

# PROGRAM



# GARROD SYMPOSIUM

*Unique Populations,  
Unique IEMs*



## MAY 7 - 9, 2020

Supported by:



in collaboration with:



more information: <https://interprofessional.ubc.ca/initiatives/garrodsymposium2020/>

Delta Hotels by Marriott London Armouries  
London, ON

# GENERAL INFORMATION

By the end of the meeting participants will be able to:

## LEARNING OBJECTIVES & PROGRAM

- Learn about new metabolic disease therapies including gene therapies, small molecule therapies, and enzyme replacement therapies
- Expand their knowledge of metabolic diseases in Indigenous populations
- Learn about metabolic diseases in Amish, Mennonite and Hutterite populations
- Share experiences in providing metabolic genetic services to new immigrants

Please note: 25% of the scientific program will be interactive.

---

## REGISTRATION

**ONLINE:** The quickest, most secure method is registering online. Registration is available for Visa/MasterCard holders on the symposium website:

<https://interprofessional.ubc.ca/initiatives/garrodsymposium2020/>

**BY FAX:** Fax your completed registration form to: 1-604-822-4835

**PHONE REGISTRATION:** Toll free within North America: 1-855-827-3112; other callers: 1-604-827-3112

**MAIL:** Send the registration form with cheque to this address:

IN 9580 REGISTRATION, Interprofessional Continuing Education, The University of British Columbia, Room 105-2194 Health Sciences Mall, Vancouver, BC, V6T 1Z3, Canada

Participants paying by credit card outside of North America: Please inform your credit card company of the transaction as some banks put a block on credit card payments made outside your country.

---

## TUITION FEES & MEETING PROGRAM

Please see the registration form (last page) for details. The tuition fee includes: welcome reception (May 7), breakfast (May 8&9), lunch (May 8&9), webinars and workshops (May 7-9), Garrod Association membership rate (if selected) and a certificate of attendance. Registration for the dinner event on Friday will also be available at an extra cost. Pre-registration before/on **March 27, 2020** is strongly recommended to take advantage of the early-bird rate. **To qualify for the Garrod Association member's rate, one must pay the fee for membership dues during the registration process.**

We will be including presenter slides (if the presenter permits) in our meeting program. We will send this program to you electronically (for free) a few days prior to the meeting.

---

## CANCELLATION & REFUND POLICY

Notifications of cancellations must be sent by email to [registration.ipce@ubc.ca](mailto:registration.ipce@ubc.ca) on or before **March 27, 2020** in order for registration fees to be refunded. If you have been invoiced but not paid the fees, you are still liable to pay the cancellation charges below:

- Up to and including **March 27, 2020:** Registration fee will be refunded less 20% cancellation fee.
- After **March 27, 2020:** No refunds on registration fees will be made under ordinary circumstances. Substitutions can be made.

---

## LOCATION

The meeting will be held at the Delta Hotels by Marriott London Armouries (325 Dundas St, London, ON). A special rate of \$159.00 for a standard guest room is available for conference delegates until April 10, 2020 or until availability. To avoid disappointment, book by calling: 855-213-0582 and specify that you are booking under the 'Garrod Symposium 2020', or book online [HERE](#). *Please specify that you are booking under the Garrod Symposium 2020 room block to receive these reduced rates.*

---

## CREDITS

More information about credits will be available soon.

---

## CONTACT

For registration enquiries, please contact 604-827-3112 or email [registration.ipce@ubc.ca](mailto:registration.ipce@ubc.ca). For other enquiries, please contact Jo at 604-822-0054 or [jo.ipce@ubc.ca](mailto:jo.ipce@ubc.ca).

# SPONSORS

We would like to acknowledge with great appreciation the financial contributions through unrestricted educational grants from the following organizations:

## PLATINUM

**B**  **OMARIN**<sup>®</sup>

## GOLD



---

## WIFI



## SILVER



## BRONZE





# SUB-MEETINGS & WEBINARS

## THURSDAY MAY 7

12:00- 14:00 **CIMDRN Meeting (Invitation Only)**  
(Room: Chelsea Green)

14:00- 16:00 **Nurse/Genetic Counselor Webinar**  
(Room: Thames Valley)  
*Session Chairs: Melanie Napier, Christina Parkinson*

