RÉSEAU CANADIEN DES MALADIES RARES

CANADIAN RARE DISEASE NETWORK

Rare Lives, Shared Strength: A Collaborative Strategy for Rare Diseases in Canada

2025 - 2030

LAND ACKNOWLEDGEMENT

The Canadian Rare Disease Network (CRDN) recognizes the importance of Indigenous knowledge, wisdom, and perspectives in the pursuit of our shared goals.
We are dedicated to building meaningful relationships with Indigenous partners, listening, and learning from their experiences, and respecting Indigenous perspectives in our work for the betterment of all communities affected by rare diseases.
We commit to upholding the principles of respect, humility, and reciprocity, guided by the understanding that the journey towards inclusivity and reconciliation is ongoing and requires collective effort.

The work of the CRDN occurs from coast to coast and on the traditional, ancestral and unceded territories of many Indigenous peoples and nations. The CRDN acknowledges the profound and enduring connections that Indigenous communities hold with the land and waters we inhabit, and the non-human relatives, and we honour, recognize, and respect these nations as the traditional knowledge keepers and stewards of these lands where we work and meet.

CONTENT

EXECUTIVE SUMMARY

Developed by the Rare Disease Community, for the Rare Disease Community

The Canadian Rare Disease Network (CRDN) – Réseau Canadien des Maladies Rares (RCMR) – presents a comprehensive, collaborative strategic plan to transform the landscape of rare disease (RD) care, research, and innovation in Canada. This plan co-developed with the RD community – individuals affected by RDs, caregivers, clinicians, researchers, and other interest holders – reflects a shared commitment to addressing the complex challenges faced by the RD community. Guided by the voices and lived/living experiences of those directly impacted by rare diseases, this strategy outlines a coordinated national approach to improve diagnostics, expand treatment options, strengthen care and support, and position Canada as a global leader in rare disease research and collaboration.

The strategic plan is built around four key objectives (pillars):

PILLAR 1: DIAGNOSTICS & REGISTRIES

Reducing the time it takes to identify rare diseases

PILLAR 2: INNOVATIVE THERAPIES

Expanding treatment possibilities

PILLAR 3: CARE, SUPPORT, & EMPOWERMENT

Supporting patients and their families

PILLAR 4: NATIONAL & GLOBAL COLLABORATION

Connecting Canada to drive rare disease breakthroughs

Each pillar addresses critical gaps and opportunities, and together, they form a comprehensive and integrated approach to RD management. Central to this strategy is a novel research-to-care framework that connects discovery, diagnostics, treatment development, and patient-centered care within a seamless continuum – fostering greater impact and sustainability. Through its implementation, Canada will strengthen its leadership in the global RD community and significantly improve the lives of those with lived/living experience of a RD.

This plan is a testament to the collective effort of the RD community, reflecting a shared vision to drive innovative care and research, and empowering all patients and families affected by RDs to live their full potential. As we move forward, we remain committed to ongoing engagement with the RD community and partners, ensuring that our actions are aligned with the needs of those we serve. By fostering collaboration, driving innovation, and amplifying the voices of patients, the CRDN is poised to make a lasting impact—improving lives, advancing global RD research, and setting new standards for comprehensive, patient-centered care in Canada.

While this plan reflects a significant step forward, CRDN acknowledges that Indigenous Peoples have not been meaningfully included in its development—an essential gap that must be addressed in the future. Indigenous individuals and communities experience distinct and disproportionate barriers to rare disease care, including challenges in access to diagnostics, treatment, and culturally safe services. Moving forward, CRDN is committed to working in partnership with Indigenous communities and leaders to explore a path that ensures their perspectives, experiences, and needs are included in RD research and care in a way that respects self-determination and cultural safety.

By fostering inclusivity, this strategy aims to address systemic inequities in rare disease care and research, and build a more equitable, representative, and effective rare disease ecosystem in Canada. We would like to extend our deepest gratitude to the entire RD community for their invaluable contributions to this strategic plan. Together, we are shaping a brighter future for those affected by RDs across Canada.

A Note on Terminology

To acknowledge different language preferences, we have chosen to use both person-first language (e.g., ‘individuals affected by rare diseases’, ‘person with lived/living experience of a rare disease') and identity-first language (e.g., 'rare disease patient’) interchangeably.

In addition, we use the term interest holders rather than stakeholders to reflect a more inclusive and respectful approach. The term “stakeholder” has colonial and proprietary connotations, whereas “interest holder” more accurately captures the broad range of people, communities, and organizations who are invested in, affected by, or contributing to the rare disease ecosystem.

PLAN ON A PAGE

RÉSEAU CANADIEN DES MALADIES RARES

CANADIAN RARE DISEASE NETWORK

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.

MISSION

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

VALUES

Patients and Families First
Collaboration and Inclusivity
Equity
Innovation
Excellence
Solidarity and Mutual Respect
Empowerment

PILLAR 1: DIAGNOSTICS AND 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.

PILLAR 2: 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 gain global recognition for its RD clinical trials and market potential, attracting investment and accelerating access to new therapies.

PILLAR 3: CARE, SUPPORT, AND 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 creates comprehensive care and support systems for RD patients and their families.

PILLAR 4: NATIONAL AND 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.

RARE DISEASES IN CANADA

A Public Health Challenge in Need of Action

Rare Diseases aren't that Rare

Rare diseases (RDs) pose a complex and often overlooked challenge within the Canadian healthcare landscape. To date over 7,000 different RDs have been discovered [1,2], and while each disease might affect a small percentage of the population, the total number of Canadians living with RDs is substantial. Approximately 1 in 12 individuals in Canada - over 3 million - are affected by a RD [3], reflecting a significant portion of the population. Globally, around 400 million people are affected by RDs [4], highlighting the widespread nature of these conditions. About 75% of those affected are children and more than 30% of them will not live to see their 5th birthday [5].

