

Peer-Reviewed Article

A Review of Genomic Data Holdings in the Atlantic Provinces

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Background: Genetics interacts with environmental, socioeconomic, and health system factors, influencing congenital conditions, medication metabolism, and susceptibility to adverse health conditions. Atlantic Canada includes populations with founder effects, such as those in Newfoundland and Labrador and Acadian populations in Nova Scotia and New Brunswick. Because individuals in the Maritimes have a distinct genetic makeup, their representation in genomic datasets is important for health research. To maximize their value for research and clinical practice, genomic data need to be findable, accessible, interoperable, and reusable (FAIR).

Goal: To identify, assemble, and document genomic datasets collected in the Atlantic provinces. This report provides a foundation for future data ecosystem development in Atlantic Canada and supports the work of Canada's Drug Agency on the Canadian Rare Disease Strategy.

Methods: There were 2 main data sources: published literature and interviews with key informants.

Findings:

- Genomic data are available across Atlantic Canada but fragmented. Research datasets are often held by individual researchers and not in a centralized database. Large datasets do exist with raw data available, mainly in Newfoundland and Labrador. Projects to provide centralized repositories (e.g., Atlantic Cancer Consortium and Atlantic PATH) have been or are being developed.
- Clinical genomic data are currently limited by lack of access to raw genetic data, siloed health systems, and lack of consent for secondary research and linkage of genomic and health administrative data. Efforts to repatriate raw data from third-party sequencers and connect genomic datasets may improve access for research as consent for secondary research and centralized electronic medical records become more common.
- A further limitation is underrepresentation of people of non-European ancestry in Maritime genomic datasets. The Canadian Precision Health Initiative was announced in 2025. It seeks to collect genomic data from Black individuals in Canada, including those living in Nova Scotia.

Key Messages

- The founder effect occurs when an isolated population inherits the genetic and phenotypic traits of a small group of founders, leading to differences from the larger surrounding population over time. Recessive genes and genetic illness can be more prevalent in these groups due to the smaller gene pools.
- Atlantic Canada is home to multiple populations shaped by founder effects, such as those in Newfoundland and Labrador and the Acadian populations in Nova Scotia and New Brunswick. However, the availability of genomic datasets and past genetic research has not yet been mapped.
- This project aimed to identify and summarize the genomic datasets available for populations in Atlantic Canada, with particular attention to the FAIR (findable, accessible, interoperable, and reusable) principles.
- Searches of the literature and communications with researchers indicate there are rich genomic data collected that have been used for secondary research (reusable), which are not necessarily easily findable, accessible, or interoperable because they are held by individual researchers not a centralized repository.

Background

A person's genetic makeup interacts with many facets of environmental, socioeconomic, and health system factors, influencing congenital conditions, medication metabolism, and their predisposition to develop adverse health conditions across the life span. The founder effect occurs when a population becomes isolated and, over generations, its descendants' phenotypic and genotypic traits still resemble the original group of founders, making the population noticeably different from the wider population.¹ Due to the smaller gene pools, recessive genes and genetic illness can be more prevalent in these groups.¹

Atlantic Canada is home to multiple populations shaped by founder effects, such as those in Newfoundland and Labrador and the Acadian populations in Nova Scotia and New Brunswick.^{2,3} The founder populations of Newfoundland and Labrador can be traced back to Irish Catholic and English Protestant settlers, as well as Indigenous Peoples.³ The Acadian population in the Maritimes is thought to descend from approximately 50 French families who settled in Canada before 1650.²

Despite the unique genetic makeup of the population living in Atlantic Canada and the potential prevalence of rare genetic mutations due to founder effects, the availability of genetic datasets and past genetic research has not yet been mapped.

The FAIR (findable, accessible, interoperable, and reusable) principles were developed in 2014 to target issues related to data access and usability. In particular, *findable* requires studies and data to be described with metadata that allow identification and the data can be registered in a searchable resource.

The purpose of this document is to provide a summary report for Canada's Drug Agency (CDA-AMC) that documents the characteristics and specifications of genomic datasets that have been collected in the Atlantic

provinces. This will allow data to be more “findable” in accordance with the FAIR principles and serve as the foundation for subsequent data ecosystem development in Atlantic Canada. In turn, development of that data ecosystem will allow more genomic data to be incorporated into real-world evidence studies, resulting in richer and more insightful analyses to support decision-making regarding approval and listing of pharmaceuticals in the Atlantic provinces.

Project Goal

As a first step to understanding the genomic data parameters of the people living in Atlantic Canada, this project aims to assemble and document the characteristics and specifications of genomic datasets that have been collected in the Atlantic provinces. This summary report will provide a foundation for subsequent data ecosystem development in Atlantic Canada and support the work of CDA-AMC on the Canadian Rare Disease Strategy.

Methods

The approach involved 2 main sources of data: published literature and interviews with key informants.

Literature Review

The literature review was based on information drawn from the bibliographies of genomics researchers and organizations in Atlantic Canada (Newfoundland and Labrador, Prince Edward Island, Nova Scotia, and New Brunswick). As a starting point, the faculty pages of Dalhousie University and Memorial University of Newfoundland were searched for researchers who listed genetics or genomics as an area of interest. From February to April 2025, the names of researchers and genomics centre contacts were also extracted from staff lists and listed partner organizations from genomic research and testing centres in Atlantic Canada (refer to Appendix 3 in the [Supplemental Material](#) document).

Up to the first 100 results (English and French) for each researcher’s bibliography were scanned by title and abstract on Google Scholar and in the University of New Brunswick (UNB) online library catalogue. Articles not referring to human genetics; not including populations from Newfoundland and Labrador, Prince Edward Island, Nova Scotia, or New Brunswick; with fewer than 25 individuals (unless the article stated that they had attempted to include all people with a genetic condition or mutation); or in languages other than English or French were excluded (refer to Appendix 1 in the Supplemental Material document for exclusion criteria). If the inclusion criteria were unclear based on the title and abstract, the full article was obtained when possible. If relevance could not be determined based on the article title and abstract, and the full article could not be obtained through UNB Libraries or other public sources, the article was excluded. If only an abstract could be obtained but there was enough information included to satisfy the criteria, information from the abstract was included in Appendix 2 in the Supplemental Material document and the abstract was noted as the source.

The review process was iterative. Researchers were added as they were identified through included studies. RE conducted the initial screening of the titles and abstracts (refer to Appendix 3 in the Supplemental

Material document for a list of the researchers and organizations searched on Google Scholar and in UNB Libraries catalogue).

Titles were selected based on the title and abstract (or the title only if the abstract was not available on Google Scholar or in the UNB Libraries catalogue). RE and LLC further screened these articles for relevance (refer to Appendix 4 in the Supplemental Material document). Relevant information from the identified articles was extracted. It is included in the table available in Appendix 2 in the Supplemental Material document. Information from the articles was extracted by only 1 reviewer (either RE or LLC) unless the relevance of an article was unclear based on the first reviewer. In these instances, the article was assessed by both RE and LLC for relevance. Due to feasibility constraints, a full systematic review of all published papers was not possible; however, any additional datasets identified through interviews or email correspondence were included, with supplementary follow-up data gathering. This process continued until no additional datasets meeting the criteria were identified.

Informant Interviews

Based on the literature search, we prepared a list of potential contacts, including names, designation, and contact details (this can be made available upon request). In addition to the contacts identified through the literature search, we also relied on existing networks and collaborations (e.g., the Maritime SPOR SUPPORT Unit, Health Data Research Network Canada, Secure Island Data Repository, Health Data Nova Scotia, Newfoundland and Labrador Health Services [NL Health Services], and CDA-AMC) to help identify relevant individuals. A letter of introduction (refer to Appendix 5 in the Supplemental Material document) that described the purpose of the project and requested follow-up was sent to each person identified. Those contacted were also requested to direct us to any additional individuals or organizations working with genomic data in Atlantic Canada, when applicable. Researchers were also asked about additional datasets they were aware of; no additional datasets were identified this way.

