2025 CAGC & CCMG Joint Conference Schedule

Banff Centre of the Art, Banff, AB, October 5-8, 2025

Sunday October 5		
8:30-8:35		Welcome to Fertility and Beyond: Exploring Management, Technology, and the Patient Journey Short Course
8:35-9:35 1 hour, including 10 mins Q&A	Plenary	**From fertility to menopause, hormonal considerations in HBOC** Speakers: Michelle Jacobson Description: *Dr. Jacobson will review fertility, contraception, risk reproduction and menopause management journey for carriers of hereditary breast and ovarian cancer genes.*
9:35-10:00		Refreshment Break
10:00-12:30 2.5 hours, including 20 mins Q&A	Plenary	**Crash Course on Preimplantation Genetic Testing** Speakers: Dr. Maude Lefebvre Dr. Svetlana Madjunkova Jaspreet Sekhon-Warren Erica Pai Charlotte Emmerson Description: *This panel presentation will provide a summary of preimplantation genetic testing options, indications for testing, and summarize current federal policy on funding for PGT.*
12:30-14:00		Lunch
14:00-15:00 1 hour, including 10 mins Q&A	**Plenary**	**Understanding Fertility Through Personal Narratives: Two Patient Journeys** Speakers: Trista Harrison Joanna Kirsh Description: *In this session, we will share two distinct patient perspectives on healthcare and fertility, offering insight into the personal challenges, triumphs, and lessons learned along the way. By exploring how these experiences have shaped our identities and influenced the work we do today, we aim to foster a deeper understanding of the emotional, physical, and psychological dimensions of fertility care.*
15:00-15:15		Refreshment Break
15:15-16:15 1 hour, including 10 mins Q&A	**Plenary**	**Monogenic causes of oocyte-factor infertility: Beyond the basics** Speaker: Meaghan Doyle Description: *Over the past decade, a significant number of genes related to infertility have been discovered. But for the 1 in 6 people of reproductive age who experience infertility and/or pregnancy loss these discoveries haven't become a routine part of their clinical care. In this session we will review some key phenotypes associated with monogenic causes of oocyte-factor infertility including oocyte/zygote/embryo maturation arrest and recurrent triploidy, while highlighting a few key genes of*

		interest. Finally, we will emphasize the important role of genetic counselling if clinicians are considering testing for these genes.
18:30-19:30 1 hour, including 10 mins Q&A		**Opening Keynote - Advancing Rare Disease Precision Health: It Takes a Country** Speaker: Dr. Kym Boycott Description: *Fifteen years ago, Canada embarked on a collaborative journey to enhance the diagnosis and management of rare diseases, aiming to deliver precision health to patients and their families. Through innovative, large-scale interdisciplinary collaborations, Canadian initiatives advanced omic technologies, national and international data sharing, and functional studies propelling the country to the forefront of the global stage. Looking ahead to the next fifteen years, we must build on this progress and apply the lessons we've learned to drive the integration of genomics across all health sectors, accelerate the development of innovative treatments, and ensure comprehensive support for patients and their families to live their best lives. The road ahead is challenging, but by working together, we can make a meaningful and lasting impact.*

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Monday October 6		
9:00-10:30 1.5 hours, including 15 mins Q&A	**Plenary**	**The future of the genetics service in Canada: evolving practices and roles in the era of genomic medicine** Speakers: Dr. Michael Mackley Dr. Kym Boycott Dr. Alison Elliot Angela Krutish Dr. Jillian Parboosingh Dr. Heleen Arts Description: *The field of medical genetics is rapidly changing. As understanding of the human genome and its role in health and human disease continues to expand, so do the indications and demand for genetic testing. Genetics services in Canada are experiencing unsustainable pressures, demonstrated through growing waitlists, while the genetics workforce remains stable. Concurrently, jurisdictions are expanding access to genomic testing to non-geneticist clinicians, with testing volumes expected to increase in response. The genetics service, and those who work in it, must adapt and rise to meet the needs of this new era.* *In this session we will discuss the genetics service in the era of genomic medicine. Through an interdisciplinary panel, including both late and early career panelists from coast-to-coast, we will explore the evolving roles of the medical geneticist, genetic counselor, and clinical laboratory geneticist. Panelists will discuss ongoing projects exploring professional identity, the delivery of genetics services, and the positioning of the genetics service in the diagnostic care pathway. We will work towards a shared vision around the future of genetics service delivery in Canada, and the unique and valuable role the genetics service plays in the broader health system.*
10:30-11:00		Break

