



Happy Holidays



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Happy Holidays and Happy New Year 2022!

In 2022, CARTaGENE will begin its 13th year of activity!

We would not have been able to take this important step without the involvement of participants helping to generate new data and the contribution of researchers who use this data to advance health research.

In this auspicious period, the CARTaGENE team wishes you a wonderful holiday season and a happy new year 2022. May this new year be synonymous with discoveries and success for all health research projects!

Updates on whole cohort genotyping and sequencing

2021 was the year of genomics for CARTaGENE. Significant funding from Génome Québec made it possible to complete the genotyping of the entire CARTaGENE cohort in 2020-2021. The *Centre d'expertise et de services Génome Québec* performed genotyping using the Global Screening Array (GSA from Illumina) chip. This was adapted following a consultation held by CARTaGENE with geneticists in Quebec (including members of the *Réseau de médecine de génétique appliquée*). Customized content of variants suggested by geneticists in Quebec (approximately 4,500 variants) has been added to the microarray. The genotyping dataset forms the basis of the whole genome sequencing (WGS) project, GenoRef-Q, funded by Génome Québec / Génome Canada.

As part of this project, 2,184 genotyped participants were selected to represent the genetic diversity of Quebec. Part of the selection focused on people of Haitian and Moroccan descent (n = 287). This resource will promote multiple activities related to personalized medicine and set the stage for projects led by the new generation of scientists. The GenoRef-Q initiative will also create a complete catalog of genetic variants present in the population of Quebec, a valuable tool for clinical geneticists and other clinicians who will be able to interpret the results of genetic tests and determine the cause of various genetic diseases. WGS data will be stored at the *Centre Québécois de Données Génomiques* (CQDG) and available to researchers during 2022.

Whole-genome genotyping data (n = 30,000) are now available for researchers

CARTaGENE webinar series

Since the spring of 2021, CARTaGENE has been holding monthly webinars during which researchers present their projects based on CARTaGENE data. This series of conferences aims to 1) provide a showcase for researchers and their students, 2) identify potential collaborators and 3) promote the use of CARTaGENE data in all fields of research. Missed previous webinars? You can watch them here (in French):

<https://cartagene.qc.ca/fr/chercheurs/documents-et-infolettres#webinaires>

If you are interested in presenting, please do not hesitate to contact us at: access@cartagene.qc.ca.

English speakers are welcome!

CARTaGENE & COVID-19

CARTaGENE launched the second phase of the COVID-19 study at the start of 2021. In addition to completing a questionnaire, participants donated a blood spot sample used to detect the presence of antibodies specific to COVID-19, especially in certain populations at higher risk of infection (elderly, people who were born outside of Canada and those living in urban areas with high infection rates).

This study is funded by the Government of Canada, through the COVID-19 Immunity Task Force (CITF) and the Canadian Institutes of Health Research (CIHR). These two funds were granted to the Canadian Partnership for Tomorrow's Health (CanPath), of which CARTaGENE is part of.

Antibody rates that reflect the start of the vaccination campaign

Of the 8,000 participants who completed the questionnaire during the summer of 2020, more than 4,500 participants agreed to fill out a new questionnaire and donate at least one blood spot sample. The results showed that in April 2021 more than 1,400 participants had antibodies to COVID-19, which is explained by vaccination. Data from the spring 2021 questionnaires and serology are available to researchers.

A COVID-19 serology follow-up is to be expected in 2022

The COVID-19 study continues to collect data and blood spot samples, longitudinally over the next year. The goal is to gather information on the impact of the pandemic on psychological well-being, vaccination and immunity to COVID-19. Contact us for more information!

New blood samples

Good news! Thanks to the COVID-19 serology study, Whatman cards containing blood spot samples from more than 4,500 participants are stored at the G nome Qu bec biobank and are now available to researchers. More than 15% of CARTaGENE participants have now provided two blood samples, increasing significantly the number of participants that could be included in a longitudinal analysis with serological data. Additional blood spot samples will be collected in 2022 on these same individuals. For more information on these Whatman cards, please do not hesitate to contact us by email at: access@cartagene.qc.ca.

CanPath Student dataset now available

Students can access a synthetic dataset to learn how to develop a research question, analyze and interpret health data, and present their findings as part of their lessons.

Known as the CanPath Student Dataset, this simulates the nationally harmonized data available to researchers, allowing students to gain hands-on experience without using or revealing actual participant information.

The [CanPath Student Dataset](#) mimics population-level data from over 40,000 participants from CanPath's cohorts, including the Ontario Health Study, the BC Generations Project, the Alberta Tomorrow Project, CARTaGENE in Quebec and Atlantic PATH. The dataset and a supporting data dictionary of over 400 variables are provided free of charge to approved teachers at Canadian universities and colleges for use in a university course.

