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CADTH Health Technology Review

An Inventory of Rare Disease Registries in the Canadian Landscape

Environmental Scan

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Abbreviations

HTA	health technology assessment
RDR	rare disease registry

Key Messages

What Was the Question?

There is a lack of centralized information about rare disease registries (RDRs) in Canada. To address this, we created an inventory of RDRs in Canada and international RDRs that include patients living in Canada.

What Did We Do?

Information about RDRs was identified through a search of published literature, grey literature, consultations with members of the rare disease community, and a survey with the registry holders.

What Did We Find?

We identified 148 RDRs, of which 66 are RDRs in Canada and 82 international RDRs.

- In total, 21% of RDRs in Canada and 11% of international RDRs capture rare cancer(s).
- About a half of RDRs in Canada (53%) and international RDRs (46%) collaborate with other national or international registries or networks.
- Most RDRs in Canada (73%) and international RDRs (86%) identify patients for RDR enrollment through health care provider referrals.
- Electronic medical records (68%), clinician-reported data (68%), and medical chart abstraction (60%) are the most common sources of data for RDRs in Canada compared to international RDRs, for which patient (79%) and caregiver (61%) surveys are most common.
- Most RDRs in Canada collect clinical data (95%), laboratory and diagnostic data (85%), health outcomes data (83%), and treatment data (78%), and less commonly patient-generated data (55%) and caregiver data (15%).

What Does This Mean?

The information in the inventory will help guide future initiatives for improving the RDR landscape in Canada. Most RDRs in Canada source data from electronic health records, clinician-reported data, and medical charts. Fewer RDRs in Canada implement patient and caregiver surveys. RDRs in Canada include information relevant to decision-makers as most collect clinical data, health outcomes data, and treatment data, and about half collect health resource utilization data. This inventory will support future initiatives to assess the suitability of RDRs for generating decision-grade real-world evidence.

Background

In 2023, the Federal Government of Canada launched the National Strategy for Drugs for Rare Diseases to help increase access to affordable and effective therapies for people with rare diseases.[1] People with rare diseases often have limited or no treatment options, and conducting clinical trials in this area is inherently

challenging. This can result in clinical uncertainty and further delays in accessing new and emerging therapies.

There is growing recognition in Canada and globally that real-world data and real-world evidence can play a significant role in optimizing both regulatory and reimbursement decision-making.[2] Rare disease registries (RDRs) have the potential to provide valuable real-world data and can complement existing regulatory and health technology assessment (HTA) frameworks. Registries can help characterize natural history, disease progression, and patient experience, as well as evaluate emerging technologies.[3]

Despite their importance, Canada currently lacks a centralized and comprehensive inventory of RDRs, including both registries in Canada and international registries with participants who live in Canada. While other registry directories exist, there is no inventory designed specifically to inform regulatory, HTA, and payer needs.

We developed an inventory of RDRs with the goal of capturing registries that collect data that have the potential for generating decision-grade real-world evidence to answer specific questions (e.g., natural history, postmarket therapy assessment). Our inventory captures additional elements, including those required for a preliminary assessment of registry capabilities (e.g., geographical coverage, sample size, data types, data sources). This is part of ongoing initiatives at our organization to better understand the RDR landscape in Canada, which will support future initiatives to assess and improve registry quality for regulatory and reimbursement decision-making.

Methods

CADTH developed the inventory of RDRs in Canada and international RDRs that include patients living in Canada in 2 phases. In phase 1, RDRs were identified through a rare disease–specific grey literature search, consultations with members of the rare disease community, and our Spring 2024 open call funding opportunity. This search was complemented by a search of disease registries in published literature and grey literature, which were further categorized into RDRs. In phase 2, the registry holders identified in phase 1 were invited to complete a survey to validate and supplement the phase 1 inventory information.

Phase 1: Identification of RDRs

RDRs were identified through several approaches, including a grey literature search using the terms *rare disease*, *registry*, *observational research*, and *Canada* in collaboration with Case Market Access Consulting Inc. and COMPASS Medical Affairs Consulting Inc.; consultations with members of the rare disease community; and through our Spring 2024 open call funding opportunity. Additional RDRs were also identified in a comprehensive search of disease registries in published and grey literature with a focus on disease registries in Canada or international disease registries that include participants who live in Canada. Registries were included in the RDR inventory if they met the inclusion criteria specified in Table 1.

Phase 2: Survey of Registry Holders

To validate and supplement the information gathered in the phase 1 search, we created an online survey for registry holders that was contracted and conducted through Medlior Health Outcomes Research Ltd from March 20, 2024, to June 10, 2024 (Appendix 1). The registry holders were invited to complete the survey and at least 2 follow-up emails were sent to encourage participation. The registry holders were able to complete the survey independently online or with a research associate by telephone or virtual meeting. An honorarium or donation was provided on completion.

Synthesis Approach

The information in the inventory is compiled from both publicly available information and the survey of registry holders. Publicly available information was populated up to January 26, 2024, from RDR websites and publications about the RDRs and entered into an Excel database (refer to Appendix 2 for additional details).

The survey of registry holders validated information gathered from publicly available information. It also gathered supplemental information, including if patient or caregiver consent is required for registry enrollment, how patients are identified as potential candidates to participate in the registry, the type(s) of data collected, the source(s) of the data, if the data have ever been linked with external data sources (e.g., other registries, administrative data, biobanks), and if the registry collaborates with any national or international registries or networks (refer to Appendix 1 for additional details on the survey questions). Registries that capture multiple diseases were able to enter specific information for up to 10 registries.

Table 1: Inclusion and Exclusion Criteria for the Inventory of Rare Disease Registries

Category	Inclusion criteria	Exclusion criteria

Geographic coverage	• Either of the following:
	  ◦ registry is led by an organization or institution in Canada
	  ◦ registry captures data from at least 1 province or territory
		• Internationally based registries in which information about the number of patients in Canada and/or number of sites in Canada was not identified

Active status	• Participant enrolment in the registry is ongoing and assessed using publicly available information (e.g., website, clinical trial registration, or publication) • Patients can continue to self-register or be included with a referral from a health care provider	• Research studies (e.g., cohort studies) that are no longer enrolling new patients • Registries that were operational for time-limited periods but were no longer running as of January 2, 2024
Rare disease or condition	• Registry collects data on at least 1 rare disease, captured if listed in the NORD Rare Disease Database[5] (primary check) or the Orphanet Database[6] (secondary check)	• Registries that did not capture people with a rare disease

Category	Inclusion criteria	Exclusion criteria
Registry type	• Registry collects clinical, demographic, social, or other observational data related to a specific disease or condition[7]	• Databases that collect participant or caregiver contact information, but do not collect or link to other clinical or patient-reported data • Databases that collect information about specific treatments (e.g., medications, dialysis) or events (e.g., hospitalizations) but are not related to a specific disease • Databases of procedures (e.g., organ replacement, implants) or health technologies not related to a specific disease • Biobanks (i.e., hold biological samples) or databases that hold genomic information without associated demographic or clinical information related to a specific disease • Databases related to immunization, occupational exposures, accidents, births, deaths, or similar records not related to a specific disease

NORD = National Organization for Rare Disorders.

Findings

Summary

A total of 148 RDRs met our inclusion criteria for the inventory summary. Of these, 66 are RDRs hosted and operated only in Canada and 82 are classified as RDRs with patients living in Canada (refer to Appendix 3). Among the 66 RDRs in Canada, 40 (61%) completed the phase 2 survey; 28 (34%) of the 82 international RDR completed the phase 2 survey.

RDRs in Canada

Information Gathered From the Public Search and Survey of Registry Holders (n = 66)

Of the 66 RDRs in Canada, 85% (n = 56) reported on the number of patients, which ranged from 17 to 55,490 patients (Table 2). Rare cancer(s) are examined in 21% (n = 14) of RDRs in Canada. Among RDRs in Canada, 50% (n = 33) include pediatric and adult patients, 23% (n = 15) exclusively include pediatric patients, and 21% (n = 14) exclusively include adult patients (Table 2). Figure 1 presents the percentage of RDRs in Canada with representation in each province and territory, where 15% (n = 10) of RDRs reported capturing data in all provinces and territories. Most RDRs capture data from Ontario (71%), Quebec (55%), and Alberta (55%) (Figure 1).

Information Gathered From the Survey of Registry Holders (n = 40)

Among the 40 RDRs in Canada that completed the survey, 93% (n = 37) require patient or caregiver consent for registry enrollment, while 5% (n = 2) collect data under a waiver of consent (Table 2). About half of RDRs in Canada collaborate with national or international registries or networks (53%, n = 21) and have been

linked with external data sources such as other registries or administrative data (43%, n = 17) (Table 2). Most RDRs in Canada identify patients for registry participation through referrals from health care providers (73%), followed by identification from electronic medical records (33%) (Figure 2). The sources of data for RDRs in Canada vary, where electronic medical records (68%), clinician-reported data (68%), and medical chart abstraction (60%) are the most common (Figure 3). Most RDRs in Canada collect clinical data (95%), laboratory and diagnostic data (85%), health outcomes data (83%), and treatment data (78%) (Figure 4). Patient-generated data (55%) and caregiver data (15%) are less common (Figure 4).

International RDRs With Patients Living in Canada

Information Gathered From the Public Search and Survey of Registry Holders (n = 82)

Of the 82 international RDRs, 87% (n = 71) reported on the number of patients, which ranged from 30 to 88,832 total patients, while 52% (n = 43) listed information about the specific number of patients in Canada, which ranged from 2 to 5,600 patients (Table 2). Rare cancers are examined in 11% (n = 9) of international RDRs. Most international registries include both pediatric and adult patients (73%, n = 60), 12% (n = 10) collect data exclusively from adult patients, and 7% (n = 6) collect data exclusively among pediatric patients (Table 2).

Among international RDRs that reported coverage of jurisdictions in Canada (73%; n = 60), 11% (n = 9) capture information in all provinces and territories. International RDRs capture data from Ontario (60%), Quebec (48%), British Colombia (42%), Alberta (42%), Manitoba (30%), Nova Scotia (29%), Saskatchewan (25%), New Brunswick (23%), Newfoundland and Labrador (23%), Prince Edward Island (14%), Yukon (13%), Northwest Territories (13%), and Nunavut (11%).

Information Gathered From the Survey of Registry Holders (n = 28)

Among the 28 international RDRs that completed the survey, 100% require patient or caregiver consent for registry enrollment (Table 2). About half of international RDRs collaborate with national or international registries or networks (46%; n = 13) and 18% (n = 5) have been linked with external data sources such as other registries or biobanks (Table 2). Most international RDRs identify patients for registry participation through referrals from health care providers (86%), patient organizations (79%), and online self-registration (79%) (Figure 2). Most data are sourced from patient (79%) and caregiver (61%) surveys (Figure 3). Most commonly, international RDRs collect clinical data (100%), sociodemographic data (93%), health outcomes data (89%), laboratory and diagnostic data (79%), treatment data (75%), and patient-generated data (75%) (Figure 4).

Table 2: Characteristics of RDRs in Canada and International RDRs With Patients in Canada

Characteristic	RDRs in Canada (n = 66), n (%)	International RDRs with patients living in Canada (n = 82), n (%)	Overall RDRs (n = 148), n (%)
Information gathered from the literature search and survey of registry holders			
Number of patients living in Canada	Range: 17 to 55,490 56 (85%)	Range: 2 to 5,600 43 (52%)	Range: 2 to 55,490 99 (67%)
Number of patients living internationally	NA	Range: 30 to 88,832[b] 71 (87%)	NA
Registry captures patients with rare cancer(s)	14 (21%)	9 (11%)	23 (16%)
Population age group captured in this registry			
Both pediatric and adult patients	33 (50%)	60 (73%)	93 (63%)
Pediatric patients	15 (23%)	6 (7%)	21 (14%)
Adult patients	14 (21%)	10 (12%)	24 (16%)
No survey response or unclear from literature search	4 (6%)	6 (7%)	10 (7%)
Information gathered from the survey of registry holders			
Number of registries that completed survey	40 (61%)	28 (34%)	68 (46%)
Informed patient or caregiver consent required for registry enrollment[c]			
Yes	37 (93%)	28 (100%)	65 (96%)
No, data obtained under a waiver of consent	2 (5%)	0 (0%)	2 (3%)
Did not respond	1 (3%)	0 (0%)	1 (1%)
Registry collaborates with national or international registries or networks[c]			
Yes	21 (53%)	13 (46%)	34 (50%)
No	13 (33%)	11 (39%)	24 (35%)
Did not respond	6 (15%)	4 (14%)	10 (15%)
Registry has been linked with external data sources (e.g., other registries, biobanks)[c]			
Yes	17 (43%)	5 (18%)	22 (32%)
No	17 (43%)	17 (61%)	34 (50%)
Did not respond	6 (15%)	6 (21%)	12 (18%)

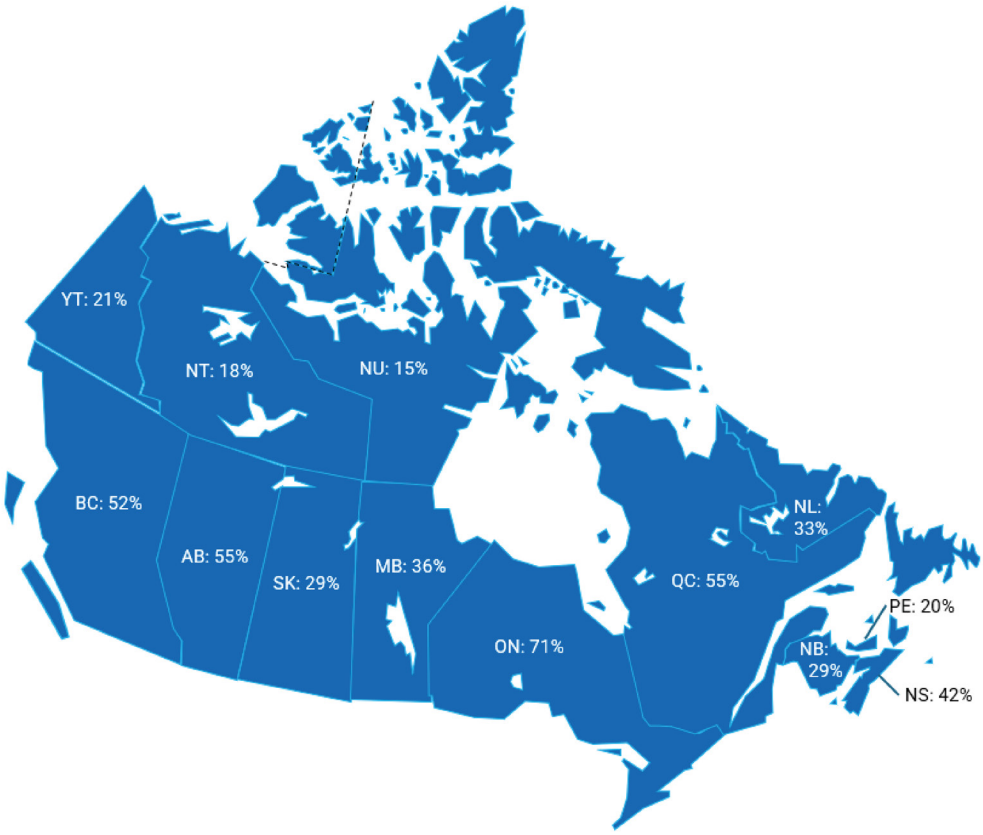
NA = not applicable; RDR = rare disease registry.

