European Rare Diseases
Research Alliance

Resource on the State of the Art of Rare Disease Activities

2025 Report for Canada

National Mirror Group

Canada has a National Mirror Group (NMG), which was launched on 23rd May 2024. The NMG is coordinated by Maternal Infant Child and Youth Research Network (MICYRN), in partnership with the Canadian Rare Disease Network (CRDN). This NMG has overseen the collection of data from Canada via a data contributing committee. Contributors are listed at the end of this report.

Definition of a Rare Disease

Canada does not have a formal definition of a rare disease but commonly uses the definition: a rare disease affects fewer than 1 in 2,000 people. This definition is adopted by the Canadian Organization for Rare Disorders (CORD).

Status Quo of any National Plan or Strategy for Rare Disease

What is the status quo?

Canada does not have, nor has it ever had, a National Plan or Strategy for rare disease. However, Canada has implemented a National Strategy for Drugs for Rare Diseases focused on improving coverage of rare disease drugs under public drug plans, screening and diagnostics activities, collection and use of data and evidence in decision-making, and research. While focused on several objectives specific to drugs and reimbursement decision-making, the strategy does not cover activities along the full continuum of care that is inclusive of wrap around patient services, social supports, rare disease education and awareness and other aspects relevant for a fully integrated strategy.

Whilst not specific to rare diseases, the Canadian regulatory body offers several pathways that can support market approval for rare disease drugs including pre-submission guidance, accelerated review pathways, aligned regulatory and health technology assessment reviews, and international collaboration and use of parallel and international reviews.

There are also examples of national plans that, whilst not specific to rare diseases, can greatly impact research and innovation in the rare disease context such as the pan-Canadian Genomics Strategy,

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and the Disruptive Technology Solutions for Cell and Gene Therapy Challenge Program. In parallel, several national and regional initiatives have been developed in recent years to address the specific needs within the rare disease community. These initiatives are summarised below:

- National Strategy for Drugs for Rare Diseases

In March 2023, the Government of Canada launched the first phase of the National Strategy for Drugs for Rare Diseases (2023 to 2027), committing $1.5 billion to improve access to, and affordability of, new and existing rare disease drugs. The strategy focuses on four key areas:

1. Seek national consistency: $1.4 billion over three years has been provided to provinces and territories through bilateral agreements to support access to new drugs, support enhanced access to existing drugs, and enhance screening and diagnostics activities. Bilateral agreements have been signed with all 13 provinces and territories. Additional funds have been allocated to support access among First Nations and Inuit populations through Indigenous Services Canada programming.
2. Support patient outcomes and sustainability: Investment of $16 million is supporting national governance structures, such as a Health Canada Secretariat, Federal, Provincial, and Territorial government working group, and an Implementation Advisory Group, a multi-stakeholder group that provides advice to Health Canada and serves as a forum to exchange information and best practices.
3. Collect and use evidence: Canada's Drug Agency (CDA) and the Canadian Institute for Health Information (CIHI) are leading several activities to improve the collection and use of evidence to support decision-making, including (1) consolidating information for public drug plans on emerging Drugs for Rare Diseases entering Canada, (2) building guidance for consistent rare disease screening for use by provinces and territories, (3) conducting customised health technology assessments, (4) building standards, best practices, and capacity among rare disease registries, (5) exploring data sources and analytic work specific to Drugs for Rare Diseases outcomes to support use in decision making on listing and reimbursement.
4. Invest in innovation: $32 million (over 5 years) has been allocated to the Canadian Institutes of Health Research (CIHR) to advance rare disease research, focusing on better diagnostic tools, methods for tracking rare diseases, readiness for clinical trials specific to gene therapies, and the establishment of a Canadian rare disease clinical trials and treatment network (called RareKidsCAN).

