

# **GARROD**

# SYMPOSIUM **2019**

## **NEUROMETABOLISM**



# **MAY 9 - 11, 2019**



**SHERATON CENTRE**  
TORONTO, ON



THE GARROD  
ASSOCIATION

in cooperation with  
THE UNIVERSITY OF BRITISH COLUMBIA  
Interprofessional  
Continuing  
Education

[INTERPROFESSIONAL.UBC.CA/INITIATIVES/GARRODSYMPOSIUM2019](http://INTERPROFESSIONAL.UBC.CA/INITIATIVES/GARRODSYMPOSIUM2019)

# GENERAL INFORMATION

## LEARNING OBJECTIVES & PROGRAM

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

- Understand different types of inherited metabolic disorders presenting with neurological features
- Expand their knowledge for treatable neurometabolic conditions and their treatments
- Learn specific neuroimaging patterns of neurometabolic disorders
- Describe organelle specific neurometabolic disorders involving mitochondria, lysosomes and peroxisomes
- Learn the neurodevelopmental outcomes and neuropsychiatric manifestations of neurometabolic disorders

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

## LOCATION & TRAVEL

The meeting will be held at the Sheraton Centre Toronto (123 Queen Street, Toronto, ON). A special rate of \$239.00 for a standard guest room (Traditional Room: single occupancy) is available for conference delegates until April 9, 2019 or until availability. To avoid disappointment, book by calling: 1.888.627.7171, please specify that you are booking under the Garrod Symposium 2019 room block to receive these reduced rates.

## REGISTRATION

**ONLINE:** The quickest, most secure method is registering online. Registration is available for Visa/MasterCard holders at the meeting organizer's website: [www.interprofessional.ubc.ca/initiatives/garrodsymposium2019](http://www.interprofessional.ubc.ca/initiatives/garrodsymposium2019)

**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 9), breakfast (May 10 & 11), lunch (May 10 & 11), webinars and workshops (May 9-11), 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 29, 2019** 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 but if you wish to purchase the paper copy that will be distributed on-site, please select the \$20 option on the registration page.

## 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 29, 2019 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 29, 2019:** Registration fee will be refunded less 20% cancellation fee.
- After **March 29, 2019:** No refunds on registration fees will be made. Substitutions can be made.

## CREDITS

This event has been approved by the Canadian Paediatric Society for a maximum of 10.75 credit hours as an Accredited Group Learning Activity (Section 1) as defined by the Maintenance of Certification program of The Royal College of Physicians and Surgeons of Canada. The specific opinions and content of this event are not necessarily those of the CPS, and are the responsibility of the organizer(s) alone.

This conference has been approved by the Canadian Association of Genetic Counsellors (CAGC) for 13.0 CECs.

## CONTACT

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

# SPONSORS

The Garrod Association and the Program Committee gratefully acknowledge the generous contributions and financial support from the following organizations:

## PLATINUM



## GOLD



## BRONZE



# PROGRAM THURSDAY

May 9, 2019

Please see [www.garrod.ca](http://www.garrod.ca) for other meetings before/after the meetings noted below.

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

**13:00-16:00** Nurse/Genetic Counselor Webinar  
(Room: Maple West)

## The Hostage Syndrome: Our Relationship with Patients

**Joe Clarke, MD, PhD, FRCPC, FCCMG, Professor Emeritus, University of Toronto, Toronto, ON**

At the end of the presentation, the listener will be able to:

1. Recount at least 4 characteristics of the "hostage syndrome" as it relates to the relationship between a health care professional and patient with a rare, inherited metabolic disease.
2. Describe how to communicate empathically with the parents of a child with a newly diagnosed inherited metabolic disease.
3. Describe what is meant by the importance of "curiosity" when managing inherited metabolic diseases.
4. Describe 3 potential obstacles to the establishment of rapport with the parents of a child with an inherited metabolic disease.

Following Dr. Clarke's presentation, there will be a closed session (~120mins) for case presentations/discussion.

