TERMS OF REFERENCE FOR A CCMG AD HOC WORKING GROUP

Clinical Practice Advisory Document (CPAD) for Genomic Data Sharing for Clinical Genetic Laboratories in Canada

Approved by CCMG Board of Directors: August 2024

Brief Background for Formation of Working Group:

The American College of Medical Genetics and Genomics (ACMG), the Association for Molecular Pathology (AMP), the National Society of Genetic Counselors (NSGC), the National Institutes of Health (NIH), Australian Genomics, the United Kingdom (Wellcome Trust), the Clinical Genome Resource (ClinGen), the GA4GH and other organizations all have policies, recommendations and / or guidelines on genomic data sharing (1-9). Furthermore, the Food and Drug Administration (FDA) is recognizing shared variant data in publicly accessible databases as valid scientific evidence to support the clinical validity of genotype-phenotype relations (10).

Canadian clinical genetic laboratories are leaders in genomic data sharing. The Canadian Open Genetics Repository (COGR – opengenetics.ca) has served as an example for other national and international data sharing initiatives (11). Several national publicly available databases have local data-sharing programs. Examples of these include the CanVar-UK (canvaruk.org), a platform for sharing data from cancer susceptibility genes by the Can-VIG-UK group, and the Shariant platform from Australian clinical genetic-testing laboratories (12-13). The COGR was designed to enhance and promote data sharing, both variant level data and interpretation drawn from existing data holdings at clinical laboratories across the country, creating collaboration between the different laboratories, and constant improvement in the quality and applications of genetic testing data for clinical purposes (11,14-16). In addition, the CCMG has a practice guideline that encourages data sharing and reporting of incidental findings (17); however, there is no formal policy or position statement on genomic or variant level data sharing that provides guidance on which data can or should be shared in the context of clinical genetic testing laboratories.

Canada has a patchwork of regional and provincial health authorities, hundreds of hospitals, home and community care centers with a large variety of IT solutions. Major differences in provincial, regional and institutional policies on the use of personal health information raise additional challenges. With evolving data sharing practices (e.g., cloud computing), laboratory information systems, interpretation and reporting standards, variant classifications changes, and availability of clinical or laboratory resources, there is sufficient complexity to data sharing practices that requires some guidance and standardization. Genomic data generated in the clinical diagnostic laboratory is an extremely valuable and ever-growing resource, which has been inaccessible to patients and providers and is at risk of being lost. Sharing genomic data from the clinical setting will lead to better quality assurance, standardizing of results between clinical laboratories and improve diagnoses. Data sharing will add value to public health testing programs and will aid clinicians in managing disease more effectively, advising family members and improve our understanding of the relationship between genes and disease.

This proposed CCMG CPAD seeks to provide some guidance and points to consider to clinical laboratories as well as to payers, regulators, and providers that encourages sharing of clinical variant data and their interpretations using modalities that assure patient and provider privacy protection. This document will reinforce the CCMG's reputation for the high quality of clinical genomic data sharing in Canada.

References

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Mandate

The purpose of the Canadian Genomic Data Sharing Working Group is to develop and publish a CPAD for the sharing of genomic and variant level data from clinical laboratories. The proposed document will outline the advantages and benefits of data sharing in establishing accurate diagnoses, effective disease management, advice to family members, and improved disease understanding. It will also summarize the perceived harms and the associated risks of misuse of genetic data, as well as the risks and harms of not sharing genetic data.

Further, the document will provide guidance and considerations on sharing of different types of genetic and clinical data, standards and minimal requirements for data sharing, as well as justifications for data elements that can be shared and what levels of permission may be needed in the context of 1) de-identified clinical variant data and interpretations from diagnostic laboratories, 2) aggregate level data and 3) sharing of genomic individual-level data with phenotype. The document will also provide a statement on the ethics and data governance framework for secure health and genomic data sharing. To ensure we are transparent in the methods used to develop this document and that the community is aware of the limitations of our recommendations, we will abide by AGREE II reporting practices (18).

