

**Canadian College
of
Medical Geneticists**

Annual Scientific Meeting



Final Program

November 12-14, 2009
The Fairmont Banff Springs — Banff, AB



CCMG 2009 Annual Scientific Meeting

Conference Learning Objectives

In addition to the specific educational objectives of each presentation, by the end of the scientific symposia, the participant will be able to

1. Recognize genetic and environmental factors which are involved in the development of complex diseases.
2. Understand the genome-wide association approach (the strengths and weaknesses) for the identification of genetic susceptibility to complex diseases.
3. Describe the potential risks of birth defects associated with assisted reproductive technologies.
4. Identify genetic pathways and molecular defects responsible for visual impairment in humans and model organisms.

Please note that 25% of each presentation will be allocated for questions, audience participation and interaction.

WEDNESDAY, NOVEMBER 11

0800-1200	BOARD OF DIRECTORS MEETING – <i>Strathcona Room</i>
1200-1800	STRATEGIC PLANNING SESSION – <i>By Invitation Only – Angus Room</i>

THURSDAY, NOVEMBER 12

0800-1200	REGISTRATION - <i>Van Horne Ballroom Foyer</i>
0800-1000	COMMITTEE MEETINGS CREDENTIALS – ROOM 1982 EDUCATION – ROOM 1976 EXAMINATIONS – ROOM 1978 NOMINATIONS – ROOM 1980
0900-1200	POSTER SET UP – <i>Van Horne Ballroom B</i>
1000-1200	COMMITTEE MEETINGS AWARDS – ROOM 1982 CLINICAL PRACTICE – ROOM 1976 ETHICS AND PUBLIC POLICY – ROOM 1978 PRENATAL DIAGNOSIS – ROOM 1980 SCIENTIFIC PROGRAM AND SPONSORSHIP – ROOM 1974
1200-1345	LUNCH AND KEYNOTE ADDRESS – <i>Van Horne Ballroom A</i> Moderator: Martin J. Somerville, PhD, FCCMG, Edmonton, AB 1200-1210 WELCOME AND OPENING REMARKS 1210-1300 LUNCH



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THURSDAY, NOVEMBER 12 (continued)

- 1300-1345 KEYNOTE ADDRESS – ENVIRONMENTAL INFLUENCES ON TRANSGENERATIONAL EFFECTS
Speaker: Judith Hall, OC, MD, FCAHS, FRCPC, FCCMG, Vancouver, BC
Speaker of the Royal College of Physicians and Surgeons of Canada
Conférencière du Collège royal des médecins et chirurgiens du Canada
Objectives: By the end of this session, participants will be able to:
1. List six adult complex diseases associated with IUGR.
 2. Distinguish monozygotic from dizygotic twins.
 3. List “new” features of a family history.
- 1345-1445 **ORAL ABSTRACT PRESENTATIONS – Van Horne Ballroom A**
- 1345-1405 RAPID ANEUPLOIDY DETECTION FOR LOW RISK PREGNANCIES: A SUITABLE REPLACEMENT FOR G-BANDING?
Speaker: Marsha Speevak, PhD, FCCMG, Mississauga, ON
- 1405-1425 HYPERPHOSPHATASIA WITH SEIZURES, NEUROLOGIC DEFICIT AND CHARACTERISTIC FACIAL FEATURES: FIVE NEW CASES OF MABRY SYNDROME
Speaker: David Cole, MD, PhD, BSc, FRCPC, FCCMG, Toronto, ON
- 1425-1445 UNDERSTANDING THE MOLECULAR PATHOGENESIS OF MALFORMATION SYNDROMES IN AN ISOLATED POPULATION
Speaker: Kym Boycott, MD, PhD, FRCPC, FCCMG, Ottawa, ON
- 1445-1645 **COMMITTEE MEETINGS**
- ACCREDITATION OF CENTRES – Room 1982
 - BIOCHEMICAL GENETICS – Room 1974
 - CONSTITUTION & BYLAWS – Room 1976
 - CYTOGENETICS – Room 1978
 - MOLECULAR GENETICS – Room 1980
- 1645-1745 **CCMG/CAGC POSTER PRESENTATIONS – Van Horne Ballroom B**
- 1800-1930 **CCMG/CAGC WELCOME RECEPTION – Mt. Stephen Hall**
- 1800-2100 **EXHIBITORS SET UP – Van Horne Ballroom C**