Contacted individuals were offered the choice of providing information on genomic datasets or research via email or a structured (virtual) interview. A questionnaire was drafted by the research team to reflect key dimensions of genomic data collection and access that were identified by CDA-AMC. The draft was reviewed for clarity and completeness by 2 individuals involved in the collection and analysis of genomic data in New Brunswick, and adjustments were made based on their feedback (refer to Appendix 6 in the Supplemental Material document).

The questionnaire was divided into 6 parts: source of data, characteristics of the dataset, data access and privacy, data use, data sharing, and data linkage. The online survey was conducted using LimeSurvey, and interviews were conducted through MS Teams. Consent to record and transcribe the interview was collected from all participants before interviewing or surveying them, and interviews were recorded for internal review purposes. Transcripts were reviewed to identify any new datasets and relevant information about these datasets, such as the purpose of the collected data (clinical or research), location of storage, current data custodian, availability of the data for secondary information, and availability of raw data.

In consultation with DataNB's privacy officer, it was determined that a research ethics board (REB) was not required for this work. The purpose of the project was to provide a summary of genomic data resources in Atlantic Canada to support future policy development around data sharing and data standards. The main objective for the interviews was to supplement the information gathered from publicly available resources. Interviewees agreed in advance to provide relevant additional information, either via the questionnaire or an interview, and at the commencement of the interview provided consent to the information they provided being incorporated into the report. Please note that the identities of the correspondents have been suppressed for confidentiality because explicit consent was not provided for in-text citations.

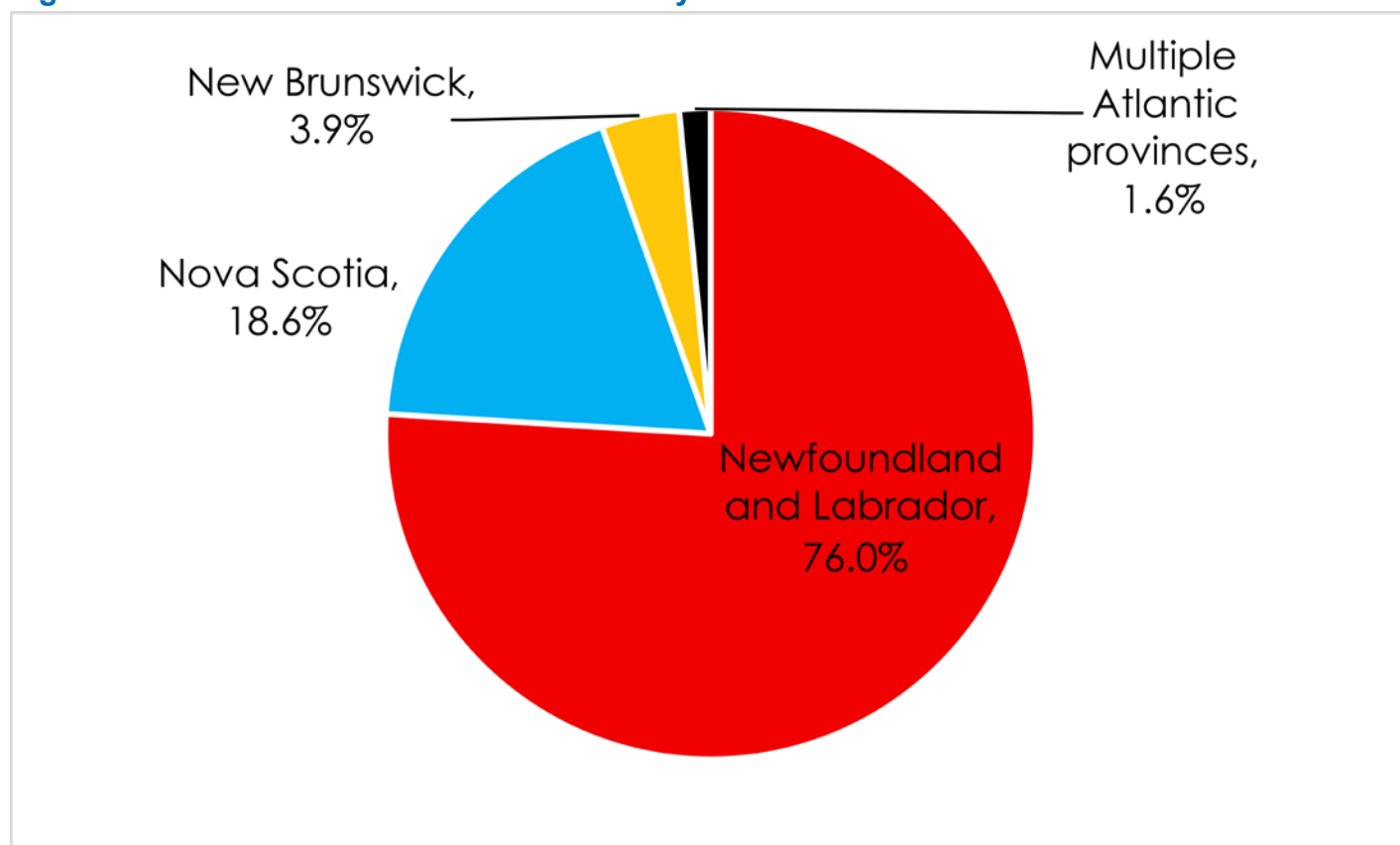
Results

Findings From the Literature Review

In total, 42 researchers and organizations based in Atlantic Canada with published work involving Atlantic Canada genomics studies were identified and searched as described (refer to Appendix 3 in the Supplemental Material document). This was approximately 8,400 titles and abstracts. Of these, **337 titles** were selected based on the titles and abstracts (or the title only if the abstract was not available on Google Scholar or in the UNB Libraries catalogue). Of the 337 titles, a total of **128 articles** were included in our literature review.

Relevant information was extracted and organized by the province of the research participants. This information is presented in Appendix 2 in the Supplemental Material document. The distribution of the included articles by province of the genetic samples is shown in [Figure 1](#). Most articles (76%) included genetic samples from Newfoundland and Labrador.

No relevant articles published by researchers in Prince Edward Island were found. The University of Prince Edward Island REB confirmed that no genomic-related research projects have been submitted for review to date. Residents of Prince Edward Island may be included in studies conducted in Nova Scotia or New Brunswick.

Figure 1: Distribution of Retrieved Articles by Province

Most information was gleaned through communications with researchers. This dataset is described further in the Findings From Surveys and Interviews section of this report.