[a]Not applicable as registries in Canada only have patients living in Canada.

[b]The TREAT-NMD Global Registry Network reported collecting data on 88,832 patients with neuromuscular diseases across multiple countries, including 65 registries.

[c]Among registries that responded to the survey of registry holders (n = 40 RDRs in Canada; n = 28 international RDRs).

Figure 1: Percentage of RDRs With at Least 1 Patient in Each Province and Territory in Canada, Among RDRs in Canada (n = 68)

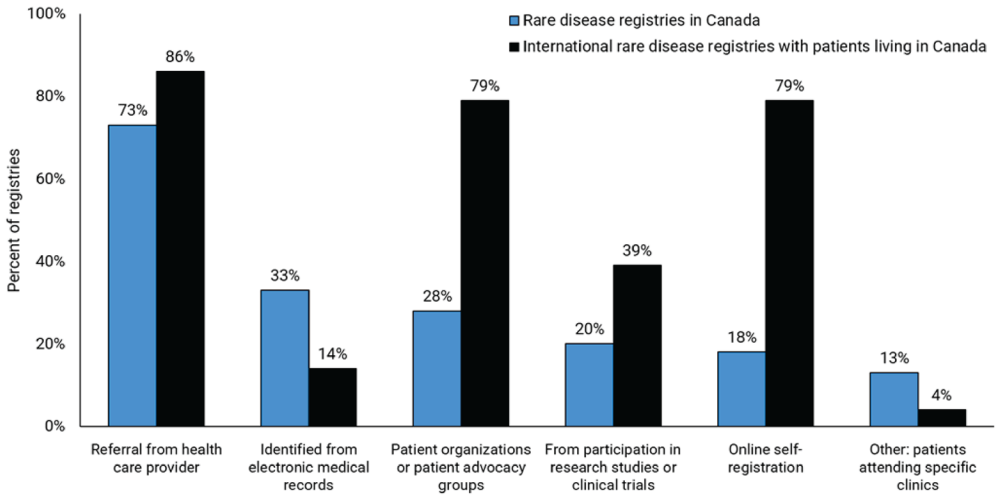


AB = Alberta; BC = British Colombia; MB = Manitoba; NB = New Brunswick; NL = Newfoundland and Labrador; NS = Nova Scotia; NT = Northwest Territories; NU = Nunavut; ON = Ontario; PE = Prince Edward Island; QC = Quebec; RDR = rare disease registry; SK = Saskatchewan; YT = Yukon.

Notes: This figure depicts the percentage of RDRs that have representation from each province and territory, not the percentage of sites per province or territory. It represents the percentage of RDRs that have at least 1 patient in each province and territory. For example, 52% of RDRs in Canada have at least 1 patient in British Columbia. The geographical coverage is presented for the overall registry; specific diseases within the registry may have differential provincial and territorial coverage. Registry holders were instructed to enter provinces and territories based on the primary residential address of patients who are currently enrolled. For example, a participant who lives in the Northwest Territories and travels to a clinic in Alberta would be included in the Northwest Territories group.

The number and percentage of registries with representation from each province and territory were, for all provinces and territories, n = 10 (15%); BC, n = 34 (52%); AB, n = 36 (55%); SK, n = 19 (29%); MB, n = 24 (36%); ON, n = 47 (71%); QC, n = 36 (55%); NB, n = 19 (29%); PE: n = 13 (20%); NS, n = 28 (42%); NL, n = 22 (33%); YT, n = 14 (21%); NT, n = 12 (18%); NU, n = 10 (15%); and registry holder did not answer survey or was unclear from the literature search, n = 9 (14%).

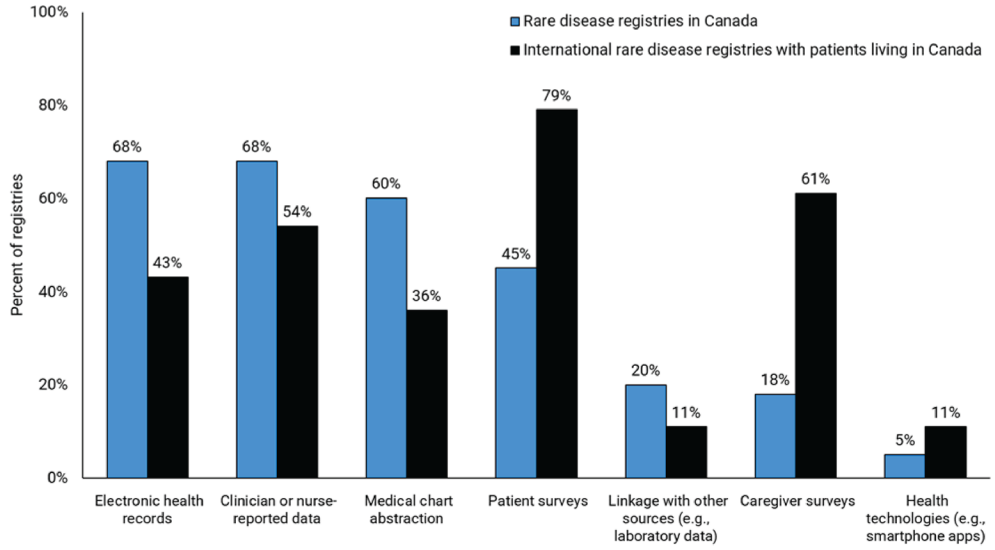
Figure 2: Method of Patient Identification for Participation in RDRs, RDRs in Canada (n = 40) and International RDRs With Patients Living in Canada (n = 28)



RDR = rare disease registry.

Notes: This information was obtained directly from a survey with the registry holders. Each registry could select multiple methods. One (3%) RDR from Canada did not respond to this question. Among registries that responded to employing "other" methods to identify patients for participation in the registry, 5 (13%) RDRs in Canada and 1 (4%) international registry with patients living in Canada responded that patients are identified to participate from specific clinics; 1 (3%) RDR in Canada identifies patients through collaborations with Statistics Canada and 1 (4%) of international RDR with patients living in Canada identifies patients through social media outreach.

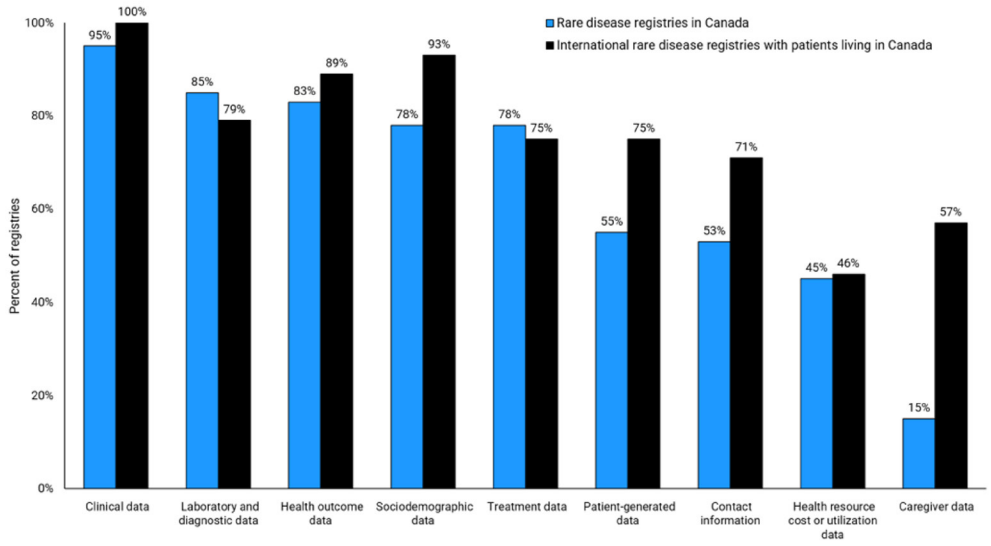
Figure 3: Source of Data, RDRs in Canada (n = 40) and International RDRs With Patients Living in Canada (n = 28)



RDR = rare disease registry.

Notes: This information was obtained directly from a survey with the registry holders. Each registry could select multiple sources. One (3%) RDR in Canada did not respond to this question.

Figure 4: Types of Data Collected, RDRs in Canada (n = 40) and International RDRs With Patients Living in Canada (n = 28)



OHIP = Ontario Health Insurance Plan; RDR = rare disease registry.

Notes: This information was obtained directly from a survey with the registry holders. Each registry could select multiple options. One (3%) RDR in Canada did not respond to this question. Among the registries that responded to collecting "other" types of data, 1 (3%) RDR in Canada reported collecting OHIP number for linkage, 1 (3%) RDR in Canada reported collecting occupational and environmental data, and 1 (4%) international RDR with patients living in Canada reported collecting behavioural data.

Strengths

This scan is the first centralized inventory of RDRs designed specifically to inform regulatory, HTA, and payer needs in support of the Government of Canada's National Strategy for Drugs for Rare Disease. The inventory includes both registries in Canada and international registries with participants who live in Canada. Information about RDRs was compiled using a comprehensive search strategy, including published literature, grey literature, consultation with the rare disease community, and a survey of the registry holders. This inventory includes key elements about RDRs that are important to decision-makers, such as coverage in Canada, sources of data, and types of data. Through this inventory, we have a better understanding of the potential capabilities of RDRs as sources of data for informing decision-making, particularly as more than 75% of RDRs collect clinical data, health outcomes data, and treatment data, and about half collect health resource utilization data. This inventory and future iterations of this inventory are well positioned to support the provinces and territories as they begin to sign bilateral agreements with the federal government to receive funds for cost-sharing new and emerging drugs for rare diseases.

Limitations

This scan provides an initial overview of RDRs in the landscape in Canada and has potential gaps. The number of RDRs in this inventory may be underestimated. Registries without prior publications, websites, or established portals for participant registration are not captured in this inventory nor are international registries that have patients in Canada but do not publicly report on geographical coverage. Although we identified additional RDRs through our spring 2024 open call funding opportunity, there may have been

additional RDRs that were not captured if they were not aware of the funding call or did not submit a letter of intent. The link to the survey for registry holders was limited to RDRs that were already on our list, which may have also reduced the number of RDRs in the inventory.

This inventory also excludes databases that collect contact information only or collect information about specific procedures or events, biobanks, and genomic datasets that are not related to at least 1 specific rare disease. It is possible these excluded databases collect disease information, although it is not publicly reported and not reflected in this inventory. In addition, registries that collect data on both rare and nonrare diseases may be excluded, unless the captured rare diseases were explicitly publicly mentioned.

Although there is no universal definition of a rare disease, for this inventory, a disease is considered rare if it is listed in the National Organization for Rare Disorders (NORD) Rare Disease Database or the Orphanet Database. However, there is variability in rare disease prevalence thresholds within these 2 databases, and prevalence estimates may be outdated or may not fully reflect the context in Canada. Given the heterogeneity of the rare disease landscape and challenges in estimating prevalence, other facets, such as unmet need and lack of or limits to access to therapeutics, should also be considered.

This inventory of RDRs relies on both publicly available information and information directly from registry holders. As a result, there is higher certainty in the inventory elements obtained from registry holders, relative to information from the literature and public search. The information gathered in the survey may not be fully representative of the RDR landscape as the response rate was 61% for RDRs in Canada and 34% for international RDRs. Although this inventory captures key elements of RDRs in Canada and international RDRs with patients living in Canada, additional information will be required to assess the quality and suitability of registries for specific HTA purposes.

Conclusion

This scan established an inventory of RDRs in Canada and internationally to better understand the rare disease registry landscape in Canada. This inventory will be periodically updated over time and the information captured may be expanded in the future to address the evolving needs of decision-makers. We plan to engage with health system partners to gather feedback about the initial inventory to ensure it remains a relevant and valuable resource for the rare disease community and decision-makers. The information captured in the inventory may help to inform future initiatives for improving the RDR landscape in Canada. RDRs in Canada include information relevant to decision-makers as most collect clinical data, health outcomes data, and treatment data, and about half collect health resource utilization data. There is a need for ongoing efforts to expand coverage in RDRs to capture patients with rare diseases across Canada. Most RDRs in Canada source data from electronic health records, clinician-reported data, and medical charts. Fewer RDRs in Canada employ patient and caregiver surveys, which could lead to gaps in data completeness and richness. This foundational work will support future initiatives to assess the suitability of registries for generating decision-grade real-world evidence.

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Appendix 1: Data Elements in Phase 2 Survey of Registry Holders

Note that this appendix has not been copy-edited.

Rare Disease Registry Inventory Survey Questions

1. Please enter the registry name.

[Free text]

2. Is this registry active as of March 2024?

Note: An active registry is defined as a registry where enrollment is open and new participants can continue to join.

Yes

No

3. Is this registry a Canadian or an international registry?

Note: Canadian registries are defined as registries that are hosted and operated in Canada.

Canadian

International

4. Please list the registry's lead institution or organization.

Note: The lead institution or organization is that which is responsible for hosting and storing registry data. You may list up to 2 lead institutions or organizations.

[Free text box 1]

[Free text box 2]

5. Please list the registry's lead contact person.

[Free text]

6. Please list the lead person's contact information for this registry.

[Free text box for information for lead person 1]

[Free text box for information for lead person 2]

7. How are patients identified as potential candidates to participate in this registry? Select all that apply.

Note: If uncertain, please select “other”.

- Referral from health care provider(s)
- Identification through electronic medical records
- Patient organization(s) or patient advocacy group(s)
- Online self-registration
- Through participation in existing research studies or clinical trials
- Other (please specify)

8. Is informed patient consent required for enrollment into this registry?

Note: Please answer “yes” even if only 1 data collection site requires patient consent.

Yes

No, data are collected under a waiver of consent

No, other (please specify)

Uncertain

9. What type(s) of data are collected within this registry? Select all that apply.

Note: If uncertain, please select “other”.

