The Development and Influence of Public Health Genomics 2023: ELSI Perspectives

Prof. Bartha Maria Knoppers, Director

Centre of Genomics and Policy,

Department of Human Genetics,

Faculty of Medicine and Health Sciences

McGill University

McGill

CGP Centre of Genomics and Policy
Centre de génomique et politiques

25 years Of Commonalities: Technologies and Individuals

Contributions and roles of industry, legislation, and patents and more recently, data sharing efforts.

Evolution of Genome mapping/ sequencing

Remarkable leadership by visionary individuals over 25 years.

Ethics in the science of genomics was perhaps the most important catalyst for the emergence of public health genomics as both were crossing fields hitherto so narrowly circumscribed/siloed. (eg HGP; HUGO)

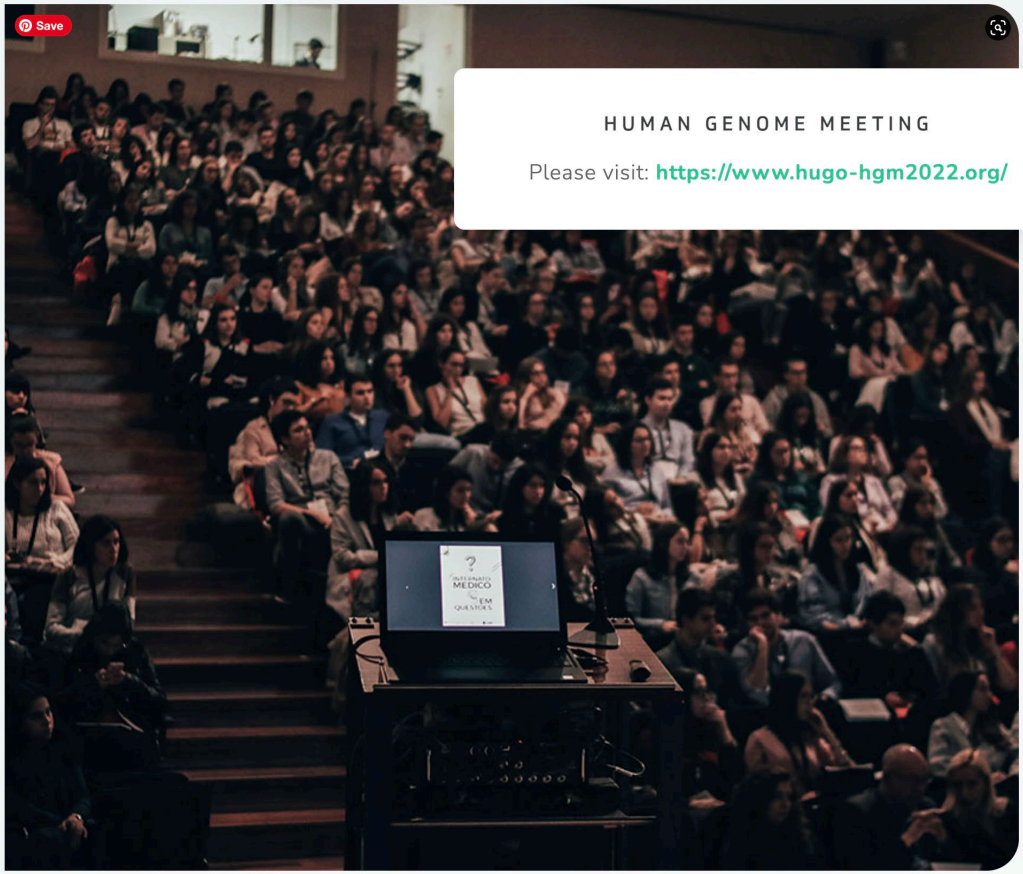
B. HUMAN GENOME ORGANIZATION

EVENTS

Coming up...

HUMAN GENOME MEETING

Human Genome Meeting (HGM) is a series of annual conferences organized by the Human Genome Organisation (HUGO). It started as a meeting dedicated for Human Genome Mapping. Over the years, with the completion of the Human Genome Project, HGM has evolved from a small targeted meeting into a scientific conference for all genetic and genomic researchers; an excellent platform for industry partners and bio-technology companies as well as pharmaceutical giants; and a fantastic reunion for fellow scientists and networking opportunity for established and young investigators.



C. THE ETHOS OF DATA SHARING – KEY TO THE SUCCESS OF THE HGP 1990-

- The HGP was followed by the creation of population biobanks (2003 -) for infrastructure science.
- Built on the foundation of both public health and population screening efforts. (eg U.K Biobanks, Estonia, Japan)

D. BIOBANKS / BIORESOURCES

E. Public Participation and Data Sharing

- National biobanks/resources → GRAPH-Int; P3G; PHG …
- Recent international response to the COVID pandemic…equity; value judgements; trade-offs; uncertainty… (E. Emmanuel et al(2022 NEJM381:17);
- The global public goods nature of public health genomics.