Newfoundland and Labrador

A total of 98 studies used genetic samples from individuals who originated from Newfoundland and Labrador. Details of these studies are provided in Table 5 in Appendix 2 in the Supplemental Material document. Various conditions and populations were studied, including the general population in Newfoundland and Labrador.³⁻⁵ Conditions studied included:

- cancers,⁶ including colorectal,⁷⁻²⁹ ovarian, and breast³⁰
- autoimmune conditions, including psoriatic arthritis and psoriasis,³¹⁻⁴⁸ spondyloarthritis,⁴⁹⁻⁵⁴ Crohn disease,⁵⁵ and autoinflammatory disease⁵⁶
- rare genetic disorders, including Lynch syndrome,⁵⁷⁻⁶³ Bardet-Biedl syndrome,⁶⁴⁻⁶⁹ Stargardt disease,⁷⁰ genetically linked hearing loss,⁷¹⁻⁷⁷ autosomal-dominant arrhythmogenic right ventricular cardiomyopathy (ARVC),⁷⁸⁻⁸² familial intracranial aneurysm,⁸³ familial and sporadic idiopathic pulmonary fibrosis,⁸⁴ and dominant hereditary spastic ataxia⁸⁵
- other conditions, including osteoarthritis,⁸⁶⁻⁹³ linkage disequilibrium,⁹⁴ food addiction,⁹⁵ and catecholaminergic polymorphic ventricular tachycardia.⁹⁶

It was noted in some of the Newfoundland and Labrador literature that there were plans to develop data repositories relating to mismatched repair genes and Lynch syndrome that were based in Newfoundland and Labrador or included samples from people in Newfoundland and Labrador.⁵⁹⁻⁶³ Woods et al. (2007)⁵⁹ reported there was a public database established at Memorial University of Newfoundland: the mismatch repair genes variant database. The amount of data stored is unclear, and the database was merged with the International Society for Gastrointestinal Hereditary Tumours (InSiGHT) database.⁶⁰⁻⁶³ This was noted in multiple articles exploring the genetics of Lynch syndrome in Newfoundland and Labrador.⁶⁰⁻⁶³ Summaries of the Newfoundland and Labrador articles along with the research contacts listed for the articles are available in Table 5 in Appendix 2 in the Supplemental Material document.

All datasets described [Table 1](#) were generated by recruiting participants for research studies rather than through retrospective clinical data. Multiple studies included individuals with spondyloarthritis⁴⁹⁻⁵⁴ and genetically linked hearing loss,⁷¹⁻⁷⁷ but it was unclear if these studies all used the same participants. Additionally, multiple studies recruited individuals with genetically linked illnesses and their families. These included studies examining Bardet-Biedl syndrome,⁶⁴⁻⁶⁹ genetically linked hearing loss,⁷¹⁻⁷⁷ familial intracranial aneurysm,⁸³ familial and sporadic idiopathic pulmonary fibrosis,⁸⁴ and dominant hereditary spastic ataxia.⁸⁵ Pedigrees were generated; researchers collected data and accessed patient charts. Peripheral blood samples were collected, and testing was conducted for specific genes known to cause disease. It was not clear how many individuals were included in the final datasets or how many had genetic sequencing. Details on some datasets were unclear from the literature and therefore are not discussed here.

Multiple datasets were identified in both the literature and interviews. The CODING dataset^{86-93,95} and the NL Genome Project dataset,^{4,5} which include data from the general Newfoundland and Labrador population and specific subpopulations with various illnesses; the Newfoundland Familial Colorectal Cancer Registry (NFCCR);^{7,8,12,13,18-28} another colorectal cancer dataset (that is not NFCCR); the Newfoundland Osteoarthritis Study (NFOAS),⁸⁷⁻⁹³ a psoriatic arthritis dataset, a psoriasis-specific dataset, and an ARVC dataset⁷⁸⁻⁸² (refer to [Table 1](#)). These datasets were collected for research purposes and were accessed for secondary use. They range in size from 300 (ARVC dataset) to 2,500 (NL Genome Project dataset) individuals.

The CODING dataset was collected for research purposes from 752 people aged 19 years or older, born in Newfoundland and Labrador, had families who had lived in Newfoundland and Labrador for at least 3 generations, and who did not have serious metabolic, cardiovascular, or endocrine diseases.^{88,95} The purpose of the CODING dataset was to provide a repository of control cases, which was used in multiple studies.⁸⁶⁻⁹³ The genomic data available for the CODING dataset varies depending on which studies used the data (personal communication, May 28, 2025). It is likely that it has been used as the source for controls (people without various conditions) in other retrieved studies, but this was not expressly stated and could not be verified.

The NL Genome Project dataset was collected more recently with the goal of generating genetic information on the general Newfoundland and Labrador population to inform medication and precision medicine studies.⁹⁷

The NFCCR, which was used in the most retrieved studies, has been used in both genome-wide association studies (GWAS)^{3,12} and whole exome sequencing (WES) in addition to panels.^{7,8,13} It has also been used in studies conducted by the Colorectal Transdisciplinary (CORECT) consortium.¹⁸⁻²⁷ Except for the non-NFCCR colorectal cancer (panel) dataset and the ARVC datasets (single mutation), whole genome sequencing (WGS), WES, or GWAS was conducted on samples in these datasets, and the researchers possessed the raw data. Genomic data from the studies retrieved were generally not uploaded to larger databases; however, raw data are available from the principal researchers who are typically the data custodians. Information on these datasets, including sample sizes, data custodians, the testing conducted, linkability to administrative data, and relevant REB approvals, is available in [Table 1](#).

Nova Scotia

Relevant information from 23 studies that included genetic samples from the Nova Scotia population were extracted (refer to Table 3 in Appendix 2 in the Supplemental Material document).⁹⁸⁻¹²⁰ These studies examined diverse health conditions, including eye conditions (familial exudative vitreoretinopathy [FEVR], exfoliation syndrome),^{108,109,111-114} congenital conditions (fetal structural anomalies, Meier-Gorlin syndrome, autosomal-dominant kidney disease),^{99,100,120} cancers (Merkel cell carcinoma, multiple myeloma, Lynch syndrome, renal cell carcinoma, colonic mixed adenoendocrine carcinoma, and neuroendocrine carcinoma),^{98,101-107,110,115} and psychiatric disorders (bipolar disorder, anxiety disorders).¹¹⁶⁻¹¹⁹

Data were collected both retrospectively from clinical data and prospectively for research purposes. Studies on eye conditions,^{108,109,111,113,114} congenital conditions,^{100,120} and psychiatric disorders¹¹⁶⁻¹¹⁹ were from prospective cohorts recruited for research, except for the fetal structural anomalies⁹⁹ study, which was retrospectively collected from clinical records. Conversely, data for cancer studies were largely collected through routine clinical practice and accessed retrospectively,^{98,101-107,110,115} although previously collected tumours were often used for prospective testing.^{102-107,110,115}

Four verified datasets were described in the literature: a bipolar disorder dataset,¹¹⁶⁻¹¹⁸ an FEVR dataset, an exfoliation syndrome dataset, and a Merkel cell carcinoma dataset (refer to [Table 2](#)). These datasets ranged from 51 individuals^{106,107} to 403 individuals.^{108,109} The raw data for all datasets are held by Nova Scotia researchers. Sample size information was unclear for the exfoliation syndrome and FEVR datasets. These values could not be confirmed by researchers. The FEVR dataset is not limited to the Nova Scotia population; a minority of the participants were reported to come from Nova Scotia (personal communication, June 2, 2025). Currently, the Nova Scotia-based datasets discussed in this section have not been deposited in centralized genomic libraries or databases; however, cancer datasets may soon be available through the Atlantic Cancer Consortium (ACC) (described further in the Findings From Surveys and Interviews section). Generally, the linkability of datasets was not described in the literature and was not confirmed by data custodians, except for the Merkel cell carcinoma dataset, which was confirmed to be nonlinkable (personal communication, June 23, 2025).

New Brunswick

Five genomic articles regarding the New Brunswick population were retrieved and information was extracted (refer to Table 3 in Appendix 2 in the Supplemental Material document).^{2,121-124} These studies included mostly

patients with various cancers, such as breast carcinoma,^{121,122} myeloid neoplasms,¹²³ and pancreatic ductal adenocarcinoma,¹²⁴ but also individuals of Acadian heritage without known disease.²

As was the case for the cancer studies in Nova Scotia, genetic samples were generally pulled from specimens previously collected for clinical purposes and genetic testing.¹²¹⁻¹²⁴ The New Brunswick carrier screening study collected questionnaires and blood samples prospectively for research purposes.²

Two datasets described in the literature were confirmed in conversation with researchers: the New Brunswick carrier screening dataset,² an ongoing data collection initiative that currently includes genetic data from 240 participants (420 when complete), and a breast and ovarian cancer genetic screening dataset, which includes genetic testing data for 306 individuals.¹²² Although not currently in place, there are plans for the New Brunswick carrier screening dataset to be included in the Pan-Canadian Genome Project. Information on these datasets, including sample sizes, data custodians, testing conducted, linkability to administrative data, and relevant REBs is available in [Table 3](#).

