2023 • EDMONTON • ALBERTA

CAGC·ACCG

ANNUAL EDUCATION CONFERENCE
CONGRES DE FORMATION ANNUEL

CAGC AEC Detailed Program

This Conference has been approved by the Canadian Association of Genetic Counsellors (CAGC) for 21.5 CECs and the National Society of Genetic Counselors (NSGC) has authorized Canadian Association of Genetic Counsellors (CAGC) to offer up to 2 CEUs or 20 Category 1 contact hours for the activity Canadian Association of Genetic Counsellors (CAGC) - AECC. The American Board of Genetic Counseling (ABGC) will accept CEUs earned at this program for the purposes of genetic counselor certification and recertification. Note this is subject to change with program changes.

SHORT COURSE
Unlocking the Genome: Tools to Advance Our Understanding of Human Health
WEDNESDAY, SEPTEMBER 27, 2023

7:00am-4:00pm **West Foyer**	**Conference Registration**
8:00am-9:30am **BC/Alberta/Yukon Ballroom**	**Elective Whole Genome Sequencing in an Ostensibly Healthy Canadian Population** *Moderator: Alison Elliott* *Speakers: Allison Hazell & Jessica Gu* Our clinic had a goal to provide Canadians with an option for high-quality, comprehensive clinically responsible whole genome sequencing designed for proactive screening purposes. Non-negotiable requirements were 1) reporting should only include Likely Pathogenic and Pathogenic variants and 2) all biological samples and data were to remain in Canada. With no commercially available laboratory options meeting these criteria, we set out to build our own. In this presentation we will share our experience collaborating with a hospital-based clinical genomics lab to build a proactive whole genome sequencing test from the ground up. We'll review the decisions we grappled with, from what to include or not include on the reports, to the development of our clinical policies, the business considerations and how we defined the client journey. We'll describe the genetic counselling process in this population, what we have learned and how we anticipate this program changing over time. Last, we will review our clinical data from the first 18-months of the program and highlight interesting cases.
9:30am-10:00am **West Foyer**	**Refreshment Break**
10:00am-11:00am **BC/Alberta/Yukon Ballroom**	**Exome / Genome Analysis: Lessons Learned from TIGeR** *Moderator: Taryn Athey* *Speakers: Matthea Sanderson, Alison Eaton, Jillian Parboosingh* TIGeR (Translational Implementation of Genomics for Rare diseases) is an Alberta-based initiative facilitating in-province clinical genome-wide sequencing (GWS). We will review our experiences to date, and through case examples we will review our approach to variant classification/prioritization, discuss limitations of exome/genome analysis and highlight the importance of phenotype in interpretation of GWS data.

11:00am-12:00pm **BC/Alberta/Yukon Ballroom**	**Genome-Wide DNA Methylation: A Functional Tool for Classifying Variants of Unknown Significance** *Moderator: Taryn Athey* *Speaker: Cheryl Cytrynbaum* The rapid advancement of genomic sequencing technologies has revolutionized the field of genetics and enhanced our ability to identify the genetic etiology of many human disorders. However, this has also led to an increase in the volume of clinically uninterpretable variants classified as variants of unknown significance (VUS). Deciphering the clinical relevance of VUS is crucial for diagnosis, accurate genetic counselling and treatment decisions. In recent years, DNA methylation, an epigenetic modification that plays a critical role in gene regulation, has emerged as a valuable resource for assessing the clinical relevance of VUS in genes that play a role in epigenetic regulation. This talk will discuss the principles and methodologies involved in utilizing DNA methylation signatures for variant classification, highlighting the utility of machine learning algorithms and statistical models for VUS classification. We will discuss data acquisition, quality control, and the significance of tissue-specific DNA methylation patterns in accurately classifying VUS and present examples of the clinical impact of successful application of utilizing DNA methylation signatures as a function tool to resolve VUS.
12:00pm-1:30pm **On own**	**Break for lunch** (lunch not provided)
1:30pm-3:30pm **BC/Alberta/Yukon Ballroom**	**How to Use ClinGen Resources to Inform Genetic Testing Results** *Moderator: Lindsay Burnell* *Speakers: Danielle Azzariti & Erin Riggs* *Panelists: Danielle Azzariti, Erin Riggs, Melanie Care, Frédéric Coulombe* Since 2013, the National Institutes of Health-funded Clinical Genome Resource (ClinGen) has been building and supporting publicly available resources to support the clinical annotation and interpretation of genes and variants. ClinGen provides extensively curated and expertly adjudicated knowledge through a process of 1) developing and implementing standards to support clinical classification of genes and variants; 2) sharing genomic and phenotypic data between clinicians, researchers, and patients through enhanced knowledge bases for clinical and research use; 3) enhancing and accelerating expert review of the clinical relevance of genes and variants; and 4) disseminating and integrating ClinGen knowledge and resources to the broader community. In this session, attendees will learn how ClinGen does this work, how to access ClinGen's curated information and use it in their practices, and how to get involved in ClinGen's data sharing efforts.
3:30pm-4:00pm **West Foyer**	**Refreshment Break**
4:00pm-5:00pm **BC/Alberta/Yukon Ballroom**	**The Translational Genomics Hub (TGH): A New Precision Medicine Tool in Edmonton** *Moderator: Dorothy Michalski* *Speaker: Oana Caluaeriu* This session will provide an overview of the Translational Genomics HUB (TGH or Hub), a research platform designed to help families affected by undiagnosed disease where a clear genetic cause could not be found through initial genetic testing and where there is a need for further experimental work to find a causative variant.

MAIN CONFERENCE
THURSDAY, SEPTEMBER 28, 2023

7:45am-8:30am **BC/Alberta/Yukon Ballroom**	**Working Towards Equitable Access to WES/WGS for Canadians** *Speaker: Jane Juusola* Breakfast generously provided by GeneDx. The uptake and clinical utilization of WES and WGS is changing rapidly. Dr. Juusola will review emerging evidence and recommendations for using WES / WGS as a first-tier test for indications such as epilepsy, autism, intellectual disability, cerebral palsy and hypotonia. We will highlight how clinical genomic sequencing can accelerate academic research through collaborations and partnerships, which are necessary for the advancement of our knowledge of genetic diseases and the improvement of patient care.
8:00am-4:00pm **West Foyer**	**Conference Registration**
8:00am-9:00am **West Foyer**	**Coffee & Tea Available**
8:45am-9:00am ***BC/Alberta/Yukon Ballroom***	**Welcoming Remarks** *Speaker: Julia Heaton*
9:00am-10:00am ***BC/Alberta/Yukon Ballroom***	**KEYNOTE - Genomics, Power, and Governance in Health: Engaging Through Critical Indigenous Political Theory** *Moderator: Susan Creighton* *Speaker: Jessica Kolopenuk* Biotechnological advances saturate society with promises of improved human health, environmental sustainability, and national security just as science, politics, economy, and law are fashioned by and to colonial power relations. Taking as a centre point that indigeneity, in empirical and heuristic forms, is a site of relationally produced scientific and political knowledges, this presentation will introduce viewers to key fields genomically re/iterating indigeneity in Canada and how these re/iterations are relationally produced with, through, and as the field of colonial power. In particular, the presentation will make a case for stronger interdisciplinary methodologies in genomic research with, about, and affecting Indigenous peoples and demonstrate the strength and future of Indigenous-led genomics.
10:00am-10:30am **Manitoba/Saskatchewan Ballroom**	**Refreshment Break**
10:10am-10:30am **Turner Valley**	**Mentor Session** *Mentors: Kelly Turner (General adult, peds, connective tissue - Nova Scotia), Deepti Babu (Founder, self-employed: Integrity Content Consulting - Alberta), Tracey Oh (Program director UBC, general genetics previously - BC)*