14:00- 16:00 **Dietitians Webinar**  
(Room: Linden)  
*Session Chairs: Amy Pender, Suzanne Ratko*

14:00 **Cultural Competency in Provision of Healthcare to Amish and Mennonite Families**  
**Victoria Mok Siu, MD, FRCPC, FCCMG, Medical Director, Medical Genetics Program of Southwestern Ontario; Associate Professor, Department of Pediatrics, Schulich School of Medicine and Dentistry, Western University, London, ON**

15:00 **Dietary Management of Maple Syrup Urine Disease Pre- and Post-Liver Transplant: A Case Presentation**  
**Laura Nagy, MSc RD, Registered Dietitian, Division of Clinical and Metabolic Genetics, The Hospital for Sick Children**

15:15 **Homocystinuria**  
**Krista Viau, PhD, RD, CSP, Metabolic Dietitian, Boston Children's Hospital, Boston, MA, USA**



15:00 **Registration for Garrod Symposium Opens**  
(Room: Armouries Ballroom Foyer)

16:00 **Introduction and Welcome to the Meeting**  
(Room: Armouries Ballroom)

**Murray Potter, MD, FRCPC, FCCMG, Biochemical Geneticist, McMaster Children's Hospital; Professor and Chair, Department of Pathology and Molecular Medicine, McMaster University, Hamilton, ON**

**Andreas Schulze, President of Garrod Association, MD, PhD, FRCPC, Professor Paediatrics and Biochemistry, University of Toronto, Toronto, ON**

16:15-18:00 **Plenary Session I: New Therapies Part I**  
(Room: Armouries Ballroom)  
*Session Chair: Tony Rupar*

16:15 **DNA: From Diagnosis to Treatment. The Modern Age of Diagnosing and Treating Metabolic Diseases**  
**Aneal Khan, MSc, MD, FRCPC, FCCMG, Professor of Medical Genetics and Pediatrics, University of Calgary Cumming School of Medicine, Alberta Children's Hospital Research Institute, Calgary, AB**

1. Summarize the technological basis for DNA and RNA transfer technologies in metabolic diseases
2. Appreciate how measures of clinical efficacy can be dependent on the technology used
3. Recognize how different types of gene transfer methods might be suitable for different types of metabolic diseases

17:00 **New Aspects of Fabry Disease Including Treatment With Pharmacological Chaperones**

**Daniel G. Bichet, MD, Professor of Medicine, Pharmacology and Physiology, University of Montreal, co-principal investigator of the Canadian Fabry Disease Initiative, Nephrologist, Hôpital du Sacré-Coeur de Montréal, Montréal, QC**

1. Become familiar with early recognition of Fabry disease
2. Recognize the concept of misfolded amenable mutations corrected by a pharmacological chaperone
3. Review the renal and cardiac changes observed after intravenous enzyme replacement therapy or oral pharmacological chaperone treatment

17:45 **Abstract #1: Development of Gene Therapy for Tay-Sachs/ Sandhoff Disease: Preparation of a Phase I/II a Clinical Trial**

**Jagdeep S Walia, Secretary/Treasurer of Garrod Association, MBBS, FRCPC, FCCMG, Clinical Geneticist and Associate Professor, Head, Division of Medical Genetics (Department of Pediatrics), Director of Research (Department of Pediatrics), Kingston Health Sciences Centre and Queen's University, Kingston, ON**

18:00 - 19:30 **WELCOME RECEPTION & EXHIBITS**  
(Room: Gunnery & Officers Club)

19:30 - 21:00 **Garrod Executive Committee Meeting (Invitation Only) (Room: Talbot)**

07:30- Registration  
08:45 (Room: Armouries Ballroom Foyer)  
Breakfast & Exhibits  
(Room: Gunnery & Officer Club)

08:45- **Plenary Session II: Unique Populations**  
09:45 (Room: Armouries Ballroom)  
*Session Chair: Chitra Prasad*

08:45 **Working With the Old Order Amish and Mennonite Populations: Successes and Challenges in the Delivery of Healthcare and Targeted Genetic Testing of Newborns and Adults**  
*Victoria Mok Siu, MD, FRCPC, FCCMG, Medical Director, Medical Genetics Program of Southwestern Ontario; Associate Professor, Department of Pediatrics, Schulich School of Medicine and Dentistry, Western University, London, ON*

