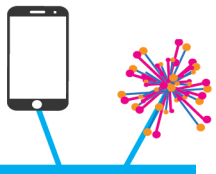
ANNUAL REPORT 2018



Data Sharing

Genomics & Epigenomics

Personalized Medicine

Rare Diseases

Paediatrics

Gene Editing

Cancer

Salmonella Genome

Food Safety

CGP Centre of Genomics and Policy
Centre de génomique et politiques

McGill

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TABLE OF CONTENTS

Message from the DIRECTOR

Dear Readers,

I am excited to share the 2018 Annual Report of the Centre of Genomics and Policy. We are pleased to showcase the work of our dedicated staff and to share their research interests with you. Once again, we have highlighted our Invited Scholars who have helped make this year exciting and fruitful. This year, our Invited Scholars joined us from Canada, the United Kingdom, the United States and South Korea.

This year's Annual Report emphasizes the CGP's innovative research program. It details completed and ongoing research projects, profiles the Invited Scholars, the Faculty's course offerings, and also highlights joint projects with P3G-IPAC and the team's publications.

In this year of the GDPR, we proudly published a "Special Issue on Data Sharing" in **Human Genetics**, and covered the diversity of ethical and legal frameworks found in Australia, Canada, China, Germany, South Korea, and the United States. During 2018, the CGP's research significantly supported the Global Alliance for Genomics and Health (GA4GH) in the development of regulatory and ethics policies and tools to enable international data sharing. CGP Members also made significant contributions in other areas such as rare diseases, epigenetics, biobanking, genetic enhancement, return of results, and, data sharing research involving vulnerable populations.

Moreover, you will learn more about our Centre's knowledge through the involvement of our researchers at the International Congress on Personalized Health Care (ICPHC) in September 2018 and its translation work through the Young Investigators Forum, our Summer Seminar Series, and our multitude of workshops.

Through these features, we hope our work is accessible and helpful to all!

We trust you will enjoy getting to know the CGP, its staff, and our work.

Bartha Maria Knoppers

Director
Centre of Genomics and Policy
McGill University

Message from the RESEARCH DIRECTOR

Dear Readers,

The year 2018 has seen the Centre continue to advance and grow as a unique, international hub for research on the ethical, legal, and policy issues of genomics and personalized health. In fact, members of the CGP have made several important contributions this year, with the publication of a number of articles on the ethical and legal issues of epigenetics, CRISPR/Cas9 and gene editing, DNA testing for family reunification, mobile health apps, genomics and food safety and, the sharing and protection of health-related data. Prof Bartha Knoppers and Erika Kleiderman have also contributed to the drafting of recommendations in the Consensus Statement on Gene Editing, Genetic Testing and Reproductive Medicine in Canada and Prof. Yann Joly participated as expert witness in the reference concerning the constitutionality of the Genetic Non-Discrimination Act. 2018 has also seen the consolidation of our collaboration with South Korea and we are pleased to welcome Hannah Kim at the CGP, an Invited Scholar from Yonsei University who is pursuing her research on data sharing, gene editing and privacy in OMICS applications.

This year marks a turn as we undertook diverse empirical initiatives to reach out and assess the needs, concerns, and expectations of different stakeholders in the field food safety, cystic fibrosis and personalized medicine. One of these initiatives is the Genetic Discrimination Observatory (GDO), an informative and collaborative platform providing access to information and tools needed to effectively counter genetic discrimination. During 2018, the GDO was used as a research platform to launch a pilot online public forum on genetic discrimination in Quebec.

I am also extremely pleased by our success in obtaining funding for 25 new research projects this year and by having had the opportunity to host 5 Invited Scholars, 4 students, 4 interns, and 1 articling law student. We look forward to collaborating with our research colleagues in upcoming national funding opportunities in 2019, where our team will continue to propose varied, multidisciplinary, and sophisticated methodologies to help in the implementation of novel science from research to the clinic.

Yann Joly

Research Director
Centre of Genomics and Policy
McGill University

ABOUT THE
CENTRE OF GENOMICS AND POLICY

Located within the Genome Québec 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, paediatrics, privacy, cancer, intellectual property, and biobanks (population genetics). These domains are approached using three guiding foundations: internationalization, policy development, and knowledge transfer. First, the CGP promotes internationalization by undertaking comparative analyses of policies and guidelines around the world. Second, the CGP actively participates in the creation of international consortia thereby promoting multidisciplinary policymaking. Finally, via its numerous workshops and lecture series, the CGP encourages knowledge transfer.

Don't forget to follow us on Twitter and suscribe to our YouTube channel.

@genomics_policy

Centre of Genomics and Policy

CGP TEAM

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ASSOCIATE PROFESSOR
JOLY Yann - RESEARCH DIRECTOR

EXECUTIVE DIRECTOR
ZAWATI Ma'n H.

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BARTHA MARIA KNOPPERS

DIRECTOR

(Comparative Medical Law), Canada Research Chair in Law and Medicine

Full Professor (PhD) and the Director of the Centre of Genomics and Policy, Faculty of Medicine, Department of Human Genetics, McGill University, Bartha Maria Knoppers is the Canada Research Chair in Law and Medicine (CRC) and holds four Doctorates Honoris Causa. She is Fellow of the AAAS, the Hastings Centre (bioethics), the Canadian Academy of Health Sciences and the Royal Society of Canada, and is Officer of the Order of Canada and of Québec.

She received the “Prix Montréal In Vivo: Secteur des sciences de la vie et des technologies de la santé” in 2012 and in 2013 was named “Champion of Genetics” by the Canadian Gene Cure Foundation. In 2014, she was named “Great Montrealer” (scientific Sector) by the Board of Trade of Metropolitan Montréal, and in 2015, she received the Medal Paul-André Crépeau for her efforts in comparative medical law (Canadian Bar Association). She also chairs the Ethics Panel of the World Anti-Doping Agency (WADA), helped found and co-chairs the Ethics and Regulatory Work Stream of the Global Alliance for Genomics and Health (GA4GH). In 2017, she gave the prestigious Galton Lecture.

RESEARCH DIRECTOR
YANN JOLY

Yann Joly, Ph.D. (DCL) Ad.E.: Lawyer Emeritus from the Québec Bar and Research Director of the Centre of Genomics and Policy (CGP). He is an Associate Professor at the Faculty of Medicine, Department of Human Genetics with a cross-appointment at the Biomedical Ethics Unit at McGill University. He is also a Research Fellow of the Fonds de Recherche du Québec-Santé (FRQS) and an Associate Researcher at the Centre de recherche en droit public at Université de Montréal. Prof. Joly chairs the Bioethics Workgroup of the International Human Epigenome Consortium (IHEC) and the UNESCO Human Variome Project (HVP) Standards Group. He is the Data Access Officer of the International Cancer Genome Consortium (ICGC) and a member of the Human Genome Organization (HUGO) Committee on Ethics, Law, and Society (CELS). In 2016 he was awarded a teaching award, and in 2017 he became a Fellow of the Canadian Academy of Health Sciences (CAHS).

EXECUTIVE DIRECTOR
MA'N H. ZAWATI

Ma'n H. Zawati (LL.B., LL.M., Ph.D. (DCL)) is the Executive Director of the Centre of Genomics and Policy in the Department of Human Genetics at McGill University. He is also an Associate Member of McGill's Biomedical Ethics Unit. His research concentrates on the legal, ethical and policy dimensions of health research and clinical care, with a special focus on biobanking, data sharing, professional liability, and the use of novel technologies (e.g. mhealth apps, WGS, WES) in both the clinical and research settings. Dr. Zawati is funded by CIHR, Genome Canada, and Genome Quebec. His work is interdisciplinary, drawing together perspectives from law, ethics, bioinformatics, genomics, and policy.

ACADEMIC ASSOCIATES

GRATIEN DALPÉ

Gratien Dalpé completed his undergraduate and master studies (B.Sc/M.Sc) in biochemistry at the University of Sherbrooke. He holds a doctorate degree (Ph.D.) in molecular biology from the University of Montreal. He later worked as a post-doctoral fellow and research associate at the Samuel Lunenfeld Research Institute in Toronto. During his career, he uncovered new molecular signalling networks regulating the development and degeneration of the nervous system. With an interest in law and bioethics, he later obtained his LL.B. in civil law at the University of Montreal and joined the Centre of Genomics and Policy in 2017 as an academic associate. He currently works on legal, ethical and social issues pertaining to genetic discrimination, health and genomic data sharing in research and personalized medicine.

PALMIRA GRANADOS MORENO

Palmira Granados Moreno is a Mexican lawyer specialized in intellectual property and information technologies, and a Doctor of Civil Law candidate at the Faculty of Law at McGill University, under the supervision of Professor Richard Gold. Her interests focus on the intersection of intellectual property, ethics, human genetic information, health, and technology. She joined the Centre of Genomics and Policy (CGP) in 2013. Her work focuses on the social, ethical, and legal aspects of research and development involving human genetic information and technology. Before joining the CGP, she became a member of the International Expert Group of the Innovation Partnership, of the New Researchers Group of VALGEN, and of the Centre for Intellectual Property and Policy of McGill University. Prior to Montreal, she obtained her law degree from la Escuela Libre de Derecho in Mexico and her LL.M from the Faculty of Law at the University of Toronto. She practiced law with a Mexican leading law firm in the area of intellectual property and information technology and was responsible for teaching the course Law and Public Policy at the Instituto Tecnologico Autonomo de Mexico. She has also been closely involved with the Free Software Foundation and the Creative Commons Mexico.

JULIE HAGAN

Julie Hagan (B.Sc/M.Sc) is an Academic Associate at the Centre of Genomics and Policy (CGP) at McGill University. She obtained a Master's degree in sociology from Université de Montréal and completed a doctoral internship at the University of São Paulo, Brazil. She is pursuing a Ph.D. in sociology at Laval University focusing on how inter-scalar governance, interdisciplinary collaborations, and public participation influence environmental policy-making in Canadian cities.

At the CGP, she contributes her experience in qualitative research methodologies and her knowledge of public participation to foster stakeholder engagement. She is currently involved in research about the social acceptability of omics approaches for the detection of Salmonella in fresh produces. She is also involved in projects examining how the changes brought about by the advances in genomics and the development of personalized medicine affects patients and health professionals as well as its effects on service delivery and policy-making.

Her other research interests include the co-construction of knowledge beyond the traditional boundaries between expertise and experience, STEM and the humanities, theoretical and applied knowledge, the natural and the social; socio-technical controversies in health and the environment; the impacts of new technologies on the urban social fabric; and planning for urban health and equity.

ERIKA KLEIDERMAN

Erika Kleiderman is a lawyer and an Academic Associate at the Centre of Genomics and Policy (CGP). She holds a civil law degree (LL.B.) from the Université de Montréal, as well as a B.Sc. in Psychology from McGill University. She was called to the Quebec Bar in 2014. Currently, her research deals with the ethical, legal, and social implications surrounding access to data and genetic information, biobanking, and the regulation of stem cells, regenerative medicine, and new reproductive technologies. Erika is engaged in the stem cell and regenerative medicine community through her involvement with the Stem Cell Network's Trainee Communications and Training & Education Committees, as well as through her coordination role in the pan-Canadian initiative aimed at assessing the adequacy of existing regulatory frameworks and considerations for reframing the Assisted Human Reproduction Act, in light of evolving reproductive technologies. She is also the Coordinator of the Canadian International Data Sharing Initiative (Can-SHARE) and the Access Officer of the Canadian Partnership for Tomorrow Project (CPTP), for which she has been actively involved in the development of controlled data and biosample access documentation and operating procedures. Erika is also interested in the potential applications of gene editing for performance enhancement (gene doping), as well as the implications of gene therapy and enhancement in minors within a sporting context. She is a member of the McGill University Health Centre Research Ethics Board.

EMMANUELLE LÉVESQUE

Emmanuelle Lévesque is a lawyer and a member of the Québec Bar. She holds a Master of Laws (specializing in biotechnology, law and society) from the Université de Montréal and an LL.B. from Université Laval. Her master's thesis is an analysis of the protections offered by the Canadian Charter and the Québec Charter against genetic discrimination in the workplace. Emmanuelle works at the Centre of Genomics and Policy at McGill University. She specializes in questions concerning ethical and legal issues in health research, particularly in biomedical, cancer and genetic research. Over the past several years, she advises researchers about the ethical and legal issues raised by the deployment of their projects, especially about the development and the framework of biobanks. She works on projects conducted in Québec, in the rest of Canada and overseas, including the PERSPECTIVE project on risk stratification in breast cancer screening. She has published several publications and held conferences on different ethical and legal issues raised by cancer research. Since 2017, she has been a member of the Comité d'excellence clinique en dépistage des maladies chroniques of the Institut national d'excellence en santé et services sociaux.

