



Thank you and Happy Holidays from the CARTaGENE team

Dear participants, you are essential collaborators in Quebec's health research ecosystem! Once again this year, you have shown your commitment and generosity by supplying new data for research (genealogy and nutrition, p. 3).

2024 marked a significant year for CARTaGENE. It was a year of transformation! We underwent a change in general management, rebranded, and for the first time, hosted events specifically for participants, including webinars and our 15th anniversary symposium.

CARTaGENE's executive director, Alexandra Obadia, who served with CARTaGENE for 16 years, including 12 years as director, resigned in January 2024. In April, CARTaGENE appointed a new director, Jean-Baptiste Rivière. More information about him can be found on page 5 of this newsletter.

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The CARTaGENE logo over the years



One of Mr. Rivière's first achievements was to help CARTaGENE reflect on its mission, values and image. This led to a rebranding exercise.

Last December, we asked you, via a survey, what would encourage you to get involved. We learned that you wanted more news from CARTaGENE, more accessible results in the form of articles, webinars and so on. The CARTaGENE team therefore decided to organize webinars for participants. In April, there was a webinar on genealogy presented by Simon Gravel (FR), and in June, a webinar on nutrition presented by Benoît Lamarche.

Finally, on October 30, we organized a symposium for participants. This event marked a turning point for CARTaGENE, as it was the first meeting between the scientific team and the participants (page 10). We really enjoyed it! Based on the discussions, we understand that you want more, and that some of you are interested in the concept of participatory research. We are thinking about ways to organise more science outreach activities and involve you more closely in the research.

Stay tuned!

Thank you once again, and we wish you a wonderful holiday season and a happy new year 2025!

The CARTaGENE team

Here is what your data creates:



In 2024, data was used by **201 research** teams in Quebec, Canada, and globally.

16 years of research advancements that will significantly influence health.



162 articles published in academic journals

67 theses and dissertations



Data collections in 2024

In 2024, CARTaGENE invited its participants in two data collection campaign, specifically the BALSAC campaign and the NutriQuébec campaign.

Genealogical research



The BALSAC population database, associated with Université du Québec à Chicoutimi, collaborates with CARTaGENE to collect genealogical data.

BALSAC is a digital database created from civil status records in Quebec. It facilitates the automatic reconstruction of genealogies throughout Quebec by tracing lineage back to the early 17th century.

This information is valuable to understand the demographic and historical elements that have influenced Quebec's genetic heritage. It enables a deeper understanding of the distribution of genes or specific diseases within the present population.

CARTaGENE genealogy and genetics data are used by population geneticists like Simon Gravel, who presented a webinar on the topic last April ([webinar \(in FR\) accessible here](#)).

CARTaGENE participants were invited to fill out the BALSAC genealogical questionnaire at recruitment. Over 8,800 participants completed it at that time. In the spring of 2024, CARTaGENE and BALSAC resent the questionnaire to participants who had not yet completed it.

Following a nine-week campaign, an additional 2,623 genealogical questionnaires were either completed or partially completed. The information gathered from these questionnaires will help in reconstructing the genealogies of CARTaGENE participants. At present, three distinct research projects use the genealogical data of CARTaGENE participants.

Nutritional research

In 2012, participants of CARTaGENE were invited to fill out a food frequency questionnaire. Over 9,300 individuals completed this questionnaire. This dataset is used extensively by researchers. For instance, 33 ongoing projects use CARTaGENE nutrition data. A scientific article detailing the nutritional profile of the CARTaGENE cohort was published last August (refer to p.9 for a summary of the key findings).

Due to the significant demand for longitudinal nutritional data, CARTaGENE has chosen to partner with the NutriQuébec project to facilitate a new collection of nutritional data in 2024.

Source of image: NutriQuébec



NutriQuébec is an initiative designed to create a dietary profile of the Quebec population to align with the objectives of the Quebec government's health prevention policy. NutriQuébec represents the most extensive project focused on nutrition and health for the residents of Quebec.

NutriQuébec gathers data on various factors associated with the health of numerous Quebec residents over an extended period of time.