[Learn more about the CanPath Student Dataset in this webinar featuring Dr. Brooks.](#)

New projects in 2021

This year again, many projects have been approved by CARTaGENE's access

committee :

Host genomics of COVID-19

Principal Investigator: **Julie Hussin**, Institut de Cardiologie de Montréal

The development of sex-specific prediction models for cardiovascular and neurological health

Principal Investigator: **Sarah Gagliano Taliun**, Institut de Cardiologie de Montréal

The relationship between pharmacotherapy and lifestyle habits in the prevention of cardiovascular disease

Principal Investigator: **Jean-Philippe Drouin Chartier**, Université Laval

Assessing the Relative and Absolute Risk for Site-Specific Cancer Mortality Attributed to Household Air Pollution

Principal Investigator: **Dean Hosgood**, Albert Einstein College of Medicine

Gene mapping and genetic correlation across complex traits

Principal Investigator: **Stuart MacGregor**, QIMR Berghofer Medical Research Institute

Improving understanding of the role of genetic variations in human disease and disease-related traits and integrating this knowledge into discovery of new drug targets and disease risk prediction

Principal Investigator: **Vincent Mooser**, McGill University

Exposure to perfluoroalkyl substances and age at menopause in the Canadian prospective cohort study CARTaGENE

Principal Investigator: **Michael Borghese**, Health Canada

The genetic epidemiology of breast cancer: CanPath Application to the Confluence Project (CanPath)

Principal Investigator: **Jennifer Brooks**, Dalla Lana School of Public Health, University of Toronto

Improving Lung Cancer Risk Prediction & Application (CanPath)

Principal Investigator: **Martin Tammemagi**, Brock University

Genotyping and Allele Calling of Complex Regions of the Human Genome (CanPath)

Principal Investigator: **Philip Awadalla**, Ontario Institute for Cancer Research

Occupational and Environmental Triggers in Development of Autoimmune Cutaneous Disease (CanPath)

Principal Investigator: **Elena Netchiporouk**, McGill University Health Centre

Mental health planning in the era of a pandemic: appropriately matching mental health needs with resources across Canada through a rapid assessment of COVID-19's impact on adults and older adults (CanPath)

Principal Investigator: **Helen-Maria Vasiliadis**, Université de Sherbrooke

There are 92 projects approved, 72 of which are ongoing.

26 new publications

Also, CARTaGENE data enabled the publication of 26 new scientific articles:

1. Adam-Poupart A, Noisel N, Irace-Cima A, Payette Y. Changements climatiques et zoonoses priorités : portrait de situation à l'aide des données de la cohorte populationnelle québécoise CARTaGENE. Institut national de santé publique du Québec, 2021
2. Béliveau A, Castilloux A-M, Tannenbaum C, Vincent P, de Moura CS, Bernatsky S, et al. Predictors of long-term use of prescription opioids in the community-dwelling population of adults without a cancer diagnosis: a retrospective cohort study. CMAJ open. 2021;9:E96
3. D'Entremont M, Couture EL, Nguyen M, Ni J, Yan A, Ko D, et al. Racial/ethnic differences in cardiovascular outcomes in a universal healthcare system: insights from the CARTaGENE cohort. European Heart Journal. 2021
4. Durrani R, Friedrich MG, Schulze KM, Awadalla P, Balasubramanian K, Black SE, et al. Effect of Cognitive Reserve on the Association of Vascular Brain Injury With Cognition: Analysis of the PURE and CAHHM Studies. Neurology. 2021