IDA NGUENG FEZE

Ida Ngueng Feze, Esq, JD, LLM, is an Academic Associate at the Centre of Genomics and Policy (CGP) at McGill University. Her research focuses on the ethical, legal, and social issues (ELSI) related to the health and environmental applications of genomic technologies. Ms Ngueng Feze's expertise and areas of interest include: genetic discrimination, bioethics within the one-health framework, pharmacogenomics, food safety policy and regulation, public health surveillance, water governance and metagenomics, ELSI related to the use of artificial intelligence, and traditional knowledge. She has provided training and presented at conferences in Canada and abroad before various stakeholders including patients' associations, health professionals, and members of the Canadian Human Rights Tribunal. She is a guest lecturer at Laval University, the University of Montreal and the University of Québec in Montréal. She is a member of the New York Bar. She holds a Master's degree (LL.M.) in International Law from the University of Montreal, a Juris Doctorate degree (J.D.) from Howard University School of Law, a certificate in Chinese Law from the China University of Political Science and Law, and a Bachelor of Arts degree (B.A.) in Law and Society with a Minor in Psychology from Ramapo College.

MINH THU NGUYEN

Minh Thu Nguyen holds a Master of Laws (LL.M.) degree in Health Law and Policy from the University of Toronto, a Civil Law (LL.B.) degree from the Université de Montréal, and a Bachelor of Science (B.Sc.) degree in Physical Therapy from McGill University. She was a fellow of the Canadian Institute for Health Research - Health Law and Policy Program (2009) and coordinator of the P3G International Paediatric Research Programme (2012-2013). She has managed the ELSI Platform for the ThéCell Network (stem cell therapies) and has been involved in several Stem Cell Network funded projects dealing with the regulation of cell and tissue therapies in Canada. She has worked on the development of model consent forms for rare disease research projects such as FORGE and CARE for RARE (pan-Canadian) and PRISMES (Quebec). Her work focuses on the socio-ethical and legal aspects of paediatric genetic research, rare disease research, cell/tissue therapy and regenerative medicine. She also has an interest in reproductive health law, particularly issues surrounding emerging reproductive technologies, such as prenatal diagnosis and pre-implantation genetic diagnosis.

MARK PHILLIPS

Mark Phillips is a practicing lawyer (Barreau du Québec) and is an Academic Associate at McGill University. He works in comparative data protection and privacy law and has an academic background in computer science. He has published analysis in law journals and scientific journals on topics including the identifiability of personal data, cloud computing, and sanctions in data protection regimes. As part of his legal practice, he has brought the first right to be forgotten claim regarding search engine privacy de-indexing to be heard before a Canadian court. He is a member of the executive of the Privacy & Access Law Section of the Canadian Bar Association, and is the co-editor of the Data Protection Task Team of the Global Alliance for Genomics and Health's Regulatory and Ethics Work Stream.

KATIE SAULNIER

Katie Saulnier graduated from Mount Allison University in 2010 with a Bachelor of Arts (Philosophy and English) focusing on ethics, and from the McGill Faculty of Law in May 2014 with a Bachelor of Civil Law (B.C.L.) and a Bachelor of Common Law (LL.B.). They were called to the Bar of the Law Society of Ontario in June 2016. They are currently pursuing an M.A. in philosophy with a specialization in bioethics from McGill University (2020), focusing on using a disability theory lens to examine the ways in which discourse around epigenetic research is changing conceptions of normative values assigned to bodies.

At the CGP, Katie is currently involved in research into the ethical, legal and social issues surrounding epigenetics and data sharing, as well as research on improving the quality of healthcare communication for intersex Canadians. Their other research interests include the ethical implications of depictions of reproduction and reproductive technologies in speculative fiction, issues with conceptions of autonomy in laws affecting women and Indigenous communities, and gender, neurodiversity and disability theory as they relate to the field of genomics.

KARINE SÉNÉCAL

Karine Sénécal holds a Master's degree in Law (Biotechnologies and Society) from the Université de Montréal and a Bachelor of Laws from the Université du Québec à Montréal. Her master's thesis focused on the legitimacy of a restricted application of germline gene therapy, from a human rights perspective and research ethics. Her thesis was published by the editor Themis, in 2007. Karine is an Academic Associate at the Centre of Genomics and Policy at McGill University. She specializes in comparative law and policies, as well as in the analysis of the ethical, legal, and social implications surrounding genomic research and modern medicine. Her main areas of research are paediatric research, genetic testing and screening of minors as well as on the governance and ethics interoperability of paediatric biobanks and databases (including access, use and sharing). Karine is a coordinator of the Paediatric Task Team of the Global Alliance for Genomics Health, a consultant for the Public Population Projects Genomics (P3G), and a member of the Quebec Network of Applied Genetics (RMGA). She sits on committees for the assessment of issues raised by biomedical technologies or on public health questions. She is author and co-author of more than 40 publications, including books or book chapters, peer-reviewed articles and policies.

LINGQIAO SONG

Lingqiao Song acquired a B.Sc in Biology and Master's Degree of Civil Law at the Chinese Academy of Social Science in China. In 2015, she completed her second Masters degree of International Business Law at the University of Montreal and was awarded "Dean's Award: Best Overall Academic Achievement". In 2016, she was admitted as a Chinese Lawyer and is working as a legal consultant for Anran Law firm (China). Currently, she is pursuing her Ph.D. degree focusing on Data sharing in Chinese biobanks at the Faculty of Law, McGill University. She is also a member of the Institutional Review Board of Faculty of Medicine, McGill University, and is the assistant to the Data Access Officer of The International Cancer Genome Consortium. At the Centre of Genomics and Policy, Lingqiao is working with Prof. Yann Joly and Ms. Ida Ngueng Feze on projects regarding Ethical, Legal, Social Issues (ELSI) of gene technology, policy approaches to address genetic discrimination, global data sharing of biobanks, and regulatory framework of microbiological genetic testing.

ANNE-MARIE TASSÉ

Anne-Marie Tassé (LL.B., LL.M., M.A., LL.D.) is a lawyer specialised in health law and bioethics. She holds a Doctorate in Law (Université de Montréal), Master's degrees in Health Law (Université de Sherbrooke), and in Bioethics (Université de Montréal), and a Certificate in Health and Social Services Management (Université du Québec). Her work looks primarily at interactions between law and ethics, in the areas of international biomedical and genetic research. Specialised in international comparative law, she is the Executive Director of the Public Population Project in Genomics and Society (P3G) and an Academic Associate at the Center of Genomics and Policy (McGill University). As such, she coordinates the legal and ethical aspects of Canadian and international research projects. Author of more than 45 book chapters, peer-reviewed articles, policies and guidelines, her work is presented in Canada and abroad.

ADRIAN THOROGOOD

Adrian Thorogood (B.A.&Sc., B.C.L./ LL.B.) is a lawyer and Academic Associate at the Centre of Genomics and Policy. He manages the Regulatory and Ethics Work Stream of the Global Alliance for Genomics and Health. This consortium frames policy and establishes technical standards to accelerate the global sharing of genomic and health related data. Adrian manages the development of policies addressing consent, privacy and security, data access, and research oversight that enable responsible data sharing between countries, institutions, and sectors. Adrian's legal research focuses on how genomic sequencing platforms, information and networking technologies, open science practices, and patient empowerment movements are disrupting biomedical research and health care. He focuses on the duties and liabilities of health professionals, and the privacy, confidentiality, and "ownership" of genomic and health data. He completed his law degree at McGill, articled at the Department of Finance Canada, and was called to the Ontario bar in 2015. Before entering law, Adrian obtained a Bachelor's degree from McGill University with a double major in health economics and biomedical sciences, and worked as an epidemiology researcher and clinical trial coordinator. He is currently pursuing his LLM at the University of Toronto.

INVITED SCHOLARS

ROSARIO ISASI

Rosario Isasi, J.D., M.P.H., is a Research Assistant Professor at the Miller School of Medicine with appointments in the Dr. J. T. Macdonald Foundation Department of Human Genetics, the Institute for Bioethics and Health Policy, the John P. Hussman Institute for Human Genomics, and the Interdisciplinary Stem Cell Institute. Her expertise is in the area of comparative law and ethics regarding genomics and regenerative medicine. Ethics Advisor to the European Commission's European Human Pluripotent Stem Cell Registry (hPSCREG), member of the American Society for Human Genetics (ASHG) Task Force on "Gene Editing," Academic Secretary of the International Stem Cell Forum Ethics Working Party, and leader of the Governance Working Group of the International Stem Cell Banking Initiative (ISCBI). She contributed to the development of harmonized ELSI and educational tools for Canadian Blood Services' National Public Cord Blood, the Centre for the Commercialization of Regenerative Medicine (CCRM), and the Bioethics Education Project of the Royal College of Physicians and Surgeons of Canada. With the CGP, she continues to collaborate in four projects related to stem cell research and regenerative cell therapy.

AMALIA ISSA

Amalia M. Issa is an internationally renowned scientist in the field of personalized genomic medicine (precision medicine). Dr. Issa was one of the first scientists to develop a unique area of translational research focused on precision medicine applications, and how they will be translated and integrated into clinical practice and health systems. She undertook some of the earliest studies of the societal and policy implications of pharmacogenomics, and continues to be engaged in leading a multidisciplinary collaborative effort to investigate and address important questions to build and develop the science of personalized genomic healthcare delivery. Dr. Issa founded the Personalized Medicine & Targeted TherapeuticsTM Center in 2001, as one of the very first centers focused on pharmacogenomics and personalized medicine. The mission of the center is to develop the evidence base for, inform decision-making about and accelerate knowledge translation of personalized medicine applications into meaningful health outcomes. She is also currently a Full Professor at the University of the Health Sciences in Health Policy and Public Health and Pharmaceutical Sciences. Dr. Issa holds leadership positions in several professional associations and national and international scientific advisory committees, and has received many awards and honours for her work. She is excited to be collaborating on several projects of mutual interest at the CGP, as well as with the Global Alliance for Genomics & Health.

HANNAH KIM

Hannah Kim M.D., Ph.D., is a research assistant professor at the College of Medicine, Yonsei University and a researcher at the Asian Institute for Bioethics and Health Law. She studied medicine and holds a PhD in healthcare law from Yonsei University. She worked as a Fellow at the Department of Medical Law at Yonsei University in 2016 –2017. She was a member of the Institutional Bioethics Board in the Severance Hospital, Yonsei University and also a member of the Committee of Direct-to Consumer Genetic Testing under the Ministry of Health and Welfare in South Korea.
With the CGP, she continues to collaborate on genomics data sharing, genome editing and genetic discrimination.

JENNIFER STODDART

Jennifer Stoddart was awarded an honourary doctorate in 2013 by the University of Ottawa and again in 2015 by McGill University. In June 2011, the Québec Bar awarded her the distinctions of Avocat émérite and Mérite Christine-Tourigny. She was appointed Officer of the Order of Canada in December 2015, for her international leadership in privacy rights and for her exemplary public service as the privacy commissioner of Canada (2003-2010). In her role as commissioner, she gave an annual report to Parliament about privacy trends and results of privacy audits of government departments. She represented Canada at the annual International Conference on Privacy and Personal Data Protection. She continues to explore her interests in personal data protection at the Centre of Genomics and Policy as an Invited Scholar. With Professor Knoppers, she is involved in the Advisory Expert Group that developed the 2017 OECD Recommendation on Health Data Governance and she co-Chaired its Advisory Expert Group.

RACHEL THOMPSON

Rachel Thompson is a final year PhD candidate at Swansea University, Wales. She holds BScs in biological and cultural Anthropology (UEL), and Medical Sciences with (medical) Humanities (Swansea). Rachel's PhD is in research ethics and governance as it applies to multi-location population scale biomedical research, with a focus on harmonization and pluralism in ethics. Rachel is co-chair of the UK Institute of Medical Ethics (IME)Postgraduate Bioethics Committee, lead organizer of the UK Postgraduate Bioethics Conference 2019, and administrates the Research Institute for Ethics and Law (Swansea University) alongside her PhD. Rachel was a visiting scholar at the CGP in September 2018, conducting qualitative research with WADA regarding potential future uses of existing data, as well collaborating with CGP colleagues on the consent of minor athletes in doping controls.

POST-DOC FELLOW

CHARLES DUPRAS

Charles Dupras, B.Sc., M.Sc., Ph.D. is a postdoctoral fellow at the Center of Genomics and Policy (CGP) at McGill University. He completed a master's degree in molecular biology at INRS-Institut Armand-Frappier, then completed a doctoral degree in bioethics at the University of Montreal. He was awarded a three-year fellowship (2017-2020) by the Canadian Institutes of Health Research (CIHR), for his research on the translation of emerging knowledge in epigenetics.

Charles is exploring Canadian laws and public policies potentially applicable to – or to be amended to accommodate – recent findings about epigenetic mechanisms, such as DNA methylation. The main objective is to ensure Canadian regulations such as the recent Genetic Non-Discrimination Act (2017) or existing guidelines for the ethical conduct of genetic research (e.g., data sharing and protection of privacy) apply consistently and justifiably to epigenetic information.

PhD STUDENTS

GABRIELLE BERTIER

"Clinical Implementation of Next-Generation Sequencing in Pediatrics: A Multidisciplinary Analysis of Policy Implications"

This thesis project analyses the current use of Next-Generation Sequencing (NGS) technologies in the clinic. Focusing on France and Quebec, it examines the ethical, legal, social and policy implications of the use of NGS technologies, in pediatric patients with undiagnosed rare diseases and relapse or refractory cancers.

Supervisor: Yann Joly, Centre of Genomics and Policy, McGill University.
Co-Supervisor: Anne Cambon-Thomsen, UMR, 1027, Insem Université Toulouse III – Paul Sabatier.

