CGP

CENTRE OF GENOMICS AND POLICY
CENTRE DE GÉNOMIQUE ET POLITIQUES
www.genomicsandpolicy.org

# ANNUAL REPORT 2009

McGill

Faculty of Medicine
Department of Human Genetics

# 1. Centre of Genomics and Policy

Located within the McGill University Genome Quebec Innovation Centre, the Centre of Genomics and Policy (CGP) is at the crossroads of the legal, medical and public policy fields. Within these fields, the CGP promotes prospective structuring and guidance for the orientation and application of research in genomic health sciences.

From a multidisciplinary perspective, and in collaboration with national and international partners, the CGP analyzes the socio-ethical and legal norms influencing multiple aspects of the promotion, prevention and protection of human health.

Currently, the CGP's research covers five areas of genomics and policy: procreation and reproductive genetics, pediatric health, privacy, public health, and personalized medicine. These domains are approached using three guiding approaches: internationalization, policy development and knowledge transfer. First, CGP promotes internationalization by undertaking comparative analyses of policies and guidelines around the world. Secondly, CGP actively participates in the creation of international consortia with a view to promoting multidisciplinary policymaking. Finally, via the HumGen law and policy database (www.humgen.org), the CGP promotes knowledge transfer of its work.

# 2. Our Team


McGill
740
Docteur-
Penfield
CGP
CENTRE OF GENOMICS AND POLICY
CENTRE DE GÉNOMIQUE ET POLITIQUES
Health
Law

**PROFESSOR**

Knoppers, Bartha Maria

**ASSOCIATE PROFESSOR**

Avard, Denise

**ASSISTANT PROFESSORS**

Joly, Yann
Wallace, Susan

**POST-DOC FELLOW**

McClellan, Kelly

**PROFESSIONAL ASSOCIATES**

Abdul-Rahman, Ma'n H.
Black, Lee
Bucci, Lucie Marisa
Isasi, Rosario
Lévesque, Emmanuelle
Nguyen, Thu Minh
Samuël, Julie
Tassé, Anne-Marie

**INTERN**

Grégoire, Gabrielle

**COLLABORATORS**

Mendez, Alberto Arellano
Sánchez , Gerardo Jiménez
Alvarez, Cesar Lara
Sánchez, Pablo Francisco Oliva
Borry, Pascal
Howard, Heidi Carmen

**RESEARCH STUDENTS**

Chagnon, Annie
Dam, Amy
Harris, Gregory
Hemmings, Francis
Kirby, Emily
Kleiderman, Erika
Le Huynh, Michael
Ouellette, Jean-François
Ramos-Paque, Emma
Rioux, Amelie

**SYSTEMS ADMINISTRATOR**

Truong, Dan-Thanh

**INVITED SCHOLARS**

Costea, Irina

**ADMINISTRATORS**

Hozyan, Rose-Marie
Racco, Eliane
Rossi, Marisa

# 3. Research Projects 2009

## 1. GRaPH-INT: Genome-based Research and Population Health International Network - Secretariat

Public Health Agency of Canada
December 2007 – March 2010

| PRINCIPAL INVESTIGATOR: | COLLABORATOR: |
|---|---|
| **AVARD Denise** | **BUCCI Lucie Marisa** |

GRaPH-Int is a global collaboration of individuals and organizations with an interest in public health genomics. The network helps transform knowledge and technologies into public policies, programmes and services for the benefit of public health. GRaPH-Int is both a Network and an Enterprise with interdisciplinary goals that support dialogue, research, education and training, communication and stakeholder engagement.

The Centre of Genomics and Policy is commissioned by the Public Health Agency of Canada to act as the International Secretariat of the Network.

## 2. ThéCell (Réseau de thérapie cellulaire et tissulaire)

Fonds de la recherche en santé (FRSQ)
April 2009 – April 2013

**PRINCIPAL INVESTIGATOR:**

GERMAIN Lucie

**CO-INVESTIGATORS:**

| | | |
|---|---|---|
| AUGER François | LAVERTY Sheila | ROY Denis-Claude |
| BERTHOD François | ROUTY Jean-Pierre | TANGUAY Jean-François |
| **KNOPPERS Bartha** | | |

**COLLABORATOR:**

**NGUYEN Thu Minh**

Created in 2009, the Cell and Tissue Therapy Network brings together some 50 researchers in order to facilitate Phase 1 and 2 clinical studies aimed at making advanced cell therapy publicly accessible by enhancing and developing technological platforms established through Québec universities and their partners. ThéCell is a lever and catalyst for mobilizing and coordinating use of, and access to, infrastructure and highly qualified personnel in the field of cell and tissue therapy. Under the wide scope of ThéCell, our team at the CGP is involved in two specific funded projects:

- Platform Project: "Ethical, Legal and Social Aspects of Cell Therapy"
  (April 2009 – April 2011)
  Principal Investigator: **KNOPPERS Bartha**

- Funded Project: "Translation clinique de la peau bi-lamellaire reconstruite in vitro par génie tissulaire - application pour le traitement des grands brûlés"
  (April 2009 – April 2011)
  Principal Investigator: AUGER François

## 3. A Prospective Study to Identify and Validate Biomarkers of Therapeutic Resistance in Colorectal Metastatic Cancer (Q-CROC)

FRSQ – Fonds de recherché en santé du Québec – and Pfizer
2009 – 2012

**PRINCIPAL INVESTIGATOR:**
BATIST Gerald

**CO-INVESTIGATORS:**

| | | |
|---|---|---|
| BASIK Mark | GUILLEMETTE Chantal | **KNOPPERS Bartha Maria** |
| CHABOT Benoît | PANASCI Lawrence | TÊTU Bernard |
| SPATZ Alan | | |

**COLLABORATORS:**

**BLACK Lee**
GRÉGOIRE Gabrielle

Q-CROC is developing a provincial-wide network of collaborators which includes oncologists, surgeons, pathologists, clinician-scientists and basic researchers. Q-CROC has the objectives of improving and developing translational and clinical research.

In this project, Q-CROC identifies and validates biomarkers specific for clinical resistance to FOLFOX and bevacizumab, a standardized first line treatment for metastatic colorectal cancer. The aims are to 1) create a unique and unprecedented clinical resistance biobank and 2) use these biospecimens to identify biomarkers that can be rapidly translated to the clinical setting. The experimental flow is a continuum of identification and validation of biomarkers starting with validation of candidates drawn from the literature or from the research labs of Q-CROC scientists, identification and validation of new biomarkers and functional validation of the most promising of these in order to obtain insights into mechanisms of resistance. Q-CROC has combined classic gene expression profiling with entirely novel approaches, including the discovery of alternative splice variants, DNA copy alterations, microRNA

expression changes and biomarker discovery, all poised to obtain novel and clinically relevant insights into the molecular biology of drug resistance. The key point is the study of samples obtained from clinically resistant patients. The project will ultimately lead to prospective biomarker-guided therapeutic studies to obtain the level 1 evidence required of truly valid biomarkers. The ultimate goal is to accelerate the development of personalized medicine through better patient and treatment selection, and to generate new knowledge on therapeutic resistance in the clinical context.

## 4. International Cancer Genome Consortium/Data Access Compliance Office (2009-2011)

Ontario Institute for Cancer Research (OICR)
2009-2011

**PRINCIPAL INVESTIGATOR:**
HUDSON Tom (ICGC Secretariat)

**CO-INVESTIGATOR:**
**KNOPPERS Bartha Maria**

**DATA ACCESS MANAGER:**
**JOLY Yann**

**COLLABORATORS:**
**WALLACE Susan** (Ethics and Policy working group)
**HEMMINGS Francis (DACO)**

The International Cancer Genome Consortium (ICGC) has been organized to launch and coordinate a large number of research projects that have the common aim of comprehensively elucidating the genomic changes present in many forms of cancers that contribute to the burden of disease in people throughout the world.

The DACO is responsible for the handling of requests for access to controlled data collected by the ICGC. It reports to both the Data Coordination Centre and the International Data Access Committee. Its objectives are to facilitate ethical, efficient and responsible transfer of controlled data to members of the scientific community who agree to the Consortium's terms and objectives.

## 5. A Research and Knowledge Network on Genetic Health Services and Policy: Building on the APOGEE-Net and CanGeneTest experiences

Canada Institutes for Health Research (CIHR)
November 2008- November 2013

**PRINCIPAL INVESTIGATORS:**

| | | |
|---|---|---|
| AMARA Nabil | GAUDET Daniel | LANDRY Réjean |
| BATTISTA Renaldo N. | GIGUERE Yves | LEDUC Nicole |
| BLANCQUAERT Ingeborg Rose | | GODARD Beatrice |
| LEGARE France | CASSIMAN Jean-Jacques | **KNOPPERS Bartha Maria** |
| MARRA Carlo A | COLE David E. C. | LABERGE Anne-Marie |
| MATTHIJS Gert | DROUIN Régen | LABERGE Claude |
| MITCHELL Grant A. | FOREST Jean-Claude | LABREQUE Michel |
| REINHARX Daniel | FOULKES William David | LAFLAMME Nathalie |
| ROUSSEAU Francois | FRIEDMAN Jan Marshall | LAMOTHE Lise |
| SIMARD Jacques R. | | |

