

**Canadian College  
of  
Medical Geneticists**

**Annual Scientific Meeting**



**Final Program**

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**October 21-23, 2010**  
The Westin Nova Scotian — Halifax, NS



# CCMG 2010 Annual Scientific Meeting

## Conference Learning Objectives

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

1. Discuss important changes that are likely to occur in the practice of genetic medicine in the next 5-10 years.
2. Describe the current status of pharmacogenetics in clinical medicine and discuss potential clinical applications of pharmacogenomics.
3. List several Mendelian conditions that are unusually common among population groups living in the Maritime Region of Canada and describe how improved scientific understanding of these disorders can be used to improve the health of these peoples.

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

## THURSDAY, OCTOBER 21

0800-1200

**REGISTRATION – *Commonwealth Foyer***

0800-1000

### COMMITTEE MEETINGS

CREDENTIALS – *Boardroom*

EDUCATION – *Fundy Room*

EXAMINATIONS – *Bedford Room*

NOMINATIONS – *Maritime Room*

0900-1200

**POSTER SET UP – *Commonwealth A***

1000-1200

### COMMITTEE MEETINGS

AWARDS – *Boardroom*

CLINICAL PRACTICE – *Fundy Room*

PRENATAL DIAGNOSIS – *Bedford Room*

SCIENTIFIC PROGRAM – *Maritime Room*

1200-1345

**LUNCH AND KEYNOTE ADDRESS – *Commonwealth A***

**Moderator: Jan Friedman, MD, PhD, FAAP, FABMG, FCCMG, FRCPC**

1200-1210 WELCOME AND OPENING REMARKS

1210-1300 LUNCH

1300-1345 KEYNOTE ADDRESS – THE FUTURE OF GENETIC MEDICINE

**Speaker: Roderick McInnes, CM, MD, PhD, FRSC, FCCMG, FRCPC**

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



# CCMG 2010 Annual Scientific Meeting

## THURSDAY, OCTOBER 21 (*continued*)

1. Indicate what the next 10 years might bring to medical, clinical, and clinical laboratory genetics.
2. Identify some of the challenges that medical geneticists, of all types, will face in both their research and practice.
3. Identify current trends in medical genetics that are changing clinical practice.

1345-1350 **BREAK**

1350-1505 **CCMG ORAL ABSTRACT PRESENTATIONS – *Commonwealth A***

1350-1405 CROSSTALK BETWEEN HISTONE MODIFICATIONS and DNA METHYLATION IN PATIENTS WITH INTELLECTUAL DISABILITY DUE TO JARID1C MUTATIONS

**Speaker: Dr. Hon-Yin Brian Chung**

1410-1425 HOW DO YOU KNOW YOUR GENETIC TEST RESULTS ARE CORRECT? EQA SCHEME RESULTS SHOW THE NEED FOR CONTINUED QUALITY IMPROVEMENT IN LABORATORY DIAGNOSTICS

**Speaker: Ms. Outi Kämäräinen**

1430-1445 IDENTIFICATION OF A HOMOZYGOUS STAR MUTATION IN FOUR MEMBERS OF TWO NEWFOUNDLAND FAMILIES WITH FAMILIAL GLUCOCORTICOID DEFICIENCY

