BRCA 2023

Moving into the Mainstream | Suivre le courant

May 2-5 | Du 2 au 5 mai MONTRÉAL, QUÉBEC, CANADA

Ninth International Symposium on Hereditary Breast and Ovarian Cancer
9e Symposium international sur le cancer héréditaire du sein et de l'ovaire

AGENDA

Tuesday, May 2, 2023

8:00 - 9:45 REGISTRATION / CONTINENTAL BREAKFAST

9:45 - 10:15 **Introduction to the Programme**

Harley Eisman, MD, Co-Founder, Hereditary Breast and Ovarian Cancer Foundation, Montréal, QC, Canada

William Foulkes, MBBS, PhD, Director, Program in Cancer Genetics, McGill University, Montréal, QC, Canada

Session 1: Mutations, variants, databases 10:15 - 11:45

CHAIR: **Lawrence Brody,** PhD, National Human Genome Research Institute, National Institutes of Health, Bethesda, MD, USA

10:15 - 10:35 **LECTURE**

Resolving ENIGMAtic VUSs, updating VCEPs and helping ClinVar

Amanda Spurdle, PhD, QIMR Berghofer Medical Research Institute, Brisbane, QLD, Australia

10:35 - 10:55 **LECTURE**

BRCA Exchange: the one-stop shop for BRCA1/2 variant information

Melissa Cline, PhD, University of California Santa Cruz Genomics Institute, Santa Cruz, CA, USA

10:55 - 11:05 **PROFFERED PAPER 1 (S1-PP1)**

Functional and clinical characterization of BRCA2 hypomorphic missense variants

Fergus J. Couch, PhD, Mayo Clinic, Rochester, MN, USA

11:05 - 11:15 **PROFFERED PAPER 2 (S1-PP2)**

CHEK2gether study of the ENIGMA consortium: functional analysis of CHEK2 missense variants identified in breast cancer patients

Petra Kleiblova, MD, PhD, Charles University, Prague, Czech Republic

Tuesday, May 2, 2023 *(cont'd)*

11:17 - 11:20 **RAPID FIRE POSTER PRESENTATION (S1-RF1)**
Poster # P040: BRCA1 frameshift variants leading to extended incorrect protein termini
Thales Nepomuceno, PhD, Moffitt Cancer Center, Tampa, FL, USA

11:20 - 11:23 **RAPID FIRE POSTER PRESENTATION (S1-RF2)**
Poster # P041: A comprehensive characterization of missense variants of uncertain significance (VUS) in RAD51C
Jean-Yves Masson, PhD, CHU de Québec-Université Laval Research Center, Québec, QC, Canada

11:25 - 11:45 **Question and discussion period (all speakers)**

11:45 - 13:15 LUNCH

Session 2: Clinically relevant pathology 13:15 - 14:45

CHAIR: **Raymond Kim**, MD, PhD, University of Toronto, Toronto, ON, Canada

13:15 - 13:35 **LECTURE**
How can molecular pathology of HBOC help the cancer genetics health professional?
Jorge S. Reis-Filho, MD, PhD, Memorial Sloan Kettering Cancer Center, New York, NY, USA

13:35 - 13:55 **LECTURE**
Non-epithelial ovarian cancers and their associated syndromes
Blaise Clarke, MD, University Health Network, Toronto, ON, Canada

13:55 - 14:05 **PROFFERED PAPER 1 (S2-PP1)**
The genomic landscape of high grade serous ovarian tumors is defined by loss of BRCA1 and BRCA2 throughout chemoresistance and relapse
Nicole Gull, PhD, Cedars-Sinai Medical Center, Los Angeles, CA, USA

14:05 - 14:15 **PROFFERED PAPER 2 (S2-PP2)**
Replication stress and defective checkpoints render Fallopian tube epithelial cells as putative driver of high-grade serous ovarian cancer
Shailja Pathania, PhD, University of Massachusetts Boston, Boston, MA, USA

14:17 - 14:20 **RAPID FIRE POSTER PRESENTATIONS (S2-RF1)**
Poster # P002: Improving the diagnosis of serous tubal intraepithelial carcinoma (STIC) using deep-learning
Joep Bogaerts, MD, PhD candidate, Radboud University Medical Center, Nijmegen, The Netherlands

14:20 - 14:23 **RAPID FIRE POSTER PRESENTATIONS (S2-RF2)**

Poster # P003: Chromothripsis and whole genome duplication are both detectable in early-stage high-grade serous carcinoma (HGSC) of upper gynaecological tract; implications for HGSC risk mitigation

Ju-Yoon Yoon, MD, PhD, Unity Health Toronto (St. Michael's Hospital), Toronto, ON, Canada