FRIDAY, NOVEMBER 13

- 0700-0830 **REGISTRATION – Van Horne Ballroom Foyer**
- 0700-0830 **CONTINENTAL BREAKFAST – Van Horne Ballroom C**
- 0700-0830 **PEACE COMMITTEE MEETING – Room 1976**
- 0730-0830 **CCMG/CAGC JOINT PLANNING COMMITTEE MEETING – Room 1974**
- 0730-1700 **EXHIBITS OPEN – Van Horne Ballroom C**



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FRIDAY, NOVEMBER 13 (continued)

0830-1200	CCMG/CAGC SYMPOSIUM – CAGC Organized – Van Horne Ballroom A Moderator: Stephanie A. Kieffer, MS, CGC, CCGC, Vancouver, BC
0830-0835	INTRODUCTION AND ANNOUNCEMENTS
0835-0920	THE FEASIBILITY OF REGULATING ONTARIO GENETIC COUNSELLORS – RESULTS OF A TARGETED SURVEY (CAGC SMALL PROJECTS GRANT UPDATE) Speakers: Christopher Trevors, MS, CGC, Toronto, ON and Nada Quercia, MSc, CCGC, CGC, Toronto, ON Objectives: By the end of this session, participants will be able to: <ol style="list-style-type: none">1. Report on the results of a provincial study entitled: The Attitudes towards and Feasibility of Professional Regulations of Genetic Counsellors in Ontario.2. Introduce the Ontario Regulated Health Professionals Act as it applies to Genetic Counsellors.3. Help the audience appreciate the complexities and difficulties of professional regulation for Genetic Counsellors in Ontario.
0920-1010	CONGENITAL ANOMALY SURVEILLANCE IN CANADA – PAST, PRESENT AND FUTURE Speaker: R. Brian Lowry, MD, DSc, FRCPC, FCCMG, Calgary, AB Objectives: By the end of this session, participants will be able to: <ol style="list-style-type: none">1. Describe surveillance.2. Distinguish between active and passive systems.3. Describe evaluation methods.
1010-1025	REFRESHMENT BREAK
1025-1115	DUCHENNE MUSCULAR DYSTROPHY AND EMERGING THERAPIES Speaker: Jean Mah, MD, MSc, FRCPC, Calgary, AB Objectives: By the end of this session, participants will be able to: <ol style="list-style-type: none">1. Outline the clinical features and natural history of Duchenne muscular dystrophy (DMD).2. Know the current management of DMD.3. Understand emerging / new therapeutic strategies for DMD.
1115-1200	BRIDGING THE GAP: THE TRANSLATIONAL CARE MODEL Speaker: Christèle du Souich, MSc, CCGC, CGC, Vancouver, BC Objectives: By the end of this session, participants will be able to: <ol style="list-style-type: none">1. Describe the Translational Care Model.2. Illustrate how this model is applied to rare diseases.3. Demonstrate how it can be used to develop approaches to ameliorate social and medical concerns.
1200-1330	LUNCH WITH EXHIBITORS – Van Horne Ballroom C
1200-1330	ROYAL COLLEGE SPECIALTY COMMITTEE IN MEDICAL GENETICS MEETING – Room 1976
1345-1630	ANNUAL GENERAL MEETING – Van Horne Ballroom A
1800	FOUNDERS AWARD DINNER – the Bison – 211 Bear Street