Multiprovince

Two multiprovince datasets were discovered through our literature: one regarding the Atlantic PATH research project¹²⁵ and a second with people who have spinal muscular atrophy (SMA) (refer to Table 2 in Appendix 2 in the Supplemental Material document). The second dataset aimed to include all people born between 2000 and 2020 with SMA from Prince Edward Island, Nova Scotia, and New Brunswick by including blood work that was positive for the SMN1 marker.¹²⁶ This dataset was small (only 30) and was not mentioned in any correspondences.¹²⁶ It would be included in the broader Germline Clinical dataset outlined in the IWK Health Clinical Data Access, Privacy, and Linkability section and in [Table 2](#). The Atlantic PATH research dataset is described in more depth under the Multiprovince section in Findings From Surveys and Interviews and in Table 4.

Findings From Surveys and Interviews

Internet searches for datasets and literature searches were useful in identifying datasets. However, interviews and other communications provided rich information regarding the flow of data and the processes required to access data. In some cases, they confirmed the existence of datasets. Overall, 50 people from different organizations or institutes across Atlantic Canada were contacted for this project. We conducted interviews with 16 researchers and received 12 email responses from multiple researchers and organizations (a list of researchers who were contacted can be provided upon request). In some datasets, no people with a condition were included; in others, no people without the specified condition were included. Datasets collected for clinical purposes only include patients (people with a specified condition). In other instances, such as the Atlantic PATH, the New Brunswick carrier screening, CODING, and NL Genome Project datasets, the goal was to collect data on the general population rather than people with a particular condition. In these instances, only an overall number is indicated rather than people with and without a condition.

Newfoundland and Labrador

Most genomic publications retrieved came from samples from Newfoundland and Labrador. We interviewed 6 clinicians and researchers from Newfoundland and Labrador regarding genomic data availability and steps

to access data. Genomic datasets were identified in Newfoundland and Labrador but, similar to the research datasets in Nova Scotia, datasets are held by individual researchers as data custodians rather than central repositories or research databases (researchers are encouraged to publish variant findings in the ClinVar database [personal communication, March 31, 2025; personal communications, March 31, 2025; March 31, 2025; April 10, 2025]). Data collection and sequencing follow different processes for different datasets. Additionally, we gained some insight into the process of accessing data collected for clinical purposes.

Newfoundland and Labrador Clinical Data

Clinical testing in Newfoundland and Labrador is outsourced out of the province (personal communication, March 31, 2025). Similar to both New Brunswick and Nova Scotia, clinical results are communicated to clinicians through the Shire system and historically accessed as paper charts, but these reports are in the process of being digitized. In some cases, reports have been uploaded into the electronic medical record system (personal communication, March 31, 2025). The approximate number of people receiving clinical genetic testing was not available.

Newfoundland and Labrador Clinical Data Access, Privacy, and Linkability

Based on discussions with other provinces, it is likely that only genetic reports, rather than raw data, would be available. Consents for clinical genomic testing currently do not address secondary use, but there are hopes that these provisions will be included in the future (personal communications, March 31, 2025; March 31, 2025; March 31, 2025). Clinical data may be considered for secondary use pending REB approval. In this instance, a local principal investigator (PI) from the Provincial Medical Genetics Program would be required (personal communication, June 11, 2025). A figure displaying the flow of genomic clinical data is available in the Supplemental Material document (Figure 8 in Appendix 7).

Newfoundland and Labrador Research Data

Overall, genomic data are project dependent and held by individual researchers rather than centralized repositories or institutions (personal communication, March 31, 2025). This can make identifying datasets a challenge; however, rich pedigrees accompanying many datasets were stated to be a strength (personal communication, March 31, 2025).

Eight verified datasets were identified that had been collected for research purposes: 2 were collected to represent the general population (CODING and NL Genome Project datasets) and 6 to collect data on people with various medical conditions (NFCCR, other colorectal, psoriasis, psoriatic arthritis, NFOAS, ARVC). Data custodians could be reached for only 3 identified datasets: NFOAS dataset, the psoriasis dataset, and the ARVC datasets.

Some additional information on the psoriatic arthritis and NFCCR datasets was gathered through researchers. The NFOAS started in 2011 by recruiting approximately 800 participants about to undergo total joint replacements due to osteoarthritis (personal communication, May 28, 2025). People undergoing total joint replacement due to injury were recruited as controls (personal communication, May 28, 2025). Blood samples, articular cartilage, subchondral bone, and synovial fluid were collected. Additional samples from people without osteoarthritis were processed through blood samples provided by participants in the CODING

study (personal communication, May 28, 2025). GWAS, WES, and RNA sequencing data (binary alignment/map [BAM] and Variant Call Format [VCF] files) are available for secondary use with this cohort (personal communication, May 28, 2025). The psoriasis dataset was collected in 1995 from 1,200 participants with psoriasis through a psoriasis clinic and 800 participants without psoriasis (personal communication, April 10, 2025). Longitudinal clinical data are still being collected in living patients (personal communication, April 10, 2025). GWAS sequencing was conducted, and raw data and samples are in the possession of the Newfoundland and Labrador-based data custodian with the possibility of secondary research projects (personal communication, April 10, 2025). Multiple articles assessed genetic aspects of psoriatic arthritis within the Newfoundland and Labrador population.^{31-37,39,50,53} A psoriatic arthritis dataset was confirmed with interviews and is reported to have raw data from GWAS, WES, and genetic panels for 200 to 300 participants (personal communication, May 28, 2025).

Newfoundland and Labrador Research Data Access, Privacy, and Linkability

The NFOAS and psoriasis datasets are linkable to administrative health data (personal communication, May 28, 2025); the NFCCR and CODING datasets are not (personal communication, May 28, 2025). More generally, the Newfoundland and Labrador genetics researchers highlighted the importance of ensuring medically actionable findings through data that can be linked back to participants, while also respecting participant privacy (personal communications, March 31, 2025; March 31, 2025; May 28, 2025).

Specific procedures are still being determined, but multiple researchers expressed the importance of having a local PI when external institutions wish to access data (personal communications, March 31, 2025; March 31, 2025). This allows for deidentification of data for external researchers, while internal researchers can maintain the ability to link findings back to patients if a medically actionable finding is discovered. Additionally, REB approval is required from the Health Research Ethics Authority of Newfoundland and Labrador, memoranda of understanding, and material transfer agreements if data are removed from researcher's laboratories (personal communications, March 31, 2025; March 31, 2025; May 28, 2025). Figure 9 in Appendix 7 in the Supplemental Material document depicts the flow of genomic data for research projects. Additionally, flow charts specific to the psoriasis dataset and NFOAS dataset are also available (Figures 10 and 11 in Appendix 7 in the Supplemental Material document).

