NEWS FROM CARTaGENE

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December 2023 Newsletter - Volume 13

Happy Holidays! Happy New Year 2024!

In 2024, CARTaGENE will enter its 15th year of operation!

We couldn't have reached this milestone without the involvement of our participants in generating new data, and without the contribution of the researchers who use this data to advance health research. At this auspicious time of year, the CARTaGENE team would like to wish you all the best for the holiday season, and a happy new year in 2024. May it be synonymous with discoveries and success for all health research projects!

Sequencing News

Since April 2023, genome-wide sequencing data on 2184 CARTaGENE participants are available to researchers. These data were produced as part of the GenoRefQ project funded by Génome Québec and Genome Canada. Genotyped participants were selected to represent the genetic diversity of Quebec.

Part of the selection involved Haitians and Moroccans (n=287). The objectives of the GenoRefQ project include the creation of a comprehensive catalog of genetic variants present in the Quebec population, a valuable tool for clinical geneticists and other clinicians who will be able to interpret genetic test results and determine the cause of various genetic diseases.

Sequencing data has already been shared in 16 research projects and, counting researchers and collaborators, more than 50 research groups have had access to this data.

Génome Québec and CARTaGENE are jointly organizing a public webinar on the progress of analyses of sequencing and other genetic data as part of the Genomics in the CARTaGENE cohort project (see next section). **This webinar will be held on January 22, 2024 from 11am to 1pm EST.** The entire scientific community is invited.

Click here to sign up for the webinar

Major Development Projects

The scientific directors are currently leading two projects to promote CARTaGENE in the scientific community.

- **Genomics in the CARTaGENE cohort**: This project aims to highlight the usefulness of genetic data in CARTaGENE by carrying out a wide range of analyses that will be published in a high-impact scientific journal. These efforts aim to enhance the CARTaGENE dataset, increase its visibility and foster future collaborations. The manuscript will provide researchers in Canada and around the world with updates on the latest genetic and phenotypic data available from CARTaGENE. It will present the potential value of these data for genetic and population health research, using multiple examples of applications in population genetics, clinical variant interpretation, genome-wide and phenome-wide association studies (GWAS/PheWAS) and polygenic risk scores (PRS). In terms of genetic data, the main analyses will use newly generated whole genome sequencing (WGS) data and the most recent version of genotypic data imputed from the TOPMed reference panel. The project is a collaborative effort by several research groups with extensive experience in population genetics and health, who are actively using the CARTaGENE biobank in their research programs.

- **Using data from the CARTaGENE cohort - from human genetics to environmental health**: This project aims to provide a baseline description of the CARTaGENE study by estimating the distribution of both environmental and personal factors, as well as the incidence and prevalence of health outcomes, particularly for chronic diseases. Furthermore, it aims to uncover meaningful associations between environmental factors and a spectrum of health outcomes, delving into the complex interplay between genetic and environmental influences on diverse health conditions.

Linked Databases

Data from several external databases were linked to those collected from CARTaGENE participants. The following data sources are used by several researchers. Will you be next?

- **BALSAC**

  The BALSAC database (Université du Québec à Chicoutimi) is based on Quebec civil registry records. These records have been computerized and linked together using a methodology for matching nominative data, enabling the automatic reconstitution of families and genealogical lines over four centuries. BALSAC has supported innovative, multidisciplinary studies in fields as diverse as genetic epidemiology and historical geography, as well as population genetics, evolutionary biology, demography and social history. Some 9,000 CARTaGENE participants are linked to the BALSAC file via their genealogical data. In addition, an upcoming research infrastructure, the Généo portal, will soon be online. The Généo portal will enable researchers to visualize and analyze genealogical, genetic and geographical data for innovative studies of the Quebec population. The Généo portal is based on a collaboration between BALSAC, the Centre interuniversitaire d'études québécoises (CIEQ) and the CARTaGENE platform.