**CO-INVESTIGATORS:**

DROUILLARD Lisa Marie
ELLIOTT David Carleton
OUELLET, Denis

**COLLABORATOR:**

**TASSÉ Anne Marie**

CanGeneTest is an international knowledge and research network studying the challenges posed by healthcare treatments and policies, including those related to genetic laboratory services. Its general objective is to improve the technological transfer of clinically useful genetic innovation to achieve a more favourable cost/efficiency ratio for the healthcare system, and to promote healthcare policy development in light of probative statistics. Using a multidisciplinary approach, we aim to study the development of the genetic screening test, from its discovery in the fundamental research lab up to its use in the clinical context. Furthermore, with the help of an electronic newsletter and a website, we hope to contribute to the dissemination of relevant and accurate information to all our colleagues interested in the research field, downstream from gene discoveries, and necessary for the translation of the clinically relevant and useful studies into innovative health care.

## 6. Emerging Team in Development of Strategies for Uptake and Analysis of Nanosequencing-Derived Data Sets and Linking to Disease

Canadian Institutes of Health Research (CIHR)
October 2008 – September 2013

**PRINCIPAL INVESTIGATOR:**

ROULEAU Guy

**CO-INVESTIGATORS:**

| | | |
|---|---|---|
| AWADALLA Phillip | DRAPEAU Pierre | L'ESPERANCE Paul |
| BOUVIER Michel | DUBÉ Marie-Pierre | MICHAUD Jacques |
| CHOUINARD Sylvain | **KNOPPERS Bartha Maria** | SAMUELS Mark |

**COLLABORATOR:**

**BUCCI Lucie Marisa**

The overall goal of this project is to create a multidisciplinary team to **develop strategies** for uptake of nanotechnology-derived data sets. Specifically, our scientific interest and model is the **identification and validation of genes that cause, or predispose to, brain diseases.** Our team will develop the required approaches for interpretation and follow-up of the genetic information arising from nanosequencing in human patients. We will use Tourette Syndrome (TS) as the major model brain disease for implementation of these goals, which can then be extended to other brain disorders.

The legal, ethical and socio-economic study will specifically:

- Review and address "intra-familial" communication issues. There are several barriers to communicating genetic information with family members including: the complexity of genomic information (there may be difficulty understanding genomic information, particularly if the results are inconclusive and the risk is uncertain); pre-conceived ideas within the family about TS; feelings of fear, guilt and resentment about the disorder and its hereditary component; and emotional or logistical distance between family members. Generating genetic information within a family may lead to psychosocial, physical, and financial difficulties and negatively affect family relationships. Awareness of genetic status is only a first step. Subsequent steps come up against the following issues: How will the information affect other members of the family and the family as a whole? Who will have access to this information? How will it be used? Is there a moral or legal duty to inform relatives of results?

- Improve education and the development of knowledge in ethical, social and legal issues by disseminating information to policy makers, researchers and the public. We have a number of knowledge exchange activities that are relevant to this research.

## 7. CIHR Team of Prediction and Communication of Familial Risks of Breast Cancer (INHERIT)

Canadian Institutes for Health Research (CIHR)
October 2008 – October 2013

**PRINCIPAL INVESTIGATOR:**

SIMARD Jacques

**CO-INVESTIGATORS:**

| | | |
|---|---|---|
| AMARA Nabil | EASTON Douglas | LESPÉRANCE Bernard |
| ANDRULIS Irene | GLENDON Greg | MAUGARD Christine M |
| ANTONIOU Antonis | GOLDGERG Mark | OUIMET Mathieu |
| **AVARD Denise** | GOLDGAR David Elliot | PLANTE Marie |
| BRIDGE Peter | KIM-SING Charmine | SINILNIKOVA Olga |
| CHIQUETTE Jocelyn | **KNOPPERS Bartha Maria** | SINNETT Daniel |
| DORVAL Michel | LAFRAMBOISE Rachel | TAVTIGIAN Sean |
| DUROCHER Francine | LANDRY Réjean | |

**COLLABORATORS:**

**BLACK Lee**

**LEVESQUE Emmanuelle**

The overarching goal of this project is to thoroughly evaluate the prediction of breast cancer risk and its communication to individuals with a family history of breast cancer. The 4 components of the program are designed:

- To determine both the contribution of uncommon or rare intermediate-risk variants in selected candidate genes to the genetic population attributable fraction for the familial relative risk of breast cancer, and to assess the robustness of these risks in women ascertained through clinic-based and population-based studies.
- To improve the estimation of breast and ovarian cancer risks associated with mutations in BRCA1 and BRCA2 genes by identifying the genetic and environmental modifiers that may influence these risks.
- To integrate the knowledge of newly identified genetic and environmental factors, as well as biological markers, in breast cancer risk prediction models providing individual risk estimates.