**13:00-16:00** Dietitians Webinar  
(Room: Linden)

## Growing up on Sapropterin: Experiences with young children on cofactor therapy

**Leslie Martell, MS, RD, LDN, Metabolic Dietitian with Boston Children's Hospital, Boston, MA, USA**

1. Describe use of Sapropterin in young children with Phenylketonuria
2. Illustrate considerations for diet liberalization in the younger population
3. Discuss strategies and approaches to working with families and young children as they grow up on Sapropterin

## "Stop, Go, Proceed with Caution": Classification of fruit and vegetables according to protein content, using a traffic light analogy (a resource for patients with phenylketonuria)

**Laura Nagy MSc, RD, Registered Dietitian with Metabolic Genetics and the PKU Program at the Hospital for Sick Children, Toronto, ON and Valerie Austin, BSc**

1. Understand the rationale for the development of a user-friendly resource, to support counting/controlling intake of protein from fruit and vegetables
2. Become familiar with the evidence that allows the protein from many fruits, and some vegetables, to not be counted/controlled within the prescription for natural protein
3. Understand the principles behind the development of the resource
4. Understand how the resource can be used to manage the diet for the treatment of PKU

## Transition to Adult PKU Clinic: Preliminary Findings

**Heather Bell, RD and Sandra Tavares MSW, RSW, Registered Dietitian and Social Worker with the Fred A. Litwin Family Centre in Genetic Medicine, University Health Network/Toronto General Hospital, Toronto, ON**

1. To increase awareness of patient-identified issues relating to transition from pediatric to adult based care



# PROGRAM THURSDAY

## May 9, 2019

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

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

*Saadet Andrews, MD, PhD, FCCMG, FRCPC, Biochemical Geneticist, The Hospital for Sick Children; Associate Professor, Division of Clinical and Metabolic Genetics, Department of Pediatrics, University of Toronto, Toronto, ON*

*Neal Sondheimer, MD, PhD, Associate Professor of Paediatrics and Molecular Genetics, The University of Toronto, Toronto, ON*

**16:15-18:00** Plenary Session I:  
Complex Neurometabolic Disorders:  
phenotypes to neuroimaging  
(Room: Willow)

*Session Chair: Saadet Andrews*

**16:15** Inborn Errors of Complex Lipid Metabolism  
and Remodelling: Focus on Neurological  
Presentations

*Jean-Marie Saudubray, MD Senior metabolic consultant  
Centre de Référence Neurométabolique Adulte, Groupe  
Hospitalier Pitié-Salpêtrière, Paris, France*

1. Summarize the complex lipid synthesis and remodeling pathways in cell membranes and roles in the nervous system
2. List inborn errors of metabolism that impair complex lipid metabolism in the nervous system
3. Recognize the prominent motor manifestations of these disorders : Spastic paraparesis, Movement disorders, Neuropathy, Myopathy.
4. Design a tentative diagnostic algorithm

### Discussion

**17:15** **Neuroimaging in Neurometabolic Disorders**  
*Susan Blaser, MD, FRCPC, Pediatric Neuroradiologist, The Hospital for Sick Children; Professor of Neuroradiology, University of Toronto, Toronto, ON*

1. Summarize van der Knaap and Valk's concept of Selective Vulnerability
2. Recognize that the vulnerability of white matter and grey matter evolves with brain maturation
3. Utilize the concepts of Selective Vulnerability and MRI pattern recognition to assess imaging studies of infants and children with possible inborn errors of metabolism

### Discussion

**18:00-19:30** WELCOME RECEPTION & EXHIBITS  
(Room: Birchwood Ballroom & Foyer)

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

May 10, 2019

# PROGRAM FRIDAY

**7:00-8:00** Registration Open & Breakfast (Provided)  
(Room: Willow)

**8:00-9:15** Plenary Session II:  
Treatable Neurometabolic Disorders  
(Room: Willow)  
*Session Chair: Neal Sondheimer*

**08:00** Creatine Deficiency Disorders: Present and Future  
*Andreas Schulze, MD, PhD, FRCPC, Professor Paediatrics and Biochemistry, University of Toronto; Head, Metabolic Genetics, Medical Director, Newborn Screening Program, Senior Associate Scientist, Research Institute, The Hospital for Sick Children, Toronto, ON*