- **CANUE**

  As part of the CANPATH consortium, CARTaGENE participants were linked to CANUE, the Canadian Urban Environmental Health Research Consortium, to gain access to a wide range of environmental data, including measures of air quality, green spaces, noise levels, and built environment, to facilitate research on the health impacts of urban living.

- **Administrative Health Data**

  CARTaGENE participants were linked to various administrative health databases in Quebec, which provide access to information regarding hospitalization history, clinical diagnoses and interventions, prescription drug use, and vital status. In addition, information on cancer is also available through the linkage to the Quebec cancer registry.

Critical Data for Research

The CARTaGENE cohort includes 43,000 Quebecers from six regions of the province, aged 40 to 69 at recruitment. The data collected on participants is broken down into several datasets that can be used for multiple research projects in epidemiology, environmental health, genetics and more. Are you already benefiting from the richness of these datasets?

Baseline questionnaires

Participant recruitment and initial data collection were carried out in two phases: phase A in 2009-2010 and phase B in 2012-2014. The questionnaires collected information on socio-demographic factors, lifestyle habits, personal and family history of illness, medication use, use of health services, female and male reproductive health, mental health and psychosocial environment. A cognitive assessment was also performed for phase A participants.

Collection years: 2009-2014

Number of participants: 43,000

Number of variables: 2500

Physical measurements

Physical measurements were taken on Phase A participants, including anthropometric measurements, blood pressure, grip strength, bone density, bioimpedance, pulmonary function, arterial stiffness, and a resting electrocardiogram. (Phase B participants, as well as health follow-up participants, self-reported their anthropometric measurements).

Collection years: 2009-2010

Number of participants: 20,000

Number of variables: 400

Environment

Phase A participants were invited to fill out a residential and occupational history questionnaire.

Collection years: 2011-2012

Number of participants: 11,500

Number of variables: 1,300

Nutrition

Along with the environment questionnaire, phase A participants were also asked to complete a questionnaire on food frequency consumption.

Collection years: 2011-2012

Number of participants: 10,000

Number of variables: 700

Health follow-up

In this first follow-up of the entire cohort, information was again collected on sociodemographic factors, lifestyle habits, personal and family history of illness, medication use, use of health services, female and male reproductive health, mental health and psychosocial environment.

Collection years: 2017-2018

Number of participants: 17,000

Number of variables: 1,300

COVID-19

During the COVID-19 pandemic, CARTaGENE participants were invited to take part in four data collections to provide information on their COVID test results, symptoms, post-COVID condition, hospitalization or medical care received, health status and risk factors, potential sources of exposure, and the impact of the pandemic on their employment status and emotional, social and financial well-being. A blood sample was also collected for serological analysis.

Collection years: 2020-2022

Number of participants: approx. 8,000 participants in one or more of the collection phases

Number of variables: 3,000

Genotyping

Genotyping was carried out in waves starting in 2012, using different chips. All participants who provided a biological sample are now genotyped with Illumina's GSA chip. This work was carried out from 2017 to 2021. Imputation data has also been available since spring 2022. Details of the methodology and quality control are available on our website.

Collection years: 2017-2021, 2022 (imputation)

Number of participants: 30,000

Number of variables: approx. 650,000 variants on the GSA chip (imputed data available)

Sequencing

Whole genome sequencing data on 2184 CARTaGENE participants are available for sharing. These data were produced as part of the GenoRefQ project. Genotyped participants were selected to represent the genetic diversity of Quebec. Part of the selection involved Haitians and Moroccans (n=287).

Collection years: 2022-2023

Number of participants: 2,180

Number of variables: 3 billion base pairs.

Collaborations

CARTaGENE collaborates with several partners to enrich its databases and provide new opportunities for collaborative research projects. The harmonization of CARTaGENE data with those of other Quebec and Canadian cohorts, and their co-analyses, provides a larger sample size which allows to investigate rare health issues, as well as to explore interactions between several risk factors. These collaborations are also inducive to comparative research between different provinces and populations in Canada.