- To assess the communication of risk prediction information by health professionals and the impact of such information on women at moderate to high risk of the disease.

We expect this program to identify specific genetic factors contributing to breast cancer susceptibility, in particular those factors which are currently poorly explored, uncommon or rare intermediate-risk variants that could potentially explain a significant proportion of missing familial risk of breast cancer. Some of these could enter clinical cancer genetics practice in the foreseeable future.

## 8. Integrated Research Network in Perinatalogy of Quebec and Eastern Ontario (IRNPQ)

Canadian Institutes for Health Research (CIHR)
September 2008 – March 2013

**PRINCIPAL INVESTIGATOR:**

FRASER William

**CO-INVESTIGATORS:**

| | | |
|---|---|---|
| **AVARD Denise** | MOUTQUIN Jean-Marie | SOMERVILLE Margaret |
| DUBOIS Lise | MUCKEL Gina | TRASLER Jacquetta |
| LUO Zhong-Cheng | SEQUIN Jean | TREMBLAY Richard E |
| MICHAUD Jacques | | |

**COLLABORATOR:**

**LEVESQUE Emmanuelle**

The mission of this multi-institutional network and its transdisciplinary research programme is to serve as a catalyst:

- to enhance the quality and impact of perinatal research in Quebec and in Canada;
- to train the next generation of researchers in an environment that reflects CIHR's four pillars; and
- to create an innovative regional/provincial clinical research model ensuring evidence-based care.

Important knowledge gaps concerning the long-term impact of various adverse exposures (environmental or genetic) during pregnancy on the health of future generations are due to the lack of prospective clinical research transcending obstetrics (pregnancy) into the neonatal and pediatric years.

## 9. Canadian Pharmacogenomics Network for Drug Safety (CPNDS)

Canadian Health Research Institutes (CIHR)
July 2008 – July 2013

**PRINCIPAL INVESTIGATORS:**

HAYDEN M.R.
CARLETON B

**CO-INVESTIGATORS:**

| | | |
|---|---|---|
| **AVARD Denise** | MARSHALL Deborah | RASSEKH Rod |
| DUBE Marie Pierre | MITTON Craig | RIDER Michael |
| KOREN Gideon | POOLE Robert | ROGERS Paul |
| LEEDER Steven | PHILIPS Michael | WASSERMAN Wyeth |
| MACLEOD Stuart | | |

**COLLABORATORS:**

**JOLY Yann**
**SAMUEL Julie**
**RAMOS-PAQUES Emma**

The Canadian Pharmacogenomics Network for Drug Safety project is a nation-wide sustainable research program that aims to identify genetic markers that are predictive of severe adverse drug reactions (ADRs) and integrate them into cost-effective diagnostic screening programs to reduce the occurrence of permanent disability and deaths from severe ADRs.

Researchers at the Centre of Genomics and Policy at McGill University investigate whether the Canadian regulatory approval process applicable to new pharmacogenomic drugs and tests is efficient and ethical or if there is a need for a fast track application process for this type of research.

## 10. Genomics and Proteomics Platforms for Vaccines and Immune Therapeutics Discovery and Development

Genome Quebec
May 2008 – May 2010

**PRINCIPAL INVESTIGATOR:**

SÉKALY Rafick-Pierre

**CO-INVESTIGATORS:**

| | | |
|---|---|---|
| AUCLAIR Claude | JOLY Yann | SHOUKRY Naglaa |
| **AVARD Denise** | KOFF Wayne | SOMOGYI Roland |
| BALDERAS Robert | MASSÉ Robert | TARTAGLIA Jim |
| BRUNEAU Julie | NICOLETTE Charles | VINCELETTE Jean |
| EL HADDAD Elias | ROUTY Jean-Pierre | WILKINSON Peter |
| GRELLER Larry | | |

**COLLABORATOR:**

**ABDUL-RAHMAN Ma'n H**

The SARS epidemic in Canada, the emergence of several other viral diseases such as West Nile and Avian flu, as well as the threat of a new Influenza pandemic in the near future have highlighted the need to develop a rapid system to produce safe and effective vaccines.

Findings emanating from this project contribute towards increasing the Canadian expertise and ensuring its dissemination worldwide and particularly in developing countries.

The Genomics and Proteomics platforms brings together a strong team composed of academic researchers in Canada, Africa, Europe and South America, plus clinicians and industrial partners with multidisciplinary and complementary expertise.

## 11. The Stem Cell Research Environment: Drawing the Evidence and Experience Together

Stem Cell Network (SCN)
2008-2011

**PRINCIPAL INVESTIGATOR:**

CAULFIELD Timothy

**CO-INVESTIGATORS:**

**KNOPPERS Bartha Maria**
MCDONALD Michael
EINSIEDEL Edna

**COLLABORATOR:**

**ISASI Rosario**

This project analyses how ethical, legal and social issues (ELSI) affect the conduct and direction of stem cell research (SCR), as well as the translation of the research into public policy.