1. Understand the function and metabolism of creatine
2. Recognize the clinical spectrum of creatine deficiency disorders
3. Utilize clinical observation, pathophysiological thinking and translational research
4. Summarize the present and future of interventions for patients with creatine deficiencies

## Discussion

**08:35** Pyridoxine-dependent epilepsies: phenotypes and treatment outcomes  
*Saadet Andrews, Saadet Andrews, MD, PhD, FCCMG, FRCPC, Biochemical Geneticist, The Hospital for Sick Children; Associate Professor, Division of Clinical and Metabolic Genetics, Department of Pediatrics, University of Toronto, Toronto, ON*

1. Describe clinical features of pyridoxine-dependent epilepsies
2. List current treatments of pyridoxine-dependent epilepsies
3. Discuss treatment outcomes of pyridoxine-dependent epilepsies

## Discussion

**9:15-9:45** Refreshment Break & Exhibitors (Birchwood Ballroom) & Posters (Pine)

**9:45-12:00** Plenary Session III:  
Organelles and Metabolic Disease  
(Room: Willow)  
*Session Chair: Andreas Schulze*

**09:45** Mitochondrial SUMOylation in Cancer and Metabolism  
*Heidi McBride, PhD, Montreal Neurological Institute, McGill University, Montreal, QC*

1. Learn how mitochondrial signaling platforms regulate glucose metabolism in liver
2. Explore a new mouse model of hepatocellular carcinoma
3. Obtain a broad view of the integration between mitochondrial dynamics/signaling and cell physiology

**10:45** Evaluating the role of Autophagy in Peroxisome Biogenesis Disorders  
*Peter Kim, PhD, Senior Scientist, Cell Biology, Hospital for Sick Children; Associate Professor, Department of Biochemistry, University of Toronto, Toronto, ON*

1. Explain the cellular mechanisms associated with peroxisome biogenesis disorder (PBD)
2. Describe the how autophagy leads to peroxisome loss some PBD
3. Discuss the use of autophagy inhibitors as potential treatment for PBD

**11:20** Zellweger Spectrum Disorder: Update and Challenges for Therapy  
*Nancy Braverman, MD, MS, FACMG, Associate Professor, Dept. of Human Genetics and Pediatrics, McGill University Dept. of Pediatrics, McGill University Health Center (MUHC), Scientist, Peroxisome Disease Program, Child Health and Human Development, Research Institute of the MUHC, Montreal, QC*

1. Prepare a diagnostic model for the different peroxisome disorders
2. Describe new contributions to peroxisome biology
3. Discuss approaches and challenges to therapies

## Discussion

**12:00-12:45** Lunch (provided)  
(Room: Willow)

**12:45-13:15** Dessert & Exhibits (provided)  
(Birchwood Ballroom)

# PROGRAM FRIDAY (continued)

May 10, 2019

**13:15-14:30** Plenary Session IV:  
Neurodevelopmental Outcomes and  
Neuropsychiatric Presentations  
(Room: Willow)  
*Session Chair: Saadet Andrews*

**13:15** **Neurodevelopmental Outcomes of  
Mucopolysaccharidosis Type I (MPS I)**  
*Michal Inbar-Feigenberg, MD, FCCMG, The Hospital for  
Sick Children, Toronto, ON*

1. Describe neurodevelopmental outcomes of patients with MPS type 1 post HSCT
2. Summarize the unique neurodevelopmental features of this population
3. Compare our findings with the ones described in the literature.

## Discussion

**13:50** **Neuropsychiatric manifestations of  
neurometabolic disorders**  
*Chantal Morel, MD, FRCPC, FCCMG, Clinical and  
Metabolic Geneticist, Fred A. Litwin Family Centre  
in Genetic Medicine, University Health Network and  
Mount Sinai Hospital; Assistant Professor, Department of  
Medicine, University of Toronto, Toronto, ON*

1. Recognize potential CNS manifestations of inborn errors of metabolism
2. Learn when to consider an inborn error of metabolism in a patient with neuropsychiatric symptoms
3. Be able to name groups of inborn errors of metabolism where neuropsychiatric presentations can occur, and what tests to arrange to assess for these

