CGP

Centre of Genomics and Policy
Centre de génomique et politiques

ANNUAL REPORT 2012

McGill

HumGen International

ANNUAL REPORT 2012

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A MESSAGE FROM THE DIRECTOR

Welcome to the Centre of Genomics and Policy's (CGP) 2012 Annual Report.

This year's Annual Report aims to showcase the CGP's work, people, and values. We hope the document will allow readers to get to know the CGP's scholars and their projects, as well as why they do the work they do.

The 2012 Annual Report invites both the general reader and the specialist to learn about the Centre's projects and activities. Through features and a newly-designed format, we hope to paint a portrait of our work that is accessible to the lay reader, yet relevant to researchers, professionals, policy-makers, and students.

Several features of this year's Annual Report highlight the CGP's innovative research programme, as well as the people who make it happen. The Report provides details about CGP: its ongoing research projects, awards and recognition received by its members, and the team's publications. We are also proud to announce a new internship program in collaboration with McGill's Department of Human Genetics. Finally, both the HumGen and CGP websites have undergone a makeover, making it is easier to search for and mobilize knowledge.

We hope you will enjoy getting to know us and our work.

Bartha Maria Knoppers

ABOUT THE CENTRE OF GENOMICS AND POLICY

Located within the Genome Quebec Innovation Centre at McGill University, the CGP works at the crossroads of law, medicine, and public policy. Applying a multidisciplinary perspective and collaborating with national and international partners, the CGP analyzes the socio-ethical and legal norms influencing the promotion, prevention and protection of human health.

Currently, the CGP's research covers six areas of genomics and policy: stem cell research and therapies, pediatrics, privacy, cancer, intellectual property, and biobanks (population genetics). These domains are approached using three guiding foundations: 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.

CENTRE OF GENOMICS AND POLICY

CGP TEAM 2012

PROFESSOR
KNOPPERS Bartha Maria

ASSOCIATE PROFESSORS
AVARD Denise
OZDEMIR Vural

ASSISTANT PROFESSOR
JOLY Yann

ACADEMIC COORDINATOR
ZAWATI Ma'n H.

ACADEMIC ASSOCIATES
ALLEN Clarissa
BLACK Lee
GOK Erdal
ISASI Rosario
LÉVESQUE Emmanuelle
MILIUS Djims
NGUYEN Thu Minh
SÉNÉCAL Karine
TASSÉ Anne Marie

POST-DOC FELLOW
MCCLELLAN Kelly

ASSOCIATE MEMBER
GOLD Richard

SCIENTIFIC CONSULTANT
LABERGE Claude

RESEARCH ASSISTANTS
BESSO Annyck
CORKERY Sarah
DAINOW Susannah
DAM Amy
DOVE Edward
DWIVEDI Supriya
FIGLARZ Marissa
HÉTU Martin
KIM Jihyun Rosel
KIRBY Emily
KLEIDERMAN Erika
LONGO Cristina
NGUENG-FEZE Ida
OUELLETTE Jean-François
PAGE Madeline
PARRY David
REIS Clarice
RIOUX Amélie
SO Derek
SOYE Kaitlin
THOROGOOD Adrian
VARDATSIKOS George

INTERN
DOLMAN Lena

INVITED SCHOLARS
BORRY Pascal
HOWARD Heidi Carmen

SYSTEM ADMINISTRATOR
OLVERA Elena

ADMINISTRATORS
HOZYAN Rose-Marie
ROSSI Marisa
THORSEN Nadine

COMPLETED RESEARCH PROJECTS 2012

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

Canadian Institutes of Health Research (CIHR)
September 2006 – March 2012

PRINCIPAL INVESTIGATORS
ARDUCKLE Tye
FRASER William

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

ACADEMIC ASSOCIATES
LÉVESQUE Emmanuelle
ZAWATI Ma'n H.

RESEARCH ASSISTANT
DAM Amy

MIREC is a national five-year research study recruiting 2,000 pregnant 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, as well as 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.

European Network for Genetic and Genomic Epidemiology (ENGAGE)

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

PRINCIPAL INVESTIGATOR
MCCARTHY Mark

CO-INVESTIGATORS
BRAZMA Alvis
ESTIVILL Xavier
GROOP Leif
HARRIS Jennifer
KAPRIO Jaakko
KNOPPERS Bartha Maria
MCCARTHY Mark
PEDERSEN Nancy
STEFANSSON Kari
VANOMMEN Gert-Jan

ACADEMIC ASSOCIATE
TASSÉ Anne Marie

The ENGAGE consortium unites 22 researchers and two companies from the biotechnology sector to improve interoperability in some of the most complete cohorts in Europe. ENGAGE aims 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 is regrouping close to 80,000 association studies of genomic characteristics to identify new medical susceptibility variants that are presently unidentifiable through individual studies. The CGP specifically addresses societal issues raised by molecular epidemiological studies that translate retrospective data and analysis.

CIHR Team in Community Genetics (ECOGENE-21)

Canadian Institutes of Health Research (CIHR)
March 2008 – June 2012

PRINCIPAL INVESTIGATOR
GAUDET Daniel

CO-INVESTIGATORS
HAMET Pavel
HUDSON Thomas
KNOPPERS Bartha Maria
SAVITRI Sonia

ACADEMIC ASSOCIATES
LÉVESQUE Emmanuelle
SÉNÉCAL Karine

ECOGENE-21 is a cohesive, transdisciplinary initiative driven by the specific needs of communities. ECOGENE-21 builds upon previous, ongoing and emerging local, national and international projects and networks of expertise in community genetics. Its objectives are to develop, validate, apply and replicate new cost-effective technologies and screening strategies to prevent recessive diseases in founder communities and common diseases at the community level. ECOGENE-21 also strives to develop the expertise and platforms needed to generate, apply, validate, and replicate within and across communities, new knowledge from genomics research.

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

Stem Cell Network (SCN)
October 2008 – September 2012

PRINCIPAL INVESTIGATOR
CAULFIELD Timothy

CO-INVESTIGATORS
EINSIEDEL Edna
KNOPPERS Bartha Maria
MCDONALD Michael

ACADEMIC ASSOCIATE
ISASI Rosario

This project analyses how ethical, legal, and social issues (ELSIs) 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; international approaches; innovative research tools to measure the return on investment in SCR and other biomedical research; consensus statements on policy positions; and ELSI guidance for various stakeholders including members of the Stem Cell Network, research ethics boards, clinical trial committees, and the public.

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

Fonds de la recherche en santé du Québec (FRSQ)
July 2009 – June 2012

PRINCIPAL INVESTIGATOR
BATIST Gerald

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

ACADEMIC ASSOCIATE
BLACK Lee

RESEARCH STUDENT
KLEIDERMAN Erika

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.

Q-CROC aims to identify and validate biomarkers specific to clinical resistance to FOLFOX and bevacizumab, in the development of 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 project starts with identification and validation of biomarkers from candidates drawn from the literature or from the research labs of Q-CROC scientists. It continues with a search for new biomarkers and functional validation of the most promising of these in order to obtain insights into mechanisms of resistance. The ultimate goal is to speed the development of personalized medicine through better patient and treatment selection, and to generate new knowledge on therapeutic resistance in the clinical context.

