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A Review of Genomic Data Holdings in the Atlantic Provinces: Supplemental Material

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Appendix 1: Inclusion and Exclusion Criteria

Please note that this appendix has not been copy edited.

Table 1: Inclusion and Exclusion Citeria

Eligibility	Inclusion	Exclusion
Criteria	• Population from Atlantic Canada (NB, NS, PEI, NL) • Human genetic data (Whole exome sequencing, whole genome sequencing, panels, individual mutations, other genetic data)	• Case studies (less than 25 people total receiving genetic testing) • Individual family units • Small samples sizes (less than 25 people), unless the small sample size is representative of all cases of a condition in a province. • Non-human genetic data (ex. from animals, viruses, bacteria, etc.)

Appendix 2: Extracted Literature on Genomic Data Collection and Analysis in Atlantic Canada

Please note that this appendix has not been copy edited.

Table 2: Genomic Datasets Collected and Analyzed in Multiple Atlantic Provinces

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/ Library/ Availability	Contact
Spinal Muscular Atrophy						
McKee-Muir, Dyack, Taillon, et al. (2023)[1]	IWK REB and Horizon Health Network REB. Waiver of full patient consent was obtained.	30 patients born from 2000–2020 with SMA due to positive SMN1 from the IWK Clinical Genomics Laboratory database and MMGS. (patients from NS, NB, and PEI)	DNA: Not specified. Additional data from health records at IWK, NS SHARE, and Horizon Health Network (NB).	Performed to detect the homozygous deletion of SMN1 gene exons 7 and 8.	DNA: MMGS clinical database Heath records: Nova Scotia Secure Health Access Record (SHARE), and Horizon Health Network No statement on data storage or availability.	Jordan Sheriko, Department of Pediatrics, Dalhousie University, Halifax jordan.sheriko@iwk.nshealth.ca
The Atlantic Partnership for Tomorrow's Health (Atlantic PATH) study						
Sweeney, Cui, DeClercq, et al. (2017)[2]	Participants were recruited between 2009 and 2015 with REB approvals in place for	Harmonized dataset for Atlantic PATH includes 34,000+ participants aged 30 to 69	• Questionnaires (demographic, comorbidities, cancer history, lifestyle habits, etc.).	Once processed, plasma, buffy coat, red blood cells, serum,	Can be linked to health data from administrative health databases: • Cancer Registries	Application for data process is found here: Data Access Process – Atlantic PATH

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/ Library/ Availability	Contact
	all 4 Atlantic provinces. Participants provided consent to be followed over a 30-year period and use of their data by Canadian and international researchers, and a research platform was designed specifically for this purpose.	(expanded to 74 after 2017).	• Physical measures (e.g., BMI, waist, hip) • Biosamples (blood, urine, saliva and toenail)s. ≥ 1 sample provided by 91% of participants (venous blood provided by 84%).	saliva and urine were transferred to 2-ml cryovials and stored in −80°C freezers. Toenail samples were collected in Ziplock bags and stored in cabinets at room temperature *Genotyped data are currently available for 1000 participants.	• Physician Billing • Hospital Discharge Abstracts • Vital Statistics (death) • Hospital-based and community-based ambulatory care, including emergency rooms.	

Table 3: Genomic Datasets Collected and Analyzed in NS

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
Lynch Syndrome/Endometrial Cancer						
Levesque, Wood, Carter, et al. (2023)[3]	Ethics approval from the Nova Scotia Health Research Ethics Board (1026033). *Consent not noted.	Nova Scotians diagnosed with endometrial cancer between 2017 and 2020. Tumour IHC staining was performed in 444 patients. 98 Received germline testing, 9 with germline tested had Lynch syndrome mutation	DNA sample (germline testing): Not stated Tumor biopsy and staining Also: Age, body mass index, tumour mismatch repair protein immuno-histochemistry results, personal and family histories (from patient charts and Tupper gynecologic database (internal database)	Specific genetic tests and level of availability of data to researchers not stated. Germline testing results, and personal and family histories of Lynch-associated malignant disease for all eligible patients were extracted from medical genetics records by an endometrial cancer, MMGS genetic counsellor. Data used in this study are available in an unidentified, coded format from the first author on reasonable request: Marianne. levesque@dal.ca.		Dr. Katharina Kieser Dalhousie University; Maritime Medical Genetics Service, IWK Health Centre; Division of Gynaecologic Oncology, Department of Obstetrics and Gynaecology, Dalhousie University, Halifax, NS kkieser@dal.ca
Fetal structural anomalies via Ultrasound						
Allen, Schollenberg, Aberg, and Brock (2025)[4]	IWK Health Research Ethics Board approval obtained May 21, 2019 (File #1024606), with annual renewal approval. IWK Health REB approved the Waiver of Consent due to impracticability.	Retrospective cohort (derived from the IWK Health Clinical Genomics Laboratory Information System) of 593 fetuses that had genetic testing performed due to structural anomalies identified by ultrasound in the Maritime Provinces from 2014 to 2022.	DNA Samples: chorionic villus sampling, amniocentesis, fetal blood, and products of conception Data combined with maternal health records review, EMR chart reviewed when appropriate.	First tier testing included rapid aneuploidy detection (RAD) and CMA, with subsequent sequence-based testing reserved for those cases meeting the selected testing criteria. Results of genetic testing, including RAD, karyotype, microarray (done in-house at IWK Health CGL), and gene sequencing (Single genes and small gene panels performed at the IWK CGL, larger gene panels and exomes performed and reported by referral laboratories.) For each eligible prenatal, cord, and newborn sample, an electronic health records chart review identified specific		Dr. Victoria Allen Department of Obstetrics and Gynaecology Dalhousie University and IWK Health, Halifax, Canada victoria.allen@dal.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				ultrasound findings, findings from other imaging modalities, involvement with medical genetic specialties, postnatal findings, and clinical outcomes including gestational age at delivery and pregnancy complications placental insufficiency, as well as newborn examination and autopsy findings. Dr. Victoria Allen Department of Obstetrics and Gynaecology Dalhousie University and IWK Health, Halifax, Canada victoria.allen@dal.ca		
Meier-Gorlin syndrome						
Guernsey, Matsuoka, Jiang, et al., (2011)[5] *Letter	Approval for the research study was obtained from the IWK Health Centre and CHU Ste-Justine research ethics boards. Informed consent was obtained for all participants in the study.	5 individuals with Meier-Gorlin syndrome: 1 Maritime-Acadian, 2 Quebec and 2 Louisiana-Acadian families (individuals 1652, 1768, 1882, 1899 and 1627) 314 local Maritime control chromosomes, which (include a substantial proportion of Acadians).	DNA Sample: blood samples Also, anthropometric measurements	High density genome-wide SNP genotyping using Illumina Human 610K panel of approximately 600,000 markers, used PLINK to search for shared haplotypes among subsets of the genotyped individuals Case and control samples sequenced at McGill University and Genome Quebec Centre for Innovation.	Illumina used to sequence data. Data from this study deposited in the RefSeq database under the following accession codes: human ORC4 mRNA, NM_002552; human ORC4 protein, NP_002543; human ORC1 mRNA, NM_004153.3; human ORC1 protein, NP_004144.2; human CDT1 mRNA, NM_030928.3;	Mark E Samuels mark.e.samuels@umontreal.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				Sequence traces analyzed using Mutation Surveyor.	human CDT1 protein, NP_112190.2.	
Renal cell carcinoma (RCC)						
Kokorovic, Thomas, Serrano-Lomelin et al., (2020)[6]	No research ethics approval listed. *Secondary data sources, no patient consent.	75 patients/family members who were evaluated for hereditary RCC syndrome and received genetic testing from January 2006 until December 2017 (collected retrospectively) received genetic testing	Clinical, pathological, and genetic analysis data.	Panel for genetic testing including FH, FLCN, MET, PTEN, SDHB, SDHC, SDHD, TMEM127, and VHL genes. Genetic testing was performed by the Clinical Genomic Laboratory, IWK, Halifax; Leeds Genetics Laboratory (Molecular Genetics), St. James' University Hospital, Leeds, UK, or Blueprint Genetics, Helsinki, Finland.	*Level of detail with genetic results available to researchers unclear *No statement on data availability.	Dr. Ricardo A. Rendon Queen Elizabeth II Health Sciences Centre, Halifax, NS, Canada; rrendon@dal.ca
Merkel cell carcinoma						
Walsh (2001)[7] Ly, Walsh, and Pasternak (2012)[8] Fleming, Ly, Pasternak et al. (2014)[9] Carter, Gaston,	study approved by the Capital Health Research Ethics Board, Halifax, Nova Scotia. (REB #1021594) *No mention of	Total of 142 study cases submitted through the Pathology Department of the Victoria General Hospital/Queen Elizabeth II Health Sciences Centre in Halifax, identified	Clinical information was obtained from pathology requisition forms, requesting physicians, and pathology reports. Thirteen MCC metastases were	No genetic testing conducted. IHC was performed on paraffin-embedded archival tissue to detect positivity for CK20 and CM2B4[7] For 83 cases in	*These cases were obtained through medical points of contact and patients required information on tests performed, therefore these are likely likable to	Dr. Sylvia Pasternak Dalhousie University Department of Pathology Halifax, Nova Scotia, sylvia.pasternak@nshealth.ca Noreen M.G. Walsh, MD,

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
Huang, et al. (2018)[10] DeCoste, Carter, Pasternak, et al. (2021)[11] DeCoste, Walsh, Gaston, et al., (2022)[12]	participant consent to study or secondary use.	through a search of laboratory computer records and review of a previously published case series of MCCs. All cases diagnosed between 1989 and 2020. Only those with available paraffin-embedded tissue samples included. 42 primary MCCs,13 MCC metastases, 57 non-MCC cutaneous lesions, 15 pulmonary neuroendocrine carcinomas and 15 GI NETs were included.	examined. Ten of these metastases were derived from 8 primary PMCC and 2 primary CMCC tumours already included in the study.	2014, new slides were cut from archived paraffin-embedded tissue blocks and stained with p63 as per the manufacturer's instructions (Clone BC4A4 [Biocare Medical, Concord, CA), CC1 antigen retrieval and iView Kit detection method [Ventana])[9] 46 cases, 21 pure Merkel cell polyomavirus (MCPyV)+, 10 pure MCPyV−, and 15 combined MCPyV− MCCs. DNA (80 ng) from each sample was probed with SNP-containing oligonucleotides targeting interrogation sites of 40 base pairs. Genome-wide CNA studies were performed for from 28 MCCs (9 MCPyV+, 9 MCPyV−, and 10 combined tumours). DNA	administrative data. DNA was extracted using QIAamp DNA FFPE (Qiagen Inc, Valencia, CA) or MagNA Pure Compact (Roche, Basel, Switzerland) commercial kits and quantitated using the Qubit dsDNA BR kit (Thermo Fisher Scientific Inc, Grand Island, NY). Affymetrix (Santa Clara, CA), conducted genome-wide CNA studies using the OncoScanFFPE gene chip probe array, Next-generation DNA sequencing data were processed using an in-house bioinformatics analysis pipeline conducted on an Illumina MiSeq gene sequencer (San Diego, CA). Processed using Picard and the Genome Analysis Toolkit following	Division of Anatomical Pathology, Queen Elizabeth II Health Sciences Centre, Halifax, NS Canada Dr. Ryan C. DeCoste Department of Pathology and Laboratory Medicine, QEII Health Sciences Centre), Halifax, NS, ryan.decoste@dal.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				from all 46 cases was analyzed by NGS using a custom set of primers for 8 tumour suppressor genes and 1 oncogene previously shown to have inactivating and activating mutations in MCC, (TP53, RB1, APC, NOTCH1, NOTCH2, MLL2, MLL3, BRCA1,and PIK3CA)[10] NGS of 51 cases were selected primary cutaneous Merkel cell carcinomas (21 MCPyV +, 13 pure MCPyV− and 17 combined MCPyV−) derived from the Maritime region of Canada (1993 to 2020). extracted DNA was analyzed via the Illumina TSO500 hybrid capture DNA panel, performed on an Illumina NextSeq550	community-developed best practices. Copy number aberration data from each sample were analyzed using Nexus Copy Number version 9.0 (BioDiscovery, El Segundo, CA).[10] Data were first analyzed using Illumina's TSO500 LocalApp workflow for alignment and variant calling of mutations 33 cases (9 MCPyV+, 10 pure MCPyV− and 14 combined MCPyV−) were sent to Mayo Clinic Laboratories (Rochester, MN) for Rb immuno-histochemical (IHC) staining, which was performed in accordance with their institutional protocols. The datasets used and/or analyzed during the current study are available from the corresponding	

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				instrument (San Diego, CA), in accordance with the manufacturer's instructions. The panel analyzes 523 cancer-related genes for small mutations, a subset of genes for copy number gains, as well as global parameters of tumour mutation burden (TMB) and microsatellite instability (MSI) Cases with median exon coverage < 100x (unique reads employed for all thresholds) were excluded from analysis[11,12]	author on reasonable request. [11,12]	
Exfoliation syndrome						
Li, Wang, Lee et al., (2021)[13]	Patients with exfoliation syndrome were enrolled following informed consent. Both studies approved by the REB of the NSHA (protocols 1011873 and 1020512).	Multiple cohorts from multiple countries included 403 cases, 392 controls included from NS (Supplemental materials) Reference controls were obtained from the DNA diagnostic laboratory serving the same	Venous blood samples Eye testing results	NS participants underwent whole exome Sequencing during first validation stage	Whole-exome sequencing and targeted sequencing (targeting the CYP39A1 [Refseq NM_016593] coding sequence) libraries were prepared using hybridization capture kits (Roche-Nimblegen SeqCap)	Chiea Chuen Khor, MD, Ph.D., Genome Institute of Singapore, 60 Biopolis St, 02-01 Genome Bldg, Singapore 138672 (khorcc@gis.a-star.edu.sg

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
		population. All participants were of self-reported European ancestry.			Cases and controls sequenced together using 2 × 151 base-pair, paired-end reads on Illumina instruments. Raw DNA sequence reads from the Illumina HiSeq 4000 and Novaseq 6000 instruments were aligned to the hg19 genome build. Variant detection was performed using exons and 50 base-pair regions flanking the exons. DNA sequence reads were mapped using Burrows-Wheeler Aligner software (version 0.7.16a-r1181). Variant calling was performed using the Genome Analysis Tool Kit (version 3.7) and the Picard (v *No statement of data availability *unclear if data can be linked to administrative data	

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Aung, Ozaki, Lee et al., (2017)[14]	For Canada, individuals with Exfoliation syndrome were enrolled following informed consent from within the Nova Scotia Health Authority (NSHA), Nova Scotia (protocols 1011873 and 1020512)	World-wide GWAS partnership study (Canada): 341 cases and 247 controls Canada Replication:136 cases, 747 controls All participants were of self-reported European ancestry	DNA samples obtained from ocular tissues	Deep sequencing was performed on a total of 5,570 XFS cases and 6,279 controls from 9 countries (excluding Canada (NS)). A genome-wide association study (GWAS) of Exfoliation syndrome cases and controls from 24 countries (including Canada) followed by replication in 18 countries (including Canada) identified 7 genome-wide significant loci.	Genome-wide genotyping was undertaken using the Illumina OmniExpress BeadChip. All 9,035 XFS cases and 17,008 controls for the GWAS discovery stage genotyped using Illumina OmniExpress array. In the replication stage, direct genotyping using the Sequenom Mass-Array and Applied Biosystems Taqman genotyping platforms was carried out for cases and controls from Nova Scotia (Supplemental materials).	Tin Aung Dept of Ophthalmology, Yong Loo Lin School of Medicine, National University of Singapore, Singapore aung.tin@singhealth.com.sg Francesca Pasutto Institute of Human Genetics, Friedrich-Alexander-Universität, Erlangen-Nürnberg, Erlangen, Germany Francesca.Pasutto@uk-erlangen.de Janey L. Wiggs Department of Ophthalmology, Harvard Medical School, Massachusetts Eye and Ear Infirmary, Boston, Massachusetts, US Janey_Wiggs@meei.harvard.edu Chiea Chuen Khor Genome Institute of Singapore, Singapore khorcc@gis.a-star.edu.sg
Multiple myeloma (MM)						
Cutler, Knopf, Campbell, et al., (2021)[15]	Conducted under ethical approval by the Nova Scotia Health Authority (NSHA)	77 patient samples (from 76 patients) bone marrow was collected from patients with	Patient bone marrow samples laboratory data, including	A custom panel to target exons of these 26 genes was designed	MM-specific targeted-sequencing approach using a custom-designed	Daniel Gaston, Ph.D., 5788 University Ave.,

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
	Research Ethics Board (1021520 and 1021397), and patients provided written informed consent for research.	plasma cell dyscrasias at the Victoria General Hospital (Halifax, NS, Canada)	albumin, $Beta_2$-microglobulin, lactate dehydrogenase, bone marrow plasma cell burden, serum M-protein quantity, immunoglobulin heavy and light chain type and quantity, serum free light chain ratio, and FISH data, coinciding with the time of bone marrow acquisition, were collected from the NSHA laboratory information system. Patient clinical data, including age, sex, diagnosis at time of bone marrow acquisition, therapies received, follow-up period, stage, and time of events, were also collected from the NSHA hospital information system.	using Illumina DesignStudio (Illumina, San Diego, CA). genes that were mutated in > 1% of the population of patients were included (25 genes) in published WES and WGS studies as well as *MYC*.	26-gene panel, the DMG26, that is applicable in a standard clinical molecular laboratory and demonstrates that the DMG26 captures prognostically relevant genomic abnormalities that are distinct from those captured by FISH. Genomic data generated during this study are available on request, with access managed by the Dalhousie Pathology Biobank. All programming scripts used in this work are available at GitLab, last accessed November 25, 2020). DNA library preparations for the DMG26 panel were performed per Illumina TruSeq and AmpliSeq for Illumina custom panel reference	Halifax, NS, Canada. dan. gaston@nshealth.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
					guides. Libraries were sequenced in 2 runs on an Illumina MiSeq at a mean depth of approximately 1000 times. FastQ files were analyzed using an in-house bioinformatic pipeline Data is linkable to administrative health data	
Familial exudative vitreoretinopathy (FEVR)						
Robitaille, MacDonald, Kaykas, et al., (2002)[16]	IWK Health Centre Research Ethics Board and the Oakland University Institutional Review Board of Human Subjects Investigation Committee approved these studies. Informed consent obtained.	81 subjects: 27 people with FEVR from NS and 54 of their family members without FEVR	DNA from blood samples	Determined genotypes of 8 family members using 6 DNA markers (D11S1887, D11S896, D11S4082, D11S1780, D11S873 and D11S1311) from the EVR1 locus. Performed Linkage analysis after collection of samples from additional family members with 4 Genethon	Genotyping using marker sequences from the Genome Database on an Applied Biosystems Prism 3100 Genetic Analyzer running GeneMapper software (Applied Biosystems), and we verified mendelian inheritance of alleles using the PedCheck program performed two-point linkage analysis using the MLINK program version 4.1p from the FASTLINK	Dr. Michael R. Hayden Xenon Genetics, Inc., Burnaby, BC mrh@cmmt.ubc.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				(D11S1887, D11S4082, D11S1780, D11S1311) and 2 Marshfield markers (D11S896, D11S873). Examined an additional 11 Genethon, 7 CHLC and 9 novel CA-repeat markers (33 total)(fine mapping). Haplotype construction using 33 polymorphic markers revealed 5 proximal and 4 distal recombination's (5 were affected, 4 unaffected). Analyzed the coding regions of FZD4 and FLJ22104 for mutations 3 family members (2 affected, 1 unaffected) by sequencing PCR amplified fragments. Used PCR–RFLP analysis and	software package, and multipoint linkage analysis using VITESSE version 1.0 and SIMWALK version 2.8, assuming a disease allele frequency of 0.000 Designed intronic primer pairs for each exon in FZD4 and FLJ22104 using the Primer3 program. Used an Applied Biosystems Prism 3100 Genetic Analyzer to detect M493_W494del and L501fsX533 mutations.	

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				fluorescence capillary electrophoresis to screen the remainder of the family and a control population for presence of mutations observed in the affected individuals. Detected both the M493_W494del and L501fsX533 mutations.		
Ells, Guernsey, Wallace et al., (2010)[17]	Approvals obtained from IWK Health Center, Calgary Health Region (Alberta Children's Hospital site), and the St Joseph Health Care London. Consent obtained according to the Canadian Tri-Council guidelines.	104 premature infants recruited both prospectively and retrospectively between March 1, 2003 and April 30, 2008, 71 with severe ROP, 33 mild to no ROP. 173 random Canadian samples *Number from NS not stated	Tissue samples (blood or buccal swabs)	Direct automated sequencing of the 2 coding exons of the FZD4 gene. Mutation detection was performed both manually and using Mutation Surveyor	Primers can be requested from the corresponding author. *No other statements of data availability.	Johane Robitaille Eye Care Team, IWK Health Center, University Ave. Halifax, Nova Scotia Canada. E-mail: jrobitai@dal.ca
Robitaille, Gillett, LeBlanc et al., (2014)[18]	Approved by IWK REB. Written informed consent obtained from participant	72 probands recruited from April 1998-September 2013. Unclear how many from Halifax.	DNA extracted from blood or saliva samples Collected clinical data retrospectively.	WES conducted. Also direct automated Sanger sequencing of 2 coding exons of the FZD4 gene, the 23 coding exons	WES conducted at McGill University and Genome Quebec Innovation Centre. DNA sequenced with 100 base pair paired-end reads (HiSeq	Dr. Johane M. Robitaille University Avenue, PO Box 9700, Halifax, NS, jrobitai@dal.ca.

