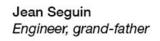
# I am making a difference...

for health research







# Newsletter

Volume 2 – January 2014

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#### Contact us!

For questions about participation, contact the call center:

1-866-366-4249

cartagene@advanis.ca

For further information:

514-343-7703

info@cartagene.qc.ca

# CARTaGENE project already enrolled 34,000 participants!

The second wave of recruitment is underway. This recruitment will cover new regions of Quebec and expand our cohort of participants, thus ensuring the sustainability of the CARTaGENE project.



Notice to participants of the second wave (since December 2012 only). The blood sample is a valuable contribution to the project and for medical research. Please, help us achieve this goal.

#### **BALSAC: the CARTaGENE genealogical option**



Among the major biobank projects in the world, CARTaGENE is one of the only ones to include a ancestry component. Indeed, BALSAC data makes it possible to reconstruct the genealogies of CARTaGENE participants.

The BALSAC population records allow for the tracing of ancestors going back almost four centuries and provide a historical dimension to research conducted using the CARTaGENE data.

BALSAC will also support researchers by providing information on the structure of the present population, which allows to refine genetic epidemiology and population genetics research.

To contact the research service of BALSAC balsac@uqac.ca, 418-545-5517

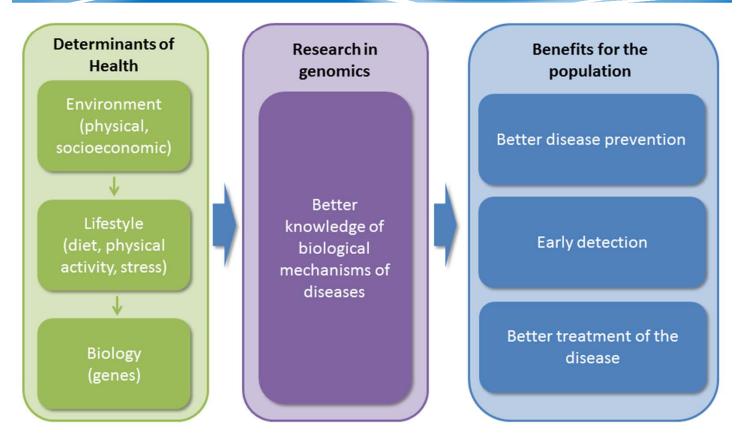
The objective of the genealogical option is to provide researchers with information on the participants' ancestors, which is invaluable for understanding the demographic and historical factors that have shaped the genetic heritage of Quebec as well as the distribution of genes or diseases in the current population. BALSAC is a computerized file built from vital event records. It allows for the automatic recreation genealogies across Quebec by tracing ancestors up until the early 17th century. BALSAC was created 40 years ago at the Université du Québec à Chicoutimi in partnership with the Université Laval, McGill University and the Université de Montréal.



**Dr. Hélène Vézina,** Director of BALSAC



# CARTaGENE enables genomics and personalized medicine



**Figure 1: Continuum of molecular medicine.** Many factors come together to affect the health of individuals and communities, including environmental factors, lifestyle and genetic factors. Genomics research explores the relationship between the environment, lifestyle and the genetic background of individuals in order to understand the mechanisms leading to diseases such as cardiovascular disease, diabetes and cancer. Increasing our knowledge of how disease begins will lead the way to better disease prevention, more appropriate screening programs and more efficient and personalized treatments.

For governments and other stakeholders the value and the benefits that platforms like CARTaGENE can bring to population health has been recognized. Strategic funding initiatives have been created to facilitate the development and use of projects focusing on genomics and personalized medicine. For instance, a partnership between **Genome Canada** and the **Canadian Institutes of Health Research (CIHR)** allowed to invest important amounts of public funding in these types projects and provides an opportunity for Canadian researchers to be leaders in the field of genomics and personalized medicine.