Simple Microfluidic System for Rapid and Robust Identification of Pathogens by Real-Time PCR at Point-of-Care

Genome Québec
January 2011 – September 2012

PRINCIPAL INVESTIGATOR
BERGERON Michel G.

CO-INVESTIGATORS
BOUDREAU Denis

KNOPPERS Bartha Maria
VERES Teodor

ACADEMIC ASSOCIATE
ZAWATI Ma'n H.

RESEARCH STUDENT
HÉTU Martin

This project aims to develop and make available real-time PCR technology at point-of-care for rapid detection of infections. Our research concentrates on the legal and ethical issues associated with the use of such a diagnostic tool. More precisely, we undertake two research objectives. The first is to examine whether healthcare institutions in Québec have a duty to integrate effective new technologies - such as real-time PCR - into their services, especially in regard to the growing problem that are nosocomial infections. The second aims to study how the use of real-time PCR technology at point-of-care will affect the legal duties of healthcare professionals.

Anticipatory Governance of Uncertainty and Futures-in-the-Present: Mapping the Foresight Methods 1990-2010

Social Sciences and Humanities Research Council of Canada (SSHRC)
February 2011 - May 2012

PRINCIPAL INVESTIGATOR
OZDEMIR Vural

CO-INVESTIGATORS
FARAJ Samer A.
KNOPPERS Bartha Maria

RESEARCH ASSISTANT
CORKERY Sarah

Whether faced by a new technology, climate change or an environmental and economic crisis, societies -- as individuals, groups and communities -- need to develop a broad capacity early on to prepare for the future impacts of such transformative events. Efforts to control or respond to these ever present societal challenges have tended to focus on "prediction" or alternatively, creation of policies that envision a deterministic future. Yet, social events with long lasting impacts such as environmental change, military conflicts or technology innovations are often unpredictable by their very nature. The traditional "predict-and-control" framework is therefore inadequate for complex social change and transformative innovations.

Anticipatory governance is a new approach for managing the uncertainties posed by future(s) of innovations and the prospective understanding of transformative social changes in rapidly moving and dynamic fields. The goal of anticipatory governance is not to predict or forecast a singular future but to develop foresight on multiple possible future(s). It builds on the principles of incrementalism and collective learning, and explores the representations of alternative future(s) in the present as perceived by a diverse set of stakeholders, both expert and lay. As such, it signals a shift towards "looking at" rather than "looking into" the future(s). The aim of this study is to characterize the methodologies used in foresight and anticipatory governance analyses over the past decade and the context in which they have been utilized.

Finding of Rare Disease Genes in Canada (FORGE)

Genome Canada
March 2011 – August 2012

PRINCIPAL INVESTIGATORS
BOYCOTT Kym
FRIEDMAN Jan
MICHAUD Jacques

CO-INVESTIGATORS
AVARD Denise
KNOPPERS Bartha Maria

ACADEMIC ASSOCIATE
NGUYEN Thu Minh

RESEARCH STUDENT
REIS Clarice

FORGE Canada (Finding of Rare Disease Genes) is a national consortium of clinicians and scientists using next-generation sequencing technology to identify genes responsible for a wide spectrum of rare pediatric-onset disorders present in the Canadian population.

The Consortium brings together clinicians from all 21 Clinical Genetics Centres (representing every province) and internationally-recognized Canadian scientists with expertise in gene identification, with the infrastructure of the Genome Canada Science and Technology (GC S&T) Innovation Centres. International collaborations have been established with clinicians in 16 countries. Two nation-wide requests for proposals have resulted in 175 disorders that met FORGE criteria; 70 of these rare disorders have been selected for study over the 18 months of this project. These disorders range from those affecting single families, to disorders with 20+ patients from across Canada and internationally recruited through the FORGE network. We are establishing a national data coordination centre to streamline and improve existing large-scale sequence analysis tools and our GE3LS team is working toward national ethical guidelines for analyzing sequence data from entire genomes and for sharing results with families.

Making Connections IV – Biobanks: Pathways to Translation?

Canadian Institutes of Health Research (CIHR)
Genome Canada, Genome Quebec, Public Population Project in Genetics, Canada Research Chair in Law and Medicine
June 2011 – March 2012

PRINCIPAL INVESTIGATOR
KNOPPERS Bartha Maria

CO-INVESTIGATORS
KAYE Jane
O'DOHERTY Kieran
PULLMAN Daryl
STRANGER Mark

ACADEMIC ASSOCIATE
ZAWATI Ma'n H.

RESEARCH ASSISTANT
DAM Amy

Making Connections is a network that brings together expert scholars to address issues surrounding the governance of biobanks. In 2011, the fourth Making Connections' workshop was held in Montreal on October 10-11, just prior to the International Congress of Human Genetics Meeting.

The workshop had four objectives:
• Continue the expansion of the international biobanks governance network.
• Examine possible pathways towards an effective and proficient use of biobanks in translational research and clinical applications.
• Publish a guidance document on translational challenges and solutions for national and international stakeholders and widely disseminate it.
• Identify opportunities for funded research and policy project.

RADICAL – Rare Disease Consortium for Autosomal Loci

Department of Human Genetics, Faculty of Medicine, McGill University
June 2011-December 2012

PRINCIPAL INVESTIGATORS
MAJEWSKI Jacek
ROSENBLATT David

ACADEMIC ASSOCIATES
NGUYEN Thu Minh
ZAWATI Ma'n H.

RESEARCH ASSISTANT
THOROGOOD Adrian

CO-INVESTIGATOR
KNOPPERS Bartha Maria

One of the biggest current challenges in human genetics is the identification of genes and variants responsible for phenotypic variability and susceptibility to common genetic disorders. We postulate that rare Mendelian disorders should be used as a tool to identify phenotypically important genes. Historically, such disorders have been neglected, both because of their rarity and because of the difficulty in identifying the underlying genes and mutations. Current sequencing technologies have drastically altered the playing field. We can now sequence individual genomes at manageable costs. In this project, we propose to set up a high throughput sequencing platform to detect mutations underlying rare Mendelian disorders. We will also establish a bioinformatics pipeline for annotation, prioritization, and selection of the genetic variants that are most likely to be responsible for the observed phenotype. Our initial approach will rely on isolating and sequencing only the coding portions of the human genome, but we anticipate that within this funding period, the rapidly decreasing cost of sequencing will render our pipeline operational at a whole-genome scale. The goal is to create a seamless technology and informatics platform allowing us to detect disease causing mutations within a week of obtaining DNA samples. We will also create cell and animal models to validate and understand the causative role of selected mutations. We aim to establish a reputation and extend a network of collaborations to position McGill as a centre for mutations detection, screening, and - in the future – genetic testing.