- Quebec Policy and Action Plan for Rare Diseases

In June 2022, Quebec became the first province in Canada to adopt a formal policy on rare diseases, followed by the Quebec Action Plan for Rare Diseases 2023–2027, formally known as the Plan d'action québécois sur les maladies rares 2023–2027. The plan outlines actions in three main areas:

1. Increasing awareness and training among healthcare professionals
2. Facilitating equitable access to diagnosis, care, and services
3. Promoting research, innovation, and data collection

The Quebec government allocated $17.2 million in 2023–2024 to support implementation. This integrated and government backed plan represents a major advancement in the coordination of rare disease care and services at the provincial level and Quebec will use the federal contribution from the National Strategy for Drugs for Rare Diseases to support the Quebec Action Plan for Rare Diseases.

Other notable initiatives include:

- In 2015, the Canadian Organization for Rare Disorders (CORD), an umbrella patient organisation, released Canada's Rare Disease Strategy, a patient-led roadmap outlining five pillars for national action: improving early detection and prevention; providing timely, equitable and evidence-informed care; enhancing community support; providing sustainable access to promising therapies; promoting innovative research. Whilst widely endorsed by the rare disease community, it has not been formally adopted by any level of government.
- In 2022, the Pan-Canadian Pharmaceutical Alliance (pCPA) introduced a Rare Disease Drug Framework to guide consistent negotiations and access to high-cost therapies across provinces and territories.
- Launched in early 2024, the Canadian Rare Disease Network (CRDN) is a national initiative that brings together the leading clinical, scientific, and patient expertise to advance coordinated rare disease research, care, and innovation across the country.

Rare Disease Research Programmes and Funding

Canada's National Strategy for Drugs for Rare Diseases includes a research component, which is led by the Canadian Institutes of Health Research (CIHR). As part of the National Strategy for Drugs for Rare Diseases, the Canadian Institutes of Health Research (CIHR) has invested $32 million over five years (2023-2028) to support:

- A paediatric clinical trial and treatment network (RareKidsCAN)
- Projects to determine the prevalence, direct cost, and burden of rare diseases in the Canadian health care systems
- Projects on improving the use of genomic testing and determining the best pathway to diagnosis
- Grants focused on increasing the readiness for clinical trials specific to gene therapies for rare diseases (in partnership with the National Research Council of Canada)

Additionally, Canada's National Strategy for Drugs for Rare Diseases also addresses:

- National RD research investment, opportunities and funding calls
- Registries or registry catalogues for rare diseases
- Ontologies, codification or data standardisation
- Diagnostics research ('solving unknown conditions')
- Clinical/Translational research

Canada does have specific research programme/funding calls dedicated to rare disease, which are funded by both public and private bodies. These funding calls are not regular but sporadic. The Canadian Institutes of Health Research (CIHR), Genome Canada and Canada's Drug Agency provide specific funding for rare disease research in Canada. Additionally, the National Research Council (NRC) of Canada funds rare disease research via the Disruptive Technology Solutions for Cell and Gene Therapy Program, which is run by the Human Health Therapeutics (HHT) Research Centre (RC) of the NRC. This program will end on 31st March 2026. However, the HHT RC will continue to promote rare disease research within the RC's research mandate.

In Canada, rare disease research is funded through a combination of federal, provincial, philanthropic, and community-based sources. The Canadian Institutes of Health Research (CIHR) supports RD-specific projects through targeted initiatives, while Genome Canada and regional genome centres (e.g., Genome Québec, Ontario Genomics) fund genomics-based rare disease programs. Provincially, the Fonds de recherche du Québec – Santé (FRQS) supports rare disease research and infrastructure and funds the RARE.Qc network. Philanthropic and community organisations such as Brain Canada Foundation, Azrieli Foundation, and Kids Brain Health Foundation fund discovery and translational studies, while disease-specific charities directly support small research grants, biobanking, and patient registries.

In 2023, through Canada's National Strategy for Drugs for Rare Diseases, funding of $32 million was granted to The Canadian Institutes of Health Research (CIHR) over five years to advance rare disease research in Canada, and ultimately to improve the lives of patients living with rare diseases and their families.