PhD Awarded on August 29, 2018.

DEREK SO

"The Role for Images of Human Genomic Engineering in Assessing Societal Views Relevant to Policy"

Recent advances in gene editing technology have renewed a longstanding bioethical debate about making heritable genetic modifications in humans. For many decades, various communities have envisaged different scenarios for the intentional selection of human traits, producing a repository of images which continue to evoke strong moral responses and to shape popular, bioethical and literary discourses alike. Studying this body of thought can help us to develop policy on gene editing by learning more about the ways people tend to conceptualize the human genome, genetic disorders, and the act of genetic modification. The aim of this thesis is to provide a theory accounting for these schemata, in order to help clarify the socio-cultural influences on stakeholder values toward gene editing. To this end, reviews of both academic and popular discourse will be performed, as well as surveys aimed at eliciting the views of different stakeholders, and a discussion paper to disseminate the resulting framework.

Supervisor: Yann Joly, Centre of Genomics and Policy, McGill University.
Co-Supervisor: Rob Sladek, Department of Human Genetics, McGill University.

VASILIKI RAHIMZADEH

"Evaluating the gap between research ethics review governance and data sharing for pediatric genomics in Canada"

Biomedical research since sequencing the human genome has become increasingly data-intensive that requires collaboration often across disciplinary and geographic borders. Delivering on the promises of precision/genomic medicine rests largely on making sound statistical associations between the human genome and disease. This requires that genomic and associated clinical data are shared widely and securely, including data involving children. Sparse policy attention has been paid, however, to the specific ethical-legal protections associated with such sharing in pediatric populations, nor what impact they might have on research collaboration. This PhD thesis adopts policy Delphi methods to examine the relationship between research ethics review policy and genomic data sharing for collaborative studies involving children across Canada (www.projectpedigree.org).

Supervisors: Bartha M. Knoppers, Centre of Genomics and Policy, McGill University and Gillian Bartlett, Department of Family Medicine, McGill University.

FOROUGH NOOHI

"Promoting Responsible Governance of Mitochondrial Replacement Therapy in Canada"

Today, more than ever, the progress of scientific technologies continues to push legal and ethical boundaries. At the center of the controversy surrounding the regulation of scientific advancements is human gene editing and more specifically the manipulation of human embryos and the human germline. Mitochondrial Replacement Therapy (MRT), a type of in vitro fertilization that aims to prevent the transmission of mitochondrial diseases to future generations, is considered a criminal offence in Canada. This PhD thesis aims to address important issues in debating the legitimacy of this controversial technique in Canada.

Supervisor: Yann Joly, Centre of Genomics and Policy, McGill University

SUMMER SEMINAR SERIES

June 5 – October 2, 2018

EVENTS AT THE CGP 2018

Each year, the CGP invites its members and Invited Speakers to present to the team and share their research findings (e.g. new publications, ongoing research results, etc). It is a great opportunity to both learn about each other's research projects and be acquainted with emerging issues in different fields. This year, some of the topics include: precision medicine, personal data, the right to benefit from science, improving communication between health care providers and intersex individuals, consenting to mHealth research, genetic counselors, minors, enhancement and sport, to name but a few.

GUEST SPEAKERS AT THE CGP

JANUARY 24

PROF. ROBERT NADON
ASSOCIATE PROFESSOR
McGill University Faculty of Medicine, Visiting Fellow of the QUEST Center for Transforming Biomedical Research at the Berlin Institute of Health
"The Importance of Transparency in Biomedical Research Study Design and Statistical Analysis"

APRIL 26

PROF. TAMRA LYSAGHT
ASSOCIATE PROFESSOR
PhD of the Centre for Biomedical Ethics, National University of Singapore
"A Framework for Encouraging Responsible Innovation with Autologous Stem Cells"

JUNE 5

DR. EDWARD DOVE
LECTURER
School of Law, University of Edinburgh
"Promoting health research and protecting participants? The impact of 'next-generation' health research regulation on NHS research ethics committees"

JULY 3

PROF. JENNIFER BYRNE
PROFESSOR
Professor of Molecular Oncology, Sydney Medical School
"Improving Biomedical Research Integrity: Better Biobanking and Literature Analyses"

AUGUST 22

DR. AMY BOMBAY
ASSISTANT PROFESSOR
Department of Psychiatry and the School of Nursing, Dalhousie University
"Creating Ethical Space for First Nations Led Biological Health Research"

OCTOBER 2

PROF. YIXIE LI
PROFESSOR OF CAS-MPG
Partner Institute of Computational Biology, Chinese Academy of Sciences
"Introduction to the Chinese National Infrastructure for Biomedical Big Data"

INTERNATIONAL CONGRESS

ON PERSONALIZED HEALTH CARE (ICPHC)

September 23-26, 2018

This important international conference gathered professionals and decision-makers interested in discussing the major stakes related to personalized medicine and health care. Based on the presence of renowned speakers and support by a call for papers process designed to take the pulse of the international community, the program proposed four days of discussion, training and networking.

YOUNG INVESTIGATORS FORUM

INTERNATIONAL CONGRESS ON PERSONALIZED HEALTH CARE

The 2nd International Conference on Personalized Health Care (ICPHC) hosted its first ever Young Investigators Forum on September 26th. The Forum was aimed at graduate students and early-career researchers in all disciplines to discuss some of the most pressing challenges facing research and academia in the modern world.

Keynote speakers walked us through issues such as the use of social media in science communication (Jonathan Jarry, Office for Science and Society, McGill University) and public speaking (Jay Olsen, PhD Candidate in Psychiatry, McGill University). Yasser Riazalhosseini (Assistant Professor, Department of Human Genetics, McGill University) hosted a Q&A session on grant applications, and several students presented throughout the day.

GE3LS CONFERENCE

COULD OPEN MODELS BE THE YELLOW BRICK ROAD TO PRECISION MEDICINE?

AN OVERVIEW OF OPEN MODELS OF COLLABORATION IN GENOMICS AND PRECISION MEDICINE

MARCH 6, 2018
LE MERIDIEN VERSAILLES HOTEL
MONTREAL, QUEBEC

Precision medicine could improve prevention and promote better healthcare. However, outside of niche markets such as rare diseases (including some cancers) and pharmacogenomics, the ultimate potential of OMICS sciences in the development of precision medicine remains uncertain.

In order to advance precision medicine, a growing number of projects are adopting models that are more open (i.e. open science, open source, peer-based production, protected commons, crowdsourcing, open innovation, etc.) than the "traditional" drug development one based on proprietary intellectual property. Other factors, such as the questioned capacity of the patent system to promote innovation in medicine and medical genetics, and the separation between basic research (academia) and the development of clinical applications (industry) becoming increasingly blurry, have also encouraged companies and researchers to opt for more open models of collaboration.

This workshop provides the forum to discuss experiences, objectives, challenges, concerns, benefits, and potential paths to follow regarding the use of open models of collaboration in the field of personalized medicine in North America: two developed countries and one developing country.

STEM CELL NETWORK

BARTHA MARIA KNOPPERS, ERIKA KLEIDERMAN, MINH THU NGUYEN AND FOROUGH NOOHI

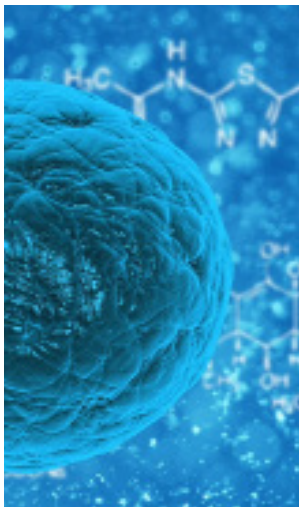
1. **Canadian College of Medical Geneticists (CCMG)**
   Harrison Hot Springs, British Columbia
   CONSULTATION (June 10, 2018)

2. **Canadian Fertility & Andrology Society (CFAS)**
   Montreal, Quebec
   CONSULTATION (September 14, 2018)

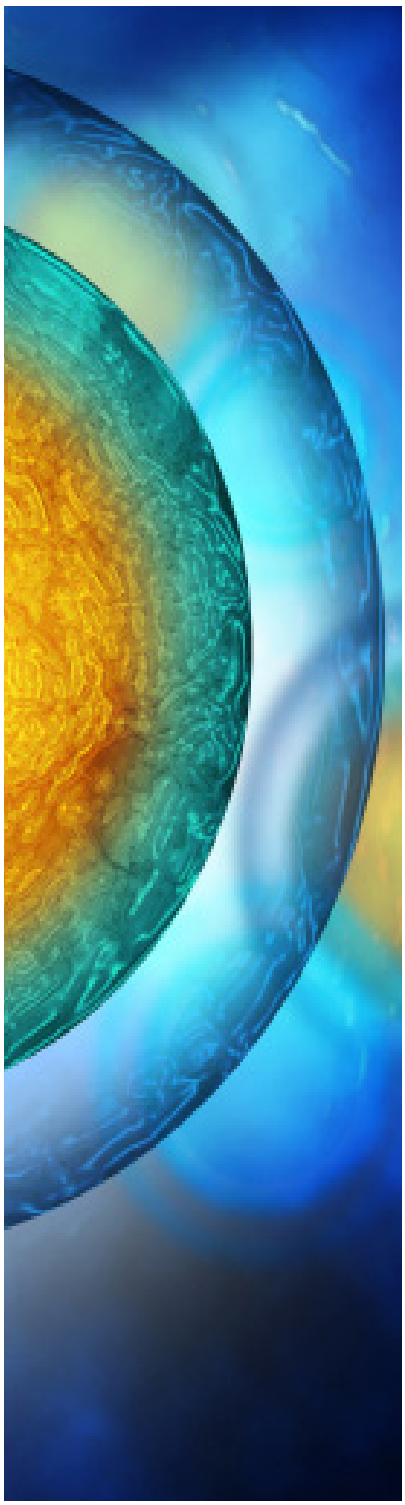
3. **Till & McCulloch Meeting**
   Ottawa, Ontario
   CONSULTATION (November 14, 2018)

4. **ThéCell**
   Montreal, Quebec
   CONSULTATION (November 21, 2018)

5. **Assisted Human Reproduction Act: Consensus Statement Deliberation / Validation**
   Ottawa, Ontario
   WORKSHOP (December 11, 2018)



COMPLETED RESEARCH PROJECTS



Enhanced CARE for RARE Genetic Diseases in Canada

April 2013 - March 2018

Genome Canada / Ontario Genomics

CARE for RARE is a collaborative pan-Canadian project configured to improve the diagnosis and treatment of rare diseases. Powerful new DNA sequencing methods such as whole-genome (WGS) and whole-exome sequencing (WES) were used to discover 60 new genes, each of which caused a rare disease. The identification of new genes provided useful biological information that gave us insight into cellular pathways significant in human health. The research lead to the implementation of WES as an effective and reliable diagnostic tool for clinical use.
The integration of WES into the clinical setting, the GE3LS component of this study, was/has been be conducted at the Centre of Genomics and Policy, included: 1) the drafting of clinical exome trial consent forms; 2) a qualitative analysis of the clinical utility of WES as a diagnostic tool for patients with rare diseases (from the patients' and clinicians' point of view); 3) the drafting of a national position statement and best practices outlining professional and ethical standards for the reporting of incidental findings found using WES; and 4) the development of clinical integration tools for use by clinics offering exome sequencing to patients with rare diseases.

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Personalized Risk Stratification for the Prevention and Early Detection of Breast Cancer (PERSPECTIVE)

April 2013 - March 2018

Fondation du cancer du sein de Québec / Le Centre de recherche du CHU de Québec-Université Laval / Genome Canada / Genome Québec

The project significantly extended the benefits of the current high-quality population breast cancer screening programs, particularly for younger women (35 to 49) by implementing a risk stratification approach targeting screening at an identifiable subset of women with relatively high risk of breast cancer (BC) who are missed by the current standard age-based screening program. This personalized risk-based approach to breast cancer screening detected cancers at an earlier stage. Significant socio-economic and health benefits were achieved since a woman's survival prospects were increased and the burden of disease and costs of treatment were reduced.

Knowledge of the genetic basis of BC and its risk factors allowed stratification of individuals into different risk groups for screening and personalized follow-up with appropriate preventive and clinical measures. However, a strategic approach was needed to facilitate the acceptance and adoption of risk-based stratification BC screening models in clinical settings, healthcare services, and policies. Our project, delivered a web-based risk stratification and communication toolbox for use by health professionals and women to facilitate the implementation of a personalized risk-based approach in BC screening and management.

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

April 2013 - March 2018

Genome Canada / Genome Québec / Le Centre de recherche du CHU de Québec-Université Laval

The goal of this project was to develop an integrated information campaign to sensitize the population to the importance of considering family history to fight effectively against breast cancer. The campaign also aimed to better equip health professionals to evaluate the risk of breast cancer on the basis of family history. This campaign was realized through a rigorous process that included partner diverse professional and community associations.

Information and sensitization tools were developed and compiled into information toolkits. These toolkits responded to three needs: to effectively collect, use, and share information on family history of breast cancer. The tools allowed 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 team included experts in genetics, epidemiology, public health, psychosocial evaluation, ethics, and public law (CGP).