Cultured Epithelial Autografts for the Treatment of Corneal Limbal Stem Cell Deficiencies

Network of Centres of Excellence
October 2011-September 2012

PRINCIPAL INVESTIGATOR
GERMAIN Lucie

CO-INVESTIGATORS
AUGER Francois
BAZIN Richard
KNOPPERS Bartha Maria

ACADEMIC ASSOCIATE
NGUYEN Thu Minh

This project will develop cell therapies for the treatment of corneal limbal stem cell deficiency ("LSCD"). LSCD is a severe disease caused by damage or depletion of the corneal stem cells in the limbus region of the eye following trauma or disease. Once the corneal stem cells are destroyed, the epithelial tissue on the surface of the cornea can no longer regenerate or heal spontaneously, resulting in chronic inflammation, conjunctivalization, and vision loss. To treat LSCD patients, we have developed a tissue engineering technique involving massive expansion of epithelial cells in vitro to produce epithelial sheets for autologous transplantation (cultured epithelial corneal autografts – "CECA"). After twenty years or more of experience using epidermal cultured autografts ("CEA") on burn patients for skin grafts, we have successfully demonstrated the effectiveness of CECA (animal models) and reconstructed human corneas in vitro (CECA). We are ready to obtain approval from Health Canada to begin CECA trials for the cornea. Our team of researchers, clinicians and ethical/legal experts will be the first in Canada to offer treatment for unilaterally blind or vision impaired patients suffering from LSCD.

Consent Protocols for UCB Donation and Banking Phase 1

Stem Cell Network / Canadian Blood Services
May 2012-December 2012

PRINCIPAL INVESTIGATOR
KNOPPERS Bartha Maria

CO-INVESTIGATORS
ALLAN David
EAVES Connie J.
HESLEGRAVE Ron J.
WALKER Mark
ZANDSTRA Peter

ACADEMIC ASSOCIATE
ISASI Rosario

RESEARCH ASSISTANT
DWIVEDI Supriya

The main objective of this project is to develop tools for optimizing access to ethically sourced umbilical cord blood (UCB) for research. UCB is considered a valuable source of stem cells for research and clinical applications. Furthmore, UCB transplantation is standard practice for the treatment of blood disorders and studies suggest their potential use for the development of novel blood and immune-based therapies. Canadian Blood Services (CBS) will establish and operate the national OneMatch Public Cord Blood Bank (OMPCBB) that will be accessible to Canadian and international patients. The OMPCBB will promote efforts that contribute to research and improved clinical care by making non bankable units available for research. In this context, the development of harmonized tools for UCB collection and uses that meets the needs of all stakeholders is critical. In partnership with CBS and other stakeholders, we will develop practical tools to enable ethical provenance of UCB stem cells as well as policy recommendations for UCB research (e.g. informed consent protocols, information pamphlets for donors and ethics review boards) to facilitate the interpretation of guidelines and ensure research is performed within a strong ethical and legal framework.

ONGOING RESEARCH PROJECTS 2012

ThéCell (Réseau de thérapie cellulaire et tissulaire)

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

PRINCIPAL INVESTIGATOR
GERMAIN Lucie

CO-INVESTIGATORS
AUGER François
BERTHOD François
KNOPPERS Bartha Maria
LAVERTY Sheila
ROUTY Jean-Pierre
ROY Denis-Claude
TANGUAY Jean-François

ACADEMIC ASSOCIATE
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 in 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 Maria

• 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

Integrated Research Network in Perinatalogy of Quebec and Eastern Ontario (IRNPQEO)

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

PRINCIPAL INVESTIGATOR
FRASER William

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

ACADEMIC ASSOCIATES
LÉVESQUE Emmanuelle
SÉNÉCAL Karine

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 (clinical, biomedical, health services, and population health); and
• To create an innovative regional/provincial clinical research model ensuring evidence-based care.
• To address 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 from obstetrics (pregnancy) into the neonatal and pediatric years.

Canadian Partnership for Tomorrow Project (CPTP)

Canadian Partnership Against Cancer
April 2009 – March 2013

PRINCIPAL INVESTIGATORS
AWADALLA Phillip
PALMER Lyle
PARKER Louise
ROBSON Paula J
SPINELLI John

ELSI & STANDING COMMITTEE CHAIR
KNOPPERS Bartha Maria

ACADEMIC ASSOCIATE
ZAWATI Ma'n H.

RESEARCH ASSISTANTS
RIOUX Amélie
THOROGOOD Adrian

The CPTP is a national research project that aims to generate new knowledge and accelerate the implementation of existing knowledge about cancer control across Canada by studying the relationships between genetics, lifestyle and the environment. CARTaGENE, as well as four other large Canadian prospective studies, are recruiting participants nation-wide.
The ELSI Standing Committee at the CGP builds the ELSI infrastructure of the CPTP platform. The goals are to bring together ELSI experts from each cohort and develop relevant policies, documents, and procedures that are needed either by the CPTP or by a specific cohort and to ensure the conformity of the platform with legislation and ethics guidelines so as to prospectively guide the cohorts. The ELSI Standing Committee mandate is broad. It ranges from developing interoperable recruitment and access policies and procedures to dealing with legal issues surrounding consent, privacy and data sharing, and proposing governance structures for the CPTP and supporting the cohorts.

Canadian Pharmacogenomics Network for Drug Safety (CPNDS)

Canadian Institutes of Health Research
January 2009-March 2013

PRINCIPAL INVESTIGATORS
CARELTON Bruce C.
HAYDEN Michael Reuben

CO-INVESTIGATORS
DUBÉ Marie-Pierre
JOLY Yann
KNOPPERS Bartha Maria
KOREN Gideon
LEVY Adrian R.
MITTON Craig R.
PHILLIPS Michael Ian
RIEDER Michael John
WASSERMAN Wyeth Wyk

Adverse drug reactions (ADRs) are in the fifth most frequent cause of death in North America North America. Half of these reactions are likely caused by inherited genetic differences and are currently considered 'unavoidable'. This project aims to understand why certain drugs are safe for use in some children but not in others and then to create easy to administer diagnostic tests that will predict and therefore prevent specific ADRs in children. To meet their goals, CPNDS is collecting data through a Canada-wide surveillance network that operates within nine of Canada's largest children's hospitals. They then perform large-scale genomic and proteomic studies to define genes critical for ADRs. These results are being used to help develop diagnostic tests to recognize a child's genetic fingerprint and allow personalized dosing recommendations to be implemented for commonly used drugs.

Maternal Infant Child & Youth Research Network (MICYRN)
Promoting Health Research Involving Children and Adolescents

MICYRN
March 2011-March 2013

PRINCIPAL INVESTIGATOR
JUNKER Anne

CO-INVESTIGATORS
AVARD Denise
KNOPPERS Bartha Maria

ACADEMIC ASSOCIATES
ALLEN Clarissa
SÉNÉCAL Karine
ZAWATI Ma'n H.

MICYRN brings together the 17 Canadian academic child / child-maternal health centres and research institutes in a multi-disciplinary national initiative committed to removing barriers and building capacity for the conduct of safe and high quality health research.
The CGP is involved in several projects with MICYRN, including: Practices for Health Research Involving Children and Adolescents, Paediatric Consent Best Practices, Research Harmonization, Newborn Bloodspots/Biobanks, Newborn Screening/WGS, and creating policy tools for a Paediatric Biobank Research Platform.

• Developing Best Practices in Pediatric Consent Forms: The analysis provides valuable insight and evidence into how consent forms address current ethical issues, reducing gaps and facilitating harmonized, yet contextualized, approaches to pediatric health research ethics.
• Evaluating a novel model for Canadian National Federated Ethics Review: Expert review of 5 new research protocols in order to test new ways of doing ethics review for pediatric research.
• P3G International Psediatric Biological Research Platform: The Pediatric Platform is meant to develop an online platform, hosted by P3G, providing research tools for researchers and REBs concerned with pediatric biobanking. Using model documents in informed consent, data sharing, and biobank governance, the platform facilitates harmonization, provides practical value for researchers, works to improve ELSI management in pediatric biobanking, and facilitates REB review.

