**NEWS FROM CARTAGENE** 

# The CARTaGENE Voice

ACCELERATE HEALTH RESEARCH

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www.cartagene.qc.ca



# What is new at CARTaGENE?

For over a decade, CARTaGENE has been supporting health research by providing access to health data and biological samples collected from 43,000 Quebec participants. This year, we wish to improve your experience with CARTaGENE by creating two new tools for you to use at your disposal soon:

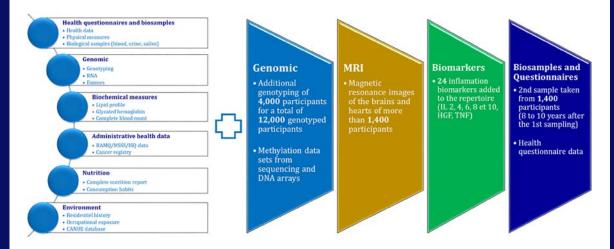
- **Genome browser**: A genotyping and sequencing data visualization tool from CARTaGENE. It will now be possible to obtain information on the targeted regions from exomes sequencing, genotyped variants or SNPs (number of genotyped participants, genotyping platforms, etc.), allele frequencies, and platforms/chips used.
- **Superset**: An aggregated data visualization and exploration tool that provides an intuitive interface for creating interactive dashboards. You will now be able to obtain the number of cases and participants' characteristics that interest you for your pre-application or grants demands.

# New data sets are available!

This year, various types of data are accessible!

### Available data

### Data to be available soon



- In 2018, CARTaGENE performed a cohort-wide recontact using its online portal to update participants' health data. This new data is now available to all health researchers. The health history of CARTaGENE participants is a valuable resource for health research. These data help in making it possible to establish links between environmental and genetic factors and the occurrence of chronic diseases.
- A collaboration with the Canadian Alliance for Healthy Hearts & Minds led to, among other things, obtain magnetic resonance images (MRI) of the brains and hearts of over 1,400 CARTaGENE participants. The health questionnaires of these participants along with the MRI images' interpretations will also be available. Stay tuned: this data will be available soon!

### **Data access**

Data Access: Rapid, Simple, Efficient

# Step 1

Create an account and submit application sdas.cartagene.qc.ca

# Step 2

Evaluation by an independent scientific committee

# Step 3

Access contract

Contact information:

access@cartagene.qc.ca / 514-345-2156

Visit our website:

cartagene.qc.ca

Submit an application:

sdas.cartagene.qc.ca

Follow us!





# **New collaborations**

Over the years, CARTaGENE has established collaborations with the public and private sectors to enrich its resources and benefit health researchers. Last year, two collaborations were initiated:

- 1. <u>PSY-CA</u>: European consortium of 10 European cohorts and 7 international cohorts focusing on the links between psychosocial factors and cancer.
- 2. <u>RMGA "Réseau de Médecine Génétique Appliquée"</u>: Quebec's research network of applied genetic medicine: integration of CARTaGENE as a key partner and offer free access to CARTaGENE's data to RMGA regular members.

**CARTaGENE** is open to new collaborations!

Follow us to stay informed of future collaborations planned for 2019  $\,$ 

\* Special offer for RMGA researchers: Free access to CARTaGENE's data \*

# **Recent projects**

For several years, CARTaGENE participants' data and biological samples have contributed in more than 50 research projects. Here are 4 projects that started in 2018:

1-Guillaume Lettre - Université de Montréal / Montreal Heart Institute - Genetic analyses of cardiometabolic diseases and traits in CARTaGENE. Dr. Lettre and his team will use statistical approaches to identify associations between genes and diseases in the CARTaGENE cohort. Research interests include cardiometabolic (myocardial infarction, hypertension, diabetes), anthropometric (height, body mass index), and blood (platelets, red and white blood cells). The results will help evaluate the potential of certain genes as therapeutic tools.

2-Patricia Tonin – McGill University - Genetic analyses of proposed new ovarian cancer predisposing gene. Although the majority of ovarian cancer cases are attributable to mutations in the BRCA genes, the inherited ovarian cancer is not solely due to these genes. Dr. Tonin and her team are working on the characterization of another gene, named FANCI that plays an important role in the development of ovarian cancer that is not attributable to the BRCA genes.

