CANSSI ONTARIO **STAGE**


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# Inherited Susceptibility to Ovarian Cancer: The Story So Far

**Friday**
**June 2, 2023**
**12:00 – 1:00 pm**

Hybrid Event I Register Here
9th Floor, Rooms 9014 & 9016
700 University Avenue
Toronto ON


**Abstract:** Epithelial ovarian cancer has a substantial inherited genetic component. Over the past 25 years, substantial progress has been made to unravel the underlying genetic architecture of risk. Linkage studies in the 1990's in multi-case, multi-generation families were successful at identifying BRCA1 and BRCA2; rare loss-of-function allele of these genes are associated with a high risk of disease. The search for common, modest risk alleles using association study designs were initially based on candidate variant/candidate gene studies, but developments in genotyping technology together with the formation of large, international consortia ushered in an era of genome-wide association studies which have been successful in identifying multiple risk variants. In the past decade large-scale, targeted sequencing of candidate genes in case-control studies has enabled the identification loss-of-function alleles of BRIP1, PALB2, RAD51C and RAD51D that are associated with intermediate disease risks. Future developments will be dependent on large scale exome and whole genome sequencing.


**Profile:** I am a research Scientist in the Department of Computational Biomedicine at Cedars Sinai Medical Center, Los Angeles. I qualified in medicine from the University of Oxford in 1986. After a series of posts in internal medicine I worked for a year in Malawi on a leprosy vaccine trial. I then completed my training in public health medicine before taking up a post as research fellow in the CRC Human Cancer Genetics group at the University of Cambridge in 1996. Having completed my doctoral studies in 1999 I won a Cancer Research UK Senior Clinical Research Fellowship. On completion of my fellowship in 2009 I was appointed Reader in Cancer Epidemiology and promoted to a personal Chair in 2012, Department of Public Health and Primary Care, University of Cambridge. I moved to Los Angeles in November 2022. My major research interests are i) common genetic variation and breast and ovarian cancer susceptibility ii) the role of germline genotype in determining the clinical and molecular pathological characteristics of breast and ovarian cancer. I have published over 600 papers reporting original research as well as numerous book chapters and review articles.


Housed at the University of Toronto Dalla Lana School of Public Health, CANSSI Ontario **STAGE** is a training program in genetic epidemiology and statistical genetics funded by CANSSI Ontario with the support of the University of Toronto Faculty of Arts & Science. Seminars are sponsored by The Hospital for Sick Children Genetics and Genome Biology Program; the Lunenfeld-Tanenbaum Research Institute, Mount Sinai Hospital; and the McLaughlin Centre at the University of Toronto.


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