International Cancer Genome Consortium/Data Access Compliance Office (DACO)

Ontario Institute for Cancer Research (OICR)
July 2009 – May 2013

PRINCIPAL INVESTIGATOR
HUDSON Tom (ICGC Secretariat)

CO-INVESTIGATOR
KNOPPERS Bartha Maria

DATA ACCESS COMPLIANCE OFFICER
JOLY Yann

ACADEMIC ASSOCIATE
MILIUS Djims

RESEARCH ASSISTANTS
DOVE Edward
NGUENG-FEZE Ida
SO Derek

The International Cancer Genome Consortium (ICGC) has been organized to launch and coordinate a large number of national cancer research projects that have the common aim of 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 terms and objectives.

Optimizing Public Cord Banking and Research in Canada Phase 2

Stem Cell Network
August 2012-July 2013

PRINCIPAL INVESTIGATOR
KNOPPERS Bartha Maria

CO-INVESTIGATORS
ALLAN David
EAVES Connie J.
HESLEGRAVE Ron J.
WALKER Mark
ZANDSTRA Peter

RESEARCH ASSOCIATE
ISASI Rosario

RESEARCH ASSISTANT
DWIVEDI Supriya

The main objective of this project is to develop tools for optimizing access to ethically sourced umbilical cord blood (UCB) for research. UCB is considered a valuable source of stem cells for research and clinical applications. Furthmore, UCB transplantation is standard practice for the treatment of blood disorders and studies suggest their potential use for the development of novel blood and immune-based therapies. Canadian Blood Services (CBS) will establish and operate the national OneMatch Public Cord Blood Bank (OMPCBB) that will be accessible to Canadian and international patients. The OMPCBB will promote efforts that contribute to research and improved clinical care by making non bankable units available for research. In this context, the development of harmonized tools for UCB collection and uses that meets the needs of all stakeholders is critical. In partnership with CBS and other stakeholders, we will develop practical tools to enable ethical provenance of UCB stem cells as well as policy recommendations for UCB research (e.g. informed consent protocols, information pamphlets for donors and ethics review boards) to facilitate the interpretation of ethical guidelines and ensure research is performed within a strong ethical and legal framework.

Returning Research Results of Pediatric Genomic Research to Participants

National Institutes of Health (NIH)
September 2011 – September 2013

PRINCIPAL INVESTIGATOR
WRIGHT CLAYTON Ellen

CO-INVESTIGATORS
KNOPPERS Bartha Maria
MCGUIRE Amy

ACADEMIC ASSOCIATE
ZAWATI Ma'n H.

RESEARCH ASSISTANTS
PARRY David
RIOUX Amélie

This project, in partnership with research groups in the United States, aims to determine what criteria should govern return of individual results in paediatric genomic research, using analysis of US law and international guidelines regarding decision making for and by minors as the foundation. This issue, which has received remarkably little attention, must be resolved if this research, which is vital to understanding the contributions of genetic variation to the health of children, is to proceed. In order to develop these criteria, the project will draw upon a host of ethical, legal, and sociocultural sources, as well as empirical data. The Centre of Genomics and Policy is analyzing international (including Canadian) policies and guidelines on the return of pediatric research results, and the US PI's are analyzing US legislation and case law that may impact on the return of pediatric research results.

The Terry Fox New Frontiers Program Project in Genomic Determinants of Childhood Leukemia

Canadian Institutes of Health Research (CIHR), Terry Fox Foundation
June 2010 – September 2013

PRINCIPAL INVESTIGATORS
AVARD Denise
AWADALLA Philip
KRAJINOVIC Maja
PASTINEN Tomi
SINNETT Daniel

CO-INVESTIGATORS
ABOU ELELA Sherif
ARROWSMITH Cheryl
KNOPPERS Bartha Maria
LAVERDIÈRE Caroline
MONTPETIT Alexandre
ROY-GAGNON M H

ACADEMIC ASSOCIATE
SÉNÉCAL Karine

This project aims to examine whole-genome sequence variations from a sample of childhood leukemia (ALL) patients with the following aims: 1) to identify novel sequence and structural variants in childhood ALL genomes; 2) to explore changes in gene expression associated with ALL by examining the transcriptome and allelic expression; 3) to assess the impact of selected genes on disease susceptibility and disease outcomes and investigate the functional significance of these genes in vitro; and 4) to translate the genetic discoveries into appropriate health care policy and services. These findings will ultimately lead to the development of powerful research and clinical tools that could improve detection, diagnosis and treatment of childhood leukemia.

The CGP aims to identify ethical, legal and social issues in the return of pediatrics research results. More specifically, our Centre is reviewing the ELSI implications of Genome-wide re-sequencing results on children and parents. This involves an analysis of international and national policy statements, the obligations and needs of researchers and health professionals regarding the return of research results, and the needs of families and their children. This analysis will culminate in the production of a comprehensive discussion document. The development of the discussion document also involves literature and policy review, interviews with healthcare providers and families, and collaboration with various stakeholders.

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
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The goal of this project is to create a multidisciplinary team to develop strategies for uptake of nanotechnology-derived data sets. The scientific interest of this project is the identification and validation of genes that cause or predispose to brain diseases. Our team is developing approaches for interpretation and follow-up of the genetic information arising from nanosequencing in human patients. Tourette Syndrome (TS) is used as the major model of a brain disease disorder for implementation of these goals.

Susceptibilité génétique au cancer du sein: échange international de données et discrimination génétique

Ministère du Développement Économique, Innovation et Exportation, Québec
June 2010 – September 2013

PRINCIPAL INVESTIGATOR
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The rules and policies of collaborative oncological gene-environment studies (COGS) as well as the laws and public policies applicable to the international exchange of genetic samples in the United Kingdom, Australia, Canada, Europe (confidentiality), and France (material transfer agreements and broad consent) will be systematically identified and analyzed by this project. Findings will be validated by consultation (teleconference or email) with experts from each of the selected countries or regions. Our analysis, complemented by a critical literature review, will allow us to outline the central similarities and differences between norms and produce recommendations aiming to harmonize the rules of EI-RFCS with those of COGS. Our second objective concerns the communication of information to third parties. We will carry out a review of the relevant law, regulations, and guidelines concerning medical liability and genetic discrimination in Quebec. This review is necessary to identify the actual practices of insurance companies in Quebec and to compare them to legal and ethical norms. We will analyze the forms available on the web-sites of personal insurance companies in Quebec (Desjardins, La Capitale, Groupe Promutuel, etc.). Norms will be organized according to the emergent issues, such as confidentiality, access, and discrimination. The contextual legal analysis of these issues will serve to identify those elements that would benefit from the issuance of recommendations addressing medical liability and genetic discrimination in Quebec.

CIHR Emerging Team in Genomics Screening

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

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CARROLL June
CASTLE David
CAULFIELD Timothy
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ETCHEGARY Holly
GRIMSHAW Jeremy
LEMEYRE Louise
LITTLE Julian
MILLER Fiona
MORIN Karine
POTTER Elizabeth
WELLS George

This CIHR Emerging Team examines two areas where genomics meets every day health care and general populations: 1) using family history information in predicting risk of common diseases and 2) screening newborns for rare but serious inborn metabolic diseases. The team is also engaging with citizen groups and health professional groups to understand their reactions to upcoming developments in 'genomic profiling', where DNA is used to assess susceptibility to common diseases in both adults and children.

