RareKids CAN

Last Updated: April 30th, 2026

RareKids-CAN Rare Disease Registry Resource Environmental Scan Protocol

Background

In 2023, Health Canada reported that approximately one in twelve Canadians are affected by a rare disease, yet effective and affordable disease-modifying treatments are unavailable for many of these disorders.[1] For example, a recent survey from the Canadian Organization for Rare Disorders reported that only 68% of Canadians with a rare disease know of an approved treatment for their condition.[2]

To increase access and affordability of treatments for rare diseases, the Canadian government launched a National Strategy for Rare Diseases in 2023.[1] This investment has inspired several rare disease initiatives, including a focus on improving real-world data and evidence to support rare disease drug regulatory and health technology assessment reviews.[1,3] Patient registries are a potential source of real-world data about rare disease drugs; such registries collect uniform observational data to evaluate specified outcomes for a population defined by one or more rare diseases.[4] As one component of a multi-faceted strategy to assess and improve the quality of rare disease registries, Canada's Drug Agency recently described 148 rare disease registries that include Canadian patients, 66 of which were developed in Canada.[5] While there is potential for these registries to generate fit-for-purpose real-world evidence about treatment effectiveness for Canadian patients with rare diseases, there are known barriers to the development and maintenance of rare disease registries, for example due to challenges associated with small sample sizes, and a recognized need for greater attention to sustainability, capacity for change, and data quality.[6]

Toward improved quality of rare disease registries, many initiatives and organizations in Canada and internationally have developed resources to support developers, holders, and users of patient registries. The purpose of this environmental scan is to catalogue key resources to provide general and tailored guidance to rare disease registry knowledge users, particularly those based in Canada. Specifically, we aim to support these knowledge users to develop, maintain, and use high-quality patient registries toward generating fit-for-purpose real-world evidence to support decision-making about treatments for rare diseases.

Objective and Design

We aim to identify and map resources in Canada, the US, Europe, and elsewhere focused on rare disease registry guidance, registry platforms, common data elements, data quality/management, interoperability (beyond core data elements), and registry inventories. To achieve this objective, we are conducting an environmental scan.

Inclusion criteria

As is typical in an environmental scan, inclusion criteria are somewhat iterative and allowed to expand and evolve as the environmental scan proceeds and in response to feedback from knowledge users.[7] Initially, to be included, a source (e.g., an initiative or organization) had to be broader than a single rare disease registry or research project and relate to one of the following categories: registry guidance, registry platforms, and common data elements. Although our focus is on rare diseases, many of the resources focus on patient registries more broadly; such resources are considered eligible if they can be applied in a rare disease setting. As the project has evolved, we have expanded the resources to include documents and articles, and added categories to cover resources that include data quality/management, interoperability (beyond common data elements), and registry inventories. These categories of eligible resources are overlapping (e.g., a resource may provide a platform and common data elements; or may provide registry guidance that largely falls within the data quality/management category). We have also expanded to include some disease-specific resources if we found them to be applicable to a broader category.

We will use a range of strategies for data collection throughout this environmental scan. Relevant sources generally fall into two broad types: 1) documents and articles, and 2) initiatives and organizations. There is some overlap between these two types of sources. For example, if an organization develops registry guidance and subsequently produces a published document from this work, the initiative/organization would be considered a source (type 2), and the published document would also be a source (type 1).

Categories

- Registry guidance: Broader category capturing documents/articles and initiatives/organizations that highlight multiple aspects of general registry guidance (i.e., governance, data collection and sharing, ethical and legal issues, planning, recruitment, consent, etc.); or that discuss one aspect of guidance that is not otherwise within one of our categories. Guidance can relate to planning a registry, developing or maintaining a registry, or using a registry; and can be specific to challenges and solutions or outline important considerations or recommendations.
- Registry platform: Computer program/software or initiative/organization that is designed to collect or host uniform observational patient data. This category may also include documents/articles focused on developing, enhancing, or assessing registry platforms.
- Common data elements: Initiatives/Organizations or documents/articles focused on developing, enhancing, assessing, or recommending common data elements for harmonization across rare diseases and patient registries. Resources may include the development of general core outcome sets or measurement tools for rare diseases, or tools specifically focused on patient-reported outcomes.
- Data quality/management: Initiatives/Organizations or documents/articles focussed on developing, enhancing, or assessing data management

protocols and data quality (i.e., completeness, accuracy, consistency, timeliness, etc.). These resources may not be specific to registries (e.g., the FAIR principles) but are relevant to registries.

- Interoperability beyond core data elements: Initiatives/Organizations or documents/articles focused on guidance for developing, enhancing, or assessing methods for data exchange between computer programs/software hosting patient registries.
- Registry inventory: Initiatives/Organizations that have catalogued existing patient registries (does not need to be specific to rare diseases) to create an inventory, catalogue, or list of registries with information being available for each registry in the inventory (e.g., owner, platform, primary contact, disease area, etc.).

Approach to identifying relevant sources

Scanning will be performed in different phases based on an iterative approach and search terms will expand from knowledge user insight. One team member will collect the data, and all resources will be verified by at least one other team member. Based on this data collection approach, our results will not reflect an exhaustive list of resources but rather a curated list that we identify as of potential interest to registry developers, holders, or users interested in rare disease patient registries as a source of real-world data.