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Élaboration d'une stratégie de dissémination des connaissances sur les cancers familiaux dans le contexte des soins palliatifs fondée sur une analyse éthico légale des points de vue des intervenants et des familles

September 2015 - August 2018

Fonds de Recherche du Québec - Santé (FRQS) / Ministère de la Santé et des Services sociaux (MSSS)

This project aimed to propose a strategy for the dissemination of knowledge in cancer genetics applied to the context of palliative care in order to help stakeholders address the needs of dying patients and their family members on issues related to their family history of cancer, all in an appropriate legal and ethical framework.

To achieve this, three activities were implemented and were aimed at: 1) identify with doctors and nurses in palliative care priority issues associated with family history of cancer in their work context, as well as the barriers and facilitating factors to address, if any, ethically with the patients end of life and members of his family; 2) identify needs and concerns related to palliative care cancer patients about their family history and how to address them properly in the context of the end of life; 3) identify the legal and ethical guidelines applicable to the communication of family history of cancer in the family members of a patient in palliative care. Ultimately, the goal was to develop knowledge dissemination activities, including a toolbox to ensure the dissemination and accessibility of information for healthcare professionals working in palliative care who have to deal with issues related to a family history of cancer.

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GE3LS Network in Genomics and Personalized Health

April 2016 - December 2018

Genome Canada / Genome Alberta / Genome Québec

The 2012 Genome Canada large-scale applied research project competition, genomics and personalized health (GAPH), funded 17 projects. Each project integrateed technology assessment and social science and humanities research under the banner of the signature genome Canada GE3LS initiative (i.e. genomics and its ethical, environmental, economic, legal and social aspects). In 2016, Genome Canada announced funding for a network to bring together GE3LS researchers from all 17 projects. The Network enableed the projects to share best practices; improve and prime future collaborative research; accelerate the progress to market of gaph technologies; and maximize the impact of investment in the gaph projects. The Network addressed four priority GE3LS themes: research ethics review; health economics and health technology assessment; knowledge transfer and implementation in health systems for 'omics technologies; and intellectual property and commercialization. This $2 million initiative was designed to accelerate the translation of research results into practical applications in healthcare for the benefit of Canadians. The government of Canada, through Genome Canada, invested $1 million in the Network, with the balance of funding to be secured from co-funders. Objectives for each Network theme were outlined by GE3LS and science representatives from each of the 17 projects at a workshop in April 2015. A Network management team (Network co-leads and theme leads) monitored progress on each objective. By having identified/identifying and directed/directing research to overarching issues that emerge from the GE3LS components of the 17 projects, the Network accelerated their common goal of transforming research results into practical applications adopted by health systems for the benefit of patients.

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CellCAN: Canadian Cell, Tissue and Virus Manufacturing for Regenerative Medicine and Cell Therapy

August 2016 - March 2018

Networks of Centres of Excellence (NCE)

Collaboration and information sharing between researchers, regulators, funding agencies and industry was essential to implement new treatments that benefited patients, and a central hub of information was crucial for better implementation and patient care. The CellCAN Regenerative Medicine and Cell Therapy Network provided the physical, management and regulatory infrastructure needed to build patient awareness of regenerative medicine and cell therapy (RMCT), and expedite regulatory approvals and promote RMCT treatments for diseases such as diabetes, cancer and heart disease by sharing and harmonizing operating procedures and clinical trial protocols. CellCAN's multidisciplinary effort involving scientists, physicians, patients, ethicists, economists, the private sector and other stakeholders increased efficiencies and reduced costs between Canada's cell manufacturing centres, while sharing procedures, patient samples, expertise and knowledge.

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Genome Quebec Policy Briefs

May 2017 - April 2018

Genome Québec

For this project, the Centre of Genomics and Policy (CGP) developed three Policy Briefs pertaining to emerging themes in genetics. More precisely, members of the CGP authored Policy Briefs on Genetic Discrimination, Gene Editing, as well as Access and Sharing of Information with Genetic Researchers.

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RMGA : Forum Québécois en ligne sur la discrimination génétique : Droits, Responsabilités et Conscientisation

RMGA : Online Quebec forum on genetic discrimination – Rights, Responsibilities and Awareness

March 2018 - December 2018

Réseau de medicine génétique appliquée (RMGA)

The use of genetic information outside of the health context can create some concerns regarding the possibility that it may negatively affect someone based on their genetic profile. This phenomenon has been referred to as genetic discrimination. Victims of genetic discrimination can be affected negatively professionally and socially, and may face psychological distress. There is presently no empirical study that has evaluated the extent of this phenomenon in Quebec.
The Genetic Discrimination Observatory (GDO) is a unique communication platform created to help prevent genetic discrimination. From July to October 2018, the GDO held an online public forum on genetic discrimination to enable residents of Quebec to share their opinion or questions on this subject. The data collected will be used to assess the concerns, needs and opinions of Quebec residents. The GDO platform will also be used as a comprehensive source information on genetic discrimination and existing approaches to prevent this phenomenon, and as a scalable tool to reach the public in Canada and internationally.

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> "...The forum on genetic discrimination enables the public to share their opinions or questions on this societal issue..."

CURRENT RESEARCH PROJECTS

CURRENT RESEARCH PROJECTS

Réseau de médecine génétique appliquée (RMGA) Infrastructure

April 2006 – March 2019

Fonds de Recherche du Québec - Santé (FRQS)

The RMGA is a Québec 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 Québec population. The Network has 350 members representing the majority of human genetics researchers in Québec. The RMGA included a Legal and Socio-Ethical Issues Infrastructure at the CGP that considered issues arising from the research activities of the RMGA members and provided ELSI guidance on emerging issues. In 2016, the CGP consolidated a decade of RMGA policies into a prospective Statement of Principles.

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ThéCell (Réseau de thérapie cellulaire et tissulaire) : enjeux socio-éthiques et juridiques des thérapies cellulaires et tissulaires

April 2009 – March 2019

Fonds de recherche du Québec - Santé (FRQS)

Created in 2009, the Cell and Tissue Therapy Network (ThéCell) 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. As the Socio-Ethical and Legal Platform, our role is to provide ad hoc consultation to researchers and clinicians on ethical and regulatory issues related to cell and tissue therapies. We provide assistance with drafting consent forms and research protocols for research ethics approval and Health Canada clinical trial applications.

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CE in Biomarker-Driven Clinical Research for Personalized Medicine in Cancer (Exactis)

April 2014 – March 2019

Networks of Centres of Excellence (NCE) / Canadian Institutes of Health Researh (CIHR) / Natural Sciences and Engineering Research Council of Canada (NSERC) / Social Sciences and Humanities Research Council (SSHERC)

The objective of the project is to create and expand a biomedical ecosystem that overcomes the major rate-limiting steps involved in realizing and expanding biomarker-driven clinical research for personalized medicine in cancer. This will be achieved through: 1) ready access to an enormous collection of engaged patients, their tumors, and clinical data collected in a prospective and longitudinal manner that conforms to the highest standards of ethics and quality; and 2) an advanced program in serial biopsies of metastatic tumors that defines the molecular signature of resistance to new targeted agents allowing the industry to modify the drug or add combinations to overcome or avoid resistance and greatly expand the clinical benefit to patients. The Centre of Genomics and Policy provides a review of the ethical and legal issues while ensuring that: 1) the consent form respects the federal and provincial legal requirements (on re-use of samples, governance, confidentiality, data-sharing, e-consent, risks, etc.); and 2) is compliant with the various ethical policies and guidelines pertaining to storage of data and samples, (access, re¬contact). Finally, it is analyzing the legal liability of health professionals undertaking such research.

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Réseau en soins de santé personnalisés: Q-CROC

April 2014 – March 2019

Fonds de partenariat pour un Québec innovant et en santé (FPQIS)

This project aims to broaden and deepen the existing Q-CROC Network which has developed internationally recognized expertise in designing and executing biopsy-driven studies to identify biomarkers in metastatic cancers. Moreover, it will use a program in which all new cancer patients are asked to consent to having their primary tumor biobanked and profiled, to having their entire clinical course anonymously recorded, and to being re-contacted for additional studies. Consistent with its prospective population-based approach, the new trans-national global network will help generate the large scale of profiled patient numbers and build an enormous biological and clinically annotated database.

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The Cancer Genome Collaboratory

April 2014 – March 2019

Natural Sciences and Engineering Research Council of Canada (NSERC)

Cancer, a disease of the genome, is the leading cause of mortality in Canada, responsible for more than 70,000 deaths per year. The Cancer Genome Collaboratory is a unique cloud computing facility that is preloaded with more than 500 TB of cancer genomic research data, hosts APIs for efficient access to the data, and provides tools for creating, using and sharing scalable cancer-genome-analysis pipelines.

This project is accelerating research for effective treatment of cancer by providing researchers with access to the world's largest open cancer genomics dataset. It is accelerating the development of precision oncology, in which the genomes of the patient and tumour inform the choice of the therapy that is most likely to benefit the patient. The project will also accelerate our understanding of tumour evolution, allowing tumours to be detected at an earlier stage and for doctors to adjust the therapy as the tumour changes. Over the long term, the project will benefit Canadians by providing cancer patients with improved diagnostics and therapeutics.

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Research Advancement through Cohort Cataloguing and Harmonization (ReACH)

April 2016 – March 2021

Canadian Institutes of Health Research (CIHR)

More and more Canadians are affected by chronic diseases such as cancer, cardiovascular disease, chronic obstructive lung disease, diabetes, and mental illnesses. Many of these conditions have their origins in early life (conception, pregnancy, infancy, and childhood). Numerous outstanding Canadian pregnancy and birth cohort studies have been implemented to explore hypotheses related to the Developmental Origins of Health and Disease (DOHaD).

The Research Advancement through Cohort Cataloguing and Harmonization (ReACH) initiative was formerly established in 2016 to provide the Canadian research community with the means to leverage and carry out leading-edge collaborative research. The ReACH initiative will provide resources in the form of a comprehensive web-based catalogue and a harmonization platform to optimize and expand the use of Canadian pregnancy and birth cohorts data and biological samples. Ultimately, the ReACH initiative will enhance the capacity for collaborative and cross-disciplinary research (outputs generated faster and at a lower cost); expand research perspectives (leverage national and international collaborations); improve quality of research practices; and foster the development of innovative and reliable evidence-based research on the Developmental Origins of Health and Disease.

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A Syst-OMICS Approach to Ensuring Food Safety and Reducing the Economic Burden of Salmonellosis

October 2015 – September 2019

Genome Québec / Genome Canada

Each year, approximately 88,000 people become sick from consuming fresh produce that is contaminated with Salmonella. The health impacts can vary between people suffering, from no ill effects to serious infection requiring medical care or even causing death. Salmonella infection is thought to cost the Canadian economy as much as $1 billion annually in terms of medical costs, work absenteeism, and economic losses (including that of the food and restaurant industries).

Using whole genome sequencing to identify the specific Salmonella strains that cause human disease, the team will develop natural bio-solutions to control the presence of Salmonella on fruits and vegetables. New tests will also be developed to quickly and efficiently detect the presence of Salmonella on fresh produces before they are sold to consumers. These new tools will allow public health officials to better determine the source of Salmonella illnesses so that contaminated foods can rapidly be removed from grocery stores and restaurants. An anticipatory governance approach will be used to conduct a regulatory assessment integrating a careful consideration of the evidentiary requirements, the economic, legal, ethical, regulatory, and global policy constraints as well as the trade implications of supporting such a paradigm shift. More precisely, the CGP team will: 1) assess the viability and implementability of a novel genomic test for Salmonella that can determine the pathogenic status of a given isolate; and 2) assess the need to shift to a risk-based approach to food safety that classifies foodborne microbial adulterants on the basis of their pathogenicity, in contrast to the current regulatory practice of classifying a microbial adulterant based on species determination alone.

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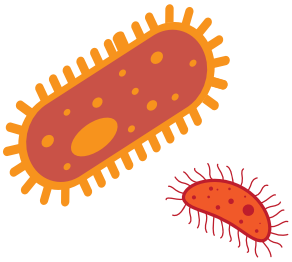

How the early environment interacts with prenatal adversity and genetic susceptibility to moderate the risk for anxious and depressive disorders from infancy to early adolescence - the moderating effect of maternal care and the mediating effect of temperament

April 2016 – March 2021

Canadian Institutes of Health Research (CIHR)

The precocious and chronic course of depression makes it the disease with the leading cause of disability, an effect marked by a 2-fold difference in the rate for girls and women as of early adolescence. Arguably what is missing is not a list of risk factors, but rather a precise knowledge of how factors interact to predict those at higher risk. For instance, differences by gender in the response to stress are well documented in early emotional development, but evidence is needed in older children and adults. Genetic differences in susceptibility to prenatal events are also important. Likewise, early maternal care might be a significant positive or negative influence on the effect of prenatal, gender and genetic risk. The project gives the opportunity to collaborate in four prolific international longitudinal cohorts to communicate, share and reproduce models and findings about early factors in the prediction of early age psychopathology. The cohorts in Canada (Maternal Adversity, Vulnerability and Neurodevelopment), the United Kingdom (Avon Longitudinal Study of Parents and Children), the Netherlands (Generation- Rotterdam) and Singapore (Growing Up in Singapore Towards Health Outcomes) include and share measures of genes, maternal care, child psychopathology, and data of a sensitive nature with different consent forms, measures, access approaches, laws and regulations. The role of the CGP is to act as a collaborator-consultant, supporting the project with analysis, reflections and recommendations that pertain to the ethics and legality of: 1. Safeguarding child genomic data, 2. Sharing data across four national jurisdictions, 3. Maintaining standards of international IRB, and 4. Issues of consent and the need to re-contact child participants once they become adults.