- Contact information (e.g., name, telephone)
- Sociodemographic data (e.g., age, gender, education, income)
- Clinical data (e.g., disease severity, medical history, medication history)
- Treatment data (e.g., treatment response, treatment adherence)
- Health outcome data (e.g., disease progression, mortality)
- Laboratory and diagnostic data (e.g., genetic tests, biomarkers)
- Health resource cost or utilization data (e.g., hospitalizations)
- Patient-generated data (e.g., patient-reported outcomes)
- Caregiver data
- Other (please specify)

10. How are data collected in this registry? Select all that apply.

- Electronic health records
- Medical chart abstraction
- Clinician-reported data
- Patient surveys

- Caregiver surveys
- Linkage with other sources (laboratory, drug utilization, and so forth)
- Health technologies (e.g., smartphone apps, wearable devices)
- Other (please specify)

11. Does this registry capture participants with rare cancer(s)?

Yes

No

12. List all rare disease(s) captured in this registry

Note: Please enter 1 disease per line. Enter the specific disease, not the disease group (e.g., enter "spinal muscular atrophy," not "neuromuscular disease"). If more than 10 diseases are captured in the registry, please list the most prevalent 10.

[Free text, where each disease is listed in a separate answer box]

13. Specify the population age group for each disease captured in this registry.

Note: Please specify the age at which follow-up with patients ceases, if applicable.

Pediatric population (< 18 years of age)

Adult population (≥ 18 years of age)

Both pediatric and adult populations

14. For each disease captured in this registry, specify the range of patient follow-up.

For example, patients with disease X are followed in this registry for 1 to 2 years.

[Free text, with a row for each disease, with a column for a lower and upper limit of follow-up range, and a dropdown tab for months and years]

For Canadian and international rare disease registries

15. For each disease captured in this registry as of February 2024, specify the total number of enrolled participants.

Note: Please specify the total number of enrolled participants, not the expected total.

[Free text, with a row for each disease]

For international rare disease registries only

16. For each disease captured in this registry as of February 2024, specify the total number of enrolled participants who identify as being from Canada.

Note: Please specify the total number of enrolled participants, not the expected total.

[Free text, with a row for each disease]

17. For each disease captured in this registry as of March 2024, check the box if you have any participants enrolled from each province and/or territory in Canada.

Note: Please specify if participants are currently enrolled in each province or territory, not based on whether participants are expected to be enrolled. If a participant travels from 1 province or territory to receive care in another province or territory, please base their listed province or territory on their primary residential address. For example, a participant who lives in the Northwest Territories and travels to a clinic in Alberta should be included in the Northwest Territories group.

Disease 1 [This question will be asked for each disease specified in question #12]

a) British Columbia

b) Alberta

c) Saskatchewan

d) Manitoba

e) Ontario

f) Quebec

g) New Brunswick

h) Prince Edward Island

i) Nova Scotia

j) Newfoundland and Labrador

k) Yukon

l) Northwest Territories

m) Nunavut

18. Have data from this registry ever been linked with external data sources (e.g., other national or international registries, administrative data, biobanks)?

Yes

No

Uncertain

If yes to question 18:

19. *Please specify the external data sources this registry has linked with. Select all that apply.*

a) Electronic health records

b) Health administrative databases

c) National health surveys

d) Public health surveillance systems

e) Other national or international registries

f) Biobanks or genetic databases

g) Clinical trial databases

h) Patient-reported data platforms

i) Other (please specify)

20. Does this registry collaborate with any national or international registries or networks?

Note: In this question, *collaborate* is defined as sharing aggregate or patient-level data or harmonizing data standards or elements, such as creating common or minimum datasets, cross-validating data, or working on joint research projects.

Yes

No

Uncertain

21. Would you be willing to update your responses to this survey annually?

Yes

No

Other (please specify)

22. If applicable, please specify any additional information you want to share about this registry or your survey responses.

[Free text: allow responders to leave this blank]

Appendix 2: Data Elements and Definitions Applied When Populating RDR Inventory With Information From the Literature and Public Search

Note that this appendix has not been copy-edited.

Table 3: Data Elements and Definitions Applied During Screening of Rare Disease Registries (Literature and Publicly Available Information)

Data element	Coding options	Data element definition
Registry name	[Free text]	Name of the registry If registry has multiple names, use the name most prominent on registry website or in logos.
Screening decision	Include Exclude Discuss	Decision if registry is to be included in the final inventory of rare disease registries. If unclear if registry should be included, reviewer codes as "Discuss" to prompt discussion among all reviewers.
Notes	[Free text]	Any notes about registry, including reason why reviewer coded screening decision as "Discuss."
Exclusion reason	NA Duplicate In development Not a registry Not rare Clinical trial registry No Canadian sites Unable to determine if any participants in Canada	Reason why registry is excluded: NA: use when registry meets inclusion criteria. Duplicate: registry is already included in the inventory. In development: new or pilot registry, which is not yet active, defined as registry without ethics approval and who has not begun patient recruitment. Not a registry: registries where patients are not defined by a particular disease, focus on procedures (e.g., organ replacement, dialysis), accidents (e.g., spinal cord injuries), occupational exposures (e.g., radiation), genomics (bioinformatic data only), track births or deaths; track immunizations, publications about need for registry, or a list of patients with a particular disease without any health outcomes collected. Not rare: not a rare disease, after searching NORD Rare Disease Database, Orphanet Database, or Canadian prevalence data. Clinical trial registry: public record system for registration in clinical trials. No Canadian sites: use for registries with no Canadian sites. Unable to determine any participants in Canada: use for registries where it is unclear how many participants or sites in Canada.
Does this registry capture participants with rare disease(s)?	Yes No Unclear	Indicate if registry collects data on at least 1 disease that is considered rare based on search of NORD Rare Disease Database, Orphanet Database, or Canadian prevalence data. Use unclear if unable to find any prevalence data.

Data element	Coding options	Data element definition
Rare disease classification	NORD EMA CA Unclear	Criteria used for coding rare disease(s) reflected in registry: NORD: disease listed in The NORD Rare Disease Database[5] based on FDA definition of rare disease.[8] EMA: disease listed in the Orphanet Rare Disease Database[6] based on EMA definition of rare disease.[9] CA: Internet scan for Canadian prevalence data that meets NORD or EMA definitions. Unclear: unable to find any prevalence data.
Website(s)	[Link(s) to registry website(s)]	Link(s) to registry website(s) that were used to populate the inventory.
Publication(s) used to populate inventory	[Link(s) to publication(s)]	Link(s) to publication(s) about the registry that were used to populate the inventory. Specify in brackets which inventory data elements you populated with the publication(s).
Is this registry active as of January 2, 2024?	Yes No Unclear	Specify if the registry is active: Yes: Active, defined as a registry with ethics approval and where patient recruitment began as of January 2, 2024. If registry is an ongoing cohort study, include. No: registry is closed, or if a cohort study is complete. Unclear: unable to determine if registry is active
Number of enrolled participants in Canada	[Free text] Unclear	List the number of participants in the registry who live in Canada. If unable to find size, use "unclear." The number of patients refers to the total number of patients enrolled in the registry, which can include both patients and parents or guardians or caregivers of patients.
Date number of enrolled participants in Canada retrieved	[Free text] Unclear	Date size (Canada) was calculated "Month, Year" or "Year" if no Month available. If unclear what date size was calculated, use publication date.
Total number of enrolled international participants	[Free text] NA Unclear	List the total number of participants in the registry. For Canadian registries, use "size (Canada)," and code "size (overall)" as "NA." If unable to find size, use "unclear."
Date number of enrolled international participants retrieved	[Free text] Unclear	Date size was calculated "Month, Year" or "Year" if no Month available. If unclear what date size was calculated, use publication date.
Population age group captured in this registry	Pediatric only Adults only Both	Among patients with the rare disease, specify the population age group represented: Pediatric only: registry includes patients ≤ 18 years old. If the parents or caregivers of pediatric patients provide data on behalf of the pediatric patients, use this option. Adults only: registry includes patients > 18 years old Both: registry includes both pediatric and adult patients.
Lead institution or organization	[Free text] Unclear	List the name of the institution that is the registry lead/ national team.

Data element	Coding options	Data element definition
Lead contact person/email	[Free text] Unclear	List the name and email of the individual that is the registry lead (e.g., director, principal investigator). If no specific lead, list the general email of the registry.
Part of an international registry or network?	Yes No NA Unclear	Specify if the registry is part of or collaborates with an international registry or international or national network: Yes: Part of an international registry or an international or national registry network. No: Explicitly stated or clear that registry is not part of an international registry or network. NA: registry is the international registry or network. Unclear: unable to determine if registry is part an international registry or network.
Specify which international registry or network	[Free text] NA Unclear	Specify which international registry or international or national network the registry is part of. For example, the Canadian Neuromuscular Disease Registry collaborates with the international network TREAT-NMD.
Is this registry a Canadian or an international registry?	Canadian International Unclear	Specify the registry's Canadian involvement: Canadian: registry is hosted in Canada and majority of sites and patients live in Canada. International: International registry (registry is hosted internationally) that includes patients who live in Canada. Unclear: unable to determine if any patients who live in Canada.
Participating countries	[Free text] Unclear	List the specific participating countries. For example, "USA and Canada." If unable to determine the specific countries, list the number of countries, for example, "23 countries." If unable to determine the number of countries, code as "unclear."
Canadian coverage	[Free text] Unclear	List the specific Canadian provinces/territories that are included. For example, "3 provinces: ON, QC, BC." If unable to determine the specific provinces, list the number of provinces/territories, for example "3 provinces." If unable to determine the number of or specific provinces/territories, use "unclear."
Does this registry capture participants with rare cancer(s)?	Yes No Unclear	Specify if the registry includes rare diseases in oncology.
Specific rare disease(s)	[Free text]	List specific name of disease(s) (e.g., spinal muscular atrophy) captured in the registry.

NORD = The National Organization for Rare Disorders; EMA = European Medicines Agency.

Appendix 3: Inventory of Rare Disease Registries Including Canadian Patients

The following presents Table 4 of RDRs in Canada and Table 5 of international RDRs that include patients living in Canada. The information is sourced from the literature and public searches populated up to January 26, 2024, and from a survey of registry holders collected March 20, 2024, to June 10, 2024. During the survey, the registry holders were asked about the number of patients as of March 2024. As information is sourced from public searches and directly from registry holders, the level of detail, availability of information, and timeliness of data varies across the registries. Please note that information may require further validation from registry holders. For any questions, please contact us.

Table 4: Canadian Rare Disease Registries (n = 66)

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
Alberta Congenital Anomalies Surveillance System (ACASS)	Vital Statistics, Ministry of Health Ministry of Registries	Health. Surveillance@gov .ab.ca	https://open.alberta .ca/publications/1710 -8594	Multiple congenital anomalies[d]	Pediatric	55,490 (2014)	AB
Alpha-1 Canadian Registry	Unclear	info@alpha1canadi anregistry.com	https://alpha1canada .ca/	Alpha-1 antitrypsin deficiency	NR[c]	290 (Dec 2013)	BC, AB, SK, MB, ON, QC, NB, NS, NL, YT, NT
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS) International Patient Registry	La Fondation de l'Ataxie Charlevoix-Saguenay	ataxie@arsacs.com	https://arsacs.com/ arsacs-international -patient-registry/	Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)	Both	NR[c]	QC
BC Glomerulonephritis (GN) Registry	BC Renal Network; BC GN Network	Sean Barbour, 604-875-5950, Sean.Barbour@vch .ca	http://www .bcrenal.ca/health -professionals/ clinical-resources/ glomerulonephritis	Glomerulo-nephritis	Both	NR[c]	BC
Brain Tumour Registry of Canada	School of Public Health, University of Alberta	Yan Yuan Yyuan@ ualberta.ca and Emily Walker Emily. walker@ulberta.ca	https:// braintumourregistry .ca/	Primary brain tumours	NR[c]	NR[c]	NR[c]
CAN-Fever Autoinflammatory Disease Registry	BC Children's Hospital	Lori Tucker, MD 604-875-3678	No RDR website identified, https:// www.bcchr.ca/ rheumatology/our -current-research	Autoinflammatory disease	Pediatric	150 (Mar 2024)	BC

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
				Chronic recurrent multifocal osteomyelitis	Pediatric	40 (Mar 2024)	BC
Canadian Acromegaly Registry	University of Calgary, Lumiio hosts and stores the data	Kirstie Lithgow info@acromegalyregistry.ca and info@lumiio.com	https://acromegalyregistry.ca/home	Acromegaly	Adult	NR[c]	AB, ON, QC, NS
Canadian Apheresis Group (CAG) Thrombotic thrombocytopenic purpura (TTP) Registry	Canadian Apheresis Group	Dr Gail Rock cag@cagcanada.ca and Albert Ebidia aebidia@rogers.com	https://www.cagcanada.ca/cag-registry	Thrombotic thrombocyto-penic purpura	Both	3,000 (Mar 2024)	All provinces/ territories
Canadian Atypical Teratoid Rhabdoid Tumors Registry	Unclear	Lucie Lafay-Cousin lucie.lafay-cousin@ahs.ca	No website identified	Atypical teratoid rhabdoid tumors	Pediatric	77 (2012)	NR[c]
Canadian Biliary Atresia Registry (CBAR)	BC Children's Hospital, Montreal Children's Hospital	Elena Guadagno elena.guadagno@muhc.mcgill.ca and Rick Schreiber	https://cbar.ca/	Biliary atresia	Pediatric	125 (Mar 2024)	BC, AB, ON, QC, NB, NS, NL
Canadian Bronchiectasis and Nontuberculous Mycobacteria Database	University of Calgary	Christina Thornton ceshaghu@ucalgary.ca and Julie Jarand julie.jarand@albertahealthservices.ca	No website identified	Bronchiectasis	Adult	150 (Mar 2024)	AB
				Nontuberculous mycobacteria	Adult	65 (Mar 2024)	AB

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
Canadian Cancer Registry	Unclear	Unclear	https://www23.statcan.gc.ca/imdb/p2SV.pl?Function=getSurvey&SDDS=3207	Multiple cancers	Both	Unclear[e]	All provinces
Canadian Cerebral Palsy Registry	Canadian Cerebral Palsy Registry	Michael Shevell michael.shevell@muhc.mcgill.ca and Maryam Oskoui maryam.oskoui@mcgill.ca	https://www.cpregistry.ca/	Cerebral palsy	Pediatric	2,700 (Unclear)	BC, AB, ON, NS, NL
Canadian Cystic Fibrosis Registry	Cystic Fibrosis Canada	Stephanie Cheng scheng@cysticfibrosis.ca	https://www.cysticfibrosis.ca/our-programs/cf-registry	Cystic fibrosis	Both	4,500 (Mar 2024)	All provinces/ territories
Canadian Fabry Disease Initiative	Canadian Fabry Disease Initiative Scientific Consortium	Michael L West mlwest@dal.ca and Kaye LeMoine kaye.lemoine@nshealth.ca	http://www.the-cfdi.ca/; https://www.fabrycanada.com/canadian-fabry-disease-initiative/	Fabry disease	Both	692 (Mar 2024)	All provinces/ territories
Canadian Familial Hypercholesterolemia Registry	Familial hyper-cholesterolemia Canada	Jacques Genest isabelle.ruel@affiliate.mcgill.ca and Liam Brun-ham liam.brunham@ubc.ca	https://www.fhcanada.net/	Familial chylomicronemia syndrome, lecithin-cholesterol acyltransferase deficiency, Tangier disease, sitosterolemia	Both	NR[c]	BC, AB, ON, QC, NS