F. CANADA'S HEALTH SYSTEM HAS A PROBLEM

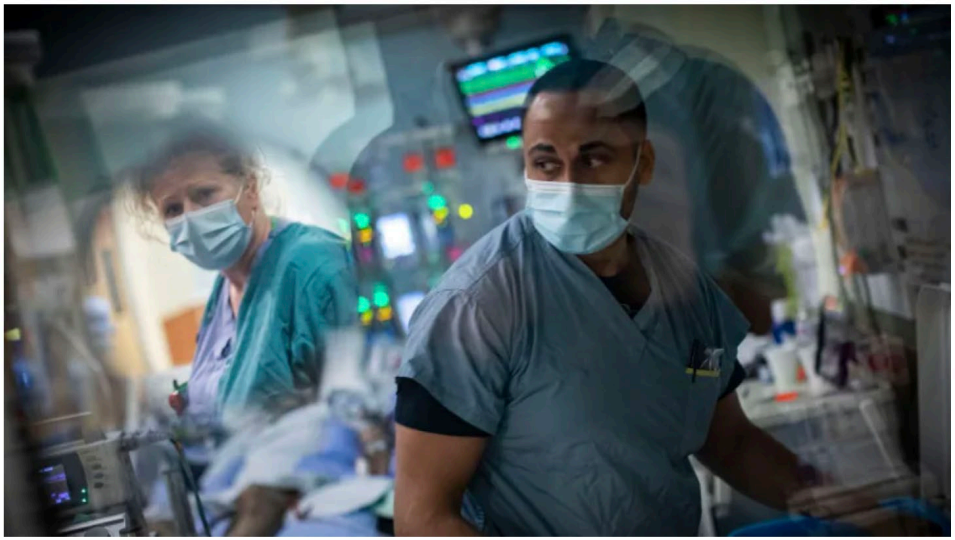
CBC NEWS

Canada

Canada's health-care system has a data problem, experts say. And it puts patients at risk

'It's not good to be flying blind,' says one doctor pushing for change

Yvette Brend · CBC News · Posted: Nov 17, 2022 4:00 AM ET | Last Updated: November 17, 2022



Nurse Dave Riar checks on a patient in the intensive care unit at the Royal Columbia Hospital in New Westminster, B.C., on Thursday, March 31, 2022. Medical front-line staff in Canada say problems persist when it comes to sharing everything from patient information to aggregate medical and staffing data. (Ben Nelms/CBC)

G. OF PUBLIC HEALTH GENOMICS: QUO VADIS?

The gradual integration of genomics into public health (and vice versa).

The emergence of precision public health genomics via population screening/stratification/scores

A new understanding of genomic susceptibilities, and of, resistance in diverse communities and subpopulations.

Polygenic risk scores illustrate this phenomenon but risk scores are not diagnoses.

H. OF SCREENING / STRATIFICATION AND SCORES

Journal of *Personalized Medicine*

MDPI

Review

Of Screening, Stratification, and Scores

Bartha M. Knoppers [1,*], Alexander Bernier [1], Palmira Granados Moreno [1] and Nora Pashayan [2]

[1] Centre of Genomics and Policy, Faculty of Medicine, McGill University, 740 Avenue Dr. Penfield, Suite 5200, Montreal, QC H3A 0G1, Canada; alexander.bernier@mcgill.ca (A.B.); palmira.granadosmoreno@mcgill.ca (P.G.M.)
[2] Department of Applied Health Research, University College London, 1-19 Torrington Place, London WC1E 7HB, UK; n.pashayan@ucl.ac.uk
* Correspondence: bartha.knoppers@mcgill.ca

Abstract: Technological innovations including risk-stratification algorithms and large databases of longitudinal population health data and genetic data are allowing us to develop a deeper understanding how individual behaviors, characteristics, and genetics are related to health risk. The clinical implementation of risk-stratified screening programmes that utilise risk scores to allocate patients into tiers of health risk is foreseeable in the future. Legal and ethical challenges associated with risk-stratified cancer care must, however, be addressed. Obtaining access to the rich health data that are required to perform risk-stratification, ensuring equitable access to risk-stratified care, ensuring that algorithms that perform risk-scoring are representative of human genetic diversity, and determining the appropriate follow-up to be provided to stratification participants to alert them to changes in their risk score are among the principal ethical and legal challenges. Accounting for the great burden that regulatory requirements could impose on access to risk-scoring technologies is another critical consideration.