About 80% of RDs are genetic in origin, while the remaining are non-genetic, including certain cancers, infections, and autoimmune disorders [6,7]. Many RDs are complex and progressive, leading to severe impacts on life expectancy, physical health, and psychosocial wellbeing for those affected [8]. The lack of effective treatments for most RDs globally (about 95% lack approved treatments) means that improving quality of life and extending life expectancy often hinges on timely and accurate diagnosis, along with appropriate care and support [4,6].

While there is large variation among RDs, people living with a RD face common challenges. One of the most pressing challenges is the "diagnostic odyssey"—a lengthy and often frustrating journey to obtain a correct diagnosis [9,10]. This difficulty is compounded by the complexity and rarity of these diseases, which are not typically covered in standard medical training [11-13]. As a result, delays in diagnosis and misdiagnoses are common, significantly impacting patient outcomes and access to appropriate treatments.

Beyond diagnostic difficulties, individuals with RDs encounter other challenges including limited treatment options, fragmented and complex healthcare and social systems, and a lack of comprehensive support. Financial burdens, such as out-of-pocket costs for care and loss of income from caregiving responsibilities, add to the strain. The emotional and psychological impacts of navigating the RD journey, alongside stigmatization and insufficient research, despite the recognized gaps in knowledge, further exacerbate the difficulties faced by RD patients and their families. Healthcare professionals, researchers, and the pharmaceutical industry also encounter obstacles in securing funding, coordinating research, and developing treatments due to the rarity and complexity of these conditions [14].

Despite global advancements, Canada is lagging behind in improving the health and well-being of RD patients and their families. In a country as vast and diverse as Canada, the multifaceted challenges posed by RDs require a coordinated, national approach—one that bridges gaps, promotes cross-jurisdictional knowledge sharing, and empowers partners through a multidisciplinary strategy that spans the research-to-care continuum [15]. This approach is crucial for overcoming existing barriers and ensuring that RD patients in Canada receive the timely and effective care they need.

ABOUT THE CANADIAN RARE DISEASE NETWORK (CRDN)

Our Vision and Mission

The Canadian Rare Disease Network (CRDN) was launched in February 2024 to reshape the way rare diseases are understood, diagnosed, treated, and managed across Canada. Our vision is for "**Innovative care and research in Canada so that all patients and families affected by a rare disease are empowered to live their full potential**". This vision embodies our commitment to creating a rare disease landscape where no one is left behind.

CRDN's mission is to "**Establish a growing network that builds connections across geographies and disease boundaries to enable timely diagnosis, screening, and access to treatment, facilitate best care, support and empowerment, and global sharing of best practices for patients and their families in Canada, ultimately enhancing their quality of life**." By uniting Canada's leading clinical, scientific, and patient expertise in an inclusive, supportive, and collaborative approach where all partners – from patients and caregivers to researchers, healthcare providers, and policymakers are engaged – we are breaking down silos and forging a unified approach that leverages the strengths of all partners to improve the lives of those affected by rare diseases (**Figure 1**).

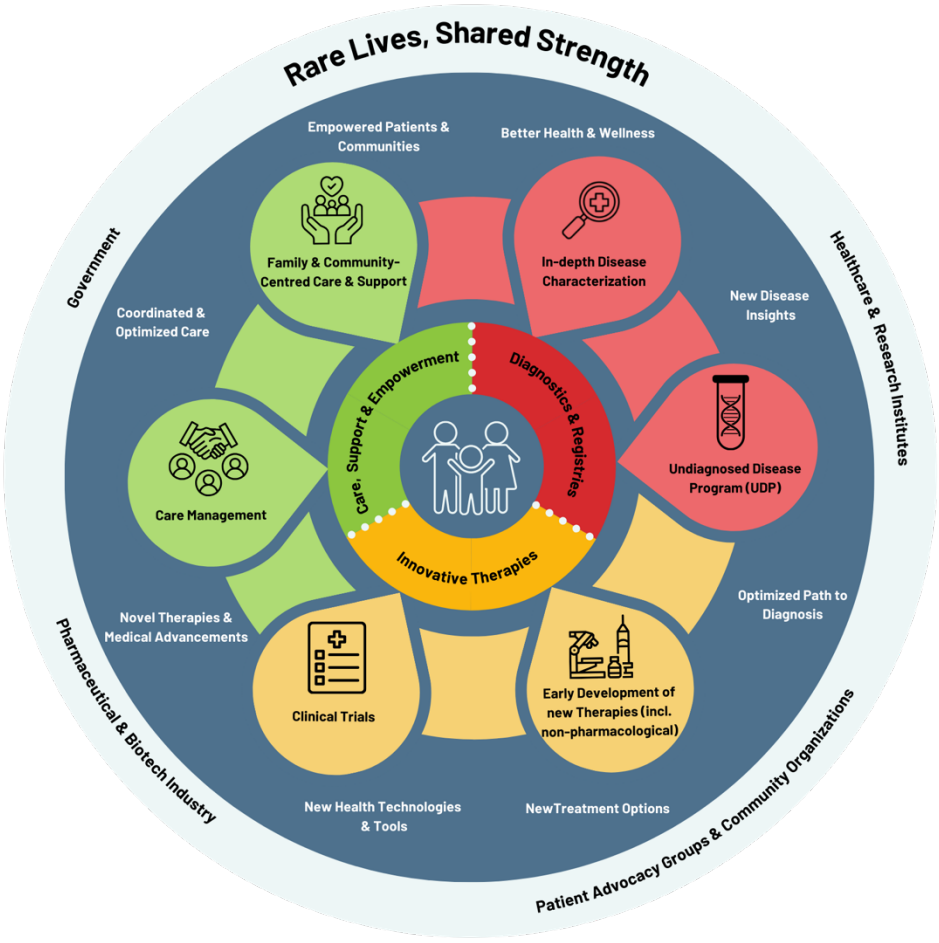

Figure 1: *Overview of CRDN's framework focused on three key challenges encountered by patients and families.*

At the core, CRDN's approach is a novel research-to-care framework that integrates every aspect of the RD journey – from research and diagnosis to treatment and patient-centered care - into a seamless continuum. This integrated framework addresses the spectrum of challenges faced by RD patients, closing gaps in knowledge and treatment while providing holistic support.