Timeline

Formation of group: July 2023

Meeting intervals: monthly meetings, starting September 2023 – February 2024

Publish date – February 2025

Membership

Co-Chairs:

Jordan Lerner-Ellis, ABMGG, FACMG (ON) – Associate Member, CCMG

Kathy Chun, FCCMG (ON)

Members:

- Ma'n Zawati, PhD (QC) – The Center for Genomics and Policy - Law and ethics
- Christian Marshall, FCCMG (ON) – Molecular Geneticist, member of the All for One project
- Kym Boycott, FCCMG (ON) – Clinical Geneticist, member of the All for One project
- Francois Bernier, FCCMG (AB) – Clinical Geneticist, member of the All for One project
- Tanya Nelson, FCCMG (BC) – Molecular Geneticist, member of the All for One project
- Ian King, ABMGG, FACMG (ON) – Molecular Geneticist
- Natascia Anastasio, FCCMG (QC) – Medical Geneticist / Molecular Geneticist
- Ron Agatep, FCCMG (MB) – Molecular Geneticist, Board Member
- Nick Antonishyn, FCCMG (SK)– Molecular Geneticist
- Darren O'Reilly, FCCMG (NFLD)- Molecular Geneticist, Chair of Laboratory Practice
- Victor Martinez, PhD (NS) – Molecular Geneticist, member of the All for One project
- Ryan Lamont, FCCMG (AB) – Molecular Geneticist
- Stephen Yip, FRCPC (BC) – Molecular Pathologist
- Yael Fisher, MD (ON) – Molecular Pathology Fellow – drafting of guidelines and administration (meetings coordination and notes)
- Taila Hartley, PhD (ON) – Genetic Counsellor, member of All for One project

Confidentiality

Matters discussed at meetings and teleconferences are confidential and may not be disclosed to others. Exclusion to this includes information that was previously published or in the public domain, or information that was already known to the member and was not acquired by the member directly or indirectly from the Working Group.

Conflict of Interest

Members should disclose any known or perceived conflicts of interest. When the main goal of the Working Group is development of educational materials, guidelines or recommendations on a specific topic, participation in similar activities (during the time frame of the Working Group activities) on the same topic led by external organizations might be perceived as a conflict of interest. If such situations arise, the member of the Working Group should discuss this with the chair(s) and Board of Directors (BoD) representative who, if required, could seek the advice of the BoD on that matter.

Appointment and term of chair

Two co-chairs are proposed for the working group: Dr. Jordan Lerner-Ellis and Dr. Kathy Chun.

Dr Jordan Lerner-Ellis is Director & Head of Advanced Molecular Diagnostics at Toronto's Mount Sinai Hospital, Sinai Health; Associate Professor in the University of Toronto's Department of Laboratory Medicine and Pathobiology and Clinician Scientist at the Lunenfeld-Tanenbaum Research Institute. Dr Lerner-Ellis is principal investigator of the Canadian Open Genetics repository, an initiative that shares genetic data both nationally and internationally. He is a member of the Human Genome Organisation's (HUGO) scientific advisory and pathogenicity committees with a focus on variant interpretation efforts aimed at improving our understanding of the relationship between DNA variants and disease.

Dr Kathy Chun is a Clinical Laboratory Director in Genome Diagnostics at The Hospital for Sick Children, where she is involved in clinical service and in translational research; Associate Professor in the University of Toronto's Department of Laboratory Medicine and Pathobiology and a Project Investigator at the Hospital for Sick Children's Research Institute. Dr. Chun has an interest in clinical laboratory quality management and has been an active member of the Genetics Scientific Committee of the Institute for Quality Management in Healthcare (IQMH) for many years, most currently as Chair. She led the development of a shared variant database in collaboration with the Institute for Quality: the Canadian Network for Public Health Intelligence for Ontario.

Conduct of Meetings

- The chair or co-chairs will convene regular meetings of the Ad Hoc Working Group, with approximately one meeting held once every two months or more frequently, if required.
- Quorum for decisions will be at least 50% of Working Group members
- Draft minutes of all meetings will be prepared and circulated to the Working Group members for comment, ideally within 10 working days of the meeting.
- All meeting minutes will be made available to the CCMG BoD via the BoD Representative.

Reporting

The co-chairs will report to the CCMG BoD via the BoD Representative for monthly updates.

After completion of the manuscript, it will be submitted to the CCMG BoD to seek feedback from the CCMG Clinical Practice and Laboratory Practice committees.

Documents will be circulated to the full CCMG membership.

The final manuscript will be submitted to the BoD for approval.