Table 1: Datasets From Newfoundland and Labrador Identified Through Literature Review and Interviews

Dataset description	NFOAS	CODING study	NFCCR	Other colorectal cancer	Psoriatic arthritis	Psoriasis	ARVC	NL Genome Project
Sample characteristics	Primarily total knee or hip joint replacement in patients with osteoarthritis and ≈ 120 samples from the CODING study (without osteoarthritis) and patients with TJR from injuries (without osteoarthritis).	3,000 volunteers (with ≥ 3 generations in Newfoundland and Labrador). Unclear how many have genetic data.	Patients diagnosed with colorectal cancer aged < 75 years at diagnosis; 750 (64%) from 708 families. People without colorectal cancer from random digit dialling in Newfoundland and Labrador matched to with colorectal cancer by sex and age. Blood sample (552 people), tumour tissue (772 tumours from 750 with colorectal cancer) (1999 to 2003).	Retrospective cohort of 280 patients followed for ≤ 12.5 years after diagnosis. From Avalon Peninsula, Newfoundland and Labrador. Nontumour colon and rectum tissues (1997 to 1998).	People with psoriatic arthritis.	Patients with psoriasis with European descent and ethnically matched individuals without psoriasis. Blood samples (some nail and buccal mucosa samples). Collected in 1995.	367 individuals from sibships and 11 families (≥ 50% known to be at high or low risk of ARVC as determined by clinical, pedigree, and/or haplotype data). Groupings were classified as affected, unaffected, or unknown. (Status known in ≥ 50% of siblings.)	Random recruitment from general practice clinics across Newfoundland and Labrador of individuals aged ≥ 18 years with valid Newfoundland and Labrador health card. Saliva samples.
Data format	Raw data	Raw data	Raw data	Raw data	Raw data	Raw data	Report	Raw data
Overall sample size, N	Approximately 1,000	3,000	Refer to Sample Characteristics and Genetic Testing Conducted	272 genotyped	200 to 300	2,000	Blood samples from 295 individuals available	2,446

Dataset description	NFOAS	CODING study	NFCCR	Other colorectal cancer	Psoriatic arthritis	Psoriasis	ARVC	NL Genome Project
With condition, n	≈ 800	NA	Unclear	272	200 to 300	1,200	Unclear	NA
Without condition, n	≈ 200	3,000	Unclear	NA	0	800	Unclear	NA
Purpose of collection	Research	Research	Research	Clinical care	Research	Research	Research and clinical care	Research
Data custodian(s)	Dr. Guangju Zhai	Dr. Guang Sun	Dr. Michael Woods	Unclear; Dr. Sevtap Savas listed as contact for papers referencing this cohort	Dr. Proton Rahman	Unknown	Dr. Kathleen Hodgkinson	Dr. Gerald Mugford, Dr. Dennis O'Keefe
REB	NL Health Research Ethics Authority							
Secondary research access	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Linkable	Yes	No	No	No	Yes	Yes	No	No
Genetic testing conducted	GWAS (1,000 people) WES (300 people) RNA sequencing	GWAS (refer to NFOAS)	GWAS (Illumina Human Omni1-Quad > 1 million SNP markers. 505 had usable data); 195 with colorectal cancer, 477 without colorectal cancer on Affymetrix Axiom platform (1.1 million SNPs). WES	Pannel (Sequenom MassARRAY technology)	GWAS WES Panel	GWAS (820,000 SNPs assessed using UK Biobank Axiom Array)	Panel testing conducted in smaller sample (to identify causal variant). Single mutation (DNA from all available individuals born at a priori 50% risk [n = 295] across the 15 ARVC families for the presence of the 1073C →	WGS (1,721,246 SNPs genotyped using the Illumina Global Diversity Array [Illumina, San Diego])

Dataset description	NFOAS	CODING study	NFCCR	Other colorectal cancer	Psoriatic arthritis	Psoriasis	ARVC	NL Genome Project
			(≥ 16 families [DeRycke et al. (2013) ¹¹]). Panel (ABI 3,700 DNA Analyzer ⁸)				T TMEM43 mutation)	
Nongenetic data collection	Cartilage DNA myelination, subchondral bone and synovial fluids (< 50), metabolic assessment 330, medical record (drugs, health status), WOMAC questionnaire (at TJR and 1 year after)	Participants provided information via a general questionnaire. Many consented to secondary use. Prospective researcher required to conduct genomic sequencing	Without condition: risk factor questionnaire With condition: permission to access their tumour specimens and medical records	For all: clinical data collected. Prognostic data collected from medical and hospital records and the Newfoundland and Labrador Centre for Health Information	NA	Physical exam characteristics. Working with Newfoundland and Labrador Centre for Health Information to provide easy linkage with administrative data	Pedigree data, annual cardiomyopathy clinic data (ECGs, Holter monitors, MRIs, ECHOs). Medical records, including “at-risk” relatives not seen in clinic and autopsy results	Information also collected on religion and the birthplace of their parental ancestors
Sources	Personal communication, May 28, 2025	Personal communication, May 28, 2025	Literature ¹¹⁻¹⁴ Personal communication, May 28, 2025	Literature ^{12,28,29}	Literature ^{31-33, 38,127-129} Personal communication, May 28, 2025	Literature ^{47,48} Personal communication, April 10, 2025	Literature ⁷⁸⁻⁸² Personal communication, March 31, 2025	Literature ^{4,5}

ARVC = autosomal-dominant arrhythmogenic right ventricular cardiomyopathy; CODING = Complex Diseases in the Newfoundland population: Environment and Genetics; ECHO = echocardiogram; GWAS = genome-wide association study; NA = not available; NFCCR = Newfoundland Colorectal Cancer Registry; NFOAS = Newfoundland Osteoarthritis Study; REB = research ethics board; SNP = single-nucleotide polymorphisms; TJR = total joint replacement; WES = whole exome sequencing; WGS = whole genome sequencing; WOMAC = Western Ontario and McMaster Universities Osteoarthritis Index. Nova Scotia

Nova Scotia has 2 sites for clinical testing: the Clinical Genomics Laboratory (CGL) at IWK Health for germline testing and the Department of Pathology and Laboratory Medicine for somatic testing at Nova Scotia Health (personal communication, March 12, 2025). Information on these data sources was based in part from interviews conducted with 3 researchers at IWK Health and 1 researcher at the Department of Pathology and Laboratory Medicine.

IWK Health Clinical Data

The CGL provides care to people across the Maritimes, and approximately 15,000 people have been referred (personal communication, March 12, 2025; personal communication, May 28, 2025). Data are processed in-house or through Blueprint. The CGL at IWK Health is the data custodian; however, the raw data generated from sequencing is currently held in the Illumina cloud (for samples processed in-house) and by external sequencing companies (personal communication, March 12, 2025; personal communication, May 28, 2025). Therefore, only reports of genetic variants are currently accessible (this report is not currently included in the patient's electronic medical record). The laboratory is in the process of repatriating data and will gain access to the BAM and VCF files in the future. [Table 3](#) provides more information on the CGL germline clinical dataset (personal communication, March 12, 2025; personal communication, May 28, 2025).

IWK Health Clinical Data Access, Privacy, and Linkability

Currently, available research has been limited to small, internal studies. Consent language does not presently account for secondary research (personal communication, March 12, 2025; personal communication, May 28, 2025). Data linkage with administrative data is feasible but complicated in practice by siloed data and privacy considerations (personal communication, March 12, 2025; personal communication, May 28, 2025). IWK Health is the data custodian; any data access would be required to go through the IWK Health REB (personal communication, March 12, 2025; personal communication, May 28, 2025). A joint access request and privacy impact assessment would be required as well as having a PI from the CGL. In future, the CGL is hoping to develop formal procedures for accessing data and a federated network to facilitate genomics research (personal communication, March 12, 2025; personal communication, May 28, 2025). Figure 6 in Appendix 7 in the Supplemental Material document displays the flow of clinical data.

No generalized or project-specific research flow charts are available for genomic data collected for Nova Scotia. Processes can differ substantially between projects. Availability of research data for secondary research is unclear. Information collected through interviews regarding individual Nova Scotia research datasets was not sufficient for flow chart creation.

Nova Scotia Health Somatic Clinical Data

The Department of Pathology and Laboratory Medicine provides somatic testing related to cancers and neoplasms to approximately 2,000 to 3,000 patients per year (since 2013) around the Maritimes. The laboratory covers various commercially available panels processed with Illumina next-generation sequencing equipment. All testing is done in-house, and the raw data (FastQs generated by the sequencer, BAM files, VCFs produced) are held in-house. Refer to [Table 2](#) for information on the clinical dataset.