Our Values

Our approach and work are guided by seven values:

- **Patients and Families First** - Prioritizing the well-being and needs of RD patients and their families in all decisions and activities.
- **Collaboration & Inclusivity** - Encouraging strong partnerships and welcoming diverse interest holders, including patients, caregivers, healthcare professionals, researchers, and other interest holders from across Canada and disease types to work together for the betterment of those with RD.
- **Equity** - Ensuring equitable access to resources, information, and support for all individuals affected by RD, regardless of their background, location, or disease type.
- **Innovation** - Embracing creative and forward-thinking approaches to advance research, diagnosis, treatment, care, support and empowerment of RD patients, families, and communities.
- **Excellence** - Committing to conducting and supporting high-quality care and research and facilitating evidence-based care, support, and empowerment so that every patient and their families can live to their full potential.
- **Solidarity & Mutual Respect** - Fostering a sense of unity and togetherness and emphasizing respect for the unique perspectives, experiences, and contributions of all network partners.
- **Empowerment** - Equipping RD patients, their families, and communities to actively participate in their care, research, and advocacy efforts.

How We Work

CRDN thrives on purposeful, collaborative relationships and is committed to driving meaningful change by working **through partnerships** with key interest holders across Canada and globally. We are not here to start from scratch – we are here to connect the dots. By working in partnership, we aim to align efforts, amplify what is working, and fill gaps together. Rather than duplicating existing work, we focus on leveraging expertise, reducing fragmentation, and creating space for new collaboration where it's needed most. Every contributor, from researchers to those delivering care, and from patients sharing their experiences to those advocating for change, and everything in between, plays an essential role in driving CRDN's mission and vision forward in the years to come.

Support

CRDN is currently enabled by the University of Calgary's One Child Every Child research project supported by the Canada First Research Excellence Fund (CFREF). As CRDN continues to grow, ensuring long-term sustainability and securing dedicated funding will be key priorities.

OUR PROCESS OF CO-DEVELOPING THE STRATEGIC PLAN

At the heart of CRDN's approach lies a commitment to a person-centred philosophy. Recognizing that those who are most impacted and invested in RDs should shape our path, we embarked on co-developing this strategic plan in close partnership with the people who it will ultimately affect so that it reflects their experiences and ideas for change. Our process was rooted in listening deeply to a diverse range of voices, including patients, clinicians, researchers, advocates, and others.

The strong sense of collaboration by, and generosity of, the RD community throughout this process have been both inspiring and instrumental. We are fiercely committed to these ambitions and excited for the positive impact we can collectively have on improving the lives of those affected by RDs.

***Figure 2**: Overview of the co-development process for the CRDN strategic plan.*

CRDN acknowledges that Indigenous Peoples have not been meaningfully included in the development of this strategic plan—an important gap that must be addressed. Recognizing the distinct barriers Indigenous individuals and communities face in accessing rare disease care, CRDN is committed to working alongside Indigenous partners to explore a path that ensures culturally appropriate engagement, equitable access, and meaningful inclusion in RD research and care.

Step 1: Strategic Engagement and Prioritization

Between May to August 2024, we conducted a series of targeted engagement sessions (N=15) with a select and diverse group of 34 key experts, totaling over 1000 minutes of meaningful dialogue. This diverse group represented various sectors within the RD community, including health systems, research, professional bodies, and patient communities. Visit our website for details about the involved experts. For those who were unable to attend engagement sessions, individual consultations were conducted. The gathered insights helped us pinpoint priority areas for each CRDN Pillar, ensuring our strategic plan is both comprehensive and reflective of the community's needs and aspirations.

Step 2 & 3: Community Feedback

To ensure our strategic plan is inclusive, transparent, and broadly reflects the expectations of the RD community, we broadened our consultation efforts to include the entire RD community. On October 31, 2024, we held a virtual 1-hour town hall to provide an overview of the initial draft strategic plan to the broader RD community, followed by Q&A and the launch of a community feedback survey. The

town hall proved to be highly successful, with over 260 individuals registering for the event. More than 160 attendees actively participated, underscoring the strong interest and engagement from the RD community. A recording of the town hall is available on our website.

For 1 month (October to November 2024), we gathered extensive feedback from the broader RD community across Canada and various sectors (e.g., research, healthcare, patient communities, industry, government) through a widely distributed public survey, receiving 115 responses from across 10 out of the 13 provinces and territories. This feedback was crucial in refining our strategic plan, making it as inclusive and representative as possible (review feedback report).

Step 4: Review and Approval

The feedback was then reviewed and discussed by the CRDN Pillars and Steering Committee, and revisions were made to the strategic plan, where appropriate. We remain dedicated to continuously reviewing and updating the plan annually, ensuring it evolves to meet the needs of the community it serves.

CRDN STRATEGIC PLAN (2025 – 2030)

The strategic plan serves as a roadmap to guide CRDN's efforts over the coming years. It is strategically centred around three pillars that are designed to address key challenges faced by the RD community. In addition, there is a fourth 'enabling' pillar, which spans across all areas and aligns efforts and partnerships at both the national and global levels.