1. Discuss cultural traditions of the Old Order populations, which need to be considered in the provision of healthcare
2. Recognize factors, which affect uptake of targeted newborn and carrier screening
3. Describe successful implementation strategies for optimal care of Old Order families with genetic/metabolic disorders

09:15 **Genetic Research with Manitoba's Hutterite and Mennonite Communities: Navigating Tradition and Innovation**  
*Cheryl Rockman-Greenberg, MD, CM, FRCPC, FCCMG, Distinguished Professor, Departments of Pediatrics & Child Health and Biochemistry & Medical Genetics, University of Manitoba and Clinician Scientist, Children's Hospital Research Institute of Manitoba, Winnipeg, MB*

1. Recognize the challenges in pursuing genetic research in Manitoba's unique populations
2. Summarize three approaches to patient engagement in research and delivery of genetic services for Inherited Metabolic Diseases in Manitoba's unique populations
3. Discuss the impact of the drug approval process in Canada on access to new therapies for unique populations

09:45 **Abstract #2: Clinical Characteristics of Patients from Quebec, Canada, with Morquio A Syndrome: A Longitudinal Cohort Study**  
*John Mitchell, Endocrinologist Montreal Children's Hospital, McGill University Health Center, Montreal, QC*

10:00- **Refreshment Break & Exhibits**  
10:45 (Room: Gunnery & Officer Club)  
& Even-numbered Posters (Room: Pine)

10:45- **Plenary Session III: Indigenous Health**  
12:00 (Room: Armouries Ballroom)  
*Session Chair: Resham Ejaz*

10:45 **Protective or Deleterious? the Unresolved Debate of the Effect of the CPT1A p.P479L Variant on Inuit and West Coast First Nations Health**  
*Laura Arbour, MSc, MD, FRCPC, FCCMG. Professor, Department of Medical Genetics, University of British Columbia, Vancouver, BC; Affiliate Professor, Division of Medical Sciences, University of Victoria, Victoria, BC*

1. Demonstrate the known population distribution of the CPT1A p.P479L variant in Canada and other circumpolar countries
2. Describe the evidence for both deleterious and protective effects of the CPT1A p.P479L variant
3. Consider the benefits and potential harms of population screening and intervention



11:30 **Pyruvate Carboxylase Deficiency in Indigenous Populations: Prevalence and Phenotype**  
*Aziz Mhanni, MD, FRCPC, FCCMG, FACMG, PhD, Consultant, Medical Genetics and Genomics, Associate Professor, Department of Pediatrics and Child Health; Department of Biochemistry and Medical Genetics, Max Rady College of Medicine, Rady Faculty of Health Sciences, University of Manitoba, Winnipeg, MB*

1. Discuss the genetic epidemiology of pyruvate carboxylase deficiency
2. Recognize the phenotypic spectrum of pyruvate carboxylase deficiency
3. Describe the neuroradiologic abnormalities in pyruvate carboxylase deficiency

12:00 - 13:00 **Lunch & Exhibits**  
(Room: Gunnery & Officer Club)

13:00-14:30 **Plenary Session IV: New Immigrants**  
(Room: Armouries Ballroom)  
*Session Chair: Andrea Yu*

13:00 **Caring for Children and Youth New to Canada: Resources and Considerations for Health Care Providers**  
*Andrea Hunter, MD, FRCPC, DTM&H (UK), Consultant Pediatrician, McMaster Children's Hospital, Refuge: Hamilton Centre for Newcomer Health; Program Director, Pediatric Residency Program; Associate Professor, McMaster University, Hamilton, ON*

1. Describe the demographics of newcomer children and youth to Canada
2. Outline how culture may impact health and developmental issues
3. Describe the extent of and the challenges and health issues facing immigrant and refugee families in Canada, and discuss steps that health providers can take in caring for, and advocating with newcomer families

