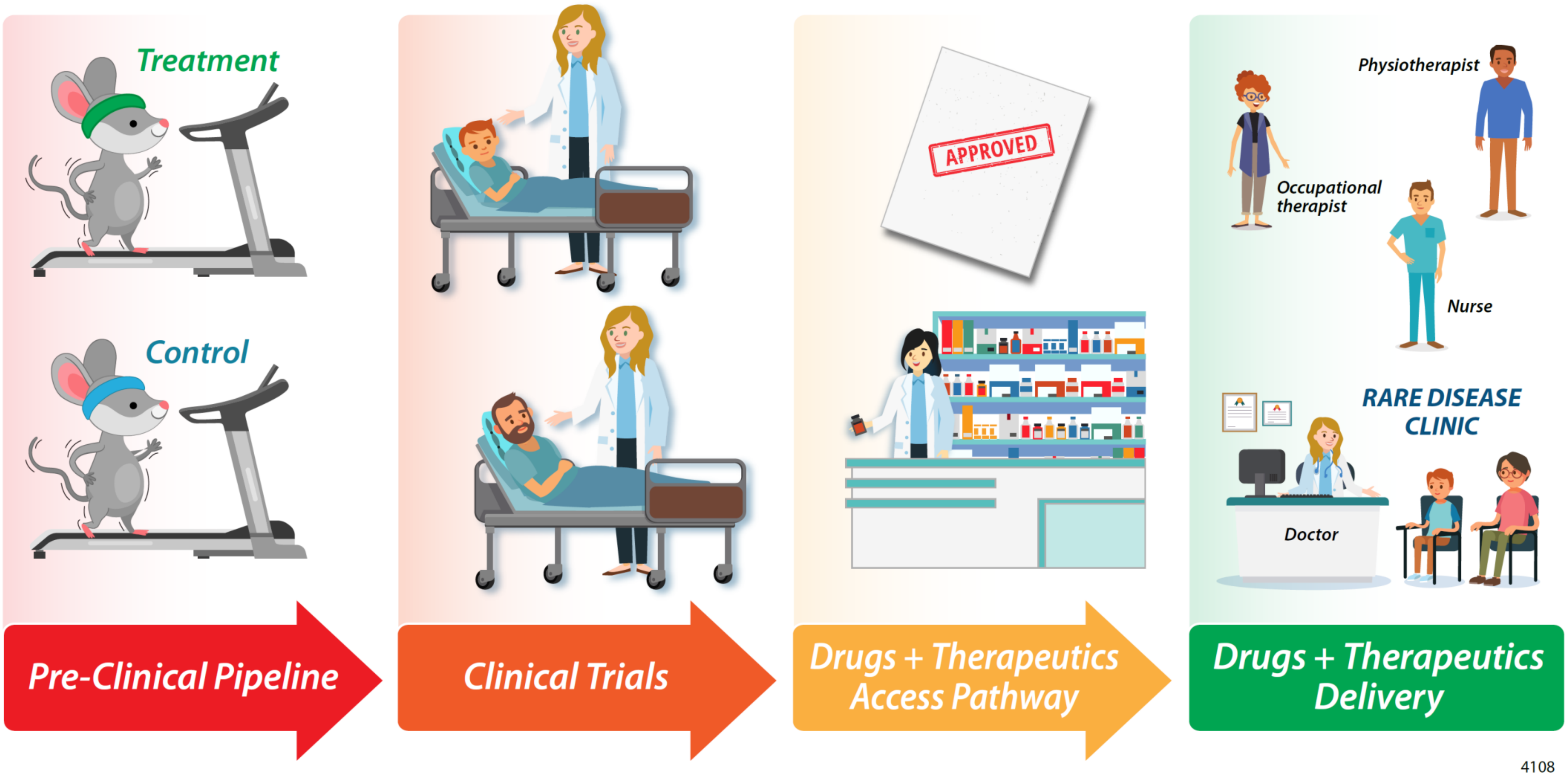
Pillar 2 – Innovative Therapies

Effective, Innovative Therapies



Pre-Clinical Pipeline

Clinical Trials

Drugs + Therapeutics Access Pathway

Drugs + Therapeutics Delivery

Pillar 2 - Members

Pillar Lead:

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

Pillar Members:

Craig Campbell
Children's Hospital LHSC,
Western University

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

Jim Dowling
Sick Kids Hospital,
University of Toronto

Heather Howley
Children's Hospital of
Eastern Ontario (CHEO)
Research Institute

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

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

Kim McBride
Alberta Children's
Hospital, University of
Calgary

Maryam Oskoui
Montreal Children's
Hospital,
McGill University

Breanne Stewart
RareKids-CAN

Risini Weeratna
National Research
Council (NRC)

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

Pillar 2 – What We Hope to Achieve

Expanding treatment possibilities

Canada will lead in the discovery and validation of novel therapeutic targets and treatments for rare disease patients

All rare disease patients, regardless of their age, location, or social context, will have equitable access to clinical trials and innovative therapies

Innovative therapies will be readily integrated into clinical practice to improve patient care and outcomes

Canada will be recognized globally for its rare disease clinical trials and market potential, thereby attracting investment and accelerating access to innovative therapies

Benefits:

- More treatment options **and** better access to innovations
- More consistent, quality care closer to home

Care, Support & Empowerment

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Pillar 3 – Care, Support, & Empowerment

Comprehensive Care and Support (wrap-around)

Community and Home-Based Care

Mental Health and Psychosocial Support

Financial Assistance

Family and Caregiver Support and Respite

Patient and Carer Wellbeing

Education, Advocacy, and Empowerment

Social Support

Care Transition

Resource Navigation and Service Access

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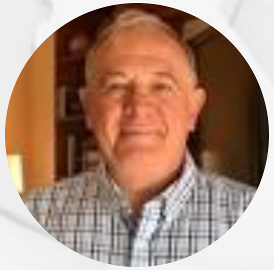
CANADIAN RARE DISEASE NETWORK

Pillar 3 - Members

Pillar Lead:

Ian Stedman
York University

Pillar Members:



John Adams
Canadian PKU and Allied Disorders (CanPKU+)

Jillian Banfield
Canadian Institutes for Health Research – Institute of Genetics (CIHR IG)

Brad Crittenden
Canadian Association of Pompe

Deborah Marshall
University of Calgary

Homira Osman
Muscular Dystrophy Canada

Gail Ouellette
iRARE Centre, RQMO

Stephen Parrott
Kidney Cancer Canada Board

Jonathan Pratt
Regroupement Québécois des maladies orphelines (RQMO)

Nicola Worsfold
World Duchenne Organization

Pillar 3 – What We Hope to Achieve

Supporting patients and their families

All rare disease patients, along with their families and caregivers, will be aware of and have equitable access to the resources and supports they need

All individuals affected by rare diseases will be empowered and engaged in meaningful opportunities in research and beyond

All rare disease patients and their families will receive the mental health and wellbeing support they need regardless of their location or social context

Canada will have a unified rare disease community that creates comprehensive care and support systems for rare disease patients and their families

Benefits:

- Greater awareness **and** more equitable access to resources
- Greater patient empowerment, knowledge sharing, and sense of community

How to Be Involved

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1. Read the **proposed strategic plan** from: https://canadianrdn.ca/public-consultation-on-the-canadian-rare-disease-network-proposed-strategic-plan/

- Summary of goals and strategies for each CRDN Pillar

2. Submit your feedback through the **online survey:** https://survey.ucalgary.ca/jfe/form/SV_4YOlL6DCpCvKYey

  - Provide feedback through a combination of free-text and multiple-choice fields

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Online feedback deadline: November 30, 2024