PILLAR 1: DIAGNOSTICS & REGISTRIES

Reducing the time it takes to identify rare diseases

PILLAR 2: INNOVATIVE THERAPIES

Expanding treatment possibilities

PILLAR 3: CARE, SUPPORT, & EMPOWERMENT

Supporting patients and their families

PILLAR 4: NATIONAL & GLOBAL COLLABORATION

Connecting Canada to drive rare disease breakthroughs

While each Pillar is presented separately below, it is essential to understand that, in reality, all pillars are interrelated and collectively contribute to a unified approach to rare disease management. Most importantly, this plan articulates how we will approach our vital work: by convening, connecting, and collaborating to enable the CRDN and its partners to fulfill their priorities and contribute to improved health for those affected by rare diseases in Canada. We are fiercely committed to these ambitions and excited for the positive impact we can collectively have on improving the lives of those affected by RDs.

Pillar 1: Diagnostics and Registries

Why is this important?

Having strong diagnostic capabilities is crucial for identifying rare diseases (RDS) early. This can lead to better outcomes for patients because it allows for timely and targeted treatments. Additionally, robust registries act as central databases that collect and analyze important information about RDs, such as how common they are (prevalence), who is affected, and how effective different treatments are. This information is essential for research and helps us understand RDs better, paving the way for new therapies. Effective screening programs, especially for newborns, help detect these diseases before symptoms develop. This early detection can prevent serious health issues and promote better long-term health outcomes. By improving our diagnostic tools, creating integrated registries, and enhancing screening methods, we can bridge critical gaps in our healthcare system. This will lead to better treatment strategies and drive innovation in RD research.

What we hope to achieve:

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

Why?	Over the next 5 years, we will:	Our work will result in:
Timely and accurate diagnosis is essential for effective RD treatment, yet access to diagnostic testing remains uneven across Canada. Many RD patients face prolonged diagnostic odysseys due to inconsistent availability of genetic and specialized testing, limited awareness among healthcare providers, and gaps in newborn screening programs. Compounding these challenges is the lack of specific medical codes (ICD codes) for RD, which makes it difficult to track patients within the healthcare system. By improving early detection, harmonizing and expanding access to diagnostic testing, and ensuring healthcare providers are equipped to identify and manage RD cases, we can streamline diagnosis, optimize healthcare resources,	i. Assess the current visibility/reporting of RD patients in the healthcare system. ii. Enhance education of non-geneticist physicians and allied healthcare professionals related to genetics/genomics to improve RD recognition and awareness of genetic testing options. iii. Develop a framework for mainstreaming of genetic tests for use by non-geneticist physicians to expand access to testing. iv. Improve and harmonize newborn screening across Canada.	More visible and accurately diagnosed RD patient population, enabling better healthcare planning; Improved genetics/genomics education among healthcare providers, leading to earlier recognition and referral; Broader use of genetic testing by non-genetics physicians, reducing diagnostic delays and improving care pathways; Harmonized newborn screening programs nationwide, ensuring all infants receive timely and equitable testing.

and build a system that provides equitable care for all RD patients—no matter where they live.		

GOAL 1.2: Genetic diagnostic laboratories across Canada will integrate resources and best practice guidelines to deliver high-quality genome-wide sequencing (GWS) for patients		
Why?	**Over the next 5 years, we will:**	**Our work will result in:**
Genome-wide sequencing (GWS) is a powerful tool for diagnosing RDs, yet disparities in resources, expertise, and best practices across Canada's genetic diagnostic laboratories result in inconsistent quality and accessibility. This variability affects diagnostic accuracy, delays treatment, and contributes to inequities in healthcare. By integrating resources, aligning standards, and fostering collaboration among genetic diagnostic laboratories**,** we can ensure consistent, high-quality genomic testing across the country**.** This will ultimately lead to timelier diagnoses, optimized patient care, and better health outcomes for individuals with RDs**.**	i. Facilitate the sharing of clinical data across diagnostic laboratories to improve the interpretation of genetic variants. ii. Develop best practice guidelines to enhance standards in genomic testing.	Nationally consistent, high-quality genomic testing practices, leading to more equitable, accurate and timely diagnoses, and faster turnaround times for test results; Stronger collaboration among genetic diagnostic laboratories, leveraging collective expertise for better patient outcomes.

GOAL 1.3: All families with diagnosed and undiagnosed RDs will have access to relevant registries for secondary research and recontact		
Why?	**Over the next 5 years, we will:**	**Our work will result in:**
Comprehensive and accessible registries are critical for advancing RD research, enabling secondary research, and facilitating patient participation in clinical trials. However, many RD families—both diagnosed and undiagnosed—struggle to	i. Support the establishment of a national network of registries to streamline and enhance the use of registry data across Canada. ii. Support the development of best practices and	National registry network, improved registry quality and data linkage, making registry-based research more efficient and impactful; Enhanced access to relevant registries and

access relevant registries. Without a unified approach, valuable clinical and genomic data remains siloed, slowing research progress and reducing opportunities for patient recontact when new treatments or studies become available. By establishing a nationally coordinated network of registries and facilitating universal access, we can ensure that all RD families have access to databases that support research and enable patient re-engagement for emerging treatments.	standards to enhance registry quality, consistency, and linkage across different databases. iii. Support the development of a national re-contact registry to facilitate the participation of patients/families and healthcare professionals in research and clinical trials.	timely recontact for all RD families, leading to greater participation in research and clinical trials which accelerates discoveries.

