CGP THE LAST 10 YEARS

MCGILL UNIVERSITY

150 YOUNG RESEARCHERS

288 RESEARCH PROJECTS

593 PUBLICATIONS

2 COURSES GIVEN TO 200+ STUDENTS

The Centre of Genomics and Policy (CGP) applies a multidisciplinary perspective, while collaborating with national and international partners, in several research areas including: stem cell research and therapies, personalized medicine, prevention and treatment of cancer, data sharing, pediatrics, genetic counseling, digital health and AI, intellectual property and open science, epigenetics, intersex and diversity in health, gene editing, genetic discrimination and biobanking (population genetics).

RESEARCH AWARDS

Canadian Research Chair in Law and Medicine
(Prof. Bartha Maria Knoppers)

Chaires de recherche du Canada / Canada Research Chairs
Canada

FRQS Chercheurs boursier Junior 1 & 2
(Prof. Yann Joly)

FRQS Chercheurs boursier Junior 1
(Prof. Ma'n H. Zawati)

Fonds de recherche Santé Québec

Bartha Maria Knoppers, Director
Yann Joly, Research Director
Ma'n H. Zawati, Executive Director
Nicole Palmour, Coordinator

Denise Avard, Past Research Director
Claude Laberge, Scientific Advisor

2023 RARE DISEASE PROJECTS

THE LAST 10 YEARS

The Centre of Genomics and Policy has been actively engaged in rare disease research, policy and tool development for more than ten years. This summary will provide an overview of the breath and depth of our engagement and expertise in this domain.

APR 2022 – SEP 2023
ALL FOR ONE HEALTH DATA ECOSYSTEM (HDE)

OCT 2020 – OCT 2022
DEVELOPMENT OF AN ETHICAL AND LEGAL FRAMEWORK FOR THE DEPLOYMENT OF RÉSEAU QUÉBEC DIAGNOSTIQUE MOLÉCULAIRE (MSSS) ACTIVITIES AND THE HARMONIZATION OF PATIENT CONSENT

MAR 2019 – MAR 2022
CANADIAN GENOMICS PARTNERSHIP FOR RARE DISEASE (ALL FOR ONE): POLICY TOOLKIT

MAR 2018 – FEB 2023
CARE4RARE CANADA: HARNESSING MULTI-OMICS TO DELIVER INNOVATIVE DIAGNOSTIC CARE FOR RARE GENETIC DISEASES IN CANADA (C4R-SOLVE)

MAR 2016 – FEB 2021
PROGRAMME DE RECHERCHE ET D'INNOVATION SUR LES MALADIES RARES (PRISMES)

MAR 2013
FEB 2017

ENHANCED CARE FOR RARE GENETIC DISEASES IN CANADA

MAR 2013
FEB 2014

RECHERCHE SUR LES MALADIES RARES: VIE PRIVÉE «BON GRÉ MAL GRÉ»? (PRISMES)

JAN 2012
FEB 2017

FRAMEWORK FOR DECISION-MAKING FOR RARE DISEASES

DEC 2011
NOV 2017

DEVELOPING A CANADIAN FRAMEWORK FOR EVALUATION AND DECISION-MAKING FOR EXPENSIVE DRUGS FOR RARE DISEASES THROUGH INNOVATION, VALUE, AND PRIORITY SETTING

MAY 2011
NOV 2012

RADICAL - RARE DISEASE CONSORTIUM FOR AUTOSOMAL LOCI

FEB 2011
JUL 2012

FINDING OF RARE DISEASE GENES IN CANADA (FORGE)

OCT 2008
SEP 2013

CIHR EMERGING TEAM IN GENOMICS SCREENING

CGP RARE DISEASE PUBLICATIONS

ARTICLES

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Driver, Hannah G., Hartley, Taila, Price, Magda E., Turinsky, Andrei L., Buske, Orion J., Osmond, Matthew, Ramani, Arun K., **Kirby, Emily**, Kernohan, Kristin D., Couse, Madeline, Le, Hannah G.B.H., Herscovich, Andrea, Marshall, Christian R., Statia, Andrew, Care Rare Canada Consortium, **Knoppers, Bartha**, Brudno, Michael, Boycott, Kym M. Genomics4RD:An integrated platform to share Canadian deep phenotype and multiomic data for international rare disease gene discovery. Human Mutation. 2022;43:800-811. Available from: https://onlinelibrary.wiley.com/doi/pdf/10.1002/humu.24354.