PERSPECTIVE
https://doi.org/10.1038/s41591-021-01549-6

nature medicine

Check for updates

Responsible use of polygenic risk scores in the clinic: potential benefits, risks and gaps

Polygenic Risk Score Task Force of the International Common Disease Alliance

Adebowale Adeyemo[1], Mary K. Balaconis[2], Deanna R. Darnes[3], Segun Fatumo[4,5,6], Palmira Granados Moreno[7], Chani J. Hodonsky[8], Michael Inouye[9,10,11,12,13,14,15] ✉, Masahiro Kanai[16,17,18,19], Kazuto Kato[20], Bartha M. Knoppers[7], Anna C. F. Lewis[21], Alicia R. Martin[16,17,18], Mark I. McCarthy[22], Michelle N. Meyer[23], Yukinori Okada[19], J. Brent Richards[24,25,26], Lucas Richter[27], Samuli Ripatti[16,18,28,29], Charles N. Rotimi[1], Saskia C. Sanderson[30,31,32], Amy C. Sturm[33], Ricardo A. Verdugo[34,35], Elisabeth Widen[29], Cristen J. Willer[36,37,38] ✉, Genevieve L. Wojcik[39] and Alicia Zhou[40]

Polygenic risk scores (PRSs) aggregate the many small effects of alleles across the human genome to estimate the risk of a disease or disease-related trait for an individual. The potential benefits of PRSs include cost-effective enhancement of primary disease prevention, more refined diagnoses and improved precision when prescribing medicines. However, these must be weighed against the potential risks, such as uncertainties and biases in PRS performance, as well as potential misunderstanding and misuse of these within medical practice and in wider society. By addressing key issues including gaps in best practices, risk communication and regulatory frameworks, PRSs can be used responsibly to improve human health. Here, the International Common Disease Alliance's PRS Task Force, a multidisciplinary group comprising expertise in genetics, law, ethics, behavioral science and more, highlights recent research to provide a comprehensive summary of the state of polygenic score research, as well as the needs and challenges as PRSs move closer to widespread use in the clinic.

I. New Risks?

- Health / Genomic data literacy
- WGS in NBS (beyond the neonatal intensive care eg. UK/USA initiatives).
- WGS neonatal futuristic report cards may damage public trust and participation and the rights of the child?

Rahimzadeh V, Friedman JM, deWert G, Knoppers BM. Exome/Genome-Wide Testing in Newborn Screening: A Proportionate Path Forward. Frontiers in Genetics. 2022; Vol. 13 1-9.

J. Genomics Eng,… Sequence 100k Newborns / USA

BIOMEDICAL RESEARCH

Sequencing projects will screen 200,000 newborns for disease

U.K. and New York City efforts face cost and ethical issues

By **Jocelyn Kaiser**

The once-futuristic idea of sequencing every newborn child's DNA to screen for genes that could shape their future health is being put to two major tests. The United Kingdom this week announced plans to sequence the genomes of 100,000 newborns for about 200 rare genetic diseases starting next year. In New York City, a similar project already underway will screen 100,000 babies.

The goal is to catch treatable diseases that standard newborn screening cannot detect. If sequencing delivers an early warning of a problem, the baby could receive care that averts permanent disability or even death.

Genome sequencing can reveal whether newborns carry any of hundreds of genetic diseases.

$1000—but is getting cheaper, could detect many more disorders, such as thyroid conditions that can cause brain damage.

Genomics England's $129 million Newborn Genomes Programme will invite expecting parents in England who are receiving care through the National Health Service (NHS) to sign up starting in late 2023. The aim is to enroll 100,000 newborns over 2 years. To avoid raising the alarm about gene variants whose risk is uncertain or that only cause disease in adulthood, parents will only receive results for 200 treatable diseases caused by well-studied genetic variants that are almost certain to cause symptoms before age 5.

The project expects to spot at least 500 newborns with genetic disease. If such testing were used across the United Kingdom, researchers estimate it would find some 3000 babies per year with these diseases.

The project has public support, but some experts argue the money would be better spent on expanding standard U.K. screening, which now covers just nine diseases. Others say following up on the screening results will tax an already overstretched NHS. "It seems like the economics is the big unanswered question," says bioethicist Josephine Johnston of the Hastings Center.

The New York City project, launched in September, is led by Columbia University geneticist Wendy Chung and supported by

K. ELSI=?

- 1990 → building the public health rationale for genomics/genetics e.g. UNESCO, 1997 Universal Declaration on the Human Genome and Human Rights;
- 2005 Universal Declaration on Bioethics and Human Rights
- 2000 → building the population /citizen rationale for biobanks (eg. Infrastructure science)
- 2013 → develop the human right to benefit from science GA4GH, Framework for the Responsible Sharing of Genomic and Health –Related Data
- 2020 → building open science (UNESCO 2021 Recommendation)
- 2023 → towards a WHO Pandemic Treaty?

L. UNESCO PROMOTE OPEN SLIDE SCIENCE

UNESCO
RECOMMENDATION
ON
OPEN SCIENCE
(NOVEMBER 2021)


CGP
Centre of Genomics and policy
Centre de génomique et politiques