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

Why?	**Over the next 5 years, we will:**	**Our work will result in:**
For RD diagnostics and research to thrive, they must be prioritized both politically and financially. Genomic medicine holds the potential to revolutionize RD diagnosis and treatment, but without dedicated funding and policy support, its full impact will remain limited. Securing this support will empower healthcare systems to adopt genomic tools and integrate them into routine care. It will foster innovation in RD research, streamline pathways for diagnosis, and provide the foundation for long-term healthcare advancements in genomics. Ultimately, prioritizing RD diagnostics and research will ensure a sustainable, future-	i. Coordinate and align national initiatives to inform the design of a genomics ecosystem and its integration into the healthcare system. ii. Evaluate the readiness of the healthcare system to integrate genomics into existing care pathways, and provide necessary guidance based on evaluation results to enhance preparedness iii. Develop policy briefs to advocate for the development and sustainability of necessary infrastructure that support genomic research and diagnostics.	Better integration of genomics into healthcare, fostering the widespread adoption of genomic tools; Better preparedness for genomic care pathways, ensuring systems are equipped to incorporate genomics into patient care; Sustained infrastructure support for genomic research and diagnostics that ensures long-term success and improved patient outcomes.

ready healthcare system that can integrate the latest advancements to meet the needs of RD patients and their families.		

GOAL 1.5: Canada will be a world leader in RD mechanism discovery and translation of new technologies into the clinic		
Why?	**Over the next 5 years, we will:**	**Our work will result in:**
For Canada to position itself as a global leader in RD research, it is critical to focus on the discovery of RD mechanisms and the translation of emerging technologies into clinical care. By contributing to global RD gene catalogues, validating new technologies, and creating a national genomic infrastructure, Canada can lead efforts in accelerating diagnostic breakthroughs and ensuring equitable access to cutting-edge innovations, which will all ultimately benefit the quality patient care.	i. Contribute to the catalogue of RD genes and mechanisms to support identification of disease and potential therapeutic targets. ii. Generate utility data on the use and effectiveness of emerging and new technologies for RD diagnostics to ensure faster integration into clinical practice. iii. Establish national genomic infrastructure and resources accessible to researchers nationally to drive cutting-edge research in RD.	Enhanced understanding of RD genes and mechanisms, improving diagnostic accuracy and enabling therapy development; Critical data on new diagnostic technologies that will inform decision-making around their clinical application; Robust national genomic infrastructure, enabling researchers to access necessary resources, collaborate and contribute to the global RD research community; Faster translation of discoveries into clinical care, ensuring that Canadian patients benefit from the latest advancements.

Pillar 2: Innovative Therapies

Why is this important?

Innovative therapies offer new hope for treating RDs, especially when traditional treatments are ineffective, limited, or don't exist. Developing new treatments that target the underlying causes of

these diseases can lead to life-changing or potentially curative options for patients. Equally important is ensuring that patients can easily access clinical trials, that effective, new treatments are approved quickly and safely, and that they are effectively integrated into clinical care to benefit patients as soon as possible. To make this possible, we need collaboration across research, healthcare, industry, and policy to build a strong infrastructure that supports pre-clinical and clinical research plus knowledge mobilization and integration into the clinic for direct patient access (both pharmacological and non-pharmacological treatments such as gene and cell therapies, digital and assistive technologies, physical and rehabilitative therapies, dietary approaches, etc.). Ensuring equitable access for all patients, regardless of location or background, is key. Additionally, providing education and ongoing support for both healthcare providers and patients will help ensure that these therapies are used effectively and safely. By focusing on these areas, we expand the treatment options available to rare disease patients, improve health outcomes, and drive progress across the field.

What we hope to achieve:

GOAL 2.1: Canada will lead in the discovery and validation of novel therapeutic targets and treatments for RD patients		
Why?	**Over the next 5 years, we will:**	**Our work will result in:**
Canada has the potential to lead in the discovery and development of novel RD therapies. However, the current landscape for RD therapy development is fragmented, with disconnected pre-clinical research efforts, limited infrastructure, and a lack of a coordinated strategies to translate research into viable therapies. By addressing these gaps through coordinated national initiatives, investments in essential technologies, and pre-clinical framework, Canada can enhance its research capabilities, foster innovation, and accelerate the development of new therapies to improve patient outcomes.	i. Coordinate and align initiatives, resources, and interest holders involved in pre-clinical research, therapy development, and production to streamline the translation of discoveries into therapeutic applications. ii. Develop rationale for creating and retaining pre-clinical therapy development capacity in Canada and create policies that advocate for “made for Canada” approaches to therapy development.	A more coordinated national approach to RD therapy development, where research, resources, and interest holders are aligned to maximize impact; Increased number of validated therapeutic targets ready for clinical trials, bringing new therapeutic options to RD patients; Enhanced policy support for necessary technologies, infrastructure, and resources necessary to develop therapies.

GOAL 2.2: All RD patients, regardless of their age, location, or social context, will have equitable access to clinical trials and innovative therapies		
Why?	**Over the next 5 years, we will:**	**Our work will result in:**

Equitable access to clinical trials and innovative therapies is critical for improving the lives of individuals affected by RDs. However, the current landscape of RD clinical trials and therapy access is fragmented and presents various barriers, particularly for certain populations. These barriers include geographic disparities, age-related gaps, and social determinants of health such as income, education, and cultural factors. Moreover, current infrastructure for RD clinical trials is underdeveloped, and there are regulatory and technical challenges that hinder timely access to treatments. By strengthening infrastructure, streamlining processes, and advocating for policies that promote equal access, we can ensure that all RD patients, regardless of location, age, or social determinants of health, can benefit from cutting-edge treatments and advancements.	i.	Strengthen and build clinical trial infrastructure to ensure equitable access to RD therapies across the lifespan (from pediatric to adult to elderly) and from coast to coast to coast (including underserved regions and rural areas).	A more streamlined national clinical trial infrastructure, enabling increased participation and access to therapies regardless of geography, age, or socio-economic background;
	ii.	Advocate for streamlined regulatory and approval processes that ensure timely approval of and access to RD therapies.	Streamlined regulatory and approval processes, ensuring timely availability and access to RD therapies across the country;
	iii.	Understand and address disparities in access to clinical trials and therapies, particularly those stemming from socio-economic, geographic, and cultural barriers, by leveraging registries and real-world data to inform improvement strategies.	Improved access to real-world evidence through registries and data collection, enabling informed decision-making on therapeutic needs and treatment efficacy.
	iv.	Promote comprehensive health economics evaluations to demonstrate the societal and clinical benefits of RD therapies, guiding evidence-based policies for access, coverage, and reimbursement.	