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> "...We would like to identify genetic and environmental factors that can predict depressive disorders in children and adults..."

Sino-Canada HeLTI: A Multifaceted Community-Family- Mother-Child Intervention Study for the Prevention of Childhood Obesity (SCHeLTI)

April 2016 – March 2021

Canadian Institutes of Health Research (CIHR) / The National Natural Science Foundation of China (NSFC)

The epidemics of obesity and metabolic syndrome related disorders are a major public health concern. Increasing evidence points to the role of early life adverse factors in the developmental origins of the vulnerability to such metabolic disorders. Reducing the risk of overweight and obesity (OWO) from early life stages will produce substantial benefits to decrease population burdens of metabolic diseases. However, current intervention measures remain insufficient to halt the increasing OWO epidemics. Building on our strengths in large birth cohort studies, clinical trials and studies on developmental programming of metabolic disorders, our transdisciplinary Sino-Canadian team will conduct a community-based multi-centre cluster-randomized controlled trial to test the effect of an evidence-based and multi-faceted early life-course community family-mother-child interventions package incorporated into the routine pre-conception, prenatal and child care systems on childhood OWO rates in 1-6 years old children in China. The mechanisms (microbiome, epigenetics, micro RNAs) that may underline the development of OWO will be explored, as well as the impacts of the intervention. The research program will for the first time evaluate an integrated intervention package from pre-conception and early pregnancy into childhood on OWO rates. The findings will produce a scalable community-based intervention package that may be recommended for implementation at regional and national level to reduce the risk of OWO and metabolic syndrome related disorders in China. The CGP develops appropriate policies and tools to facilitate the use of the cohorts included in this project. More specifically, the CGP develops the governance framework as well as the data access and biospecimen sharing policy. The tools created by the CGP will aim to facilitate policy interoperability and access authorizations as well as streamline the ethical and legal aspects of international collaborative research. Furthermore, the CGP offers to SCHeLTI researchers ongoing ethics and policy support throughout the project.

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The gender specific effects of prenatal adversity on the development of anxious and depressive psychopathology in early adolescence – the moderating effect of genes and early maternal care

April 2016 – March 2021

Canadian Institutes of Health Research (CIHR)

There is a considerable amount of evidence for gender dimorphic effects of prenatal stress. Gender effects have been found on the placenta, the fetus, the infant and the child, although inconsistently. Girls may be more susceptible to the effects of fetal adversity on fearful temperament, emotional reactivity and internalizing problems increasing their risk for the development of affective problems. Very little research has directly examined whether gender differences in the effect of fetal adversity are maintained in the prediction of anxious and depressive psychopathology in older children. A landmark study reports that maternal prenatal depression is associated with an increased risk of depressive symptoms in 18 year-old offspring in females. Although inconsistent findings can be explained by multiple factors (including for example variations in the measurement of prenatal stress exposure), this study suggests the need to carefully consider how gender and prenatal adversity interact in a longitudinal design, and to consider the role of genotype and the postnatal environment. Accordingly, gender considerations will be approached as follows: (1) carefully examination of gender-based age-specific trends in the developmental of anxious and depressive psychopathology from preschool age to pre-adolescent age; (2) the moderation of gender effects for anxious and depressive psychopathology by genetic susceptibility; (3) the role of early maternal care; and, (4) early temperamental signals of vulnerability to anxious and depressive psychopathology. The CGP's role is to design the guidelines for the ethics and legality of: (i) the safeguarding of genomic data from children; (ii) the sharing across four national jurisdictions of biologic data; (iii) the maintenance of standards by international IRB's; and, (iv) the consent for use of data from children as subjects become adults.

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CanDIG: Canadian Distributed Cyber-Infrastructure for Genomics

April 2016- March 2020

Canadian Foundation for Innovation (CFI)

The Distributed Infrastructure for Genomics Data Sharing and Analysis project is divided into four activities. In Activity 1, they will develop a broad Canadian data sharing framework, using the APIs that we and others are developing under the auspices of the Global Alliance for Genomics and Health (GA4GH); Activity 2, they will continue the development of GenAP: a computational gateway for data analysis in life sciences that is configured to take advantage of Compute Canada infrastructure; Activity 3, they will build a data-sharing platform that will allow for the collection of standardized clinical data, dynamic definition of cohorts, and performance of genome analytics across datasets that are being stored on various Compute Canada nodes; and, to enable genome-guided clinical trials across Canada, in Activity 4 they will establish the Canadian Molecular Profiling in Cancer Trials (CAMPACT) Interchange. Together, the four Activities will utilize Compute Canada infrastructure to build a distributed and secure computational framework for the analysis of genomic datasets relevant to human diseases and beyond. The Centre of Genomics and Policy will also contribute to the implementation of the data sharing and privacy policy framework of the International Human Epigenome Consortium (IHEC) as well as the GA4GH.

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Programme de Recherche et d'Innovation Sur les Maladies rarES (PRISMES)

April 2016 – March 2021

Le Centre de recherche du CHU de Québec-Université Laval

In a framework of scientific collaboration, the Centre of Genomics and Policy (CGP) oversees and prepares the ethical documents required for initial approval by the Research Ethics Committee of CHU of the Québec-Université de Laval. Subsequently, the CGP will provide support on ethical and legal issues, including changes/modifications to ethical documents or preparation of documents arising from the evolution of the research Program and the initiation of new collaborators with groups of international research.

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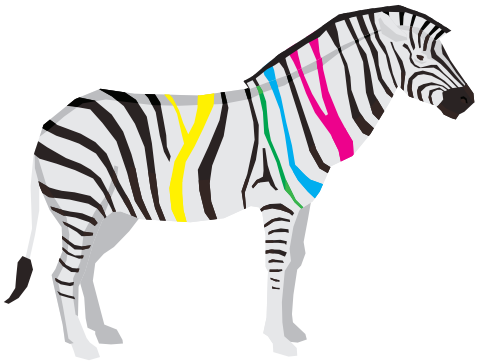

Predisposition, Prediction et Prevention du Cancer du Sein (PREVENTION)

April 2016 – March 2019

Fondation du cancer du sein de Québec / Ministère de l'Économie, de la Science et de l'Innovation (MEIE) / CHU Laval / Fondation Cancer du Sein

Most breast cancers develop in a small proportion of women at high-risk. These women are currently identified based on family history and BRCA1/2 mutations. Recent breakthroughs in genomics allowed the identification of a large number of genetic variations associated with breast cancer. These include common variations (SNPs), which can be combined into a polygenic risk profile to stratify women according to their individual risk. It is imperative to conduct studies using large international cohorts to obtain reliable estimates of the individual risk of breast cancer and to facilitate the identification of women who can most benefit from prevention approaches. Two major projects were set up: PERSPECTIVE, led by the Québec team, and BRIDGES, funded by the European Commission-Horizon 2020 (09/2015-08/2020), of which Université Laval is a partner without funding. Professor Simard's participation was decisive and the results of PERSPECTIVE will be instrumental to the realization of several BRIDGES objectives. The funding of the MEIE will be timely, consolidating the leadership of the Québec team in the BRIDGES project. Our results will improve targeted disease prevention in oncogenetic clinics and the Québec breast cancer screening program.

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Multidimensional Epigenomics Mapping Centre (EMC) at McGill

February 2017 - January 2021

Canadian Institutes of Health Research (CIHR)

To join global efforts, the International Human Epigenome Consortium (IHEC) has established an Epigenome Mapping Centre (EMC) at McGill University that applies epigenome mapping in order to understand interactions between environment and genome in human blood cells, interprets diseases impacting metabolism using tissue samples, and studies how epigenetic changes can alter function of the brain. The large-scale generation and sharing of human epigenome data presents challenges to the informed consent process that are managed first through the integration of existing cohort data with EMC McGill, using a special template developed in conjunction with the Public Project in Genomics and Society (P3G), and subsequently by prospectively developing a model consent template that ensures all IHEC consent, policy, and ethics requirements are met. Throughout this, we will continue to actively participate in discussions on the development of a more comprehensive ethical policy framework at the IHEC level. Both the EMC and Epigenomic Data Coordination Centre (EDCC) projects also involve the development of and support for a bioethics workgroup for the IHEC.

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Healthy Life Trajectories (HeLTI): Governance Framework

January 2018 - March 2022

Canadian Institutes of Health Research (CIHR)

The HeLTI program was developed by the Canadian Institutes of Health Research (CIHR), Institute of Human Development, Child and Youth Health led by Dr Shoo Lee. The program represents a partnership between CIHR, the South African MRC, the Department of Biotechnology, India, the National Natural Science Foundation of China and the World Health Organization. HeLTI was developed to address the increasing burden of non-communicable diseases (including obesity, diabetes, cardiovascular disease and poor mental health) around the world. There are four separate but harmonized projects that are commencing in Soweto (South Africa), Mysore (India), Shanghai (China), and across Canada. All projects are focused towards developing evidence-based interventions that span from pre-conception across pregnancy and into the postnatal period with a goal improving maternal, infant and child health.

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Regulation of International Direct-to-participant (DTP) Genomic Research

March 2018 - January 2021

National Institutes of Health (NIH)

The proposed study is designed to generate a rich body of data concerning the legal and ethical issues of international Direct-To-Participant (DTP) genomic research using a diverse sample of 32 countries. The aims described below are designed to identify and frame the issues in a way to maximize the utility of the research contributions of the international experts. Ultimately, the goal is to produce well-conceived conclusions and actionable policy options.
Aim 1: Convene three expert working groups of researchers and IRB leaders to identify and prioritize the key issues for a standard questionnaire and template to distribute to the study's 32 international experts on country-specific laws and research ethics.
Aim 2: Distribute the questionnaires to the international experts, consult with and advise them as they prepare their responses to the questionnaires and summaries of country-specific legal and ethical issues, compile and analyze the responses, and draft conclusions and policy options.
Aim 3: Distribute the draft conclusions and policy options to the three expert working groups, international consultants, and advisory board members, and obtain their feedback; analyze the responses and use them to prepare a final draft; write and publish a consensus article presenting the findings of the study; and disseminate the results in presentations to various groups of stakeholders.

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Toward effective health communication with intersex Canadians: a study of ethical and legal challenges

March 2018 - March 2022

Social Sciences and Humanities Research Council (SSHERC)

The belief that all human beings develop into only one of two neatly defined sexes – male and female – is deeply inaccurate. As much as 2% of the population falls under the category of intersex, with chromosomes, gonads, hormones, or genitalia that exist along a spectrum of sex differences. Intersex individuals face egregious human rights violations, discrimination, and stigmatization worldwide. Historically, these issues have been exacerbated by healthcare practitioners (HCPs) via unnecessary surgical interventions, the withholding of accurate health information, and an overemphasis of incidental health risks. For adult intersex individuals, distrust of the medical system due to poor treatment is exacerbated by poor health communication, generating reluctance to seek medical care and greatly impacting quality of life. Despite a pressing need, qualitative research from which to base public policies for ending these harms remains underdeveloped, and no Canadian-specific guidance exists.

This project engages two communities of stakeholders: intersex individuals and HCPs. The core objectives of the project are to generate much-needed qualitative evidence on the experiences of intersex individuals in the healthcare system, and to use this evidence to develop improved standards of care. Following a comparative review of relevant laws and policies, we will conduct a series of semi-structured interviews with intersex adults and with the HCPs implicated in their care. From these, we will develop: 1) a guidance document on effective HC with intersex patients in the Canadian context; 2) an issues paper highlighting the challenges of conducting person-centered research with vulnerable populations; and 3) information sheets in both French and English for intersex individuals navigating the Canadian healthcare system.

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Precision Oncology For Young People (PROFYLE 2)

April 2018 - March 2019

Terry Fox Research Institute

Cancer is the leading cause of disease-related death in children, adolescents, and young adults (CAYA) beyond the newborn period. While the overall survival rate in Canada approaches 88% for all CAYA patients (0-29 years old), the prognosis for those with refractory, relapsed or metastatic ('hard-to-treat') disease is grim, and progress has stagnated for many disease groups over the last three decades.

To address this gap, and to make new therapeutic for CAYA patients with hard-to-treat childhood cancers, we initiated the PRecision Oncology For Young peopLE (PROFYLE) program. Overall, the program aims to transform the care of CAYA patients by using next-generation molecular tools and cancer model systems to identify disease and patient-specific biomarkers. The core of PROFYLE consists of real-time molecular profiling to personalize cancer treatment with either existing oncology drugs or repurposed drugs. The project includes the incorporation of new technologies and an emphasis on the evaluation and development of minimally invasive patient-specific biomarkers that can track the patient's molecular profile over time. To enable these activities, and to propel profiling results into new discoveries and future novel therapies, the core molecular profiling stream will be supported by two unique program components – a pan-Canadian network for disease modeling and a nextgeneration-biobank & data repository. This platform will have an immediate impact on outcomes by providing treatment options for patients with hard to-treat tumours.