Rare Disease Registration and Biobanking

Canada has a national registry for rare disease, known as the Connect Registry, which sits under the All for One Connect Governance Framework (2023). The registry is hosted by the Children's Hospital of Eastern Ontario Research Institute (CHEO-RI). $35 million was invested by Genome Canada to build regional genomics capacity, promote the equitable and ethical uptake of precision health tools, and address barriers to genomic data sharing. This registry aims to link individuals with a rare disease to approved research opportunities that may be of interest to them. These may include natural history studies, gene discovery programs, clinical trials using emerging therapeutics, or evaluation of new clinical workflows. Researchers can submit a proposal to access data, which is then reviewed by a committee. The registry is currently only available in two provinces in Canada. Further information is available here.

Canada does have disease-specific registries for rare conditions. The Canada Drug Agency (CDA) has created a comprehensive, live, inventory of rare disease registries in Canada which is searchable and continuously updated. The CDA conducted a study in 2024 which found that there are 66 rare disease registries in Canada. About half of these registries collaborated with international partners and networks. Electronic medical records (68%), clinician-reported data (68%), and medical chart abstraction (60%) are the most common sources of data collected in rare disease registries in Canada. Most rare disease registries in Canada collect clinical data (95%), laboratory and diagnostic data (85%), health outcomes data (83%), and treatment data (78%). Less commonly collected are patient-

generated data (55%) and caregiver data (15%). Canada also participates in international registry collaborations, which are important for ultra-rare disorders where national registries may not be feasible. Each registry may use different coding systems and are not consistent across Canada. There are elements of national approaches but there is not one consistent standard.

Some rare disease registries in Canada embed FAIR (FAIR stands for Findable, Accessible, Interoperable, Reusable) data principles. ICD10 coding is used within registries but codes vary depending on province and healthcare system used. The Human Phenotype Ontology (HPO) is used by Canada's national registry for rare disease, the Connect Registry, but its use is currently not standardised across the country.

Canada does not have a national or regional biobank for rare Disease. Canada has several biobanks that collect and store biosamples relevant to rare diseases, but these efforts are currently fragmented across institutions and disease areas rather than coordinated through a single national infrastructure. National initiatives such as the Canadian Tissue Repository Network (CTRNet) provide frameworks and capacity for biobanking and data linkage. Disease-specific initiatives such as the Neuromuscular Disease Network for Canada (NMD4C) and metabolic disease programs at major paediatric centres maintain specialised collections of samples.

Newborn Screening

In Canada, Newborn Screening (NBS) is organised at the provincial and territorial level. Each jurisdiction decides which conditions to include, although there is coordination through federal–provincial/territorial committees and expert bodies such as the Canada's Drug Agency (CDA). There are eight newborn screening programs in Canada, some programs cover multiple jurisdictions. The current programs include:

- The Alberta Newborn Screening Program, which provides screening for Alberta, the Northwest Territories, and Nunavut (Kitikmeot)
- Newborn Screening BC, which provides screening for British Columbia and Yukon
- The Manitoba Newborn Screening Program which provides screening for Manitoba and Nunavut (Kivalliq)
- The Maritime Newborn Screening Program, which provides screening for New Brunswick, Nova Scotia, and Prince Edward Island
- The Newborn Screening Program in Newfoundland and Labrador
- Newborn Screening Ontario, which provides screening for Ontario and Nunavut (Qikiqtaaluk)
- Quebec's Neonatal Blood and Urine Screening Program
- Saskatchewan's Universal Newborn Screening Program

The number of conditions screened for by provinces and territories are:

- Alberta (AB) - 25
- British Columbia (BC) - 27
- Manitoba (MB) - 28
- New Brunswick (NB) - 26

- Newfoundland and Labrador (NL) - 23
- Nova Scotia (NS) - 26
- Ontario (ON) - 29
- Prince Edward Island (PE) - 26
- Quebec (QC) - 15
- Saskatchewan (SK) - 28
- Nunavut (NU) (Qikiqtaaluk – Qik) - 29
- Nunavut (NU) (Kitikmeot – Kit) - 27
- Nunavut (NU) (Kivalliq – Kiv) - 26
- Yukon Territories (YT) – 25
- Northwest Territories (NT) – 24