14:25 - 14:45 **Question and discussion period (all speakers)**

POSTER SESSION 1 *(with break)* 14:45 - 16:15

Session 3: SPECIAL ROUND TABLE: Mainstreaming genetic testing for HBOC: A paradigm shift 16:15 - 17:45

MODERATOR:
Laura Palma, MSc, CGC, CCGC, McGill University Health Centre, Montréal, QC, Canada

Where to Begin? Implementation & evaluation of the mainstreaming genetic testing model

Jeanna McCuaig, MSc, PhD, University Health Network, Toronto, ON, Canada

Patient understanding and responses to mainstreaming genetic testing

Jada G. Hamilton, PhD, MPH, Memorial Sloan Kettering Cancer Center, New York, NY, USA

Cost-effectiveness and population impact of mainstreaming for breast and ovarian cancer

Ranjit Manchanda, MD, MRCOG, PhD, Wolfson Institute of Population Health, Queen Mary University of London, London, UK

Wednesday, May 3, 2023

8:00 - 9:00 CONTINENTAL BREAKFAST

9:00 - 9:15 **Introduction to the Programme**

Harley Eisman, MD, Co-Founder, Hereditary Breast and Ovarian Cancer Foundation, Montréal, QC, Canada

William Foulkes, MBBS, PhD, Director, Program in Cancer Genetics, McGill University, Montréal, QC, Canada

Session 4: The breast cancer genes: How to implement what we know and what to expect next — 9:15 - 10:45

CHAIR: Britta Weigelt, PhD, Memorial Sloan Kettering Cancer Center, New York, NY, USA

9:15 - 9:35 **LECTURE**

CanRisk: personalising cancer risk prediction for prevention and early detection

Antonis Antoniou, PhD, University of Cambridge, Cambridge, UK

9:35 - 9:55 **LECTURE**

The breast cancer susceptibility gene scorecard: What is in, what is out and what is left to find?

Clare Turnbull, MD, PhD, MFPH, Institute of Cancer Research, London, UK

9:55 - 10:05 **PROFFERED PAPER 1 (S4-PP1)**

Finding new causes of hereditary breast cancer: resolving the role of inherited promoter hypermethylation in known hereditary breast cancer genes and mutations in non-coding regions of BRCA1, BRCA2 and PALB2

Ian Campbell, PhD, Peter MacCallum Cancer Centre, Melbourne, VIC, Australia

10:05 - 10:15 **PROFFERED PAPER 2 (S4-PP2)**

Multiplexed assays of variant effect for all possible missense alterations located in the DNA Binding Domain of BRCA2

Fergus J. Couch, PhD, Mayo Clinic, Rochester, MN, USA

10:17 - 10:20 **RAPID FIRE POSTER PRESENTATIONS (S4-RF1)**

Poster # P059: Exome sequencing identifies novel susceptibility genes and defines the contribution of coding variants to breast cancer risk

Naomi Wilcox, BSc, MPhil, PhD Student, University of Cambridge, Cambridge, UK

10:20 - 10:23 **RAPID FIRE POSTER PRESENTATIONS (S4-RF2)**

Poster # P083: Universal whole genome tumour and germline sequencing of newly diagnosed breast cancer

Ian Campbell, PhD, Peter MacCallum Cancer Centre, Melbourne, VIC, Australia

10:25 - 10:45 **Question and discussion period (all speakers)**

Wednesday, May 3, 2023 *(cont'd)*

10:45 - 11:15 BREAK

Session 5: New approaches to the early diagnosis of hereditary cancers 11:15 - 12:45

CHAIR: Alan Spatz, MD, MSc, McGill University, Montréal, QC, Canada

11:15 - 11:35 **LECTURE**
Hereditary Cancer Surveillance Using Cell-free DNA Sequencing
Trevor Pugh, PhD, Princess Margaret Cancer Centre, Ontario Institute for Cancer Research, Toronto, ON, Canada

11:35 - 11:55 **LECTURE**
Li-Fraumeni as a case study of the effect of early diagnosis on outcomes
David Malkin, MD, The Hospital for Sick Children (SickKids), Toronto, ON, Canada

11:55 - 12:05 **PROFFERED PAPER 1 (S5-PP1)**
Increase in tumor DNA testing during nationwide implementation of the Tumor-First workflow to optimize identification of heredity in epithelial ovarian cancer
Vera M. Witjes, MSc, PhD Candidate, Radboud University Medical Center, Nijmegen, The Netherlands