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				of the LRP5 gene, and 22 coding exons of the KIF11 gene.	2000 sequencer; Illumina). Genomic variants called using Genome Analysis Toolkit pipeline and annotated with ANNOVAR. Specific exons sequenced using Sanger sequencing performed with ABI PRISM 3100 Genetic Analyzer and mutation calling was done by Mutation Surveyor, version 3.97)	
van der Ende, Bedard, Wallace, et al., (2025)[19]	Approved by the REB of the IWK Health Centre. Consent obtained in accordance with Declaration of Helsinki.	Patients with retinal features compatible with FEVR, affected relatives, and 1st-degree relatives were recruited between 1998 and 2024. Excluded people with retinopathy of prematurity. 94 people (76 had WES, 18 had Sanger sequencing data.)	DNA was extracted from peripheral blood or saliva samples Also: results from eye examinations and IV fluorescein angiography and fundus photography (if available). Ethnicity provided at discretion of referring physician.	Sanger sequencing was used to identify rare variants in known genes, and this method alone identified a molecular diagnosis for 18 probands. 76, including 2 diagnosed with Sanger sequencing in prior publications. A virtual FEVR gene panel was used in those 76 probands to	WES completed using Illumina NextSeq 550. Genomic variants called using the Genome Analysis Toolkit and Picard. Variants were annotated with Annovar and compared against dbSNP, 1000 Genomes project, and the genome aggregation dataset version 4. No statement of data availability.	Dr. Johane M. Robitaille University Avenue, PO Box 9700, Halifax, NS, jrobitai@dal.ca. Christopher R. McMaster, Department of Pharmacology, Dalhousie University Halifax, NS, christopher.mcmaster@dal.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				identify variants in *FZD4*, *LRP5*, *TSPAN12*, *NDP*, *ZNF408*, *CTNNB1*, *KIF11*, *ATOH7*, *ILK*, *JAG1*, *TGFBR2*, *RCBTB1*, *DOCK6*, *ARHGAP31* (, *NOTCH1*, *CTNNA1*, *CTNND1*, *DLG1*, *COL9A1*, *LRP6*, *TUBGCP4*, *TUBGCP6*, *PLK4*, *CDK19*, and *LAMA1.* At minimum, the Gene, Nucleotide Change, Amino Acid Change, Variant Type, REVEL, CADD, SpliceAI, gnomAD, Sex, ACMG ranking, SVI evidence were collected.	Data likely linkable to administrative health data as patents recruited through health care system.	
Colonic mixed adenoneuroendocrine carcinoma and neuroendocrine carcinoma						
Sinha, Gaston, Manders, et al., (2018)[20]	Approved by our institutional research ethics board.	All colonic tumours (269?) with a previous diagnosis of MANEC, high-grade NEC, and adenocarcinoma with neuroendocrine differentiation were	Tumor samples. Samples deidentified, and tissue cores obtained from an area containing at least 80% viable tumour cells.	Sought to identify unique chromosomal alterations of MANEC and NEC by comparing their genome-wide	comparison colorectal adenocarcinomas investigated and analyzed by the OncoScan FFPE assay	Dr. Weei-Yuarn Huang Division of Anatomical Pathology, Queen Elizabeth II Health Sciences Centre and Dalhousie University, Halifax, Nova

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
		retrieved from the archives of the Department of Pathology, QE II Health Sciences Center, between July 2000 and July 2015. 269 colorectal adenocarcinomas used as controls	Immuno-histochemistry (IHC) of tumour sections	CNAs to those of 269 conventional adenocarcinomas characterized the genome-wide copy number aberrations of 14 MANECs and 5 neuroendocrine carcinomas using the OncoScan FFPE (Affymetrix, Santa Clara, CA) assay. Genomic Identification of Significant Targets in Cancers algorithm identified 7 peaks that drive the tumorgenesis of MANEC. Genomic Identification of Significant Targets in Cancer (GISTIC) analysis was used to distinguish significant chromosomal aberrations from random background. CNA data generated for colonic MANEC	and Nexus copy number software the OncoScan FFPE assay from Affymetrix (Santa Clara, CA) was performed on 19 samples. Raw data analyzed using Nexus Copy Number 7.5 Discovery Edition software (BioDiscovery, El Segundo, CA).	Scotia, Canada B3H 1V8. huangw@nshealth.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				and NEC were compared colorectal adenocarcinomas. For statistical comparison of copy number alternations between MANEC and adenocarcinoma, false discovery rate–corrected *P*s were then obtained using the Benjamini-Hochberg procedure. MSI kit from Promega (Madison, WI): The presence of the *BRAF* V600 mutation was detected using a SNaPshot-based assay that can detect the V600E, V600K, V600M, V600R, and V600D mutations		
Bipolar disorder						
Hou, Heilbronner, Degenhard, et al. (2016)[21]	Written informed consent was obtained from all participants. Ethical and regulatory	GWAS of lithium response in 2563 patients with DSM III or DSM-IV diagnosis	Whole blood samples. assessment	largest-ever set of genomic data from BD patients treated with lithium	Samples were genotyped at the National Institute of Mental Health	Thomas G Schulze, Institute of Psychiatric Phenomics and Genomics (IPPG), Medical Center of

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
Stone, Nunes, Akiyama, et al. (2021)[22] O'Connell, Lee, Koromina, et al. (2025)[23]	approvals were obtained at each site that contributed anonymized data and DNA to the analysis.	of a bipolar spectrum disorder, collected by 22 participating sites from the International Consortium on Lithium Genetics. Patients required to have taken lithium for ≥ 6 months with no additional mood stabilizer (2008 and 2013). One site was Halifax. They conducted GWAS in 2 parts: Halifax: 353 total, GWAS 1: 241, GWAS 2: 112	methods: semi-structured clinical interviews (clinical), medical records, registries and questionnaire data (community) and self-reported surveys)	for maintenance therapy over a duration of at least 1 year, from 14 international site members of the Consortium on Lithium Genetics (ConLiGen) and 79 cohorts. A total of 3193 participants were genotyped; 2563 remained after quality control (1162 in GWAS 1 and 1401 in GWAS 2). Data from common single nucleotide polymorphisms (SNPs) were tested for association with categorical and continuous ratings of lithium response. Lithium response was measured using a well established scale (Alda scale). Identified 337 linkage disequilibrium-independent genome-wide significant variants	(Bethesda, MD, US), Life and Brain Center at the University of Bonn (Bonn, Germany), or Broad Institute (Cambridge, MA, US) using either Affymetrix or Illumina SNP arrays Halifax: 241 individuals genotyped with an SNP array from Illumina Omni1-Quad in GWAS1 and 112 individuals genotyped with Illumina OmniExpress 1·1 in GWAS 2 (supplementary materials). After basic quality control and linkage disequilibrium (LD) pruning, about 40K directly genotyped SNPs shared across all SNP arrays were used for the relatedness testing by PLINK.	the University of Munich, Munich, Germany thomas.schulze@med.uni-muenchen.de

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				that map to 298 loci		
Youth Anxiety disorders						
McAusland, Burton, Bagnell, (2024)[24] *Study protocol* Not yet conducted	Eligible youth will complete the consent form and may opt-in to long-term storage of their DNA sample. In some cases parental consent will be obtained with youth assenting to participate.	The goal is to recruit 13,000 youth aged 10 to 19 of which 50% are expected to endorse symptoms of anxiety that indicate the presence of an anxiety disorder. The GAYA study with local sites in Calgary, Halifax (IWK Health Centre community), Hamilton, and Vancouver. Clinical recruitment approach and online recruitment (Social media content and visuals)	An OG-600 Oragene saliva DNA sample kit or choose to have an OCR-100 ORA collect DNA sample kit mailed to their home with a pre-paid return envelope. Also, participants will be asked to complete the Screen for Child Anxiety Related Emotional Disorders (SCARED), the Short Mood and Feeling Questionnaire (SMFQ), the CPSS-SR-5, the Behavioural Inhibition and Behavioural Activation System (BIS/BAS) scales. Also 5 optional surveys.	Genetic Architecture of Youth Anxiety (GAYA): De-identified research IDs will link data and saliva samples. The linked IDs will be logged in REDCap to document and track the handling of biologic samples. DNA extraction from saliva is performed at each individual site following best practice and according to the manufacturers' protocols. Subsequent genotyping will be performed in 3 batches. All individuals will be genotyped.	genotyped (using DNA from the saliva samples) with Illumina's Global Screening Array v3.0 (GSA). For genotyping calling Illumina's GenomeStudio will be used. Spit for Science genotyping will be done locally using the same Illumina array and their genetic data are sent to the Halifax site once the participant has completed the questionnaires and/ or app portion of the study.	Dr. Laina McAusland Department of Psychiatry, University of Calgary, Calgary, AB, Canada

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
Autosomal Dominant kidney disease						
Kmochová, Kidd, Orr, et al. (2024)[25]	Approved by Nova Scotia Health REB	5 families found to have autosomal dominant medullary amyloidosis due to 2 different pathogenic APOA4 variants. From sites in United States, Czech Republic, and Nova Scotia, Canada (at least 32 people from 1 family)	Whole blood or whole saliva	Whole exome of whole genome sequencing underwent whole genome sequencing with identification of a chr11:116692578 G>C (hg19) variant encoding the missense mutation p.L66V of the ApoA4 protein.	Sanger Sequencing	Dr. Anthony Bleyer Wakefeild School of Medicine Winston-Salem, North Carolina ableyer@wakehealth.edu

Table 4: Genomic Datasets Collected and Analyzed in NB

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
Breast carcinoma						
Ouellette, Richard, and Maïcas, (2004)[26]	REB approval not specified. Consent for both surgical procedures and SLN analysis was obtained from all patients.	53 consecutive patients (42 patients retained for analysis) with breast CA (cases) and 25 patients with melanoma (controls) at CHU Dumont from 1999 to 2002.	SLN biopsy	RNA and DNA from samples assessed for genes coding for MGB1 and MGB2 using PT-PCR testing.	Not specified.	Not specified. Dr. Rodney Ouellette, MD Ph.D. Dr Georges-L. Dumont Hospital University of Moncton
Gauvin, Allain, Bouhamdani, Williams, et al. (2025)[27]	Approved by the Vitalité Health Network REB (#101703). No consent due to retrospective design	445 participants from NB (Acadian and French Canadian, European descent (non-French or Acadian), South-American, Native American, African or Asian origins) tested for	Not specified. Data on other health information collected from patient charts.	Multi-gene panel testing (268), or targeted predictive testing for a known familial variant (39) for genes known to be associated with breast CA.	Paired-end sequencing with Illumina sequencers. Data is available upon request.	Eric Allain eric.allain@vitalitenb.ca Mouna Ben Amor mouna.benamor@vitalitenb.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
		hereditary breast C gene from 2019 to 2022		Genetic testing performed by Fulgent Genetics or GeneDx.		
Myeloid neoplasms						
Northrup, Maybank, Carson, et al. (2020)[28]	Approved by the Horizon Health Network REB	178 (156 from Saint John, 22 other locations) randomly selected specimens from patients with Myeloid Neoplasms submitted for the NGS Ion AmpliSeq Myeloid Panel (2017) or the Oncomine Myeloid Panel (Thermo Fisher)(2018)	43 bone marrow aspirates and 136 peripheral blood samples	NGS of a genetic panel of genes related to hematologic malignancies Additionally, karyotyping done in 75 cases	Library preparation performed using Ion AmpliSeq Myeloid Panel (145 samples) or Oncomine Myeloid Panel (33 samples) at the Saint John Regional Hospital.	Victoria Northrup, MSc, Dalhousie Medicine NB, Saint John, Canada; and Departments of Laboratory Medicine and Research Services, Saint John Regional Hospital, Saint John, Canada.
Healthy participants with Acadian heritage						
Robichaud, Allain, Belbraouet et al. (2022)[29]	Vitalité Health Network REB Consent obtained from all subjects for DNA collection and project participation	60 from Southeast NB (all at least 1 Acadian grandparent born in the region) *Plans to expand to more areas of NB	DNA samples collected using the Oracollect DNA buccal swab from DNA Genotek inc.	DNA from oral swabs was extracted by Fulgent Genetics. WES of 312 autosomal recessive and 30 X-linked diseases connected with French ancestry	NGS libraries generated using modified KAPA DNA Library. Prepared DNA libraries sequenced using a HiSeqX or NovaSeq 6000 from Illumina inc. (San Diego, CA, US). Data restricted due to higher risk of re-identification from genetic information	Dr. Mouna Ben Amor Department of Medical Genetics, Vitalité Health Network, Dr. Georges L.-Dumont University Hospital Centre, Moncton mouna.benamor@vitalitenb.ca
Pancreatic ductal adenocarcinoma						
Roy, Wajnberg, Ouellette, et al. (2023)[30]	Approval obtained from Vitalité Health	16 PDAC patients and 13 non-affected individuals	Plasma samples	Assessed utility of the Vn96 synthetic peptide	Tumoral Biobank at CHU Dumont	Stephen M. Lewis Atlantic Cancer

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
	Network REB consent obtained from patients			for the isolation of extracellular vesicles from human plasma and the subsequent detection of small RNA biomarkers of PDAC by NGS analysis.	and the Ontario Pancreas CA Study at Mount Sinai Hospital (Toronto, ON). No statements of data availability.	Research Institute, Moncton, NB, Canada Beatrice Hunter Cancer Research Institute, Halifax, NS, Canada Department of Chemistry and Biochemistry, Université de Moncton, Moncton, NB, Canada stephenl@canceratl.ca

Table 5: Genomic Datasets Collected and Analyzed in NL

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
General Newfoundland and Labrador population						
Zurel, Bhérer, et al. (2025)[31] Gilbert, Zurel, et al. (2023)[32]	NLGP protocol was approved by the NL Health REB (#: 2018.243). Participants consented to secondary use, pending access requests for scientific research through Sequence Bio board.	Random recruitment of participants from general practice clinics across the province aged ≥ 18 years, possessed a valid NL health card, and provided written consent. 2,500 participants total, including people of Indigenous heritage, 1,807 NL individuals of European descent for the NLGP cohort. Additionally, included data from the Irish DNA Atlas and the People of the British Isles.	Saliva samples were obtained and medical histories provided by all 2,500 participants. Information also collected on religion and the birthplace of their parental ancestors.	Targeted SNP genotyping: 1,110 Y chromosomes from an NL-based cohort were analyzed using 5,761 Y-specific SNPs. Y-chromosome haplogroup comparison – compared SNP data with Irish DNA Atlas and People of the British Isles. Rare variant analysis – checked 2,114 Y-SNPs against the gnomAD database to find population-specific markers. Phylogenic tree was constructed using the SNP data from the haplotype analysis. WGS was conducted. A quality threshold of 99.2% SNP pass rate per sample resulted in 2,446 individuals and 1,721,246 SNPs passing initial genotyping QC. Used Linkage disequilibrium and haplotype analyses comparing to SNPs from other datasets to determine participants with European ancestry from this paper. This resulted in 1,807	DNA extracted from these samples was genotyped using the Illumina Global Diversity Array (GDA; Illumina, San Diego, CA). fineSTRUCTURE. was used for LD and haplotype analyses. The genotype and sample metadata from the NLGP are not publicly available due to participant recruitment conditions and consent agreements that protect the privacy of NLGP participants. Researchers interested in accessing the NLGP data are encouraged to contact Sequence Bioinformatics (info@sequencebio.com).	Michael S. Phillips Sequence Bioinformatics Inc., St. John's, NL pgxdoc@gmail.com Dr Edmund Gilbert School of Pharmacy and Biomolecular Sciences, Royal College of Surgeons in Ireland, Dublin, Ireland FutureNeuro SFI Research Centre, Royal College of Surgeons in Ireland, Dublin, Ireland edmundgilbert@rcsi.ie

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				individuals and 685,221 common SNPs. performed haplotype-based clustering using *fineSTRUCTURE* identified 22 discrete clusters which summarize *fineSTRUCTURE*'s		
Rahman, Jones, Curtis, et al. (2003)[33]	No mention of consent or REB	200 consecutive unrelated individuals presenting to a blood collection service in St John's. Recruited subjects from St John's because it represents the 'worst case' scenario in demonstrating extended LD in NL	Blood samples	Genotyped > 1,300 individual SNP sites but > 200 of these sites on the chip consistently failed, yielding most of the no-signal calls so only 1,064 markers were analyzed. Estimated the LD between 2 SNPs using the standardized multiallelic disequilibrium coefficient D genotyped 200 done by estimating haplotype frequencies for the 200 subjects using the EH program limited analyses to the 591 marker pairs.	Genotyped using the Affymetrix HuSNP protocol according to the manufacturer's instructions. No statements of data availability.	Dr. Proton Rahman had full access to data Memorial University of NL, St. John's, NL prahman@mun.ca
Zhai, Zhou, Woods, et al. (2016)[34]	Approved by the NL Health REB. As part of a colorectal cancer research study	494 unaffected individuals recruited between 2001 and 2003 by random digital dialing across the NL to serve as controls Age matched to	Provided a blood sample for DNA extraction Gave information on their family sufficient to construct ≥ 3-generation pedigree Used public records to determine the historic	WGS using 1.2 million SNPs multidimensional scaling (MDS) analysis were carried out using PLINK. First included all 14 populations; the second included the NL, IRE, BBC and NAS data; the third included only the NL, IRE and BBC data Linkage disequilibrium	genome-wide SNP data genotyped using an Affymetrix Axiom Genome-Wide Array The genotype data of the NL samples are available with accession number GSE74392. Religion/ ethnicity data of the NL samples are available	Dr G Zhai Discipline of Genetics, Faculty of Medicine, Memorial University of NL, St John's, NL guangju.zhai@med.mun.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
		cancer patients but only inclusion criterion was the absence of a personal history of colorectal cancer. 52 individuals with < 2 generations of NL ancestry were excluded. Reference populations include Irish (211), British (450), Native American (108) and HapMap3 populations. Individuals with > 3% missing SNP data were excluded, as were SNPs that were not successfully typed in > 5% of participants.	religious affiliation of each family Provided questionnaire that gave information on personal, parental and grand-parental ethnicity. Public list of names on the Qalipu Mi'kmaq First Nation Band Order, with genealogical methods, used to confirm ancestral connection to Aboriginal populations.	was also conducted 86 individuals randomly selected from each population.	at request to the author (guangju.zhai@med .mun.ca) subject to an appropriate ethics approval. genotyping of the NL samples conducted by Dr Stephen B Gruber, USC Norris Comprehensive Cancer Center, University of Southern California as part of the ColoRectal Transdisciplinary Study (CORECT). The study makes use of data generated by the Wellcome Trust Case-Control Consortium.	
Crohn disease						
Zipperlen, Peddle, Melay et al. (2005)[35]	Approved by MUN REB. Consent obtained from patients.	128 with Crohn disease and 103 controls, all white and of NL ancestry.	Peripheral whole blood samples Collected demographic and health data from participants.	DNA extracted from lymphocytes with the Wizard Genomic DNA Purification Kit from Promega (Madison, WI). Genotyped for 5 SNPs of the TNF-Alpha gene	The products cleaned and spotted onto a SpectroChip. Chip scanned with a mass spectrometry workstation (Bruker). Resulting spectra analyzed with Sequenom SpectroTYPER-RT	Dr. Proton Rahman had full access to data Memorial University of NL, St. John's, NL prahman@mun.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
					software. *No statement about data availability	
Psoriatic Arthritis/Psoriasis						
Gladman, Farewell, Pellett, et al. (2003)[36]	Not specified.	65 sibling pairs from a PsA Clinic in NL (50 pairs affected by PsA, and/or psoriasis), 117 from a clinic in Toronto.	DNA sample not specified. Collected clinical history, physical, and radiologic data.	Extracted DNA assessed for SNP for each HLA-A, -B, -C, -DR, and -DQ loci. Also, clinical history, physical examination by a rheumatologist, and radiologic assessment	Not specified.	Dafna D Gladman Centre for Prognosis Studies in The Rheumatic Diseases, Toronto Western Hospital, Toronto, ON dafna.gladman@utoronto.ca
Butt, Sun, Greenwood, et al. (2005)[37] *Letter to the editor, unable to find initial reference	Approved by MUN REB. Consent obtained from all PsA patients	259 PsA probands and 238 ethnically matched controls all from the NL founder population	Not specified.	All subjects genotyped for: 3 SNPs in *SLC22A4* (rs3792876, 1050152, rs3763112), 1 SNP in *SLC22A5* (rs2631367), 1 SNP in *SLC9A3R1* (rs734232), and 1 SNP in *RUNX1* (rs2268277)	DNA was extracted using the Promega Wizard Genomic DNA purification kit. The Sequenom MassArray platform using time-of-flight mass spectrometry used for sequencing	Dr. C. Butt provided this in correspondence with: Dr. Proton Rahman Memorial University of NL, St. John's, NL prahman@mun.ca
Butt, Sun, Peddle, et al. (2005)[38]	Approved by MUN REB. Consent obtained from all participants.	234 patients with PsA and 88 ethnically matched, unrelated controls from NL founder population.	Whole blood samples	All subjects genotyped using SNP technology for: NFkB1-94delATTG RelA rs2009453 RelA rs6591183 NFKBIA-410 (rs2233409) NFKBIA-673 (rs2233407) NFKBIA-949 (rs2233406) NFKBIA 2578 (rs696)	DNA extracted using Promega Wizard Genomic platform. Polymerase chain and extension reactions designed using MassARRAY design software. Primer extension products cleaned and spotted onto a SpectroChip. Genotypes determined using the Sequenom SpectroTYPER-RT software.	Unclear who to contact but this contact is provided in other articles with this group: Dr. Proton Rahman full access to data. Memorial University of NL, St. John's, NL prahman@mun.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
					*No statement about data availability	
Butt, Peddle, Greenwood, et al. (2006)[39]	Approved by MUN REB and UofT REB. Consent obtained from all participants.	238 NL PsA patients and 149 healthy NL controls. All PsA probands and controls similar ethnicity to the patients (cases). *410 participants were also included from Toronto.	Whole blood samples Collected demographic information and data on disease onset and patterns.	All subjects genotyped using SNP technology for polymorphisms: *PPTN22* (rs2476601, R620W) and *tp53* (rs1042522, Pro72Arg).	DNA extracted using Promega Wizard Genomic DNA purification Kit. PCR and extension reactions designed using MassARRAY design software. Primer extension products cleaned and spotted onto a SpectroChip. Genotypes determined using the Sequenom SpectroTYPER-RT software. *No statement about data availability	Dr. Proton Rahman had full access to data. Memorial University of NL, St. John's, NL prahman@mun.ca
Rahman, Siannis, Butt, et al. (2006)[40]	Approved by MUN REB and the UofT. Consent obtained from all patients.	237 PsA cases and 103 controls from NL and 203 cases 101 controls from Toronto. Control subjects were of similar ethnicity to cases.	Peripheral blood samples (lymphocytes) Collected demographic information and data on disease onset and patterns.	DNA genotyped for 5 TNF variants by 5 SNPs were in the 5′ flanking region of TNFalpha gene positions: −1031 (T→C), −863 (C→A), −857 (C→T), −308 (G→A), and −238 (G→A).	Genotyped using time of flight mass spectrometry using Sequenom platform. DNA extracted using Wizard Genomic DNA purification kit from Promega. The products cleaned and spotted onto a SpectroChip. The chip scanned with mass spectrometry workstation (Bruker). Resulting spectra analyzed with Sequenom SpectroTYPER-RT software. *No statement about data availability	Dr. Proton Rahman Memorial University of NL, St. John's, NL prahman@mun.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
Rahman, Sun, Peddle, et al. (2006)[41]	Approved by MUN REB. Consent was obtained from all participants.	212 PsA cases and 150 ethnically matched controls. Controls were volunteers from NL who participated in as a result of a local campaign seeking controls for genetics studies.	Whole blood samples Information collected including age at onset of psoriasis and PsA, and disease pattern.	DNA extracted using Wizard Genomic DNA Purification kit. DNA genotyped by 29 SNPs within the *IL1* gene cluster (but specifically located within the *IL1A*, *IL1B*, and *IL1F5–10* genes).	Genotyped using time of flight mass spectrometry using Sequenom platform. DNA extracted using Wizard Genomic DNA purification kit from Promega. Products cleaned and spotted onto a SpectroChip. Chip scanned with mass spectrometry workstation (Bruker). Resulting spectra analyzed with Sequenom SpectroTYPER-RT software. *No statement about data availability	Dr. Proton Rahman Memorial University of NL, St. John's, NL prahman@mun.ca
Butt, Lim, Greenwood, et al. (2007)[42]	Approved by MUN REB. Consent was obtained from all participants.	258 PsA probands and 154 unrelated controls. All from the NL founder population.	Whole blood samples. Information collected including age at onset of psoriasis, PsA and disease pattern	Participants genotyped for SNPs: VEGF gene (rs3025039, rs699947, rs1570360, and rs2010963) FGF1 gene (rs34011) FGF2 gene (rs1048201) EGF gene (rs4444903, rs11568943 and rs2237051)	DNA extracted using the Promega Wizard Genomic DNA purification kit. Polymorphisms typed using Sequenom chipbased MALDI-TOF mass spectrometry platform using MassArray software (Sequenom). Products cleaned and spotted onto SpectroChip. Chip scanned with mass spectrometry workstation (Bruker). Resulting spectra analyzed with Sequenom SpectroTYPER-RT software. *No statement about data availability	Dr. Proton Rahman Memorial University of NL, St. John's, NL prahman@mun.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
Rahman, Inman, Maksymowych, et al. (2009)[43]	Approval from MUN REB and UHN, UofT. Specimens collected from patients with PsA with informed consent.	496 (247 NL, 249 Toronto) PsA probands and 476 controls (228 from NL, 248 from Toronto). All patients and controls mostly of North European ancestry. Controls for the NL population were volunteers who participated from a local campaign seeking population-based controls for genetic studies.	DNA: Not specified. Information collected including age at onset of psoriasis and PsA, and disease pattern.	Cases and controls genotyped for 11 SNPs in IL-23R: (rs1004819, rs7517847, rs7530511, rs10489629, rs2201841, rs11465804, rs11209026, rs1343151, rs10889677, rs11209032, and rs1495965.)	The Sequenom MassARRAY system used to genotype each study participant. Genotypes determined using a MassARRAY compact analyzer (Sequenom). *No statement about data availability	Dr. Proton Rahman Memorial University of NL, St. John's, NL prahman@mun.ca
Pollock, Chandran, Pellett, et al. (2013)[44]	Study approved by the UHN REB and all subjects provided written informed consent	374 PsA/PsC) probands and 115 health ethnically matched controls recruited from MUN, St. John's. *745 cases and 547 controls from Toronto also included.	Whole blood samples Data including age at onset of psoriasis and PsA, disease pattern, rheumatologist assessment	Genomic DNA extracted using the Gentra Pure Gene Blood Kit (Qiagen, Mississauga, ON). SNP technology used to genotype HLA-B and HLA-C and MICA-129 polymorphism rs1051792	Unclear, genotyping done through Life Technologies, Mississauga, ON. No statement provided regarding data availability.	Dr Dafna D. Gladman PsA Program Centre for Prognosis Studies in the Rheumatic Diseases Toronto Western Research Institute University of Toronto Toronto, ON dafna.gladman@utoronto.ca
O'Rielly, Pollock, Zhang, et al.	*Not stated	Investigated the differential methylation pattern among	*Not stated. Likely a blood sample	Genome-wide DNA methylation profiling was performed.	Genome-wide DNA methylation profiling was performed using Illumina HumanMethylation450k	*Not provided