Nova Scotia Health Somatic Clinical Data Access, Privacy, and Linkability

No formal processes are in place for external institutions to access data from the Department of Pathology and Laboratory Medicine for research purposes. Consent language for clinical somatic testing does not specifically allow for research use. REB approval from the Nova Scotia Health Authority would be required, and a local PI would need to be included to access data. Linkage may technically be possible but would be complicated in practice due to lack of explicit patient consent. Refer to Figure 7 in Appendix 7 in the Supplemental Material document for the flow of somatic genomic data and access to genomic data.

Table 2: Datasets From Nova Scotia Identified Through Literature Review and Interviews

Dataset description	Merkel cell carcinoma dataset	Exfoliation syndrome dataset	Somatic clinical dataset	FEVR dataset	Bipolar disorder dataset	Germline clinical dataset
Sample characteristics	Primary cutaneous MCC samples from Maritime Canada through Pathology Department of Victoria General Hospital and QEII Health Sciences Centre in Halifax (1993 to 2020).	Blood samples (WES) and ocular tissues (GWAS) from patients with exfoliation syndrome enrolled. Control samples drawn from discarded blood samples at Core lab. Deidentified, no reidentification.	All clinical NGS somatic testing from across Nova Scotia and Prince Edward Island and some from New Brunswick and Newfoundland and Labrador. Consent obtained. (Since 2017.)	Patients and family members affected by FEVR. Blood or saliva samples collected (1998 to ongoing). Note: minority (number unspecified) from Nova Scotia.	People with <i>DSM-III</i> or <i>DSM-IV</i> diagnosis of a bipolar spectrum disorder in Nova Scotia, who have been taking lithium ≥ 6 months.	Patients in Nova Scotia, New Brunswick, Prince Edward Island, and sometimes Newfoundland and Labrador requiring germline genetic testing in Clinical Genomics Lab (IWK Health). Includes testing for various genetic mutations and conditions (all ages).
Data format	Raw data (BAM and VCF files); report	Raw data	Raw data (FASTQ, BAM, and VCF files); report	Raw data; report	Raw data	Report
Overall sample size, N	51	Unknown	2,000 to 3,000 per year	Unclear, ≥ 94	353	$\approx 15,000$
With condition, n	51	403 (WES), 341 (GWAS), 136 (replication cohort)	2,000 to 3,000 per year	Unclear	353	$\approx 15,000$
Without condition, n	NA	392 (WES), 247 (GWAS), 747 (replication cohort)	NA	Unclear	NA	NA
Purpose of collection	Research	Research	Clinical care	Research	Research	Clinical care
Data custodian	Dr. Michael Carter	Dr. Andrew Orr	Dr. Daniel Gaston	Dr. Johane Robitaille	Dr. Martin Alda	Dr. Victor Martinez
REB	NSH REB	NSH REB	NSH REB	IWK REB	NSH REB	IWK REB
Secondary research access	Yes	Unclear	Yes	Unclear	Unclear	No
Linkable	No	Unclear	Yes	Unclear	Unclear	Unclear

Dataset description	Merkel cell carcinoma dataset	Exfoliation syndrome dataset	Somatic clinical dataset	FEVR dataset	Bipolar disorder dataset	Germline clinical dataset
Genetic testing conducted	Panel (523 cancer-related genes)	GWAS (Illumina OmniExpress BeadChip) WES (Illumina HiSeq 4000 and NovaSeq 6000 instruments)	Panel (≈ 50 genes) RNA sequencing Single mutation	WES (Illumina NextSeq 550) Panel (Applied Biosystems Prism 3100 Genetic Analyzer)	SNP array from Illumina Omni1-Quad in GWAS1 (n = 241) Illumina OmniExpress 1·1) (n = 121)	WGS GWAS WES Panel
Nongenetic data collection	NA	Eye testing results available for people with condition	Pathology reports	Eye exams, IV fluorescein angiography fundus photography	Clinical interviews, medical records, self-reported surveys	Phenotypic data collected
Sources	Literature ^{106,107} Personal communications: May 29, 2025 June 13, 2025 June 23, 2025	Literature ^{108,109} Personal communication: May 29, 2025	Personal communication: May 29, 2025	Literature ^{111,113,114} Personal communication: May 29, 2025	Literature ¹¹⁶⁻¹¹⁸	Personal communications: March 12, 2025 May 28, 2025

BAM = binary alignment map; DSM = *Diagnostic and Statistical Manual of Mental Disorders*; FEVR = familial exudative vitreoretinopathy; GWAS = genome-wide association study; MCC = Merkel cell carcinoma; NA = not applicable; NGS = next-generation sequencing; NSH = Nova Scotia Health; QEII = Queen Elizabeth II; REB = research ethics board; SNP = single-nucleotide polymorphism; VCF = variant call format; WES = whole exome sequencing; WGS = whole genome sequencing. New Brunswick

Four New Brunswick researchers were interviewed from both regional health authorities: 3 affiliated with Vitalité Health Network and 1 affiliated with Horizon Health Network. These researchers and clinicians were affiliated with 2 centres in New Brunswick involved in genomic testing: the Dr. Georges-L.-Dumont University Hospital Molecular Genetics Laboratory and the Molecular Diagnostics Division of Laboratory Medicine at Saint John Regional Hospital. No other centres were named in the literature or in interviews with researchers within New Brunswick.

Vitalité Clinical Data

There are both a research group and a clinical geneticist serving people in New Brunswick in the Vitalité Health Network through the Dr. Georges-L.-Dumont University Hospital. Through interviews, we gained insight into the flow of data as well as the availability of data for secondary use and linkability to administrative data. Approximately 1,000 individuals per year are assessed in the genetics clinic (personal communication, March 20, 2025). Although most individuals are from New Brunswick, a minority are from Nova Scotia and Prince Edward Island (personal communication, March 20, 2025).

Patients are genetically tested through a variety of modalities, largely through third-party sequencing companies such as Blueprint Genetics, Cento-gene, and MNG (personal communication, April 25, 2025); however, targeted gene testing is performed in-house at the Dr. Georges-L.-Dumont University Hospital (personal communication, March 20, 2025). Raw data are typically generated and processed into a report by third-party sequencing companies and not made available for clinical purposes or retained by the third-party sequencers unless requested at the time of sequencing. Only paper copies of genetic reports provided by the third-party sequencers are retrieved and kept in patient charts and in the genetics clinic (personal communication, March 20, 2025). Information regarding the clinical dataset is presented in [Table 3](#). A flow chart displaying the genetic data creation and access process is available in Figure 3 in Appendix 7 in the Supplemental Material document.

Vitalité Research Data

One research-generated dataset was mentioned in interviews and communications, the New Brunswick Carrier Screening Project dataset, which aims to conduct a 342-gene panel on 60 people from each of New Brunswick's 7 health zones (420 people total, with 240 from 4 health zones collected to date) who have at least 1 grandparent with Acadian ancestry born in the region. These data will eventually be available (deidentified) through the Pan-Canadian Genome Project. All participants will also complete a survey including self-reported family history and demographics which will be collected in Excel tables (personal communication, March 20, 2025). This project was sequenced with Fulgent Genetics at McGill University. Raw data are not held by external labs unless requested (personal communication, April 25, 2025); for this project, the raw data are maintained at McGill University, and some have been repatriated to Moncton. Reports are also being generated for clinicians. These data will eventually be available (deidentified) through the Pan-Canadian Genome Project. A flow chart displaying the genetic data creation and access process is available in Figure 4 in Appendix 7 in the Supplemental Material document.