In this phase of studies, we utilize new and existing legal and social science research tools to carry out a more nuanced analysis of the complex institutional and social structures that have emerged around SCR – from the research networks to special funding and regulatory mechanisms. The immediate goal of this work is to inform research policy, such as the upcoming review of Canada's embryo research legislation, the *Assisted Human Reproduction Act* (AHRA). More broadly, this work produces the following: data describing the nature and impact of the research environments; policy recommendations and best practices; innovative research tools to measure the return on investment in SCR and other biomedical research; consensus statements on policy positions; and ELSI guidance to various stakeholders, including members of the Stem Cell Network, research ethics boards, clinical trial committees, and the public.

Specific Project Objectives include:

- providing a detailed description and analysis of the international SCR environment, thereby informing international collaboration and policy development, while serving as a research resource;
- providing an analysis of the impact of various variables (e.g., research ethics rules, IP policy, availability of funds) on the nature and direction of SCR, thereby informing the development of best practices, commercialization policy and measurements of return on investment;
- providing an analysis of, and recommendations concerning, public representations of SCR. This will inform the development of best practice guidelines for the communication of developments in SCR, as well as for the reception of popular messages about the research by policymakers. It will also proactively inform regulatory bodies by developing normative ethical guidelines for resolving challenges to, and controversies posed by, emerging SCR processes and techniques. Finally, this will directly inform the review of the AHRA in 2009.

## 12. Material Transfer Agreements (MTA's)

Canadian Institutes for Health Research (CIHR)
2008-2010

**PRINCIPAL INVESTIGATOR:**

**KNOPPERS Bartha Maria**

**COLLABORATOR:**

PATHMASIRI Saminda

- This study focuses on how publicly funded infrastructures (DNA repositories) can use a Uniform Material Transfer Agreement (UMTA) to: better define and respect the rights of participants, funding agencies and potential researchers;
- preserve the open-source approach of publicly funded infrastructures;

- preserve the patentability and licensing of resulting intellectual property;
- clearly define research limits in light of public purpose and the participant's informed consent;
- establish national and international transfer modalities for biological samples and data; and
- define related risks and responsibilities.

A proper analysis of these aspects may reveal that MTAs can, at least provisionally, in the absence of specific legislation, be used as a flexible and efficient tool in the context of research infrastructures improving predictability, public protection (and incidentally confidence), decreasing negotiation delays and setting important standards in the field. It is hoped that these proposed standards will influence the private sector as well.

## 13. European Network for Genetic and Genomic Epidemiology (ENGAGE)

European Commission under the 7th Framework Program
January 2008-December 2012

**PRINCIPAL INVESTIGATOR:**

PELTONEN Leena

**CO-INVESTIGATORS:**

| | | |
|---|---|---|
| BRAZMA Alvis | KAPRIO Jaakko | PEDERSEN Nancy |
| ESTIVILL Xavier | **KNOPPERS Bartha Maria** | STEFANSSON Kari |
| GROOP Leif | MCCARTHY Mark | VAN OMMEN Gert-Jan |
| HARRIS Jennifer | | |

**COLLABORATOR:**

**TASSÉ Anne Marie**

**CHAGNON Annie**

The ENGAGE consortium unites 22 research organizations and two biotechnology companies. Partners of ENGAGE provide access to some of the most complete cohorts in Europe, in order to translate the richness of information obtained from some of the most important genetics and genomics research projects on European cohorts (and others) to information relevant to clinical applications. In order to do so, ENGAGE aims to regroup close to 80,000 association studies of genomic characteristics, to identify new medical susceptibility variants, presently unidentifiable through individual studies.

## 14. Communication of Hereditary Breast Cancer Risk Information with Families: Ethical and Legal Framework

Canadian Breast Cancer Research Alliance (CBCRA)
July 2007 – June 2010

**PRINCIPAL INVESTIGATOR:**
**KNOPPERS Bartha Maria**

**CO-INVESTIGATOR**
**AVARD Denise**

**COLLABORATORS:**
**BLACK Lee**
**MCCLELLAN Kelly**

The CBCRA is Canada's primary granting agency for breast cancer research and plays a national leadership role in setting priorities and directions for breast cancer research.

Breast cancer (BC) and cancers "run in families." Diagnosing a breast cancer gene in an individual implies that other members of the family may be affected. During the INHERIT BRCAs project, funded by Canadian Institutes of Health Research (CIHR), we investigated the ethical and legal implications for professionals of communicating research results to participants, recontacting patients and warning relatives.