10:30am-12:00pm
BC/Alberta/Yukon Ballroom

GenCOUNSEL: Improving Access to Genetic Counselling in Canada
Moderator: Catherine Kentros
Speakers: Alison Elliott, Nicola Kopac, Kennedy Borle, Ma'n Zawati, Courtney Cook & Shelin Adam

In this plenary session, we will discuss five sub activities of the GenCOUNSEL Study:

1) GenCOUNSEL: The Big Picture

Since its inception in July 2018, the GenCOUNSEL study has been conducting research to examine the clinical, ethical, and economic aspects of genetic counselling provision in Canada with the aim of improving access for all Canadians. We will be presenting an overview of the GenCOUNSEL study, describe the four main research activities and their corresponding sub activities and progress to date.

2) Understanding workforce dynamics for genetic counsellors in Canada - Workforce need and supply models

The need for clinical genetic services in Canada has rapidly increased over the last decade, however the clinical workforce to support the delivery of genetic counselling and testing has not been able to keep up. We designed Canada-specific need and workforce supply models to estimate the current and future gaps between the patients who need services and the number of genetic counsellors available to provide care. We will discuss the results from our need and supply models, the challenges in workforce expansion for genetic counsellors, and opportunities for the future to reduce the gap between need and the capacity of the workforce.

3) Understanding workforce dynamics for genetic counsellors in Canada - Genetic counsellor career preferences

The size of the genetic counselling workforce has been steadily increasing over the last 20 years, however there are still gaps between the need for genetic counsellors and the available supply. One of the factors impacting this gap is the large proportion of genetic counsellors working in non-direct patient care positions. We conducted a discrete choice experiment to better understand which factors are most important for genetic counsellors in North America in determining how they would make choices about job opportunities. We will present the findings from this study and describe how these different factors could be useful for recruiting and retaining genetic counsellors in direct patient care roles in Canada

4) Using e-tools to support families through genomic testing when there is no genetic counsellor

In many specialty clinics, there are no genetic counsellors to inform and support families though decision-making prior to genomic testing and when results are received. In order to address this issue, we have developed two multi-language, patient-friendly tools. DECIDE is an interactive online tool that provides information and decision-support to people considering genomic sequencing. The Genomic Results Booklet (GRB) is a parent co-designed, customizable tool describing each family's specific results, with relevant resources. Using a mixed methods approach, we explored the benefits of clinical implementation of these tools. We will present how these e-tools were found to promote

equity in access by reducing geographic and linguistic barriers and empowering families to understand and navigate their genomic journey.

5) Evaluating family-centered care: parents'/caregivers', adolescent patients', and health care providers' perspectives

Family-centered care is a method of service delivery that emphasizes collaborative caregiving in which families are recognized as the experts on the child's abilities and needs and therefore work with healthcare providers to make informed decisions about services received. Using a mixed methods approach we explored the experiences of families seen in three specialty clinics at BC Children's Hospital that do not currently have a genetic counsellor as part of the care team. By administering a variety of validated tools (MPOC-20, GYV-20, MPOC-SP, Peds-QL), we evaluated the perspectives of parents, adolescent patients, and care providers, respectively. We conducted semi-structured interviews to gain a deeper understanding of parents' perspectives. We will discuss the results from our analyses exploring associations between MPOC score, PedsQL scores and potential demographic variable predictors and present on our model of the strengths and challenges parents experience with FCC over time and across service sectors.

6) The Future Legal Recognition of Genetic Counselling in Canada: Legal and Stakeholder Perspectives

Imagine receiving treatment from someone that has not received proper training? It's hard to imagine given the strict rules we have on medical acts. Now, imagine receiving counselling from someone that has no training in genetic counselling. That is also hard to imagine, but legally possible. This is because genetic counselling is not a legally recognized profession in the majority of Canadian provinces and territories. In 2019, the province of Manitoba, however, legally recognized genetic counselling through a limited form of delegation from physicians to genetic counsellors. It remains the only Canadian jurisdiction to do so. Legal recognition serves to regulate professions which, if not properly regulated, may expose the public to risk of harm. This potential for risk, in conjunction with the increasing demand for genetic counselling services, warrant the implementation of regulatory mechanisms to ensure the safer provision of genetic counselling. In this plenary, we will update the community on the stakeholder meetings undertaken by GenCOUNSEL to study this issue. We will present the outcomes of these deliberations and summarize their key outcomes. These perspectives will help inform future legal recognition initiatives in Canada.

12:00pm-2:00pm **On Own**	**Exhibit Hall & Posters Open** **Break for lunch** (lunch not provided)
12:15pm-1:00pm **BC/Alberta/Yukon Ballroom**	**Moving Towards Provincial Equity of Hereditary Testing for Canadian Breast Cancer Patients** *Speaker: Karen Panabaker* Lunch generously provided by Merck / AstraZeneca.

2:00pm-3:30pm
BC/Alberta/Yukon Ballroom

Abstract Session
Moderator: Julia Tagoe

O6 - The Creation of an Online Training Module for the Genetic Counseling Skills Checklist (GCSC) - *Heather Zierhut*
Communication is an integral facet of the genetic counseling process, yet the skills used in practice are not well described. The Genetic Counseling Skills Checklist (GCSC) is a novel process measure that was created in order to provide a standardized and reliable method for capturing and categorizing multiple communication skills observed in genetic counseling for research purposes. The checklist has 8 categories: (1) Building Rapport, (2) Mutual Agenda Setting & Structuring, (3) Risk Communication, (4) Recognizing & Responding to Emotions and Prior Experiences, (5) Educating, (6) Checking for Understanding, (7) Facilitating Decision Making, and (8) Promoting Patient Activation. Each category has 5 to 8 different observable skills. For the GCSC to be used in research, a standardized training process was needed to help ensure adequate inter-rater reliability among coders.

Objective: This project aimed to create an online training module for reliable use of the GCSC in individuals with no prior exposure to genetic counseling.

Methods: Detailed descriptions for each category and associated skill items within the categories were created through a series of discussions with a team of experienced coders and researchers. After skill descriptions were finalized, illustrative videos of the skill used and multiple-choice questions were created to train coders to recognize the skills and understand inclusion criteria for checking each skill item.

Results: The final training module was completed in the Canvas Learning Management System. It contains a total of 56 skill descriptions, 56 skill checks, 8 category quizzes to check for skill identification, 3 practice quizzes to familiarize novice users with coding, and 9 reliability tests. To demonstrate accurate and reliable use of the GCSC, pass the training module, and receive a certificate of completion, coders must complete all modules and achieve at least 80% accuracy on 2 reliability tests.

Conclusion: The GCSC training module provides a flexible and convenient self-paced training environment to teach coders how to use the checklist reliably. The GCSC training modules will now allow researchers to test its effectiveness, ease of use, and time needed to reach reliable use of the instrument in genetic counseling sessions.""