The partnership with NutriQuébec enables CARTaGENE to enhance its expertise in nutrition and to bonify its databases for researchers.

Last June, the NutriQuébec campaign started for CARTaGENE participants. A few days in, a webinar on NutriQuébec was presented to participants. Throughout the nine-week campaign, over 3,000 individuals consented to take part in the project.

Health Follow-up 2025

Your involvement is essential!

Our last general health follow-up was 7 years ago (in 2016-2017) !

In 2025, you will be asked to fill out a new health questionnaire.

Here is a glimpse of the topics of the forthcoming questionnaire:

- Demographic attributes (education, employment, income, etc.)
- Lifestyle habits (physical activity, alcohol intake, sleep, etc.)
- Diseases
- Medication
- Consultations with a healthcare provider
- Physical measures
- Psychological well-being

When the campaign is launched, you will receive an email inviting you to participate, which will include a link to our online questionnaire. Your ongoing participation in CARTaGENE is crucial for gaining insights into how your health changes over time. The information you provide will be instrumental in disease prevention and enhancing care for everyone. Every response is valuable.

We sincerely appreciate your dedication to this vital research. The insights obtained can enhance treatments, public health strategies, and prevention efforts for the entire community.

Interview with a researcher

Jean-Baptiste Rivière has been serving as the executive director of CARTaGENE since April 8.

As the newest member of the team, Jean-Baptiste introduces fresh ideas for the development of CARTaGENE, bringing a new perspective on its future.

This interview took place on December 3.



CARTaGENE (CaG): Hi Jean-Baptiste, we appreciate your participation in this interview. You have been appointed as the new executive director of CARTaGENE. Could you share the journey that brought you to this position?

Jean-Baptiste Rivière (JBR)*: Hello Catherine, my education encompasses a PhD in human genetics from the University of Montreal, a postdoctoral fellowship in genomics and bioinformatics in Seattle, USA, and clinical postdoctoral training in molecular genetics, certified by the Canadian College of Medical Geneticists at McGill University. Throughout my career, I have worked in university hospitals, initially in Dijon, France, and subsequently at the McGill University Health Centre (MUHC) in Montreal.

I have experience in genetic diagnosis for rare diseases and cancers within clinical laboratories, in bioinformatics by leading teams of data analysts, and in academic research in genetics. In Dijon, I contributed to the establishment of exome sequencing, which involves the analysis of all genes in the genome, for the rapid diagnosis of children with rare diseases, during the early stages of this technology. This advancement has significantly improved the management of rare diseases over the past decade.

In 2015, while in Montreal, I contributed to the modernization of the MUHC's molecular diagnostic laboratory by incorporating new genomic and computer technologies into patient care.

CaG: Could you share the circumstances under which you first learned about the CARTaGENE project?

JBR: I became aware of CARTaGENE shortly after its launch, or perhaps even before. At that time, I was pursuing my doctoral studies, or possibly my master's. I was a student in the laboratory of Guy Rouleau, a prominent neurogeneticist renowned in Quebec and beyond (he would later serve as co-principal investigator of CARTaGENE). My understanding at that time was that CARTaGENE constitutes a significant resource for health research in Quebec. CARTaGENE exemplifies the kind of infrastructure or societal initiative, that nations with a robust research foundation develop.

CaG: In 2024, what significance does CARTaGENE hold for you?

JBR: CARTaGENE has reached the milestone of 15 years, a significant achievement worth emphasizing. This moment also serves as a chance to reflect on all that has been accomplished and to consider the path forward in the upcoming years. In 2024, CARTaGENE continues to be a vital resource in addressing health challenges in Quebec.

For instance, we understand the difficulty of obtaining quality data for research, which subsequently informs health actions grounded in evidence. This principle is the foundation of CARTaGENE: access to high-quality, extensive, and varied health data.

In Quebec, as in other industrialized nations, it is recognized that chronic diseases place considerable strain on our healthcare system. For instance, fifty percent of the adult population have one chronic health issue, and twenty-five percent have at least two. CARTaGENE was specifically established to investigate chronic diseases.