12:05 - 12:15 **PROFFERED PAPER 2 (S5-PP2)**
MRI surveillance and breast cancer mortality in BRCA1 and BRCA2 mutation carriers
Steven Narod, MD, PhD, Women's College Hospital, Toronto, ON, Canada

12:17 - 12:20 **RAPID FIRE POSTER PRESENTATIONS (S5-RF1)**
Poster # P029: Somatic testing lags far behind germline testing in patients with epithelial ovarian cancer: a missed opportunity
Kayla Ostiller, MD, Kaiser Permanente San Francisco, San Francisco, CA, USA

12:20 - 12:23 **RAPID FIRE POSTER PRESENTATIONS (S5-RF2)**
Poster # P031: Facilitated cascade testing for families with identified variants associated with hereditary gynecologic cancers
Sarah S. Lee, MD, MBA, NYU Langone Health, New York, NY, USA

12:25 - 12:45 **Question and discussion period (all speakers)**

12:45 - 14:15 LUNCH

Wednesday Special Interest Groups (SIGs) 14:15 - 15:45

Moderated 90-minute interactive discussions *with the aim of fostering bilateral knowledge transfer. Choice of one of the following 3 SIGs.*

SIG 1
Other BRCA-associated Cancers (pancreas and prostate): Germline Testing, Early Detection and Targeted Cancer Therapies

- **George Zogopoulos**, MD, PhD, McGill University Health Centre, Montréal, QC, Canada
- **Danny Vesprini**, MD, MSc, Sunnybrook Health Sciences Centre, Toronto, ON, Canada

SIG 2
Professional ethical duty and liability in a context of increasing decentralization and de-specialisation of genetic tests

- **Yann Joly,** PhD (DCL), FCAHS, AdE, Centre of Genomics and Policy, McGill University, Montréal, QC, Canada *(session lead)*
- **David J. Peloquin**, JD, Ropes & Gray LLP, Boston, MA, USA
- **Yvonne Bombard,** PhD, Unity Health Toronto and University of Toronto, Toronto, ON, Canada

SIG 3
Cancer Concerns for Transgender and Gender Diverse People

- **Joshua D. Safer**, MD, Mount Sinai Center for Transgender Medicine and Surgery, Mount Sinai Health System, New York, NY, USA *(session lead)*
- **Sharon Bober,** PhD, Dana-Farber Cancer Institute/Harvard Medical School, Boston, MA, USA
- **Sarah Roth**, PhD Candidate, Johns Hopkins University, Baltimore, MD, USA
- **Sasha Weiss**, PhD Student, Indiana University, Bloomington, IN, USA

15:45 - 16:15 BREAK

Session 6: SPECIAL ROUND TABLE: Disparities in Hereditary Cancer Genetics – What are we doing to improve the situation? 16:15 - 17:45

MODERATORS:
Lawrence Brody, PhD, National Human Genome Research Institute, Bethesda, MD, USA;
Laurence Baret, MA, MSc, CGC, CCGC, McGill University Health Centre, Montréal, QC, Canada

Tuya Pal, MDCM, Vanderbilt – Ingram Cancer Center, Nashville, TN, USA

Yvonne Bombard, PhD, Unity Health Toronto and University of Toronto, Toronto, ON, Canada

Funmi Olopade, MD, FACP, OON, Center for Clinical Cancer Genetics, University of Chicago Medicine, Chicago, IL, USA

Grace-Ann Fasaye, ScM, CGC, National Institutes of Health, National Cancer Institute, Bethesda, MD, USA

Thursday, May 4, 2023

8:00 – 9:00 CONTINENTAL BREAKFAST

9:00 – 9:30

Introduction to the Programme

Harley Eisman, MD, Co-Founder, Hereditary Breast and Ovarian Cancer Foundation, Montréal, QC, Canada

William Foulkes, MBBS, PhD, Director, Program in Cancer Genetics, McGill University, Montréal, QC, Canada

Remembering David Livingston, MD (1941-2021)

Introduction:
William Foulkes, MBBS, PhD, Director, Program in Cancer Genetics, McGill University, Montréal, QC, Canada

Speaker:
Judy Garber, MD, MPH, Dana-Farber Cancer Institute, Boston, MA, USA

Session 7: New developments in HBOC management 9:30 - 11:00

CHAIR: Tuya Pal, MDCM, Vanderbilt – Ingram Cancer Center, Nashville, TN, USA

9:30 - 9:50

LECTURE

Treatment (and prevention) of BRCA1/2-related breast cancer – Platinums, PARPs and beyond

Judy Garber, MD, MPH, Dana-Farber Cancer Institute, Boston, MA, USA

9:50 - 10:10

LECTURE

Is cure of hereditary ovarian cancer an achievable goal?