This study follows previous research on the legal and ethical obligations associated with the communication of genetic information. It investigates whether family members have different obligations than other parties (e.g. doctors, "third parties" who are interested in the results but are not directly involved) in communicating breast cancer information.

Using our results, we evaluate current legal obligations in Canada, make suggestions about how (and if) this communication should take place, and determine participants' opinions about these ethical and legal issues.

## 15. Nanomedicine: Whither Policy and Regulation

Social Sciences and Humanities Research Council of Canada
April 2007 – March 2010

**PRINCIPAL INVESTIGATOR:**
**KNOPPERS Bartha Maria**

**COLLABORATORS:**
**BUCCI Lucie Marisa**
SHEREMETA LORRAINE

This research project explores the relevance of the current framework for the regulation of nanomedicine products falling under the purview of Health Canada's Health Products and Food Branch. The main objectives of this project are to:

- identify the salient issues and statutory and regulatory gaps associated with the current approval process for nanomedicine products (e.g. drugs, medical devices, natural health products and cosmetics);
- identify, describe and analyse the legal and regulatory requirements considered and/or employed in other jurisdictions to address these issues and gaps identified earlier;
- compare the legal and regulatory requirements considered and/or employed in other jurisdictions with the Canadian framework; and
- identify and frame policy options for the regulation of nanomedicine for consideration by Canadian policy-makers;
- communicate the themes and goals of this research to the broader publics.

## 16. CARTaGENE

Genome Canada and Genome Québec
2007-2010

**PRINCIPAL INVESTIGATOR:**

**KNOPPERS Bartha Maria**

**CO-INVESTIGATORS:**

| | | |
|---|---|---|
| LABERGE Claude | GAUDET Daniel | AWADALLA Philip |
| GODARD Béatrice | FORTIER Isabel | |

**COLLABORATORS:**

**BÉDARD Karine**

**ABDUL-RAHMAN Ma'n H**

CARTaGENE creates a resource for the advancement of genetic research, with the aim of improving the health of Quebecers. This public resource operates under a governance framework and consists of a databank and a biobank of 20,000 individuals. The database contains environmental, demographic and health data. The biobank contains DNA, blood and urine samples. More information can be found at www.cartagene.qc.ca.

## 17. Canadian Partnership for Tomorrow Project (CPTP)

Genome Canada and Genome Québec
2007-2010

**PRINCIPAL INVESTIGATORS:**

| | | |
|---|---|---|
| **KNOPPERS Bartha Maria** | GALLAGHER Richard | PARKER Louise |
| BORUGIAN Marilyn | ROBSON Paula J. | McLAUGHLIN John |

The Canadian Partnership for Tomorrow Project (CPTP) is an organization that aims to instigate new knowledge and accelerate the implementation of existing knowledge about cancer control across Canada by studying the relationship between genetics, lifestyle and the environment. CARTaGENE, as well as four other large Canadian prospective studies, are recruiting 300,000 participants nation-wide between the years 2009-2012.

## 18. Maternal-Infant Research on Environmental Chemicals: A National Profile of In Utero and Lactational Exposure (MIREC)

Instituts de recherche en santé du Canada (IRSC)
Septembre 2006 – Mars 2012

**PRINCIPAL INVESTIGATORS:**

ARDUCKLE Tye
FRASER William

**CO-INVESTIGATORS:**

| | | |
|---|---|---|
| **AVARD Denise** | LEGRAND Melissa | TITTLEMIER Sheryl |
| COCKELL Kevin | LUO Zhong Cheng | VILLENEUVE Maya |
| ETTINGER Adrienne | MITCHELL Grant | VINCENT Renaud |
| KUMARATHASAN Premkumari | | PLATT Robert |
| WEBER Jean-Philippe | | |

**COLLABORATOR:**

**LÉVESQUE Emmanuelle**
**DAM Amy**

MIREC is a national five-year research study that is recruiting about 2,000 women from the following cities: Vancouver, Calgary, Winnipeg, Sudbury, Ottawa, Kingston, Hamilton, Toronto, Montreal and Halifax. Women will be

recruited during the first trimester of pregnancy and followed through pregnancy, up to eight weeks after birth. The main goals of this study are:

- to measure the extent to which pregnant women and their babies are exposed to environmental chemicals and tobacco smoke;
- to assess what pregnancy health risks, if any, are associated with exposure to heavy metals (lead, mercury, cadmium, arsenic and manganese);
- to measure the levels of environmental chemicals and some of the beneficial components (nutritional and immune constituents) of breast milk.