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Interrogating and implementing Omics for precision medicine in Acute Myeloid Leukemia

April 2018 - March 2022

Genome Canada / Genome Québec

Acute myeloid leukemia (AML) is one of the leading causes of cancer-related death in young adults and a highly lethal disease in older adults. Most AML patients do not survive longer than two years after diagnosis, due to a lack of effective treatment options and inadequate molecular tolls to monitor disease prognosis. Treating patients considered favourable (with chemotherapy) or adverse (with stem cell transplantation) is relatively straightforward. It is significantly more difficult, however, to treat patients who fall into the middle, intermediate risk category. The Leucegene Project was inaugurated in 2009 to address this gap in treatment. It has since become a world leader in deciphering genetic abnormalities associated with AML.

Leucegene's work has led to the development and implementation of a new prognostic test that enables physicians to predict responses to available AML treatments for patients. Despite great improvement, physicians remain unable to identify optimal treatment for 30% of patients. Over the coming years, the Leucegene project aims to reduce this number to less than 10% using genomic technologies to identify previously unknown genetic variants and to develop novel prognostic testing regimes.

In parallel, the Leucegene project will develop a prognostic and therapeutic web portal to make this work available to researchers, clinicians, and patients. The CGP will contribute to this effort by developing internal ethical and legal policy documents that will facilitate implementation of the portal. We will review policy instruments adopted for similar functions and extensively engage with patients, caregivers, and healthcare providers to inform these efforts. The CGP will, drawing on international policy and jurisprudence, similarly work to develop a Good Practices document to inform the development of prognostic and therapeutic web portals in other healthcare contexts.

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GenCOUNSEL: Optimization of Genetic Counselling for Clinical Implementation of Genome-Wide Sequencing

April 2018 - March 2022

Genome Canada / Genome British Columbia / Genome Québec

Genome-wide sequencing (GWS; whole genome or exome sequencing) is a powerful new genetic test that analyzes a person's entire genetic make-up. While valuable, it can be problematic, by revealing disorders or disease risk factors unrelated to the original reason for testing, or by generating complex findings that are difficult for non-expert health providers to interpret. While not currently routinely available, genome-wide sequencing will soon be in more widespread use for patients who need it – increasing demand for genetic counselling, to which access is already limited in Canada.

Genetic counsellors provide education and emotional and decisional support to patients and families, helping them to make informed decisions about genetic testing and its results. Because of lack of legal recognition of genetic counsellors in Canada, most of them are found in academic centres rather than in the community.

GenCOUNSEL, which brings together experts in genetic counselling, genomics, ethics, health services implementation and health economics research, is the first project to examine the genetic counselling issues associated with clinical implementation of GWS. It will determine the most efficient socio-economic, clinical, legal and economic methods of providing genetic counselling once GWS is available in the clinic. It will create an understanding of current and future needs for genetic counselling, develop best practices for the delivery of genetic counselling, improve access to the counselling, particularly for underserved patient populations, and study the feasibility of different models of legal recognition of genetic counsellors. The result will be increased access, patient satisfaction and cost-efficiency while helping to make genetic counselling available to all Canadians who need it.

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Personalized risk assessment for prevention and early detection of breast cancer: Integration and Implementation (PERSPECTIVE II)

April 2018 - March 2021

Genome Canada / Genome Québec / Ontario Genomics

Breast cancer is the most common cancer and the second leading cause of cancer death in Canadian women. Current screening recommendations invite all women aged 50-74 to have a mammogram every two-to-three years. This research project will provide evidence supporting the transition of current age-based screening programs to a risk-based approach. Personalized risk assessment using genomic profiling and other risk factors would allow prevention and screening recommendations tailored to individual risk. The evidence generated will also improve genetic counselling about screening and risk reduction strategies for cancer genetic clinics patients with a family history of breast cancer. The research team, led by Drs. Jacques Simard (Université Laval) and Anna Maria Chiarelli (Cancer Care Ontario, University of Toronto), will study large cohorts of women using high-throughput genomic technologies, together with statistical and epidemiological methods, to develop and validate a multi-gene panel test based on genetic variations associated with breast cancer. The team will also assess the acceptability, feasibility, and outcomes of risk-based screening using a new, comprehensive risk-prediction web tool and a genomic profiling test within existing mammography centers. An improved personalized risk assessment will enable earlier detection, prevention, and treatment of breast cancer, saving lives and providing health and socio-economic benefits.

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Personalized therapy for individuals with cystic fibrosis

April 2018 - April 2022

Genome Canada / Ontario Genomics

Cystic fibrosis (CF) is the most common fatal genetic disease, affecting 4,000 Canadians and 80,000 people throughout the world. The debilitating disease causes difficulties in breathing, lung infections, and digestive disorders and those affected die at a median age of 35 in Canada. Treatments can ease symptoms, but there is currently no cure. Newer drugs can address the underlying genetic defect that causes CF, but only some patients respond positively to them, while others do not. Given the side effects and the high cost of these drugs, there is a pressing need for robust predictors of who will respond to what treatment. Dr. Felix Ratjen (Hospital for Sick Children) and his team are developing predictive tools to help clinicians determine the right medicine for the right patient. The team will examine how genetic factors can help predict individual treatment responses and examine if drug testing on tissue samples can be used to inform the potential clinical response to drugs by each patient. The team will work with industry partners, patient organizations and the Ontario Ministry of Health to integrate these strategies into patient care. The result of the team's work will be a shift toward individualized treatment for CF, assistance for clinicians in making treatment decisions, guidance for policymakers on reimbursement and better health outcomes for patients.

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Reforming Canadian Stem Cell Policy: Moving Beyond the Assisted Human Reproduction Act (AHRA)

April 2018 - February 2019

Networks of Centres of Excellence (NCE) / Canadian Institutes of Health Researh (CIHR) / Natural Sciences and Engineering Research Council of Canada (NSERC) / Social Sciences and Humanities Research Council (SSHERC) / Stem Cell Network (SCN)

Before 2004, there were few policies or research guidelines in Canada that addressed stem cell use, specifically the use of embryonic stem cells. Credible, research-based evidence was urgently needed to inform policy discussions on balancing moral and religious views with the needs of researchers and people seeking treatment for injury and disease.

To overcome this challenge, SCN brought together scientists, legal scholars and policy-makers to share their insights, which resulted in informed government legislation and research guidelines implemented by national research funders, such as the Canadian Institutes of Health Research (CIHR). That evidence and expertise also informed the Assisted Human Reproduction Act in 2004. Approximately 20% of SCN's network members appeared as witnesses at the draft and committee stages of the AHRA. In addition, SCN provided briefing materials to every MP and Senator, and held informational workshops on Parliament Hill.

In 2016-17 the Stem Cell Network provided support for members of its policy community to hold a series of workshops focused on genetic reproductive technologies relevant to the federal Assisted Human Reproduction Act (AHRA). In November 2017, Bartha Knoppers, from the Centre of Genomics and Policy, presented a report at the annual Till & McCulloch meetings. The policy paper and recommendations do not represent the official policy or position of the Stem Cell Network. Rather, it is an output generated to inform policy development in relation to updates to the AHRA.

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Autism Speaks MSSNG Project

April 2018 - March 2021

Autism Speaks Inc.

Autism Speaks launched the MSSNG project with the overall goal to store and analyze the world's largest collection of genomes of people with autism and their families. As part of the MSSNG Database project, P3G is offering Data Access Compliance Office (DACO) services to the MSSNG project. From January 1st to December 31st 2017, DACO processed 39 applications (including renewals).

With our goal of sequencing the DNA of 10,000 families affected by autism, MSSNG will help answer the many questions we still have about the disorder. Thanks to the Google Cloud, this vast sea of information will be made accessible for free to researchers. The greatest minds in science from around the world will be able to study trillions of data points in one single database. Already, MSSNG has led to new discoveries about autism and its associated health conditions. With 10,000 or more whole genomes and the help of the global science community, we can far outreach what has been accomplished so far. MSSNG will identify many subtypes of autism, with the goal of developing more personalized and effective treatments.

As part of the MSSNG Database project, P3G2 is offering Data Access Compliance Office (DACO) services to the MSSNG project. From May 1st to October 31st 2018, DACO processed 15 applications (including renewals).

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McGill University and Génome Québec Innovation Centre

April 2018 - March 2022

Genome Canada / Genome Québec

The McGill University and Génome Québec Innovation Centre is a world class research facility for genomics and proteomics. Founded in 2002, the Centre has developed a world-renowned expertise in complex genetic disorders such as cardiac disease, asthma and Type 2 diabetes, and has become a resource and a networking site for various research initiatives in human health, forestry, infectious diseases, agriculture and environment.

Ambitious projects in recent years are a testimony of the ability of Génome Québec to provide data of exceptional quality in the pursuit of various genomics studies. The Innovation Centre provides complete DNA and RNA analysis services, from a few samples to several tens of thousands per week. Large-scale genomics and proteomics services at the Innovation Centre are articulated around sequencing (including massively parallel sequencing), genotyping, functional genomics and extraction supported by a solid infrastructure, tools (Nanuq), unique expertise in bioinformatics and nucleic acid extraction. The Innovation Center also hosts the Canadian Center for Computational Genomics (C3G) which offers bioinformatics services.

All services work in parallel to provide comprehensive, reliable services to the Québec, Canadian and international scientific community. Located on the campus of McGill University in the heart of Montreal, the Innovation Centre acts as a vast resource of knowledge and technology to the academic and industrial sectors.

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Genomics, Islamic Ethics and Public Engagement (GIEPE): Towards Bridging the Knowledge and Communication Gaps

May 2018 - April 2021

Qatar National Research Fund

As genomic medicine and personalized healthcare becomes increasingly prevalent in the Gulf region, the public and policymakers face difficult ethical questions. Since 2003, there has been a great deal of institutional emphasis on developing genomic medicine and personalized healthcare in the Gulf. As healthcare systems increasingly emphasize these emerging fields, the public should be exposed both to the science of genomic medicine and to associated issues in Islamic ethics. The latter, being a crucial element of the public's 'moral world,' underscores how ethics and policy norms may be developed for the regulation of genomics practices within the Islamic tradition.

There has been little scholarly or public debate on the implications of genetics on Islamic ethics. To address these gaps, this project attempts to develop an understanding of public engagement for the development of policies and regulations in the genomics and personalized medicine contexts that is both religiously and culturally sensitive. As a long-term objective, the project aims to lay the foundation for a Qatar-based consortium of experts and specialists interested in fostering public understanding and engagement with science. This project's research team will be composed of experts in such fields as Islamic ethics, genomics, international bioethics, medical social sciences, translation, and interdisciplinary and intercultural communication. The Centre of Genomics and Policy will perform a comparative analysis of international genomics norms that will provide the team with international perspectives they can emulate.

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Epigenome-wise: ethical, legal and societal issues of new assays for DNA-methylation in cancer diagnostics and screening

July 2018 - April 2020

Netherlands Organisation for Scientific Research (NWO) / Erasmus Medical Center

Epigenetics is the study of molecular mechanisms for the regulation of gene expression, such as DNA methylation, that switch genes off or on without altering the DNA sequence. The Building Blocks of Life project 'Cut out for the future!' is developing a novel assay (MeD-seq technology) for genome-wide DNA methylation profiling in colon and cervical cancer. This new technology will help to understand the role of DNA-methylation in the pathogenesis of cancer, and may ultimately improve the ability to predict disease progression and treatment outcome in colon and cervical cancer and potentially a variety of other cancers. Due to its unique features epigenetic technology - uncovering potentially modifiable risk factors at high resolution and very low cost - may increase individual autonomy, health and well-being by offering personalised preventive interventions. However, these features also give rise to pressing ethical, legal and societal issues regarding autonomy, informed consent and unsolicited findings, harms & benefits of screening tests, privacy, personal responsibility and solidarity.

Through close cooperation with the BBoL project, we will identify and address ethical, legal and societal issues arising in the early phase of research and development of MeD-seq technology. Moreover, we will anticipate and explore issues that may arise in the future when epigenetic technologies may be introduced into the domain of population screening. We will develop practical guidelines to help investigators, test developers and policy makers to consider these issues in the phase of research & development and in anticipation of its potential implementation in population screening.

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Genetic Counselling in Quebec for Prenatal Testing for Intersex Conditions

September 2018 - August 2019

McGill Observatory on Health and Social Services Reforms

As our scientific understanding of intersex conditions increases, and as we increase the frequency of testing that can detect intersex conditions prenatally via genetic testing, we will increase awareness of intersex conditions in otherwise asymptomatic individuals. As is true of other biological differences with the potential for prenatal diagnosis, this has the potential to increase pathologization, particularly given the option to terminate affected pregnancies. Genetic counselling is one avenue through which parents might be helped to understand the full spectrum of intersexuality and realistic expectations for their children. Despite a pressing need, policies and regulations for genetic testing and genetic counselling in the context of intersex conditions are severely lacking, and there has not been a commensurate rise in training and education for genetic counselors on this subject.