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
(2014)[45] *Abstract only		paternally (n = 24) and maternally (n = 24) transmitted PsA. 48 samples provided by 48 participants		Methylation analysis was performed on 382,024 of the 485,512 CpG sites after filtering. Revealed 90 significant CpG sites (P < 0.05). The 3 most significant CpG sites were hypermethylated regions located on chromosome 8 that did not reside on or adjacent to a gene.	Beadchip, which measures up 480,000 different CpG sites per sample and covers 96% of RefSeq genes. No information on data availability	
Chandran, O'Reilly, Haddad, et al. (2014)[46] *Only abstract available*	*Not provided, only abstract available*	All patients were Caucasians of Northern European Ancestry. 24 PsA patients with mutilans (Patients with ≥ 1 joint with grade 4 damage were classified as having arthritis mutilans) and 24 patients without arthritis mutilans (no damage following similar duration of follow up).	blood DNA samples	to investigate DNA methylation	methylation profiling was performed using Illumina HumanMethylation450k Beadchips, which measures up ~480,000 different CpG sites per sample and covers 96% of RefSeq genes. No information on data availability.	*Not provided.
O'Rielly, Zhang, Codner, et al. (2015)[47] *Only abstract available	Not specified.	41 from NL with PsA treated with TNF-alpha Inhibitors (21 considered TNFi responders, 20	Not specified.	Genome-wide DNA methylation profiling performed using Illumina HumanMethylation450k Beadchip (480,000 different CpG sites per sample and	Not specified.	Not specified. All authors from Faculty of Medicine, Memorial University of NL, St. John's, NL Dr. Darren O'Rielly

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
		secondary TNFi failures		covers 96% of RefSeq genes)		St. John's, NL Darren.orielly@med.mun.ca
Pollock, Thavaneswaran, Pellett, et al. (2015)[48]	The study was conducted with the approval of the UHN REB, and all subjects provided written informed consent.	Cutaneous psoriasis without arthritis (PsC) and PsA 92 NL PsA probands	DNA-Not specified. Family history	PsC or PsA susceptibility alleles *HLA–C*01, C*02, C*06, C*12, HLA–B*07, B*08, B*27, B*38, B*39, B*57, HLA–DR4, DR7, DQ*0303*, and *MICA–129* **Included in analysis, not specified how this was collected.*	*Not specified.	Dafna D. Gladman, MD, FRCPC, Full access to data in study University of Toronto Psoriatic Arthritis Program, Toronto, ON dafna.gladman@utoronto.ca
Stuart, Nair, Ellinghaus, et al. (2010)[49]	All participating subjects gave informed consent and protocols were reviewed and approved by local institutional review boards.	Replication study following meta analysis, including 4,064 cases and 4,685 controls from Michigan, Toronto, NL (n=368 cases, 358 controls) and Germany	Not stated	Collaborative Association Study of Psoriasis (CASP) was included in the initial meta-analysis (includes 368 cases and 358 controls from NL). Following meta selected 102 SNPs for replication based on their p value rankings either in a subset of 350 **CASP** PsA cases vs. normal controls Replication sample consisted of up to 4,064 PsV cases and 4,685 controls from Michigan, Toronto, NL, and Germany. a meta-analysis of 2 recent psoriasis genome-wide association studies First-stage replication genotyped all selected	Sequencing software, and data libraries not stated No statement of data availability.	James T Elder Ann Arbor Veterans Affairs Hospital, Ann Arbor, Michigan, US, jelder@umich.edu

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				SNPs in the large Michigan sample; 91 SNPs passed all quality control (QC) filters. Second stage replication typed all but 1 of 51 QC-filtered SNPs from set A of the PsA GWAS in the samples from Toronto and NL, which contain most of the PsA cases. SNPs with the most promising association results were then followed-up with third stage typing--8 from sets B and C in the 2 Canadian samples, and 6 from all 3 sets in the German sample.		
Stuart, Philip, Nair, et al. (2015)[50]	All participants provided written informed consent and were recruited according to the protocols approved by the institutional review board of each institution.	1430 cases (323 from NL) and 1417 controls (323 from NL) (no PsA or PsC) from multiple studies included* Also included data from previous studies for meta-analysis. PsC (individuals affected with PsV for ≥ 10 yrs without developing any signs of PsA).	*Not stated, likely blood samples	Wanted to discern differences in genetic risk factors for PsA and cutaneous-only psoriasis (PsC) Used GWAS of 1,430 PsA case subjects and 1,417 unaffected control subjects (yielded high-quality genotypes at 791,217 SNPs for 1,430 PsA case subjects and 1,417 control subjects). After replication, also identified a PsV-associated SNP near *CDKAL1* (rs4712528, odds ratio = 1.16, $P = 8.4 \times 10^{-11}$).	Samples for the PsA GWAS were genotyped with the Illumina HumanOmni1-Quad BeadChip array (Illumina). Genotypes were obtained for 1,020,596 autosomal SNPs genotyped 23 markers to validate and replicate promising signals from the discovery analysis. *No statement of data availability or where sequencing was conducted.	James T Elder Ann Arbor Veterans Affairs Hospital, Ann Arbor, Michigan, US, jelder@umich.edu

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				All PsV case subjects were diagnosed by a dermatologist; a large subset were also evaluated for PsA by a rheumatologist.		
Rahmati, O'Rielly, Li, et al. (2020)[51]	Ethics approval was granted for this study by the Health Research Ethics Authority in NL (2016.195). All patients consented to be part of study	40 PsA patients recruited by the PsA clinic at MUN. All patients satisfied the Classification of Psoriatic Arthritis (CASPAR) criteria	Blood sample. Both DNA and total RNA extracted. Peripheral blood mononuclear cells separated from whole blood with Ficoll-Paque Plus density gradient (Cat # GE-1440-02, Millipore Sigma). Isolated $CD4^+$ cells and $CD14^+$ cells from PsA patients to determine the most discriminatory cell type. Consecutive patients initiating TNFi or IL-17Ai therapy were invited to participate in this study	Assessed PsA patients initiating either TNFi (golimumab, adalimumab, etanercept, infliximab, and certolizumab pegol) or interleukin-17A inhibitors (17Ai) 17Ai (secukinumab and ixekizumab) for active PsA at baseline and 3-months after initiating use. $CD4^+$ transcript profiling performed using transcriptomic data at initiation of therapy. Identified > 100 DEGs that differentiated IL-17Ai response from non-response and TNFi response from non-response. Regarding PsA, 161 genes were down-regulated, and 27 genes were upregulated in patients treated with etanercept as compared with untreated controls Network and pathway analysis: only overlap of DEGs (fold change ≥ 1.4 and raw *p* value ≤ 0.05) between response	NEBNext Ultra II Directional RNA Library Prep Kit for Illumina (Cat # NEB-E7760L, D-Mark Biosciences) and the NEBNext rRNA Depletion Kit (Cat # NEB_E6310X, D-Mark Biosciences) used to create sequencing libraries from total RNA. Double-stranded cDNA purified with Agencourt AMPure XP beads (Cat # A63881, Beckman Coulter). Followed by end repair and dA-tail addition before adaptor ligation. Adaptor-ligated DNA further purified then unique dual index barcodes attached (NEBNext Dual Indexed Primer Set 1, Cat # E7760D-Mark) Final libraries size-selected with bead purification and the quality and quantity assessed with Tape Station D1000 kit (Agilent) and the KAPA Library Quantification Kit (Cat# KK4828, Roche). Library concentrations	Dr. Proton Rahman Memorial University of NL, St. John's, NL prahman@mun.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				groups to each biologic at baseline and DEGs between responders to the 2 biologics were selected: generated 2 lists of statistically significant DEGs. To capture biologically meaningful DEGs, gene sets were obtained by overlaying differential expression values onto physical protein interaction network obtained from Integrated Interaction Database (IID, version 2018 to 11). Obtained *ROCK2* and *RAC1* protein expression data across 19 different tissues from the Human Protein Atlas Gene expression analysis: Raw FASTQ files with RNA reads aligned to the human hg19 reference genome. Gene expression for each sample quantified as fragments per kilobase of transcript per million mapped reads using Cufflinks (version 2.2.1) and normalized within-sample to transcripts per million reads values. Also collected DAPSA activity scores (includes a 68/66 joint count summed with a patient global score,	normalized and pooled for sequencing on Illumina NovaSeq 6000 for 2 × 150 bp reads and ~40 million reads/sample. No statement of data availability. Does not state where DNA analysis was completed	

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				patient pain score, and CRP level) at baseline and 3 months into treatment.		
Psoriasis						
Nair, Duffin, Helms, et al. (2009)[52] *A letter	Protocols reviewed and approved by local institutional REBs. All participants subjects gave consent.	Pulled samples from multiple sources (identified psoriasis susceptibility loci, by genotyping 1,409 psoriasis cases and 1,436 controls of European ancestry. Followed up 21 SNPs in 5,048 psoriasis cases (368 cases, 358 controls from NL) *NL involvement stated in supple-mental materials	Not specified.	Identified psoriasis susceptibility loci in initial samples (1,359 psoriasis cases and 1,400 controls to impute). WGS genotyped for 438,670 autosomal SNPs. From these, 21 SNPs (representing 18 loci) with strongest evidence for association genotyped in an additional 5,048 cases and 5,051 controls (including the NL cases and controls)	Follow-up samples genotyped using Applied Biosystems Taqman assays, Sequenom single base extension assays, or allele-specific kinetic PCR. dbGAP: genotype and phenotype data deposited with the accession code phs000019.v1.p1. NCBI GEO: microarray data were deposited under accession number GSE13355.	James T Elder Ann Arbor Veterans Affairs Hospital, Ann Arbor, Michigan, US, jelder@umich.edu Gerald G Krueger Department of Dermatology, University of Utah, Salt Lake City, Utah, US gerald.krueger@hsc.utah.edu Anne M Bowcock Division of Human Genetics, Department of Genetics, Washington University School of Medicine, St. Louis, Missouri, US, bowcock@wustl.edu Gonçalo R Abecasis Department of Biostatistics, Center for Statistical Genetics, University of Michigan, Ann Arbor, Michigan, US goncalo@umich.edu

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
Das, Stuart, Ding, et al. (2015)[53]	Not specified.	5067 subjects of European ancestry from 5 collaborating centers, including 783 (470 psoriasis cases, 313 controls) from NL *58 samples pulled from Tsoi et al (a previous study)	Not specified.	All subjects genotyped to include all genes of interest in their entirety and all SNPs in 8 psoriasis susceptibility regions (MHC, IL12B, IL23A, IL23R, TNIP1, TNFAIP3, IL13 and RNF114)	Illumina technology used. Information uploaded to NCBI dbGaP database at (dbGaP accession number: phs000019.v1.p1).	Dr GR Abecasis Department of Biostatistics, Center for Statistical Genetics, University of Michigan, Ann Arbor, MI goncalo@umich.edu
Spondyloarthritis						
O'Rielly, Uddin, Codner, et al. (2016)[54]	The REBs from all participating hospitals approved study (HREA #1999.172). Patient consent obtained.	Single multi-generational family from NL with Axial spondyloarthritis conditions (number of participants unclear). Replication cohorts provided by The Spondyloarthritis Consortium of Canada: provided clinical and radiographic data, as well as DNA samples for the replication cohorts.	Whole blood samples	*HLA-B*27* genotyping and exome sequencing performed by Life Technologies (US)	Samples sequenced targeting whole genome exons using Illumina HiSeq 2000. Mapping of reads aligned using Burrows Wheeler Alignment V.0.7.10, and the genome analysis toolkit V.1.1.28 used to call variants against the reference genome. *No statement about data availability	Dr. Proton Rahman Memorial University of NL, St. John's, NL prahman@mun.ca

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Lehr, Rahman, and O'Rielly (2017)[55] *Letter to the editor	Institutional ethics was approved from the Health REB at MUN (Health Research Ethics Authority reference #15.051). Patient consent was obtained.	1,000 consecutive patient samples (572 men and 428 women) received by the clinical genetics laboratory at Eastern Health to determine the *HLA-B*27* genotyping status. There were 172 *HLA-B*27*–positive and 828 *HLA-B*27*–negative patients.	Whole blood samples	Targeted analysis of the HLA-B locus by Genotyping of rs4349859 (NM_001289154.1:c.2921G > A) and rs116488202 (NC_000006.12:-g.31377139C > T) as a test of who has axial spondylo-arthritis	Targeted analysis of the HLA-B locus was performed using LABType SSO HLA-B locus kit on a Luminex 100/200 platform (One Lambda, ThermoFisher Scientific). Genotypes of rs4349859 (NM_001289154.1: c.-2921G > A) and rs116488202 (NC_000006.12:-g.31377139C > T) were assigned using the end point genotyping analysis software (Applied Biosystems). *Conducted at the Molecular Genetics Laboratory, Eastern Health,	Dr. Darren O'Rielly Faculty of Medicine, Memorial University of NL, Craig L. Dobbin Genetics Research Centre, St. John's, NL darren.orielly@med.mun.ca
Osteoarthritis						
Spector, Reneland, Mah, et al. (2006)[56]	Approved by REBs at each clinical centre. All participants provided consent for the study.	London, UK: 343 cases with OA, 303 controls without OA NL Canada: 378 cases with OA (211 with knee OA, 99 with hand OA, and 68 with hip OA), 264 controls without OA *All European ancestry	Not specified. Health history and rheumatologist examination	Genotyping of all participants: A set of 25,494 SNP markers located within 10 kb of 13,735 Entrez Gene annotated genes.	SNP annotation based on the NCBI dbSNP database, reference SNP, build 118. Genomic annotation was based on NCBI genome build 34. Gene annotation based on Entrez Gene genes for which NCBI was providing positions on the Map Viewer FTP site. *No statement of data availability, but NF and UK data analyzed together. Site of processing and data library not specified.	Dr. Tim D. Spector St. Thomas' Hospital, London, UK Twin Research and Genetic Epidemiology Unit, St. Thomas' Hospital, London UK tim.spector@kcl.ac.uk

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
Aref-Eshghi, Zhang, Liu et al. (2015)[57] And Gill, Liu, Sun, et al. (2024)[58] are both part of the NFOAS is ongoing, initiated in 2011, Aref-Eshghi (above), Gill (Below)	Approved by the HREA of NL (NFOAS-# 2011.311 and CODING- #10.33) Consent obtained from participants.	5 patients with hip OA, 6 with knee OA, and 7 OA-free hip controls. OA Patients having joint replacements due to primary OA recruited between 2011 and 2013, and OA-free controls (joint replacement for alternate reason) recruited in same NL hospital.	Cartilage samples, pathology reports, Demographic information (obtained by a self-administered questionnaire) height and weight from medical records.	Looked at the methylation status of 485,000 methylation sites throughout genome, covering 99% of RefSeq genes at an average of 17 CpG sites per gene across the 5-UTR, gene promoter regions, first exon, gene body, and 3-UTR.	Genome wide DNA methylation analysis conducted using the Illumina Infinium HumanMethylation450 BeadChip. *The* genome-wide DNA methylation data *available* with accession number GSE73626.	Guangju Zhai Memorial University of NL, St. John's, NL gzhai@mun.ca
Aref-Eshghi, Zhang, Liu et al. (2015)[57] and Gill, Liu, Sun, et al. (2024)[58] are both part of the NFOAS is ongoing, initiated in 2011, Aref-Eshghi (above), Gill (Below)	Approved by the Health Research Ethics Authority of NL (NFOAS-# 2011.311 and CODING- #10.33) Consent obtained from participants.	559 OA patients (172 hip, 387 knee), 118 healthy controls (from CODING study) study all from NL. *Replication cohort from UK	Whole blood samples	WGS done by the Illumina Human Omni2.5 to 8 microarray platform (genotypes 2.5 million SNPs) or the Human Global Diversity Array (genotypes 1.9 million SNPs) at The Centre for Applied Genomics in Toronto .	Data imputed against 1000 Genome Project phase 3 data by the Sanger Imputation Server which generated data on a total of 81,706,022 SNPs across the entire genome. Datasets available from the corresponding author on reasonable request	Guangju Zhai Memorial University of NL, St. John's, NL gzhai@mun.ca
Zhai, Aref-Eshghi, Rahman, et al. (2014)[59]	The study was approved by Health Research Ethics Authority (HREA) of NL and	A case-control study (part of the NFOAS). 126 patients undergoing total	Blood samples obtained from all study participants. DNA was extracted by using the standard	105 SNPs were genotyped (selected based on SNPs reported to be associated with OA in GWAS and some candidate studies).	Genotyped using either the Sequenom iPLEX Gold method (OA cases) using 384-well plate chip and loaded onto the	Guangju Zhai Memorial University of NL, St. John's, NL gzhai@mun.ca

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	written consent was obtained from all the participants.	hip/knee joint replacements between 2011 and 2013 in local hospital due to severe OA (cases). 348 healthy controls who did not have any evidence of either knee or hip OA were selected from previous genetic association study for type 2 diabetes and obesity. Controls had no musculo-skeletal pain, a prior diagnosis of osteoarthritis, were not taking acetaminophen or NSAIDs and had a normal physical examin-ation, as it relates to the skeletal system	protocol. Controls completed questionnaire regarding ongoing symptoms, previous diagnosis and medication history. Each patient examined by a rheumatologist. Height and weight measurements were obtained from patient's hospital medical record. Body mass index (BMI) was calculated		Mass Spectrometer for reading, or using Illumina HumanHap550-Duo BeadChip at Centrillion Biosciences at Palo Alto, California (completed previously for controls). Cross-validation of genotyping quality was carried out on 31 controls that were genotyped by both Sequenom iPLEX Gold method and IlluminaHumanHap550-Duo BeadChip. Data availability not reported. Quebec Genome and Innovation Centre and Centrillion Biosciences, CA, conducted gentyping.	
Aref-Eshghi, Liu, Harper, et al. (2015)[60]	*See below The study protocol was approved by the Health Research Ethics Authority of	Part of the ongoing NFOAS that was initiated in 2011, A total of 32 OA cases (25 hip OA and 7 knee OA) and 21	Human cartilage samples were collected from patients undergoing total hip/ knee joint replacement surgery due to primary OA or hip fractures as	Compared the expression levels of TGFB1 and BMP2 as ligands, SMAD3 as an intracellular mediator, and MMP13 as a targeted gene between human osteoarthritic and healthy	*See below, same processes used. *Sequencing performed in-house.	Guangju Zhai Memorial University of NL, St. John's, NL gzhai@mun.ca

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	NL and written consent was obtained from all participants.	healthy controls were included. *See below	controls. *See below			
Aref-Eshghi, Liu, Razavi-Lopez, et al. (2016)[61]	Refer to above	Part of the NFOAS initiated in 2011. 58 patients with OA undergoing total hip/knee replacements and 55 controls with hip fractures. Recruited between November 2011 and December 2013 in St. Clare's Mercy Hospital and Health Science Centre General Hospital in St. John's, NL.	DNA/RNA: Four pieces (□ 200 mg each) of cartilage tissues retained from either tibial plateaus or femoral heads collected from cases and controls. Also, pathology reports, demographic information (self-administered questionnaire), and Anthropometric data including BMI from hospital admission and medical records and age was at the time of surgery.	Investigated *SMAD3* gene expression and its promoter DNA methylation in cartilage tissues. PCR was performed to measure gene expression. One microlitre of the converted cDNA was subject to quality control by PCR amplification of the *SMAD3* and *GAPDH* genes followed by agarose gel electrophoresis.	Sequenom EpiTyper was used to assay DNA methylation. Complementary DNA (cDNA) synthesis from the extracted RNA was done using the Maxima H Minus First Strand cDNA Synthesis Kit (K1682; Thermo Scientific, Vilnius, Lithuania). Quantification of *SMAD3* was performed using the ABI-7900HT Fast Real-Time PCR system on a 96-well plate. Bisulfate conversion of DNA was conducted using the EpiTect Bisulfite Kit *SMAD3* promoter DNA methylation was quantified using the Sequenom EpiTyper platform The mass signals generated were translated into quantitative DNA methylation levels (beta value range 0 to 1) by MassArray EpiTyper Analyzer software.	Guangju Zhai Memorial University of NL, St. John's, NL gzhai@mun.ca