Jonathan Ledermann, MD, University College London Cancer Institute, London, UK

10:10 - 10:20

PROFFERED PAPER 1 (S7-PP1)

Risk-reducing mastectomy and breast cancer mortality in women with a BRCA1 or BRCA2 pathogenic variant: an international analysis

Kelly Metcalfe, RN, PhD, University of Toronto, Toronto, ON, Canada

10:24 - 10:27

RAPID FIRE POSTER PRESENTATIONS (S7-RF1)

Poster # P129: Early salpingectomy with delayed oophorectomy as alternative for risk-reducing salpingo-oophorectomy in BRCA1/2 pathogenic variant carriers - Update of the TUBA-WISP II study

Majke van Bommel, PhD candidate; and **Marleen Kets**, MD, PhD, Radboud University Medical Center, Nijmegen, The Netherlands

Thursday, May 4, 2023 *(cont'd)*

10:27 - 10:30 **RAPID FIRE POSTER PRESENTATIONS (S7-RF2)**
Poster # P073: Bilateral oophorectomy and all-cause mortality in women with BRCA1 pathogenic variants
Joanne Kotsopoulos, MSc, PhD, Women's College Hospital, University of Toronto, Toronto, ON, Canada

10:30 - 10:33 **RAPID FIRE POSTER PRESENTATIONS (S7-RF3)**
Poster # P121: Describing the real world experience of implementing a ore-test genetic counselling portal within a provincial hereditary cancer program
Jennifer Nuk, MSc, BC Cancer Hereditary Cancer Program, Vancouver, BC, Canada

10:33 - 10:36 **RAPID FIRE POSTER PRESENTATIONS (S7-RF4)**
Poster # P084: Novel blood-based biomarker to monitor neoadjuvant therapy outcome In triple-negative breast cancer patients
Srinidi Mohan, PhD, University of New England School of Pharmacy, Portland, ME, USA

10:40 - 11:00 **Question and discussion period (all speakers)**

11:00 - 11:30 BREAK

Thursday Special Interest Groups (SIGs) 11:30 - 13:00

Moderated 90-minute interactive discussions *with the aim of fostering bilateral knowledge transfer. Choice of one of the following 3 SIGs.*

SIG 4
The Emotional Pedigree: Addressing the Heart of Complex Psychosocial Issues in GC sessions with HBOC Patients

- **Karen Hurley**, PhD, Cleveland Clinic, Cleveland, OH, USA

SIG 5
Exploring cut-offs: when is the risk high enough to consider salpingo-oophorectomy?

- **Helen Hanson**, MBBS, MD(Res), St George's Hospital, London, UK *(session lead)*
- **Lesa Dawson**, MD, University of British Columbia, Vancouver, BC, Canada; and Memorial University, St. John's, NL, Canada
- **Ranjit Manchanda**, MD, MRCOG, PhD, Wolfson Institute of Population Health, Queen Mary University of London, London, UK

SIG 6
CanRisk Workshop – Multifactorial breast and ovarian cancer risk prediction

- **Antonis Antoniou**, PhD, University of Cambridge, Cambridge, UK *(session lead)*
- **Marc Tischkowitz,** MD, PhD, University of Cambridge, Cambridge, UK

13:00 - 14:30 LUNCH

Thursday, May 4, 2023 *(cont'd)*

Session 8: Polygenic risk scores in practice 14:30 - 16:00

CHAIR: Jacques Simard, PhD, FRSC, FCAHS, Centre de recherche du CHU de Québec - Université Laval, Québec, QC, Canada

14:30 - 14:50 **LECTURE**
Polygenic risk scores and moderate risk genes
Paul James, MD, PhD, Parkville Familial Cancer Centre, Peter MacCallum Cancer Centre, Melbourne, VIC, Australia

14:50 - 15:10 **LECTURE**
Will polygenic risk scores for cancer ever be clinically useful?
Clare Turnbull, MD, PhD, MFPH, Institute of Cancer Research, London, UK

15:10 - 15:20 **PROFFERED PAPER 1** (S8-PP1)
Implementation of risk stratified breast cancer screening: clinical use of multifactorial risk assessment including polygenic risk scores to guide recommendations
Anna Maria Chiarelli, PhD, Ontario Health, Toronto, ON, Canada

15:20 - 15:30 **PROFFERED PAPER 2** (S8-PP2)
Addition of breast density (BD) and polygenic risk score (PRS) In breast cancer (BC) risk estimation among women with family history (PRiSma Study)
Judith Balmaña, MD, PhD, Vall d'Hebron University Hospital, Barcelona, Spain