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Nguyen, Minh Thu, Goldblatt, Jack, Isasi, Rosario, Jagut, Marlene, Jonker Anneliene, Hechtelt, Kaufmann, Petra, Ouillade, Laetitia, Molnar-Gabor, Fruszina, Shabani, Mahsa, Sid, Eric, **Tassé, Anne Marie**, Wong-Rieger, Durhane, **Knoppers, Bartha Maria**, on behalf of the IRDiRC-GA4GH Model Consent Clauses Task Force. Model consent clauses for rare disease research. BMC Medical Ethics. 2019;20(1):55.
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GENETIC DISCRIMINATION OBSERVATORY

Genetic discrimination involves treating differently and negatively or unfairly profiling individuals or a group relative to the rest of the population based on actual or presumed genomic and other predictive data.

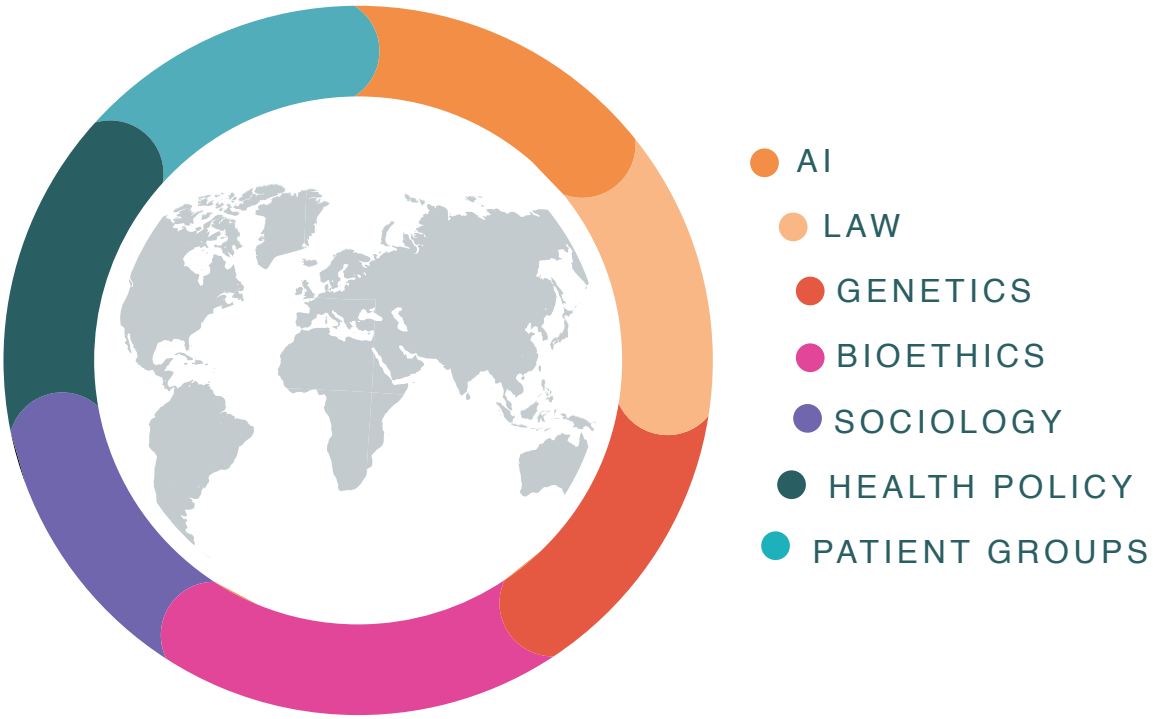
The Genetic Discrimination Observatory (GDO) is a network of international experts and collaborators from over 23 jurisdictions dedicated to researching and preventing genetic discrimination.

GDO MAIN OBJECTIVES

1. Document the issue of genetic discrimination in a scientific and evidence-based manner.

2. Engage the public, policymakers and other stakeholders in a collective debate about genetic discrimination.

3. Use this information to assess existing normative models work best and develop new ones.



From January to November 2022,
the GDO website reached
164,000 VISITORS
(an average of 13,700 visits/month)

COLLABORATIVE RESEARCH

GDO is now in collaboration with the Global Alliance for Genomic and Health (GA4GH) to develop tools and policies to address genetic discrimination. The first product of this collaboration is the information brief Genetic Discrimination: Implications for Data Sharing Projects (GeDI).

www.ga4gh.org/wp-content/uploads/Genetic-Discrimination-Dec.-2-2021.docx.pdf
approved by GA4GH/GDO in January 2022

GDO INTERNATIONAL COLLABORATIONS

26 COUNTRIES 27 JURISDICTIONS

- CANADA
- UNITED STATES
- MEXICO
- COLUMBIA
- CHILE
- ICELAND
- IRELAND
- UNITED KINGDOM
- DENMARK
- BELGIUM
- FRANCE
- GERMANY
- SPAIN
- SOUTH AFRICA
- UGANDA
- UKRAINE
- LEBANON
- ISRAEL
- KAZAKHSTAN
- INDIA
- CHINA
- HONG KONG
- TAIWAN
- SOUTH KOREA
- JAPAN
- AUSTRALIA

GDO PUBLICATIONS

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GenomeCanada

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