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

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

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CHIQUETTE Jocelyn
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DUROCHER Francine
EASTON Douglas GLENDON Greg
GOLDGAR David Elliot
GOLDGERG Mark
JOLY Yann
KIM-SING Charmine
KNOPPERS Bartha Maria
LAFRAMBOISE Rachel
LANDRY Réjean
LESPÉRANCE Bernard
MAUGARD Christine M
OUIMET Mathieu
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POST-DOC FELLOW
McCLELLAN Kelly

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 the contribution of uncommon or rare intermediate-risk variants in selected candidate genes to the genetic population attributable fraction and the familial relative risk of breast cancer, and to assess the robustness of their risk in women ascertained through clinic-based and population-based studies.

•To improve 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 on newly identified genetic an environmental factors, as well as biologic markers, in breast cancer risk prediction models providing individual risk estimates.

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

This program is identifying specific genetic factors contributing to breast cancer susceptibility, in particular, yet poorly explored, uncommon or rare intermediate-risk variants that 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.

A Research and Knowledge Network on Genetic Health Services and Policy: Building on the APOGEE-Net and CanGeneTest Experiences

Canada Institutes of Health Research (CIHR)
November 2008 – November 2013

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Consent has long been considered as the crystallization of the researcher's duty to inform research participants. Indeed, providing consent is based on the right of participants to exercise full autonomy in decisions affecting their personal privacy. That being said, as the number of participants recruited in large-scale longitudinal studies – for example – grows, obtaining and maintaining consents will become increasingly onerous and complex. Hence, research studies are gradually using interactive, electronic media for consent procedures – which are seen as more accurate, dynamic and cost-effective. It is unclear, however, how and under what conditions such an approach will satisfy the legal and ethical requirements related to consent. Outcomes from this research will interest various stakeholders, including clinical researchers, health policy advisors, lawyers as well as technology and computer specialists, as it will promote the research-to-practice transition and provide preliminary data and guidelines for the legal and ethical design, implementation and approval of projects using e-consent procedures. More generally, the future use of e-consent will likely require that decision-makers provide guidelines and rules specifically addressing the role of new technologies in this field, thereby impacting the research, ethical and legal fields.

The Leucegene Project: Transcriptome Sequencing to Identify Novel Prognostic Markers and Therapeutic Targets in Acute Myeloid Leukemia

Genome Quebec
January 2011 – January 2014

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BOUVIER Michel
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The project proposes to identify novel prognostic markers and therapeutic targets in acute myeloid leukemia. The ELSI portion of this project, for which the Centre of Genomics and Policy has primary responsibility, analyzes the 2010 Tri-Council Policy Statement requirement that material incidental findings be disclosed to research participants. Of particular interest are how diligent must researchers be in looking for 'incidental findings', how should they be disclosed, and how is this obligation funded (or not). We will also be pursuing a qualitative study that will ask researchers about difficulties they have had with this obligation, whether it be know what to disclose, how to disclose it, or how to fund the staff to assist in the disclosure if the researcher is not qualified to do so.

Next Generation Predictive Signatures for Breast Cancer

Genome Quebec
January 2011 – January 2014

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The project proposes to develop a prognostic gene signature and validate a "made in Quebec" clinical test for ER-positive breast cancer patients. The ELSI portion of this project, for which the Centre of Genomics and Policy has primary responsibility, revolves around the barriers to the adoption of new technologies into clinical practice. We are focusing first on the educational factors that might limit uptake or understanding of these tests. That is, does the education received by physicians and nurses as part of primary or continuing education curricula adequately take into account the growth of genetic medicine in everyday medical practice? We will also be pursuing a qualitative study beginning in the spring of this year. For this, we will be interviewing or sending questionnaires to clinical care providers to obtain their input on what barriers exist for them, if any, for the use of genetic testing in their practices. From this we hope to develop a discussion paper on barriers to uptake of new genetic technologies, including educational and any other that arise from the qualitative study.

The Cartography of Intestinal Microbial Communities in a NHP Model System

Genome Quebec
January 2011 – January 2014

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The scientific objectives of this project are to evaluate in a nonhuman primate (NHP; vervet monkey) model differences in the microbiome of the gastro-intestinal tract at different locations, determine whether stool is a relevant material for microbiome studies, and assess how age, sex, genetics and diet influence the diversity of the microbiome at points along the gastrointestinal tract. Microbiome based therapies, namely fecal transplantation (FT) therapeutics, are thought to hold great potential for treatment of a number of disorders. However, potential FT based therapeutics raise GE3LS issues that are unique to microbiome research and implementation. The project aims to investigate the regulatory hurdles of FT based therapies, namely: 1) How would FT based therapeutics, as they are currently prepared and administered, be treated under Canadian and International legislative and regulatory regimes? 2) What are the legislative, regulatory, and/or local administrative hurdles that both current, and anticipated FT based therapeutics face in Canada and Internationally?

From the Lab to the Clinic: ELS Issues in Cancer Stem Cell Research

Cancer Stem Cell Consortium (CSCC)
May 2010-April 2014

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From the Lab to the Clinic: ELS Issues in Cancer Stem Cell Research is a project key to a larger Cancer Stem Cell Consortium (CSCC) funded initiative that aims to identify, characterize and develop methods of destroying leukemia stem cells (LSC). This latter research project involves three key phases of the innovation process: the use of a large tissue bank (basic research), the engagement of industry partners (commercialization) and the development of valuable therapeutics (translation) for patients with intractable leukemia. The ELS initiative led by HeaLS Research Director Timothy Caulfield explores the ethical, legal, social and policy issues affiliated with each realm of research associated with the innovative process. The team will be investigating ELSI challenges that characterize tissue banking and the commercialization process, in addition to those associated with the marketing of therapies. This latter component is being done through the lens of medical tourism.

Biobank Standardisation and Harmonisation for Research Excellence in the European Union (BioSHaRE-EU)

The European Commission
June 2010 – May 2014

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TASSÉ Anne Marie

BioSHaRE-EU has assembled a consortium of leading international researchers from all domains of biobanking science. These experts are working to develop and apply methods and tools that will provide a foundation for an ambitious program of harmonization and standardization in European biobanks and major biomedical studies. This facilitates the full participation of European bioscience in the next phase of international aetiological research that demands access to studies that have 3 complementary characteristics: 1) participants must be comprehensively assessed not only for genotype, but also for phenotype; 2) measurement quality must be high; and 3) because no single study will provide adequate numbers of subjects for certain questions, biobanks must therefore be harmonized and standardized so that studies can pool biobank data in valid and effective ways. The CGP is involved in the development of ethical, legal and social guidance in order to harmonize the treatment of environmental risk and personal life-style data in and from different European biobanks. The CGP also offers BioSHaRE its ethical expertise in issues of privacy and retrospective access to samples and data.