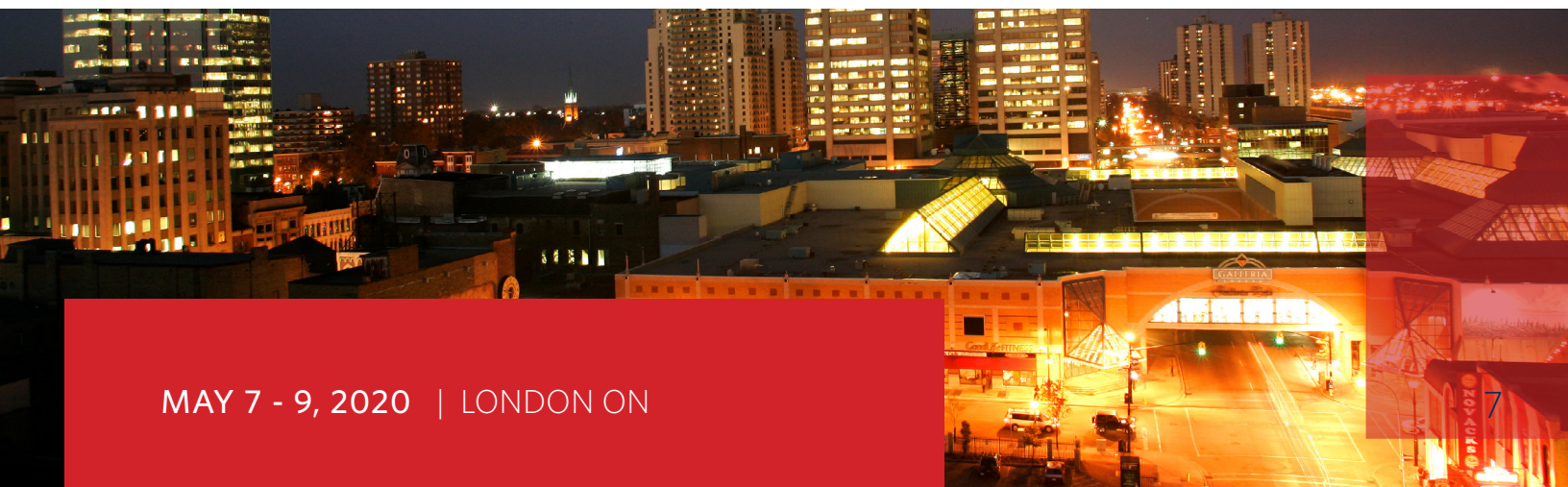
13:45 Patient Family Experiences: Hamilton Case

14:00 Patient Family Experiences: London Case

14:15 **Abstract #3: Lysinuric Protein Intolerance Mimicking N-acetylglutamate Synthase Deficiency in a Nine-year-old Refugee Boy**  
*Sarah Al-Qattan*

14:30-15:15 **Refreshment Break & Exhibitors**  
(Room: Gunnery & Officer Club) & **Odd-numbered Posters** (Room: Pine)

15:15-17:00 **Plenary Session V: New Therapies Part II**  
(Room: Armouries Ballroom)  
*Session Chair: Murray Potter*



15:15 **Palynziq™: Novel Enzyme Substitution Therapy for Adults with Phenylketonuria (PKU)**

**Cary Harding, MD**, Professor, Molecular and Medical Genetics, and Pediatrics, Oregon Health & Science University, Portland, OR, USA

1. Review the Phase 3 trial data that forms the basis of regulatory approval for the use of pegvaliase (Palynziq™) treatment in PKU
2. Provide a practical approach to the induction and titration of pegvaliase therapy using specific case studies
3. Illustrate the management of pegvaliase related hypersensitivity reactions

16:00 **Nutrition Management of Patients Treated with Pegvaliase**

**Krista Viau, PhD, RD, CSP**, Metabolic Dietitian, Boston Children's Hospital, Boston, MA, USA

1. Identify the dietitian's roles in the initiation and treatment of adults with PKU with pegvaliase
2. Describe nutrition assessment and management strategies during titration and maintenance pegvaliase therapy
3. Discuss nutrition counseling considerations for increasing dietary protein intake

16:45 **Abstract #4: Landscape of Treatable Rare Diseases in a Founder Population**

**Alison Eaton,**

17:00 **Mini Break**

17:15-  
18:15 **Interesting Cases and New Diagnostics**

(Room: Armouries Ballroom)

Session Chair: Mariya Kozenko

17:15 **Abstract #5: Whole Genome Sequencing Identifies a Rare Case of Moderate Zellweger Spectrum Disorder Caused by a PEX3 Defect**

**Whiwon Lee, MS, CGC**, Genetic Counsellor, Centre for Genetic Medicine, The Hospital for Sick Children, Toronto, ON



08:00-  
09:00 Breakfast  
(Room: Gunnery & Officer Club)