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					No statement of data availability.	
Werdyani, Liu, Xie, et al. (2021)[62] *Only Abstract Available	Refer to above	Primary OA patients from the NFOAS that comprised total knee or hip replacement and recruited before 2016. 83 primary OA patients (44 responders, 39 non-responders) included in analysis.	Whole blood DNA samples Study participants completed their pre-operation and 3.99 ± 1.38 years post-surgery outcome assessment using the Western Ontario and McMaster Universities Osteoarthritis Index (WOMAC).	WGS completed. Pre-association quality control filtering conducted for raw genotyping data using PLINK 1.7 program, and genotype imputation performed using IMPUTE2 algorithm with multiple population reference data (1000 Genome Project). The imputed data with □3.1 million variants used to test the association with non-responders to TJR using the additive genetic model.	DNA samples were genotyped using the genome-wide Illumina HumanOmni2.58 genotyping microarray containing 2.4 million SNPs.	Werdyani, Liu, Xie, et al (2020) *Only Abstract Available
Werdyani, Liu, Furey, et al (2022)[63] *Only Abstract Available	Refer to above	Part of the NFOAS total hip or knee replacement patients due to primary OA, recruited before 2017 in St John's, NL. Self-reported OA-free controls derived from same source population, originally recruited to the CODING study. 566 OA patients and 120 unaffected	DNA samples were extracted from whole blood	WGS conducted. 2 genotype datasets were merged and the common SNPs between both genotyping microarrays were used in the analysis. Pre-association quality control filtering and population structure analyses performed for study participants based on genotyping data. Individuals excluded from cohort if they: a) had a heterozygosity rate > 3SD, b) had discordant sex information; c) were non-Caucasian; and d) had call rate < 95%. Also, variants quality control	genotyped using the Illumina HumanOmni2.58 and Infinium Global Diversity 8 v1.0 genotyping microarrays. No statement of data availability in the abstract.	Werdyani, Liu, Furey, et al (2022) *Only Abstract Available

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		passed the QC check and were included in the analysis.		filtering was performed to exclude SNPs that had call rate < 0.0001).		
Ankylosing spondylitis						
Maksymowych, Rahman, Reeve, et al. (2006)[64]	Ethical approval for the study was obtained from the MUN Bioethics Research Committee. The ethnic composition of this cohort is relatively restricted to individuals of Irish/southwest English descent.	3 independent case-control cohorts AB, NL (112 unrelated white AS patients attending the outpatient rheumatology department of MUN, 10 had IBD, 20 had psoriasis, and 4 had reactive arthritis. Controls were 150 healthy unrelated white blood donors from St. John's, NL. Cohort is relatively restricted to individuals of Irish/ southwest English descent), and Toronto that have been pheno-typically well-characterized in a systematic	DNA was extracted from peripheral blood and the final concentration adjusted to 5 ng/μL.	Samples were genotyped using a panel of 38 single-nucleotide polymorphism (SNP) markers within the *IL1* gene cluster. Data from 20 informative and nonredundant SNP markers were analyzed using several association test strategies. First, we used the program WHAP to identify single-marker associations. Second, we used WHAP to analyze "sliding windows" of 3 contiguous markers along the entire extent of the *IL1* gene cluster to identify haplotypic associations. Third, we used the linkage disequilibrium mapping program DMLE to estimate the posterior probability distribution of a disease locus. First, 2.5 ng of genomic DNA was amplified under standard conditions using forward and reverse primer pairs.	Genotyped by time-of-flight mass spectrometry using the Sequenom platform (Sequenom, San Diego, CA) The primer extension products were then cleaned and spotted onto a SpectroChip. Measures of pairwise LD were determined using Haploview	Dr. W. P. Maksymowych University of Alberta Department of Medicine, Edmonton, AB walter.maksymowych@ualberta.ca *has full access to all the data

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		manner by investigators collaborating in the Spondylo-arthritis Research Consortium of Canada disease network.				
Snelgrove, Lim, Greenwood, et al. (2007)[65] *Only had access to article draft	Approved by MUN REB. Consent obtained from all patients.	101 AS patients and 100 ethnically matched controls (all Caucasian). Controls from volunteers from local campaign seeking controls for genetic studies.	Whole-blood samples Data collected included measures of disease activity and function Index	SNP testing of 2 functional variants in the TLR4 gene: Asp299Gly (A/G polymorphism) and Thr399Ile (C/T polymorphism)	DNA extracted using the Promega Wizard Genomic DNA purification kit. polymorphisms typed using Sequenom chipbased MALDI-TOF mass spectrometry platform using MassArray software (Sequenom). Products cleaned and spotted onto a SpectroChip. Chip scanned with mass spectrometry workstation (Bruker). Resulting spectra analyzed with the Sequenom SpectroTYPER-RT software. *No statement about data availability	Dr. Proton Rahman Memorial University of NL, St. John's, NL prahman@mun.ca
Rahman, Inman, Gladman, et al. (2008)[66]	Ethics approval for this study was obtained at each of the participating universities.	3 cohorts (NL, AB, ON) of AS patients from established rheumatic disease centers in Canada. (majority of Caucasians	Whole-blood samples Data collected included measures of disease activity and function Index	SNPs were rs1004819 (C > T), rs7517847 (t > G), rs10489629 (G > A), rs2201841 (t > C), rs11465804 (t > G), rs11209026 (G > A), rs1343151 (t > C), rs10889677 (A > C), rs11209032 (G > A), and	The MassArray system (Sequenom, San Diego, CA) was used to genotype each study participant using a single multiplex reaction that was designed using AssayDesigner 3.0. T	Dr. Proton Rahman Memorial University of NL, St. John's, NL prahman@mun.ca *had full access to all of the data in the study

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		of northern European descent), (NL: 121 AS probands and 219 controls)		rs1495965 (G > A). (of these were incorporated in the haplotype analysis. Allele and haplotype associations were calculated using the WHAP software package. P values for haplotype associations were calculated using a permutation test.	*No statement about data availability	
Maksymowych, Inman, Gladman, et al. (2009)[67] *Earlier study Rahman (2008) drew from same cohort	Approval obtained from each participating university. Patients provided consent.	992 AS cases and 1,437 controls from 3 centers (472 cases and 451 controls from AB, 138 (22 had IBD, 28 had psoriasis in addition to AS) cases and 392 controls from *NL* founder population, and 382 cases and 594 controls from Toronto).	Whole blood sample	All genotyped for 13 coding-region SNPs, 6 in the *ERAP1* gene, 4 in the *TAP2* gene, 1 in the *TAP1* gene, 1 in the *LMP2* gene, and 1 in the *LMP7* gene	The MassARRAY system (Sequenom, San Diego, CA) used to genotype study participants. Genotypes determined using MassARRAY Typer software, version 4.0. *No statement regarding data availability	Dr. W. P. Maksymowych has full access to all of the data University of Alberta Department of Medicine, Edmonton, AB walter.maksymowych@ualberta.ca
Uddin, Maksymowych, Inman, et al. (2013)[68]	Informed consent for participation in the study was obtained from participants or, where participants are children, a parent or guardian. approved by the MUN Human	NL: 298 cases 299 controls AB: 289 cases 285 controls All cases and controls were of North European	Each individual was assessed clinically including imaging studies.	Custom genome-wide tiling microarray consisted of 2 × 1 million probes covering the genome with a mean spacing of 280 bp. Approximately 1700 CNVs were detected in each individual sample. Segregated gene-centric CNV analysis (i.e., a CNV	Samples were analyzed using the ViiA™ 7 Real-Time PCR System (Life Technologies) and analyzed using CopyCaller Software (Life Technologies, PN 4412907). *No statement of Data Availability	Dr. Proton Rahman Memorial University of NL, St. John's, NL prahman@mun.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
	Investigation Committee.	ancestry. *Cases satisfied the modified New York criteria		that consists of or overlaps with a gene) revealed that 56 CNVs are enriched in affected family members (at least three) and absent in the unaffected family members. Affected family members carried 2 copies of the UGT2B17 gene, whereas unaffected members carried a single copy. The breakpoint encompassing the UGT2B17 gene region was covered with probes with 280 bp spacing, providing high resolution to detect genomic aberrations. *Analysis conducted in Quebec and Toronto.		
Hearing Loss						
Young, Ives, Lynch (2001)[69]	Obtained informed consent from each individual including relatives	6-gen family from NL with mutations in the Wolfram Syndrome Gene (WFS1). 62 family members identified with low frequency hearing loss. Additionally unaffected family members were included.	peripheral blood (83 people) DNA was isolated using a salting out extraction method. family and medical histories (from 314 informative relatives) and pure tone audiometric evaluations (performed on 81 relatives.)	Linkage analysis: 47 family members included in genome-wide scan using 386 polymorphic markers. Additional polymorphic markers were genotyped on family members in the vicinity of D4S431 to define the *DFNA38* critical region. Sequencing: Coding sequence, regulatory regions and flanking intronic sequence of candidate	Genome wide scan completed using the ABI Prism Linkage Mapping Set-MD10. PCR products were size fractionated on an ABI 377 sequencer machine and alleles were scored with Genotyper software (v. 2.0 Applied Biosystems). 2-point linkage analyses were performed using the MLINK subroutine of FASTLINK (V4.0P) Genome-wide.	Dr. Terry-Lynn Young Memorial University of NL St. Johns, NL tlyoung@mun.ca

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		150 unrelated individuals also included as controls for genetic analyses.		genes were sequenced from genomic DNA. 11 variant sites in *WFS1* were detected in all deaf individuals. 5 variants were known polymorphisms; 6 novel, of which 3 were amino acid substitutions: Gly107Arg, Ala716Thr and Val871Met. The A716T mutation abolishes an *Hae*II restriction enzyme site, all family members who were not included in the genome scan were tested for the mutation by restriction enzyme analysis using *WFS1* specific primers. Restriction digest analysis was also used to test for mitochondrial mutations associated with hearing impairment: A1555G, A3243G, A7445G, 7472 ins C, T7510C, T7511C, T7512C, and 961_TC (n) ins. 150 controls were also genotyped for the A716T mutation.	No statement of data availability. Unclear where analysis was performed.	
Ahmed, Cindy Li, Powell, et al. (2004)[70]	Approval from the NL Labrador Medical Genetics Program, Health Sciences Centre REB. Participants provided consent.	32 people from a large six-generation pedigree from Labrador Additionally, families in	Whole blood samples Family pedigrees, pure tone air and bone conduction audiometry and Pneumatic otoscopy results	WES to identify mutant alleles of *TMPRSS3* locus. PCR amplified products were sub-cloned into pGEM-T Easy vector (Promega) and sequenced.	Dr. Zubair M Ahmed performed DNA sequencing. No additional information on data availability.	Dr. Edward R Wilcox Laboratory of Molecular Genetics, National Institute on Deafness and Other Communication Disorders, National

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		Pakistan were included.				Institutes of Health, Rockville, MD, US US wilcoxe@nidcd.nih.gov
Abdelfatah, McComiskey, Doucette, et al. (2013)[71]	Informed consent, family history and permission to access medical records and audiograms were obtained as per approved protocol #01.186 (Human REB, St John's, NL).	A 6 generation (n = 28 affected) family of English extraction from NL with sensory neural hearing loss. part of a large study of hereditary hearing loss in province.	DNA extracted from peripheral leukocytes of all available family members. Phenotypic data reviewed by audiologists and classified according to the GENDEAF study group recommend-ations	1. Genomic DNA from a proband (Figure 1; IV-11) was screened by Sanger sequencing for population specific mutations in WFS1, TMPRSS3 and PCHD15, a mutations in GJB2 and GJB6. 2. Targeted sequencing of exons 5 to 7 in KCNQ4, exons 4, 5, 12 in COCH and exons 5, 9 to 14, 17, 18, 20 in TECTA based on their association with bilateral symmetric, sloping, SNHL 3. To check for other potential pathogenic mutations in the DFNA2 locus, we fully sequenced the remaining exons of KCNQ4 and the entirety of GJB3 (DFNA2B) in selected family members 4. Variants of interest subjected to cascade sequencing (affected n = 23; unaffected n = 19) and allele	Determined using ethnically matched population controls from NL. Sanger sequencing was carried out on an ABI PRISM 3130XL DNA Analyzer (Applied Biosystems, Foster City, CA, US) Markers and alleles scored with GeneMapper software (v.4.0; Applied Biosystems) used for step 6. Linkage calculations were performed using the Linkage Program MLINK, version 5.1.9	Dr. Terry-Lynn Young Memorial University of NL St. Johns, NL tlyoung@mun.ca

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				frequencies determined using ethnically matched population controls from NL. 5. To identify other candidate genes to screen, audiograms from selected subjects were submitted to Audiogene for computerized comparison with known average audiograms of 16 AD loci. 6. All available family members were genotyped for DFNA2A/B-linked. Haplotypes were manually constructed. 7. Two-point linkage analysis was carried out using MLINK on 4 microsatellite markers (D1S195, D1S472, D1S2706 and D1S2130		
Pater, Benteau, Griffin, et al. (2017)[72]	Approval from Human REB, St. John's, NL). Protocol #01.186 Consent for family history and permission to access medical records and audiograms	4 members of a NL family with HL, relatives, 169 HL cases and 175 from NL founder population.	Whole blood samples Family histories Retrospective and prospective audiograms Medical data from medical charts Family pedigrees	4 cases vetted for population-specific deafness alleles. More loci selected based on audiograms. WES libraries created using the Ion Torrent AmpliSeq RDY Exome Kit. Based on these results, samples from relatives and 175 controls and 169 HL cases	Bidirectional Sanger sequencing Used Mutation Surveyor Software to select quality reads and analyze DNA sequences. Purified libraries obtained from WES of 4 probands quantified using the Ion Library Quantification Kit	Dr. Terry-Lynn Young Memorial University of NL St. Johns, NL tlyoung@mun.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
	obtained from all participants			had Cascade sequencing and haplotype analysis on potential mutations in Microsatellites flanking candidate genes.	(Life Technologies, Cat. #4468802) and loaded onto a PI v3 chip and sequenced with Ion Torrent Proton Sequencer. SNVs and insertion/deletions were called (GATK, v3.5) and annotated using SnpEff (v4.1;). Cascade and haploid screening using GeneMapper software v4.0.	
Pater, Penney, O'Rielly, et al. (2022)[73]	Approval from Human REB, St. John's, NL). Protocol #01.186 Consent for family history. Participant consent obtained.	5 affected and 2 unaffected family members of a 6 generation NL family with a variable form of bilateral sensori-neural HL, 202 sensorineural HL cases and 326 ethnically matched controls all from NL.	Peripheral blood sample	1 family member sequenced for all pathogenic variants causing HL in NL population, the 34 select autosomal dominant genes matching proband's audiometric data. Next 4 affected and 2 unaffected family members had genome-wide SNP genotyping. Candidate variants amplified using standard touchdown PCR protocol and sequenced in family members and compared to sensorineural HL cases and controls using Gene Mapper software.	Genomic DNA libraries (using the Lucigen Shotgun NxSeq AmpFREE Low DNA Library Kit) were prepared on 4 affected and 2 unaffected members of the NL family. Prepared libraries were loaded on Illumina. *The datasets generated during and/or analyzed during study available from the corresponding author on reasonable request.*	Dr. Terry-Lynn Young Memorial University of NL St. Johns, NL tlyoung@mun.ca
Abdelfatah, Mostafa, French, et al. (2022)[74]	Approved by MUN REB (#1.186), Western University (#103,679) and the Danish Research Ethical Committee (KF 01-234/02 and	5 people with otosclerosis and 17 relatives in 1 NL family. As well as 2 controls with no HL for first pass screening.	Leukocytes isolated from peripheral blood samples	5 affected family members and 2 controls genotyped for markers spanning each *OTSC* disease interval and bracketing 3 OTSC susceptibility genes using WES. Unaffected	WES was outsourced to the Genome Centre (McGill University, QC,) including library preparation (TrueSeq Prep Kit). Positional candidate genes	Dr. Terry-Lynn Young Memorial University of NL St. Johns, NL tlyoung@mun.ca

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	KF 01-108/03). Participants consented regarding publishing their data	Unrelated cases with otosclerosis recruited from NL (*n* = 82), Finland (*n* = 35) and the Faroe Islands (*n* = 20) and used as validation cohorts.		family members and cases genotyped with 9 extra markers (*D16S518, D16S3049, D16S3098, D16S422, D16S2625, D16S520, D16S413, D16S3023, D16S3026*) mapping qter of the *OTSC4* disease interval.	selected based on function and/or gene expression. Samples Sanger sequenced on an ABI 3130xl/3730. Samples run on Illumina Hiseq 2000. Variants of interest validated by cascade sequencing in NL family. Heterozygous variants co-segregating with otosclerosis in an autosomal dominant pattern tested in the unrelated otosclerosis cases. *The datasets generated during and/or analysed during study available from the corresponding author on reasonable request.*	
Singh, Penney, Griffin, et al. (2023)[75]	Approval from Human REB. St. Johns (protocol #01.186), Consent from participants to access medical/ audiometric records, collect biological samples.	22 people from a NL family (first Canadian family known to have profound HL and be *POU4F3* c.37del carriers) Plus 187 HL cases and 71 ethnically matched controls from NL	Peripheral blood or saliva samples of 22 family members (8 for WGS, 14 for WES) Additionally, audiogram, and 7 generation family pedigree Data Blood samples from HL cases and controls.	A WGS and WES done with SNP technology done with Human610-Quad chip on family members. Sanger sequencing used to screen cases and controls variants that co-segregated with significant sensory neural HL identified with WGS and WES of affected family.	Libraries multiplexed and sequenced using a S2 Reagent Kit (200 cycles) NovaSeq 6000 (Illumina Inc.) Sequence data analyzed by Mutation Surveyor and Alamut Visual on the combined linkage analysis with WES. Exome library preparation made with the Nextera DNA kit (Illumina Inc.) *Datasets generated and analyzed are available from the corresponding author on reasonable request. Have been uploaded*	Dr. Terry-Lynn Young Memorial University of NL St. Johns, NL tlyoung@mun.ca

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					to ClinVar (accession SCV002556358).	
Stargardt disease						
Green, O'Rielly, Pater, et al. (2020)[76]	Approved by Human REB of Eastern Health in St. John's NL. (protocol # 02.116)	Population-based clinical recruitment of patients with STGD1/STGD1 like disease and their extended families (29 families [DNA samples from 82 people] since 1978)	Whole blood samples Family interviews and public archival records providing historical religious affiliations and community of origin	WGS of the *ABCA4* gene	Location of sequencing and data library not specified. Primer sequences are available upon request. *Data from this study have been submitted to the public Leiden Open Variation Database.*	Dr. Terry-Lynn Young Memorial University of NL St. Johns, NL tlyoung@mun.ca
Bardet–Biedl and Obesity						
Fan, Rahman, Peddle, et al. (2004)[77]	Not specified. Recruitment through advertise-ments in local newspaper and family doctors.	200 people with BMI ≥ 30 and 200 controls with BMI ≤ 30. All European ancestry from west coast of NL. Plus, previously collected data from 6 BBS1 families from same region (8 affected, 37 carriers, 18 noncarriers)	Whole blood samples	DNA extracted using simple saltingout method. BBS1 exons screened for the M390R mutation by direct sequence analysis.	Location of sequencing and data library not specified. Sequencing was carried out with ABI 377 automated sequencer. Sequences analyzed using Sequencer 3.1.1. *No statements about data availability.	Dr. William S. Davidson, Department of Molecular Biology and Biochemistry, Simon Fraser University, 8888 University Drive, Burnaby, British Columbia william _davidson@sfu.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
Linkage disequilibrium in populations						
Service, DeYoung, Karayiorgou, et al. (2006)[78]	Approved by University of California, Los Angeles Institutional REB. Consent obtained from participants.	Samples from 12 populations including NL. 200 independent persons geno-typed from each population.	Not specified.	SNP technology used to analyze magnitude and distribution of linkage disequilibrium across the entire length of chromosome 22.	Genotyping reactions performed using Illumina BeadLab kit reagents and protocols25. Performed genotyping, using 250 ng of normalized genomic DNA from each sample as template for the Illumina GoldenGate genotype assay, on the Illumina BeadLab 1000 platform. Data collection accomplished using the BeadScan (Illumina) software package. No statement regarding data availability.	Dr. Nelson Freimer (nfreimer@mednet.ucla .edu).
Ovarian Cancer						
Dawson, Smith, Werdyani, et al. (2010)[80]	Approval from Human Research Ethics Authority of NL (protocols #2016.1914 and #2018.0391) Consent and permission to access medical records obtained from all participants.	70 cases recruited from Provincial Medical Genetics Program and NL HBOC Study and extended family members from 5 cases. 880 NL ethnically matched controls screened for the *RAD51C* c.571 + 4A > G variant.	Whole blood samples from all 70 case participants and 880 controls. Family history and pedigree evaluations and clinical risk for 5 probands	Performed panel of moderate-high-risk CA susceptibility genes for 5 probands at external facilities using the OncoGeneDx Custom Panel (GeneDX), the Common Hereditary CAs Panel (Invitae) or the Ovarian CA Focus Panel (Fulgent Genetics). Performed WES and WGS microarray analysis to detect additional variants. Based on this, remaining 65 cases, family members, and 880 controls	WES and WGS performed at Centre for Translational Genomics (St. John's, NL) WES Libraries were quantified by quantitative PCR and loaded onto the Ion Proton platform for high-throughput sequencing. DNA sequences analyzed using Mutation Surveyor Software v5.0.0 (SoftGenetics LLC). Data analyzed using TaqMan Genotyper Software v1.4 (Applied Biosystems). GeneMapper Software	Dr. Darren O'Rielly Faculty of Medicine, Memorial University of NL, Craig L. Dobbin Genetics Research Centre, St. John's, NL darren.orielly@med.mun .ca

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				screened for *RAD51C* c.571 + 4A > G variant. Haplotype analysis on markers flanking the recurrent *RAD51C* c.571 + 4A > G conducted on family members	used to conduct haplotype analysis on family members.	
Food addiction						
Pedram, Zhai, Gulliver et al. (2017)[81]	Approved by the Health Research Ethics Authority (HREA), MUN project identification code: #10.33, (latest date of approval: February 10, 2016). All partici-pants provided consent.	Stage 1: 24 subjects 8 obese with high FA, 8 obese with low/ zero FA clinical symptom score (FAO, NFO), and 8 healthy controls with normal BMI and low/zero FA symptom score (Ctrl). Stage 2: 752 subjects (all taken from the CODING study)	DNA sample: 5 ml of whole blood	Stage 1: Genome-wide screening study using a whole-exome sequencing method (sent to BGI Americas). Top 100 SNPs identified and categorized into 5 subgroups based on gene functions: addiction (Ad), psychological disorders, energy metabolism and obesity, and cancer, unknown function or with other diseases. Stage 2: Top 19 SNPs in the Addiction subgroup genotyped in all subjects (Génome Québec Innovation Centre).	Exome sequencing done using latest Illumina HiSeq 4000 system with 100 bp and 150 bp paired-end sequencing Sequencing of top 19 SNPs done using Sequenom iPLEX Gold genotyping technology *No statement of data availability.	E-mail addresses: Dr. Pardis Pedram p.pedram@mun.ca Guangju Zhai guangju.zhai@med.mun.ca Dr. Hongwei Zhang hzhang@mun.ca Dr. Guang Sun gsun@mun.ca
Bardet–Biedl syndrome (BBS)						
Young, Woods, Parfrey, et al. (1998)[82]	Appropriate informed consent was obtained and medical records were reviewed.	17 NL families	WBCs from whole blood medical records A physical examination was performed and measurements were	includedD3S1776, D3S1251, D3S1752, D3S1271, D3S1753,D11S1298, D11S480, FGF3, D11S1369, D15S216,D15S131, D15S204, D15S114, D15S211, D16S419,D16S390,	*Here only Department of Biochemistry, Memorial University of NL, St. John's, NL, was mentioned	Dr. Terry-Lynn Young, Faculty of Medicine, Memorial University of NL, St. John's, NL tlyoung@mun.ca