GOAL 2.3: Innovative therapies will be readily integrated into clinical practice to improve patient care and outcomes			
Why?	**Over the next 5 years, we will:**		**Our work will result in:**
Innovative therapies are transforming the treatment landscape for RDs, offering new hope where few or no options	i.	Improve education of non-RD physicians and allied healthcare providers on current and emerging RD	More consistent access to therapies, improving patients' health

previously existed. However, for patients to fully benefit, these therapies must be rapidly and effectively integrated into clinical practice**.** Currently, many healthcare providers—especially those not specialized in RDs—may lack the knowledge and resources needed to deliver and monitor these treatments. Without proper education, support, and care coordination, patients risk inconsistent access, suboptimal care, or treatment delays. By enhancing provider education, creating expert support networks, and strengthening care pathways across Canada, we can ensure that RD patients receive timely, appropriate, and high-quality care close to home.	therapies, delivery protocols, and long-term monitoring. ii. Establish expert support networks to provide virtual support, guidance, consultation, and mentorship to healthcare providers managing RD patients, both in urban and rural settings. iii. Build a comprehensive healthcare provider network that spans all levels of care, from tertiary to community settings, ensuring appropriate and continuous patient management.	outcomes and quality of life; Enhanced awareness, knowledge, and confidence of healthcare providers to deliver and monitor RD therapies, leading to more informed and effective care; Specialized support and guidance, when needed, ensuring that healthcare teams can make informed decision and expand their local capacity to deliver "care closer to home", reducing the strain on families and healthcare providers; A more cohesive healthcare provider network with care pathways, ensuring patients receive consistent support throughout their journey.

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

Why?	**Over the next 5 years, we will:**	**Our work will result in:**
Global recognition of Canada's RD clinical trial capabilities and market potential is vital for attracting investment, fostering international partnerships, and accelerating innovation. By strengthening our clinical trial	i. Showcase Canada's capabilities and potential in the RD clinical trial and therapy market to national and international interest holders, including, industry,	Increased global recognition of Canada's RD clinical trial capacity, attracting new investment and fostering partnerships to drive innovation;

infrastructure and highlighting Canada's expertise, we can position the country as a leader in RD research and development. This will not only drive economic growth but also improve access to life-changing therapies for RD patients, both domestically and internationally.		investors, and research partners. ii. Enhance and leverage public-private partnerships by fostering collaboration between government, industry, patient organizations, and research institutions to increase investment, drive innovation, and expand RD clinical trial capacity.	Improved patient outcomes, as increased trial activity leads to accelerated access to therapies and better treatment options; Enhanced research and development opportunities, creating a more dynamic RD clinical trial and therapy ecosystem.

Pillar 3: Care, Support, and Empowerment

Why is this important?

Effective care for individuals affected by RD and their families extends far beyond just medical care – it requires a holistic, wrap-around approach that addresses their full spectrum of needs throughout their entire journey. This includes psychological, emotional, and mental health supports, access to disability, financial assistance and income programs, education accommodations, as well as social connection, and practical day-to-day needs. By recognizing and responding to these multi-dimensional needs, we can build a more compassionate and effective care system. Addressing these needs will foster a more cohesive, family- and community-centered care system, significantly improving quality of life. Beyond meeting individual needs, this approach will strengthen the entire RD community—driving greater collaboration, advocacy, and innovative solutions across the healthcare system. It will also promote a more inclusive, patient-driven model of care, where individuals and families feel seen, supported, and empowered throughout their journey.

What we hope to achieve:

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

Why?	Over the next 5 years, we will:	Our work will result in:
Living with a RD is incredibly complex and challenging, not only due to the disease itself but also because of the difficulties in finding and accessing appropriate information, resources, and support. Although various resources and services exist, patients and families often	i. Consolidate and enhance awareness of available RD resources and support services for patients, including those who are undiagnosed, their families, caregivers, and healthcare professionals.	Improved awareness and access to essential resources and support services for RD patients and families, leading to better outcomes, reduced financial strain, and enhanced quality of life.

struggle to locate them, creating disparities in access. By consolidating information, improving visibility, and addressing regional gaps, we can ensure that all RD patients and their families—regardless of where they live—can access the support they need to improve their quality of life.	ii. Develop strategies to increase awareness and accessibility of available financial aid programs (federal, provincial, and private) and advocate for financial initiatives that address gaps (e.g., out-of-pocket expenses, travel support) to reduce financial strains on patients and families	

GOAL 3.2: All individuals affected by RDs will have the opportunity to be empowered and engaged in meaningful opportunities in research and beyond		
Why?	**Over the next 5 years, we will:**	**Our work will result in:**
Individuals affected by RDs and their families have unique, first-hand insights into the challenges and gaps in care, research, and policy. Yet, they are often excluded from key decision-making processes that directly impact their lives. Empowering individuals with lived/living experience of RDs to actively participate in research, healthcare policy, and advocacy is essential to ensuring that their voices shape solutions. When patients and families are meaningfully involved, research becomes more relevant, healthcare policies better reflect their needs, and advocacy efforts become more impactful.	i. Ensure patient and family representation in CRDN's leadership and activities by integrating them as equal and active partners in governance, strategy, and execution. ii. Ensure meaningful patient engagement by creating connection and facilitating direct involvement in research, research design, healthcare policy, and advocacy. iii. Support skill development for patient partners by working alongside patient engagement initiatives.	Greater patient empowerment and engagement in research, healthcare policy, and advocacy, ensuring patient voices shape the RD landscape; More patient-centered efforts, improving relevance, quality and impact of efforts by directly addressing needs and priorities of the RD community; Stronger collaborations and partnerships between patients, researchers, healthcare providers, and policymakers, driving meaningful change.