09:00-  
12:00 **Plenary Session VI: Adult Metabolics**  
(Room: Armouries Ballroom)  
*Session Chair: Natalya Karp*

09:00- **Polygenic Effects in Disease**  
**Robert A. Hegele, MD, FRCPC, FACP, Distinguished University Professor of Medicine and Biochemistry, University of Western Ontario, London, ON**

1. Contrast polygenic versus monogenic effects on phenotypes
2. Derive polygenic scores
3. Apply polygenic scores to research and clinical applications

09:45 **Abstract #9: Long-term Migalastat Treatment Slowed the Decline of Renal Function in Patients with Fabry Disease and Amenable GLA Variants**  
**Daniel Bichet, MD, Physician, Hôpital Du Sacré-Coeur Université de Montréal, Montréal, QC**

10:00-  
10:30 **Refreshment Break & Exhibitors**  
(Room: Gunnery & Officer Club)

10:30 **Adults Are Not Large Children: Care of Adults With Inborn Metabolic Diseases**  
**Sandra Sirrs, MD, FRCPC, Medical Director, Adult Metabolic Diseases Clinic, Clinical Professor, UBC Division of Endocrinology, Vancouver, BC**

1. List challenges faced by adults with IMDs
2. Describe resource requirements clinics for adults with IMDs
3. Discuss common situations in adults where evaluation for IMDs is indicated

11:00 **(Adult Management)**  
**Hanna Faghfoury**

1. TBD
2. TBD
3. TBD

11:30 **(Dietary Management)**  
**Heather Bell**

1. TBD
2. TBD
3. TBD

12:00 **Closing Remarks**  
**Chitra Prasad, MD FRCPC FCCMG, Director of Metabolic Clinic, London Health Sciences Centre, Professor, Department of Paediatrics, Section of Genetics and Metabolism, Western University, London, ON**

**Paula J Waters, PhD, FCCMG, Vice President of Garrod Association, Biochemical Geneticist and Affiliate Professor, Medical Genetics Service, Department of Pediatrics, CHUS- Université de Sherbrooke, Sherbrooke, QC**

12:05-  
13:00 **Garrod Association Membership Meeting & Lunch**  
(Room: Armouries Ballroom)

# GARROD 2020 COMMITTEES

## Garrod 2020 Symposium Presidents

**Murray Potter**, MD, FRCPC, FCCMG, Biochemical Geneticist, McMaster Children's Hospital; Professor and Chair, Department of Pathology and Molecular Medicine, McMaster University, Hamilton, ON

**Suzanne Ratko**, RD, HBSc, BSc, Registered Dietitian, Children's Hospital, London, Health Sciences Centre, London, ON

## Garrod 2020 Symposium Scientific Committee

**Resham Ejaz**, MD, FRCPC, Assistant Professor, Division of Genetics, Department of Pediatrics, McMaster University, Hamilton, ON

**Natalya Karp**, MD, FRCPC, FCCMG, MSc, Medical Geneticist, London Health Sciences Centre, Assistant Professor, Department of Paediatrics, University of Western Ontario, London, ON

**Mariya Kozenko**, MD, FRCPC, FCCMG, Associate Professor, Division of Genetics, Department of Pediatrics, McMaster University, Hamilton, ON

**Melanie Napier**, MSc, MSc, CGC, CCGC, Genetic Counsellor, London Health Sciences Centre, London, ON

**Christina Parkinson**, MSc, CGC, CCGC, Genetic Counsellor, Department of Pediatrics, McMaster Children's Hospital, Hamilton, ON

**Amy Pender**, MSc, RD, Registered Dietitian, Department of Pediatrics, McMaster Children's Hospital, Hamilton, ON

**Murray Potter**, MD, FRCPC, FCCMG, Biochemical Geneticist, McMaster Children's Hospital; Professor and Chair, Department of Pathology and Molecular Medicine, McMaster University, Hamilton, ON

**Chitra Prasad**, MD FRCPC FCCMG, Director of Metabolic clinic, London Health Sciences Centre, Professor, Department of Paediatrics, Section of Genetics and Metabolism, Western university, London, ON

**Suzanne Ratko**, RD, HBSc, BSc, Registered Dietitian, Children's Hospital, London, Health Sciences Centre, London, ON  
Children's Hospital – London Health Sciences Centre, London, ON