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			made of height and weight.	D16S408, D16S526, and D16S265. Additional markers typed for haplotype analysis on chromo-some 3		
Woods, Young, Parfrey, et al. (1999)[83]	Protocol for clinical investigation approved by the Human Investi-gations Committee of the Faculty of Medicine, MUN, and the Medical Advisory Council of the St. John's General Hospital. *Patient consents not specified.	Genotyped members of 17 NL families (20 males and 14 females with BBS and from 111 unaffected family members) identified through the registry of the Canadian National Institute of the Blind and the Nephrology Unit at the Health Science Center, St. John's, NL.	DNA was prepared from whole blood using a simple salting-out method.	≥ 4 polymorphic microsatellite markers spanning the critical regions of the 4 known the 4 known loci (BBS1 (11q), BBS2 (16q), BBS3 (3p), and BBS4 (15q), were examined in all families. PCR using 100 to 200 ng of template DNA was conducted with primers purchased from Research Genetics, Inc. Haplotypes were constructed for each of the 4 known loci and linkage analysis was tested with two-point linkage Two-point linkage analysis was performed using the MLINK subroutine of FASTLINK to corroborate haplotype analysis.	*Information on likability with administrative data and availability of data not provided.	Dr. Micheal O. Woods Discipline of Genetics, Faculty of Medicine, Health Sciences Centre, St. John's, NL mwoods@mun.ca
Young, Woods, Parfrey, et al. (1999)[84]	Stated that Informed consent had been obtained previously.	17 BBS families of English ancestry were haplotyped at the BBS1, BBS2, BBS3, and BBS4 loci.	DNA was extracted from the lymphocytes of venous blood by a simple salting-out procedure	Markers D11S1298, D11S956, D11S480, D11S4205, D11S1883, D11S4945, PYGM, D11S4946, D11S4940, D11S4938, D11S449, D11S4941, D11S913, and FGF3 were typed in key family members. 1 family was excluded	Accession number and URLs for data in this article are as follows: Online Mendelian Inheritance in Man (OMIM), (for BBS [MIM 209900]) Sixth International SCW 11 Workshop (Nice, France, 1998). Institute of Molecular Biology and Biochemistry,	Dr. Terry-Lynn Young, Faculty of Medicine, Memorial University of NL, St. John's, NL tlyoung@mun.ca

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				from the study, because several samples failed to amplify. Haplotypes were constructed, for each family, to give the minimum number of recombinations. Disease haplotypes (DH) were identified from alleles that were transmitted from both unaffected parents to affected offspring. LOD scores were calculated by the MLINK subroutine program of FASTLINK (V3.0P). marker-allele frequencies in disease and normal (non-transmitted) chromosomes of the 10 obligate BBS carriers (parents) were compared. Normal alleles from each parent were used as population controls, to avoid the possibility of inadvertently including BBS1 disease alleles from random carriers in the population. DHs were compared between families in the search for (i) common BBS1 haplotypes that would indicate that the parents of ≥ 2 families were distantly related and (ii) a common ancestral DH.	Simon Fraser University, Burnaby, British Columbia Credited in paper.	

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Katsanis, Beales, Woods, et al. (2000)[85] *Letter	Full informed consent was obtained in concert with the Institutional Review Boards for Human Subject Research at the sponsoring institutions	Included 5 NL and 2 European American BBS pedigrees. (40 total individuals, 31 individuals from NL)	extracted DNA from venous lymphocytes A detailed family history and pedigree were obtained through personal inter views with appropriate family members. Available pediatric, ophthalmologic, electrophysiologic, otologic, renal, orthopedic, endocrinologic and genetic records were recovered and reviewed.	Haplotype and mutation analysis.	lymphocytes for exclusion studies from all known BBS loci by genotyping all available individuals with published markers from the respective critical intervals and constructing haplotypes. Suitable pedigrees were subsequently used for a genome screen with the CHLC 10 cM screening set version 8 (Research Genetics); alleles were analysed and lod scores were tabulated. Fine-mapping studies used additional microsatellites derived from the Genome Database. Amplified PCR products from patients, relatives and control samples were purified with the Qiagen 96-PCR purification kit (Qiagen) and sequenced with dye-primer chemistry (Applied Biosystems) in an ABI 377 automated sequencer (Applied Biosystems). PCR products were also cloned into the TA vector (Invitrogen) and sequenced to separate the different alleles. Resulting sequences were aligned and mutations were evaluated by the Sequencher sequence	Dr. James R. Lupski, Department of Molecular and Human Genetics Baylor College of Medicine, Room 609E, One Baylor Plaza, Houston, TX jlupski@bcm.tmc.edu

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					alignment program (ACGT Codes). MKKS exon amplification primers are available. *Unclear if data can be linked to administrative data or who holds the data.	
Beales, Katsanis, Lewis, et al. (2001)[86]	Informed consent obtained from patients. Protocols approved by the appropriate oversight committees at each institution	163 BBS pedigrees were screened for mutations in *MKKS (BBS6).* 50 (27 North American/ European and 2 NL pedigrees for which *BBS1* was excluded through haplotype analysis, and 21 consanguineous pedigrees of Turkish, Iraqi, Pakistani, and Indian origin) were included. BBS6 gene McKusick-Kaufman (MKKS) syndrome gene (MKKS) on chromosome 20 and for potential assignment of the disorder to any of the other known	Blood samples provided and DNA was extracted by a salting-out process In several cases, the diagnosis was ascertained by local physicians and verified through extensive examination of medical records.	Combinatorial strategy of linkage disequilibrium and haplotype analysis. For the genetic analyses, a total of 54 custom-synthesized (MWG/ Sigma-Genosys) fluorescent microsatellite STRPs (short-Tandem-repeat polymorphisms) were typed for each family member: for *BBS1: D11S4205, D11S1883, D11S599, D11S449, D11S1889, D11S4909, D11S4946, PYGM, D11S4945,* and *D11S4944;* for *BBS2: D16S411, D16S415, D16S419, D16S390, D16S408, D16S526, D16S3089, D16S265, D16S3034, D16S408, D16S3057, D16S514, D16S503, D16S400, D16S421,* and *D16S515;* for *BBS3: D3S1566, D3S1276, D3S3634, D3S1603, D3S1251, D3S2419, D3S1271,* and *D3S1278;* for	mutations in the MKKS gene Products were resolved on an ABI 377 automated sequencer, and alleles were assigned with GENESCAN v2.3 and GENOTYPER v2.1 software (Applied Biosystems). Sequence data were managed on Sequencher (Genecodes Corporation). Linkage analysis was performed, where appropriate, with the LINKAGE package of programs: two-point linkage data were generated with MLINK (assuming a gene frequency of 0.005 and a penetrance of 0.9); multipoint analyses were performed with LINKMAP. No information provided on data availability or ability to link with administrative data. Primers were provided by the Lupski lab in Houston, TX.	Dr. James R. Lupski, Department of Molecular and Human Genetics Baylor College of Medicine, Room 609E, One Baylor Plaza, Houston, TX jlupski@ bcm.tmc.edu

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		BBS loci in the human genome. *unclear how many individuals from NL are included.		*BBS4: D15S117, D15S153, D15S125, D15S988, D15S814, D15S650, D15S131 D15S204, D15S114,* and *D15S205;* for *BBS5: D2S151, D2S142, D2S156, D2S2330, D2S335,* and *D2S326;* and for *BBS6: D20S115, D20S851, D20S189,* and *D20S186.*		
Moore, Green, Fan et. al (2005)[87]	Approval obtained from Human Investigation Committee of MUN and Simon Fraser University REB. Informed consent was obtained for all participants in the study	46 patients (26 males, 20 females) from 26 families.	Blood sample; Also, neurologic assessments, anthropometric measurements, and clinical photographs	Linkage to *BBS1, BBS2, BBS3, BBS4*, or *BBS5* was assessed using microsatellite markers. Linkage to *BBS7* or *BBS8* was examined by haplotype analysis	*No statement regarding where genetic data was deposited *No statement on data availability.	Dr. Patrick S. Parfrey Clinical Epidemiology Unit, Health Sciences Centre, Memorial University, St John's, NL pparfrey@mun.ca
Lynch syndrome						
Stuckless, Parfrey, Woods, et al. (2007)[88]	Informed consent obtained from all subjects or an appropriate proxy. Ethics approval was granted by the Human Investigations Committee of the Faculty of Medicine, MUN, the Health Care Corporation of St. John's and the	More than 300 families with high or intermediate risk for hereditary CRC have been referred to the Medical Genetics Program of NL. 52 families met either the Amsterdam I or Amsterdam II criteria. 290	DNA from all available family members at 50% risk of inheriting an MSH2 mutation was prepared from whole blood using a simple salting-out method.	The point mutation in the splice donor site of intron 5 in MSH2 (c.942 + 3A > T) was determined by restriction fragment analysis. Exon deletions in MSH2 were detected by Multiplex Ligation-dependent Probe Amplification (MLPA) using genomic DNA	Genotyping, using microsatellite markers, for families segregating the exon 8 deletion performed on either an ABI 3100 Genetic Analyzer (Applied Biosystems) or a Beckman Coulter CEQ 8000 Genetic Analysis System (Beckman Coulter Inc., Fullerton, CA, US). M. MLPA, using the HNPCC probes (kit no. SALSA P003) conducted	Dr. Jane S. Green Department of Genetics, Faculty of Medicine, Health Sciences Centre, Memorial University of NL, A1B 3V6, St. John's, NL

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	Avalon Peninsula Health Board.	people from 18 families confirmed to have MSH2 mutation Mut.: intron 5 splice site; fam #: 12; # Individ: 151 Mut.: exon 8 deletion; fam. #: 5; # individ.: 74 Mut.: exon 4-16 deletion; fam #: 1; individ.: 65 235 individuals (81%) found to be mutation positive, remaining 55 (19%) considered presumed positive			and analyzed according to protocol provided by MRC-Holland (Amsterdam, Holland) on an ABI 377 Genetic Analyzer (Applied Biosystems, Foster City, CA, US). Marker positions on chromosome 2 were identified using the Genome Browser from the UCSC Genome Bioinformatics website.	
Campbell, Edwards, McLaughlin, et al. (2007)[89]	Written informed consent was obtained from all subjects for use of their DNA samples for research purposes consistent with this study. The ethics review board of	Genotyped 146 Caucasian people with a Lynch syndrome mutation (5′-untranslated region poly-morphism in cytochrome P450 17A1 (*CYP17*; c.−34T→C, exon	Genomic DNA was extracted from leukocytes using a common salting out procedure People had data on age and site of cancer diagnoses, colon screening history, polyp detection, and vital status	genotyped 146 Caucasian Lynch syndrome mutation carriers for a 5′-untranslated region polymorphism in cytochrome P450 17A1 (*CYP17*; c.−34T→C) and an exon 4 polymorphism in catechol *O*-methyltransferase (*COMT*; c.472G→A); 50 mutation carriers had	Genotypes determined blinded to the subjects' cancer status. *YP17* and *COMT* genotypes determined by PCR and RFLP techniques. 459 bp fragment of *CYP17* was amplified using the following primers: forward 5′-TTC TTC CAC AAG GCA AGA GA-3′; reverse 5′-TTG GGC	Dr. Micheal O. Woods Discipline of Genetics, Faculty of Medicine, Health Sciences Centre, St. John's, NL mwoods@mun.ca

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	the MUN approved this study.	4). Included (*n* = 161). From 161 samples, 15 were excluded for low DNA yield. 92 DNA samples from population-based controls in NL to compare allele frequencies in the current study with that of the general population (identified through random-digit dialing and frequency-matched on age and sex to the case population)		developed colon or rectal cancer at last contact.	CAA AAC AAA TAA GC-3′. The 25 µL PCR reaction for *CYP17* included 100 ng of genomic DNA, 1 × reaction buffer, 3.0 mmol/L of $MgCl_2$, 0.2 mmol/L of each deoxynucleotide triphosphate, 0.04 units/ µL of Taq polymerase, and 0.6 µmol/L each of forward and reverse primers. The region containing the *COMT* exon 4 polymorphism was amplified using the primers: forward 5′-TAC TGT GG CTA CTC AGC TGT GC-3′; and reverse 5′-GTG AAC GTG GTG TGA ACA CC-3′. The 20 µL PCR reaction for *COMT* included 200 ng of genomic DNA, 1 × reaction buffer, 0.2 mmol/L of each deoxynucleotide triphosphate, 0.05 units/µL of Taq polymerase, and 0.2 µmol/L each of forward and reverse primers.	
Woods, Williams, Careen, et al. (2007)[90] Kohonen-Corish., Macrae, Genuardi, et al. (2011)[91] Plazzer, Sijmons, Woods, et al. (2013)[92]	N/A	The InSiGHT database is the primary store of public information of inherited gastrointestinal cancer gene variants. The intended uses of the	If the InSiGHT database can be realized by detailed analysis of the variant spectrum, by gene and exon, especially when cross-referenced to patient demographics and disease information.	A database entry is defined as a single record of a variant in the database. A unique variant is a summary account of all the entries of a variant. sharing of variant information from individual clinics and regional or national organisations. 1 central and public	"Probably the most used and referenced database is that which is associated with the International Society for Gastro-intestinal Hereditary Tumours (InSiGHT; www.insight-group.org). However, this is primarily a submission catalogue and INSIGHT has made no	Curator of the InSiGHT database: John-Paul.Plazzer@mh.org.au MMR Genes Variant Database: Dr. Micheal O. Woods Discipline of Genetics, Faculty of Medicine,

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Thompson, Spurdle, Plazzer, et al. (2014)[93]		database are to assist clinicians in providing accurate health care to their patients and to facilitate biological and clinical studies on variants associated with hereditary GI syndromes. As of May 2012, there are 12,538 entries for 3,072 unique variants across the MMR genes in the InSiGHT database.	Additionally, the results of the interpretation process will allow the calibration of automated methods of variant classification	web-accessible database powered by the Leiden Open Variation Database (LOVD) [1] platform InSiGHT which had 3 separate databases which could be combined. Thus, the InSiGHT database became a model system of the HVP for other gene/ disease groups to emulate.	attempt to actively assemble published data. It does have the advantage, however, of having an extensive list of unpublished variants describing the geographical locale of the variant carrier and whether the alteration was considered pathogenic or nonpathogenic by the submitter." This database was then merged into the InSiGHT database The International Society for Gastrointestinal Hereditary Tumours (InSiGHT) is a database with information on variants contributing to GI cancers. It includes information on the. There has been data submitted on NL participants. In 2007, a public database was established at MUN: the Mismatch Repair Genes Variant Database (MMRGVD Literature) [4]. The curators of this database obtained variants solely from published articles, consisting of 6,136 entries on 2,260 unique variants. This would go on to represent the largest single component of the	Health Sciences Centre, St. John's, NL mwoods@mun.ca

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					InSiGHT database with 49% of total entries and covering 74% of unique variants. Of these, 1366 unique variants had not been submitted directly by any other contributor to the InSiGHT database (Table 2). 580 disease phenotype descriptions were listed, associated with 372 unique variants. 259 reports had Amsterdam or Bethesda criteria (positive or negative) listed. All of this information was obtained from 1,100 published articles InSiGHT, as a GDSDb will receive data from HVP Country Nodes, such as the Australian Node which is currently expanding to cover 15 laboratories. The Human Variome Project (HVP) aims to collect all human genetic variation affecting health to a central resource that is freely available, and mobilize the scientific and medical community worldwide to participate in this effort. The International Society for Gastrointestinal Hereditary Tumours (InSiGHT, www.insight-group.org) established a pilot program	

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					with the HVP in 2007 to collect all inherited variation affecting colon cancer susceptibility genes. multiple gene mutation/ variant repositories have been merged or linked forming the InSiGHT Colon Cancer Gene Variant Databases Other work includes fostering the worldwide MMR consortium, which is an initiative between InSiGHT and the NCI Colon Cancer Family Registry, born at the Washington meeting of the 2 groups on April 27, 2010. The InSiGHT Databases now have over 3,800 unique variants listed. Initially, gene-specific data from the original InSiGHT Mutation database was merged with 2 other MMR gene databases from NL[90] and from the Netherlands. accompanying phenotype information was absolutely essential for the genotypic data to be useful.	
Win, Dowty, Reece, et al. (2021)[94]	Study approved by the institutional human ethics committees, institutional review	Data from the International Mismatch Repair Consortium (IMRC)	For each family, data were collected on identification number; the mismatch repair gene with	—	Data for this study were contributed by the IMRC investigators. Availability of these data will depend on the agreement of	Dr. Micheal O. Woods Discipline of Genetics, Faculty of Medicine, Health Sciences

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	boards, or central national authorities of participating centres. Participants provided written or oral informed consent, whenever required by these committees.	(organized by InSiGHT Used secondary from 5585 (173 families from Canada) families (collected between July 11, 2014, and Dec 31, 2018)	the pathogenic variant; method of ascertainment of the family (ie, a population-based source, such as a cancer registry, or after presentation to a familial cancer clinic or genetics clinic); the date the family was ascertained; and the person in the family first identified as carrying the pathogenic variant (the proband). For each family member, data were collected on the personal, mother, and father identification numbers; sex; carrier status of a pathogenic variant (ie, a carrier, non-carrier, or untested); genetic testing date; cancer diagnoses (anatomic site and age at diagnosis); polypectomies and bowel surgery (age at the time of surgery); ages at the time of pedigree collection and at last contact or		the investigators who contributed the data to the IMRC. Upon agreement, deidentified, individual participant data that underlie the results reported in this Article will be made available, together with data dictionaries and the study protocol, with publication of all IMRC prespecified manuscripts. Data will be available to researchers who provide a methodologically sound proposal for use in achieving the goals of the approved proposal. Proposals can be submitted according to the instructions provided. To gain access, data requestors will need to sign a data access agreement with the University of Melbourne and participating IMRC centres.	Centre, St. John's, NL mwoods@mun.ca

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			death; and country of residence.			
Colorectal Cancer (CRC)						
Woods, Hyde, Curtis, et al. (2005)[95] *Only abstract available. Wish, Hyde, Parfrey, et al. (2010)[96] Woods, Younghusband, Parfrey, et al. (2010)[97] Clarke, Green, Green, et al. (2012)[98] DeRycke, Gunawardena, Middha, et al. (2013)[99] Haja Mohideen, Hyde, Squires, et al. (2014)[100] Haja Mohideen, Dicks, Parfrey, et al. (2015)[101] Xu, Green, Green, et al.(2015)[102] Zhu, Wang, Zhai, et al. (2017)[103] Zhu, Wang, Zhai, et al. (2018)[104]	Ethics approval obtained from the REB of MUN. All patients or their proxies provided informed consent for access to previously collected tumour/ tissue samples and medical records. *Secondary use studies approved, including studies doing additional sample testing	NL Familial Colon Cancer Registry (NFCCR): Patients were eligible for the study if diagnosed with colorectal carcinomas (pure adenomas not included) between 1 January 1999 and 31 December 2003 and were <75 years of age at diagnosis. 750 (64%) from 708 different families consented to be in study. Controls were identified by random digit dialing from the residents of the province, and matched to the cases on sex and 5 year age group. Controls provided a blood	Blood sample (552 people), Tumour tissue (772 tumours from 750 cases) people) and medical records (728 patients from 685 families) Family questionnaire collected, which enabled pedigrees to be constructed. From the family history questionnaire, recorded the type of cancer, age at diagnosis, and age at death or age at last follow-up for all first-degree relatives (FDR). Cancer diagnoses in consenting family members confirmed by medical records when possible. Pathology reports available. 16 families selected based on the presumption of a genetic predisposition to disease due to	Pannels: DNA sequencing and multiplex ligation-dependent probe amplifications of MMR genes and APC was undertaken. DNA from all patients was screened for MUTYH mutations. The presence of the BRAF variant, p.V600E, and of MLH1 promoter methylation was also tested in tumours.[96-98] Genotyping experiments were performed for 539 patients with available prognostic data at a service provider (Centrillion Biosciences, US) using the Illumina Human Omni1-Quad SNP genotyping platform. This cohort contains > 1 million SNP markers. After quality control, 505 had usable data.[97] Investigate the associations of polymorphisms from the selected genes (*HIF1A*, *HIF1B*, *HIF2A*, *LOX*, *MIF* and *CXCL12*) in a larger patient cohort. 77 SNPs to be included. the 77 SNPs	Pannels: Methylation was detected using the MS-MLPA kit ME001B (MRC-Holland) Automated sequencing was done on an ABI 3700 DNA Analyzer (Applied Biosystems). Transgenomic Wave 3500HT System, Omaha, Nebraska, US) was used to detect MUTYH mutations.[96-98] In total, 2 pathogenic variants were identified in at least 11 probands (3: c.907G > A (p.Asp303Asn, 1: c.1187A > G (p.Tyr396Cys)) variant) from 4 different families All variants have been submitted to the LOVD CRC database (www.lovd.nl/GALNT12) (this was only 4 people) Genotypes were obtained using the Illumina Human Omni1-Quad Bead Chip at a service provider (Centrillion Genomic Services, US).[100] Whole-genome SNP genotyping using the Illumina® Omni1-Quad	Patrick S. Parfrey Health Sciences Centre, 300 Prince Philip Drive, St. John's, NL A1B 3V6 pparfrey@mun.ca Dr. Micheal O. Woods Discipline of Genetics, Faculty of Medicine, Health Sciences Centre, St. John's, NL mwoods@mun.ca Dr. Sevtap Savas Memorial University, Division of Biomedical Sciences, Human Genetics and Genomics savas@mun.ca[100,101] Peizhong Peter Wang Division of Community Health and Humanities, Faculty of Medicine, Memorial University of NL, St. John's, NL pwang@mun.ca[104] Request to access the datasets should be directed to NL Colorectal Cancer Registry (PP; pparfrey@mun.ca) and Research, Grant,