11:00-12:00 1 hour, including 10 mins Q&A	**Plenary**	**All About Antisense Oligonucleotides: Eligibility For, Communication About, and Development of Individualized ASOs** Speakers: David Cheerie Amy Pan Dr. Gregory Costain Description: *As genome-wide sequencing shortens diagnostic odysseys, individuals are increasingly embarking on therapeutic odysseys. Proof-of-concept exists for antisense oligonucleotides (ASOs) as a genetic medicine customized for an individual's specific genetic variant and/or ultra-rare condition. Questions about ASOs - in our laboratories and in our clinics - are increasingly common. Our presenters are Canadian members of the N=1 Collaborative (N1C) (n1collaborative.org/), a global initiative to standardize ultra-rare "n-of-1/few" therapy development for safe and equitable delivery to individuals with rare diseases. In this session, we will present the N1C VARIANT (Variant Assessments towards Eligibility for Antisense Oligonucleotide Treatment) guidelines (https://www.medrxiv.org/content/10.1101/2024.09.27.24314122v1; Am J Hum Genet, accepted). We will provide resources/tools that facilitate this variant triage process, work through specific real-world examples, and show data that a meaningful proportion of genetic diagnoses identified by genome-wide sequencing may be amenable to ASO treatment (https://www.medrxiv.org/content/10.1101/2025.02.10.25321921v1). We will discuss frequently asked questions and specifically outline an approach to explaining why an ASO may not be feasible for a given patient. Last, we will critically review factors necessary for pre-clinical development and clinical administration, and the resource allocation, ethical, and equity concerns that shape this emerging field of genetics.*
12:00-14:00		Lunch in Exhibit Hall (posters 12:00-13:30 with judging – author's present)
14:00-15:30 1.5 hours, including 15 mins Q&A	**Plenary**	**The Canadian All for One Precision Health Initiative: Insights and lessons from the past five years** Speakers: Dr. Kym Boycott Dr. Taila Hartley Dr. Victor Martinez Dr. Jacques Michaud Camille Varin-Tremblay Meredith Gillespie Dr. Cheryl Rockman-Greenberg Angela Krutish Dr. Francois Bernier Dr. Tanya Nelson Description: *The All for One Precision Health Initiative is a groundbreaking national effort to transform access to high-quality clinical genomic testing for suspected rare disease across Canada. Launched by Genome Canada in 2018, this initiative includes six provincial Genomic Applications Partnership Program (GAPP) projects and two national projects, one devoted to providing policy support to regions and the other focused on co-design and pilot of a national Health Data Ecosystem. Together, the All for One partnership represents a cohesive strategy to address disparities in access, foster genomic capacity, and enable data-sharing across the country. This plenary session will highlight the high-level outcomes from these eight projects, showcasing their collective impact on precision health. Each project will be presented briefly,*

		offering insights into unique provincial contexts and lessons learned in local environments. Collectively, All for One represents an exceptional effort to align and unify highly disparate and siloed systems to improve genomic medicine for all Canadians. The session will conclude with a panel discussion on the future of Canadian precision health, focusing on pathways to sustain equitable access, fostering interprovincial collaboration, and strengthening Canada's leadership in genomic medicine.
15:30-16:00		Refreshment Break
16:00-17:00 1 hour, including 10 mins Q&A	**Plenary**	**Germline versus somatic: variants of uncertain origin (VUO) in hereditary cancer testing** Speakers: Dr. My Linh Thibodeau Dr. Laila Schenkel Description: *This presentation explores the complexities of interpreting genetic variants of uncertain origin (VUO) identified through blood DNA testing. Attendees will learn to recognize clinical and laboratory cues that suggest a variant may not be of germline origin, with a discussion of potential sources such as mosaicism and somatic mutations. Drawing on genetics clinic experience, the presentation outlines a standard approach to investigating VUOs, including testing methods and clinical follow-up considerations. Real-world case studies will illustrate diagnostic challenges and management strategies for patients with VUO findings. Finally, current guidelines and diagnostic strategies for evaluating VUOs will be assessed to support informed clinical decision-making.*