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
Toronto Glomerulonephritis Registry	University Health Network	Heather Reich heather.reich@uhn.ca and Arenn Jauhal arenn.jauhal@uhn.ca	https://cansolveckd.ca/gnregistry	IgA nephropathy	Adult	600 (Mar 2024)	ON
				Membranous nephropathy	Adult	400 (Mar 2024)	ON
				Focal segmental glomerulo-nephritis	Adult	400 (Mar 2024)	ON
				Minimal change disease	Adult	100 (Mar 2024)	ON
Canadian Homozygous Familial Hypercholesterolemia (HoFH) Registry	McGill; University of BC	Jacques Genest isabelle.ruel@affiliate.mcgill.ca and Liam Brunham liam.brunham@ubc.ca	No website identified	Homozygous familial hypercholes-terolemia	Both	45 (Mar 2024)	BC, MB, ON, QC
Canadian Inflammatory Myopathy Study (CIMS)	Jewish General Hospital	Marie Hudson, Jewish General Hospital	https://www.canadianims.org/	Dermatomyositis	Adult	75 (Mar 2024)	BC, MB, ON, QC
				Polymyositis	Adult	< 6 (Mar 2024)	QC
				Inclusion body myositis	Adult	20 (Mar 2024)	AB, QC
				Immune-mediated necrotizing myositis	Adult	10 (Mar 2024)	QC

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
				Scleromyositis	Adult	50 (Mar 2024)	QC
				Antisynthetase syndrome	Adult	40 (Mar 2024)	BC, ON, QC
				Overlap myositis	Adult	20 (Mar 2024)	QC
Canadian Morphea Registry (C-MORE)	McGill University Health Centre	Elena Netchiporouk Elena. netchiporouk@mail .mcgill.ca	https://skincanada .org/c-nest/	Morphea	Both	174 (Mar 2024)	SK, ON, QC
				Eosinophillic fasciitis	Both	< 6 (Mar 2024)	ON, QC
Canadian Myelodysplastic syndromes (MDS-CAN)	Sunnybrook Health Sciences Center	Rena Buckstein and Cherry Blushi	https://www.mds-can .ca/	Myelodysplastic syndromes	Adult	1,300 (Mar 2024)	BC, AB, SK, MB, ON, QC, NB, NS
				Chronic myelomonocytic leukemia	Adult	100 (Mar 2024)	BC, AB, SK, MB, ON, QC, NB, NS
				Oligoblastic acute myeloid leukemia	Adult	50 (Mar 2024)	BC, AB, SK, MB, ON, QC, NB, NS
Canadian National Hypoparathyroidism Registry	McMaster University	Aliya A. Khan aliya@ mcmaster.ca	No website identified	Hypopara-thyroidism, and subsyndromes	Adult	130 (Jan 2020)	ON
Canadian Network for Autoimmune Liver Disease (CaNAL)	Toronto General Hospital; University of Alberta	Gideon Hirschfield gideon.hirschfield@ uhn.ca and Andrew	https://www .canalregistry.ca/	Autoimmune hepatitis	Adult	1,780 (Mar 2024)	BC, AB, ON, QC, NS

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
		Mason am16@ualberta.ca					
				Primary biliary cholangitis	Adult	3494 (Mar 2024)	BC, AB, ON, QC, NS
Canadian Neuromuscular Disease Registry (CNDR)	University of Calgary	Lawrence Korngut lwkorngu@ucalgary.ca and Victoria Hodgkinson vhogkin@ucalgary.ca	https://cndr.org/	Spinal muscular atrophy	Both	364 (Mar 2024)	BC, AB, SK, MB, ON, QC, NB, PE, NS, YT, NT
				Duchenne muscular dystrophy	Both	374 (Mar 2024)	All provinces/ territories
				Amyotrophic lateral sclerosis	Both	2,260 (Mar 2024)	All provinces/ territories
				Limb girdle muscular dystrophy	Both	200 (Mar 2024)	BC, AB, SK, ON, QC, NB
				Myotonic dystrophy	Both	578 (Mar 2024)	All provinces/ territories
				Congenital myasthenic syndrome	Both	14 (Mar 2024)	BC, AB, SK, ON, QC, NB
				Facioscapulo-humeral muscular dystrophy	Both	255 (Mar 2024)	BC, AB, SK, MB, ON, QC, NB, NS

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
				135 other neuromuscular diseases[f] (only diagnosis collected)	Both	1,635 (Mar 2024)	All provinces/ territories
Canadian Paroxysmal Nocturnal Hemoglobinuria (PNH) Registry[i]	Unclear	Christopher Patriquin Christopher.patriquin@medportal.ca and medinfo@alexion.com	https://pnhregistry.com/physicians	Paroxysmal nocturnal hemoglobinuria	Both	107 (Jan 2018)	BC, AB, ON, QC, NS, NL
Canadian Pediatric Neuroinflammatory Disorders Registry[g]	Hospital for Sick Children, University of Toronto	E. Ann Yeh ann.yeh@sickkids.ca	https://lab.research.sickkids.ca/neuroinflamm/research/	Pediatric onset multiple sclerosis	Pediatric	200 (Mar 2024)	All provinces/ territories
				Myelin oligodendrocyte glycoprotein antibody-associated disease	Pediatric	200 (Mar 2024)	All provinces/ territories
				Neuromyelitis optica spectrum disorder-seronegative	Pediatric	20 (Mar 2024)	All provinces/ territories
				Neuromyelitis optica spectrum disorder - aquaporin-4 positive	Pediatric	10 (Mar 2024)	All provinces/ territories

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
				Optic neuritis, transverse myelitis, acute necrotizing encephalopathy of childhood, opsoclonus myoclonus syndrome, monophasic acquired demyelinating syndromes	NR[c]	NR[c]	NR[c]
Canadian Prospective Cohort Study to Understand Progression in MS	NR[c]	a.prat@umontreal.ca	No website identified	Multiple sclerosis	Adult	NR[c]	BC, AB, ON, QC
Canadian Pulmonary Hypertension Registry	The University of British Columbia; Vancouver Coastal Health; VGH & UBC Hospital Foundation	Freda Tom freda.tom@vch.ca and Dr. John Swiston swiston@mail.ubc.ca	https://www.phacanada.ca/canadianphregistry	Pulmonary hypertension	Both	2,212 (Mar 2024)	BC, AB, MB, ON, QC, NB, NS, NL, YT
Canadian Registry for Amyloidosis Research (CRAR)	University of Calgary	Nowell Fine nowell.fine@ahs.ca	https://amyloidregistry.ca	Amyloid light-chain light chain amyloidosis	Adult	100 (Mar 2024)	BC, AB, ON, NS
				Transthyretin amyloidosis	Adult	300 (Mar 2024)	BC, AB, ON, NS
Canadian Registry for Pulmonary Fibrosis[g]	University of British Columbia	Christopher Ryerson chris.ryerson@hli.ubc.ca	No website identified	Idioapthic pulmonary fibrosis	Adult	1,200 (Mar 2024)	BC, AB, SK, ON, QC, YT

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
				Connective tissue disease-associated interstitial lung disease	Adult	2,000 (Mar 2024)	BC, AB, SK, ON, QC, YT
				Hypersensitivity pneumonitis	Adult	350 (Mar 2024)	BC, AB, SK, ON, QC
				Unclassified interstitial lung disease	Adult	1,100 (Mar 2024)	BC, AB, SK, ON, QC, YT
				Drug-induced interstitial lung disease	Adult	50 (Mar 2024)	BC, AB, SK, ON, QC, YT
				Sarcoidosis	Adult	200 (Mar 2024)	BC, AB, SK, ON, QC, YT
				Asbestosis	Adult	30 (Mar 2024)	BC, AB, SK, ON, QC
				Silicosis	Adult	20 (Mar 2024)	BC, AB, SK, ON, QC, YT
				Post-COVID pulmonary fibrosis	Adult	50 (Mar 2024)	BC, AB, SK, ON, QC, YT
				Cryptogenic organizing pneumonia	Adult	50 (Mar 2024)	BC, AB, SK, ON, QC, YT
Canadian Rett Syndrome Registry	ON Rett Syndrome Association	info@rett.ca	https://canadianrettsyndromeregistry.com/	Rett syndrome	NR[c]	NR[c]	NR[c]

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
Canadian Sarcoma Research and Clinical Collaboration (CanSaRCC)	University Health Network	Dr. Abha Gupta Abha.Gupta@uhn.ca and Dr. Hagit Peretz Soroka Hagit.Peretz@uhn.ca	https://www.cansarcc.ca/about-cansarcc	Sarcoma	Both	4,250 (2023)	BC, AB, SK, MB, ON, QC, NS, NL
Canadian Scleroderma Research Group	St Joseph's Health care Hamilton; Canadian Scleroderma Research Group	Maggie Larche 905-528-0489 and Stephanie Densmore-Farnworth 905-528-0489	http://www.canadiansclerodermaresearchgroup.org	Systemic sclerosis	Both	1,570 (Mar 2024)	BC, AB, SK, MB, ON, QC, NB, NS, NL
				Scleroderma	Both	150 (Mar 2024)	AB, MB, ON, QC, NB, NS
Canadian Vasculitis Network (CanVasc) Registry	Canadian Vasculitis Network	Lillian Barra Lillian.Barra@sjhc.london.on.ca	https://canvasc.ca/	Vasculitis[j]	Adult	425 (June 2024)	NR[c]
Cancer in Young People in Canada (CYP-C)	C17 Council; Public Health Agency of Canada	Randy Barber randy.barber@c17.ca	https://www.c17.ca/	Pediatric cancers	Pediatric	13,500 (Mar 2024)	All provinces/ territories
CAN-OPTICS: the Canadian Neuromyelitis Optica Spectrum Disorder and other atypical demyelinating diseases Cohort Study	Unity Health Toronto	Dr. Dalia Rotstein 416-864-5660	No website identified	Seropositive neuromyelitis optica spectrum disorder	Adult	135 (Mar 2024)	BC, AB, MB, ON, NS

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
				Seronegative neuromyelitis optica spectrum disorder	Adult	15 (Mar 2024)	AB, MB, ON, NS
				Myelin oligodendrocyte glycoprotein antibody disease	Adult	85 (Mar 2024)	BC, AB, MB, ON, NS
				Glial fibrillary acidic protein encephalomyelitis	Adult	< 6 (Mar 2024)	BC, AB, MB, ON, NS
Clinical Von Hippel-Lindau disease (VHL) Database	University Health Network	Raymond Kim raymond.kim@uhn.ca	No website identified	Von Hippel-Lindau disease	Both	86 (2019)	ON
Discovering the Periodic Fever Syndrome Population at Hamilton Health Sciences	Hamilton Health Sciences	Liane Heale healel@mcmaster.ca	No website identified	PFAPA (periodic fever, aphthous stomatitis, pharyngitis, adenitis)	Pediatric	14 (Mar 2024)	ON
				Familial Mediterranean fever	Both	17 (Mar 2024)	ON
				Deficiency of adenosine deaminase 2	Pediatric	< 6 (Mar 2024)	ON
				Yao syndrome	Adult	< 6 (Mar 2024)	ON
				Behcet's disease	Pediatric	< 6 (Mar 2024)	ON

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
				NLRP3-associated auto-inflammatory syndrome	Pediatric	< 6 (Mar 2024)	ON
				SURF (syndrome of undifferentiated recurrent fever)	Both	6 (Mar 2024)	ON
Fighting Blindness Canada Inherited Retinal Disease Patient Registry[g]	The Hospital for Sick Children	Elise Heon elise.heon@sickkids.ca and Larissa Moniz lmoniz@fightingblindness.ca	https://www.fightingblindness.ca/patient-registry/	Retinitis pigmentosa	Both	1,147 (Mar 2024)	BC, AB, SK, MB, ON, QC, NB, PE, NS, NL, YT, NT
				Stargardt's disease	Both	236 (Mar 2024)	BC, AB, SK, ON, QC, NS, NT
				Usher syndrome	Both	175 (Mar 2024)	BC, AB, SK, ON, QC, NB, NS
				Leber congenital amaurosis	Both	133 (Mar 2024)	BC, AB, SK, MB, ON, QC, NS
				Cone-rod dystrophy	Both	92 (Mar 2024)	BC, AB, SK, ON, QC, NB, PE, NS
				Achromatopsia	Both	73 (Mar 2024)	BC, AB, ON, QC, NB, NS
				Choroideremia	Both	69 (Mar 2024)	BC, AB, MB, ON, QC, NS

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
				Retinoschisis	Both	57 (Mar 2024)	BC, AB, SK, ON
				Congenital stationary night blindness	Both	55 (Mar 2024)	BC, AB, SK, ON, QC, PE, YT
				Rod-cone dystrophy	Both	35 (Mar 2024)	BC, AB, ON, NB, NL
Genetic Studies of Chronic Kidney Disease[g]	University of BC, Providence Health Research	Dr. Mark Elliott melliott1@providencehealth.bc.ca	http://www.bcrenal.ca/health-professionals/clinical-resources/polycystic-kidney-disease#ADPKD--Registry	Genetic chronic kidney diseases[k]	Both	1,076 (Jan 2020)	BC
Genodermatoses Registry[g]	The Hospital for Sick Children	Irene Lara-Corrales irene.lara-corrales@sickkids.ca and Yiming Wang yiming.wang@sickkids.ca	No website identified	Ichthyosis (multiple)	Pediatric	8 (Mar 2024)	ON
				Skin fragility disorders	Pediatric	< 6 (Mar 2024)	ON
				Palmoplantar keratodermas	Pediatric	7 (Mar 2024)	ON
				Pigmentary disorders	Pediatric	8 (Mar 2024)	ON
				Ectodermal dysplasias	Pediatric	< 6 (Mar 2024)	ON

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
				Cancer predisposition syndromes	Pediatric	< 6 (Mar 2024)	ON
				Hair disorders	Pediatric	< 6 (Mar 2024)	ON
				Nail disorders	Pediatric	< 6 (Mar 2024)	ON
				Photosensitive syndromes	Pediatric	6 (Mar 2024)	ON
				Other rare genetic skin disorders	Pediatric	40 (Mar 2024)	ON
KidCOM	The Hospital for Sick Children; Nationwide Children's Hospita	Christoph Licht christoph.licht@sickkids.ca and William E. Smoyer william.smoyer@nationwidechildrens.org	No website identified	Atypical hemolyticuremic syndrome	Both	40 (Mar 2024)	BC, AB, ON, QC
				IC-membranopro-liferative glomerulo-nephritis/C3 glomerulopathy	Both	80 (Mar 2024)	BC, AB, ON, QC
Mastocytosis Assessment and Treatment Evaluation Registry	McGill University Health Centre	Greg Shand masterstudymch@gmail.com	https://www.mastocytosis.ca/en/get-involved/join-the-mastocytosis-registry	Mastocytosis	Both	23 (Feb 2020)	NR[c]
Mito Canada Patient Registry[g]	Lumiio	Kate Murray Kate.Murray@MitoCanada.org	https://mitocanada.org/patient-contact-registry/; https://-	MELAS (mitochondrial encephalo-myopathy,	Both	14 (Mar 2024)	AB, ON, NL