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
Curtis, Yu, Carey, et al. (2022)[105] Savas, Xu, Werdyani, et al. (2015)[106]		sample and filled out a risk factor questionnaire. *Cohort 2 in Haja Mohideen, Hyde, Squires, et al. (2014)[100]	1) large numbers of affected relatives and 2) younger ages at diagnosis Forty affected individuals were chosen for sequencing based on genetic relatedness (preferring distant relatives), including 3 cases per family where possible.[99] *149 controls, 118 probands (only 4 with the variant inputted into the LOVID CRC database)*[98]	were obtained as a part of a whole genome SNP genotyping study (535 patients). Primer sequences for each of the 10 exons of GALNT12 were described by Guda et al. All exons andintron/exon boundaries were screened for variants using standard protocols for automated direct sequencing.[98] Patients were genotyped 6 (Mitochondrial DNA) mtDNA polymorphisms (16189 (T/C; rs55749223), 10398 (A/G; rs2853826), (MitoT479C, rs41442247; MitoT491C, rs28625645; MitoT10035C, rs41347846; MitoA13781G, rs41358152). In addition, out of 736 patients, 276 patients (274 have results) were investigated for the mtDNA copy number change based on the availability of their DNA samples extracted from both the tumour and non-tumour colon or rectum tissue; 210 of these patients were also investigated in the genotyping analysis. (performed at an outsourced genotyping facility (Centrillion Genomics Services, CA, US).)	human SNP genotyping platform (service provider: Centrillion Biosciences, US).[102] Genotyping conducted using the Illumina Human Omni-Quad Beadchip that contains about 1.1 million SNPs at Centrillion Biosciences (US). Control individuals were genotyped in the Laboratory of Dr. Stephen Gruber (Director, USC Norris Comprehensive Cancer Center, Los Angeles) using the Affymetrix Axiom® myDesign™ GW Array Plate, which contains 1.3 million probes.[104]	and Contract Services (rgcs@mun.ca) at Memorial University of NL, St. John's, NL, and the ethics approval shall be obtained from the Health Research Ethics Board (HREB), Ethics Office, Health Research Ethics Authority.[105]

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				The nucleotide positions of these SNPs are annotated based on the Cambridge Reference Sequence (NC_001807.4), whereas the positions of the TaqMan® genotyped polymorphisms are based on the revised Cambridge Reference Sequence (NC_012920.1)[101] Initially, a total of 539 patients with available prognostic data and germline DNA were subject to whole-genome SNP genotyping. QC analyses on the genetic data were performed using PLINK v1.07 and Eigensoft 4.2. After QC analyses, a total of 505 subjects and 729,737 SNPs were included in the final analysis.[102] Genotyping of peripheral blood DNA samples was performed using the Illumina Human Omni-Quad Bead chip that contains about 1.1 million SNPs at Centrillion Biosciences (US). For quality control purposes, genomic DNA from 200 duplicate samples were sent to the Laboratory of Dr Stephen Gruber (Director,		

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				USC Norris Comprehensive Cancer Center, Los Angeles) for genotyping using the Affymetrix Axiom my Design GW Array Plate, which contains 1.3 million probes. SNPs with genotype concordance < 97% between the 2 platforms were dropped from all analyses."[103] Genotyping for the GC rs2282679 allele To monitor quality and consistency between the 2 platforms, DNA samples from 200 CRC patients were typed on both platforms. As the DNA from cases and controls were genotyped on different platforms, a genotype imputation strategy was implemented to integrate the 2 datasets using IMPUTE2 with multi-population reference panels from 1000 Genomes (Phase 1). The imputation approach was validated based on the overlapping SNPs between the 2 platforms and the genotypes from 200 CRC samples that were typed on both platforms. SNPs with genotype concordance < 97% across the 2 platforms were removed		

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				from further analysis. For the purpose of the current study, directly measured data from both arrays on rs2282679 were retrieved from the genome-wide SNP genotype database of the NFCCR.[104] Whole exome sequencing was completed using SNP technology to identify new colon cancer associated loci. This was done differently in different families. It is unclear how NL families conducted sequencing and library preparation.[99]		
Dorani, Hu, Woods, and Zhai. (2018)[107] Schumacher, Schmit, Jiao, et al. (2015)[108] Bien, Su, Conti, et al. (2019)[109] Guo, Lin, Wen, et al. (2021)[110] Thomas, Sakoda, Hoffmeister et al. (2020)[111] Archambault, Su, Jeon, et al. (2020)[112] Archambault,	Original data collection approved by MUN Health Research Ethics Authority (HREA) with the approval number HIC 01.70. Present studies recognized by HREA as the use of secondary data which have already been collected and de-identified, and did not require a clearance.	***NFCCR dataset used, see above.*** These samples were processed for larger studies developed by the colorectal cancer Transdisciplinary (CORECT) consortium *(672 people: 195 cases, 477 controls on the Affymetrix Axiom platform. This was also stated as being*	See above.	GWAS array data (with approximately 1.3 million SNPs and indels on 2 physical genotyping chips (pegs). Prior to imputation, SNPs with < 95% call rate, concordance < 95% with 1000 Genomes in samples genotyped for quality control, or HWE P < 10^{-4} in controls were excluded. All SNPs overlapping 1000 Genomes were matched to the forward strand.) from NL was done at McGill University and Génome Québec Innovation Centre, Montréal, QC, for	The CRC Transdisciplinary (CORECT) consortium coordinated the genotyping of data. Genotyping was conducted using a custom Affymetrix genome-wide platform (the Axiom CORECT Set) on 2 physical genotyping chips (pegs) for 2 datasets with around 1.2 and 1.1 million SNPs. The first dataset has 1,236,084 SNPs and 696 samples with 200 cases and 496 controls and the genotyping rate was 0.997. In GWAS, genotyping rate is computed as the percentage of	Dr. Ting Hu Department of Computer Science, Memorial University, St. John's, NL ting.hu@mun.ca Dr. Micheal O. Woods Discipline of Genetics, Faculty of Medicine, Health Sciences Centre, St. John's, NL mwoods@mun.ca Contact: Ulrike Peters, Email: upeters@fredhutch.org .[111] Dr. Richard B. Hayes

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
Jeon, Lin, et al. (2022)[113] Schmit, Edlund, Schumacher, et al (2019)[114] Huyghe, Bien, Harrison, et al. (2019)[115] Huyghe, Harrison, Bien, et al. (2021)[116]		*part of the CORECT study (supplemental materials*.[108] 17 cases and 68 controls from NFCCR, collected from 2000 to 2004)[112,113]		genotyping the Sequenom panel in on the Affymetrix Axiom Platform) the NFCCR samples For 65 SNPs initially discovered in the GECCO and CORECT studies, estimated the log-odds ratios from a model fit with overall CRC (no age restrictions) as the outcome and the 141 SNPs as independent variables, adjusted for age, sex, principal components, and genotype platform[112,113] Huyghe, Bien, Harrison, et al. (2019)[115] imputed genotype datasets (BAM files) from previous studies to the Haplotype Reference Consortium panel (39.2 million variants) using the University of Michigan Imputation Server.	samples (including both cases and controls) that are successfully genotyped. The second dataset has 1,134,514 SNPs and 656 cases with a genotyping rate of 0.888. Using PLINK, a tool for analyzing genetic data, we merged these 2 datasets based on their common SNPs and obtained a dataset of 265,195 SNPs and 1,152 unique samples. Among the samples, 656 were cases and 496 were controls. After the steps of quality control and imputation, the final preprocessed and balanced dataset had 186,251 SNPs and 944 samples (472 being cases and 472 being controls) Data availability: Colorectal Transdisciplinary (CORECT) Study GitHub Data underlying article (SNP (ex, rs72647484), Locus (ex. 1p36.12), Risk/other (ex. T/C), allele Risk allele freq (ex. 0.9107), and PMID (ex. 25990418) provided for 141 risk variants.) accessed from the Fred Hutchinson Cancer Center. The derived	NYU Langone Health New York, NY Richard.B.Hayes@nyulangone.org).[112,113]

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
					data generated shared on reasonable request to the corresponding author with permission of the Fred Hutchinson Cancer Center.[112,113]	
Savas, Hyde, Stuckless, et al. (2012)[117] Haja Mohideen, Hyde, Squires, et al. (2014)[100] Haja Mohideen, Dicks, Parfrey, et al. (2015)[101] Dan, Werdyani, Xu, et al. (2016)[118]	Approved by the Human Investigation Committee of MUN and the Regional Health Boards. Informed consent not required by local ethics board as study was considered an anonymous chart review. (#12.206, #10.133).	Population-based retrospective cohort of 280 patients followed for ≤ 12.5 yrs after diagnosis. From the Avalon Peninsula of NL, new cases (diagnosed in 2 St. John's health facilities, ascer-tained between Jan 1, 1997 and Dec 31, 1998. *Cohort 1 in Haja Mohideen, Hyde, Squires, et al. (2014)[100]	Surgical specimens available for 280 (272 could be genotyped) of 292 patients identified. DNA extracted from formalin-fixed paraffin-embedded non-tumour colon and rectum tissues. For all 280 patients the clinical data was collected. Prognostic data of these patients was collected from the medical and hospital records and the NL and Labrador Centre for Health Information. DNA samples were extracted from either peripheral blood samples (*n* = 40) or the nontumour colon or rectum tissues obtained during the surgery (*n* = 207)	initially selected 5 tagSNPs in *SLC6A4* (rs11080121, rs12150214, rs2066713, rs4251417, and rs140700) and 2 tagSNPs in *BDNF* (rs7124442 and rs6265) and 1 tagSNP in *AVPR1B* (rs35369693) but, rs11080121 and rs2066713 in *SLC6A4* and rs7124442 in *BDNF* could not be genotyped. Data from the 5 remaining SNPs were included in this study. At least 5% of the samples were genotyped 2 and all duplicated genotypes were concordant.[117] Selected 6 hypoxia pathway genes (*HIF1A*, *HIF1B*, *HIF2A*, *LOX*, *MIF* and *CXCL12*). 49 SNPs from these genes (both tagged and untagged) were successfully genotyped.[100]	No linkability. Due to lack of consents Genotyped using the Sequenom MassArray® technology by The Centre for Applied Genomics Facility, Toronto, ON [117] Sequenom MassArray technology at an outsourcing genotyping facility (UHN Analytical Genetics Technology Centre, ON; n = 35 SNPs) or in-house TaqMan SNP genotyping assays (n = 14 SNPs).[100] Out of the 6 SNPs genotyped, 2 SNPs [16189 (T/C; rs55749223) and 10398 (A/G; rs2853826)] were genotyped using the TaqMan® SNP genotyping technique. The genotype data for the remaining 4 mtDNA polymorphisms (MitoT479C, rs41442247; MitoT491C, rs28625645; MitoT10035C, rs41347846; MitoA13781G, rs41358152) were obtained	Dr. Sevtap Savas Memorial University, Division of Biomedical Sciences, Human Genetics and Genomics savas@mun.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
					using a genome wide SNP genotyping technique (Illumina Human Omni1-Quad genome wide SNP genotyping platform) as part of another project of our team.[101] Sequenom MassArray® technique at a service provider (Clinical Genomics Centre, Mount Sinai Hospital, Toronto, ON). 15 DNA samples genotyped twice (6%) and in all cases the genotypes obtained were identical. All SNPs in this cohort had MAFs ≥ 5% and their genotype frequencies were in HWE equilibrium.[118]	
Familial and sporadic idiopathic pulmonary fibrosis						
Fernandez, Fox, Bhatia, et al. (2012)[119]	MUN's Human Investigations Committee approved this study (#02.26) and all individuals consented to be in the study.	Recruited people with pulmonary fibrosis with the 5 respirologists in NL from January 2006 to July 2011 (90% consented to study. 78 pulmonary fibrosis patients (28 familial plus the recruitment of an additional 49 affected relatives [first degree	DNA was extracted from whole blood using either a simple salting out method or by using the Wizard Genomic DNA Purification Kit. Additional: family history form, medical history form, questionnaire on environmental exposures, medical records, biopsy proven UIP on a	DNA sequencing in both directions was performed for all exons and exon/intron boundaries for 4 of the known genes causing familial pulmonary fibrosis were sequenced in 28 FPF probands and 50 sporadic PF patients: *TERT* (telomerase reverse transcriptase) and *TERC* (telomerase RNA component) Autosomal dominant mutations in surfactant protein C	Automated sequencing was performed on either an ABI 3130 Genetic Analyzer or an ABI 3730 Genetic Analyzer (Applied Biosystems, Foster City, CA). *Only NL researchers credited with DNA sequencing. Likely housed in NL. Was connected with medical records and therefore linkable with administrative data.	Dr. Micheal O. Woods Discipline of Genetics, Faculty of Medicine, Health Sciences Centre, St. John's, NL mwoods@mun.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
		relatives] and 50 sporadic cases). Excluded patients with significant environmental exposures known to be associated with lung fibrosis, or with known collagen vascular diseases,	clinical pathology report or, in the absence of surgical lung biopsy, met American Thoracic Society/European Respiratory Society criteria for IPF Relatives completed Pulmonary Function Tests (PFTs) and a high-resolution chest CT scan (HRCT).	(*SFTPC*) and in surfactant protein A2 (*SFTPA2*). Variant nomenclature and primers were derived from the following RefSeq accession numbers: NG_009265.1 (*TERT*), NG_016363.1 (*TERC*), NG_016968.1 (*SFTPC*) and NG_013046.1 (*SFTPA2*). Telomere length assays carried out on 5 affected individuals with *TERT* mutations from 2 families.	*No stamen of data availability.	
Familial Intracranial Aneurysm						
Powell, Bridget, Fernandez, et al, (2019)[120]	The Health REB (Reference Number 04.89) for MUN approved this study, and all participants provided consent.	Over a 5-year recruitment period assembled 53 IA families containing 154 affected individuals and 415 unaffected family members for study. through collaboration between MUN's Discipline of Genetics and the Department of Surgery (Division of Neurosurgery, only neuro department in NL) at Eastern Health. Referred	Whole blood samples provided by people affected by IA and family members. Diagnosis of IA confirmed through CT imaging of the Circle of Willis or by magnetic resonance angiography. Consenting relatives obtained through family history offered CT angiography, underwent a comprehensive medical and family history assessment.	For this specific paper: WES completed for a cohort of 12 affected individuals (5 from 1 family, 7 from the other) from 2 multiplex families (> 11 affected family members) Filtered variants were prioritized based on validation by Sanger sequencing and segregation within the families. Following this filtering process, variants C4orf6 c.A1G (p.M1V) and SPDYE4 c.C103T (p.P35S) were sequenced using Sanger sequencing in 100 population controls.	Genomic DNA was extracted from whole blood using either a simple salting out method or the Wizard® Genomic WES was completed at McGill University and Genome Quebec Innovation Center (MUGQIC, Montreal, QC). The Agilent SureSelect 50 Mb All Exon kit was used for exome capture and paired-end sequencing was completed on the Illumina® HiSeq 2000 platform. *No statement of data availability. Seems to be on-going. Likely likable with administrative data through	Dr. Micheal O. Woods Discipline of Genetics, Faculty of Medicine, Health Sciences Centre, St. John's, NL mwoods@mun.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
		to study if presented or had family member that presented with ruptured or unruptured IA. *Not all participants couple provide a DNA sample. For this specific paper, 100 controls from CRC study (described in CRC section also included).			medical insurance number since obtained through clinical interactions and findings from this study can impact patient health.	
Autosomal dominant arrhythmogenic right ventricular cardiomyopathy (ARVC)						
Hodgkinson, Parfrey, Bassett, et al. (2005)[121]	Consent for analysis of past and current cardiology records and for DNA analysis was obtained from each subject (or surrogate) in compliance with the Human Investigation Committee requirements of the Health Care Corporation of St. John's. All available clinical records and autopsy reports were obtained.	367 subjects (212 male, 155 female) from "well-ascertained" sibships (those where ≥ 50% of siblings were known to be at high or low risk of ARVC as determined on the basis of clinical, pedigree, and/or haplotype data.) from 11 families were studied.	Blood sample: DNA extracted from peripheral lymphocytes (201 out of the 367 subjects)	Part 1: A NL family with autosomal-dominant ARVC was reported in 1988. An affected-only linkage analysis was performed. 17 affected individuals who had DNA samples available for molecular analysis. Linkage of disease to 3p25 was confirmed with a maximum multipoint logarithm of the odds score of 9.3 at D3S1585. Analysis of 8 markers allowed the construction of a chromosome 3p25 HR haplotype with D3S3610-Fibulin2-D3S2385-D3S1585-D3S1554 to	Part 2: Fluorescent-labelled primers used to amplify di-, tri-, or tetra nucleotide repeats by the polymerase chain reaction, and fragments analyzed by capillary gel electrophoresis by the use of an ABI 310 genetic analyzer (Applied Biosystems, Foster City, California).	Dr. Kathleen Hodgkinson Discipline of Genetics, Faculty of Medicine, Health Sciences Centre, St. John's, NL, khodgkin@mun.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				708d1CA-316A10CA-D3S3613 ordered from telomere to centromere shared by all affected family members. Part 2: 10 additional families with ARVC were studied, and the critical region was reduced to 2cM, at which locus the identical set of DNA markers (haplotype) co-segregates with ARVC. The presence or absence of the 2cM HR haplotype was determined for each family member assessed.		
Merner, Hodgkinson, Haywood, et al. (2008)[122]	Informed consent was obtained in compliance with the Human Investigation Committee requirements of the Eastern Health Corporation of St. John's, NL (study number 00-176).	15 unrelated families referred to the NL Provincial Medical Genetics Program or the NL Labrador genetics cardiomyopathy clinic for family history of cardiomyopathy and sudden death. Families determined to have ARVC on the basis of clinical testing with established criteria. Total of	Blood samples from 295 subjects born at a priori 50% risk collected. (201 not DNA tested for mutation)	Clinical data prospectively collected over 11 years. Step 1: Screening candidate genes. Mutation-screening panel established with genomic DNA from 4 clinically affected subjects from 3 families and 3 spouses (controls). All coding and noncoding exons and intron-exon boundaries of positional candidate genes for *ARVD5* sequenced. Sequencing variants found exclusively in clinically affected subjects on the mutation-screening panel were experimentally determined to reside on the *ARVD5* haplotype by	Step 1: Purified PCR products were cycle sequenced in both forward and reverse directions with the use of BigDye Terminator V3.1 cycle sequencing kit on an automated ABI 3700 DNA analyzer (Applied Biosystems) Sequencing electropherograms were inspected manually and analyzed with Mutation Surveyor software (Transition Technologies). The URLs for data presented herein are as follows: • NCBI dbSNP	Dr. Terry-Lynn Young Memorial University of NL St. Johns, NL tlyoung@mun.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
		496 subjects born at a priori 50% risk (clinically affected and their first-degree relatives) available for study. Only subjects from well-ascertained sibships in study.		segregation analysis. Step 2: Genotyped 18 polymorphic microsatellite markers on clinically affected subjects across all families to identify a disease-associated haplotype at 3p25. Identified key recombinations and reduced the disease region to a 2.36 Mb interval containing 20 annotated genes. Bidirectional resequencing of the 20 physical candidate ARVD5 genes revealed 240 variants. Nineteen variants found exclusively in clinically affected subjects on the mutation-screening panel (based on also screening 161 population controls from a colorectal study, and 47 spouses), and 11 were determined to reside on the ARVD5 ancestral haplotype through segregation analysis in 1 family. Only 1 of the 5, *TMEM43* 1073C→T (S358L), shared by all clinically affected subjects Step 3: Sequenced genomic DNA from all available subjects born at a priori 50% risk (n = 295) across the 15 ARVC families for the	• Online Mendelian Inheritance in Man (OMIM) • UCSC Genome Browser	

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				presence of the 1073C→T *TMEM43* mutation. **Clinical data:** Annual visits by subjects to genetics cardiomyopathy clinic in which 12 lead ECGs, Holter monitors, MRIs, signal-averaged ECGs, and echocardiograms were done. Clinical data obtained retrospectively from medical records including "at-risk" relatives not seen in clinic and autopsy results. Subjects categorized as affected, unaffected, or unknown. (in which disease status was known in ≥ 50% of siblings) were included in this study.		
Haywood, Merner, Hodgkinson, et al. (2013)[123]	Study complies with the Declaration of Helsinki; informed consent was obtained in compliance with the Human Investigation Committee requirements of the Eastern Health Care Corporation of St John's, NL (study # 00-176)	*Study included both UK and NL probands. NL population-based controls (*n* = 161) were obtained through random phone dialling, as part of a large colorectal cancer study.	Blood samples.	To identify which recurrent mutations were due to potential founder effects, comparison of the *ARVC*-haplotypes was carried out. DNA from NL probands was genotyped for 5 microsatellite markers spanning *TMEM43* on chromosome 3p25, and disease haplotypes inferred by comparing alleles in UK probands to those residing on the disease haplotypes in NL patients with the same mutation.	Variant analysis was carried out using the Mutation Surveyor v3.25 software (SoftGenetics, LLC, PA, US). No mention of data availability.	Dr. Terry-Lynn Young Memorial University of NL St. Johns, NL tlyoung@mun.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				33 variants identified. 15 variants identified from bidirectional sanger sequencing of UK cohort had been found in control NL population previously so were non-pathogenic. Of the 18 variants remaining from the original 33 identified in UK probands and not seen in the NL controls, 5 were present in > 1% of the UK population control panels and therefore classified as 'no known pathogenicity'. This included 2 synonymous (p.P74P and p.S303S) and 3 intronic (c.443 to 48 G > A, c.1203 + 719 G > T, c.1203 + 771_775 delCAAAA) variants (*Table 2*). Our data support the current classification of 3 of these 5 variants (p.P74P, p.S303S, c.443 to 48 G > A) as having unknown pathogenicity in the ARVD/C database.		
Hodgkinson, Connors, Merner, et al. (2013)[124]	Human REB of the Eastern Health Care Corporation of St. John's study # 00-176 Participants provided informed consent	15 NL families referred to a genetic cardio-myopathy clinic with a history of sudden cardiac death and were found to have a p.S358L mutation in *TMEM43* gene.	DNA was extracted from peripheral lymphocytes using standard methods Additionally accessed medical records for all subjects. Prospective serial 12-lead ECGs [≥ 1 available for 300 subjects],	Subjects (*n* = 295) initially had DNA haplotype analysis at the 3p25 *ARVD5* locus and subsequent direct mutation analysis for the p.S358L mutation in *TMEM43* (present = 144, absent *n* = 151).	No information provided on sequencing software or data availability.	Dr. Kathleen Hodgkinson Discipline of Genetics, Faculty of Medicine, Health Sciences Centre, St. John's, NL khodgkin@mun.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
		Subjects were unaffected (*n* = 164) if they did not have *TMEM43* p.S358L. 412 subjects were included in the study: **258** affected) and **154** unaffected.	echocardiograms [≥ 1 available for 248 subjects] and Holter monitors were [≥ 1available for 239 subjects] available from 1998 onward. All clinical assessments were repeated at 1- to 3-year intervals.			
Hodgkinson, Howes, Boland, et al. (2016)[125]	Consent for retrospective and current chart review and for genetic analysis was obtained from each individual (or surrogate) in compliance with institutional review committee and regional ethics board (study no. 00.176).	Studied 24 multiplex families followed for ≤ 20 years segregating an autosomal dominant p.S358L mutation in TMEM43. 229 unaffected, 393 affected, 240 unknown. Family histories obtained from which 862 individuals who were born at an a-priori 50% pedigree risk were identified	*No new information provided, see above*	Genetic results obtained initially using a research-generated disease-associated haplotype[24] and later direct mutation analysis.	Managed at the Provincial Cardiac Genetics Clinic No information provided on sequencing software or data availability.	Dr. Kathleen Hodgkinson Discipline of Genetics, Faculty of Medicine, Health Sciences Centre, St. John's, NL khodgkin@mun.ca
Spastic Ataxia						
Bourassa, Meijer, Merner, et al. (2012)[126]	Approved by the relevant ethic committees, and	110 study participants (107 from 4	blood was collected for DNA analyses.	A mutation screening panel of 53 genes, composed of 2 affected individuals	PCR analysis: analyzed with Mutation Surveyor v.3.0 (SoftGenetics)	Dr. Guy A. Rouleau Department of Medicine, Université de Montréal,