Vitalité Data Access, Privacy, and Linkability

Previously, external organizations have not obtained retrospective genetic report data (personal communication, March 20, 2025). The consents obtained for clinical genetic testing do not currently include permissions for secondary research. Studies may be allowed through the Vitalité Health Network REB if data are deidentified, no additional testing is needed, and more than 60 people are included in the proposed study (personal communication, March 20, 2025), such as in the study by Gauvin et al. (2025).¹²² Additional processes would be required to access medical genetic reports from third-party sequencing companies (personal communication, April 25, 2025). A local PI would typically be required; formal processes are not in place for external researchers or organizations to access data. Genetic reports are theoretically linkable to administrative health data but, in practice, this would be quite complicated because the administrative data are siloed and linkage would have to be performed manually (personal communication, March 20, 2025). Research ethics approval from the Vitalité Health Network REB would be required for any secondary use of data initially obtained for clinical purposes (personal communication, March 20, 2025).

Obtaining access to genetic data collected for research purposes follows a similar process to that for clinical data, although more raw data have been repatriated and more sequencing is conducted in-house. Genomic data are stored on servers maintained by the Digital Research Alliance of Canada (DRAC) or in-house (personal communication, March 20, 2025).

Horizon Clinical Data

We spoke with 1 clinician within Horizon Health involved in referring patients for clinical genetic testing at the Molecular Diagnostics Division of Laboratory Medicine at Saint John Regional Hospital. The Molecular Diagnostics Division of Laboratory Medicine has processed genomic data (mostly from cancerous tumour samples) for hundreds of patients of Horizon Health over the past 10 years. Samples are processed in-house from commercially available genetic panels including approximately 500 variants (with fewer variants available in genetic panels from earlier years). Raw data (BAM files and data processing files) are housed within the laboratory, and clinical reports are provided to referring physicians.

Horizon Data Access, Privacy, and Linkability

Data are not generated for research purposes, and official processes are not in place for accessing data for research programs through Horizon. Consent forms signed by patients do not include provisions for secondary use; however, it has been used for internal research purposes on a small number of occasions for samples sizes greater than 100 persons (personal communication, April 10, 2025) after Horizon Health Network REB review. More official processes are being put in place. External organizations would likely require a local PI if accessing data because the raw data are only available directly within the Molecular Diagnostics Division of Laboratory Medicine at Saint John Regional Hospital. Although linkage is theoretically possible, due to siloed health information and paper charting, such linkage would need to be conducted manually using patient charts. A flow chart displaying the flow of clinical data is available in Figure 5 in Appendix 7 in the Supplemental Material document. Datasets available from New Brunswick are outlined in [Table 3](#).

Table 3: Datasets From New Brunswick Identified Through Literature Review and Interviews

Dataset description	Retrospective breast and ovarian cancer dataset	Vitalité clinical dataset	Horizon Health clinical dataset	New Brunswick carrier screening dataset
Sample characteristics	A retrospective study of people who received a genetics consultation at the Medical Genetics Clinic in Moncton, New Brunswick, between October 2019 and July 2022, with a personal or family history of breast or ovarian cancer and had genetic testing.	Individuals requiring germline testing (multiple ages, ethnicities, conditions, and phenotypic presentations). Majority of individuals are from New Brunswick with some from Nova Scotia and Prince Edward Island.	Tumour and blood samples (majority cancer genomics cases providing tumour samples) in New Brunswick health zones served by Horizon Health Network over the past 10 years, processed at the Molecular Diagnostics Division of Laboratory Medicine at Saint John Regional Hospital.	60 (7 regions total) people from each health zone in New Brunswick with at least 1 Acadian grandparent born in the region.
Data format	Report	Report	Raw data; report	Raw data; report
Overall sample size, N	306 with genetic testing	≈ 1,000 per year	> 100 per year	240 (420 when complete)
With condition, n	306	≈ 1,000 per year	> 100 per year	NA
Without condition, n	NA	NA	NA	NA
Purpose of collection	Clinical care	Clinical care	Clinical care	Research
Data custodians	Dr. Eric Allain, Dr. Mouna Ben Amor	Dr. Mouna Ben Amor	Dr. Doha Itani	Dr. Eric Allain
REB	Vitalité Health Network REB	Vitalité Health Network REB	Horizon Health Network REB	Vitalité Health Network REB
Accessed for secondary research	Yes	No	Yes	Yes
Linkable to administrative data	Yes	No	Yes	No
Genetic testing conducted	Panel (268 people) Single mutation (39 people)	WGS WES Panel RNA sequencing Single mutation	Panel (covers approximately 500 variants)	Panel (342 genes)
Nongenetic data collection	Data on cancer history and some demographic information included	NA	NA	Survey information (age, sex, ethnicity, socioeconomic level), stored in Excel format

Dataset description	Retrospective breast and ovarian cancer dataset	Vitalité clinical dataset	Horizon Health clinical dataset	New Brunswick carrier screening dataset
Sources	Literature: Gauvin et al. (2025) ¹²² Personal communication, March 20, 2025	Personal communication, March 20, 2025 Personal communication, April 25, 2025	Personal communication, April 10, 2025	Literature: Robichaud et al. (2022) ² Personal communication, March 20, 2025 Personal communication, April 25, 2025

NA = not available; REB = research ethics board; WES = whole exome sequencing; WGS = whole genome sequencing.

Multiprovince

Two multiprovincial datasets were identified through internet and literature searches: the ACC and Atlantic PATH. Researchers from Atlantic PATH and the ACC provided insight into the data acquisition and availability of these multiprovincial datasets (refer to [Table 4](#)).

Atlantic Cancer Consortium (Research)

Four researchers working on the ACC provided insight into the project. The ACC is part of the Terry Fox Marathon of Hope Cancer Centres Network. It aims to create a federated network that will be accessible to researchers who become consortium members. Newfoundland and Labrador, Nova Scotia, and New Brunswick will each have a local PI, and health facilities within each province will submit blood and tumour tissue samples (from consenting patients with multiple forms of cancer) for WGS and RNA sequencing (personal communication, April 10, 2025; personal communication, May 29, 2025). Although the samples may be collected retrospectively (from previously collected tumour and blood samples) or prospectively, all samples receive the same sequencing. So far, 1,000 participants have been sequenced (personal communication, November 19, 2025). Deidentified data will be uploaded to the consortium portal (federated system facilitated by CanDIG), and researchers will be able to search cancer genetic demographic data in the portal, such as how many cancer cases can be attributed to a particular mutation (personal communication, May 29, 2025). The type of questions the dataset is oriented to answer (e.g., correlation studies, clinical, health technology assessment) was not specified; however, raw data from WGS and RNA sequencing will be available to researchers through a data application process in lossless .bam or .fastq.gz format (personal communication, May 29, 2025).^{130,131} Information on the dataset is available in [Table 4](#).

Atlantic Cancer Consortium Data Access, Privacy, Linkability

Consent allows for access to raw data for research (personal communication, April 10, 2025; personal communication, May 29, 2025). Samples will be deidentified at the site where they are obtained and could be reidentified to provide reports to physicians for patient care, but ACC consent forms explicitly state that participants will not be reidentified for additional purposes. Therefore, data linkage to administrative health data for secondary research would not be feasible through the ACC (personal communication, April 10, 2025; personal communication, May 29, 2025). To obtain data, membership with the consortium is expected in addition to an REB application to the applicant's local institution and an access application to the Marathon of Hope Access Committee (personal communication, May 29, 2025). A flow chart displaying the genetic data creation and access process is available in Figure 1 in Appendix 7 in the Supplemental Material document.