Réseau de médecine génétique appliquée (RMGA) Infrastructure

Fonds de la recherche en santé du Québec (FRSQ)
June 2010 – May 2014

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The RMGA is a network of multi- and trans-disciplinary researchers. Its objective is to support and develop basic and applied human genetic research by creating, managing, and promoting scientific infrastructures and knowledge transfer through health services and interventions, for the benefit of the Quebec population. The Network has close to 350 members, representing the majority of human genetics researchers in Quebec. The RMGA includes a Legal and Socio-Ethical Issues Infrastructure that considers those issues arising from secondary use of biological materials and data in health research.

Applied Metagenomics of the Watershed Microbiome

Genome BC, Genome Canada
July 2011 - June 2014

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Water quality is primarily assessed at the tap using coliform bacterial species as indicators of microbial pollution, a paradigm which reflects an anthropocentric focus on drinking water and human health. Metagenomics is a "culture-independent" method for analysis of multiple microbial genomes, for example, in the drinking water. Importantly, water safety and genomics together create a highly volatile postgenomics innovation trajectory for metagenomics applications in public health and ecosystem health.

This study comprises three stages. First, we will identify the metagenomics stakeholders for water safety in consultation with the water and metagenomics experts and through metagenomics and water safety document analyses and interviews. Second, the issues associated with a new watershed test that may impact each stakeholder group will be identified through complementary social science methodologies including document analyses, literature reviews, surveys, focus groups and interviews. Third, we will hold a multi-stakeholder workshop to present, negotiate and validate the identified social, legal, ethical and policy issues in the form of a "points-to-consider" document to inform prospective policy.

Design and Evaluation of Electronic Consent and Governance Processes for Clinical Research

Canadian Institutes of Health Research
February 2011-July 2014

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The objective of this study is to create Canadian recommendations and other resources for electronic consent for future research use of data and biological materials. Informed consent signals the primary duty to inform and protect research participants. Providing consent is based on the right of research participants to exercise full autonomy in decisions affecting their health and personal privacy. As technologies in medical research improve and research questions become increasingly complex, there is a need to recruit new participants, as well as use previously-collected data and biological materials to increase statistical power and minimize the burden on research populations. This application addresses the need for guidance in the use of research data and biological materials to answer questions that were not planned or known at the time of collection. This guidance will provide recommendations for the use of data and biological materials that have already been collected and provide recommendations and a model electronic consent process for data and biological materials that will be collected in the future.

Specifically we will: 1. Evaluate the feasibility of creating a digital governance system in Canada by identifying the legal and ethical issues relating to the use of an electronic approach to consent for future research use of data and biological materials; 2. Draft an Electronic Consent Governance Framework Report with actionable recommendations, and validate it with pertinent stakeholders (ethics boards, patient organizations); 3. Develop a model form for electronic consent, which could be adapted to different types of research and sites, across disease groups, and across populations with an emphasis on the needs of the aging population.

Translation Challenges, Science Policy and Stem Cell Research

Stem Cell Network
October 2011 – March 2015

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Intellectual property (IP) is perceived as playing an important role in the commercialization process, and the role of patents in particular has received a considerable amount of attention in the literature (Golden 2010). Our focus in this phase will be on the relationship between translation and commercialization pressure (including IP policies), and data access policies. We will investigate current restrictions to access, as reflected in (for example) international stem cell banking policies, including restrictions to future IP claims, and compare them with existing open access policies (e.g., UK Stem Cell Initiative and the CIRM iPS biobank). This work will include an analysis of whether certain commercialization policies, and concomitant IP approaches (such as restrictive patenting practices) do in fact conflict with emerging open access approaches and policies, as reflected, for example, in the UK Stem Cell Bank.

Subvention de formation des IRSC-FRSQ en médecine génétique apliquée

Canadian Institutes of Health Research
April 2009-March 2015

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SIMARD Jacques R.
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The objective of the RMGA is to develop the four key goals of its program: training workshops, Genetics Days, training scholarships, and a multi- and trans-disciplinary educational program through collaborative supervision. RMGA hopes to widen its scope and access to its training in specific domains that the scientific community and the general population both recognize as essential.

From Banking to International Governance: Fostering Innovation in Stem Cell Research

Networks of Centres of Excellence (NCE)
October 2011 – March 2015

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International initiatives are emerging to address harmonization and standardization processes for SCR and banking (e.g. International Society for Stem Cell Research (ISSCR) and the International Stem Cell Banking Initiative (ISCBI)). Until recently however, these efforts adopted an 'embryo-centric' approach, leaving behind other timely and promising sources (e.g. induced pluripotent stem (iPs) cells, cells derived from placentas, etc.).

While certain socio-ethical and legal (ELSI) concerns are specific to the nature of SCBs, can they thrive by applying the lessons learned in biobanking generally? To answer this, we will examine the current national and international SC banking landscape against the biobanking models for human tissues generally, with a view to evaluating existing governance, commercialization and regulatory frameworks and to proposing policy recommendations to increase the upstream understanding of the factors which encourage or hinder SC translation. We will develop "international governance models" and a "Points to Consider" thereby providing a wide range of stakeholders and receptors (e.g. researchers, SC bankers, policy-makers and the general public), with analyses, strategies and solutions for moving towards translational SCR within Canada and on the global stage. Furthermore, we will build capacity by training and mentoring future ELSI researchers.

Quebec Training Network in Perinatal Research (QTNPR)

Canadian Institutes of Health Research (CIHR)
April 2009 – May 2015

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The QTNPR network is creating a multidisciplinary curriculum on the impact of environmental exposures on maternal and child health. The objectives of QTNPR are to 1) provide trainees the knowledge, skills, and values that will allow them to address the complex interdisciplinary challenges of the current reproductive and perinatal health environment; 2) integrate into a single training network several research groups with complementary expertise in reproductive, perinatal, and infant health research; 3) link state-of-the-art, discipline specific teaching to crosscutting core competencies in the form of a transdisciplinary training grid; and 4) establish and maintain national and international partnerships with relevant complementary training programs.

International Stem Cell Forum Ethics Working Party (EWP)

International Stem Cell Forum, Medical Research Council (UK), Canadian Institutes of Health Research (CIHR)
April 2004 – May 2015

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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 health ministeries 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) affecting collaboration at the national and international levels.

The Ethics Working Party (EWP) initiative was set up on behalf of the International Stem Cell Forum by its Canadian member organization, the CIHR, and is now supported by the Canadian Stem Cell Network. The EWP is comprised of independent experts in the area, appointed by each of the Forum's member organizations. It is chaired by Dr. Bartha Maria Knoppers and its Secretariat is housed at the CGP in the Genome Quebec / McGill University Innovation Centre. 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.

Reconciling Law and Ethics with Open Science in Biotechnology Research

Fonds de la recherche en santé du Québec (FRSQ)
July 2011 – June 2015

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Do the current ethical and legal policies applicable to research with genomic databases sufficiently account for the new reality of open biotechnology? How could the current policy framework be improved to facilitate the transition to a more transparent, collaborative research context? Our research will investigate the impact of open biotechnology on research ethics and legal policies with a particular focus placed on informed consent (scope of consent, privacy, data ownership) to large open database projects. We will use a combination of quantitative and qualitative research strategies that will offer complementary applied legal and ethical data on the impact of open biotechnology on the governance of genomic research. The use of a common research methodology in all streams of the project will facilitate comparisons and integration of our results. Our methods will include comparative legal and ethical research (policy review, legal research), questionnaire analysis and focus group interviews. To validate our findings, we will engage stakeholders at the annual meetings of two major organizations involved in research with open databases: The Public Population Project in Genomics (P3G) and the International Cancer Genome Consortium (ICGC).