The aim of research in genomics and personalized medicine is not only to make health systems more cost-effective, but also to ensure that the findings will be translated into benefits for patients and the population.





# Canadian Alliance for Healthy Hearts and Minds of the Canadian Partnership for Tomorrow Project

The Canadian Partnership for Tomorrow Project (CPTP) is a prospective cohort study funded by the federal government whose aim is to recruit 300,000 participants to create a national database. This project on the health of the population will accelerate the fight against cancer and other chronic diseases for the benefit of all Canadians. CARTaGENE represents the Quebec cohort of this pan-Canadian project.

The Canadian Alliance for Healthy Hearts and Minds (CVCD) is a large nationwide study of cardiovascular disease and cognitive impairment. Cardiovascular dysfunction and vascular cognitive impairment have a strong impact on the quality of life, costs of health care and longevity in Canada and around the world. It is therefore of paramount importance to understand their early determinants and progression to clinical manifestations.

# The CVCD project is a unique opportunity to bring together Canadian experts to:

- 1 Characterize contextual, cultural and individual factors of cardiovascular health in Canada by linking clinical manifestations and use of health services;
- 2 Enrich the CPTP cohort with magnetic resonance data to better understand the impact of risk on the first subtle pathophysiological changes and identify early markers of disease.

This study including 9,700 people of the pan-Canadian CPTP cohort is unique because of its size, the depth of its clinical information, its geographic and ethnic diversity and the integration of public health data. This initiative therefore has unparalleled potential to fill our gaps and provide critical information to help health prevention by combining the assessment of contextual factors, the detailed assessment of individual risk factors, and the inclusion of new markers based on imaging. In this project, 1300 CARTaGENE participants will be selected to contribute to the CVCD project. Therefore, CARTaGENE participants may be contacted to participate in this important project.





# CARTaGENE participants can be proud to have advanced research in health

#### **Ongoing projects**

The following projects have used CARTaGENE data and / or samples:

#### **Environment, Epigenetics and Rhumatic Diseases**

- McGill University Health Centre, Division of Clinical Epidemiology
- Abstract: There is a strong interest in epigenetic diseases such as rheumatoid arthritis (RA). Exact pathways for triggering the RA process are still poorly understood. The aim of our project is to investigate whether exposure to air pollution is associated with immune disorders such as RA by an epigenetic change.
  Dr. Sa



Dr. Sasha Bernatsky, MD, FRCPC, PhD

#### **Medical Consequences of Human Herpes Virus 6 Chromosomal Integration**

- Université Laval, Division of Microbiology, Infectious Diseases and Immunology
- Abstract: The Human Herpes Virus Type 6 is known to be able to integrate into the host chromosome. In fact, 1% of the world population has a chromosomal integration of the virus. Our goal is to determine the sites of chromosomal integration and to assess whether the virus is a risk factor for certain diseases.



Dr. Louis Flamand, PhD, MBA

#### Genealogical Characteristics of the CARTaGENE Participants

- Université du Québec à Chicoutimi, BALSAC Project
- Abstract: The objective of this research project is to draw a portrait of the genealogical structure of the CARTaGENE participants based on genealogical data and vital event records. About 5400 genealogies will be constructed for the first time on a large sample of the Quebec population.



Dr. Marc Tremblay, Ph.D

#### **Prevalence of Chronic Kidney Disease in the CARTaGENE Cohort**

- Montreal Sacré-Cœur Hospital Research Centre
- Abstract: In recent years, many studies have suggested that the UMOD gene plays a role in the development of chronic kidney disease. The UMOD gene produces a protein called uromodulin, whose function is still unknown. Our goal is to study in detail the variations of the UMOD gene, its relation with uromodulin production and renal function.