5. Elkholi IE, Di Iorio M, Fahiminiya S, Arcand SL, Han H, Nogué C, et al. Investigating the causal role of MRE11A p. E506* in breast and ovarian cancer. *Scientific Reports*. 2021;11:1-9
6. Ferri F, Deschênes SS, Power N, Schmitz N. Associations between cognitive function, metabolic factors and depression: A prospective study in Quebec, Canada. *Journal of Affective Disorders*. 2021;283:77-83.
7. Fierheller CT, Guitton-Sert L, Alenezi WM, Revil T, Oros KK, Gao Y, et al. A functionally impaired missense variant identified in French Canadian families implicates FANCI as a candidate ovarian cancer-predisposing gene. *Genome Med*. 2021
8. Gamache I, Legault M-A, Grenier J-C, Sanchez R, Rhéaume E, Asgari S, et al. A sex-specific evolutionary interaction between ADCY9 and CETP. *eLife*. 10
9. Graham E, Deschênes SS, Rosella LC, Schmitz N. Measures of depression and incident type 2 diabetes in a community sample. *Annals of Epidemiology*. 2021;55:4-9
10. Huguet G, Schramm C, Douard E, Tamer P, Main A, Monin P, et al. Genome-wide analysis of gene dosage in 24,092 individuals estimates that 10,000 genes modulate cognitive ability. *Molecular psychiatry*. 2021:1-14
11. Jantzen R, Noisel N, Camilleri-Broet S, Labbe C, Malliard T, Payette Y, et al. Epidemiological characteristics of the COVID-19 spring outbreak in Quebec, Canada: a population-based study. *BMC Infect Dis*. 2021
12. Jantzen R, Payette Y, de Malliard T, Labbe C, Noisel N, Broet P. Validation of breast cancer risk assessment tools on a French-Canadian population-based cohort. *BMJ Open*. 2021
13. Labrecque M, Touma L, Bhérer C, Duquette A, Tétreault M. Estimated prevalence of Niemann–Pick type C disease in Quebec. *Scientific Reports*. 2021

14. Li N, Zethoven M, McInerney S, Devereux L, Huang Y-K, Thio N, et al. Evaluation of the association of heterozygous germline variants in NTHL1 with breast cancer predisposition: an international multi-center study of 47,180 subjects. *NPJ Breast Cancer*. 2021;7:1-12
15. Lona-Durazo F, Mendes M, Thakur R, Funderburk K, Zhang T, Kovacs MA, et al. A large Canadian cohort provides insights into the genetic architecture of human hair colour. *Communications Biology*. 2021
16. Luu JM, Sergeant AK, Anand SS, Desai D, Schulze K, Knoppers BM, et al. The impact of reporting magnetic resonance imaging incidental findings in the Canadian alliance for healthy hearts and minds cohort. *BMC Medical Ethics*. 2021;22:1-15
17. Meng T, Kubow S, Nielsen DE. Common variants in the CD36 gene are associated with dietary fat intake, high-fat food consumption and serum triglycerides in a cohort of Quebec adults. *International Journal of Obesity*. 2021
18. Moura CS, Payette Y, Boileau C, Abrahamowicz M, Pilote L, Bernatsky S. Agreement in the CARTaGENE cohort between self-reported medication use and claim data. *Chronic Illness*. 2021
19. Noisel N, Pasquet R, Richardson L, Siemiatycki J, Broet P. RF-367 Occupational exposure to agents and substances in the CARTaGENE cohort. *Occupational and Environmental Medicine*. 2021
20. Nouwen A, Deschênes SS, Balkhiyarova Z, Albertorio-Díaz JR, Prokopenko I, Schmitz N. Measurement invariance testing of the patient health questionnaire-9 (PHQ-9) across people with and without diabetes mellitus from the NHANES, EMHS and UK Biobank datasets. *Journal of Affective Disorders*. 2021
21. Sen A, Schmitz N, Brazeau A-S, Deschenes S, Melgar-Quiñonez HR. The Role of Ultra-Processed Food and Depression on the Incidence of Type 2 Diabetes: A Community Study from Quebec, Canada. 2021 Virtual Diabetes Canada/CSEM Professional Conference. Virtual event: Elsevier; 2021:S27-S28.

22. Touma L, Labrecque M, Tetreault M, Duquette A. Identification and Classification of Rare Variants in NPC1 and NPC2 in Quebec. Sci Rep. 2021
23. van Tuijl LA, Voogd AC, de Graeff A, Hoogendoorn AW, Ranchor AV, Pan KY, et al. Psychosocial factors and cancer incidence (PSY-CA): Protocol for individual participant data meta-analyses. Brain Behav. 2021
24. Veillette S, Lamarche F, Agharazii M, Wassertheurer S, Hametner B, Madore F, et al. POS-296 Cardiovascular risk prediction with aortic pulse wave velocity: A CARTaGENE Study. Kidney International Reports. 2021;6:S127
25. Võsa U, Claringbould A, Westra H-J, Bonder MJ, Deelen P, Zeng B, et al. Large-scale cis-and trans-eQTL analyses identify thousands of genetic loci and polygenic scores that regulate blood gene expression. Nature genetics. 2021;53:1300-1310
26. Zeng Y, Zhao K, Oros Klein K, Shao X, Fritzier MJ, Hudson M, et al. Thousands of CpGs Show DNA Methylation Differences in ACPA-Positive Individuals. Genes. 2021;12:1349

Projects and publications using CARTaGENE's data pique your interest? [Visit our website](#)

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