Towards Systems Medicine for Fatty Liver Disease

Fonds de la recherche en santé du Québec (FRSQ)
September 2011-September 2015

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POSNER Barry
ROZEN Rima
SLADEK Rob
VIDAL Silvia

This project focuses on three questions relating to a disease-specific biobank:

Given its current ethical framework, can the liver biobank re-contact research participants for updates to enrich the quality of the bank? If so, what measures will need to be taken to meet Canadian and international ethical standards?

How can a policy to return incidental findings be developed that takes advantage of a double-coding system of privacy protection but is streamlined and efficient?

What is the scientific, ethical, legal and policy framework applicable to the return of results in the context of a personalized medicine biobank project?

Risk Stratification for Prevention and Early Detection of Breast Cancer: Development and Implementation of Communication Tools

La fondation du cancer du sein du Québec
September 2012 – September 2016

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LESPÉRANCE Bernard

The goal of this project is to develop an integrated information campaign that aims to sensitize the population to the importance of considering family history to fight effectively against breast cancer. The campaign also aims to better equip health professionals to evaluate the risk of breast cancer on the basis of family history. This campaign will be realized through a rigourous process that will partner diverse professional and community associations. Information and sensitization tools will be developed and compiled into information toolkits. These toolkits will respond to three needs: to effectively collect, use, and share information on family history of breast cancer. The tools will allow users, for example, to answer the following questions: From whom should I obtain information? What kind of medical information do I need? How can I obtain this information from my family? With whom should I share the information I gather on breast cancer risk?

The present team is a group of experts in genetics, epidemiology, public health, psychosocial evaluation, ethics, and public law, all of whom possess a specific expertise in breast cancer.

Multidimensional Epigenomics Mapping Centre (EMC) at McGill

Canadian Institutes of Health Research
March 2012-February 2017

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SLADEK Robert Gaye
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TURECKI Gustavo Xavier

To join global efforts (The International Human Epigenome Consortium), we will establish an Epigenome Mapping Centre (EMC) at McGill University that builds upon a high-throughput sequencing infrastructure with a critical mass of expertise and technology available to contribute significantly in deciphering the functional code of the human genome. Our work is internationally coordinated and will support research initiatives across the Canadian research community. We apply epigenome mapping to understand interactions between environment and genome in human blood cells, to interpret diseases impacting metabolism using tissue samples and to study how epigenetic changes can alter function of the brain. EMC McGill is a national hub housing a critical mass of epigenomics expertise supported by state-of-the-art genomics infrastructure. Our integrated operation also includes a legal and ethics component. It will contribute to Canadian leadership in epigenome research in biomedicine.

Integrative Epigenomic Data Coordination Centre (EDCC) at McGill

Canadian Institutes of Health Research
March 2012-February 2017

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HALLETT Michael Trevor

This project proposes an integrative Epigenomic Data Coordination Centre (EDCC) at McGill, which will be a national hub to support data collection, processing, storage and dissemination for projects funded under the CEEHRC initiative and facilitate integration with the IHEC. The outcome will include data pipelines and tools using standardized formats and vocabularies for verification, validation, and analyses across the CEEHRC network. The EDCC McGill will also develop a framework that leverages Compute Canada national resources to support large-scale processing, sharing and visualization of epigenomics data. The platform will enable epigenetic researchers on a national level to query and exploit this valuable resource.

Framework for Decision-Making for Rare Diseases

Canadian Institutes of Health Research
March 2012-March 2017

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JOLY Yann
KLEIN Peter
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MARRA Carlo
MILLER Fiona
OZDEMIR Vural
SIRRS Sandra

As our understanding of diseases and how to treat them evolves, so too must our decision-making procedures for providing fair and cost effective treatments for those living with an illness. Today, one area of policy and decision-making in particular lags behind, that for treating rare diseases. At present there is no policy framework to help decision makers navigate the complex factors involved when making decisions about paying for orphan drugs. This project brings together a multidisciplinary team of experts in matters relating to treatment for rare diseases, and will incorporate input from the public and key stakeholders to develop such a framework. It will facilitate priority setting for orphan treatment decisions constrained by a limited budget, that considers the relevant developmental, clinical and economic factors and ethical principles, as well as being consistent with the values of society at large.

RECOGNITIONS AND AWARDS

The Centre for Genomics and Policy congratulates its scholars on the following special achievements.

Prof. Bartha Maria Knoppers

Prof. Bartha Maria Knoppers, Director of the Centre for Genomics and Policy, became an Officer of the Ordre national du Québec this year for her work in health law and ethics research and policy.

The Ordre cited her tireless engagement and international expertise on topical issues including biobanks, stem cell research, cloning, population research, assisted reproduction, rare diseases, pharmacogenomics and the future of public health. The awarding committee also highlighted her leadership roles in Genome Canada and Genome Quebec, in CARTaGENE, UNESCO, and in the Human Genome Organization (HUGO).

Prof. Yann Joly

This year, the CGP's Prof. Yann Joly was awarded the Quebec Bar Association's prestigious Mérite Innovations prize, designed for innovators in access to justice or judicial administration. The award recognizes individuals or organizations that have made advances in law related to specific issues in technological, communications, or administrative developments.

Prof. Joly was awarded the prize for his pioneering work in two key areas where privacy and health research law intersect: medical genetics and information technologies. Together with CGP colleagues, Prof. Joly developed normative solutions to protect privacy in health research for the International Cancer Genome Consortium (ICGC). He also prepared contracts for accessing individual data that aim to protect the identities of the ICGC's research participants.

In addition to the Mérite Innovations award, Prof. Joly has also received the distinction of Lawyer Emeritus from the Quebec Bar Association this year.

INTERVIEW WITH INTERN LENA DOLMAN

The Centre for Genomics and Policy inaugurated its academic internship program this year, in partnership with the McGill University Department of Human Genetics (HGEN 697, Advanced Readings in Human Genetics). Lena Dolman, the Centre's first intern, shared her thoughts about what participating in the internship means for her.

Lena Dolman

1.What drew you to ethical, legal and social questions in general and the internship in particular?

I am currently completing a Master's degree in Human Genetics at McGill University, in which I am studying the involvement of the BRCA2 gene in ovarian cancer. In contrast with the black-and-white nature of my laboratory molecular research, I have been drawn towards the ethical and social grey areas of genetics research, where issues are interdisciplinary, controversial, and occasionally inconclusive. As genetic testing for BRCA2 cancer susceptibility has considerable connections to these sorts of issues, I became interested in this internship as an opportunity to expand my understanding of the unresolved issues facing families with a history of cancer and the physicians treating them, as well as to develop skills in qualitative and legal research methodology.

2.What research are you pursuing? What has it revealed so far?