**Tony Rupar**, PhD, FCCMG, Section Head, Biochemical Genetics Laboratory, London Health Sciences Centre Professor, Pathology and Laboratory Medicine, Pediatrics and Biochemistry Western University, London, ON

**Andrea C Yu**, MD FRCPC FCCMG, Medical Geneticist, London Health Sciences Centre, Assistant Professor, Department of Paediatrics, University of Western Ontario, London, ON

## Garrod 2020 Symposium Organizer

**Jo Nam**, Senior Education Manager, Interprofessional Continuing Education, University of British Columbia, Vancouver, BC

## Garrod Association Executive Committee

**Andreas Schulze**, President of Garrod Association, MD, PhD, FRCPC, Professor Paediatrics and Biochemistry, University of Toronto, Toronto, ON

**Jagdeep S Walia**, Secretary/Treasurer of Garrod Association, MBBS, FRCPC, FCCMG, Clinical Geneticist and Associate Professor, Head, Division of Medical Genetics (Department of Pediatrics), Director of Research (Department of Pediatrics), Kingston Health Sciences Centre and Queen's University, Kingston, ON

**Paula J Waters**, PhD, FCCMG, Vice President of Garrod Association, Biochemical Geneticist and Affiliate Professor, Medical Genetics Service, Department of Pediatrics, CHUS-Université de Sherbrooke, Sherbrooke, QC



### AFFILIATION / PROFESSION

(please select only one)

- Physician   
  Registered Nurse   
  Dietitian   
  Genetic Counsellor   
  Lab  
 Other: \_\_\_\_\_

### DELEGATE INFORMATION

Please write in block letters and use one registration form per person. Please photocopy the form if needed.

- Dr.   
  Mr.   
  Ms.

Last Name	First Name	Initials
Institution / Organization		
Mailing Address		
City	Prov/State	Postal Code
Daytime Telephone Number / Local		Email Address for Meeting Correspondence

(If applicable) Please inform us of any dietary requirements for the meeting: \_\_\_\_\_

### REGISTRATION RATES

(Rates include all applicable taxes)

	On or before March 27, 2020	After March 27, 2020 or Onsite
Non-Members	<input type="checkbox"/> \$450	<input type="checkbox"/> \$550
Members**	<input type="checkbox"/> \$330	<input type="checkbox"/> \$430
<b>(This rate includes the \$30 Garrod Association membership dues)</b>		
Student/ Trainee	<input type="checkbox"/> \$100	<input type="checkbox"/> \$150
Allied Health Professional	<input type="checkbox"/> \$100	<input type="checkbox"/> \$150

### SOCIAL EVENTS

Additional\* Guest for Welcome Reception (Thursday, May 7)

\*The welcome reception is included for Garrod registrants.

Gala Dinner at Museum London (Friday, May )

Regular Garrod Attendee

Additional guest(s)

\$50

\$80

\$120

### METHOD OF PAYMENT

#### Payment by Credit Card *Most secure and easy method*

- Complete the full registration online at [www.interprofessional.ubc.ca](http://www.interprofessional.ubc.ca) with your Visa or MasterCard.
- Fax\* the registration form to +1-604-822-4835 and indicate that you would like to pay with Visa or MasterCard. We will send you the secure online link to enter your credit card information.
- Register and pay over the phone: Local/International: +1-604-827-3112 or toll free within Canada/USA: +1-855-827-3112.

\*Please do not fax your credit card information

#### Payment by Cheque

Please make your cheque payable to the University of British Columbia and send it along with this form to: Interprofessional Continuing Education, The University of British Columbia, Room 105-2194 Health Sciences Mall, Vancouver, BC V6T 1Z3.

#### Alternative Payment Methods

Mail or fax complete registration form along with one of the following:

- Signed purchase order (PO)
- Letter of Authorization (LOA) from the manager on the organization's letterhead stating that they will be paying the registration fees. Must include the amount of registration fees, name and contact information of the manager
- Signed cheque requisition form (ChReq)

Please indicate below how you would like to pay:

- Cheque:** Payment is enclosed with mailed registration form  
 **Credit Card:** Please email me a secure link to enter my credit card number  
 **PO/LOA/ChReq:** Purchase order/letter of authorization/cheque requisition form is enclosed with faxed/mailed registration form