Canadian Partnership for Tomorrow's Health (CanPath)

CanPath is a pan-Canadian cohort consortium that constitutes the largest population health cohort in Canada. CanPath's objective is to create a harmonized research platform facilitating epidemiological research in Canada, in order to investigate the causes of chronic diseases including cancer. To achieve this, the consortium brings together six cohorts representing different provinces (CARTaGENE for Quebec) for a total of 330,000 participants. Through the participating cohorts, CanPath collected data on the health, lifestyle, behaviours and environment of Canadians. A significant number of participants also provided physical measurements and biological samples and consented to the matching of their data with various administrative health and environmental databases. The size of the cohort and the richness of its epidemiological, clinical and biological data position Canada among the world leaders in longitudinal research on cancer and other chronic diseases.

Public Health Agency of Canada's COVID-19 Immunity Task Force (CITF)

The mandate of the CITF is to support and harmonize knowledge on immunity to COVID-19 in order to assist federal, provincial and territorial decision-makers in their efforts to protect Canadians and limit the impact of the pandemic. The working group also supports laboratory studies aimed at establishing the advantages and limitations of immunity tests and associated technologies, with a view to generating knowledge on optimal methods of using and interpreting these tests. More than 70 cohorts across Canada contributed to CITF by collecting data throughout the pandemic. CARTaGENE participants were invited to take part in four data collections, including the collection of a blood sample, from the start of the pandemic in spring 2020 until the end of the global health emergency in 2022.

Centre québécois de données génomiques (CQDG)

The Centre québécois de données génomiques is a platform for harmonizing and disseminating genomic data generated by clinical and research studies in Quebec. Researchers can explore, visualize, and analyze genomic data online through the CQDG. CARTaGENE contributes to this initiative by sharing its

participants' genotyping and sequencing data on this platform. In this way, CARTaGENE's genetic data is documented according to this Quebec standard, making it much easier for researchers interested to explore. The high-performance computing environment developed by the CQDG will also support analyses on these datasets.

CARTaGENE in Numbers

Data used by 234 research groups in Quebec, Canada and around the world!

15 years of research advances that will have a major impact on health!

144 articles published in scientific journals!

59 theses and dissertations!

Survey

Help us improve CARTaGENE by answering this short survey about your user experience.

Click here to fill out the survey

Theses and Dissertations 2023

CARTaGENE data is used by many students as part of their master's or doctoral work. CARTaGENE is proud to participate in student training. Has your student published a thesis or dissertation that includes CARTaGENE data? Please let the CARTaGENE team know.

Fourteen theses and dissertations published this year involve analysis of CARTaGENE data.

Université Laval

Under the supervision of **Jean-Philippe Drouin-Chartier**, Faculty of pharmacy.

- **Amélie Bélanger**, Msc. These titled: Relation entre la qualité de l'alimentation et l'utilisation des statines chez les adultes de la cohorte CARTaGENE ayant un syndrome métabolique.
- **Clémence Desjardins**, MSc. Thesis titled: Étude de la relation entre la qualité de l'alimentation et l'intensité de la pharmacothérapie antidiabétique dans le traitement du diabète de type 2
- **Laurie Dessureault**, MSc. Thesis titled: Habitudes de vie et santé cardiovasculaire dans l'hypercholestérolémie familiale hétérozygote

McGill University

Under the supervision of **Patricia Tonin**, Human Genetics Department.

- **Wejdan Alenezi**, PhD. Dissertation titled: The genetic analyses of DNA repair pathway genes in French Canadians of Quebec identified new candidate risk variants implicated in hereditary ovarian cancer
- **Caitlin Fierheller**, PhD. Dissertation titled: A molecular genetic investigation of FANCI as a new candidate ovarian cancer predisposing gene

Université de Montréal

Under the supervision of **Vikki Ho**, École de Santé publique de l'Université de Montréal.

- **Alice Nguyen**, MSc. Thesis titled: DNA methylation of F2RL3 and AHRR and lung cancer risk
- **Michael Pham**, MSc. Thesis titled: The influence of smoking and occupational exposures on DNA methylation in the AHRR and F2RL3 genes

Under the supervision of **Julie Hussin**, Department of Biochemistry and Molecular Medicine, Faculty of Medicine.