## 19. International Stem Cell Forum Ethics Working Party

International Stem Cell Forum, MRC, CIHR
2004-2010

**PRINCIPAL INVESTIGATOR:**

**KNOPPERS Bartha Maria**

**CO-INVESTIGATORS:**

| | | |
|---|---|---|
| MORRIS Clive | RAGER Bracha | PENG Lee Hin |
| LOMAX Geoffrey | MURRAY Thomas | KIM Ock-Joo |
| ZENG Fanyi | PERRY Margery | WAHLSTROM Jan |
| ZHOU Qi | SIPP Douglas | RICHARDSON Genevra |
| TANNER Klaus | | |

**COLLABORATOR:**

**ISASI Rosario**

Scientific collaboration is a key aspect of the globalization of research. It is essential for the feasibility of any international collaborative project such as the International Stem Cell Forum (ISCF). The ISCF is composed of twenty-one partners and research funding institutions from around the world. It faces the challenge of conflicting regulatory and policy approaches regarding the exchange of materials and data adopted by its various partners. The divergent policy frameworks and governing regulations affect the permissibility of conducting stem cell research (i.e. procurement, derivation, banking, distribution and use of stem cell lines) and thus, they could potentially inhibit collaboration at the national and international level.

The Ethics Working Party (EWP) initiative was set up on behalf of the International Stem Cell Forum by its Canadian member organization, the Canadian Institutes of Health Research (CIHR). The EWP is comprised of

independent experts in the area, who are appointed by the Forum's member organizations. It is chaired by Dr. Bartha Maria Knoppers and its Secretariat is housed at the CGP in the Genome Innovation Centre of the Faculty of Medicine of McGill University. The primary purpose of the Ethics Working Party is to assist member countries to undertake stem cell research within a transparent and well-considered ethical framework. The EWP seeks to identify prospective strategies to foster the scientific and ethical integrity of research in a global context.

## 20. Developing Best Practices for Research Involving Children and Adolescents

National Council on Ethics in Human Research (NCEHR)
2009-2010

**PRINCIPAL INVESTIGATORS:**

**AVARD Denise**

**KNOPPERS Bartha Maria**

**CO-INVESTIGATORS:**

| | | |
|---|---|---|
| BOIVIN Michel | DESFAITS Anne-Cécile | UNKER Anne |
| BOWER Tara | DUBOIS-FLYNN Geneviève J | |
| WALTON Nancy | CARPENTIER Richard | |

**COLLABORATOR:**

**SAMUËL Julie**

The National Council on Ethics in Human Research (NCEHR) project is to study the ethical and legal issues of research involving children and adolescents in four areas:

- Genetic Research;
- Pharmaceutical Research;
- Longitudinal Studies; and
- Palliative Care Research.

This project is orchestrated through NCEHR's Emerging Issues Analysis Committee, and operated in collaboration with the Centre of Genomics and Policy, CIHR's Institute of Human Development, Child and Youth Health (IHDCYH) the CIHR Ethics Office, Health Canada, and other key organizations such as the Maternal, Infant, Child, Youth, Research Network (MICYRN).

The main goals of this project are:

- to identify the issues that have emerged since the publication of the Report on Research Involving Children in 1993;

- to identify the new policies and guidelines on research involving children;
- to build a consensus among policy-makers, researchers, and health professionals on the best practices; and
- to develop a durable partnership in policy and practice with regards to research involving children and adolescents.

## 21. Informing Evidence-Based Policy Expanded Newborn Screening

Canadian Institutes for Health Research (CIHR)
2008-2011

**PRINCIPAL INVESTIGATOR:**

MILLER Fiona

**CO-INVESTIGATORS:**

| | | |
|---|---|---|
| ALLANSON Judith Elizabeth | | CHAKRABORTY Pranesh |
| KLITTLE Julian | **AVARD Denise** | GIGUERE Yves |
| WILSON Brenda Joyce | CARROLL June C | HAYEEMS Robin A |

**COLLABORATORS:**

**SAMUËL Julie**

**KLEIDERMAN Erika**

This project proposes empirical research in two Canadian jurisdictions, Ontario and Quebec, which have addressed the challenge of expanded newborn screening quite differently. The main objectives of this project are:

- to gain empirical and theoretical insight into the perceptions and expectations of parents and health professionals with regards to the governance of newborn screening programs, the option-provision, and the provision of information and/or decision support to parents;
- to develop and conduct initial validation of an aid for parents; and
    - to explore ways of enhancing the capacity of public health services and clinical and public health professionals to provide information and support to parents prior to the newborn screening test. This project also includes a background literature review on: informed choice in newborn screening;
    - perceptions regarding storage and secondary use of newborn screening dried blood spots for research; and
    - the roles and responsibilities of clinical and public health professionals in the provision of information and decisional support for parents.