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
			mitocanada.lumiio.com/home	lactic acidosis, and strokelike episodes)			
				Leigh syndrome	Pediatric	< 6 (Mar 2024)	ON
				CPEO (chronic progressive external ophthalmoplegia)	Adult	6 (Mar 2024)	BC, ON, NB
				Complex IV/COX (cytochrome c oxidase) deficiency	Both	< 6 (Mar 2024)	AB, ON
				MIDD (maternally inherited diabetes and deafness)	Adult	< 6 (Mar 2024)	BC, ON
				MERFF (myoclonic epi-lepsy with ragged red fibers)	Adult	< 6 (Mar 2024)	BC, ON
				Progressive external ophthalmoplegia	Adult	< 6 (Mar 2024)	BC, AB
				Lactic acidosis	Both	6 (Mar 2024)	AB, ON
				MNGIE (mitochondrial neurogastro-intestinal encephalopathy)	Adult	< 6 (Mar 2024)	BC

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
				Mitochondrial encephalopathy	Both	< 6 (Mar 2024)	ON
Myeloma Canada Research Network (MCRN) Canadian Multiple Myeloma Database	Canadian Myeloma Research Group	info@cmrg.ca	https://cmrg.ca/research/real-world-evidence/	Multiple myeloma	Both	8,387 (2014)	BC, AB, SK, MB, ON, QC, NB, NS, NL
Myeloproliferative Neoplasms Patient Registry	University Health Network	Vikas Gupta vikas.gupta@uhn.ca	No website identified	Myeloproliferative neoplasms	Both	5,000 (2023)	ON
National Hearts in Rhythm Organization (HiRO) Registry[g]	University of British Columbia	Dr. Andrew Krahn akrahn@mail.ubc.ca and Brianna Davies BDavies@providencehealth.bc.ca	https://hiro.heartsinrhythm.ca/	Unexplained cardiac arrest syndromes	Both	513 (Mar 2024)	BC, AB, MB, ON, QC, NS
				Sudden arrhythmogenic death syndrome	Both	35 (Mar 2024)	BC, AB, MB, ON, QC, NS
				Arrhythmogenic right ventricular cardiomyopathy	Both	700 (Mar 2024)	BC, AB, MB, ON, QC, NS
				Long QT syndrome	Both	1,388 (Mar 2024)	BC, AB, MB, ON, QC, NS
				Brugada syndrome	Both	483 (Mar 2024)	BC, AB, MB, ON, QC, NS

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
				Catecholaminergic polymorphic ventricular tachycardiac	Both	119 (Mar 2024)	BC, AB, MB, ON, QC, NS
				Calcium release deficiency syndrome	Both	< 6 (Mar 2024)	BC, AB, MB, ON, QC, NS
				Short QT syndrome	Both	< 6 (Mar 2024)	BC, AB, MB, ON, QC, NS
				Hypertrophic cardiomyopathy	Both	181 (Mar 2024)	BC, AB, MB, ON, QC, NS
				Dilated cardiomyopathy	Both	73 (Mar 2024)	BC, AB, MB, ON, QC, NS
Orphan Disease Center CDKL5 Deficiency Disorder International Patient Registry[h]	Not Applicable	Daniel J. Lavery	https://www.cdkl5.com/cdkl5-international-registry-database/	*CDKL5* deficiency disorder	Both	NR[c]	NR[c]
Pediatric Neurofibromatosis Registry	The Hospital for Sick Children	Dr. Patricia Parkin patricia.parkin@sickkids.ca and Keenjal Mistry keenjal.mistry@sickkids.ca	No website identified	Neurofibromatosis type 1	Pediatric	1,500 (Mar 2024)	ON
Pediatric Oncology Group of ON Networked Information System (POGONIS)	Pediatric Oncology Group of ON (POGO)	Bruna DiMonte bdimonte@pogo.ca	https://www.pogo.ca/research-data/pogonis-childhood-cancer-database/data-anatomy/	Pediatric cancers	Pediatric	19,900 (2020)	ON

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
Prospective Longitudinal Study to Assess Long-Term Safety of Treatments and the Epidemiology of Bleeding in Immune Thrombocytopenia	McMaster University Michael G. DeGroote Centre for Trans-fusion Research	Dr. Donald Arnold arnold@mcmaster.ca	No website identified	Immune thrombocy-topenia	Both	1,065 (Mar 2024)	ON
Province of ON Neurodevelopmental Disorders (POND) Network OBI: POND Registry	Unclear	Alana Iaboni aiaboni@hollandbloorview.ca	https://pond-network.ca/areas-of-study/, https://braininstitute.ca/year-in-review/advancing-knowledge-22	Attention-deficit/ hyperactivity disorder, autism spectrum disorder, intellectual disability, obsessive-compulsive disorder, Tourette syndrome, Rett syndrome, Down syndrome, fragile X syndrome, Other	Pediatric	2,000 (May 2021)	ON
QC congenital heart disease registry	Université de Sherbrooke	Frédéric Dallaire frederic.a.dallaire@usherbrooke.ca	https://ccpcrn.ca/portfolio/the-QC-congenital-heart-disease-registry/	Congenital heart malformations	Both	54,000 (Mar 2024)	QC
QC Myotonic Dystrophy Registry/ Registre québécois sur la dystrophie myotonique de type 1 Q-DMR	CIUSSS du Saguenay-Lac-Saint-Jean	Jean Mathieu and Cynthia Gagnon cynthia5_gagnon@uqac.ca	No website identified	Myotonic dystrophy	Both	1,410 (June 2023)	QC

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
QC Trophoblastic Disease Network	Réseau des Maladies Trophoblastiques du Québec (RMTQ)	Catherine De RAVINEL catherine.de.ravinel.chum@ssss.gouv.qc.ca	https://rmtq.ca/en/rmtq-mission/rmtq-software/	Trophoblastic diseases	NR[c]	925 (Dec 2019)	QC
Registry of Rare Diseases in Pregnancy (Groupe d'etude en medecine obstetricale du Québéc)	Groupe d'étude en médecine obstétricale du QC (QEMOQ)	Lucie Terriault 514-350-5118	https://gemoq.ca/welcome-to-the-gemoq/	Rare medical conditions in pregnancy	Both	NR[c]	QC
SickKids Lupus Registry	The Hospital for Sick Children	Linda Hiraki linda.hiraki@sickkids.ca	No website identified	Childhood-onset systemic lupus erythematosus	Pediatric	250 (Mar 2024)	ON
				Secondary hemophagocytic lymphohistio-cytosis/ macrophage activation syndrome	Pediatric	25 (Mar 2024)	ON
				Monogenic lupus	Pediatric	20 (Mar 2024)	ON
Southern Alberta Registry for Systemic Lupus Erythematosus: STARLET	University of Calgary	Dr. Ann Clarke aeclarke@ucalgary.ca	https://connect.invitae.com/en/org/tdhlf	Systemic lupus erythematosus	Adult	450 (Mar 2024)	AB
STXBP1.CA	BC Children's Hospital	Cyrus Boelman Cyrus.boelman@cw.bc.ca	https://www.stxbp1.ca/	STXBP1 encephalopathy	Both	25 (Mar 2024)	All provinces/ territories

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
The Canadian Alliance of Pediatric Rheumatology Investigators Juvenile Idiopathic (CAPRI) Registry	University of British Columbia	Jaime Guzman jguzman@cw.bc.ca	No RDR website identified, https://www.bcchr.ca/rheumatology/our-current-research	Oligoarthritis	Pediatric	521 (Mar 2024)	All provinces/ territories
				Polyarthritis rheumatoid factor negative	Pediatric	218 (Mar 2024)	All provinces/ territories
				Polyarthritis rheumatoid factor positive	Pediatric	41 (Mar 2024)	All provinces/ territories
				Enthesitis related arthritis	Pediatric	169 (Mar 2024)	All provinces/ territories
				Psoriatic arthritis	Pediatric	69 (Mar 2024)	All provinces/ territories
				Systemic arthritis	Pediatric	54 (Mar 2024)	All provinces/ territories
				Undifferentiated juvenile arthritis	Pediatric	76 (Mar 2024)	All provinces/ territories
The Canadian Bleeding Disorders Registry (CBDR)	Association of Hemophilia McMaster	Alfonso Iorio iorioa@mcmaster.ca and Arun Keepanasseril keeppaa@mcmaster.ca	https://www.ahcdc.ca/cbdr	Hemophilia, Von Willebrand disease, rare coagulation factor deficiencies	Both	3,200 (2021)	All provinces

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients in Canada (date retrieved)[b]	Coverage in Canada
The Canadian Inherited Marrow Failure Registry (CIMFR)	The Hospital for Sick Children	Bozana Zlateska cimf.registry@sickkids.ca and Yigal Dror yigal.dror@sickkids.ca	https://www.sickkids.ca/en/care-services/clinical-departments/cimfr/	More than 30 inherited bone marrow failure syndromes	Both	600 (Unclear)	NR[c]
The Canadian Inherited Metabolic Diseases Network (CIMDRN)	Children's Hospital of Eastern ON Research Institute, INFORM RARE	Beth Potter bpotter@uottawa.ca and informrare@uottawa.ca	https://www.informrare.ca/cimdrn	Inherited metabolic diseases[l]	Pediatric	798 (June 2019)	BC, AB, MB, ON, QC, NS, NL
The Canadian Mucopoly-saccharidosis Registry	CHEO Research Institute	Emma Lynn elynn@cheo.on.ca	https://www.mpsregistry.ca/	Mucopoly-saccharide and related diseases	Pediatric	17 (Mar 2024)	All provinces/ territories
The University Health Network and SickKids Hospital Neurofibromatosis type 1 Registry	University Health Network; The Hospital for Sick Children	Carolina Barnett-Tapia c.barnetttapia@utoronto.ca and Patricia Parkin patricia.parkin@sickkids.ca	No website identified	Neurofibro-matosis type 1	Both	1200 (Mar 2024)	ON
Toronto Hereditary Hemorrhagic Telangiectasia Database	St Michael's Hospital, Unity Health Toronto	Marie Faughnan marie.faughnan@unityhealth.to and Negar Bagheri Negar.bagheri@unityhealth.to	No website identified	Hereditary hemorrhagic telangiectasi	Adult	1400 (Mar 2024)	All provinces/ territories

AB = Alberta; BC = British Colombia; Dec = December; Jan = January; Mar = March; MB = Manitoba; NB = New Brunswick; NL = Newfoundland and Labrador; NR = not reported or not available; NS = Nova Scotia; NT = Northwest Territories; NU = Nunavut; ON = Ontario; PE = Prince Edward Island; PROMIS = Patient Records and Outcome Management Information System; QC = Quebec; RDR = rare disease registry; SK = Saskatchewan; YT = Yukon.

[a]Websites sourced from publicly available information.

[b]Among registries captured from the literature search, information is available overall, and not per disease.

[c]Not reported or available because of reliance on information available in the literature or a public search or partial completion of the registry holder survey. If the registry holder did not complete information, literature and/or public search information was inputted if available.

[d]Multiple congenital anomalies, including neural tube defects, anencephaly, spina bifida, anotia or microtia, orofacial clefts, cleft lip with or without cleft palate, cleft palate only, anorectal malformations, congenital heart defect, transposition of the great arteries, tetralogy of Fallot, ventricular septal defect, atrial septal defect, hypoplastic left heart syndrome, hypospadias, undescended testes, limb reduction, gastroschisis, omphalocele, Down syndrome, and other congenital anomalies.

[e]The Canadian Cancer Registry includes at least 1,000,000,000 individuals diagnosed with cancer; however, the total number of individuals with rare cancers is unclear.

[f]CNDR collects diagnosis (no clinical data) for 135 other neuromuscular disorders, including autosomal dominant paramyotonia congenita (SCN4A), congenital myopathy (unspecified), hereditary inclusion body myopathy, Kearns-Sayre syndrome, King-Denborough syndrome, mitochondrial myopathy, myotonia congenita — Thomsen disease, nondystrophic myotonia, paramyotonia congenita, phosphorylase deficiency (McArdles Disease), primary myopathy, PTEN myopathy, rippling muscle disease, *RYR1*-related myopathy, X-linked myopathy with excessive autophagy, congenital muscular dystrophy, congenital muscular dystrophy, merosin deficient, manifesting dystrophinopathy carrier, muscular dystrophy (unspecified), oculopharyngeal muscular dystrophy, titinopathy, dermatomyositis, inclusion body myositis, polymyositis, Lambert-Eaton syndrome, myasthenia gravis (all types), myasthenic syndrome, Charcot-Marie-Tooth disease (all types), Dejerine-Sottas disease, Freidreich ataxia, hereditary motor and sensory neuropathy with agenesis of the corpus callosum (Andermann syndrome), hereditary neuralgia amyotrophy, hereditary neuropathy with liability to pressure palsies, hereditary sensory autonomic neuropathy type IV, hereditary sensory neuropathy type 1, syndrome of neuropathy ataxia and retinitis pigmentosa, Tangier disease, diabetic neuropathy, glucose intolerance neuropathy, idiopathic neuropathy, neuropathy (unspecified), peripheral neuropathy (unclassified), chronic inflammatory demyelinating polyneuropathy, Guillain-Barré syndrome, Miller Fisher syndrome, multifocal motor neuropathy, acquired neuromyotonia (Isaacs syndrome), POEMS neuropathy, Sjogren peripheral neuropathy, vasculitis, polyneuropathy, sensory polyneuropathy, small fibre peripheral neuropathy, motor neuron disease (unspecified), primary lateral sclerosis, progressive muscular atrophy, spinal bulbar muscular atrophy, post-polio syndrome, Andersen Tawil syndrome, hyperkalemic periodic paralysis, hypokalemic periodic paralysis, hereditary spastic paraparesis, human T-cell lymphotropic virus type 1–associated myelopathy or tropic spastic paraparesis, myofibrillar myopathy (LDB3), stiff person syndrome, adrenoleukodystrophy, amyoplasia, arthrogryposis, autosomal recessive spastic ataxia of charlevoix-saguenay, D-bifunctional protein deficiency, Freeman-Sheldon syndrome, metabolic ataxia, Poland syndrome, spinal atrophy and paraplegia, and spinocerebellar ataxia.

[g]During the survey with registry holders, registries with more than 10 rare diseases were instructed to enter data for the 10 most prevalent diseases at this stage.

[h]Registry holder reported registry as a registry in Canada.

[i]Please note that there is also an International PNH Registry (funded by Alexion Pharmaceuticals), which will soon be rolling into an academic-run broader registry under the auspices of the International PNH Interest Group, which will permit all patients with PNH regardless of therapeutic (e.g., those not made by Alexion) to join to be broader and more representative of the current PNH treatment landscape.