In Quebec, there is a significant challenge of population aging that is prevalent in nearly all industrialized nations. By 2031, one-quarter of the Quebec population will be aged 65 years and older. Globally, only Japan has a higher rate of population aging. CARTaGENE is a longitudinal study that tracks individuals as they age. The current median age of participants is 66 years.

Our contemporary lifestyle harms the environment and worsens numerous health issues. For instance, forest fires and heat waves affect the well-being of older adults. CARTaGENE was established to support research on the environmental factors contributing to diseases and the link between genetics and environment in the susceptibility to diseases.

Social inequalities, which remain a persistent issue, significantly affect health outcomes. Poverty, in particular, is associated with lower life expectancy, a higher prevalence of chronic diseases, and limited access to healthcare, among other factors.

CARTaGENE possesses a variety of data that enables us to investigate these matters in research and to suggest measures that can be implemented in public health policy.

Ultimately, education and the distribution of biomedical knowledge to the general public are crucial in addressing the challenges I have outlined, especially in an environment rife with misinformation. I believe that CARTaGENE can collaborate with provincial partners to promote high-quality knowledge for health education and prevention.

CaG: What do you believe should be the priority for CARTaGENE?

JBR: The primary goal should be to position CARTaGENE as a key reference for population health research in Quebec. We need to increase usage and visibility, which requires several actions. (1) We must boost the use of data by research teams across all health research domains. We will accomplish this by enhancing the resource.

Therefore, (2) we must persist in converting biological samples into data, which includes sequencing the entire genomes of thousands more participants and employing other innovative technologies that enable us to gain a deeper understanding of the factors that influence the development of chronic diseases.

(3) It is essential to establish and strengthen partnerships with various entities in Quebec, other cohorts, and research biobanks to combine efforts, as well as with health organizations to enhance the use of CARTaGENE in health decision-making.

(4) It is essential to create a component for the communication and dissemination of knowledge for both participants and the general public. To achieve this, we must engage CARTaGENE participants who are willing to assist us in addressing this challenge. The focus is on developing participatory research to enhance the usefulness and visibility of the results derived from our data for the general public.

JBR: I see this year as a time of transformation during which we establish the groundwork for our future vision, adjusting the steering wheel to chart a course and keep it steady.

(1) By optimizing the use of data in research. (2) By helping to convert them into health initiatives. (3) By featuring a substantial element of knowledge sharing and collaborative research. That applies to the first year.

Within five years, I anticipate that we will have converted our samples into data utilizing the most advanced biomedical technologies available. This enhancement will result in a fivefold increase in access requests and the establishment of strong partnerships with governmental and paragonovernmental organizations in public health. Additionally, we will have created a dynamic component for knowledge dissemination by engaging participants.

CaG: Thank you, Jean-Baptiste, and welcome to CARTaGENE.

*The content of this interview has been minimally revised for improved readability.



What does your nutrition data tell us?

You probably should consider taking vitamin D supplements.

An analysis of CARTaGENE nutrition data from 2011-2012 suggests this conclusion.

Vikki Ho and her team evaluated nutritional intakes using a metric known as the Healthy Eating Index (HEI), which measures the extent to which an individual adheres to the recommendations outlined in Canada's Food Guide.

The research team indicated that the quality and quantity of nutritional intakes differed among participants, yet generally adhered to Health Canada recommendations. However, there were exceptions. Fewer than 50% of participants achieved the recommendations for fibre, folate, sodium, and vitamin D intakes.

The most significant deficiency was in daily vitamin D consumption. In fact, fewer than 5% of CARTaGENE participants met the recommended daily intake of vitamin D.

HEI scores fluctuate according to specific demographic traits, lifestyle habits, and chronic health conditions.

Scores were elevated for individuals who had never smoked, those who had attained at least a high school education, and those who engaged in a high level of physical activity, in comparison to current smokers, individuals with less than a high school education, and those with a low level of physical activity, respectively.