The process for adding a new condition typically begins with a nomination from clinicians, researchers, or patient advocacy groups. These nominations are reviewed by provincial NBS advisory committees. When evaluating whether to add a condition, provinces consider criteria such as the availability of effective treatment, the strength of evidence that early detection improves outcomes, the accuracy and feasibility of the test, the seriousness of the disease if left undetected, and the healthcare system's ability to provide follow-up and treatment. Once a provincial NBS advisory committee makes a recommendation, a request for funding is submitted to the provincial Ministry of Health. The funding for national coordination of NBS is currently being provided through the National Strategy for Drugs for Rare Diseases. This funding is being provided until 2027. It is unclear what the path or funding support will be for NBS after 2027.

Canadian provinces and territories regularly look at international evidence and precedents when deciding whether to add new conditions to their NBS programmes. For example, Canadian experts and ministries of health often review outcomes from the U.S. Recommended Uniform Screening Panel (RUSP) and programs in the U.K. or Europe when evaluating rare disease screening.

Diagnostics

Canada does not currently have a single, comprehensive, national registry of laboratories providing genetic testing for rare diseases. Information is fragmented across provincial and institutional directories. Ontario maintains a detailed Genetic Test Directory covering provincially funded tests, whilst provinces like Alberta and British Columbia list their own publicly available tests through health service websites. The NIH Genetic Testing Registry (GTR) also includes many Canadian laboratories, although participation is voluntary and incomplete. Private providers such as LifeLabs Genetics offer additional testing options, and some information can be found through professional resources like Genetics Education Canada (GEC-KO).

Reimbursement for genetic tests are largely provincially governed, as healthcare delivery is a provincial responsibility under the public system. Most clinically indicated genetic tests for rare diseases are covered when ordered through publicly funded hospitals or accredited laboratories, but coverage policies vary by province and by the type of test (e.g. single-gene, gene panels, exome sequencing). Some provinces have specific funding programs for high-cost or specialised tests, while private insurance companies may cover additional tests not included in provincial plans. However, many tests

such as Whole Exome Sequencing (WES) and Whole Genome Sequencing (WGS) are not widely adopted or covered. This leads to further variation between provinces. If the test is not approved/funded, patients can either pay privately or sometimes it may be included in academic research studies through the investigator.

Links to province specific genetic testing programmes are shown below:

- Ontario
- British Columbia
- Alberta
- Saskatchewan
- Manitoba
- Quebec

In Canada, there are no formal nationwide agreements with other countries specifically for cross-border genetic testing of rare diseases. Canadian provinces primarily rely on domestic accredited laboratories for clinically indicated genetic tests. However, when a test is not available in Canada or is highly specialised, clinicians may request testing through international laboratories on a case-by-case basis, typically via hospitals or university-affiliated partnerships, research collaborations, or specialised programs. Canada's National Strategy for Drugs for Rare Disease and related provincial policies acknowledge the importance of international collaboration and access to global expertise, particularly for ultra-rare conditions, but they do not include formalised mechanisms for cross-border testing.

In Canada, there are several strategies in place to address patients with undiagnosed or currently undiagnosable conditions, though there is no single, formalised national programme for all undiagnosed patients. The Canadian healthcare system primarily identifies and supports undiagnosed patients through specialised clinical and research programs (e.g. Care4Rare). Canadian clinicians and researchers also engage in global collaborations such as Undiagnosed Diseases Network International (UDNI) and the International Rare Disease Research Consortium (IRDiRC), which facilitates cross-border clinical and genomic data sharing, case studies, and access to international expertise to help identify undiagnosed cases.

There is a policy in place to provide genetic counselling for patients with suspected or confirmed rare disease in Canada. Genetic counselling is provided through provincial programs, typically located at academic or paediatric hospitals.