## 22. P3G - Public Population Project in Genomics

Genome Canada and Genome Quebec
2007 – 2010

**PRINCIPAL INVESTIGATOR:**

**KNOPPERS Bartha Maria**
HUDSON Thomas J.

**CO-INVESTIGATORS:**

| | | |
|---|---|---|
| LABERGE Claude | DESCHÊNES Mylène | FORTIER Isabel |
| BURTON Paul | PELTONEN Leena | LITTLE Julian |
| OLLIER William | METSPALU Andrès | FERRETI Vincent |
| KHOURY Muin | PALMER Lyle | |

**Collaborator:**

REGNIER Marie-Hélène

The Public Population Project in Genomics (P3G) is a not-for-profit international consortium which seeks to promote collaboration between researchers in the field of population genomics.

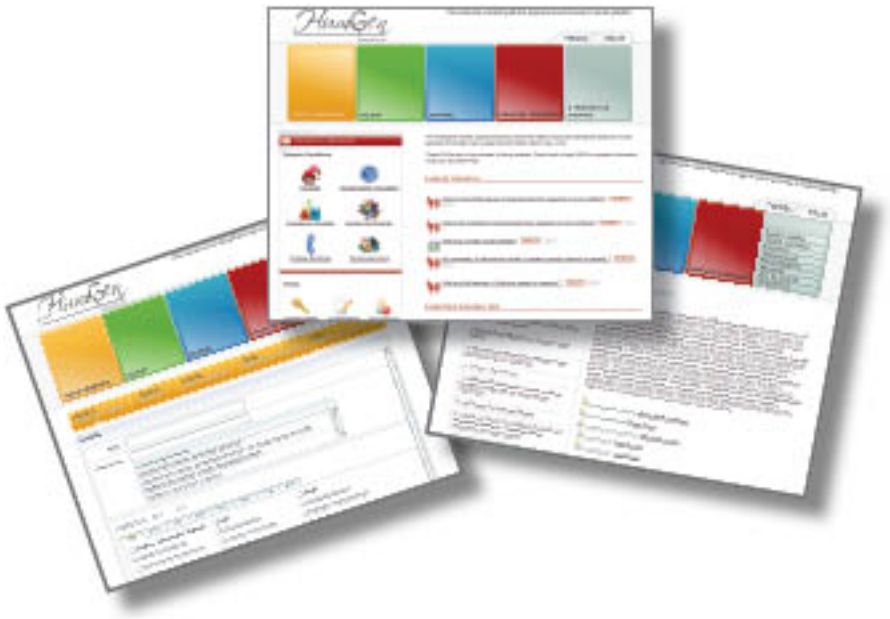
It has been launched in order to provide the international population genomics community with the resources, tools and know-how to facilitate data management for improved methods of knowledge transfer and sharing. Its main objective consists in the creation of open, public and accessible tools for international collaboration between population biobanks knowledge database. Its motto is transparency and collaboration.

# 4. HumGen - Database

HumGen International is the central "work-tool" and also the most valued product of the CGP (www.humgen.org). The HumGen website/database incorporates 5 specialized modules: PopGen, PediaGen, StemGen, IPGEN and DTCGen. HumGen International was created for the dissemination of information relevant to policy-making, the goal being to make the information accessible to as many people as possible, by removing geographic and financial barriers.

In 2009 the HumGen website attracted a total of 177,647 users, with an average of 486 users per day (60% of all requests are from outside Canada).This server also acts as a mirror site for the mega search engine http://bioportal.graphint.org/bioportal/, developed by Dr. Knopper's team. It enables data searches on HumGen (McGill University), the participating institutions at Cambridge University, U.K. (Public Health Genomics at the University of Cambridge) and the Public Health Genomics Population database at the CDC in Atlanta, U.SA.

**HumGen**:



With HumGen, these are also special thematic modules of databases of information in selected topics. These modules regroup both policy and literature.

**PopGen**: a database of international, regional and national laws and policies as well as a database of literature addressing population genetics research and

biobanks, grouped under 7 main themes: Access - Consent - Benefit Sharing - Commercialization - Governance - Communication of Results – Confidentiality**.**

**PediaGen**: a database of international, regional and national laws and policies, relevant to both genetic research and their clinical applications involving children, as well as a selective review of literature, grouped under 6 main themes: Banking - Research - Gene Therapy - Genetic Testing - Genetic Screening – Pharmacogenomics

**StemGen**: a database of international, regional and national laws and policies and selected literature concerning the socio-ethical and legal aspects of stem cell research and related therapies. A unique feature of StemGen is the STEM CELL WORLD MAP, which describes the policy approaches adopted in over 50 countries.

**IPGen**: a database of international, regional and national laws and policies addressing issues concerning intellectual property and human genetics. It also includes a literature section grouped under five main themes in intellectual property:

- property,
- commercialization,
- benefit sharing,
- patents, and
- copyright.

# 5. Team Publications 2009

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