[j]Multiple types of vasculitis, including giant cell arteritis, Takayasu arteritis, polyarteritis nodosa, Behcet disease, Cogan disease, relapsing polychondritis, antineutrophilic cytoplasmic antibody–associated vasculitis, IgA vasculitis, cryoglobulinemic vasculitis, cutaneous limited vasculitis, hypocomplementemic vasculitis, primary angiitis of the central nervous system IgG4-related disease, and secondary vasculitis.

[k]Genetic chronic kidney diseases, including Alport syndrome, autosomal dominant polycystic kidney disease, chronic kidney disease of unknown etiology, *CUBN*-associated proteinuria, genetic focal segmental glomerulosclerosis, autosomal dominant tubulointerstitial kidney disease, genetic tubular disorders, C3 glomerulopathy and thrombotic microangiopathy, cystic kidney disease, and *APOL1*-assocaited kidney disease.

[l]Inherited metabolic diseases, including phenylalanine hydroxylase deficiency, homocystinuria, maple syrup urine disease, tyrosinemia, arginase deficiency, argininosuccinic acidemia, carbamoyl phosphate synthetase deficiency, citrin deficiency, citrullinemia, hyperornithinemia-hyperammonemia-homocitrullinuria syndrome, N-acetylglutamate synthetase deficiency, ornithine transcarbamylase deficiency, beta-ketothiolase deficiency, glutaric acidemia type I, hydroxymethylglutaryl-CoA lyase deficiency, isovaleric academia, 3-methylcrotonyol-CoA carboxylase deficiency, methylmalonic acidemias, propionic acidemia, medium-chain acyl-CoA dehydrogenase deficiency, very long-chain acyl-CoA dehydrogenase deficiency, carnitine uptake defect, long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency, trifunctional protein deficiency, guanidinoacetate methyltransferase deficiency, mucopolysaccharidosis type I, Farber disease, galactosemia, glycogen storage disease type I, multiple carboxylase deficiency or biotinidase deficiency, and pyridoxine-dependent epilepsy.

Table 5: International Rare Disease Registries Including Patients Living in Canada (n = 82)

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
Alpha-1 International Registry	Unclear	Robert Stockley r.a.stockley@bham.ac.uk	No website identified	Alpha-1 antitrypsin deficiency	Both	4,615 (2014)	290 (Dec 2013)	BC, AB, SK, MB, ON, QC, NB, NS, NL, YT, NT
APS ACTION International Clinical Database and Repository	APS ACTION	JoAann Vega vegaj@hss.edu	https://apsaction.com/research/	Antiphospho-lipid syndrome	Adult	1,300 (2022)	31 (Feb 2022)	AB, QC
Autosomal Recessive Cerebellar Ataxia Global Registry	Unclear	Matthis Synofzik matthis.synofzik@uni-tuebingen.de	No website identified	Autosomal recessive cerebellar ataxia (ARCA)	Both	800 (2021)	NR[c]	QC
Barth Syndrome Registry and Repository	Barth Syndrome Foundation	Melissa Huang melissa.huang@barthsyndrome.org	https://barthsyndromeregistry.patientcrossroads.org/	Barth syndrome	Both	148 (Mar 2024)	12 (Mar 2024)	BC, AB, SK, ON, QC
Brain Vascular Malformation Consortium (BVMC)	Rare Diseases Clinical Research Network	rd.dmcc@cchmc.org	https://bvmc.rarediseasesnetwork.org/our-consortium	Brain vascular malformations	Both	33 (Dec 2011)	NR[c]	ON
Canadian Cholangio-carcinoma Collaborative Registry	Ottawa Hospital Research Institute	Rebecca Auer rauer@toh.ca	https://www.cholangio.ca/patients	Cholangio-carcinoma	Adult	30 (Mar 2024)	30 (Mar 2024)	All provinces

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
CDKL5 International Patient Registry	University of Pennsylvania Perelman School of Medicine, the Orphan Disease Center and University of Western Australia	odcregistry@pennmedicine.upenn.edu and info@cdkl5canada.ca	https://www.cdkl5canada.ca/cdkl5-international-patient-registry	CDKL5 deficiency disorder	Pediatric	NR[c]	NR[c]	NR[c]
Clinical Research Consortium for the Study of Cerebellar Ataxia (CRC-SCA)	National Ataxia Foundation and Ataxia Canada	Laura Crespo laura@ataxia.org and Francois-Olivier Theberge francois.theberge@lacaf.org	https://www.ataxia.org/crc-sca/	SCA2	Adult	204 (Mar 2024)	< 6 (Mar 2024)	All provinces/ territories
				SCA3	Adult	345 (Mar 2024)	< 6 (Mar 2024)	All provinces/ territories
				SCA6	Adult	168 (Mar 2024)	< 6 (Mar 2024)	All provinces/ territories
				SCA7	Adult	26 (Mar 2024)	< 6 (Mar 2024)	All provinces/ territories
				SCA8	Adult	42 (Mar 2024)	< 6 (Mar 2024)	All provinces/ territories
Consortium for Clinical Investigation of Neurologic Channelopathies	Rare Diseases Clinical Research Network Patient Contact Registry	Richard Barohn, University of Missouri	https://neuromuscularstudygroup.org/2016/04/07/consortium-for-clinical-investigation-of-neurologic-channelopathies-cinch/	Neurologic channelo-pathies	NR[c]	NR[c]	NR[c]	ON

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
CoRDS Ataxia Patient Registry[d]	National Ataxia Foundation and Sanford Research	Lauren Moore lauren@ataxia.org and Alyssa Mendel alyssa.mendel@sanfordhealth.org	https://research.sanfordhealth.org/rare-disease-registry	SCA2	Adult	110 (Mar 2024)	15 (Mar 2024)	All provinces/ territories
				SCA3	Adult	258 (Mar 2024)	14 (Mar 2024)	All provinces/ territories
				SCA (Unknown Subtype)	Adult	150 (Mar 2024)	11 (Mar 2024)	All provinces/ territories
				SCA6	Adult	185 (Mar 2024)	8 (Mar 2024)	All provinces/ territories
				SCA8	Adult	75 (Mar 2024)	< 6 (Mar 2024)	All provinces/ territories
				Friedreich ataxia	Adult	73 (Mar 2024)	6 (Mar 2024)	All provinces/ territories
				Episodic Ataxia	Adult	54 (Mar 2024)	< 6 (Mar 2024)	All provinces/ territories,
				SCA1	Adult	74 (Mar 2024)	< 6 (Mar 2024)	All provinces/ territories
				SCA5	Adult	27 (Mar 2024)	< 6 (Mar 2024)	All provinces/ territories
				SCA7	Adult	30 (Mar 2024)	< 6 (Mar 2024)	All provinces/ territories

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
Diamond Blackfan Anemia Registry	Feinstein Institutes for Medical Research at Northwell Health	Adrianna Vlachos 516-562-1505	https://www.dbar.org/home	Diamond Blackfan anemia	Both	877 (Mar 2024)	54 (Mar 2024)	BC, AB, SK, MB, ON, QC, NS, NL
EBCare Patient Insights Network	Unclear	Unclear	https://ebcare.patientcrossroads.org/	Epidermolysis bullosa	Both	214 (2022)	10 (2022)	NR[c]
Eosinophilic gastrointestinal disease Partners	University of North Carolina at Chapel Hill School of Medicine	info_egidpartners@unc.edu	https://egidpartners.org/pages/partners_statistics_index	Eosinophilic gastrointes-tinal disease (EGID)	Both	694 (Jan 2024)	19 (Jan 2024)	ON
Enroll-HD	Unclear	Noopur Modi Info@Enroll-HD.org	https://www.enroll-hd.org/	Huntington disease	Adult	20,000 (2023)	NR[c]	BC, AB, ON, QC, NS
Fibrodysplasia Ossificans Progressiva Registry	Inter-national Fibrodysplasia Ossificans Progression Association (IFOPA)	Samantha Kile sammi.kile@ifopa.org and Michelle Davis michelle.davis@ifopa.org	https://www.ifopa.org/fop_registry_hcp	Fibrodysplasia Ossificans Progressiva	Both	355 (Mar 2024)	9 (Mar 2024)	ON, NT
Frontotemporal Degeneration (FTD) Disorders Registry[e]	FTD Disorders Registry, LLC	Lakecia Vincent lvincent@ftdregistry.org and Carrie Milliard cmilliard@ftdregistry.org	https://ftdregistry.org/	Frontotem-poral dementia/ Behavioural variant	Adult	1,225 (Mar 2024)	19 (Mar 2024)	BC, ON, QC
				Progressive Supranuclear Palsy	Adult	184 (Mar 2024)	< 6 (Mar 2024)	ON

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
				FTD with motor neuron disease	Pediatric	84 (Mar 2024)	< 6 (Mar 2024)	BC, ON
				Primary Progressive Aphasia	Adult	420 (Mar 2024)	6 (Mar 2024)	ON, NS
Fungi-Scope	Unclear	Unclear	http://www.fungiscope.net/index.php	Invasive fungal infections	Both	1,268 (2021)	NR[c]	NR[c]
Genetic Disorders of Mucociliary Clearance Consortium	Rare Diseases Clinical Research Network Patient Contact Registry	Kelli Sullivan Kelli_Sullivan@med.unc.edu, 919-962-9786	https://www1.rarediseasesnetwork.org/cms/gdmcc/	Primary ciliary dyskinesia, primary immunedeficiencies, pseudohypoaldosteronism, nontuberculous mycobacterium pulmonary disease, cystic fibrosis, idiopathic bronchiectasis	Both	534 (Sep 2012)	NR[c]	ON, QC
Genetic of Intellectual Disability and Autism Spectrum Disorders (GENiDA)[d]	Université de Strasbourg	Pauline Burger burgerp@igbmc.fr and Jean-Louis Mandel jlmandel@igbmc.fr	https://genida.unistra.fr	Koolen-de Vries syndrome	Both	276 (Mar 2024)	< 6 (Mar 2024)	Does not capture

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
				Kleefstra syndrome	Both	219 (Mar 2024)	7 (Mar 2024)	Does not capture
				DDX3X syndrome	Both	63 (Mar 2024)	< 6 (Mar 2024)	Does not capture
				KBG syndrome	Both	56 (Mar 2024)	< 6 (Mar 2024)	Does not capture
				MED13L syndrome	Both	47 (Mar 2024)	< 6 (Mar 2024)	Does not capture
				MECP2 duplication syndrome	Both	46 (Mar 2024)	< 6 (Mar 2024)	Does not capture
				SETD5 syndrome	Both	37 (Mar 2024)	< 6 (Mar 2024)	Does not capture
				DYRK1A syndrome	Both	33 (Mar 2024)	< 6 (Mar 2024)	Does not capture
				White-Sutton syndrome	Both	32 (Mar 2024)	< 6 (Mar 2024)	Does not capture
				Wiedemann-Steiner syndrome	Both	32 (Mar 2024)	< 6 (Mar 2024)	Does not capture
Global Angelman Syndrome Registry	Unclear	curator@angelmanregistry.info	https://www.angelmanregistry.info/	Angelman syndrome	Both	2,333 (Unclear)	115 (Unclear)	NR[c]
Global Dystonia Registry	Invitae	Unclear	https://www.globaldystoniaregistry.org/	Dystonia	NR[c]	6,500 (2024)	NR[c]	NR[c]

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
Global Mucopoly-saccharidosis type I (MPS 1) Registry	Sanofi	Contact-Us@sanofi.com and help@MPSIRegistry.com	https://www.mps1disease.com/get-support/mps1-registry	Mucopoly-saccharidosis type I syndromes	Both	1,500 (May 2023)	NR[c]	BC, AB, MB, ON, QC, NB
Global Patient Registry for Smith-Kingsmore Syndrome	Coordination of Rare Diseases at Sanford; Smith-Kingsmore Syndrome Foundation	Sarah Lepore Sarah.lepore@smithkingsmore.org	https://smithkingsmore.org/patient-registry/	Smith-Kingsmore Syndrome	Both	80 (Mar 2024)	< 6 (Mar 2024)	All provinces/ territories
Global Prader-Willi Syndrome (PWS) Registry	National Organization for Rare Disorders; Foundation for Prader-Willi Research	Aliza Fink aliza.fink@nord.com and Theresa Strong jessica.bohonowych@fpwr.org	https://pwsregistry.org/	Prader-Willi Syndrome	Both	1,936 (Mar 2024)	179 (Mar 2024)	BC, AB, ON, QC, NB, NS
GNAO1 International Registry	Invitae, The Bow Foundation	Emily@bowfoundation.org	https://connect.invitae.com/en/org/gnao1	GNAO1-related neurodevelop-mental disorders	Both	63 (2020)	< 6 (2020)	NR[c]
Hyperinsulinism International Global Registry (HIGR)	Congenital Hyperinsulinism International	Lauren Lopez llopez@congenitalhi.org and Tai Pasquini tpasquini@congenitalhi.org	https://congenitalhi.org/higlobalregistry/	Congenital hyperinsu-linism	Both	500 (Mar 2024)	20 (Mar 2024)	BC, SK, ON, QC
International Diffuse intrinsic pontine glioma (DIPG)/ diffuse	Cincinnati Children's Hospital Medical Center (CCHMC)	referrals@dipgregistry.org	https://www.dipgregistry.org/	Diffuse intrinsic pontine	Both	1,100 (2018)	NR[c]	NR[c]

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
midline glioma (DMG) Registry				glioma, diffuse midline glioma				
International Fabry Registry	Genzyme (a Sanofi company)	Contact-Us@sanofi.com	https://clinicaltrials.gov/study/NCT00196742	Fabry disease	Both	9,000 (Jul 2023)	NR[c]	BC, AB, MB, ON, QC, NB, NS, NL
International Family Registry for Centronuclear and Myotubular Myopathies	Myotubular Trust	connect@joshuafrase.org	https://mtmcnmregistry.org/	Centronuclear and myotubular myopathies	Both	444 (2023)	7 (2023)	NR[c]
International Fanconi Anemia Registry	The Rockefeller University	Agata Smogorzewska MDPhD Asmogorzewska@rockefeller.edu	https://lab.rockefeller.edu/smogorzewska/ifar/	Fanconi anemia	Both	NR[c]	NR[c]	NR[c]
International Fragile X Premutation Registry	UC Davis MIND Institute, National Fragile X Foundation	hilary@fragilex.org (202) 747-6207	https://fragilex.org/our-research/projects/premutation-registry/	Fragile X syndrome	Adult	747 (2022)	16 (2022)	NR[c]
International Gaucher Registry	Unclear	Dr Aneal Khan khaa@ucalgary.ca and Contact-Us@sanofi.com	https://www.gaucherdisease.org/blog/medical-history-international-gaucher-registry/	Gaucher disease, cerebroside lipidosis syndrome, glucocerebrosidase deficiency disease, glucosylceramide beta-glucosidase	NR[c]	NR[c]	NR[c]	AB