National Alliances of Rare Disease Patient Organisations

Canada does have a national alliance for rare disease patient organisations, which is the Canadian Organisation for Rare Disorders (CORD). Additionally, the Regroupement québécois des maladies orphelines (RQMO) is a strong provincial organisation for Quebec (French speaking). Canada's national alliance for rare disease is involved in Canada's National Mirror Group (NMG). They are also involved in setting the strategic direction for rare disease research by being actively involved in advocacy work and national strategy development. They provide webinars and other forms of learning and capacity-building on patient engagement in rare disease research, further information is available here.

Information Resources for Rare Disease

Canada does not have a government funded national helpline for rare disease but there are national and provincial resources that provide information, guidance, and referral support including iRARE centres. They help people with a lived experience of a rare disease navigate the healthcare system, connect individuals to specialists or clinical trials, and link families to patient organisations. They provide education, resources, and psychosocial support, while also contributing to registries and research initiatives to strengthen Canada's rare disease knowledge base.

Training and Education

In Canada, there are multiple entities that provide education and training materials about rare disease but there are no nationally recognised bodies that coordinate training on rare disease. Individual organisations offer some training opportunities on rare disease including but not limited to: Care4Rare, Canadian Rare Disease Network (CRDN), Canadian Organisation for Rare Disorders (CORD), RareKids-CAN, The Maternal Infant Child and Youth Research Network (MICYRN), Genome Canada, and iRare centres. There are currently no dedicated university programs or full courses in Canada solely focused on rare disease. However, aspects of rare disease are integrated into broader curricula in medical genetics, genetic counselling, biomedical sciences, and health sciences programs at several universities. Additionally, research-focused fellowships and professional development opportunities are available through some networks.

Orphan Medicinal Products (OMPs)

There are eight OMPs available in Canada:

- Poteligeo (Mycosis fungoides or Sézary syndrome)
- Oxlumo (Primary hyperoxaluria type 1)
- Epkinly (Relapsed or refractory diffuse large B-cell lymphoma)
- Welireg (Von Hippel-Lindau disease)
- Yescarta (Follicular lymphoma; 2nd line treatment of diffuse large B-cell lymphoma or high-grade large B-cell lymphoma)
- Koselugo (Neurofibromatosis type 1)
- Sohonos (Fibrodysplasia ossificans progressiva (FOP))
- Imcivree (Bardet-Biedl syndrome)

Canada does have Early Access Programmes/Expanded Access programmes to help patients access medicinal products for rare conditions. Health Canada Special Access Program (SAP) provides a regulatory pathway that allows healthcare practitioners to request access to drugs, including pharmaceuticals, biologics, and radiopharmaceuticals, that are not authorised for sale in Canada for patients with serious or life-threatening conditions when conventional therapies have failed, are

unsuitable, or unavailable. While the SAP is not intended to replace clinical trials or bypass regulatory review, it serves as a compassionate, case-by-case mechanism, to bridge the gap for patients with rare diseases who lack approved treatment options in Canada. To use the program, a physician must initiate the request, justify the need with clinical evidence, confirm the manufacturer's willingness to supply the product, and follow all safety and reporting requirements. Although the SAP has facilitated access to investigational or unlicensed drugs for some rare disease patients, its limitations include case-by-case approval, potential manufacturer refusal, and possible delays or denials. Overall, the SAP provides a vital but restricted route for accessing orphan or rare disease medicines in exceptional circumstances within the Canadian healthcare system.

Canada is not involved in any formal efforts or initiatives with other countries to support access to medicines and therapies for rare disease.

After approval, OMPs undergo HTA assessment by Canada's Drug Agency (CDA) and Institut national d'excellence en santé et services sociaux (INESSS). The recommendation is provided to each of the provinces who decide reimbursement, and prices are negotiated through the pan-Canadian Pharma Alliance. The final funding decision is made by each province and territories, meaning drugs may be reimbursed in some provinces and territories but not others. This results in delays in access as well as major gaps in access for patients and limited transparency for caregivers and providers.

ERDERA European Rare Diseases Research Alliance

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