**Speaker: Dr. Jane S. Green**

1450-1505 ARRAY CGH IN REPRODUCTIVE GENETICS

**Speaker: Dr. Evica Rajcan-Separovic**

1515-1715 **COMMITTEE MEETINGS**

ACCREDITATION OF CENTRES – *Boardroom*

BIOCHEMICAL GENETICS – *Fundy Room*

CONSTITUTION & BYLAWS – *Bedford Room*

CYTOGENETICS – *Cornwallis Room*

ETHICS & PUBLIC POLICY – *Northumberland Room*

MOLECULAR GENETICS – *Business Suite 250*

1730-1830 **CCMG/CAGC POSTER PRESENTATIONS – *Commonwealth A***

1800-2100 **EXHIBITORS SET UP – *Commonwealth B***

1830-2000 **CCMG/CAGC WELCOME RECEPTION – *Harbour Suites A & B***



# CCMG 2010 Annual Scientific Meeting

FRIDAY, OCTOBER 22

- 0700-0830 REGISTRATION – *Commonwealth Foyer*
- 0700-0830 BREAKFAST – *Commonwealth B*
- 0700-0830 PEACE COMMITTEE MEETING – *Maritime Room*
- 0730-1330 EXHIBITS OPEN – *Commonwealth B*
- 0830-1200 CCMG/CAGC SYMPOSIUM – CAGC ORGANIZED – *Commonwealth A*  
**Moderator: Pat Steele**
- 0830-0835 WELCOME AND OPENING REMARKS
- 0835-0920 INSURANCE AND GENETIC TESTING – HOW WILL IT REALLY IMPACT YOUR PATIENTS?  
**Speaker: Richard Sturk, BA**  
Objectives: By the end of this session, participants will be able to:
1. Describe the various types of insurance.
  2. Review how family history relates to insurance.
  3. Discuss: What advice can you give your patients.
- 0920-1010 SUSPECTING AND DIAGNOSING GENETIC CARDIAC DISEASE: THE ROLE OF AN INTERDISCIPLINARY CARDIAC GENETICS CLINIC  
**Speakers: Martin Gardner, MD, FRCPC, FAAC, Amy Crowley MS, CGC, and Janet Marcadier, MSc, CGC, CCGC**  
Objectives: By the end of this session, participants will be able to:
1. Discuss the history and evolution of the Inherited Heart Disease clinic.
  2. Describe examples of patients and families seen in the clinic with outcomes.
  3. Review the role of each health care professional in the IHD clinic – cardiologist, geneticist, genetic counsellor.
  4. Examine a case example using our Eskasoni outreach clinic.
  5. Describe future areas of development and collaboration, such as pathology and research involvement.
- 1010-1025 REFRESHMENT BREAK
- 1025-1115 STREAMLINING PSYCHOLOGICAL SERVICES TOWARDS THOSE IN MOST NEED: HOW? AND WHO ARE THEY?  
**Speaker: Mary Jane Esplen, PhD, RN**  
Objectives: By the end of this session, participants will be able to:
1. Describe the psychological impact of genetic testing.
  2. Illustrate specific risk factors associated with poorer psychological adjustment to genetics information.
  3. Define who to offer psychological services to during the genetic testing process.



# CCMG 2010 Annual Scientific Meeting

## FRIDAY, OCTOBER 22 (*continued*)

1115-1200	THE PATH TO A NEW CLINIC MODEL <b>Speaker: Sonya Grewal, MSc, CCGC</b> Objectives: By the end of this session, participants will be able to: <ol style="list-style-type: none"><li>1. Describe the purpose of LEAN in health care.</li><li>2. Discuss how we used the LEAN process to develop and implement a new clinic model.</li><li>3. Describe the challenges of developing new processes.</li></ol>
1200-1330	CCMG LUNCH WITH EXHIBITORS – <i>Commonwealth B</i>
1200-1330	ROYAL COLLEGE SPECIALTY COMMITTEE IN MEDICAL GENETICS MEETING – <i>Maritime Room</i>
1345-1630	CCMG ANNUAL GENERAL MEETING – <i>Commonwealth A</i>
1800	CCMG FOUNDERS AWARD DINNER – <i>Waterfront Warehouse - 1549 Lower Water Street</i>

## SATURDAY, OCTOBER 23

0800-0830	BREAKFAST – <i>Commonwealth B</i>
0800-1200	REGISTRATION – <i>Commonwealth Foyer</i>
0800-1300	EXHIBITS OPEN – <i>Commonwealth B</i>
0830-1200	CCMG/CAGC SYMPOSIUM – CCMG ORGANIZED– PHARMACOGENOMICS – <i>Commonwealth A</i> <b>Moderator: Jan Friedman, MD, PhD, FAAP, FABMG, FCCMG, FRCPC</b>
0830-0835	INTRODUCTION AND ANNOUNCEMENTS
0835-0920	THE CANADIAN PHARMACOGENOMICS NETWORK FOR DRUG SAFETY <b>Speaker: Bruce Carleton, BSc, PharmD</b> Objectives: By the end of this session, participants will be able to: <ol style="list-style-type: none"><li>1. Cite the areas in which pharmacogenomics have applications at the bedside.</li><li>2. Identify ways pharmacogenomics are transforming practical clinical drug dosing strategies.</li><li>3. List the barriers that remain to the advancement of bench to bedside care.</li></ol>
0920-1010	PHARMACOGENETICS AND THE TREATMENT OF CARDIOVASCULAR DISEASE <b>Speaker: Michael Phillips, PhD</b> Objectives: By the end of this session, participants will be able to: <ol style="list-style-type: none"><li>1. Explain how genetic variation in drug metabolism genes can affect patient drug response and safety.</li></ol>