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
	informed consent was obtained from all subjects	NL families and 3 from ON probands) + 169 NL population controls (used from previous NFCCR cohort)	Diagnosed by single neurologist	with the ancestral disease haplotype from 2 different families and an unaffected control, was established. PCR analysis of all coding exons and flanking intron regions of the 53 genes and predicted genes located in the 1.9 Mb disease haplotype (sequenced at the McGill University and Génome Québec Innovation Centre) was conducted. Searched for rare variants (those with less than 1% allele frequency) that were exclusively present in the 2 affected individuals Then, determined the allele frequency of remaining interesting variants by screening 169 NL population controls who were previously collected for NFCCR study. single variant of interest was observed in vesicle-associated membrane protein 1 (*VAMP1* [MIM 185880]) at position chr12: g.6574054T > G. *VAMP1* has 3 annotated isoforms, *VAMP1A*, *VAMP1B*, and *VAMP1D*, that only differ by their last exon: *VAMP1A* and *VAMP1B* is c.340 + 2T > G, and the *VAMP1D*		Montréal, Quebec guy.rouleau@umontreal.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				mutation is c.342T > G (p.Ser114Arg). The c.340 + 2T > G variant is thought to be the disease-causing variant. Segregation of this particular variant was tested with genomic DNA from every participant, and the variant segregated with the disease in all 4 families, as well as in the 3 single cases from ON. The nucleotide mutation was also assessed in variant databases.		
Catecholaminergic polymorphic ventricular tachycardia						
Abdel-Razek, Collier, Predham, et al. (2017)[127]	Not stated.	66 total (33 affected, only 16 diagnosed by mutation) and both those affected and their family members (individuals born at an a-priori 50% risk) from 3 pedigrees.	Uknown	*May not be relevant, just a chart review* Individuals were included if disease status of ≥ 50% of their sibship was known (n = 66), but this included people diagnosed through testing for mutation and/or obligate carriers (OC) and/ or documented SCD < 50 years so not necessarily genomic data. 66 total (33 affected, only 16 diagnosed by mutation) and both those affected and their family members. Collected genetic and clinical records from medical data.	Unknown	Unknown

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
Breast Carcinoma						
Etchegary, Pike, Puddester, et al. (2022)[128]	study was approved by the provincial Health REB and Eastern Health's Research Proposal Approvals Committee (Ref#: 2022.125).	Not available	Not available.	This is a protocol for a dataset of BRCA and LS mutation carriers in NL (identified from 2006 (CPS testing start date) to present day) To assess (1) Health care utilization (uptake of risk management interventions—screening, surgeries, including prophylactic surgery, specialist appointments), (2) adherence to recommended risk management interventions, (3) cancer outcomes (cancer incidence, stage at cancer diagnosis). NLCHI will link the data collected from the PMGP in Obj. 1. Wiil include people with or without a history of cancer with any molecularly confirmed hereditary cancer syndrome [HCS] (any 1 or a combination of any hereditary cancer genes including *LS* genes, *BRCA1/2*, *ATM*, *CHEK2*, *PALB2*, *RAD51C*, *CDH1*, *MEN1*, *VHL*, etc.) This will also include qualitative data. *Data about the mutations themselves is not the priority but the mutations	Not available.	Dr. Holly Etchegary Department of Clinical Epidemiology, Faculty of Medicine, Memorial University of NL, St. John's holly.etchegary@mun.ca

Article	Ethics Approval	Population and sample size	Type of sample	Test performed	Linkability/Library/ Availability	Contact
				of the people will likely be included* Unclear if this has actually been established.		
MethMotif Database						
Dyer, Lin, Shapoval, et al. (2024)[129]	Not available	Not available	Not available	MethMotif is a database publicly available database that provides a comprehensive repository of transcription factor (TF)-binding profiles, enriched with DNA methylation patterns. It includes multiple species. Incorporates a vast array of ChIP-seq and Whole Genome Bisulfite sequencing (WGBS) or Enzymatic Methyl Sequencing (EM-seq) datasets from diverse sources Release Notes At least 1 set of Human NL samples is available in the database: GEO Accession viewer	Not available	Touati Benoukraf Division of BioMedical Sciences, Faculty of Medicine, Memorial University of NL, St. John's tbenoukraf@mun.ca

AB-Alberta; AS-Ankylosing Spondylitis; BBS1-Bardet-Biedl Syndrome; CA-Cancer; CASP-Collaborative Association Study of Psoriasis; CIPARS- Canadian Integrated Program for Antimicrobial Resistance Surveillance; CHU Dumont-Centre-Hospitalier Universitaire Dr. Georges L. Dumont; CODING study-Complex Disease in the Newfoundland Population; Environment and Genetics; dbGAP-Database of Genotypes and Phenotypes; DEG-differentially expressed genes; GWAS-Genome-Wide Association Study; HBOC-Hereditary Breast and Ovarian Cancer Study; HL-Hearing loss; IWK-Izaak Walton Killam Health; JANL-HIP-Janssen and Newfoundland and Labrador Health Innovation Partnership; MGB1-Mammaglobin1; MGB2-Mammaglobin2; MMGS-Maritime Medical Genetics Service; MUN-Memorial University of Newfoundland; NB-New Brunswick; NCBI-National Center for Biotechnical Information; NFOAS-Newfoundland Osteoarthritis study; NGS-Next generation sequencing; NL-Newfoundland; NLGP-Newfoundland Genome Project; NS-Nova Scotia; OA-Osteoarthritis; ON-Ontario; PCR-Polymerase chain reaction; PDAC-Pancreatic Ductal Adenocarcinoma; PEI-Prince Edward Island; PsA-Psoriatic arthritis; PsC-Cutaneous psoriasis; REB-Research ethics board; RT-PCR-Reverse transcription polymerase chain reaction; RNA-Ribonucleic Acid; SHARE-Secure Health Access Record; SMA-Spinal muscular atrophy; SMN1-Survival motor neuron 1; SLN-Sentinel lymph ntode; SNP-Single-nucleotide polymorphism; SNV-Single-nucleotide variant; STGD1-Stargart disease; TNF-Tumor Necrosis Factor; UHN-University Health Network; UofT-University of Toronto; WES-Whole exome sequencing; WGS-Whole genome sequencing

Appendix 3: Researchers and Organizations Searched for Relevant Genomics Studies

Please note that this appendix has not been copy edited.

Table 6: Researchers Who Were Searched for Genomic Publications

Province	Genomics related researchers/Institutions
Newfoundland	**Researchers:** 1. Dr. Kathy Hodgkinson (Ph.D.): Memorial University 2. Dr. Darren O'Rielly (Ph.D., FCCMG. Memorial University Molecular Genetics Laboratory (Director), Eastern Health 3. Dr. Proton Rahman (MD., FRCPC.): Memorial University 4. Dr. Neetu Singh (Ph.D.): Marathon of Hope - Atlantic Cancer Consortium 5. Dr. Curtis French (Ph.D.): Memorial University 6. Dr. Touati Benoukraf (Ph.D.): Memorial University, Bioinformatics for Personalized Medicine (Canada Research Chair) 7. Dr. Sevtap Savas (Ph.D.): Memorial University, Beatrice Hunter Cancer Research Institute 8. Dr. Michael Woods (Ph.D.): Memorial University, National Board of Directors of the Canadian Cancer Society 9. Dr. Terry-Lynn Young (Ph.D.): Memorial University, Craig L. Dobbin Genetic Research Centre (Director, Genomics Research), Laboratory Medicine Program, Eastern Health (Medical Scientist) 10. Dr. Guangju Zhai (Ph.D.): Memorial University 11. Dr. Gerald Mugford (Ph.D): Memorial University 12. Dr. Dennis O'Keefe: The NL Genome Project (Co-Principal Investigator) **Institutions:** 1. Provincial Medical Genetics Program 2. Central Newfoundland Satellite Genetics Centre
Nova Scotia	**Researchers:** 1. Lynette Penney (MD.): Dalhousie University, IWK Health (Division Head of Medical Genetics) 2. Steve Armstrong (Ph.D): Genome Atlantic (President/CEO) 3. Kristin Tweel (Ph.D): Genome Atlantic (Director, Sector Innovation) 4. Britta Fiander (Peng): Genome Atlantic (Director, Innovation Programs) 5. Dr. Victor Martinez (Ph.D): Dalhousie University, IWK Health (Clinical Genomics Specialist) 6. Dr. Karen Bedard (Ph.D): Dalhousie University 7. Dr. Jo-Ann Brock (MD.): IWK Health (Medical geneticist, Division Head, Pathology and Laboratory Medicine) 8. Dr. Sarah Dyack (MD.): IWK Health (Medical geneticist, Division Head, Medical Genetics): Dalhousie University 9. Dr. Anthony Vandersteen (MD. Ph.D FRCP. FRCPC. FCCMG.): IWK Health (Medical Geneticist) 10. Dr. Robin Urquhart (PH.D.): Dalhousie University, Population Cancer Research (Endowed Chair), Atlantic PATH (Scientific Director) 11. Dr. Jason Hicks (M.Sc.): Atlantic PATH (Executive Director) 12. Dr. Ellen Sweeney (Ph.D.): Atlantic PATH (Research Director), Dalhousie University 13. Yunsong Cui (M.Sc.): Atlantic PATH (Data Analyst) 14. Dr. Luke Chen (MD., FRCPC., MMEd.): Dalhousie University

Province	Genomics related researchers/Institutions
	15. Dr. Dan Gaston (PhD.): Dalhousie University 16. Dr. Manuel Mattheisen (MD.): Dalhousie University 17. Dr. Christopher McMaster (Ph.D): Dalhousie University, CIHR Institute of Genetics 18. Dr. Johane Robitaille (MDCM.): Dalhousie University **Institutions:** 1. Maritime Medical genetics 2. Genome Atlantic
New Brunswick	**Researchers:** 1. Dr. Rodney J. Ouellette (MD.): Atlantic Canada Research Institute (Founder and Senior Scientist), CHU Dumont (Laboratory Medical Head-Molecular Genetics) 2. Dr. Gilles Robichaud (Ph.D): Université de Moncton, BHCRI (Senior Scientist), Atlantic Canada Research Institute (Adjunct Scientist) 3. Dr. Mouna Ben Amor (MD.): Clinical Genetics Vitalité Health Network, Université de Moncton 4. Dr. Eric Allain (Ph.D.): Université de Moncton, Vitalité Health Network, Atlantic Cancer Research Institute, BHCRI (Associate member) 5. Dr. Nicolas Crapoulet (Ph.D.): Vitalité Health Network (Clinic Specialist in Molecular Genetics) 6. Dr. Dominique Comeau (Ph.D.): Vitalité Health Network 7. Dr. Nadia Bouhamdani (Ph.D.): Vitalité Health Network (Clinical Specialist and Researcher)
Prince Edward Island	1. Dr. Denise Lockhart: PEI Cancer Treatment Centre (Manager)

Appendix 4: Literature Review Tool

Please note that this appendix has not been copy edited.

Items from the form completed for titles retrieved following bibliography scans of researchers/organizations listed in Appendix 3 include the following:

- author
- paper information
- province
  - included (yes/no)
- if included, extracted (yes/no)
- comments.

Appendix 5: Contact Email Template

Please note that this appendix has not been copy edited.

Dear (Name of researcher),

Greetings from NB IRDT. If you are not familiar with our organization, we have included a brief description of our mandate and role.

We are currently working on a project in collaboration with Canada's Drug Agency (CDA) to assemble and document the characteristics and specifications of genomic datasets that have been collected in the Atlantic Provinces. A summary report and asset map will be produced that will provide a foundation for subsequent data ecosystem development in Atlantic Canada, and support CDA's work on the Canadian Rare Disease Strategy.

We learned about (*A brief details of what they are working on*)

We are particularly looking for some specific details about the database. They are given as follows:

- Characteristics of the dataset (sample size, relevant population, genomic information typed, etc)
- Linkability with administrative data (consent to link, identifiers for linkage, technical requirements)
- Access protocols and requirements – who can access the data, how is data access made available, what form of data are made available
- Data ontologies, labelling, and use of common data models, if applicable
- Federated analysis options and linkages with other repositories of genomic data elsewhere in the region, country or internationally, if available.

We would welcome an opportunity to engage with you to assemble this information. This can be via a written questionnaire or interview or both, but at this stage we are hoping you could suggest who would be the best person to connect with for follow-up.

Thank you in advance for your cooperation, and please feel free to contact us, if you have any questions or concerns.

Appendix 6: Interview Questions

Please note that this appendix has not been copy edited.

Genomic Data Holdings in the Atlantic Provinces - Survey

Information and Consent Form

The New Brunswick Institute for Research, Data and Training (NB-IRDT) is conducting a study to assemble and document the characteristics and specifications of genomic datasets that have been collected in the Atlantic Provinces. We are conducting survey/interview with Key organisations that have either collected or worked with genomic dataset. A summary report and asset map will be produced that will provide a foundation for subsequent data ecosystem development in Atlantic Canada, and support CDA-AMC's mandate more broadly. The survey is being conducted by the New Brunswick Institute for Research, Data and Training (NB-IRDT), at the University of New Brunswick, on behalf of the Canada's Drug Agency (CDA-AMC).

Invitation to Participate

This is an invitation to participate in the survey. It includes questions related to data characteristics, data usage and access, linkability to administrative data, federated analysis options and linkages with other repositories of genomic data elsewhere in the region, country or internationally.

Your individual responses will be summarized, and a collated information will be presented in the final report. For any questions or concerns about your participation, please contact us at nb-irdt@unb.ca.

By clicking on "Agree" below, you are consenting to participate in the survey:

___ ***Agree*** I have read and understood all the statements above and agree to participate in this survey.

Questions

Source of Data

1. Does your organization collect genetic data itself?
2. In what ways are you involved with the collection of genomic data - e.g., for clinical practice, as the testing facility, for research, etc.
3. Are the genetic datasets collected for people in Atlantic Canada?

   Yes

   No, data collection is not limited to people in the region
4. If '**No**' for which provinces or regions outside of Atlantic Canada are the data collected?
5. Is the primary purpose of the original data collection for clinical practice or research? Both?
6. Can you trace the pathway of data processing from collection to data sequencing, to disclosure of results: (If a separate process exists for clinical and research data, please explain both)?

   Research purposes:

   Clinical purposes:
7. Are consents to secondary use of information included in the data collection process? *If not answered above*
8. Does the data collection process need ethics approval? *If not answered above*
9. If ethics approval is required, what is the process?
10. Are the data also contained in or combined with other genomic datasets or databases?
11. If 'yes', which database?
12. If 'no', would they be able to be part of any genome libraries or databases? (for example, the Pan-Canadian Genome Library)?

Data Characteristics

13. What type of genetic information is collected? Whole genome? Whole exome? Specific genes or mutations?

14. How many unique individuals are in your data collection?' (if there are multiple distinct data collections, please report for each)
15. Are the datasets collected for any specific age group or cohorts or from populations that have a higher rate of rare diseases? (Acadians, Newfoundlanders, etc)
16. What is included in the data holding? (for example, physical measures or blood samples, patient chart information)

Data Access and Privacy

17. Are the datasets accessible by anyone outside of your organization (e.g., clinicians, researchers, health authorities)

    Yes

    No
18. If '**yes**', what is the process they follow?
19. Are there additional requirements for sharing the data across organizations versus borders?
20. Who is the custodian of the data and who has the authority to approve access and use?
21. Besides requiring ethic's board approval and participant consent, what measures are used to protect the people who have information in the datasets?
22. Does the data include Indigenous people or have any indicators of Indigenous status?
23. If yes, what safeguards, protection and governance are in place to reflect indigenous data sovereignty?

Data Linkage

24. Is the data linkable with administrative data?
25. Would any additional consents be required to link with administrative data?
26. Are there any privacy concerns with linkage to administrative data?
27. How would the identifiers for linkage work?
28. Who does the linkage?
29. Is it linkable with other genomic data?
30. If yes, are the genomic data going out for linkage to other data or are other data coming into the location of the genomic data for linkage?

If '**worked on genome data collected by another organization**', please answer the following questions:

31. Which organization collected the data?
32. How did you access the data (requirements of accessing the data)
33. How did you use the data?
34. What was the sample size of the dataset you worked on?
35. Is it for any specific age group or cohorts (specific genomes or rare diseases)?

36. Are there any other organizations working with you on the dataset?

    Yes

    No

37. If there are other(s) in your organization who you feel would be better suited to answer some of these questions, could you provide us with their name(s) and contact details?

Appendix 7: Flow of Genomic Data in Clinical and Research Settings

Please note that this appendix has not been copy edited.

Figure 1: Flow of Genomic Data Collected for The Atlantic Cancer Consortium (Research Based)

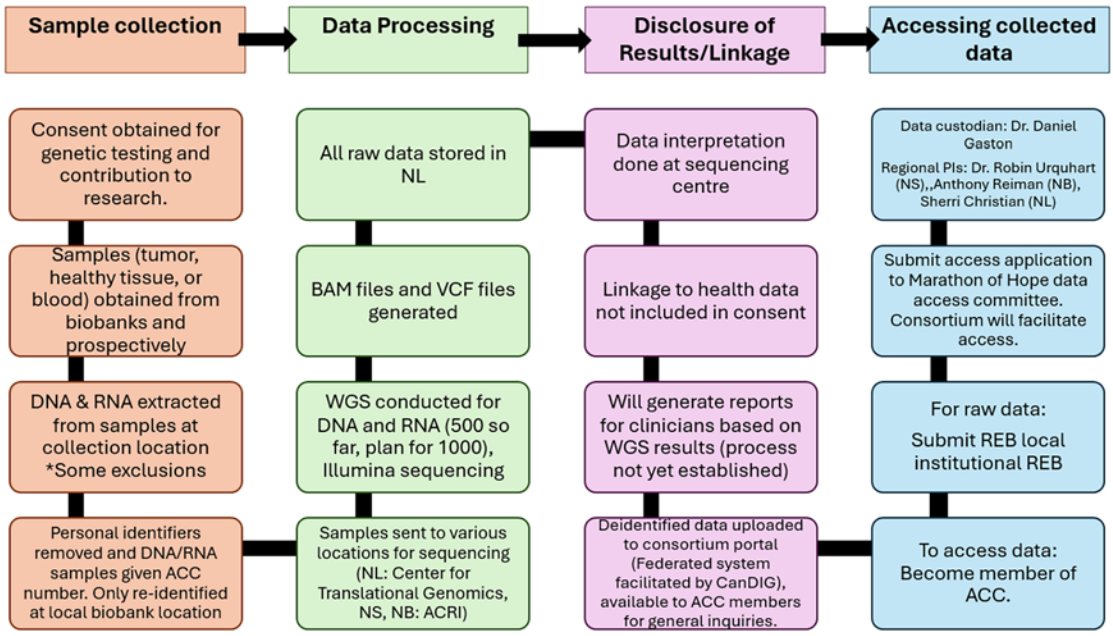


Figure 2: Atlantic PATH — Flow of Genomic Data Collected for Atlantic PATH Dataset

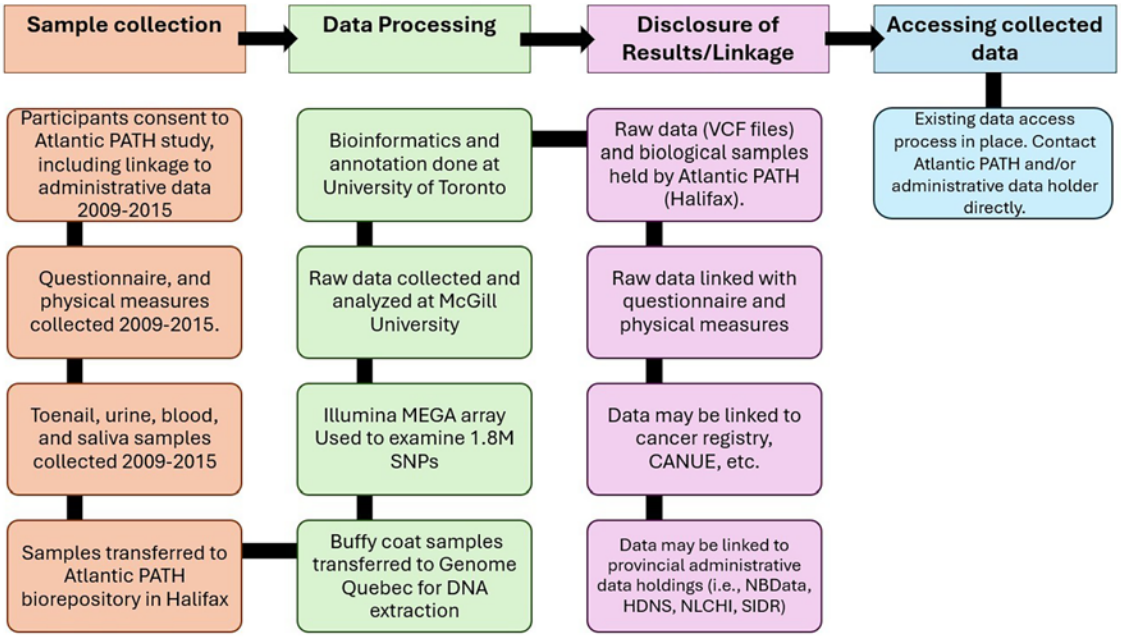


Figure 3: New Brunswick, Vitalité Health Network — Flow of Genomic Data Collected in Clinical Setting

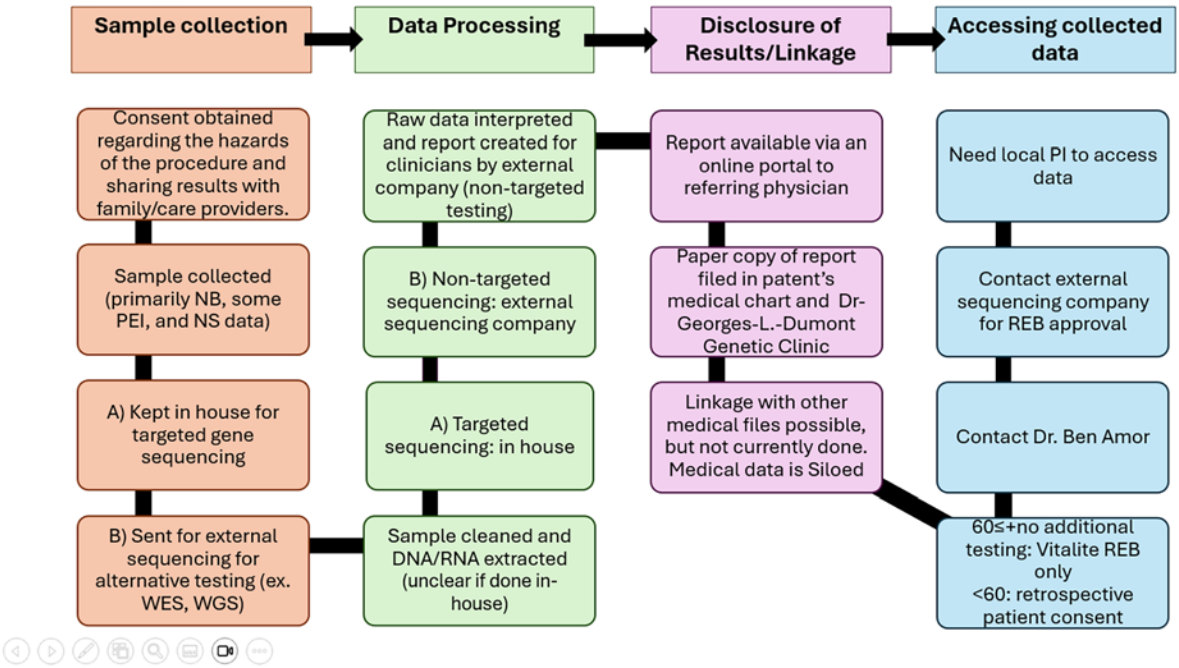


Figure 4: New Brunswick, Vitalité Health Network — Flow of Genomic Data Collected for Carrier Screening Project