15:30 - 15:40 **PROFFERED PAPER 3** (S8-PP3)
Comparison of the impact of integrating BRCA1 and BRCA2 pathogenic variants and polygenic risk score for individual genetic risk estimation in three cancer types
Ryoko Yamada, PhD, RIKEN, Yokohama, Kanagawa, Japan

15:40 - 16:00 **Question and discussion period (all speakers)**

POSTER SESSION 2 *(with break)* 16:00 - 17:30

Friday, May 5, 2023

8:00 - 9:00 CONTINENTAL BREAKFAST

9:00 - 9:15 **Introduction to the Programme**

Harley Eisman, MD, Co-Founder, Hereditary Breast and Ovarian Cancer Foundation, Montréal, QC, Canada

William Foulkes, MBBS, PhD, Director, Program in Cancer Genetics, McGill University, Montréal, QC, Canada

Session 9: Population-based testing for HBOC-related genes – Pros and cons 9:15 - 10:45

CHAIR: Ranjit Manchanda, MD, MRCOG, PhD, Wolfson Institute of Population Health, Queen Mary University of London, London, UK

9:15 - 9:35 **LECTURE**

Testing the limits of population-based testing

Mark E. Robson, MD, Memorial Sloan Kettering Cancer Center, New York, NY, USA

9:35 - 9:55 **LECTURE**

Population testing for HBOC genes: time for action

Funmi Olopade, MD, FACP, OON, Center for Clinical Cancer Genetics, University of Chicago Medicine, Chicago, IL, USA

9:55 - 10:05 **PROFFERED PAPER 1 (S9-PP1)**

Hereditary cancer testing In an unselected 'Healthy' Canadian population: Is it time to consider population screening?

Jessica Gu, MSc, MS, Medcan, Toronto, ON, Canada

10:05 - 10:15 **PROFFERED PAPER 2 (S9-PP2)**

Implementation and outcomes of a program to increase hereditary cancer testing ordered by genetics and non-genetics providers in over 12,500 patients

Mónica Alvarado, MS, LGC, Kaiser Permanente Southern California, Pasadena, CA, USA

10:17 - 10:20 **RAPID FIRE POSTER PRESENTATIONS (S9-RF1)**

Poster # P111: Genetic contribution of BRCA1 in hereditary breast and ovarian cancer in Senegalese women

Rokhaya Ndiaye, PharmD, PhD, Université Cheikh Anta Diop, Dakar, Sénégal

10:20 - 10:23 **RAPID FIRE POSTER PRESENTATIONS (S9-RF2)**

Poster # P104: Early insights from the DNA screen study, an Australian pilot study of population genomic screening

Tu Nguyen-Dumont, PhD, Monash University, Clayton, VIC, Australia

10:25 - 10:45 **Question and discussion period (all speakers)**

10:45 - 11:15 BREAK

Friday Special Interest Groups (SIGs) 11:15 - 12:45

Moderated 90-minute interactive discussions *with the aim of fostering bilateral knowledge transfer. Choice of one of the following 2 SIGs.*

SIG 7
Alternative Models of Service Delivery

- **Intan Schrader**, MBBS, PhD, BC Cancer, University of British Columbia, Vancouver, BC, Canada *(session lead)*
- **Kelly Metcalfe**, RN, PhD, University of Toronto, ON, Canada
- **Nicoline Hoogerbrugge,** MD, PhD, Radboud University Medical Center, Nijmegen, The Netherlands

SIG 8
The lower risk and rarer breast and/or ovarian cancer genes – What do we know and what should we do with what we know?
(e.g. CHEK2, ATM, BRIP1, BARD1, NTHL1)

- **Tuya Pal,** MDCM, Vanderbilt – Ingram Cancer Center, Nashville, TN, USA *(session lead)*
- **Helen Hanson**, MBBS, MD(Res), St George's Hospital, London, UK
- **Raymond Kim**, MD, PhD, University of Toronto, Toronto, ON, Canada
- **Marc Tischkowitz,** MD, PhD, University of Cambridge, Cambridge, UK

Closing Session: The Marla Miller Memorial Lecture 13:00 - 14:15

13:00 - 14:15

Introductory Remarks

William Foulkes, MBBS, PhD,
Director, McGill University Program in Cancer Genetics,
Montréal, QC, Canada

LECTURE
Optimizing Treatment of Breast Cancer in BRCA1 Carriers

Steven Narod, MD, PhD
Senior Scientist, Women's College Hospital, Toronto, ON, Canada

Presentation by the Miller Family

Closing Remarks

Harley Eisman, MD,
Co-Founder, Hereditary Breast and Ovarian Cancer Foundation,
Montréal, Québec, Canada