O7 - Modeling Cellular Evidence: Approaches for Generating, Validating and Incorporating Data from Functional Assays to Improve Clinical Variant Classification - *Britt Johnson*
Objectives: Variants of uncertain significance (VUS) in clinical genetic testing are a significant source of frustration for clinicians, patients, and laboratories because the uncertainty hinders diagnoses and clinical management. Evidence of a variant's effect on protein function has the potential to reduce VUS during clinical variant interpretation (VI). In recent years, cellular based functional experiments, called Multiplex Assays of Variant Effects (MAVEs), have been developed to generate variant level functional data for a variety of genes. We investigated the impact that MAVE data have on VUS reclassification at a large commercial laboratory.

Methods: Using an internal machine learning platform, we assessed the performance of 49 MAVE datasets from 22 publications. We also generated 44 MAVE datasets internally

using a single cell RNAseq methodology to develop functional evidence for clinically relevant genes and variants. Performance of all MAVE datasets were assessed by calculating the area under a receiver-operating characteristic (AUROC) curve which measures the model's performance at distinguishing between benign and pathogenic variants. MAVE datasets with high performance (AUROC>0.8) were incorporated into clinical VI. The impact of incorporating this standardized functional information on VUS reduction was assessed.

Results: Of the 49 published MAVE datasets, seven datasets (one each for BRCA1, BRCA2, MSH2 and SCN5A, and three for TP53) met the performance threshold (AUROC ≥ 0.8), underscoring the need for quality control (QC) before integration into VI. Among the 7 datasets that passed QC, ~3,000 unique variants achieved sufficiently confident evidence (>80% NPV or >80% PPV) to be incorporated into VI, which resulted in 66 unique reclassified VUS in 172 patients. Of the 44 internally generated MAVE datasets, 19 met the predictive performance threshold for integration into VI as of Nov 2022. For these 19 datasets, we observed a mean reclassification rate of 4.6% of the targeted VUS, which resulted in 41 unique reclassified VUS in 303 patients. For each reclassified VUS, there were an additional 18 VUS that received evidence in our VI framework, moving them one step closer to a future reclassification.

Conclusion: We have introduced a framework to QC and integrate both publicly available and internally generated highly performing MAVE datasets. Incorporation of functional evidence from these large-scale data has shown to improve VI and reduce VUS.

O8 - Diagnostic Utility of Whole Genome Sequencing After Exome Reanalysis for Rare Disease Diagnosis: A Cohort Study - *Andrea Goodman*
Objective: Care4Rare-SOLVE is a Canada-wide research study, one aim of which is to identify the etiologies of rare diseases with the use of exome sequencing (ES) reanalysis, whole genome sequencing (WGS) and other 'omic approaches. We sought to determine the utility of WGS after exome reanalysis.

Methods: We recruited and consented participants and relevant family members to Care4Rare-SOLVE through the genetics clinic at the Children's Hospital of Eastern Ontario. All participants were suspected of having monogenic disease, had non-diagnostic clinical testing performed on ES backbones, and had non-diagnostic research ES reanalysis by the Care4Rare team. Participant DNA samples were sequenced by short-read WGS at The Centre for Applied Genomics. The sequencing data was then run through Care4Rare's WGS bioinformatics pipeline to identify and annotate coding variants, non-coding variants in genes associated with the participant's phenotype (collected as Human Phenotype Ontology terms), copy number variants, repeat expansions, and other structural variants. The output files of each family were then analyzed in collaboration with the referring clinicians. Results were categorized based on whether they were variants of interest, whether the variants required WGS analyses (i.e., whether they were intractable to ES) and if these variants were in known or novel disease genes.

Results: Our cohort consisted of 91 participants (57% biologically male) and their family members. Overall, we identified compelling variants in 31 of 91 families (34% of cohort). We identified intronic variants and copy number variants that are intractable to ES in 12 families (13% of cohort). Most of these findings (11/12) were within known disease genes and we have upgraded 5 from compelling candidates to likely diagnoses after collecting additional evidence. In 19 families (21% of cohort) we identified compelling coding variants that could have been found by ES reanalysis. In most cases, the variant was present and

flagged by analysts at the time of the original ES reanalysis, but there was insufficient evidence available to consider it a compelling candidate.

Conclusion: WGS after ES reanalysis yielded a compelling candidate for 1/3 of our cohort, but just over 1/3 of those compelling candidates needed WGS to be found. This study shows the added yield of WGS is modest but can help determine the etiology of particular genetic diseases.

O9 - Exploring Canadian Genetic Counsellors' Experiences and Perspectives with Discussing Medical Assistance in Dying (MAiD) - *Rebecca Candlish*
Objective: Canada's 2021 changes to eligibility criteria for Medical Assistance in Dying (MAiD) have resulted in Canada having the world's most permissive assisted dying regime. The aims of this study were to describe Canadian genetic counsellors' experiences, knowledge, and preparedness to discuss MAiD with their patients and to provide suggested learning objectives to assist genetic counsellors in their preparation to discuss assisted death.

Methods: Survey responses were collected from Canadian genetic counsellors (n=44) through advertisement to the Canadian Association of Genetic Counsellors (CAGC) distribution list (n=386). Semi-structured interviews were conducted with 13 genetic counsellors, including those who had and had not discussed MAiD with their patients. Survey data were analyzed using descriptive statistics, and interview transcripts were analyzed by two independent coders using phronetic iterative analysis and an interpretive description approach.

Results: Survey data revealed that genetic counsellors have discussed MAiD with patients referred for cancer, neurologic, metabolic, connective tissue, and cardiac indications (n=18, 40.6%). While most participants thought that it was important for genetic counsellors to be knowledgeable of (n=42, 95.5%) and prepared to discuss MAiD (n=43, 97.7%), many were not familiar with the eligibility criteria (n=26, 59.1%), the process of accessing MAID (n=28, 63.6%), or the changes to MAiD eligibility criteria in 2021 (n=30; 68.2%). Interview participants described discussions about MAiD that were either initiated by themselves or their patients. Analysis of interview transcripts demonstrated that most participants felt prepared to discuss patients' thoughts about MAID but did not feel confident or well prepared to share detailed information about MAiD with their patients. Participants felt discussions about MAiD were likely to increase. They also expressed discomfort with discussing MAiD with patients who planned to access it prior to their natural death being foreseeable. Genetic counsellors were interested in guidelines, education sessions, and resources that could assist them in preparing to discuss MAiD with patients. They suggested topics that could be covered in MAiD education, which led to the development of our learning objectives.

Conclusions: As genetic counsellors continue engaging in discussions about MAiD, it is critical that these sensitive conversations are approached with increased knowledge and awareness of MAiD laws, the ethical issues surrounding MAiD in Canada, and relevant patient resources. Learning objectives can assist in genetic counsellors' clinical work and self-directed research and will aid in the development of professional guidelines, educational materials, and the integration of MAiD education into genetic counselling training program curricula.

O20 - An Approach to a Holistic Admissions Process at the University of Manitoba Genetic Counselling Program - *Nicole Yang*

Introduction: Since the inception of the Genetic Counselling Program at the University of Manitoba (U of M) in 2017, a formal admissions committee was charged with the evaluation of candidates on their capacity for successful matriculation into the U of M program and the genetic counselling profession. Having identified possible biases within previous cycles, the committee advocated for a more holistic review. As defined by the Association of American Medical Colleges, holistic review is an individualized and balanced assessment of a candidate's experiences, academic ability, and other attributes. Holistic review is important as the program continues to align to its values of diversity, equity, and inclusion to train genetic counsellors who are as diverse as our patient population.