Dr. François Madore, MD, M.Sc

#### **CARTaGENE** is proud to contribute to research

#### Ongoing projects (continue)

### Testing the accumulation of deleterious mutations during expansion space by exome sequencing

- University of Bern (Switzerland), Department of Ecology and Evolution
- Abstract: The spatial population expansion occurs when individuals spread to vacant spaces and leads to the accumulation of deleterious mutations at a higher rate than stationary populations. Some regions like Saguenay Lac-St-Jean (SLSJ) experienced spatial expansions and have a high rate of genetic diseases which could be partly due to expansion. We propose to compare the genetics diversity of people with ancestors from wave expansion from which with stationary ancestors. These results would otherwise explain the high incidence of certain genetic diseases in areas like SLSJ.

  Dr. Laure



Dr. Laurent Excoffier, Ph.D

#### **Upcoming projects**

The new projects using CARTaGENE data and / or samples that will be starting soon :

### Molecular Dissection of Congenital Heart Disease in the French-Canadian Population

- CHU Ste-Justine, Department Foeto-maternal and neonatal pathologies
- Research interests: Congenital heart disease, founder effect, new traits, population genetics, autozygotes



Dr. Gregor Angelfinger, MD, Ph.D

## Clinical Implementation and Evaluation of the Results of Blood Biomarkers for Chronic Obstructive Pulmonary Disease (COPD)

- Genome British Columbia, St. Paul's Hospital, UBC, PROOF Centre for Excellence
- Research interests: biomarkers, economic health, genomics, proteomics, diagnostics, evaluation of health technologies

Dr.Don D. Sin, MD, MPH.

# Electrocardiogram (ECG) Abnormalities in Systemic Sclerosis and other Systemic Rheumatic Autoimmune Diseases

- Jewish General Hospital
- Research interests: ECG abnormalities, systemic autoimmune rheumatic diseases, disease management, personalized treatment

Dr. Marie Hudson, MD, MPH.

### Quebec's news in genetics

#### Dr. Luigi Bouchard rewarded!

Chicoutimi, Canada, November 2, 2013. Dr. Luigi Bouchard was awarded the Plourde-Gaudreault prize at the 30th edition of the Regional Scientific Merit. This award is intended as a recognition of the professional contributions in the field of health and social services. The award specifically recognizes the impact of his work with the population, among others, the screening for genetic diseases that are more common in the Saguenay region.

# Stay informed!

Visit our website:

www.cartagene.qc.ca
You'll find the latest
news in the field of
genetics and an
updated list of research
projects that are
underway.

# An innovative partnership between the CHU Sainte-Justine and Genome Quebec

Montreal, Canada, October 21, 2013. The creation of the first pediatric clinical genomics center built in Canada was announced this past October. This highly innovative center will develop new solutions for genetic diseases in children. This initiative will transform the quality of care and improve prevention. This is a first in Canada and brings a breath of hope to sick children and their families.

# Unexpected Genomic Evolution in 400 Years of the French-Canadian history

Montreal, Canada, October 8, 2013. Our principal investigator, Dr. Philip Awadalla, in collaboration with researchers at the CHU Sainte-Justine Research Centre and Université de Montréal have discovered a unique genomic signature bequeathed to the current French Canadians by the early French settlers. This signature experienced unprecedented development in human history and could serve as a model to study the effect of demographic processes on human genetic diversity.

#### What participants think about CARTaGENE:

"It is today that we work for our future. By putting our children into the world, we want the best for them. We want them to be healthy. Participating in this program allows my generation to help the next ones have a healthy life!"

"I believe that a better understanding of our genes and our behavior that affect our health can help identify our diseases and find treatments that fit. Therefore, this requires a large participation of Quebecers!"

"I'm glad I could contribute to the advancement of science and health here in Quebec, to help society. It is important that some of us help-out in this way because it does not always mean that others will!"



# Thank you for making a difference!

#### In partnership with:



Centre de Recherche du CHU Sainte-Justine

Le centre hospitalier universitaire mère-enfant

Pour l'amour des enfants



















Genome Québec relier la science à la vie



**Genome**Canada