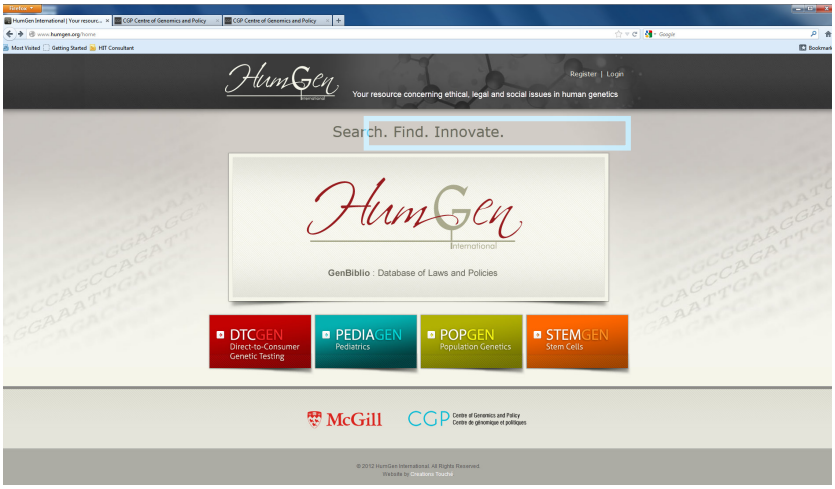
Through my internship, I am researching the highly unresolved issue of physician and patient duties to warn third parties (i.e. relatives) of genetic cancer risks identified during genetic testing. Looking specifically at the province of Quebec, my research has involved determining the extent to which patients communicate test results to their relatives, and what obligations apply to physicians in terms of warning these relatives when patients choose to withhold the information. My research has revealed an alarming deficit in intra-familial communication, particularly to relatives outside of the nuclear family. Furthermore, through analysis of recent Canadian case law and policy, my research has shown that physicians may not justifiably breach confidentiality in order to warn relatives of genetic risk, leaving the responsibility with the patients.

3.How will this internship impact your future career plans?

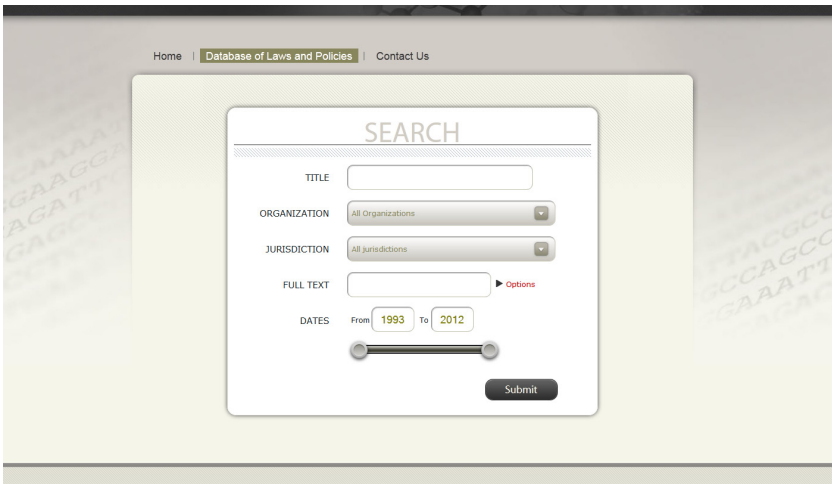
This internship has introduced me to the broad range of issues and challenges that inter-connect with medical practice and research. It has also allowed me to feel confident in approaching literature of a whole different kind, including qualitative studies, policy, and court judgements. As I am currently exploring options for continued academic study after my Master's (potentially in the area of global health), this internship has motivated me to seek out interdisciplinary fields that will allow me to incorporate considerations of socio-political, ethical, and legal concepts into health-related research. Most immediately, I will make use of the skills acquired at the CGP in a short-term research project on cancer screening policy that I will be conducting with the International Agency for Research on Cancer, a part of the World Health Organization), in summer 2013.

HUMGEN – DATABASE

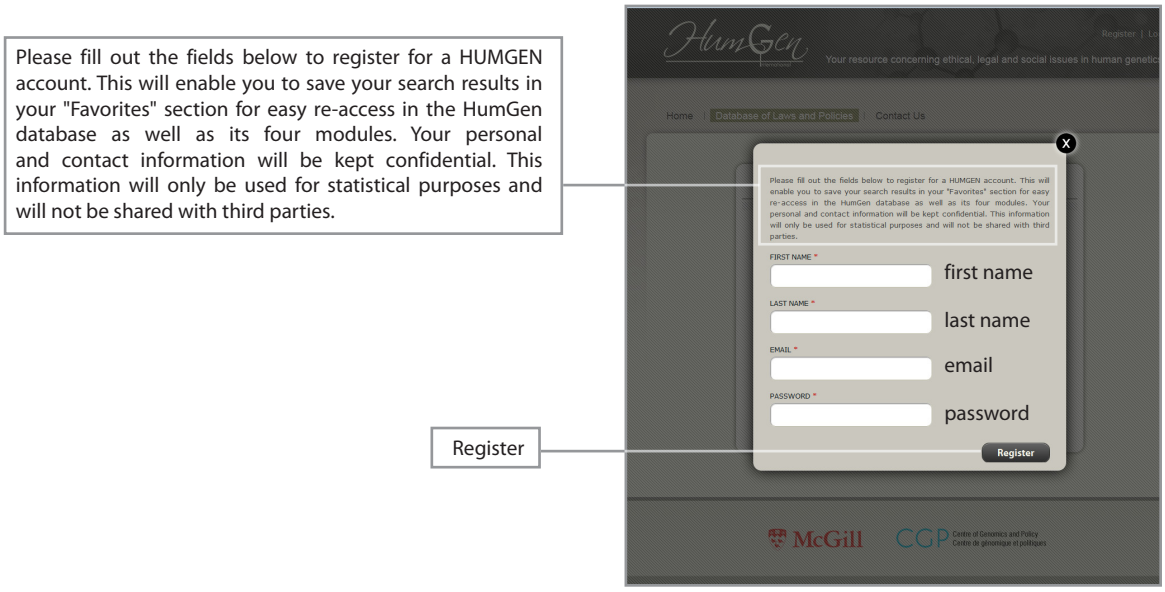
In 2012, the Centre for Genomics and Policy optimized its HumGen international database search engine to promote online access to information on laws, policies, and guidelines in human genetics research. This year, the HumGen website has undergone a makeover to optimize searches through four modules in order to make it easier for users to conduct research into ethical, legal and social issues in human genetics, and to personalize the user's experience at the same time.



The newly-optimized search engine has been redesigned to facilitate access to normative documents (laws, policies and guidelines), and to word and phrase searches. Search results are displayed in four subsections of international, national, provincial, and regional documents, giving a sense of socio-geographical context to the findings. HumGen's new search functions make research easier to conduct, organize and follow international developments.



Registration is also open. It enables you, the user, to create and save a personalized favourites list. This function is useful for when you want to bookmark especially interesting results, or when you want to return to certain documents at a later date. The list is your creation within the site; indeed, the HumGen experience for the registered user has been personalized in several respects.



It is also easy to personalize the HumGen experience by sharing your findings with colleagues and friends. Click the "share" button at the bottom of a search result to send a link to the document to whomever you choose. A useful tool for study as well as for informal reading, the share feature is another aspect of HumGen's evolving role in ELSI research.

HumGen has been built to serve multiple audiences. The search engine is a streamlined research tool for researchers, professionals, policymakers, and students alike. We hope the new HumGen search engine will assist you in exploring the world of ethics in human genetics research.

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WE ARE DEEPLY GRATEFUL TO ALL WHO SUPPORT OUR WORK