Search strategy

- Literature searches (peer-reviewed and grey literature)
  - Sources (to date): Google browser, PubMed, snowball searching (links and references in already-included sources)
    - Google Browser and PubMed Search Terms (to date): patient registry initiatives, registry initiatives, rare disease registry initiatives, rare disease registry standards, rare disease registry guidance, patient registry initiatives in Europe, rare disease registry standards in Canada, patient registry resources, building a patient registry, how to start a patient registry, patient registry governance, data handling governance, common data elements, interoperability for patient registries, rare disease registry platforms, internet based patient registry, rare disease registry software
- Survey of knowledge producers and users affiliated with the RareKids-CAN network
- Key informant, semi-structured interviews, or focus groups
- Person contact (e.g., emails, advisory groups, networks, expert panels, knowledge users, conferences, related knowledge synthesis projects undertaken by team members)
- Other methods (e.g., snowball searching)

Data extracted from documents and articles (type 1)

- Title
- First Author
- Year of Publication

- Journal
- Digital Object Identifier
- PubMed ID
- Document/Article Category
  - Registry guidance
  - Registry platform
  - Common data elements
  - Data quality/management
  - Interoperability (beyond core data elements)
- Description
- Type of resource
  - Rare disease specific
  - Registry specific
  - Generic
- Audience
  - Registry developers/holders
  - Registry users
- Hyperlinks

Data extracted from initiatives and organizations (type 2)

- Initiative/Organization name
- Initiative/Organization category
  - Registry guidance
    - Patient and Family Partners or Patient Organizations involved in guidance development
  - Registry platform
    - Number of registries hosted on the platform
    - Number of rare disease registries hosted on the platform
    - Multilingual platform
    - Canadian-led platform
    - Types of data supported on the platform
    - Data security described on the platform's public website
  - Common data elements
    - Scope of common data elements initiative
    - Patient and Family Partners or Patient Organizations involved in common data elements development
    - Measurement tools described for common data elements
  - Data quality/management
  - Interoperability (beyond core data elements)
  - Registry inventory
    - Inventory updated in the last two years
    - Inventory geographical reach (Note: we have specifically searched on Canadian, American, and European initiatives)
    - Inventory publicly available
    - Inventory self-enrollment available

- Canada's Drug Agency ***Best Practices and Standards to Enhance the Quality of Rare Disease Registries in Canada*** Tag
    - Governance Best Practices and Standards
    - Data Best Practices and Standards
    - Information Technology Infrastructure Best Practices and Standards
    - Not applicable
- Description
- Type of resource
    - Rare disease specific
    - Registry specific
    - Generic
- Year of initiative/organization establishment
- Audience
    - Registry developers/holders
    - Registry users
- Geographical Origin/Host (Note: we have specifically searched on Canadian, American, and European initiatives)
    - Canada
    - United States
    - Europe
    - International
    - Other (specify)
- Current Status (as of January 2026)
    - Active
    - Time-limited
    - Retired
- Hyperlinks
- Files

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References

1. Health Canada. Government of Canada improves access to affordable and effective drugs for rare diseases. March 22, 2023. Accessed September 22, 2024. https://www.canada.ca/en/health-canada/news/2023/03/government-of-canada-improves-access-to-affordable-and-effective-drugs-for-rare-diseases.html
2. Canadian Organization for Rare Disorders (CORD). EXPERIENCES OF RARE DISEASE PATIENTS. 2023. Accessed August 14, 2024. https://www.raredisorders.ca/uploads/Documents/CORD-Rare-Disease-Survey_Full-Report_Feb-2870-2.pdf
3. Canada's Drug Agency. Drugs for Rare Diseases - Rare Disease-Based Registrie. Accessed September 22, 2024. https://www.cda-amc.ca/drugs-rare-diseases-rare-disease-based-registries
4. Gliklich, Leavy, Dreyer. *Registries for Evaluating Patient Outcomes: A User's Guide*. (Gliklich RE, Leavy MB, Dreyer NA, eds.).; 2020. doi:10.23970/AHRQEPCREGISTRIES4
5. Canada's Drug Agency. Understanding the Rare Disease Registries Landscape in Canada. September 4, 2024. Accessed September 22, 2024. https://www.cda-amc.ca/news/understanding-rare-disease-registries-landscape-canada
6. Hageman IC, van Rooij IALM, de Blaauw I, Trajanovska M, King SK. A systematic overview of rare disease patient registries: challenges in design, quality management, and maintenance. *Orphanet J Rare Dis*. 2023;18(1):106. doi:10.1186/S13023-023-02719-0
7. Wilburn A, Vanderpool RC, Knight JR. Peer Reviewed: Environmental Scanning as a Public Health Tool: Kentucky's Human Papillomavirus Vaccination Project. *Prev Chronic Dis*. 2016;13(8). doi:10.5888/PCD13.160165
8. Graham P, Evitts T, Thomas-MacLean R. Environmental scans: How useful are they for primary care research? *Canadian Family Physician*. 2008;54(7):1022. Accessed October 1, 2024. /pmc/articles/PMC2464800/

APPENDIX A. Environmental Scan Guiding Models and Frameworks

- Choo CW. Environmental scanning as information seeking and organizational learning. *Inf Res* 2001;7:1–26.
- Blanken RL, Liff A. *Facing the future: preparing your association to thrive: emerging trends from the ASAE Foundation's ongoing environmental scan research*. Washington, DC: American Society of Association Executives, 1999.
- Wilburn A, Vanderpool RC, Knight JR. Environmental scanning as a public health tool: Kentucky's human papillomavirus vaccination project. *Prev Chronic Dis* 2016;13:E109.
- Graham P, Evitts T, Thomas-MacLean R. Environmental scans: how useful are they for primary care research? *Can Fam Physician* 2008;54:1022–3.
- Rowel R, Moore ND, Nowrojee S. The utility of the environmental scan for public health practice: lessons from an urban program to increase cancer screening. *J Natl Med Assoc* 2005;97:527–34.