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SATURDAY, NOVEMBER 14

0800-0830 CONTINENTAL BREAKFAST – *Van Horne Ballroom C*

0800-1200 REGISTRATION – *Van Horne Ballroom Foyer*

0800-1400 EXHIBITS OPEN – *Van Horne Ballroom C*

0830-1200 CCMG/CAGC SYMPOSIUM – CCMG Organized – WHOLE GENOME ASSOCIATION STUDIES – *Van Horne Ballroom A*

Moderator: Jan M. Friedman, MSc, MD, PhD, FABMG, FACMG, FRCPC, FCCMG, Vancouver, BC

0830-0835 INTRODUCTION AND ANNOUNCEMENTS

0835-0920 PLAYING TOGETHER: EPIDEMIOLOGICAL ISSUES IN GENOME-WIDE ASSOCIATION STUDIES

Speaker: Julian Little, PhD, Ottawa, ON

Objectives: By the end of this session, participants will be able to:

1. Identify epidemiological aspects of the design, conduct and analysis of genome-wide association studies.
2. Describe potential biases that may influence the results of genome-wide association studies, such as in the selection of study participants, population stratification, and genotyping error (differential, non-differential).
3. Assess critically the public health value of genome-wide association studies, in particular implications for disease prediction and for gene-environment joint effects.

0920-1010 GWAS AND POSTGWAS IN COMPLEX DISEASES.

Speaker: Jerome I. Rotter, MD, Los Angeles, CA

Objectives: By the end of this session, participants will be able to:

1. Summarize recent findings of family, epidemiologic, physiologic, and linkage studies of the common inflammatory bowel diseases, Crohn's disease and ulcerative colitis.
2. Explain how understanding the genetic susceptibility to etiologically complex gastrointestinal/inflammatory diseases can improve our knowledge of their pathogenesis.
3. Describe how the identification of genetically high-risk individuals can be used to improve prognostic assessment and guide therapy.

1010-1025 REFRESHMENT BREAK

1025-1115 TYPE 1 DIABETES – LIFE IN THE POST-GWA ERA

Speaker: Constantin Polychronakos, MD, FRCPC, Montreal, QC

Objectives: By the end of this session, participants will be able to:

1. Understand what the results of genome-wide association studies tell us and what questions remain unanswered.
2. Understand the possible explanations for the portion of genetics of complex diseases that remain unexplained.
3. If rare but highly penetrant variants do turn out to be a substantial part of the genetic architecture of complex traits, the audience should be ready to use their accumulating knowledge in the development of personalized medicine.



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SATURDAY, NOVEMBER 14 (*continued*)

- 1115-1200 THE CANADIAN PHARMACOGENOMIC NETWORK FOR DRUG SAFETY: FINDING DRUG SAFETY SOLUTIONS TO SERIOUS ADVERSE DRUG REACTIONS
Speaker: Bruce Carleton, PharmD, Vancouver, BC
Objectives: By the end of this session, participants will be able to:
1. Cite the areas in which pharmacogenomics have applications at the bedside.
 2. Identify ways pharmacogenomics are transforming practical clinical drug dosing strategies.
 3. List the barriers that remain to the advancement of bench to bedside care.

1200-1300

CCMG SYMPOSIUM – HOT TOPIC: Clinical Application of Whole Genome Sequencing – *Van Horne Ballroom A*

Moderator: Jan M. Friedman, MSc, MD, PhD, FABMG, FACMG, FRCPC, FCCMG, Vancouver, BC

NEW GENERATION SEQUENCING FOR GENOME ANALYSIS

Speaker: Marco Marra, PhD (Genetics), FRSC, Vancouver, BC

Objectives: By the end of this session, participants will be able to:

1. Summarize the advantages and limitations of highly parallel sequencing in comparison to conventional Sanger sequencing.
2. Describe how highly parallel sequencing can be used to identify genetic variants that cause or are associated with constitutional disease.
3. Describe how highly parallel sequencing can be used to identify genetic variants that cause or are associated with the development of cancers.