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Tuesday October 7		
8:00-9:00 1 hour, including 10 mins Q&A	**Breakfast Symposium**	**Achondroplasia: what is new in 2025** Speaker: Peter Kannu Description: *Achondroplasia is an autosomal dominant skeletal dysplasia characterized by disproportionate short stature and associated complications such as motor delay, obstructive sleep apnea, recurrent otitis media, and bowing of the lower limbs. There is an elevated risk of sudden death during infancy, primarily due to foramen magnum stenosis. Management has traditionally relied on a multidisciplinary team approach to monitor and address the evolving needs of the child throughout development. Vosoritide is the only FDA approved therapy for use in individuals with achondroplasia starting from birth. In addition, several investigational therapies are undergoing evaluation in Phase 2 and 3 clinical trials. The overarching goal of treatment is to reduce morbidity and mortality while enhancing quality of life for affected individuals.*
9:00-10:15 75 mins, including 15 mins Q&A	**Breakout Sessions** (x3 rooms)	• **Where efforts at the CAGC and CCMG around EDI are going** Speakers: Dr. Lauren Chad Dr. Elaine Goh Laura Redondo Kenna Kelly-Turner Description: *This interactive and focused conversation is intended to update participants on the current state of EDI in the CAGC-CCMG communities as well as identify priority activities for these two groups to advance EDI in the genetics space moving forward.* • **Practicalities, Planning and Pitfalls of Mainstreaming General Genetic Testing** Speakers: Dr. Andrea Guerin Rachelle Dinchong Angelique Lang Description: *Mainstreaming has been used for many years within cancer genetics. Although many expert groups call for expansion of mainstreamed testing, it has yet to be widely adopted for general genetics due inadequate education for non-genetics care providers, lack of time and educational resources coupled with logistical barriers. However, both Saskatchewan Health Authority and Queen's University set up 'mainstream' testing based on common referral indications, such as developmental delay and familial variant testing, to combat long wait times in Genetics. Clinicians that order testing are supported by genetic counsellors/ genetic assistants. This presentation will outline each province's implementation process of 'generalized' mainstreaming including the process behind selection of indications for testing, logistics of ordering, review results of testing and the impact on patient wait times.* • **Bridging the Evidence Gap: A Measurement System for Evaluating the Utility of Genetic Testing** Speakers: Dr. Robin Hayeems Christine Armour Wendy Ungar Salma Shickh

		Description: *As genetic testing options continue to expand in complexity and costs, determining the full value of these technologies requires a set of metrics that extends beyond laboratory-based performance. This session will present novel, validated measures of personal and clinical utility that can be used to generate evidence for policymakers, payors, and healthcare decision-makers committed to providing value-based genomic medicine. Aligned with measurement science best practices and using multi-step processes of item generation, item reduction, and validity testing across seven Canadian sites, the Patient-reported Genetic testing Utility InDEx (P-GUIDE) will be presented as a standardized approach for gathering patient preference data in five distinct clinical settings (i.e. prenatal care, outpatient genetics, precision oncology, pharmacogenomics, adolescent medicine). Complementary to P-GUIDE and using international expert-driven consensus panels and multi-site validity testing, the Clinician-reported Genetic testing Utility InDEx (C-GUIDE) will be presented as an additional standardized approach for gathering clinical utility data in four distinct clinical settings (i.e. prenatal care, critical care, outpatient genetics, genomic newborn screening). As a robust measurement system for quantifying the personal and clinical utility of genetic testing across the lifespan and across screening and diagnostic testing applications, these tools fill a critical evidentiary gap in genomic medicine.*
10:15- 10:45		Break
10:45- 12:15 1.5 hours, including 15 mins Q&A	**Workshop (CIHR)** (x2 rooms)	**"Nothing about us without us": How to build a strong partnership with patient organizations** Speakers: Dr. Gail Ouellette Dr. Genevieve Currie Svenja Espenhahn Kateryna Kratzer Dr. Thierry Lacaze-Masmonteil Breanne Stewart Description: *In Canada, our public medical system is based on the premise that medical care is provided in a family centered manner, a philosophy that prioritizes the family's role in caregiving. It's a partnership between the family and healthcare providers that focuses on the family's strengths and needs. This aligns perfectly with the focus genetics services have in evaluating patients with genetic disorders and their implications for families at large. Partnering in a more meaningful way with patient groups is a strategy that organizations like CCMG and CAGC would like to embrace. This workshop will tackle a variety of concrete approaches to help implement this goal.*
12:15- 13:45		**Lunch in Exhibit Hall (posters – repeat of Monday's posters)**
13:45- 15:00 75 mins, including 15 mins Q&A		**Abstract Oral Presentation Session** (4x15) • **Surveying Canadian prenatal genetics clinics: identifying work practices and innovations to address growing patient referrals** – speaker: Katherine Skora • **From knowledge to action for rare disease: Development of the K2A-RD knowledge mobilization strategy to advance equitable genome-wide sequencing for rare disease diagnosis in Canada** – speaker: Caitlin Slomp