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

Why?	Over the next 5 years, we will:	Our work will result in:
Living with a RD places a significant emotional and psychological burden on patients and their families. Feelings of isolation, uncertainty, and distress are common, yet mental health needs are often unmet due to limited resources, lack of awareness, and disparities in access across regions. Ensuring that RD patients and their families have access to timely, specialized mental health support—no matter where they live or their social determinants of health—is essential for improving their overall quality of life.	i. Co-create actionable strategies and promote available mental health resources to enhance mental health and wellbeing of RD patients and their families. ii. Raise awareness of the mental health impacts of living with a RD and advocate for systemic changes in mental health support to ensure RD patients and their families (parents, siblings) are provided with appropriate support and services.	Improved access to mental health and wellbeing supports, improving quality of life and resilience; Greater awareness and recognition of mental health needs in RD care, leading to more compassionate and comprehensive support systems.

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

Why?	Over the next 5 years, we will:	Our work will result in:
While many RD patient organizations and other partners are doing vital work, they often operate in silos, limiting their collective impact. Fragmentation reduces the effectiveness of advocacy, support services, and care coordination. By uniting the RD community, fostering collaboration, and enhancing resource sharing, we can build a stronger, more coordinated network. This will amplify advocacy efforts, improve access to resources, and ensure that RD patients and their families receive more seamless and comprehensive support.	i. Establish a Canadian RD community of practice to foster collaboration, collective action, and mutual learning and resource sharing. ii. Enhance the visibility of RD efforts happening across Canada by promoting initiatives in research, diagnostics, treatment, care and support services, making it easier for interest holders to align their work and identify partnership opportunities.	A cohesive Canadian RD community that fosters collaboration, amplifies advocacy efforts, and drives collective action to improve care and support; Greater visibility of RD initiatives, enabling stronger partnerships and reducing duplication of efforts; More streamlined, patient-centered support systems, enhancing quality of life of RD patients and their families.

Pillar 4: National and Global Collaboration

Why is this important?

Effective collaboration on both national and global levels enables the sharing of knowledge, resources, and best practices, which accelerates progress and enhances outcomes. By fostering strong relationships among diverse partners—such as research institutions, healthcare providers, patient organizations, and international networks—we can collectively address complex challenges, drive innovation, and implement effective solutions. This pillar emphasizes the importance of creating and sustaining collaborative frameworks that facilitate cross-border knowledge exchange, joint research initiatives, and coordinated advocacy efforts. Through these partnerships, we can leverage global expertise, harmonize approaches, and address disparities in RD care and research. By strengthening national and international ties, we not only enhance our capacity to tackle RD but also contribute to a more unified and impactful global RD community. This collaborative approach ensures that we are better equipped to address the needs of patients, support groundbreaking research, and drive meaningful improvements in RD care worldwide.

What we hope to achieve:

GOAL 4.1: Canada will have a unified national approach to RDs that drives innovation and improves care for all affected by RDs		
Why?	**Over the next 5 years, we will:**	**Our work will result in:**
Despite growing efforts, Canada lacks a cohesive national strategy for RDs. The current landscape is fragmented, with care, research, and support initiatives varying widely across provinces and territories. This inconsistency creates inequities in access to care and hinders the adoption of innovative solutions. A unified, national approach is essential to streamline efforts, enhance collaboration, and ensure that all RD patients benefit from the latest advancements in research, diagnostics, and care, regardless of where they live.	i. Foster collaboration among key interest holders – including RD research institutions, healthcare providers, patient organizations, government agencies, and other relevant partners - to advance a cohesive, national RD framework that integrates research and care. ii. Develop and promote national, cross-disciplinary initiatives and policies that address key RD challenges, promote best practices, and ensure equitable access to care across the country.	A national, comprehensive RD framework that integrates research and care, enhancing coordination; More equitable and consistent care across Canada, ensuring patients have access to the same high-quality diagnostics, treatments, and support, regardless of their location; Accelerated innovation, faster adoption of best practices, improved efficiency and effectiveness of RD initiatives, leading to

	iii.	Promote national data sharing and infrastructure alignment to enable better coordination.	better resource utilization, enhanced patient experiences, and greater impact.

GOAL 4.2: Canada will be recognized as a key global player in RD research, innovation, and knowledge exchange, benefiting patients worldwide			
Why?	**Over the next 5 years, we will:**		**Our work will result in:**
RDs affect small and dispersed patient populations, making it difficult for any single country to generate the necessary evidence, develop treatments, or implement best practices on its own. Global collaboration is essential to overcome these challenges by enabling the exchange of knowledge, data, and expertise. By forging strong international partnerships, Canada can contribute to and benefit from global advancements, accelerating RD research, improving access to cutting-edge therapies, and enhancing care for patients at home and abroad.	i.	Forge global partnerships with and contribute to major global RD initiatives and policy dialogues to foster cross-border knowledge exchange and best practices, s.	Stronger global partnerships and knowledge exchange, leading to collaborative initiatives, shared adoption of best practices, and more innovation; Accelerated development and access to diagnostic tools, treatments, and care models, enhancing patient outcomes in Canada; Increased global visibility and influence for Canada, positioning the country as a key partner in RD research, care, and policy.
	ii.	Promote Canadian RD research, data, and innovations on the global stage.	

Cross-cutting themes

Patient Partnership and Co-Creation

Patient partnership and co-creation are fundamental to CRDN's approach to improving the lives of those affected by rare diseases and we will continue to explore the best ways to formalize and expand these partnerships. By involving patients and caregivers as active collaborators in all aspects of the strategic plan—whether it's in research, care, or policy development—we ensure that their lived experiences and unique insights are central to shaping solutions. However, we also acknowledge the need to find the best methods of engagement to ensure that participation does not add an undue burden on patients and caregivers. By collaborating with rare disease organizations, we aim to leverage existing networks and platforms, ensuring that patient partnership is both effective, reciprocal, and sustainable.