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
				deficiency disease				
International GNE Myopathy Registry	John Walton Muscular Dystrophy Research Centre at Newcastle University	gnem@newcastle.ac.uk	https://www.gne-registry.org/	Hereditary inclusion body myopathy, quadriceps-sparing myopathy, Nonaka myopathy, inclusion body myopathy type 2	Adult	269 (Oct 2016)	< 6 (Oct 2016)	ON
International Hereditary Thrombotic Thrombocy-topenic Purpura Registry	Hematology and Central Hematological Laboratory, Inselspital Bern University Hospital, University of Bern	Johanna A. Kremer Hovinga johanna.kremer@insel.ch and Marissa Schraner marissa.schraner@insel.ch	https://ttpregistry.net/	Hereditary thrombotic thrombocy-topenic purpura	Both	265 (Mar 2024)	< 6 (Mar 2024)	AB, ON
International Kawasaki Disease Registry (IKDR)	Unclear	Brian W. McCrindle brian.mccrindle@sickkids.ca	No website identified	Kawasaki disease	Both	NR[c]	NR[c]	ON, QC
International LGDA Registry for Complex Lymphatic Anomalies	Lymphangio-matosis & Gorham's Disease Alliance (LGDA)	coordinator@lgdaregistry.org	https://lgdalliance.org/patients-caregivers/patient-registry.html	Lymphatic anomalies	Both	464 (2014)	NR[c]	NR[c]

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
International Niemann-Pick Disease Registry	International Niemann Pick Disease Registry; OpenApp Ltd, Dublin, Ireland	Conan Donnelly conan.donnelly@inpdr.org	https://inpdr.org/	Niemann-Pick Type C	Both	330 (Mar 2024)	9 (Mar 2024)	ON
				Acid sphingo-myelinase deficiency	Both	100 (Mar 2024)	< 6 (Mar 2024)	ON
International Pachyonychia Congenita Research Registry[d]	Pachyonychia Congenita Project	Janice Schwartz jan.schwartz@pachyonychia.org and Holly Evans holly.evans@pachyonychia.org	https://www.pachyonychia.org/patient-registry/	K6a, K6b, K6c, K16, K17 Pachyonychia Congenita	Both	1,190 (Mar 2024)	41 (Mar 2024)	BC, AB, SK, ON, QC
				TRPV3/ Olmsted Syndrome	Both	17 (Mar 2024)	0 (Mar 2024)	All provinces/ territories
				DSG1-desmoglien	Both	22 (Mar 2024)	0 (Mar 2024)	BC
				DSP-desmoplakin	Both	< 6 (Mar 2024)	0 (Mar 2024)	All provinces/ territories
				GJB2-Connexin 26	Both	< 6 (Mar 2024)	0 (Mar 2024)	All provinces/ territories
				GJB6-Connexin 30	Both	33 (Mar 2024)	0 (Mar 2024)	All provinces/ territories

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
				RHBDF2-Tylosis with esophageal cancer	Both	< 6 (Mar 2024)	0 (Mar 2024)	All provinces/ territories
				WNT10	Both	< 6 (Mar 2024)	0 (Mar 2024)	All provinces/ territories
				K1	Both	< 6 (Mar 2024)	0 (Mar 2024)	All provinces/ territories
				K9	Both	12 (Mar 2024)	0 (Mar 2024)	All provinces/ territories
International Pediatric Acute-Onset Neuro-psychiatric syndrome (PANS) Registry	University of Washington	Erin Masterson emaster@uw.edu and info@pansregistry.org	https://pansregistry.org	PANS, PANDAS	Both	1,823 (2024)	NR[c]	NR[c]
International Paroxysmal Nocturnal Hemoglobinuria (PNH) Registry[f]	Alexion Pharma-ceuticals[h]	Dr. Jeff Szer jeff.szer@mh.org.au and Dr. Christopher Patriquin christopher.patriquin@uhn.ca	https://pnh.iamrare.org	Paroxysmal nocturnal hemoglo-binuria	Both	4,000 (Mar 2024)	205 (Mar 2024)	BC, AB, ON, QC, NS, NL
International Pyridoxine-dependent epilepsy (PDE) Registry	PDE Consortium	PDE@amsterdamumc.nl	https://www.pdeonline.org/pderegistry.html	Pyridoxine-dependent epilepsy	Both	130 (2021)	NR[c]	BC

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
International Pompe Registry	Sanofi Genzyme	Contact-Us@sanofi.com	https://www.lumizyme.com/patients/resources/the_pompe_registry	Pompe disease	Both	1,753 (Jul 2017)	NR[c]	BC, AB, MB, ON, QC, NB
International Registry For Pediatric Systemic Vasculitis (Pedvas) Initiative	BC Children's Hospital	Dr. David Cabral dcabral@cw.bc.ca and Dr. Kelly Brown kbrown@bcchr.ca	No website identified, information about registry on https://www.bcchr.ca/rheumatology/our-current-research	Granulomatosis with polyangiitis, microscopic polyangiitis, eosinophilic granulomatosis with polyangiitis, polyarteritis nodosa, Takayasu's arteritis, Deficiency of Adenosine Deaminase 2, undifferentiated vasculitis	NR[c]	NR[c]	NR[c]	NR[c]
International Registry of Acute Aortic Dissection	University of Michigan	Elise Woznicki elisew@umich.edu and Kim Eagle keagle@umich.edu	https://www.iradonline.org/home	Acute aortic dissection	Adult	14,356 (Mar 2024)	575 (Mar 2024)	AB, ON
International SCN8A Registry Research Study	Unclear	scn8a.info@gmail.com and Michael Hammer	https://scn8a.net/	SCN8A epilepsy	Pediatric	381 (Dec 2021)	NR[c]	NR[c]

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
International Study Group of Pediatric Pancreatitis: In search for a cure: INSPPIRE	University of Iowa	Aliye Uc: 319-384-6032 aliye-uc@uiowa.edu	https://medicine.uiowa.edu/pediatrics/research/pediatric-centers-and-programs/insppire-pediatric-pancreatitis-research-project	Pediatric pancreatitis	Pediatric	867 (Nov 2022)	NR[c]	ON, QC
Immune thrombocytopenia (ITP) Natural History Study Registry	Platelet Disorder Support Association (PDSA); NORD	Jennifer DiRaimo jdiraimo@pdsa.org and Caroline Kruse ckruse@pdsa.org	https://itpstudy.iamrare.org/	Primary ITP	Both	2,345 (Mar 2024)	89 (Mar 2024)	All provinces/ territories
				Secondary ITP	Both	45 (Mar 2024)	< 6 (Mar 2024)	All provinces/ territories
				Inherited platelet disorders	Both	7 (Mar 2024)	< 6 (Mar 2024)	All provinces/ territories
				Other auto-immune conditions co-existing with ITP	Both	29 (Mar 2024)	< 6 (Mar 2024)	All provinces/ territories
				Other comorbid conditions co-existing with ITP	Both	152 (Mar 2024)	14 (Mar 2024)	All provinces/ territories

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
KIF1A Associated Neurologic Disorder (KAND) Natural History Study	Boston Children's Hospital	Wendy Chung wendy.chung@childrens.harvard.edu	https://www.kif1a.org/research/natural-history-study/	KAND	Both	150 (Mar 2024)	< 6 (Mar 2024)	All provinces/ territories
Leber hereditary optic neuropathy (LHON) Data Collection Program	RARE-X, LHON, LHON Canada, LHON Society	support@rare-x.org, (716) 427-2739	https://lhon.rare-x.org/	Leber hereditary optic neuropathy	Both	NR[c]	NR[c]	NR[c]
Life Raft Group Gastrointestinal stromal tumours Patient Registry	Life Raft Group	info@liferaftgroup.org	https://liferaftgroup.org/patient-registry/	Gastrointes-tinal stromal tumours	Both	2,441 (2021)	63 (Unclear)	NR[c]
Mito-SHARE	United Mito-chondrial Disease Foundation	Philip Yeske philip.yeske@umdf.org and Nicole Wilson nicole@umdf.org	http://www.umdf.org/registry	Mitochondrial disease	Both	1,800 (Mar 2024)	35 (Mar 2024)	BC, AB, SK, MB, ON, QC, NB
Moebius Syndrome Foundation Contact Registry	Moebius Syndrome Foundation Contact Registry	info@moebiussyndrome.org	https://moebiussyndrome.org/membership-page/	Moebius syndrome	Both	NR[c]	NR[c]	NR[c]
Myotubular and Centronuclear Myopathies Patient Registry	John Walton Muscular Disease Research Centre, Newcastle University	Julie Bohill julie.bohill@ncl.ac.uk and Chiara Marini Betollo chiara.marini-betollo@ncl.ac.uk	https://mtmcnmregistry.org/	Myotubular Myopathy	Both	275 (Mar 2024)	6 (Mar 2024)	All provinces/ territories

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
				Centronuclear Myopathies	Both	103 (Mar 2024)	< 6 (Mar 2024)	SK
The Nephrotic Syndrome Study Network: NEPTUNE	Rare Diseases Clinical Research Network Patient Contact Registry	NEPTUNE-STUDY@umich.edu; Heather Reich	https://www.neptune-study.org/	Nephrotic syndrome, Alport syndrome	Both	1,100 (2022)	NR[c]	ON
New-Onset Refractory Status Epilepticus Prospective Observational Study Registry	Norse Institute	Nicolas Gaspard nicolas.gaspard@erasme.ulb.ac.be and Lawrence Hirsch lawrence.hirsch@yale.edu	https://www.norseinstitute.org/norse-registry-2	New-onset refractory status epilepticus, febrile infection-related epilepsy syndrome	Both	NR[c]	NR[c]	BC, MB, ON
NORSE/ FIRES Family Registry	NORSE Institute, Western University	Teneille Gofton teneille.gofton@lhsc.on.ca	https://www.norseinstitute.org/norse-registry-2	NORSE (new-onset refractory status epilepticus), FIRES (febrile infection-related epilepsy syndrome)	Both	NR[c]	NR[c]	ON
North American Diamond Blackfan Anemia Registry (DBAR)	Cohen Children's Medical Hospital in New York	Eva Atsidaftos eatsidaf@nshs.edu; DBARegistry@northwell.edu	https://dbafoundation.org/families-and-individuals/dba-registry/	Diamond Blackfan anemia	Both	900 (Apr 2023)	NR[c]	NR[c]

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
North American Malignant Hyperthermia Registry	University of Florida	agunnett@anest.ufl.edu, 888-274-7899	https://anest.ufl.edu/namhr/	Malignant hyperthermia	Both	725 (Mar 2018)	NR[c]	ON
North American Mitochondrial Disease Consortium	Columbia University	Dr. Michio Hirano mh29@cumc.columbia.edu	https://www1.rarediseasesnetwork.org/cms/namdc/	Mitochondrial myopathy	Both	500 (Mar 2024)	75 (Mar 2024)	ON
Primary Ciliary Dyskinesia (PCD) Foundation Registry	PCD Foundation	Carey Kauffman cakauffman@pcdfoundation.org and Michele Manion mmanion@pcdfoundation.org	https://pcdfoundation.org/registry/	Primary ciliary dyskinesia	Both	492 (Mar 2024)	114 (Mar 2024)	ON, QC
Pediatric Cardiomyopathy Registry	Children's Cardiomyopathy Foundation	info@childrenscardiomyopathy.org	https://www.childrenscardiomyopathy.org/pages/physician-resources/pediatric-cardiomyopathy-registry/	Cardiomyo-pathy	Pediatric	3,500 (Unclear)	90 (2023)	AB
Paroxysmal nocturnal hemoglobinuria (PNH) Registry	Unclear	medinfo@alexion.com	https://pnhregistry.com/data-collection	Paroxysmal nocturnal hemoglobi-nuria	Both	5,700 (Oct 2022)	107 (Jan 2018)	BC, AB, ON, QC, NS, NL
Primary Immune Deficiency Treatment Consortium	The Rare Diseases Clinical Research Network Patient Contact Registry	elizabeth.dunn@ucsf.edu	https://pidtc.rarediseasesnetwork.org/our-consortium	Primary immune deficiencies	Both	900 (2018)	NR[c]	BC, AB, MB, ON, QC

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
Primary sclerosing cholangitis (PSC) Partners Patient Registry	PSC Partners Seeking Cure (US)	Rachel Gomel, registrycoordinator@pscpartners.org	https://pscpartners.org/about/participate/patient-registry.html	Primary sclerosing cholangitis	Both	2,595 (Unclear)	NR[c]	BC, AB, SK, MB, ON, QC, NS
Rare Brain Tumor Consortium Global Registry	Huang Lab, The Hospital for Sick Children	rbt.consortium@sickkids.ca and Mei Lu meilu@sickkids.ca	https://lab.research.sickkids.ca/annie-huang/rbtc/about-rbtc/	Rare brain tumours[g]	Pediatric[h]	1,904 (2020)	NR[c]	BC, AB, SK, MB, ON, QC, NS
RARE-X CHD2 - Data Collection Program[d]	RARE-X; The Coalition To Cure CHD2	Zohreh Talebizadeh zohreh.talebizadeh@globalgenes.org	https://chd2.rare-x.org/	LHON (Leber hereditary optic neuropathy), WSS (Wiedemann–Steiner syndrome), CHD2, STXBP1, Koolen-de Vries Syndrome, FOXP1 (forkhead box protein P1), Usher Syndrome, CACNA1A (calcium voltage-gated channel subunit	Both	155 (Dec 2022)	6 (Dec 2022)	NR[c]

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
				alpha1 A), Huntington's, Pompe				
Simons Searchlight[d]	Boston Children's Hospital; Geisinger	Dr. Wendy Chung coordinator@ simonssearchlight .org and Dr. Cora Taylor coordinator@ simonssearchlight.org	https://www .simonssearchlight .org/	16p11.2 deletion syndrome	Both	275 (Mar 2024)	16 (Mar 2024)	All provinces/ territories
				16p11.2 duplication syndrome	Both	199 (Mar 2024)	8 (Mar 2024)	All provinces/ territories
				SCN2A-Related Disorders	Both	235 (Mar 2024)	7 (Mar 2024)	All provinces/ territories
				STXBP1 Encephalo-pathy	Both	217 (Mar 2024)	10 (Mar 2024)	All provinces/ territories
				CTNNB1-Related Syndrome	Both	205 (Mar 2024)	11 (Mar 2024)	All provinces/ territories
				PPP2R5D-Related Syndrome	Both	185 (Mar 2024)	10 (Mar 2024)	All provinces/ territories
				SLC6A1-Related Syndrome	Both	158 (Mar 2024)	< 6 (Mar 2024)	All provinces/ territories