Methods: Over the period of 2020-2022, the program evaluated aspects of the admissions process in an effort to promote a holistic review of applications in order reduce possible bias. This included:

1) Applications review. The purpose and importance of each item within the application was examined, considering the possibility of unfair expectations/biases. Additionally, we introduced a "horizontal review" such that reviewers assess certain aspects of the application and are blinded to other items, which may have biased scoring in the past.
2)Interviews. Two interview formats (multiple mini-interviews and leadership interviews) are used to assess candidates. We evaluated the strengths and limitations of each, and looked for redundancies, appropriate repetition, and ways we could welcome greater diversity and inclusion into this process.
3)General Accessibility. Advice was sought from the Genetic Counsellors with Disabilities group to implement strategies to make the admissions process more inclusive and accessible.

Results: This self-study suggested several potential biases which may have influenced individuals involved in admissions in the past. To illustrate, most individuals conducting MMIs station were genetic counsellors associated with the program, which may have resulted in a narrow and biased view. As such, in the most recent cycle, interviewers with personal or professional knowledge of / experience with genetic counselling were included to gather a more diverse perspective of each interviewee.

Conclusion: Our goal is to align with our values of diversity, equity, and inclusion to allow for the genetic counselling students at the U of M to represent the population served. Upon review of our admissions process, we identified biases within our process and acknowledge the need to create a more equitable and holistic review of applicants. While numerous positive changes were made, we recognize the importance of continual examination from multiple stakeholders to maintain our ascribed values.

References: Amiri, L. et al., (2023) Holistic considerations for the admission cycle. Association of American Medical Colleges, 1.

O23 - A Genomic Results E-Booklet Helps Support Parents after Genomic Sequencing - *Patricia Gombas*
Objectives: Families undergoing genomic sequencing (GS) told us they felt "abandoned and lost" after receiving their results. In response, with parent-partners, we co-developed and tested the Genomics Results e-Booklet (GRB) in a research setting. The GRB

provides individual genomic results, implications, and customized resources, in family-friendly language. There are two forms of the GRB: one for pathogenic findings, and the other for null results. The aim of the current study is to test both versions of GRB in a clinical setting.

Method: Families undergoing GS in the Pediatric Neurology Clinic at BC Children's Hospital, where there is no genetic counsellor, received an emailed GRB 8-14 weeks after receipt of results from their neurologist. To assess the utility of GRB, we conducted semi-structured, recorded, phone interviews. Two authors coded the transcriptions using interpretive description to derive primary and secondary codes and develop context, themes, and subthemes from the data.

Results: We interviewed 13 parents, 12 mothers, and one father, most of which were in their 30s. Five families received a GRB with a specific diagnosis and seven families received a GRB with a null result. Interviews ranged from 10-29 minutes. Parents expressed many challenges and emotional burdens after result receipt and this context impacted the four themes that emerged: 1. the value of information; 2. the importance of a written tool; 3. the desire for situation-appropriate resources and supports; 4. the need for a clear path forward.

Conclusion: All parents shared their challenges, emotional burdens, and needs associated with caring for a child with a complex condition. Most parents described how the GRB helped them better understand the genetic information, gave them a sense of direction, and provided support after GS. Additionally, most parents shared that the GRB helped them take some action, such as accessing financial resources or facilitating the sharing of their child's genetic results with others. Our study demonstrates that the GRB was able to address some of the needs of families undergoing GS, and thus reduce some of the challenges and emotional burdens that these parents face. We found that the GRB has value as a clinical resource and support tool for families and may help fill the void many families feel after GS. This may be especially helpful in stretching limited genetic counselling resources, or in clinical settings where there is no genetic counsellor.

3:30pm-4:00pm
Manitoba/Saskatchewan Ballroom

Refreshment Break

4:00pm-5:15pm
BC/Alberta/Yukon Ballroom

Concurrent Sessions:

Thinking Outside the Clinic: A Novel Model of Genetics Service Delivery Provides Efficient Care in a Historically Underserved Canadian Province
Moderator: Zoe Lohn
Speaker: Katherine Hodson

This presentation will highlight the experience running a publicly funded Genetics service in the province of New Brunswick. New Brunswick, an Atlantic province with a population of 775 000 people is historically underserved in terms of Genetics care. Most patients were traditionally referred out of province to Quebec or Nova Scotia and were faced with logistical issues and long wait times. In 2018, the first-ever Medical Genetics clinic in New Brunswick was established at the Georges L. Dumont Hospital in Moncton. The clinic employs an innovative solution to address the lack of genetic counselling professionals in the province. Genetic counsellors employed by private organizations in the rest of Canada provide provincially funded genetic counselling by telemedicine. By utilizing public-private partnerships, the clinic provides genetic counselling to approximately 450 patients per year and has demonstrably shorter wait times than neighboring provinces. This presentation will outline the clinic model of service delivery and the possibility of broader application, while highlighting the increased job satisfaction for genetic counsellors providing direct patient care while employed in an industry setting.

4:00pm-5:15pm
Turner Valley

Can Technology Build the Bridge? Using EMR Tools to Increase Access to Genetic Services
Moderator: Simina McDermott
Speakers: Grace Ediae & Alexandre White-Brown

The widespread adoption of electronic medical records (EMRs) and development of health information technologies has provided an unprecedented opportunity to support genetic services across a patient's diagnostic process. In this platform presentation, we will summarize the landscape of technologies that can be applied to genetic services and primarily focus on the first step of the diagnostic process - the referral. There are several ways that health information technology has been utilized to increase access to genetic services; and with that comes challenges and considerations. As a collaborative, multidisciplinary research team, we developed an algorithm known as 'ThinkRare' that uses structured EMR data and clinical criteria to help identify patients who may have a rare genetic disease but have not yet been referred for genetic evaluation. Building on the interest generated from last year's 'ThinkRare' abstract presentation, we hope to continue the conversation. Specifically, our algorithm has demonstrated several benefits and limitations regarding the effectiveness of technology in identifying patients eligible for genetic services. Here, we present what we have learned through our journey of developing the 'ThinkRare' algorithm, and how we can move towards the responsible harnessing of this technology to improve access to genetic services.

4:00pm-5:15pm
Leduc

Public Health as a Framework to Improve the Diagnosis and Genetic Screening for the Long-Term Health of Individuals Diagnosed with Familial Hypercholesterolemia: Opportunities & Barriers for GCs
Moderator: Lindsay Burnell
Speakers: Heather Zierhut, Michael Khoury, Cathy Huculak, Susan Christian

The early diagnosis and treatment of hereditary cardiovascular diseases such as familial hypercholesterolemia (FH) is an illustrative example of how public health and genetics services can be integrated to significantly reduce the health and economic burden of disease for patients. The goal of public health is to prevent disease, prolong life and promote health through an organized societal effort. FH is a common, inherited, life-threatening, and treatable disease that if untreated, individuals have an 18-fold increased risk of cardiovascular disease (Hu et al. 2020). Universal lipid screening in childhood is

a powerful public health tool to identify individuals affected and start treatment in childhood to normalize the risk of cardiovascular disease (Luirink et al. 2019). However, research has shown that less than 10% of individuals with FH are diagnosed and treated. Combination approaches including electronic medical record screening, genetic counseling, cascade screening paired with public health approaches are ways to overcome the significant underdiagnosis. The aim of this education session is to explore the state of FH screening in Canada, to identify opportunities and challenges to applying the public health framework and suggest genetic counseling strategies for overcoming the barriers to screening and increasing uptake of cascade testing.