Of the five genetic counseling training programs available in Canada, two are currently located in Quebec, at McGill University and Université de Montréal. For this research, we are investigating the training available for genetic counselors in Quebec on intersex conditions, as well as the number and content of genetic tests available to Quebec residents that permit testing for intersex conditions. Simultaneously, we are undertaking a scoping review of existing literature and applicable policy recommendations on the communication of prenatal test results involving intersex conditions to parents. Out of this, we will develop a discussion paper drawing attention to the implications of the rise of prenatal genetic testing for intersex individuals.

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Epigenomics Secure Data Sharing Platform for Integrative Analyses (EpiShare)

October 2018 - September 2021

Genome Canada / Genome Québec

Advances in next-generation sequencing have led to a vast increase in available human epigenetic data, including transcriptomic data (via RNA-seq) and chromatin data (via ChIP-seq). These epigenetic datasets have led to the development of expression-wide association studies (EWAS) and chromatin-wide association studies (CWAS). These approaches may lead to improved biomedical applications by providing mechanistic explanations and key insights into the interpretation of genome-wide association studies (GWAS). However, one of the main issues for users of these datasets is that obtaining the raw data stored at multiple controlled access repositories can be a very challenging task. That is because access to this raw data needs to be controlled in order to protect research participants' right to privacy.

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EUCANCan: a federated network of aligned and interoperable infrastructures for the homogeneous analysis, management and sharing of genomic oncology data for Personalized Medicine

December 2018 - November 2022

Canadian Institutes for Health Research (CIHR) / Fonds de Recherche du Québec - Santé (FRQS)

EUCanCan aims to federate existing European and Canadian infrastructures to analye and manage genomic oncology data. Each member projects, in Amsterdam, Barcelona, Berlin, Heidelberg, Paris and Toronto, has established a strong genomic oncology research and clinical program. EUCanCan will provide oncology researchers with a uniform computing environment to securely share and analyze harmonized cancer genome and phenome data in the context of clinical research. The CGP is co-leading the development of guidance and ethico-legal tools regarding international sharing of clinical and research oncology data. The aims are to (1) perform a legal, policy, and literature review to develop guidance aimed at Canadian oncology projects when sharing personal health data with European countries and within Canada, (2) analyze the requirements Canadian projects will have to satisfy pursuant to the EU General Data Protection Regulation when receiving personal health data from European partners, (3) delivering a Report these first two topics, (4) developing overarching guidance for the project on the use and sharing of clinical cancer-related genomic and other health related data, and (5) describe generalized compliance tools to be used by future international collaborations, whether they are studying cancer or other diseases.

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The Cancer Genome Collaboratory

October 2018 - September 2021

Canadian Foundation for Innovation (CFI)

The Cancer Genome Collaboratory (CGC) is a unique Canadian cloud compute facility that holds the world's most comprehensive public collection of cancer genomes and associated clinical information. The proposed work will extend the CGC's data holdings, improve accessibility to the data, add a series of high-quality vetted pipelines for standardized cancer genomics analysis, and implement services that apply new cutting-edge algorithms for the interpretation of cancer genomes. Our design and implementation plans are based on feedback from our Canadian and international user community, and on our own hands-on experience coordinating international genome analysis projects across petabyte-scale data sets.

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Open Science and Third-Country Personal Data Transfers: "Open" Unlimited?

April 2018 - March 2019

Tanenbaum Open Science Institute

Open Science is among the most recent branches of the various open content movements to attract widespread attention and significant investment on both sides of the Atlantic. The European Commission's "EU Open Science Cloud" was detailed in its October 2017 EOSC Declaration. In Canada, meanwhile, the 2017 launch of the Tanenbaum Open Science Institute came with a commitment forgo intellectual property claims on the data associated with their research, and otherwise make their data broadly available. These initiatives correspond with the lead-up period to the enforcement of the European Union's new General Data Protection Regulation (GDPR), whose rules on the transfer of personal data to countries outside the EU are part of a network of data protection frameworks including Canada's Personal Information Protection and Electrotic Documents Act. Because open content movements, including Open Science, have rarely engaged with the implications of data protection on their overarching mission, the risk of contradictory, incoherent, and ill-suited practice and frameworks looms large in this area. This article seeks to trace how the values underlying Open Science and data protection might be harmonized in the context of data transfer, and in particular with key transfer frameworks sketched out in the GDPR: adequacy decisions, model contractual clauses, and codes of conduct.

> "...the values underlying Open Science and data protection might be harmonized in the context of data transfer, and in particular with key transfer frameworks sketched out in the GDPR..."

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Care4Rare Canada: Harnessing multi-omics to deliver innovative diagnostic care for rare genetic diseases in Canada (C4R-SOLVE)

April 2018 - March 2022

Genome Canada / Genome Alberta / Genome British Columnbia / Ontario Genomics

To understand unsolved rare diseases (RDs), C4R-SOLVE explores new sequencing technologies and seeks to improve data sharing world-wide, enabling the discovery of new causes of RDs. The goal of C4R-SOLVE, aligned with Canada's national RD strategy, is to fully understand the molecular pathogenesis of unsolved RDs and facilitate timely access to clinical genome-wide sequencing. As part of its activities, C4R-SOLVE will develop a pan-Canadian RD repository (Genomics4RD) to optimize data sharing and analysis. P3G2 developped a governance framework to oversee the Genomics4RD database. This framework outlines the policies with respect to the creation, management, and use of the Genomics4RD repository. It includes, amongst others, provisions on the identification of eligible participants, core consent requirements (prospective/retrospective), terms of the collection and integration of data from different sources (e.g., genomic data, clinical data, cost data, patient-contributed data, etc.), re-contact options and, where relevant, mechanisms for the return of results. Furthermore, an assessment of legacy (retrospective) datasets to be shared with Genomics4RD and PhenomeCentral was undertaken.

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euCanSHare: An EU-Canada joint infrastructure for next-generation multi-Study Heart research

October 2018 - September 2022

Canadian Institutes for Health Research (CIHR) / Fonds de Recherche du Québec - Santé (FRQS)

H2020 euCanSHare aims to build the first one-stop-shop platform for multi-cohort cardiac data integration and exploitation, integrating within a unified ecosystem metadata catalogues, computational tools and data models for facilitating data sharing and re-use in cardiovascular research. Through this consortium, the platform will build upon existing collaborative initiatives on both sides of the Atlantic, namely the Canadian Alliance for Healthy Hearts and Minds (8 cohorts), the European BiomarCare project (>30 cohorts), and other large cohorts such as the UK Biobank (500,000 participants). Addressing their current limitations and exploiting the accumulated know-how and experience, euCanSHare will provide new possibilities for personalised medicine research in the field of cardiology. As the leaders of WP1 (Socio-ethical and legal interoperability analysis) (and in collaboration with P. Borry (KU Leuven)), P3G2 coordinates the ELSI research activities regarding the legal and ethical implications of implementing blockchain technology, and associated smart contracts, in a Canadian-European data sharing context.

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P3G2
Policy Partnerships Project for Genomic Governance

As a project under the Center of Genomics and Policy (CGP), the Policy Partnerships Project for Genomic Governance (P3G2) provides interoperability services to assist international researchers in meeting ethical and legal regulatory requirements governing genetic/genomic research in their home countries. P3G2 provides two main types of service:

THE IPAC PROVIDES TWO MAIN TYPES OF SERVICE:

ELSI INTEROPERABILITY

This service creates a wide range of customized tools (consent forms, data access policies, material/data transfer agreements, re-contact, return of results, publications and intellectual property policies, etc.) to foster the interoperability of international and national research consortia and projects.

DATA ACCESS COMPLIANCE OFFICE

The Data Access Compliance (DAC) office services both international and national research projects. It receives and reviews data access applications for access to controlled datasets, in conformity with the goals and policies of the project.

IN 2018

P3G2 has been involved in 7 projects at the CGP: Human Cell Atlas (HCA) (see pg 56); Transforming Autism Care Consortium (TACC) (see pg 54); Care4Rare-SOLVE (C4R-SOLVE) (see pg 50); Healthy Brains for Healthy Lives - Knowledge Mobilization Program (see pg 54); Canadian Healthy Infant Longitudinal Development (CHILD) (see pg 55); NacBio (see pg 55); and EU-Canada joint network for Sharing Heart research data, tools and know-how (euCanSHare) (see pg 50).

CGP & P3G ▸ IPAC COLLABORATIONS

Canadian Partnership for Tomorrow Project (CPTP)

April 2009 - March 2018

Canadian Partnership Against Cancer (CPAC)

The Canadian Partnership for Tomorrow Project (CPTP) has enrolled 300,000 Canadians between the ages of 35 and 69 years, who have agreed to be followed for their adult lifetime, to explore how genetics, environment, lifestyle, and behavior interact and contribute to the development of cancer and other chronic diseases. This pan-Canadian project has five participating cohorts (Atlantic PATH, CARTaGENE, Ontario Health Study, Alberta Tomorrow Project, BC Generations Project).

Hosted at the Public Population Project in Genomics and Society (P3G), the ELSI Standing Committee 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, access policies, and procedures to dealing with ethical issues surrounding consent, privacy, data sharing, and proposing governance structures for the CPTP. P3G also runs the Access Office (AO) of the CPTP, which reviews access requests from national and international researchers.

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

July 2009 – March 2018

Ontario Institute for Cancer Research (OICR)

The International Cancer Genome Consortium (ICGC) has coordinated 90 national cancer research projects that have the common aim of elucidating the genomic changes present in many forms of cancers contributing to the burden of disease in people throughout the world.
Housed at the CGP, the ICGC's Data Access Compliance Office (DACO) grants ICGC Controlled Data Access to international researchers in genomics, bioinformatics, and other related fields. A total of 335 applications, submitted by researchers in 31 countries, are currently approved for access. To date, DACO has processed over 2000 applications for ICGC Controlled Data Access (including resubmissions and renewals).

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Transforming Autism Care Consortium (TACC)

April 2018 - March 2021

Fonds de Recherche du Québec - Santé (FRQS)

Launched in 2018, the Transforming Autism Care Consortium (TACC) network aims to bring together the autism research community in Quebec, by improving access and availability of resources and integrating knowledge into practice. The Quebec 1000 families project (the "Q1K project") is a TACC network's flagship project. It provides a platform to facilitate research by creating a large cohort ASD family trios (proband participant, and first-degree relatives) in families where a child has been diagnosed with an autism spectrum disorder. P3G2 assists the TACC with the development of a governance framework, template consent forms/assent forms, relevant policies (privacy, publication, data access, code of ethics/conduct), template agreements (contributor, data sharing) and support to research ethics board submission.

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Healthy Brains for Healthy Lives Open Science Policy Knowledge Mobilization

August 2018 - August 2019

Canada First Research Excellence Fund

As part of the Open Science Policy Knowledge Mobilization project, P3G2 develops a brief on an international comparative research, to ensure that consent forms are designed in a manner that increases patient awareness of and promotes data sharing and open science principles, in line with international best practices.

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NAC BIO

September 2018 - September 2019

NAC Bio Inc.

NAC Bio is a health information company aiming to advance clinical research into the medicinal benefits of cannabis in the treatment of chronic disease and illness, by using the latest developments in data-science, human genomics and digital health. Headquartered in Vancouver, British Columbia, it was founded by National Access Cannabis Corp. and by Dr. Tyler Wish. As part of this project, P3G2 is planning to support the development of the following documents and policies: Governance Framework, Template Participant Information Sheet and Consent Forms, Recruitment and Consent Policy, Re-Contact and Re-Consent Policy, Return of Results and Incidental Findings Policy, Data and Samples Access Policy.

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Building a Governance Toolkit for Canadian Healthy Infant Longitudinal Development (CHILD)

October 2018 - September 2019

AllerGen/National Centres Excellence (NCE)

The Canadian Health Infant Longitudinal Development (CHILD) project was launched in 2007. It is a multidisciplinary, longitudinal, population-based birth-cohort study. This study is powered to test multiple hypotheses within the theme that specific environmental exposures together with genetic and immunological determinants, lead to pathophysiological allergic responses, and that clinical outcomes including asthma may be further modified by lung growth, hormonal and metabolic influences and psychosocial environment. The cohort provides a platform for many current and future studies of childhood allergy and asthma and is an offshoot of AllerGen. P3G2 assists the project in the development of a governance framework for the CHILD database and biobank, consent clauses, relevant policies on: data and material access, re-contact/re-consent, return of results, intellectual property, publication, and, guidance documents (memoranda for tech transfer offices and research ethics boards).

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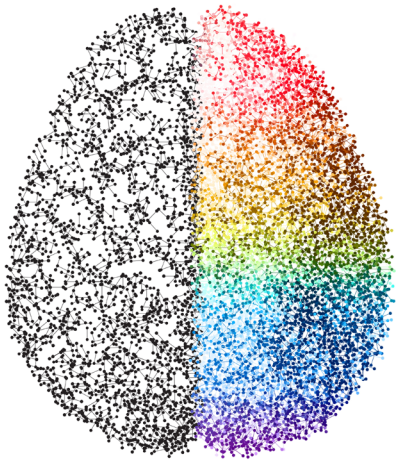
Human Cell Atlas

November 2018 - October 2020

The ChanZuckerberg Initiative / The Leona M. and Harry B. Helmsley Charitable Trust / The Klarman Family Foundation

The Human Cell Atlas (HCA) is a global biomedical research collaboration to create a reference map of all human cells – an international, public resource for better understanding human health and disease. The HCA's success relies on the expertise of researchers across several countries, undertaking work in different research domains and settings. P3G2 assists the HCA in the ongoing coordination of its Ethics Working Group (EWG), the development of an ethics and data governance framework, template consent forms, relevant tools (retrospective consent filter, ethics submission guidance), template agreements (material/data transfer agreement templates, data submission agreement template, data use agreement template) and a helpdesk to support the HCA community.