# CCMG 2010 Annual Scientific Meeting

## SATURDAY, OCTOBER 23 (continued)

2. Summarize which cardiovascular drugs have been identified that display pharmacogenomic variation.
3. List which pharmacogenomic tests can be used to help direct Warfarin dosing.
- 1010-1025 REFRESHMENT BREAK
- 1025-1115 PHARMACOGENETICS AND DRUG REGULATION  
**Speaker: Lawrence Lesko, PhD, FCP**  
Objectives: By the end of this session, participants will be able to:
1. Identify three regulatory decisions that have significantly impacted pharmacogenomics.
  2. Understand the importance of evidence generation to support the use of genetic tests of drug response.
  3. Learn what the five major barriers are to further evolution of personalized medicine.
- 1115-1200 DIRECT-TO-CONSUMER GENETIC TESTING AND PHARMACOGENETICS  
**Speaker: Tanya Nelson, PhD, FCCMG**  
Objectives: By the end of this session, participants will be able to:
1. Distinguish the difference between the business model of direct-to-consumer genetic testing and that of genetic testing provided by a private, for profit, clinical laboratory.
  2. Describe at least three types of direct-to-consumer genetic testing currently available.
  3. Discuss the perceived benefits and risks of direct-to-consumer genetic testing.
- 1200-1300 CCMG LUNCH WITH EXHIBITORS – *Commonwealth B*
- 1200-1300 POSTER REMOVAL – *Commonwealth A*
- 1300-1400 CCMG SYMPOSIUM – HOT TOPICS: CLINICAL APPLICATIONS OF EXOME SEQUENCING – *Commonwealth A*  
**Speaker: Jan Friedman, MD, PhD, FAAP, FABMG, FCCMG, FRCPC**  
Objectives: By the end of this session, participants will be able to:
1. Describe what exome sequencing is and how it is done.
  2. Discuss the strengths and limitations of whole exome sequencing in comparison to conventional gene-by-gene sequencing on one hand and whole genome sequencing on the other.
  3. Recognize clinical conditions for which exome is most likely to be useful.
- 1400-1715 CCMG SYMPOSIUM – MARITIME PROVINCES SESSION – *Commonwealth A*  
**Moderator: Sarah Dyack, MD, FCCMG, FRCPC**
- 1400-1445 GROWING UP WITH CHARGE SYNDROME  
**Speaker: Kim Blake, MBs (London), MRCPC**



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## SATURDAY, OCTOBER 23 (continued)

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

1. Describe the adolescent continuum of CHARGE syndrome.
2. Discuss the use of botulinum toxin type A for decreasing oral secretions.
3. Describe how to identify and treat Sleep Apnea in CHARGE syndrome.

1445-1530

THE MI'KMAQ PEOPLE: A MARITIME GENETIC ISOLATE

**Speaker: Sarah Dyack, MD, FCCMG, FRCPC**

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

1. Review the history of Mi'kmaq people post European contact.
2. Discuss genetic conditions that are prevalent in this group.
3. Discuss the challenges of providing genetic care to this population.

1530-1545

REFRESHMENT BREAK

1545-1630

THE ACADIANS OF THE MARITIME: HISTORY, GENETICS AND GENE DISCOVERY

**Speaker: David Skidmore, MD, FCCMG, FRCPC**

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

1. Summarize the history of the Acadian peoples and how it predisposed to the creation of both local and panethnic founder affects.
2. Identify some of the different, medically significant autosomal recessive conditions believed to be endemic in this population.
3. Discuss the results and implications of some of the ongoing gene discovery projects within this population.

1630-1715

LEVERAGING A PUBLICLY-FUNDED HEALTH CARE SYSTEM TO ASCERTAIN AND CHARACTERIZE NATURALLY OCCURRING HUMAN MUTATIONS

**Speaker: Andrew Orr, MD, FRCSC**

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

1. Learn more about a large-scale Mendelian discovery project in Atlantic Canada.
2. Learn about new, rapid methods in gene identification.

1400-1700

EXHIBITORS TEAR DOWN – *Commonwealth B*

1900

CCMG SOCIAL EVENT – *Join us for an evening Halifax style!*

*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.*

## 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 34<sup>th</sup> Annual Scientific Meeting.

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