5:15pm-6:00pm **Free Time**

6:00pm-8:00pm
Manitoba/Saskatchewan Ballroom

Welcome Reception & Poster Session

FRIDAY, SEPTEMBER 29, 2023

7:30am - 8:15am **BC/Alberta/Yukon Ballroom**	**Next Dimension in Rare Disease Diagnostics with Transcriptomics** *Speaker: Peter Bauer* Breakfast generously provided by Centogene. Centogene is on a mission to provide data drive, life changing answers to patients, physicians, and pharma companies. We have the world's largest growing real world integrated data repository in Multiomics for rare and neurodegenerative diseases and a state of the art genomic and multiomic reference laboratory to support testing. While genomic analysis is the typical start point in diagnosis, integrating other important molecular data may yield faster and more conclusive results, and can lead to more accurate prognosis and therapy for a given patient. The Multiomics approach, when used first line, supports earlier diagnosis by integrating phenomics, genomics, transcriptomics, proteomics and metabolomics data at once for a patient in a single test result.
8:00am-4:00pm **West Foyer**	**Conference Registration**
8:00am-8:30am **West Foyer**	**Coffee & Tea Available**
8:20am-8:30am **BC/Alberta/Yukon Ballroom**	**Opening Remarks** *Speaker: Julia Heaton*
8:30am-9:15am **BC/Alberta/Yukon Ballroom**	**Presidential Address** *Moderator: Dorothy Michalski* *Speaker: Danna Hull* This address will provide an overview of current and ongoing initiatives of the CAGC through the lens of the board of directors. Topics include an update on CAGC's progress on diversity, equity, inclusion and justice (DEIJ) including the DEIJ Task Force's recommendations, leveraging partnerships with related organizations, and recognizing the value of our CAGC volunteers.

9:15am-10:15am
BC/Alberta/Yukon Ballroom

Abstract Session
Moderator: Dorothy Michalski

O11 - Refining the Activities of Genetic Assistants: Development of Task Statements Across Practice Settings - *Jessica Hartley*

Objective: As the current genetic counsellor workforce is not large enough to meet the growing demand for services, many institutions have implemented genetic assistant (GA) positions. Through role substitution, GAs assume tasks that are typically assigned to genetic counsellors but do not require their specialized skillset. While the literature suggests that GAs improve productivity, studies have also demonstrated that the activities performed by GAs are variable, which may cause role overlap and ultimately limit the impact of their work. Therefore, this study aimed to define the tasks that GAs perform across work settings and specialties.

Methods: Tasks performed by GAs were extracted from peer-reviewed articles, publicly available theses, and job postings, then analyzed using deductive content analysis. Briefly, task statements were coded using broad categories defined by original research, with new categories added as emergent. Coded tasks were condensed and combined to produce a final task list, which was reviewed by subject matter experts.

Results: Sixty-one task statements were extracted from original research and 334 task statements were extracted from job descriptions. Directed content analysis produced a list of 40 unique tasks under ten categories (eight from original research and two from the data).

Conclusion: This study design resulted in a well-rounded, reliable list of GA tasks, applicable across work settings and specialties, which is an essential step towards defining the scope of GA work. Beyond the human resource applications of a standardized job description, this work may also benefit genetics services by reducing role overlap, improving efficiencies, improving job satisfaction, and streamlining training.

O12 - Genome Sequencing Performed in Ostensibly Healthy Adults from Ontario Identifies Secondary Findings related to Personal Disease Risks - *Selina Casalino*

Background & Aim: Genome sequencing (GS) may be used in the context of clinical diagnosis and preventative screening. GS may reveal secondary findings (SF) related to inherited predisposition for diseases or a personal or family history that was previously undiagnosed. Advancements in GS and the growing field of genomic medicine warrants exploration into how these technologies may be used in the population at scale. We aim to evaluate the yield of clinically significant SF from GS in a representative population of adults from Ontario.

Methods: Ostensibly healthy adults (N=1,131) unselected for disease status with a prior diagnosis of COVID-19 were recruited for GS and consented to return of SF through the GENCOV study. Demographics and medical history were collected by intake survey at the time of recruitment. Pre-test and results counselling were provided. Clinical confirmation and follow-up (including complete medical and family history) were initiated for clinically significant findings impacting personal disease risk. This excludes common, higher frequency conditions (e.g. hemochromatosis, thrombophilia) for which a letter was given with recommendations for follow-up.

Results: Based on 967/1,106 (87%) survey responses, participants were mostly female (56%), White/European (51%), 25 to 44 years of age (42%), with a Bachelor’s degree or

higher (66%). To date, 638 genomes have been analyzed and GS results were returned to 447 participants. Of those who received results, 64 (14%) had clinically significant SF impacting personal risk for disease requiring clinical follow-up and 17 (4%) had findings consistent with a reported medical or family history. Several participants are pending clinical assessment, which may reveal further history consistent with SF.

Conclusion: Our findings illustrate that population GS screening may identify SF related to hereditary disease risk in adults, which in some cases was consistent with a previously unexplained medical or family history. These findings have a significant impact on management, treatment, and risk counselling for patients and their families, and demonstrate the importance of involving Genetics specialists in adult patient care. GS findings may provide overdue answers for patients with undiagnosed symptoms by ending long diagnostic cascades that impose significant healthcare costs. Future work will aim to evaluate the diagnostic yield of GS in individuals from our cohort selected for specific disease status.

O14 - “It's Just Another Tool in the Box”: Patient Attitudes Toward Supported Direct Contact to Inform Relatives About Genetic Testing - *Jennifer Nuk*
Introduction: Most hereditary cancer programs use a family-mediated approach to tell relatives about cascade carrier testing. To improve testing uptake, some have implemented a direct contact approach whereby the program contacts family members to share the option of genetic testing, typically in parallel to patients notifying their family. To ensure that the implementation of such an approach is acceptable to patients, an understanding of their preferences, concerns and expectations is required. The purpose of this study is to explore the attitudes of hereditary cancer patients from a Canadian provincial program on a supported direct contact approach.

Methods: Individuals carrying a pathogenic variant in a hereditary cancer gene were identified; purposive sampling guided the selection of participants to support diversity. Semi-structured interviews were conducted by three of the investigators via Zoom or telephone. The interview guide was reviewed by patient partners. Interviews were recorded, professionally transcribed, de-identified, and reviewed for accuracy. Coding and analysis was performed by two analysts using N-vivo qualitative coding software.

Results: Fifteen participants were interviewed; 11 females and 4 males between ages 35 and 70 years. In response to reported challenges communicating hereditary cancer risk information with relatives, many participants were supportive of enhanced assistance from hereditary cancer programs. There was variation in preferences for a supported direct contact approach centered around timing and level of genetic counsellor involvement, as well as the detail and format of information provided to family members. Perceived benefits included reducing the burden for those in cancer treatment and limited relationships with extended family, concerns included violations of privacy and difficulties with learning about cancer in the family this way. Three individuals had lived experience with direct contact.