Education and Capacity Building

Education and capacity building are key to empowering the rare disease community, ensuring that both healthcare professionals and individuals living with rare diseases are equipped with the knowledge and tools needed to navigate complex healthcare systems. CRDN recognizes the need to foster ongoing learning and professional development across sectors to build a more informed, responsive healthcare environment. Through collaborative efforts and innovative educational initiatives, we strive to ensure that knowledge is shared, skills are enhanced, and that individuals in the rare disease ecosystem are prepared to contribute to advancing care, research, and advocacy.

Equity, Diversity, Inclusion, and Accessibility

Equity, Diversity, Inclusion, and Accessibility (EDIA) are fundamental to CRDN's vision for creating an inclusive and equitable healthcare and research environment for all those affected by rare diseases. The diversity of the rare disease community, including patients, caregivers, researchers, and healthcare providers, demands a commitment to creating an environment that is not only accessible but also inclusive of different backgrounds, experiences, and needs. Our efforts to advance equity, diversity, inclusion, and accessibility will be embedded in all our initiatives. As we move forward, we will engage with diverse voices and partners to co-create solutions that address inequities. This includes considering social determinants of health, such as socioeconomic status, race, gender, geographic location, and language, to ensure that all individuals, regardless of their circumstances, have the resources, care, and opportunities they deserve.

Data and Technologies

Digital technologies, data, and emerging tools are essential enablers across CRDN's strategic pillars. These innovations hold the potential to transform how we diagnose rare diseases, develop therapies, and provide care. Whether through telemedicine, mobile apps, data sharing platforms, or advance in genomic sequencing and artificial intelligence (AI), these technologies can enhance collaboration and improve equitable access to care, while supporting patient empowerment through online platforms and wearable technologies. As we build and implement our strategic plan, we recognize that digital solutions will undoubtedly shape our efforts to drive progress and create a more connected, supportive, and inclusive rare disease ecosystem.

Knowledge Mobilization and Impact Assessment

Knowledge mobilization (KM) plays a critical role in the CRDN's strategic plan by ensuring that the insights and findings from research, clinical trials, and community experiences are actively disseminated and applied to improve the care and outcomes for people affected by RDs. By fostering a culture of sharing knowledge, we can bridge the gap between research and practice, ensuring that the latest scientific advancements and innovative practices reach those who need them most. Equally important is the systematic assessment of the impact of CRDN's initiatives. By tracking and evaluating the outcomes of our efforts, we will ensure that our strategies are not only effective but also responsive to the evolving needs of the RD community. This process will help us identify areas of success, recognize challenges, and make adjustments where necessary to maximize our collective impact.

MOVING FORWARD

The CRDN's strategic plan is a product of a collaborative effort, crafted by diverse input from those deeply invested in advancing RD care and research. It is a roadmap born out of the collective vision of those who are most directly impacted—patients, caregivers, healthcare providers, and researchers. The plan's core focus is to drive innovation and improve the quality of care for those living with RD, ensuring they are empowered to live their fullest potential.

As can be seen from this first iteration of the CRDN's strategic plan, it is clear that there is much work to be done. The implementation of this ambitious plan is already underway and will unfold in stages. Some goals and strategies can be achieved relatively quickly by leveraging existing resources and scaling proven solutions, while others will require more time and long-term investment and be subject to funding. Regardless of the pace, CRDN is committed to continuously engaging with the RD community, including patients, healthcare providers, researchers, and relevant organizations, to maximize the impact of our efforts.

By uniting leaders and communities, CRDN will foster connections, synergies, and collaboration across the rare disease space. This network approach has already driven positive outcomes, such as partnerships, resource sharing, and more. Moving forward, this will further enhance our ability to address the unique challenges faced by the rare disease community and ensure that our strategic plan remains aligned with their needs and flexible in responding to emerging developments in healthcare, research, and policy. As the landscape of rare disease care and research evolves, CRDN will remain agile, keeping the voice of individuals with lived/living experience at the heart of every decision. This adaptability will be crucial to navigating the rapid pace of change in the health and social care sectors.

By implementing this strategic plan, we anticipate significant improvements in diagnostics, treatment accessibility, and overall patient care. By enhancing research capabilities, fostering stronger national and global collaborations, and promoting innovative therapies, we aim to create a more integrated and effective RD research-to-care framework in Canada. These improvements will not only benefit patients and their families but will also enhance the broader healthcare system's capacity to address the challenges posed by rare diseases. Ultimately, the success of this strategic plan will be measured by its ability to create lasting, tangible improvements in the lives of those affected by RD. By working together with the RD community and building on our shared vision, we will contribute to a more inclusive, effective, and sustainable rare disease ecosystem in Canada.

WITH GRATITUDE TO THE RARE DISEASE COMMUNITY

This strategy represents a collective vision shaped by the insights, experiences, and unwavering commitment of Canada's RD community. We extend our deepest gratitude to all who have contributed throughout the development of this plan, by sharing ideas, participating in extensive engagement sessions, online surveys, or contributing feedback on drafts along the way. Your voices guided our priorities. Your experiences grounded our understanding. Your leadership will make this strategy possible. A special thank you to our Pillar Group and Steering Committee members who come from across Canada and bring together a range of expertise and experience to drive forward our work (more details about the below members can be found on CRDN's website).

While this strategy is an important milestone, we recognize that it is only the beginning. Our continued progress will depend on the same spirit of collaboration and partnership that brought this work to life. Together, we are driving meaningful change—for today and for generations to come.

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