Atlantic PATH (Research)

Atlantic PATH enrolled participants aged 30 to 74 years from 2009 to 2015 in Newfoundland and Labrador, Prince Edward Island, Nova Scotia, and New Brunswick (email correspondence, February 19, 2025; May 23, 2025). Participants provided toenail, urine, blood, and saliva samples; completed questionnaires; and had physical measures collected (email correspondence; February 19, 2025; May 23, 2025). To date, 1,000 genetic samples have undergone WGS, with plans to process 22,000 samples. Data and biosamples are retained in Atlantic PATH (Halifax) (email correspondence; February 19, 2025; May 23, 2025). Information on the dataset is available in [Table 4](#).

Atlantic PATH Access, Privacy, and Linkability

Atlantic PATH has a data access process for researchers interested in using its data holdings and biosamples. Requirements include REB approval. Consent allows for data linkage to cancer registries and other administrative health data through institutional linkage agreements. Atlantic PATH currently holds cancer registry data for Nova Scotia and Newfoundland and Labrador. For administrative health data, agreements are currently in place with Health Data Nova Scotia and DataNB (previously the New Brunswick Institute for Research Data and Training), and in progress with the Secure Island Data Repository and Newfoundland and Labrador Health Services (email correspondence, February 19, 2025; May 23, 2025). Health Data Nova Scotia and DataNB hold Atlantic PATH data for their respective provinces that can be linked to administrative data holdings (email correspondence, February 19, 2025, May 23, 2025). There is an existing access process between Atlantic PATH, administrative data holders, CanPath, and The Health Data Resource Network (depending on the number of provinces and so on). Additional details can be provided by contacting Atlantic PATH and/or the administrative data holder directly at Ellen.Sweeney@dal.ca or visiting the Atlantic PATH website¹³⁰ (email correspondence, February 19, 2025; May 23, 2025). A flow chart displaying the genetic data creation and access process is available in Figure 2 in Appendix 7 in the Supplemental Material document.

Table 4: Datasets From Multiple Atlantic Provinces Identified Through Literature Review and Interviews

Dataset description	Atlantic Cancer Consortium	Atlantic PATH (genotyped)
Province	Newfoundland and Labrador, Nova Scotia, New Brunswick	Newfoundland and Labrador, Prince Edward Island, Nova Scotia, New Brunswick
Sample characteristics	People with colorectal and lung cancer; both tumour samples and peripheral blood samples provided (germline testing)	Toenail, urine, blood, and saliva samples collected from 2009 to 2015 from individuals aged 30 to 74 years
Data format	Raw data	Raw data
Overall sample size, N	1,000	Genotyped: 1,000 Future: ≈ 22,000 additional participants
With condition, n	1,000 (people with various malignancies)	NA
Without condition, n	None	NA
Purpose of collection	Research	Research
Data custodians	Dr. Sherri Christian (Newfoundland and Labrador), Dr. Robin Urquhart (Nova Scotia), Dr. Anthony Reiman (New Brunswick)	Atlantic PATH Data Platform and Biorepository (Research Director: Dr. Ellen Sweeney)
REB application process	Become member of ACC Apply to local institutional REB Submit access application to Marathon of Hope data access committee	PI's Home institution REB Nova Scotia Health REB and/or Dalhousie University REB (facilitated by Atlantic PATH)
Accessed for secondary research	Yes	Yes

Dataset description	Atlantic Cancer Consortium	Atlantic PATH (genotyped)
Linkable to administrative data	No	Yes
Genetic testing conducted	WGS (minimum 80 × tumour, 30 × normal or 30 × tumour, 30 × normal) RNA sequencing (minimum 80 million)	WGS (Illumina MEGA array, 1.8 million SNPs)
Nongenetic data collection	Data such as clinical outcomes, stage, treatment	Demographics, comorbidities, cancer history, lifestyle habits, and so on; physical measurements (height, weight, waist and hip circumference, body composition, and blood pressure)
Sources	Literature: MOHCCN Technology Working Group (2026); ¹³¹ MOHCCN Technology Working Group (2025) ¹³² Personal communication, April 10, 2025; personal communication, April 23, 2025; personal communication, May 29, 2025	Literature: Sweeney et al. (2017) ¹²⁵ Email correspondence, February 19, 2025, and May 23, 2025

ACC = Atlantic Cancer Consortium; MOHCCN = Marathon of Hope Cancer Centres Network; NA = not applicable; PATH = Partnership for Tomorrow's Health; PI = principal investigator; REB = research ethics board; SNP = single-nucleotide polymorphism; WGS = whole genome sequencing.

Summary of Main Findings

Limited Genomic Data in Atlantic Canada

FAIR standards require data to be findable, accessible, interoperable, and reusable.¹³³ Our search of the literature and communications with researchers found that there are rich genomic data collected that have been used for secondary research (reusable), but are not necessarily easily findable, accessible, or interoperable.

Data are generally held by individual data researchers rather than held in a centralized repository, and metadata on particular datasets are not typically held in any centrally accessed resource. Appendix 2 in the Supplemental Material document provides multiple genomics-related publications by province and the medical condition being investigated. Details could not be verified for some datasets that were cited in the published literature, including for spondylarthritis,⁴⁹⁻⁵³ hearing loss,⁷¹⁻⁷⁷ Stargardt disease,⁷⁰ Bardet-Biedl syndrome,⁶⁴⁻⁶⁹ and others, particularly in the Newfoundland and Labrador population. Additionally, not all researchers could be contacted; therefore, additional attempts may be warranted by researchers with an interest in specific areas of genomics (refer to Appendix 2 in the Supplemental Material document). This showcases the challenge of identifying relevant datasets, which in turn hampers accessibility of these resources. Notably, large datasets including genetic data on hundreds or thousands of individuals do exist with raw data available, mainly in Newfoundland and Labrador (refer to [Tables 1](#) to [Table 4](#)).

Clinical data are held in fewer locations and are more findable than datasets developed for research purposes. Clinical data poses accessibility and interoperability limitations. Clinics such as the clinical laboratories Maritime Medical Genetics in Halifax and the Dr. Georges-L.-Dumont University Hospital

Genetics Clinic do not currently allow access to the majority of raw genetic data. For Maritime Medical Genetics Service, which does most of the germline medical testing for New Brunswick, Nova Scotia, and Prince Edward Island, steps are being taken to repatriate raw data from third-party sequencers. Such data could provide a rich source of genomic data for research, provided a federated research platform is established. The use of these datasets for secondary research and linkage of genomic data to health administrative data is complicated by a lack of provision for these processes in clinical consent forms and siloed health data. Maritime Medical Genetics Service, Vitalité Health Network, and IWK Health have expressed interest in making data more widely available in a potentially linkable form. These organizations have taken steps to increase access to genomics data on individuals in Atlantic Canada. As more clinics consider secondary research in the medical consent process, and as electronic and centralized medical records become the norm, genomic data may become more accessible and interoperable.

In addition to researchers expressing interest in developing models that respect patient and participant privacy while also improving availability of genomic data for research, there are projects currently providing findable, assessable, and interoperable data on people in the Atlantic provinces. The ACC and Atlantic PATH ([Table 4](#)) have datasets and centralized repositories for data with publicly accessible protocols and processes to access both genomic data and support medical information. The ACC will eventually have easily accessible metadata through internet searches.

Finally, there is a lack of data on individuals of non-European ancestry within research projects conducted in the Maritimes. Genome Atlantic noted that the Canadian Precision Health Initiative seeks to collect genomic data specifically on Black individuals in Canada. The project was only announced in 2025, but the project plans to include people in Nova Scotia (personal communication, March 25, 2025).¹³⁴

Conclusion

This project aimed to identify and summarize the genomic datasets available for populations in Atlantic Canada. By summarizing the available data, our hope is that more researchers will gain awareness of where to find relevant datasets. This project also identified key genomics researchers in the Atlantic Canada region and areas of growth regarding access to genomic data. More data are needed on privacy policies and processes in place to improve accessibility while protecting patient and participant privacy and health.

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Conflicts of Interest

No conflicts of interest were declared.

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