Vitamin D

Health Canada advises individuals aged 51 and older to:

1. Take a daily supplement containing 400 IU (10 µg) of vitamin D
2. Continue to eat foods that contain vitamin D as part of healthy eating

Examples of foods that are rich in vitamin D include:

- eggs (yolk)
- soft margarine
- fatty fish, such as:
 - salmon
 - arctic char
 - rainbow trout
- unsweetened lower-fat milk
- unsweetened fortified plant-based beverages

Reference:

<https://www.canada.ca/en/health-canada/services/nutrients/vitamin-d.html>

Nutrition data was collected in 2011-2012 from participants in the first phase of CARTaGENE recruitment. The standardized questionnaire comprised over 160 questions regarding food, beverages, and dietary supplements. Participants were asked about the frequency of consumption per year, month, or day for various items. More than 9,300 participants completed this nutritional questionnaire.

CARTaGENE nutritional data, which can be integrated with additional information from health questionnaires, physical measurements, and biological samples, continues to be one of the most comprehensive resources for nutrition research in Quebec and Canada. Numerous researchers use CARTaGENE nutrition data to examine its connections with the onset of various diseases. The newly gathered data in partnership with the NutriQuébec project will further enrich this resource.

The CARTaGENE nutrition data profile was released last August in the BMJ Open journal.

Reference: Ho V, Csizmadia I, Boucher BA, McInerney M, Boileau C, Noisel N, Payette Y, Awadalla P, Koushik A. Cohort profile: the CARTaGENE Cohort Nutrition Study (Quebec, Canada). BMJ Open. 2024 Aug 17;14(8):e083425.

CARTaGENE's 15th Anniversary Symposium

On October 30, CARTaGENE held its 15th Anniversary Symposium of CARTaGENE at CHU Sainte-Justine in Montreal.

This event aimed to celebrate this milestone with the participants. Approximately 75 of you attended in person, while around 90 joined online.

The afternoon was split into two parts. Scientific directors introduced the CARTaGENE platform. This was followed by presentations from six researchers with active research projects on various topics.



The second part, organized by the team from the Center of Excellence on Partnership with Patients and the Public (CEPPP), was a discussion forum designed to gather insights about the participants. Guided by two general questions, the facilitators conducted a 60-minute interactive session between the participants and the CARTaGENE team.



Question 1: What are the most important themes or research topics relevant to your health and wellness?

Participants are interested in a variety of subjects, including chronic illnesses, neurological disorders, mental well-being, nutrition, and more.

Question 2: How would you like to be involved in CARTaGENE's activities?

Overall, the participants who voiced their opinions would like greater involvement through workshops or discussion groups. They also seek increased communication from CARTaGENE, including webinars, written updates, and website information. Additionally, they would appreciate personalized feedback on their results.

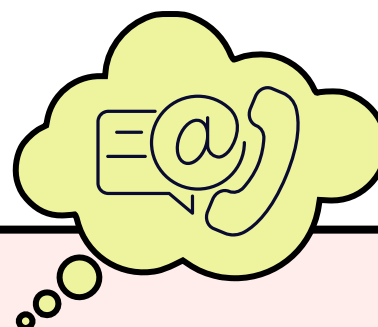
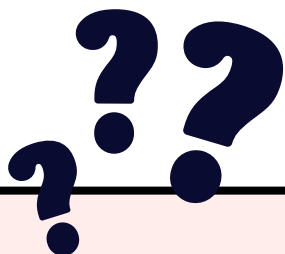
Ultimately, numerous participants indicated that it would be beneficial to recruit new participants from the general population or their family members.

The CARTaGENE team greatly enjoyed the Symposium and is considering future steps. We are assessing funding opportunities to initiate participatory science activities and collaboration with participants.

One thing is clear: there will be no turning back!

We encourage participants to watch the video of this unforgettable afternoon.





Have you relocated, retired, updated your email address, or changed your phone number?

Please assist us in maintaining the accuracy of your information.

Please take a moment to provide us with your updated contact information. Even if you relocate outside of Quebec or Canada, you can continue to be a participant!

Maintaining communication with you is crucial for the success of this project!

Get in touch with us!

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Our collaborators