Conclusion: Based on our program’s experience in supporting direct contact for a number of families, a draft clinical guideline was developed after legal and ethical review for use by all medical genetics services in our provincial health authority. A collaborative approach is desired, and a tailored approach to sharing needs to be part of the genetic counselling process. Results from this study will inform the final version of the guideline and lead to implementation of a patient values-informed supported-direct contact approach to facilitating cascade carrier testing in our province.

O17 - Exploring Non-Genetic Healthcare Providers' Experiences and Perspectives with Rapid Genome-Wide Sequencing in Canadian NICUs - *Lauren Piers*
Objective: Rapid genome-wide sequencing (rGWS) continues to transform the care provided to infants with genetic conditions in Neonatal Intensive Care Units (NICUs). Previous research has demonstrated that rGWS has immediate effects on patient management in NICUs, improving the health outcomes of critically ill neonates. However, little is known about non-genetic healthcare providers' (HCPs) experiences and perspectives of working with rGWS and supporting families through the rGWS testing process in Canadian NICUs. The objectives of this study were to explore the perspectives and experiences of non-genetic HCPs to 1) understand the barriers encountered when accessing and implementing rGWS in NICUs, and supporting families through the rGWS testing process; 2) inform the development of practice guidelines on the use of rGWS in Canadian NICUs to support non-genetic HCPs.

Methods: Non-genetic HCPs of diverse professions were recruited from two NICUs in British Columbia. Participation in this study consisted of two parts: completion of an online demographic survey followed by a one-on-one semi-structured interview. The survey was completed by 28 non-genetic HCPs with representation across different specialties. The survey data was analyzed using descriptive statistics. Ten participants completed a semi-structured interview. An interpretive description approach was used to analyze interview transcripts to identify patterns and variations in non-genetic HCPs experiences and perceptions of rGWS.

Results: Participants were found to have varying degrees of exposure to rGWS and levels of comfort with different elements of the rGWS testing process. Numerous barriers affecting the implementation of rGWS were identified, including low levels of comprehension of rGWS, longer turn-around times for results than expected, and having to apply for provincial government approval to access testing. Participants viewed a collaborative partnership with genetics as imperative for the successful implementation of rGWS in NICUs. Further, participants desired more education on rGWS, clear guidelines on the use of rGWS in NICUs, and resources for non-genetic HCPs and parents in the NICU to support implementation.

Conclusions: The results from this study can be used to inform the development of practice guidelines and educational resources on the use of rGWS in Canadian NICUs. A practice guideline and other educational resources can help ensure that all members of the NICU multidisciplinary team are supported to help optimize the implementation of rGWS in NICUs and improve the care provided to critically ill neonates and their families. Further, the results of this study provide support for the integration of genetic counsellors into NICU facilities to provide support to families undergoing rGWS and non-genetic healthcare providers implementing this technology in NICUs.

10:15am-10:45am
Manitoba/Saskatchewan Ballroom

Refreshment Break

10:25am-10:45am **Turner Valley**	**Mentor Session** Mentors: Kennedy Borle *(PhD Candidate, Fellowship with Ministry of Health - BC)*, Prescilla Carrion *(Research Manager, Medcan, psychiatric GC, primary care - BC)*, Jessica Gu *(Clinical Director, Medcan, personalized preventative medicine - Ontario)*, Danna Hull *(Prenatal Screening, CAGC President* - Ontario*)*, Andrea Rideout *(Research, cancer, aortopathy, connective tissue, cardiac 22q11DS and NBS/hemoglobinopathies - Nova Scotia)*
10:45am-11:45am **BC/Alberta/Yukon Ballroom**	**Unraveling Genetic Tapestry: Exploring Special Populations and Hidden Stories of Genetic Disorders in Alberta, Canada** *Moderator: Angela Inglis* *Speaker: Melissa MacPherson* In this session you will learn about special populations in Alberta and the historical, social, and genetic factors contributing to genetic disorders in these populations. We will also discuss commonly observed genetic disorders and the challenges associated with genetic counseling.
11:45am-2:00pm **Manitoba/Saskatchewan Ballroom**	**Networking in the Exhibit Hall**
11:45am-2:00pm **Provided by CAGC**	**Break for lunch** (lunch provided by CAGC)
1:30pm-1:50pm **Turner Valley**	**Mentor Session** *Mentors: Emily Fox (General GC - cancer, general, prenatal, preconception - Ontario), Alison Elliott (Associate Professor, Research GC Project Lead: GenCOUNSEL - BC),* Jessica Lawrence *(General - metabolics and ocular, Lab - Winnipeg)*
2:00pm-3:15pm **BC/Alberta/Yukon Ballroom**	**Concurrent Sessions:** **Prenatal WES: Diagnostic Yield, Syndromic Landscape, and Incidence and Uptake of Secondary Findings** *Moderator: Julia Heaton* *Speakers: Harriet Druker & Kayleigh Avello* Last year, the Canadian College of Medical Geneticists (CCMG) issued a series of recommendations for the use of prenatal genome-wide sequencing (GWS)1. The authors acknowledge a relative lack of literature surrounding the diagnostic yield and utility of GWS during the prenatal period; however, they've found that the available evidence supports the use of GWS for certain fetal indications. To contribute to the growing need for more data surrounding the potential benefits of GWS, we have examined the diagnostic yield and types of conditions previously uncovered by our prenatal whole exome sequencing (WES) test. Although not expressly recommended by the CCMG, the American College of Medical Genetics and Genomics (ACMG) added to their position statement that prenatal WES should include for opt-in their list of medically-actionable genes and genes that can cause a childhood-onset disorder2. These secondary findings categories were added to our prenatal WES tests in 2021, and we discuss the uptake, incidence of positive reporting, and types of conditions that have surfaced through that portion of the analysis.

2:00pm-3:15pm
Turner Valley

A Practical Guide to ChatGPT in the Genetic Counselling Profession
Moderator: Simina McDermott
Speaker: Justin Lorentz

On November 30, 2022, OpenAI released ChatGPT, a free access chatbot that provides detailed responses to a prompt. How does it work? How can we use it in our profession? Is there anything we should consider before using it?

I was thinking there would be two components to this talk, a didactic and an interactive (audience participation) component. In a didactic format I was planning to review the technical limitations of ChatGPT and how ChatGPT sources the information it has. I was also hoping to explore real-life case examples of how ChatGPT has been used in other professions as both aspirational stories and cautionary tales. I was also planning to review the literature on the use of chatbots in the medical and genetic counselling sphere (there are multiple publications in JGC for example on chatbots).

On the interactive side I was hoping to show how to setup a ChatGPT account and explain what personal information is needed to do so in real-time. I would like to review strategies on how to write clear prompts to elicit more comprehensive responses from ChatGPT and discuss the real-time responses to these prompts as a group.

2:00pm-3:15pm
Leduc

Pancreatic Cancer Screening in High-Risk Populations - Canadian Context & Clinical Conundrums
Moderator: Zoe Lohn
Speakers: Carol Cremin, Spring Holter, Adeline Cuggia

This session will discuss evolving evidence and recommendations in pancreatic cancer screening for high-risk individuals and how GCs are navigating the tricky waters between what is possible and what is clinically available.