3-Brent Richards - Institut Lady Davis Institute, Hôpital Général Juif - Pharmacogenetic study exploring the impact of a genetic variant in the CYP2R1 gene on the response to vitamin D replacement therapy. Dr. Richards' team uses the CARTaGENE platform to evaluate the impact of a genetic mutation in response to vitamin D supplements. The latter could be lower in individuals carrying this mutation.

4-Josée Laganière - Héma-Québec - <u>Prevalence study of the weak D antigen blood group type 42</u>. For several years, blood banks have been turning to genetic analyzes to avoid incompatibilities and the risk of fatal reactions during transfusions. Dr. Laganière and her team are studying the prevalence of significant genetic variations of this blood group in the Quebec population.

To view all the projects in progress, visit:

https://www.cartagene.qc.ca/en/researchers/projects-and-publications

# **Miscellaneous**

### Discover our Super Users

With 3 ongoing projects, Dr. Alain Moreau is one of our super users. Dr. Moreau's team uses the CARTaGENE data and biological samples to answer different research questions according to the following projects: Identification of genes involved in adolescent idiopathic scoliosis (since 2015); SNP discovery involved in

myalgic encephalomyelitis (since 2017); Role of prohibitins and SUMOylation in osteoarthritis pathogenesis (in 2018).

### New blood in health research!

Did you know that in the past two years, 7 young researchers have used CARTaGENE data with/without biological samples for their research projects? This represents a significant number of CARTaGENE's users in that period. Congratulations to the next research generation!

### Letter of Support from CARTaGENE

Did you know that CARTaGENE could support you with your grant applications by providing support letters for any research projects already using or expecting to use CARTaGENE's data with/without biological samples?

To obtain a letter of support, simply send an email to access@cartagene.qc.ca as soon as possible before the submissions deadlines (at least five working days).

Make sure to include the following information: the name(s) of the researcher(s), the name of the grant and granting agency, submission deadline, and a summary of the research project including the contribution of CARTAGENE.

### Do not hesitate to contact us for more information

### Devoting to participants

This year, CARTaGENE is multiplying its efforts to improve the commitment of its participants. "The CARTaGENE Voice" now exists in two distinct versions: a version for you, the researchers, and one for the participants, focused on their interests. Participants' commitment is a crucial matter for the sustenance and the success of a population cohort such as CARTaGENE. It is a guarantee of success for future health monitoring as well as for researcher recontact projects.

### Our team

### The scientific directors of CARTaGENE:

**Dr. Sébastien Jacquemont:** Dr. Jacquemont's research focuses on neuropsychiatric disorders caused by genetic factors. His team combines genetics, neuroimaging, biomarkers, and the clinical aspect to

understand how genetic mutations lead to neuropsychiatric symptoms and disability in patients. Dr. Jacquemont and his team are also actively investigating structural genomic variants (deletions and duplications) associated with autism and schizophrenia.

**Dr. Philippe Broët:** Dr. Broët, a clinical researcher at CHU Sainte-Justine, joined the CARTaGENE team as scientific director during the past year. Dr. Broët is a full university professor - hospital practitioner at the "Assistance Publique - Hôpitaux de Paris" since 2017. After a specialty internship, he continued his training with a doctorate in biostatistics. His areas of expertise are clinical genomic epidemiology and predictive medicine. His research focuses on the development of new analytical strategies adapted to genomic studies in multifactorial diseases.

**Dr. Aikaterini Kritikou:** Since 2013, Dr. Kritikou is the assistant director of the CHU Sainte-Justine research center. She previously led the biobank of the Montreal Heart Institute. She holds a Ph.D. in Developmental Biology from the University of Cambridge. Dr. Kritikou joined the CARTaGENE team in 2017.

### The CARTaGENE team:



CARTaGENE is 43,000 participants devoted to advancing health research in Quebec and beyond!

The CARTaGENE team provides quality data and biological samples to help you with your research quests.

We would like to thank each one of you for your continued research contribution in the past 10 years!





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