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> "...The Human Atlas is a reference map of all human cells..."

The Centre of Genomics and Policy | and WYNG TRUST Visiting Scholar Program

2016-2019

Prof. Bartha Maria Knoppers, PhD

Thanks to a collaborative agreement with the Centre for Medical Ethics and Law of the University of Hong Kong (CMEL) and the WYNG Foundation, we are pleased to introduce the CGP / Wyng Trust Visiting Scholars Program.

The research of our Visiting Scholars will be dedicated to the emerging topics of (i) policy and regulatory responses to new genomic and reproductive technologies, (ii) international data sharing and (iii) epigenetics. This emphasis will cement existing connections with academics working in the Law and Technology Centre on issues of privacy and data sharing and the 'Children of 1997' project supported by the WYNG Foundation. This year, Professor Bartha Knoppers and Erika Kleiderman, a CGP Academic Associate, participated and joined Hong Kong (CMEL) partners at the "Policy and Regulatory Responses to New Genomic and Reproductive Technologies Symposium" on April 11, 2018, at The University of Hong Kong, Hong Kong. Professor Yann Joly participated in the International Human Epigenome Consortium Annual Meeting 2018 held at the University of Hong Kong on October 26, 2018, and in the Science days, held on October 27-28, 2018 where he had the opportunity to meet with CMEL colleagues.

The CGP would like to thank the WYNG Foundation for this award.

McGill CGP-CMEL SUMMER INTERNSHIPS

The CGP has proposed summer internships for two CMEL LLM students per year, providing one month supervision at CGP with appropriate mentorship and training.

RESEARCH AWARDS

CANADA RESEARCH CHAIR IN LAW AND MEDICINE

Prof. Bartha Maria Knoppers, PhD

Sequencing the human genome has provided the biomedical community with unprecedented opportunities for unlocking answers to some of the most complex human diseases.
Biomedical research is a collaborative, international endeavor and relies on the sharing of data to accelerate innovation and discovery. To share data in this way requires the harmonization of guidelines, policies and laws that span data protection, research ethics and health policy. The proposed Canada Research Chair in Law and Medicine seeks to enhance data sharing capacities among international researchers by activating, testing and most importantly implementing the human right of citizens' to benefit from science advances.

Chaires de recherche du Canada
Canada Research Chairs
Canada

RECHERCHE TRANSLATIONNELLE EN MÉDECINE PERSONNALISÉE, PERSPECTIVES ET ENJEUX POUR LE QUÉBEC

Prof. Yann Joly, PhD (DCL), Ad.E.

Designated as "Chercheur boursier niveau Junior 2" since July 2015, the awarded research grant will contribute to the development of a new type of infrastructure covering specific categories of diseases moving the translational domain closer to the clinic (eg. Q-CROC, Biobank Cohort of hospital ICM and Hepatopancreatobiliary (HPB) and Transplant Biobank Research at McGill University). These facilities are used to conduct research, monitor patients in real time and inform therapeutic discoveries (choices) or treatments relevant to the patient's specific genetic profile. This research grant will allow the conception of legal and ethical policies needed to establish optimal translational research infrastructure for safe, patient centered personalized medicine in Québec.

Fonds de recherche Santé Québec

COURSES

HGEN-660 B
GENETICS, ETHICS AND LAW

Instructor: Prof. Yann Joly, PhD (DCL), Ad.E.

The objectives of this course are to: 1) Introduce students to legal, ethical, and policy scholarship in genetics and related "omics" disciplines; 2) promote interdisciplinary collaboration and debate as a means of enriching scientific practices; 3) enable students to develop analytical research skills and to identify and critically evaluate the legal, ethical and policy issues that arise in genetic research and in clinical genetics.

The classes will be taught in seminar style, complemented by thematic class discussions and case studies. Themes covered in this course include, but are not limited to: genetic testing, genetic counseling, personalized medicine, privacy and confidentiality, population genetics, regenerative medicine, commercialization and intellectual property, genetic discrimination, and genetic analysis of social and behavioral traits. Through class lectures, case studies and discussions on a series of selected readings, students will be asked to reflect on the complex relationships between science, law, and ethics. Each member of the class will participate and contribute to the learning that occurs. Such a collaborative learning experience will be reflected in the way that the course is structured and the student's work is evaluated.

HGEN-674
RESEARCH INTERNSHIPS IN GENOMICS AND POLICY

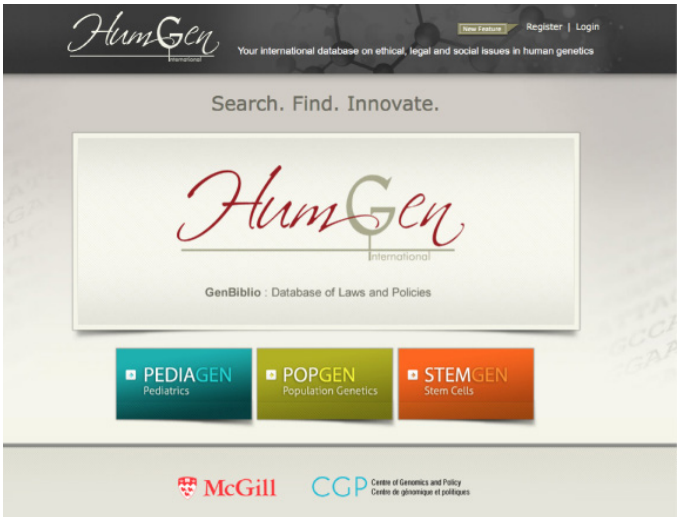
Instructor: Dr. Ma'n H. Zawati, LL.B., LL.M.

The Research Internships in Genomics and Policy course aims at providing 1 to 2 graduate students in the Human Genetics program with an opportunity to do research on the ethico-legal and policy issues in human genetics. More specifically, graduate students will be 1) introduced to the ethical, legal, and policy issues in human genetics in both the research and clinical settings; and 2) familiarized with social science research methodologies, especially international comparative analysis of normative policy and legal instruments. As an internship, these objectives will be achieved through active research under the supervision of a mentor working in the student's area of interest. Specific areas of research at the Centre of Genomics and Policy include but are not limited to: population genomics, biobanks, stem cells, reproductive technologies, paediatric genetic research, data protection, direct-to-consumer genetic testing, gene therapy, personalized medicine, and genetic counseling. Interested students are encouraged to explore the CGP website (www.genomicsandpolicy.org) to identify areas of interest. Undertaking an internship at the Centre of Genomics and Policy allows students to benefit from a close collaboration with experts at the crossroads of the ethico-legal, medical, and policy fields.

HUMGEN 2000-2018

Every great journey has an end. It is with sadness that we announce the end of a long journey for the HumGen International Database. Created in the year 2000 when the team was stationed at the Centre de recherche en droit public (CRDP) at the Université de Montréal, HumGen quickly became a vital source for a multitude of users interested in the ethical, legal and social issues in human genetics, whether researchers, clinicians, students or members of the public curious to learn about the challenges associated with this new field.

Indeed, faced with the rapid advances in human genetics research, HumGen provided multiple stakeholders with access to legislation, policy, guidelines, and recommendations from government and non-governmental organizations worldwide. The database incorporated more than 3000 documents emanating from more than 800 organizations around the world. Throughout the years, and thanks to a dedicated list of staff members, HumGen became a unique source of international genetic policy information and has been cited as a critical resource for geneticists who are interested in examining the wider implications of their work.



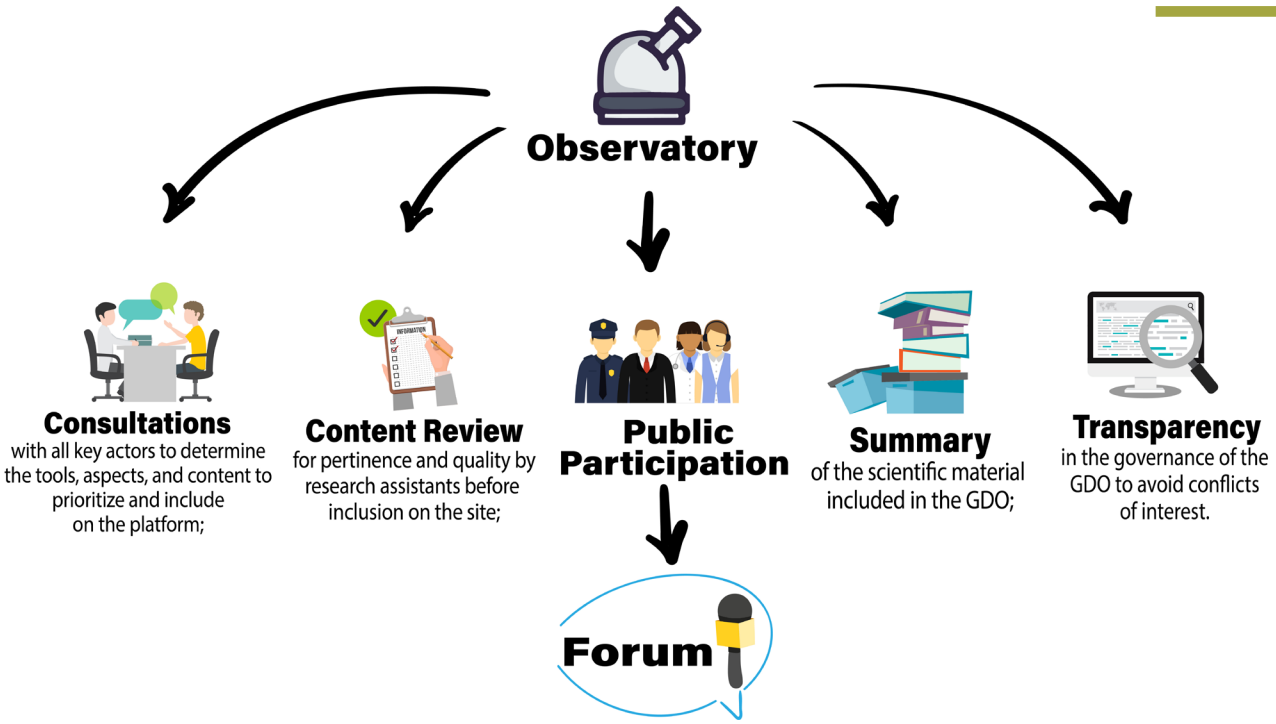
In 2012, the Centre of Genomics and Policy optimized its HumGen international database search engine to facilitate access to normative documents and to word and phrase searches. HumGen's new search functions at the time made research easier to conduct and for users to organize and follow international developments. A registration mechanism was put in place to enable users to create and save a personalized favourites list. More importantly, it allowed users to share their findings with colleagues and friends.

However, in the wake of the proliferation of databases around the world and the increased sophistication of search engines, we have decided to stop updating the database and archive its content for internal use at the Centre of Genomics and Policy as of the end of 2018.

We would like to thank all the dedicated members of the Centre of Genomics and Policy that have shared their expertise with the larger community by collecting, coding and uploading normative documents for the past 18 years. We also would like to thank our funders who made this knowledge translation possible. Finally, all our thanks go to our devoted users for their trust over the years. We appreciated your support and hope that you will continue to search, find and innovate.

- The CGP Team

GENETIC DISCRIMINATION OBSERVATORY



Fifteen years after the end of the Human Genome project, medical benefits are beginning to emerge. However, Canadians have concerns about the possibility that their genetic information be used in a way that violates human rights and ethics. These fears undermine participation in research with biobanks as well as the conduct of genetic tests recommended by a physician. To address genetic discrimination, the Canadian Law to Ban and Prevent Genetic Discrimination, S-201, was adopted in 2017. This law has benefited from the support of patients, geneticists and politicians. The legitimacy of S-201, however, has been put in question in an important advisory opinion of the Court of Appeal of Quebec and its application remains limited. Although S-201 is an important first step, it does not solve all the challenges posed by genetic discrimination. The goal of the GDO is to create a single communication platform to collectively improve the prevention of genetic discrimination.

The GDO will serve as a comprehensive online resource that offers an informative and collaborative platform providing access to the information and tools needed to effectively counter genetic discrimination. Specific aspects of the GDO will include a secure management system, integrated registration for participants and a database of projects, new developments, and events. Our overall approach will be guided by four fundamental principles:

- Consultations with all key stakeholders to determine the tools, aspects and content to be prioritized in the inclusion on the platform;
- Content reviewed for relevance and quality by research assistants prior to inclusion on the site;
- A summary of the scientific material included in the GDO; and
- Transparency in the governance of the GDO to avoid conflicts of interest.

Visit us at: https://gdo.global for more information.

TEAM PUBLICATIONS

BOOK CHAPTERS

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