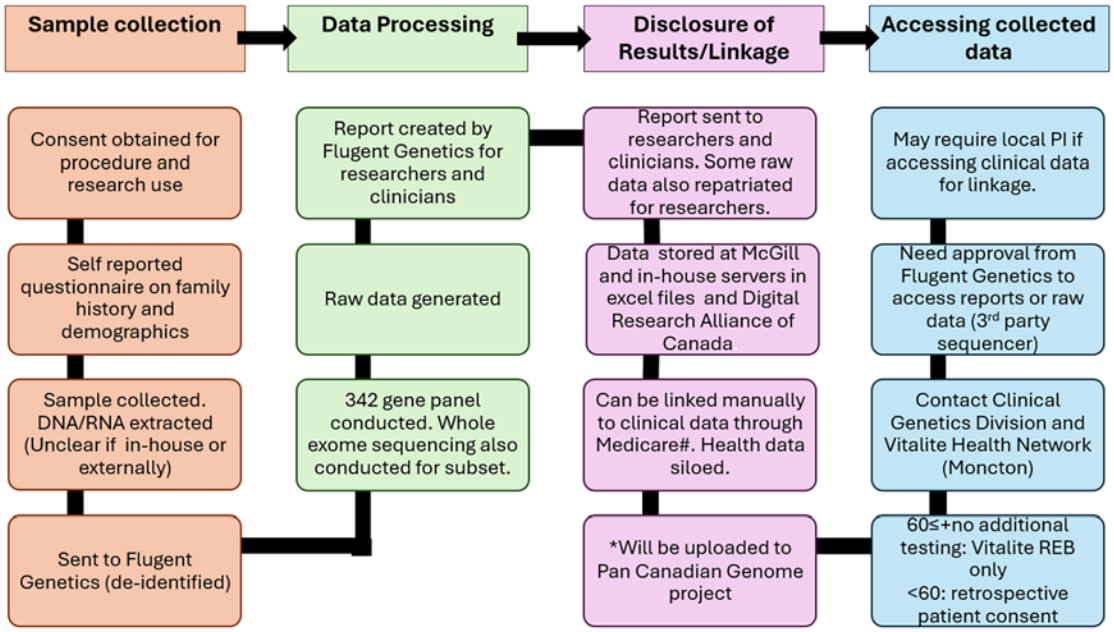


Figure 5: New Brunswick, Horizon Health Network — Flow of Genomic Data Collected in Clinical Setting

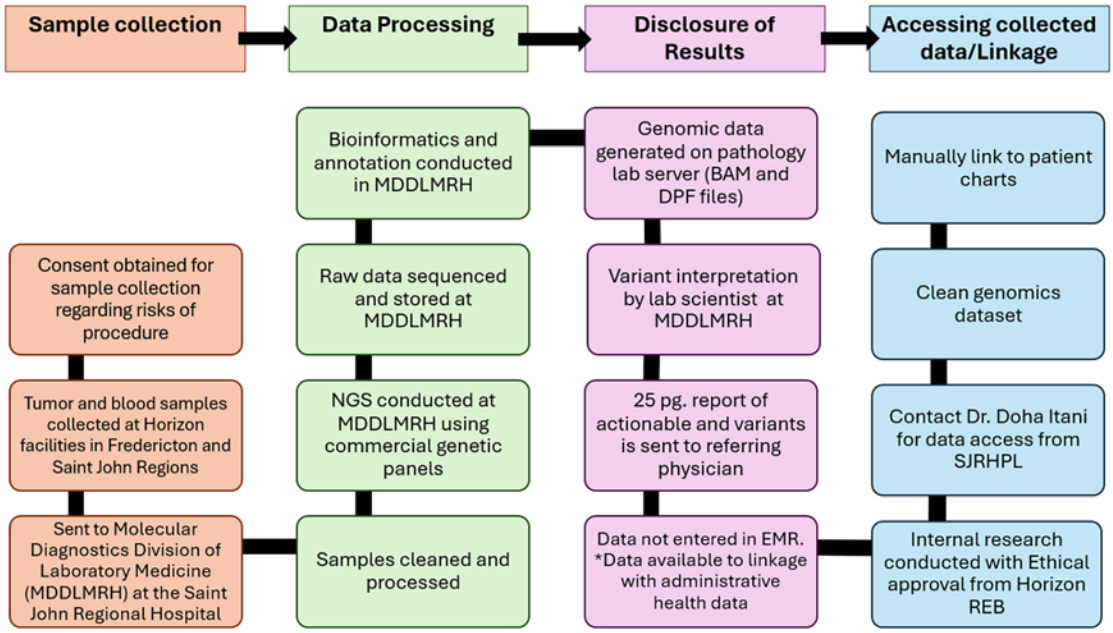


Figure 6: Nova Scotia, IWK Health — Flow of Genomic Data Collected in Clinical Setting

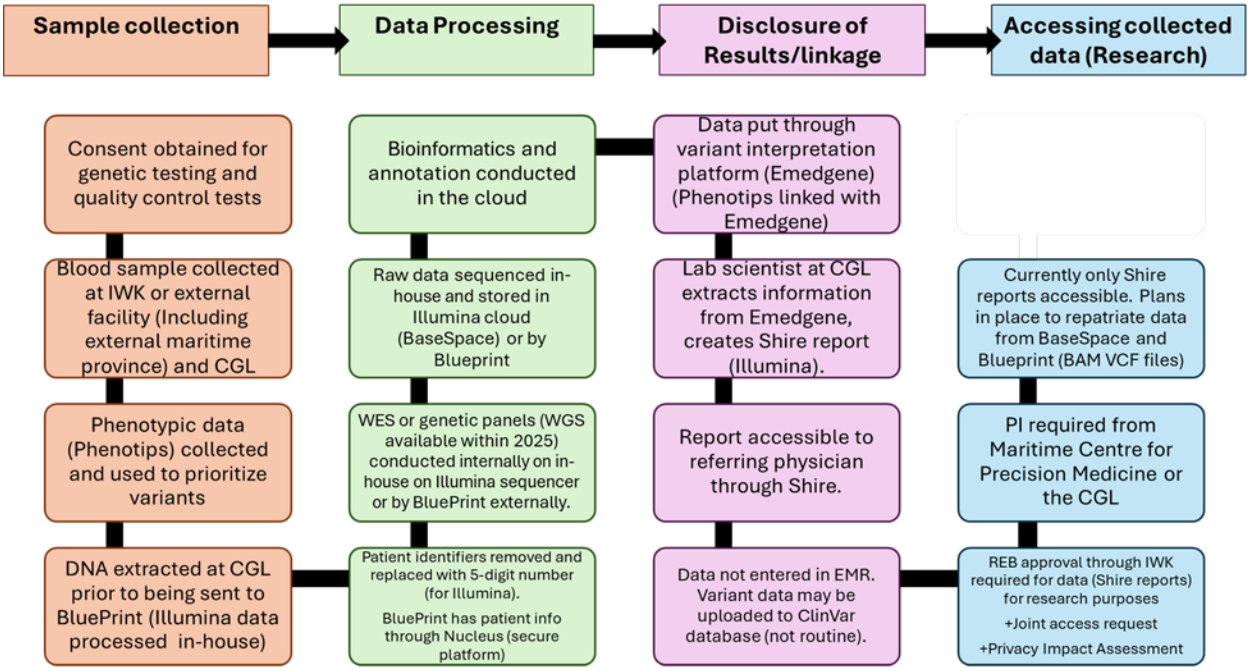


Figure 7: Nova Scotia Health Authority — Flow of Genomic Data Collected in Clinical Setting With Somatic Data

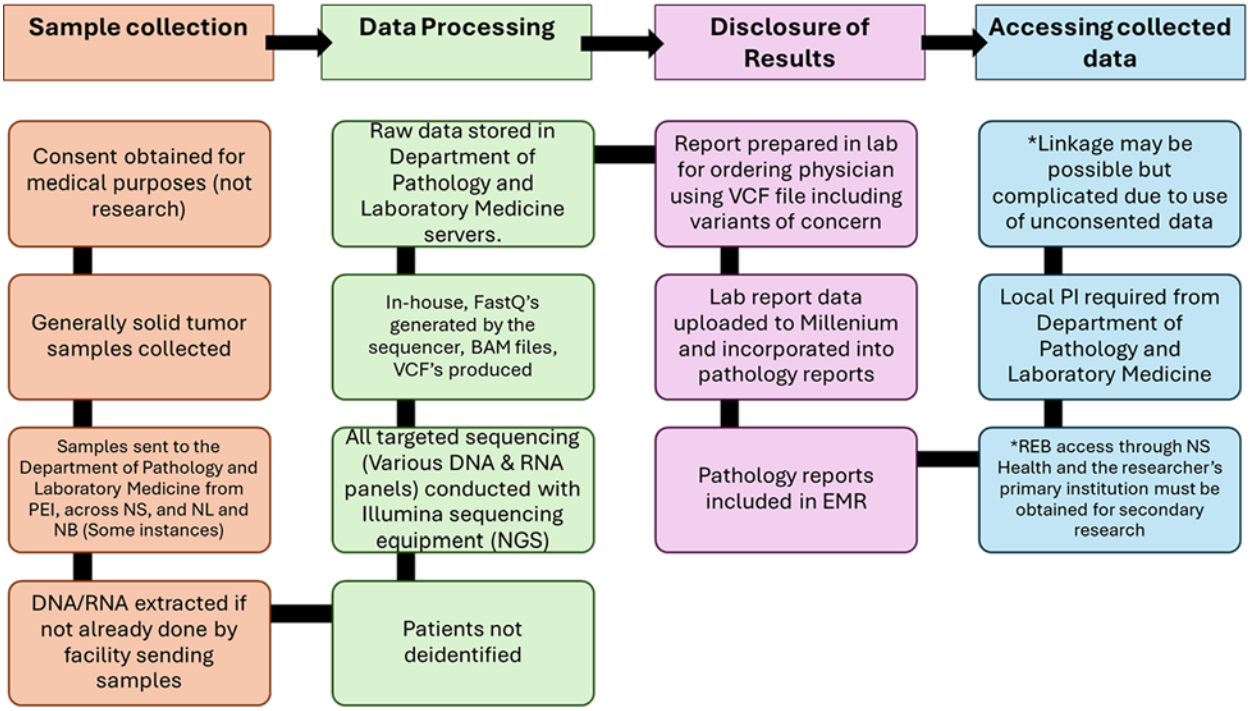


Figure 8: Newfoundland and Labrador — Flow of Genomic Data Collected in Clinical Setting

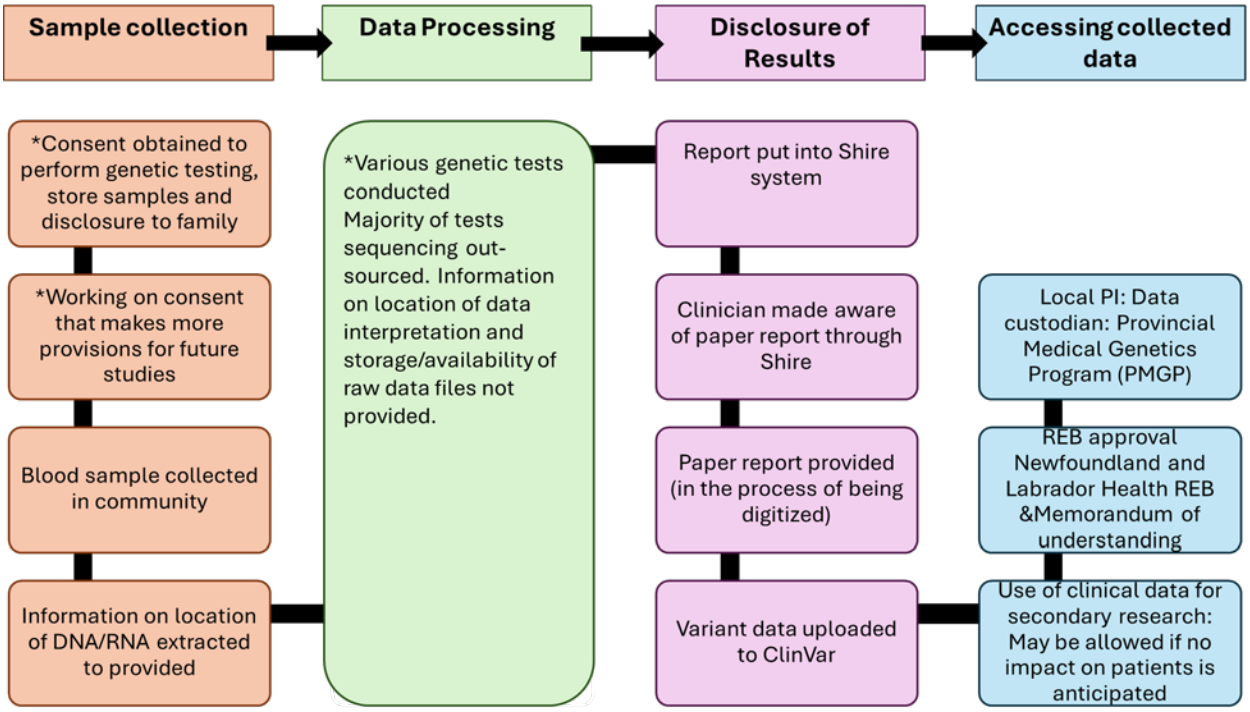


Figure 9: Newfoundland and Labrador — Flow of Genomic Data Collected for Research

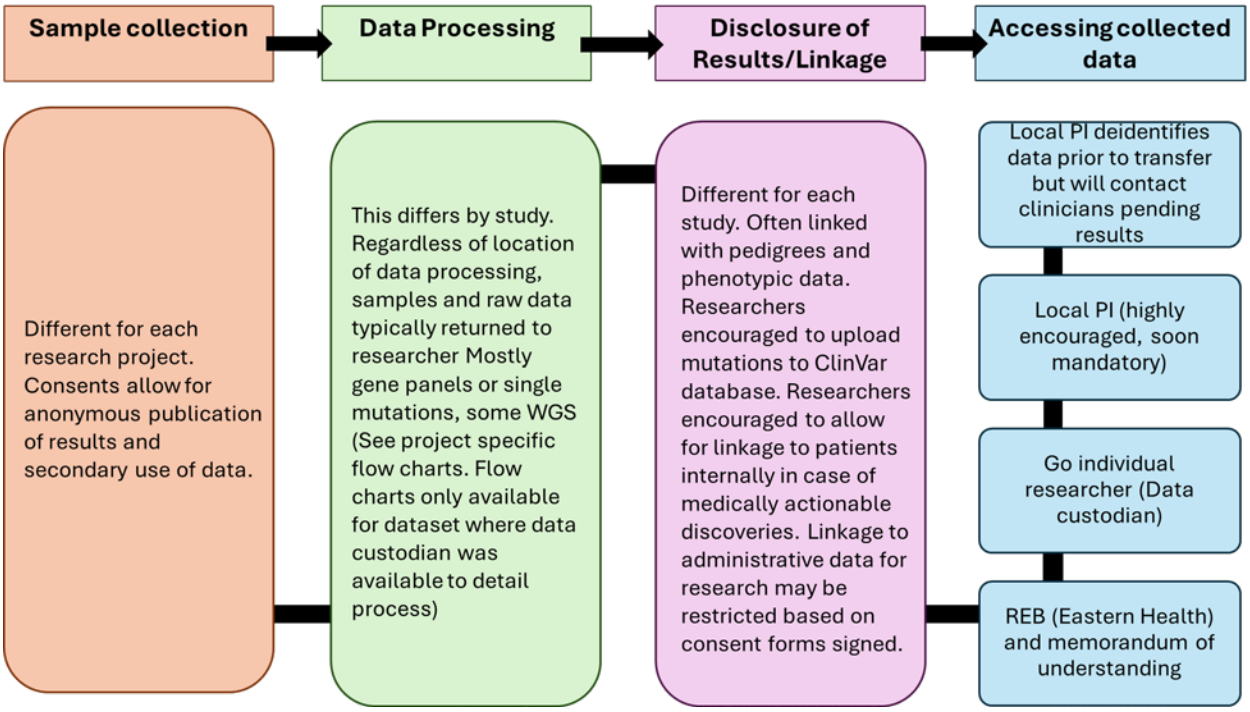


Figure 10: Newfoundland and Labrador — Flow of Genomic Data Collected for Psoriasis Cohort Research (Dr. Gulliver)

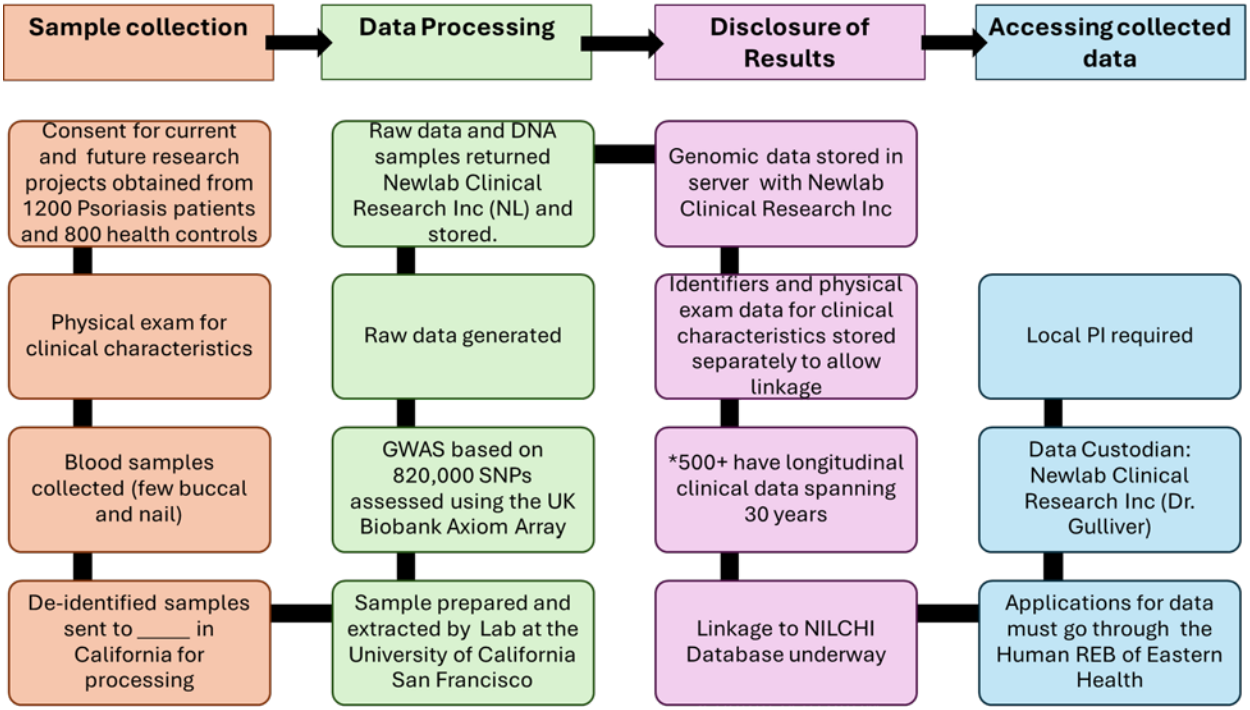
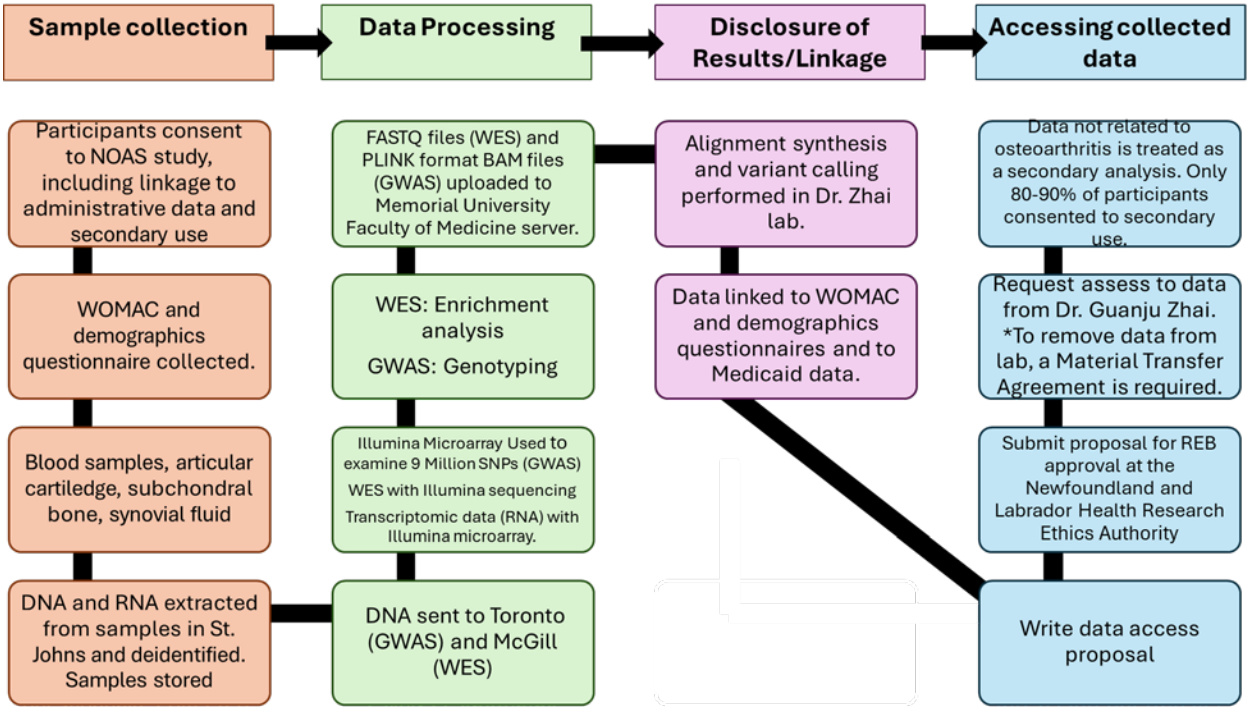


Figure 11: Flow of Genomic Data Collected in Newfoundland Osteoarthritis (Research)



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