All three speakers specialize in pancreatic cancer and are from the three Canadian sites (Quebec, Ontario, and BC) that are participating in the PRECEDE consortium (international consortium on pancreatic cancer screening and early detection (precede.org)).

3:15pm-3:45pm
Manitoba/Saskatchewan Ballroom

Refreshment Break

3:45pm-5:00pm
BC/Alberta/Yukon Ballroom

Concurrent Sessions:

Aortopathy & Connective Tissue Disorder Workshop
Moderator: Lacey Benoit
Speakers: Andrea Rideout & Kelly Turner

This session will be a review of aortopathy conditions & genes, upcoming clinical trials, models of care.

3:45pm-5:00pm
Turner Valley

A Practical Guide to ChatGPT in the Genetic Counselling Profession
Moderator: Jocelyn Carter-Sim
Speaker: Justin Lorentz

See above at 2:00pm.

3:45pm-5:00pm **Leduc**	**Crucial Considerations for Unilateral Retinoblastoma Patients** *Moderator: Catherine Kentros* *Speaker: Jamie Jessen* This session will review the commonly asked questions relating to the unilateral retinoblastoma patient population.
5:00pm-6:30pm	**Free Time**
6:30pm-10:00pm Campio Brewing Co. 10257 105 St. NW Edmonton	**CAGC Social Soirée** *Tickets required*

TRUTH & RECONCILIATION DAY

SATURDAY, SEPTEMBER 30, 2023

8:00am-10:00am **West Foyer**	**Conference Registration**
8:00am-8:30am **West Foyer**	**Coffee & Tea Available**
8:30am-9:00am **BC/Alberta/Yukon Ballroom**	**Opening Remarks** *Speaker: Julia Heaton*
9:00am-10:30am **BC/Alberta/Yukon Ballroom**	**Abstract Session** *Moderator: Angela Inglis* **O3** - Exploring Professional Liability Insurance for Genetic Counsellors in Canada - *Amy Davis* Objective: Professional liability insurance provides medical practitioners legal coverage when complaints are made arising from negligence, malpractice, mistakes or misrepresentation. We sought to understand the current status of liability coverage and explore personal liability insurance options for genetic counsellors practicing in Canada. Methods: In 2021, the Alberta Association of Genetic Counsellors created a Liability Insurance Working Group (LIWG) in collaboration with the Canadian Association of Genetic Counsellors. Informal interviews with genetic counsellors in Canada were conducted to understand the current landscape of liability coverage. Formal discussions with insurance brokers were used to better understand possible liability coverage options. Results: There was hesitancy on the part of genetic counsellors to approach institutions about liability coverage. Many genetic counsellors who work in hospital settings are believed to have liability coverage through their employer, however most do not know specific details about the coverage. Most genetic counsellors in Canada do not currently purchase additional liability insurance. Some genetic counsellors have been informed that personal liability insurance would be a requirement for provincial regulation. Genetic counsellors who did purchase insurance reported that annual premiums ranged from $1,700-3,000 for at least $1 million of coverage. Insurance brokers had an overall poor understanding of the genetic counselling profession. Questions included scope of practice, training and education requirements, settings where genetic counsellors work, local insurance requirements per province, type of insurance that is currently purchased,

and number of genetic counsellors in Canada. Conversations with the insurance broker about insurance policy options are ongoing.

Conclusion: Further discussions within the genetic counselling profession are needed to better understand the insurance coverage genetic counsellors have through employers, and what additional coverage would be desired or required in the event of provincial regulation. Determining how many genetic counsellors would be interested in purchasing liability insurance and minimum coverage requirements will help facilitate ongoing conversations between the LIWG and insurance brokers.

O16 - What Are Patients Saying About Virtual Genetic Counselling Care? The Sickkids' Experience - *Stacy Hewson*
Objective: Genetic counselling care transitioned rapidly to virtual during the COVID19 pandemic. While virtual care in other specialty areas has seen a decrease, virtual genetic counselling visits at SickKids, facilitated by Epic-Integrated Zoom, have remained high. To ensure that virtual genetic counselling meets the needs of patients and families, we reviewed data from the annual SickKids Patient & Family Virtual Care Experience Survey specific to genetic counselling completed in 2022 and 2023.

Methods: The Patient & Family Virtual Care Experience Survey comprises multiple choice/Likert scales and open-ended responses. Responses were anonymous, collecting high level demographic and user experience feedback in domains of satisfaction, effectiveness, timeliness, cost savings, and preferences. The survey was circulated by email, sent automatically via the electronic health record the day following an integrated video visit, for a period of approximately six consecutive weeks in 2022 and 2023. Respondents indicated if they were seen by a genetic counsellor.

Results: At SickKids, 1,610 virtual visits with a genetic counsellor were completed between January 2022 and March 2023. Nearly all (95%) completed visits were virtual with the majority (61%) completed by video and the remaining 39% by telephone. The percentage of virtual visits remained over 91% in 2022 and currently remains at 95%.

There were 26 participants who completed the survey after a virtual genetic counselling appointment in 2022 and 2023. All respondents reported being satisfied with their virtual visit and all reported that their virtual visit experience was the same or better when compared to an in-person visit. Wait times to start a virtual visit were less than 5 minutes for 83% of respondents. All participants reported that they received instructions before the virtual visit, that they felt high confidence in information privacy and that they understood the next steps at the end of the visit. From available data, families indicated an average savings of $78 leading to an estimated total cost savings of $1245 from not having to travel to the appointment (94%), not having to take time off from work (62%), and not having to arrange care (39%). Themes from open-ended comments highlight the convenience of virtual visits and the ability to have thorough, uninterrupted discussions.

Conclusion: The data presented here is based on a small sample of convenience. The study highlights high patient satisfaction with virtual genetic counselling, timely quality care and the savings of time and money, however future studies are needed for validation. To ensure equitable access to genetic counselling, these studies should also aim to assess benefits and potential barriers to virtual genetic counselling as virtual genetic counselling is likely to remain a mainstay of clinical practice.

O21 - Yield of Genetic Testing in Patients with a Family History of Pancreatic Cancer - *Carol Cremin*

Objective: Germline pathogenic variants (PVs) are found in up to 15% of individuals with pancreatic ductal adenocarcinoma (PDAC) and National Comprehensive Cancer Network Guidelines (NCCN) have recommended genetic testing in all patients since 2019.1,2 Current NCCN guidelines extend the testing recommendation to individuals with a first degree relative with PDAC, though the frequency and spectrum of PVs have not been well described in this group.1 Starting in 2022, BC Cancer's Hereditary Cancer Program (HCP) piloted offering testing to individuals with a first or second degree relative with PDAC. The aim of this pilot was to asses the pathogenic variant (PV) detection rate and impact of the expanded criteria.

Methods: Patients were individuals referred to the HCP who received pre-test information and signed consent for genetic testing. Family cancer history and personal medical history were obtained through a questionnaire on a web-based portal or by telephone call with a genetic counsellor. The panel was primarily a 76 gene hereditary cancer panel through Ambry Genetics. The HCP Progeny database was used to identify all pedigrees that contained both 1) an individual who had genetic testing in 2022 and 2) an individual who had pancreatic cancer. Pedigrees were reviewed for personal and cancer family history, reason for referral and genetic test results.