- **Justin Pelletier**, MSc. Thesis titled: Évaluation de l'imputation des données génétiques Canadiennes-Françaises

Under the supervision of **Sébastien Jacquemont**, Department of Biochemistry and Molecular Medicine, Faculty of Medicine.

- **Catherine Proulx**, MSc. Thesis titled: Effet du dosage génique sur les phénotypes cognitifs et psychiatriques

Under the supervision of **Martine Tétreault**, Department of Biochemistry and Molecular Medicine, Faculty of Medicine.

- **Marjorie Labrecque**, MSc. Thesis titled: Caractérisation de variants génétiques pour estimer la prévalence de Niemann-Pick type C au Québec

Under the supervision of **Éric Robitaille**, École de Santé publique de l'Université de Montréal.

- **Sirpal Sanjeev**, MSc. Thesis titled: Association entre l'accessibilité géographique aux commerces d'alimentation et le diabète type II chez les adultes québécois : analyse de la banque de données de santé CARTaGENE.

University of Toronto

Under the supervision of **Philippe Awadalla**, Molecular Genetics Department.

- **Elyssa Bader**, PhD. Dissertation titled: Genomic Architecture of the Aging Hematopoietic System
- **Kimberly Skead**, PhD. Dissertation titled: Predicting Health Outcomes from Hematopoietic Evolution
- **Michelle Harwood**, PhD. Dissertation titled: Genetic and Epigenetic Factors Influencing Population Variability of Allele-Specific Expression

List of publications 2023 by our researchers

A. Sen, A.S. Brazeau, S. Deschenes, H.R. Melgar-Quiñonez, and N. Schmitz. The role of ultra-processed food consumption and depression on type 2 diabetes incidence: a prospective community study in Quebec, Canada. Public Health Nutr. 26(11): 2294-2303.

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A. Bélanger, C. Desjardins, L. Leblay, M. Filiatrault, O. Barbier, et al. Relationship between diet quality and statin use among adults with metabolic syndrome from the CARTaGENE cohort. CJC Open.

C. Desjardins, L. Leblay, A. Bélanger, M. Filiatrault, O. Barbier, et al. Relationship between diet quality and glucose-lowering medication intensity among adults with type 2 diabetes: results from the CARTaGENE cohort. CJC Open.

H.-M. Vasiliadis, J. Spagnolo, M.-J. Fleury, J.-P. Gouin, P. Roberge, et al. Factors associated with mental health service use during the pandemic: Initiation and barriers. Int J Soc Psychiatry.

L.-C. Desbiens, A.-C. Nadeau-Fredette, F. Madore, M. Agharazii, R. Goupil. Impact of Successive Office Blood Pressure Measurements During a Single Visit on Cardiovascular Risk Prediction: Analysis of CARTaGENE. Hypertension. 80(10): 2209-2217.

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Y. Wu, S.B. Goleva, L.B. Breidenbach, M. Kim, S. MacGregor, et al. 150 risk variants for diverticular disease of intestine prioritize cell types and enable polygenic prediction of disease susceptibility, Cell Genom. 3(7): 100326.

D.E. O'Sullivan, T.W.R. Hillier, D.R. Brenner, C.E. Peters, W.D. King. Time spent in the sun and the risk of developing non-Hodgkin lymphoma: a Canadian cohort study, Cancer Causes Control. 34(9): 791-799.

M. Bahda, J. Ricard, S.L. Girard, M. Maziade, M. Isabelle, A. Bureau Multivariate extension of penalized regression on summary statistics to construct polygenic risk scores for correlated traits. HGG Adv. 4(3): 100209.

M. Drouin, S. Rochette, M. St-Louis, A. Lewin, J. Laganière Prevalence of weak D phenotypes in the general population of Québec, Canada: A focus on weak D type 42. Vox Sang. 118(7): 577-581.

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