		• **ThinkRare: Harnessing AI and electronic medical record data to shorten the diagnostic odyssey for rare disease patients in Canada** – speaker: Caitlin Chisholm • **From infants to elders: Understanding the impact of long QT syndrome in a Northern BC First Nation across the life course** – speaker: Alexa McAdam
15:00-15:30		Refreshment Break
15:30-16:45 75 mins, including 15 mins Q&A	**Breakout Sessions** (x3 rooms)	• **Inter-professional education in medical genetics** Speakers: Dr. Danielle Lynch Tracey Oh Description: *Inter-professional delivery of care in medical genetics uniquely centres around a team of genetic counsellors, laboratory geneticists, and medical geneticists. Developing competency in collaborative care is a goal of training in all three professions, and one which requires thoughtful graduated exposure. In this session, we propose to focus on modalities of inter-disciplinary education in medical genetics, introducing two initiatives at the University of British Columbia. Both are the result of trainee requests for more interaction with other types of trainees and professionals in medical genetics. First, we will discuss a workshop centred around a patient's digital story , wherein trainees recognize each other's roles and scope of practice and collaborate to make a plan for patient care. We will then describe how this is put in practice in a trainee-led multi-disciplinary clinic. We explore how patients can benefit from an inter-professional team in which there is strong cohesion, and how this can impact trainee development and well-being. Group discussion will focus on ways to implement inter-professional learning at medical genetics centres with different combinations of training programs.* • **Bridging Theory and Practice: Designing Effective Non-Clinical Rotations in Genetic Counselling** Speaker: Dr. Leichelle Little Description: *This interactive session will guide participants in developing non-clinical rotations that incorporate activities and learning outcomes aligned with the Canadian Association of Genetic Counsellors (CAGC) Practice-Based Competencies. Participants will examine how to apply foundational teaching principles, such as backward design, scaffolding, and active learning, to enhance the educational impact of their rotation designs. The session will also feature a case study of a successfully implemented non-clinical rotation, demonstrating how educational theory can be effectively translated into practice. By the end of the session, participants will have taken the first steps towards creating a non-clinical rotation plan. Participants will also leave with practical tools and increased confidence in their ability to support the training of future genetic counsellors in non-clinical settings.* • **Education Webinars as an Alternative Tool for Counselling Individuals with Hypermobile Ehlers-Danlos Syndrome** Speaker: Taryn Athey Description: *The hypermobile form of Ehlers-Danlos syndrome (hEDS) is the most common form of EDS. Individuals with hEDS have many health complications including chronic joint*

		pain, postural orthostatic tachycardia, mast cell activation syndrome, and gastrointestinal concerns. The underlying genetic cause for hEDS remains unknown, and the diagnosis of this condition is based on clinical features. Due to the high prevalence of the condition, genetics departments receive an overwhelming number of referrals for individuals with hEDS. Because of this, many genetics departments have decided to no longer accept referrals for individuals with hEDS. However, patients with hEDS report that their family doctors are not educated in their complex needs and that they are not taken seriously by the healthcare system. As well, many individuals with hEDS are concerned that they may have a more severe form of EDS that is not being properly managed. In order to combat this gap in care, the Edmonton Medical Genetics Department is now providing an hEDS education webinar for patients to learn about their condition and the management that may be available to them. In this session, we would like to discuss our experiences in Edmonton and the feedback we have received on our hEDS webinar.