Results: The Progeny search identified 1249 potential cases. Cases were excluded due to: missing or incomplete data (n=439), personal history of pancreatic cancer (n=254), family history of pancreatic cancer not in a first or second degree (n=345). A total of 211 remaining cases had testing in 2022 and >1 first degree relative (n=102) or >1 second-degree relative (n=115) with pancreatic cancer. 58 of these patients had testing for a known familial PV and 153 patients had index testing. The overall PV rate in the index group was 12.4% (19/153) and PVs in PC-susceptibility genes accounted for the majority (n=14, BRCA2 (4), BRCA1 (2), ATM (4), PALB2 (2), MSH2 (1), CDKN2A (1). Patients with a personal history of a HBOC/Lynch or other syndrome related cancer had a significantly higher rate of PVs (16/103= 15.5%) compared to those without (3/50=6%, PALB2, MSH2 and CFTR). The rate of actionable pancreatic cancer susceptibility PVs in patients without a personal history of syndrome-related cancers but with a family history of pancreatic cancer was 4%.

Conclusions: We found a 12.4% overall rate of PVs among individuals who have a family history of PDAC. Testing unaffected patients due to a family history of PDAC identified an actionable pancreatic cancer susceptibility PV in 4%. As germline genetic testing guidelines contemplate testing in individuals with ≥5% likelihood of carrying a pathogenic/likely pathogenic variant, real world data suggest that testing of first- and second-degree relatives of patients with PDAC may meet this bar.

O29 - Psychiatric Genetic Counselling for Inpatients with Treatment Resistant Psychosis - *Prescilla Carrion*

Objective: To use validated measures to evaluate the outcomes of psychiatric genetic counselling (pGC) in an inpatient population of adults with treatment resistant psychosis, (including, but not limited to people with diagnoses of schizophrenia, schizoaffective disorder and bipolar disorder), and their family members.

Methods: We conducted a proof of concept (pretest-post-test design) study. Adults with treatment resistant psychosis (n=76) and their family members (n=76) were recruited from a tertiary care inpatient unit in Vancouver, Canada. Participants completed the Genetic Counseling Outcome Scale (GCOS-24) and Illness Management Self-Efficacy Scale (IMSES) at two time points during a "control" period prior to receiving pGC (1-2 weeks prior to pGC, immediately pre-pGC), and then completed them again 1-month, and 6-months after pGC. Unadjusted linear regressions with generalized estimating equations for repeated measures were used to calculate mean differences (MD) with 95% confidence intervals (CI) and p-values for the GCOS-24 and IMSES scores over time.

Results: There were no significant changes in empowerment (mean GCOS-24 scores) for inpatient participants, and no significant changes in mean IMSES scores for patients or family members. However, for family members, there were significant increases in empowerment scores 1-month post pGC (MD=4.4, 95% CI=1.2-7.5, p=0.007) and 6 months post pGC (MD=7.4, 95% CI=2.9-11.8, p=0.001).

Conclusion: Larger samples sizes of individuals stratified by the severity of their symptoms of psychosis could help with further determining the impact of pGC in this inpatient population. Furthermore, the observed mean differences in empowerment scores for family members suggest small-moderate effect sizes. This is the first study to our knowledge, to demonstrate longer lasting effects (6 months post-pGC) of pGC on empowerment for family members of individuals with serious mental illness (schizophrenia, schizoaffective disorder, and bipolar disorder with psychosis).

O31 - Concurrent Tumour and Germline Multi-Gene Panel Testing in Epithelial Ovarian Cancer: A Prospective Study - *Melanie Care*

Objective: Next-generation sequencing (NGS) of BRCA1 and BRCA2 is routinely performed on tumour samples from individuals with epithelial ovarian cancer (EOC). Targeted tumour testing can identify individuals potentially at risk for BRCA1/2-associated hereditary cancer but not other inherited cancer syndromes. We sought to determine the feasibility of tumour-first testing as a means by using a large multi-gene panel to analyze paired tumour and germline samples.

Methods: Tumour and blood samples were collected prospectively from individuals with newly diagnosed EOC. An NGS multi-gene hereditary breast/ovarian/prostate (HBOP) cancer panel (BRCA1, BRCA2, MLH1, MSH2, MSH6, PMS2, EPCAM, RAD51C, RAD51D, BRIP1, ATM, BARD1, CDH1, CHEK2, PALB2, POLD1, POLE, PTEN, STK11, TP53) was completed for both samples. Results were compared to determine the sensitivity of tumour NGS in identifying germline variants and the prevalence of inherited cancer syndromes in EOC.

Results: A total of 73 individuals had paired HBOP panel testing, with a mean of 2 variants detected per tumour (range 0-6). Excluding TP53, 45/73 patients (62%) had at least one potentially significant variant identified in their tumour tissue; 25/45 (56%) had at ≥1 likely pathogenic/pathogenic (LP/P) variant and 20/45 (44%) had only variants of uncertain significance (VUS). 11/73 patients (15%) were identified to carry germline LP/P variants (BRCA1[6]; BRCA2[3], RAD51C[1]; ATM[1]). Of those with only VUS in tumour, 16 (80%) had ≥1 confirmed to be germline. Twenty-two large copy number variants (CNVs) were identified in 13 tumour samples including 10 full gene deletions, 4 multi-exon deletions, 7

	full gene duplications, and 1 multi-exon duplication. Of these, only two BRCA1 multi-exon deletions were confirmed to be germline. All germline variants including LP/P and VUS were independently identified by tumour testing. Conclusion: In our cohort of EOC patients, 12% were identified to have BRCA1/2-associated hereditary cancer and 3% were identified to have another cancer syndrome. Tumour multi-gene panel testing identified all germline variants, though comparison with a germline sample remains a requirement for confirmation of variant origin. Excluding TP53, 38% of patients had no significant variants identified by tumour testing. These results suggest a tumour-first testing approach may be feasible to identify individuals at risk for inherited cancer syndromes and potentially reducing the number of germline multi-gene panel tests required in this population.
10:30am-11:00am **West Foyer**	**Refreshment Break**
11:00am-11:45am **BC/Alberta/Yukon Ballroom**	**Use of Race in Multiple Marker Screening** *Moderator: Susan Creighton* *Speaker: Danna Hull* An overview of the work Prenatal Screening Ontario has done in collaboration with our Ontario Multiple Marker Screening Laboratories on the use of race in Multiple Marker Screening (MMS).
11:45am-12:30pm **BC/Alberta/Yukon Ballroom**	**Genetic Counselling in Aboriginal Australian Peoples Living in Remote Australia** *Moderator: Susan Creighton* *Speaker: Lindsay Tuer* A unique personal account, including cultural insights and challenges, based on the experience of working as the state representative genetic counsellor in the Northern Territory of Australia for five years. Discussion will also be derived from the long-term genetic counselling work within the MJD Foundation. The MJD Foundation partners with Aboriginal & Torres Strait Islander communities to support families living with Machado-Joseph Disease (MJD) and Spinocerebellar Ataxia Type 7 (SCA7).
12:30pm-12:45pm **BC/Alberta/Yukon Ballroom**	**Closing Remarks** *Speaker: Julia Heaton*
12:45pm	**CAGC Conference Concludes**

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