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
				SYNGAP1-Related Syndrome	Both	141 (Mar 2024)	< 6 (Mar 2024)	All provinces/ territories
				1q21.1 duplication syndrome	Both	90 (Mar 2024)	< 6 (Mar 2024)	All provinces/ territories
				GRIN2B-Related Syndrome	Both	132 (Mar 2024)	7 (Mar 2024)	All provinces/ territories
The Duchenne Registry	Parent Project Muscular Dystrophy	Ann Martin ann@parentprojectmd.org; coordinator@duchenneregistry.org	https://www.duchenneregistry.org/about-the-registry/	Duchenne Muscular Dystrophy	Both	4,395 (Mar 2024)	150 (Mar 2024)	BC, AB, SK, MB, ON, QC, NB, PE, NS, NL, YT
				Becker Muscular Dystrophy	Both	892 (Mar 2024)	24 (Mar 2024)	BC, AB, ON, QC
The International Registry and Natural History Study for Adaptor-Protein 4- related Hereditary Spastic Paraplegia	Boston Children's Hospital, Cure AP-4	AP4HSP.Research@childrens.harvard.edu	https://cureap4.org/research.php	Adaptor-Protein 4-associated hereditary spastic paraplegia	Both	400 (2023)	< 6 (Unclear)	QC
The International Replication Repair Deficiency Consortium (IRRDC)	The Hospital for Sick Children	Uri Tabori uri.tabori@sickkids.ca and replication.repair@sickkids.ca	https://replicationrepair.ca/	Constitutional Mismatch Repair Deficiency Syndrome,	NR[c]	NR[c]	NR[c]	NR[c]

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
				Other replication repair deficiency syndromes				
The International Schwanno-matosis Database	Unclear	Dr. Allan Belzberg Abelzbe1@jhmi.edu	http://sid2011.squarespace.com/	Schwanno-matosis	NR[c]	385 (2016)	NR[c]	BC
The North American Antiepileptic Drug Pregnancy Registry	Harvard Medical School	1-888-233-2334	https://www.aedpregnancyregistry.org/	Epilepsy	Both	10,215 (2023)	432 (2023)	BC, AB, SK, MB, ON, QC, NB, NS, NL
The Progeria Research Foundation International Registry	The Progeria Research Foundation	info@progeriaresearch.org	https://www.progeriaresearch.org/international-registry-2/	Progeria, other possible progeroid syndromes	Pediatric	364 (Sep 2022)	NR[c]	SK, MB
Rare Diseases Clinical Research Network Patient Contact Registry	Rare Diseases Clinical Research Network	support@rdcrn.org	https://www.rarediseasesnetwork.org/registry	Several rare diseases[i]	Adult	8,861 (2012)	443 (2012)	NR[c]
The Scleroderma Patient-centred Intervention Network (SPIN) Cohort	McGill University	Dr. Brett Thombs brett.thombs@mcgill.ca, spingeneral@gmail.com	https://spinsclero.com/en/cohort	Scleroderma	Adult	1,800 (2021)	NR[c]	BC, AB, MB, ON, QC, NS

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
The US Immuno-deficiency Network	USIDNET Consortium	contact@USIDNET.org	https://usidnet.org/about-the-registry/enrolling-institutions/	Chronic granulo-matous disease, X-linked agamma-globulinemia, common variable immune deficiency, X-linked hyper IgM, leukocyte adhesion deficiency, severe combined immune deficiency, DiGeorge syndrome, Wiskott-Aldrich syndrome	Both	5,000 (2022)	NR[c]	QC
TREAT-NMD global registry network	TREAT-NMD	registries@treat-nmd.com and John McKenna john.mckenna@treat-nmd.com	https://www.treat-nmd.org/what-we-do/global-registry-network/	Neuro-muscular diseases (e.g., spinal muscular atrophy)	Both	88,832 (2024)	5,600 (2023)	All provinces (BC, AB, SK, MB, ON, QC, NB, PE, NS, NL

Registry name	Lead organization(s)	Lead contact name and email	Registry website[a]	Disease(s)	Age group[b]	Number of patients internationally (date retrieved)	Number of patients in Canada (date retrieved)[b]	Coverage in Canada[b]
Tuberous Sclerosis Complex Natural History Database and Biorepository (TSC Alliance)	TSC Alliance	Elizabeth Cassidy ecassidy@tscalliance.org and Steve Roberds sroberds@tscalliance.org	https://www.tscalliance.org/researchers/natural-history-database/	Tuberous Sclerosis Complex (TSC)	Both	2,704 (Mar 2024)	76 (Mar 2024)	AB, QC
				Lymphangioleiomyomatosis (LAM)	Both	50 (Mar 2024)	< 6 (Mar 2024)	QC
Unique Families Global Network	Unique	info@rarechromo.org and Sarah Wynn sarah@rarechromo.org	https://rarechromo.org/	Rare chromosome disorders	Both	27,789 (2024)	1,034 (2024)	NR[c]
Urea Cycle Disorders Consortium	The Rare Diseases Clinical Research Network Patient Contact Registry	jseminar@childrensnational.org	https://ucdc.rarediseasesnetwork.org/	Urea cycle disorders[j]	Both	NR[c]	NR[c]	ON
Vasculitis Clinical Research Consortium	The Rare Diseases Clinical Research Network Patient Contact Registry	Unclear	https://www1.rarediseasesnetwork.org/cms/vcrc	Multiple types of vasculitis[k]	Both	3,151 (Dec 2011)	NR[c]	BC, AB, ON, QC
VISION Registry	Melanoma Research Foundation	cureom@melanoma.org	https://melanoma.org/visionregistry/	Ocular melanoma	Both	421 (2023)	34 (2023)	BC, NL

AB = Alberta; Apr = April; BC = British Colombia; Dec = December; Feb = February; Jan = January; Jul = July; Mar = March; MB = Manitoba; NB = New Brunswick; NL = Newfoundland and Labrador; Nov = November; NR = not reported or not available; NS = Nova Scotia; NT = Northwest Territories; NU = Nunavut; Oct = October; ON = Ontario; PE = Prince Edward Island; QC = Quebec; Sep = September; SK = Saskatchewan; YT = Yukon.

[a]Websites sourced from publicly available information.

[b]Among registries captured from the literature search, information is available overall, and not per disease.

[c]Not reported or available because of reliance on information available in a literature and/or public search or partial completion of registry holder survey. If the registry holder did not complete information, literature and/or public search information was inputted if available.

[d]During the survey with registry holders, registries with more than 10 rare diseases were instructed to enter data for the 10 most prevalent diseases at this stage.

[e]Registry has a total of 6204 participants in our registry as have registrants who participate on two levels: contact or research registrants. Additionally, FTD disorders is a complex set of diseases, therefore, participants may display symptoms from multiple FTD disorders on the spectrum. It is possible a participant may not know their diagnosis when joining the registry. The registry is currently undergoing a platform upgrade and is implementing strategies to better understand diagnoses and disease progression of its participants.

[f]This registry (funded by Alexion Pharmaceuticals) will soon be rolling into an academic-run broader registry under the auspices of the International PNH Interest Group (IPIG) which will permit all patients with PNH regardless of therapeutic (e.g., those not made by Alexion) to join to be broader and more representative of the current PNH treatment landscape.

[g]Rare Brain Tumor Consortium Global collects data on rare brain tumours, including atypical teratoid rhabdoid tumours, embryonal tumours with multilayered rosettes, pineoblastomas, embryonal tumours with abundant neuropil and true rosettes, supratentorial primitive neuroectodermal tumours, central nervous system embryonal tumours, medulloepitheliomas, ependymoblastomas, and other rare embryonal tumours.

[h]The Rare Brain Tumor Consortium Global Registry includes participants aged 0 to 21 years old.

[i]Acute intermittent porphyria, adrenoleukodystrophy and adrenomyeloneuropathy, Aicardi-Goutières syndrome, Alexander disease, *ALG12*-congenital disorder of glycosylation, *ALG13*-congenital disorder of glycosylation, *ALG3*-congenital disorder of glycosylation, *ALG6*-congenital disorder of glycosylation, *ALG8*-congenital disorder of glycosylation, Alpers syndrome, Alport syndrome, amyotrophic lateral sclerosis and related disorders, aminoglycoside-induced deafness, amyotrophic lateral sclerosis, amyotrophic lateral sclerosis-frontotemporal dementia, aortitis, arginase deficiency, argininosuccinate lyase deficiency, argininosuccinate synthetase deficiency, ATP6AP1-congenital disorder of glycosylation, ATP6AP2-congenital disorder of glycosylation, Barth syndrome, Behcet disease, biopterin synthesis or recycling defects, blepharospasm and Meige syndrome, brain vascular malformation, brittle bone disorders and osteogenesis imperfecta, carbamyl phosphate synthetase deficiency, cerebral cavernous malformations, cervical dystonia, Charcot-Marie-Tooth disease CMT1A, Charcot-Marie-Tooth disease CMT1B, Charcot-Marie-Tooth disease CMT2A, Charcot-Marie-Tooth disease CMT4, Charcot-Marie-Tooth disease CMTX, chronic granulomatous disease, chronic progressive external ophthalmoplegia, citrullinemia II, central nervous system vasculitis, complex I deficiency, complex II deficiency, complex III deficiency, complex IV deficiency, complex V deficiency, congenital disorders of glycosylation, congenital erythropoietic porphyria, congenital infections, coenzyme deficiency, cryoglobulinemic vasculitis, cutaneous vasculitis, cystic fibrosis, cytomegalovirus, developmental synaptopathies, diabetes and deafness, dihydropteridine reductase deficiency, DNAJC12 deficiency, DPAGT1-congenital disorder of glycosylation, drug-induced vasculitis, dystonia, *EDEM3*-congenital disorder of glycosylation, encephalomyopathy, encephalopathy, enterovirus, eosinophilic colitis, eosinophilic enteritis, eosinophilic esophagitis, eosinophilic gastritis, eosinophilic gastrointestinal disorders, eosinophilic granulomatosis with polyangiitis, erythropoietic protoporphyria, Fabry disease, familial bilateral striatal necrosis, focal and segmental glomerulosclerosis, frontotemporal dementia, FUT8-congenital disorder of glycosylation, *GALNT2*-congenital disorder of glycosylation, generalized dystonia, genetic mucociliary disorders, giant cell (temporal) arteritis, granulomatosis with polyangiitis, GTP cyclohydrolase 1 deficiency (recessive form), hepatocerebral disease, hepatoerythropoietic porphyria, hereditary coproporphyria, hereditary hemorrhagic telangiectasia, hereditary spastic paraplegia, herpes simplex virus, Hunter syndrome, Hurler-Scheie syndrome, Hurler syndrome, hyperphenylalaninemia, idiopathic aortitis, idiopathic bronchiectasis, IgA vasculitis, inherited neuropathies, Kearns-Sayre syndrome, Krabbe disease, laryngeal dystonia, Leber hereditary optic neuropathy, Leber hereditary optic neuropathy-plus, Leigh syndrome, leukodystrophies, leukoencephalopathy, limb dystonia, lysosomal disorders, *MAN1B1*-congenital disorder of glycosylation, *MAN2B2*-congenital disorder of glycosylation, Maroteaux-Lamy syndrome, maternally inherited Leigh syndrome, membranous nephropathy, metachromatic leukodystrophy, microscopic polyangiitis, minimal change disease, mitochondrial disease, mitochondrial DNA depletion syndrome, mitochondrial encephalomyopathy lactic acidosis with stroke-like episodes, mitochondrial neurogastrointestinal encephalomyopathy, Morquio syndrome, MPI-congenital disorder of glycosylation, mucopolysaccharidoses, multifocal dystonia, multiple deletions of mitochondrial DNA, multiple respiratory chain enzyme deficiencies, multisystem proteinopathy, myasthenia gravis, myoclonus epilepsy ragged-red fibres, N-acetylglutamate synthase deficiency, nephrotic syndrome, neuropathy, ataxia, and retinitis pigmentosa syndrome, NGLY1 deficiency, nontuberculous mycobacterium pulmonary disease, ocular myasthenia, ornithine transcarbamylase deficiency, ornithine translocase deficiency, other known Charcot-Marie-Tooth peripheral neuropathy, other unknown Charcot-Marie-Tooth peripheral neuropathy, Pearson syndrome, Pelizaeus-Merzbacher disease, peripheral neuropathy, *PGAP3*-congenital disorder of glycosylation, *PGM1*-congenital disorder of glycosylation, Phelan-McDermid syndrome, phenylalanine hydroxylase deficiency, phenylketonuria, *PIGA*-congenital disorder of glycosylation, *PIGN*-congenital disorder of glycosylation, *PIGT*-congenital disorder of glycosylation, *PMM2*-congenital disorder of glycosylation, polyarteritis nodosa, Pompe disease, porphyria cutanea tarda, porphyrias, primary ciliary dyskinesia, primary immune deficiency disorders, primary immune regulatory disorders, primary lateral sclerosis, progressive muscular atrophy, pseudohypoaldosteronism, PTEN hamartoma tumour syndrome, pterin-4a-carbinolamine dehydratase deficiency, pyruvate dehydrogenase complex deficiencies, Sanfilippo syndrome A, Sanfilippo syndrome B, Sanfilippo syndrome C, Sanfilippo syndrome D, Scheie syndrome, segmental dystonia, sensory ataxia neuropathy dysarthria and ophthalmoplegia, severe combined immunodeficiency, *SLC35A2*-congenital disorder of glycosylation, *SLC35C1*-congenital disorder of glycosylation, *SLC39A8*-congenital disorder of glycosylation, Sly syndrome, *SRD5A3*-congenital disorder of glycosylation, *SSR4*-congenital disorder of glycosylation, *STT3A*-congenital disorder of glycosylation, Sturge-Weber syndrome, Takayasu arteritis, tuberous sclerosis complex, urea cycle disorders, urticarial vasculitis, variegate porphyria, vasculitis disorders, *VMA21*-congenital disorder of glycosylation, Wiskott-Aldrich syndrome, and X-linked protoporphyria.

[j]N-acetylglutamate synthase (NAGS) deficiency, carbamoyl-phosphate synthase 1 deficiency, ornithine transcarbamylase deficiency, argininosuccinate synthase deficiency (also known as citrullinemia type I), citrin deficiency (also called citrullinemia type II), argininosuccinate lyase deficiency (also known as argininosuccinic aciduria), arginase deficiency (also known as hyperargininemia), and ornithine translocase deficiency (also known as hyperornithinemia-hyperammonemia-homocitrullinuria or HHH syndrome).

[k]Aortitis, Behcet disease, central nervous system vasculitis, cryoglobulinemic vasculitis, cutaneous vasculitis, drug-induced vasculitis, eosinophilic granulomatosis with polyangiitis (Churg-Strauss), giant cell (temporal) arteritis, granulomatosis with polyangiitis (Wegener), idiopathic aortitis, IgA vasculitis (Henoch-Schonlein purpura), microscopic polyangiitis, polyarteritis nodosa, Takayasu arteritis, urticarial vasculitis, and other types of vasculitis.

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