1300-1400

CCMG LUNCH WITH EXHIBITORS – *Van Horne Ballroom C*

1300-1400

POSTER REMOVAL – *Van Horne Ballroom B*

1400-1715

CCMG SYMPOSIUM – CALGARY SESSION – *Van Horne Ballroom A*

Moderators: Ross McLeod, MD, FRCPC, FCCMG, and Judy Chernos, PhD, FCCMG, Calgary, AB

1400-1530 REPRODUCTIVE GENETICS

DOES ART INCREASE THE RISK OF BIRTH DEFECTS? - EVIDENCE FROM THE BENCH

Speaker: Renée Martin, PhD, FCCMG, Calgary, AB

Objectives: By the end of this session, participants will be able to:

1. Discuss data regarding the likelihood of normal progeny following ART.
2. Discuss the particular risks of progeny from ICSI.
3. Realize that infertility per se may increase the risk of children with birth defects.

DOES ART INCREASE THE RISK OF BIRTH DEFECTS? - EVIDENCE FROM THE BEDSIDE

Speaker: R. Douglas Wilson, MD, FRCPC, Calgary, AB

Objectives: By the end of this session, participants will be able to:

1. Describe the congenital anomalies that are associated with Assisted Reproductive Technology (ART).
2. Discuss the monozygotic twinning outcomes related to ART.
3. Review the rare genetic conditions that are at an increased risk following ART as determined in large population studies.



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SATURDAY, NOVEMBER 14 (continued)

1530-1545 REFRESHMENT BREAK

1545-1715 OPHTHALMIC GENETICS

CHOROIDEREMIA: NEW FINDINGS, NEW INSIGHTS

Speaker: Ian MacDonald, MD, FRCPC, FCCMG, Edmonton, AB

Objectives: By the end of this session, participants will be able to:

1. Recognize choroideremia as an inherited disorder of trafficking.
2. Identify the key clinical characteristics and laboratory findings of choroideremia.
3. Assess the potential of gene therapy in retinal disorders with the example of choroideremia.

GROWTH FACTORS, GROWTH CONES AND BRAIN MAPS

Speaker: Sarah McFarlane, PhD, Calgary, AB

Objectives: By the end of this session, participants will be able to:

1. Be introduced to the use of *Xenopus laevis* as a model system to understand the development of the visual system and in particular the optic nerve.
2. Explain the importance of using simple model systems to understand events in the developing embryo, and how conservation between species allows for the transference of information learned using one model system to be applied to others.
3. Describe the basic mechanisms by which connections between neurons in the developing brain are formed.
4. Describe the proper formation of molecular patterns within the developing brain that are key to the formation of neural networks within the brain
5. Discuss disorders/diseases of optic nerve formation that occur perinatally and postnatally in infants and children.

1400-1700 EXHIBITORS TEAR DOWN – *Van Horne Ballroom C*

1730-2030 CCMG BOARD OF DIRECTORS MEETING

1800 WESTERN NIGHT – *Wild Bill's Legendary Saloon – 201 Banff Ave*

SUNDAY, NOVEMBER 15

0900-1200 PHAC SESSION – *Room 1974* – A NATIONAL MECHANISM FOR INFORMATION SHARING – BY INVITATION ONLY

The Division of Continuous Professional Learning at the University of Alberta has approved this as an Accredited Group Learning Activity under Section 1 of the Framework of Continuing Professional Development options for the Maintenance of Certification Program of the Royal College of Physicians and Surgeons of Canada.

The Royal College of Physicians and Surgeons of Canada has provided a National Specialty Society Annual Meeting Grant in support of our meeting / Le Collège royal des médecins et chirurgiens du Canada appuie la présente réunion par l'octroi d'une Subvention pour la réunion annuelle d'associations nationales de spécialistes.

THANK YOU TO OUR SPONSORS

The Canadian College of Medical Geneticists gratefully acknowledges our sponsors for generously providing unrestricted educational grants in support of the CCMG 33rd Annual Scientific Meeting.

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OTHER CONTRIBUTORS



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