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Wednesday October 8		
7:00-8:30 Talk will be held from 7:30-8:00am and is **30 mins**, including 5 Q&A		**CAGC Presidential Address and Awards** Speaker: Jessica Hartley Description: *Join the CAGC-ACCG President for a reflection on the past year, including key accomplishments aligned with the Strategic Plan, highlights of major initiatives and collaborations, and recognition of award recipients. This address will also look ahead, reinforcing our shared vision and encouraging continued engagement across the genetic counselling community.* *Note: the presidential address is 30 mins including 5 mins for Q&A. We are not seeking accreditation for the award presentation.*
8:30-10:15 1.75 hours, including 20 mins Q&A		**Abstract Oral Presentation Session** (6x15) • **POWER (Precision Oncology at Western University): Evaluating clinical impact of comprehensive genomic profiling of solid tumors in patient management** – speaker: Pratibha Bhai • **Understanding KAT6 Syndromes: A Combined Clinical and Experimental Approach to Phenotypic Characterization** – speaker: Zoe Goldstone-Joubert • **Clinical utility of optical genome mapping (OGM) to replace chromosomal microarray analysis for investigating constitutional chromosome abnormalities** – speaker: Zeynep Onkal • **Barriers and facilitators to clinical genome-wide sequencing implementation in Canada: A comparative case study.** – speaker: Whiwon Lee • **Understanding parents' experiences and perspectives on the personal utility of pharmacogenetic testing for their children.** – speaker: Kulisara Sirinawasathian • **Clinical Applications of and Molecular Insights from RNA-Sequencing in a Rare Disease Cohort** – speaker: Jamie Stark
10:15-10:45		Refreshment Break
10:45-11:45 1 hour, including 10 mins Q&A	**Plenary**	**Augmenting Human Ability in Clinical Genetics: From Innovation to Patient Care** Speakers: Melanie Napier Dr. Maria J. Guillen Sacoto, Dr. Bekim Sadikovic, Justin Lorentz, Maelenn Corfmat Description: *Artificial intelligence (AI) has become ubiquitous in today's society and has permeated the field of clinical genetics and genomics. The development and adoption of machine learning in research and clinical diagnostic laboratories has become crucial to workflows and continues to require innovation to allow quality service, at scale, for patients and clinicians.* *Additionally, as the volume of patients referred to clinical genetic services continues to grow along with waitlists, the use of large language models (LLMs) and other AI tools is being explored and implemented as part of alternative service delivery*

		models. While we adopt AI applications into clinical care, we must consider ethical, legal and regulatory aspects of their integration. This session will highlight how AI can be leveraged in research, clinical diagnostic, and patient care settings to augment our human ability with the goal to ultimately benefit our patients and our profession, while at the same time examining how we can achieve this responsibly.
11:45-12:45 1 hour, including 10 mins Q&A	**Plenary**	**Advancing Equity in Precision Genomics: Updates from the Indigenous Background Variant Library and Pan Canadian Genomic Library** Speakers: Dr. Anna Lehman Dr. Laura Arbour Dr. Wyeth Wasserman Claude Bhérer Dr. Dean Regier Description: *It is essential to include Canada's diverse ancestries in genomic databases in order to achieve accurate variant interpretation and precision health care. This session will provide updates on the Indigenous Background Variant Library (IBVL) and the Pan- Canadian Genome Library (PCGL), two initiatives designed to improve genetic diagnosis by incorporating underrepresented variants. This session will also review current standards for applying allele frequency information toward variant classification. We will discuss the rationale for their creation, their impact on reducing health disparities, and the ethical considerations in their development and use, particularly as related to Indigenous sovereignty.*