## Discussion

**14:30-15:00** Refreshment Break & Exhibitors (Birchwood Ballroom)  
& Posters (Pine)

**15:00-16:30** Plenary Session V:  
Selected Abstracts  
(Room: Willow)  
*Session Chair: Neal Sondheim*

**Platform Presentations (Selected Abstracts)**  
Six abstracts (15 minutes each)

**16:30-17:45** Poster Walk (Pine) & Light Refreshments  
& Exhibits (Birchwood Ballroom)

**18:45-23:00** Garrod Symposium Gala Dinner  
*Pioneer Award Presentation and Entertainment*  
(Location: The Hockey Hall of Fame)



## PROGRAM SATURDAY

**08:00–09:00** Breakfast (Provided)  
(Room: Willow)

**9:00–11:00** **Plenary Session VI:  
Mitochondrial Neurometabolics**  
(Room: Willow)  
*Session Chair: Lauren MacNeil*

**09:00** **Exercise and Nutraceutical Approached for  
the Treatment of Mitochondrial Cytopathies  
and Diseases Associated with Mitochondrial  
Dysfunction**

*Mark Tarnopolsky, MD, PhD, FRCP(C), Professor  
of Pediatrics and Medicine, CEO and CSO, Exerkine  
Corporation, Director of Neuromuscular and  
Neurometabolic Clinic, McMaster University Medical  
Center, Hamilton, ON*

1. Summarize the pathophysiological consequences of mitochondrial dysfunction.
2. Understand the theory behind mitochondrial therapies.
3. Describe the mechanism of action of mitochondrial therapies.
4. Review the mitochondrial cocktail studies.
5. Review the exercise intervention studies.
6. Understand the basis of novel therapies for mitochondrial disease.

**09:50** **Laboratory Diagnosis of Mitochondrial  
Diseases**

*Tony Rupar, PhD, FCCMG, Professor in the Depts.  
of Pathology & Laboratory Medicine, Pediatrics and  
Biochemistry at Western University and the London  
Health Sciences Centre in London Ontario. Head,  
Biochemical Genetics Laboratory*

1. Recognize the roles of biochemical, genetic and pathological testing
2. Describe the significance of heteroplasmy in diagnosing mitochondrial diseases
3. Plan a diagnostic strategy to diagnose mitochondrial disease

**10:25** **A small-molecule approach to heteroplasmy  
shifting**

*Neal Sondheimer, MD, PhD, Associate Professor of  
Paediatrics and Molecular Genetics, The University of  
Toronto, Toronto, ON*

1. Analyze the impact of heteroplasmy on the phenotype of mitochondrial diseases caused by mtDNA mutation
2. Critique various approaches to heteroplasmy shifting
3. Assess the impact of G-quadruplex formation on DNA replication

### Discussion

**11:00–11:30** Refreshment Break & Exhibitors (Birchwood Ballroom)  
& Posters (Pine)

**11:30–13:00** Garrod Association Membership Meeting  
(Room: Willow)

*Please see [www.garrod.ca](http://www.garrod.ca) for other meetings after the  
official program adjourns.*



# GARROD SYMPOSIUM 2019 COMMITTEES

## Garrod 2019 Symposium Presidents

**Saadet Andrews**, MD, PhD, FCCMG, FRCPC, Biochemical Geneticist, The Hospital for Sick Children; Associate Professor, Division of Clinical and Metabolic Genetics, Department of Pediatrics, University of Toronto, Toronto, ON

**Neal Sondheimer**, MD, PhD, Associate Professor of Paediatrics and Molecular Genetics, The University of Toronto, Toronto, ON

## Garrod 2019 Symposium Scientific Committee

**Saadet Andrews**, MD, PhD, FCCMG, FRCPC, Biochemical Geneticist, The Hospital for Sick Children; Associate Professor, Division of Clinical and Metabolic Genetics, Department of Pediatrics, University of Toronto, Toronto, ON

**Michal Inbar-Feigenberg**, MD, FCCM, Assistant Professor Division of Clinical and Metabolic Genetics, Department of Pediatrics, University of Toronto, The Hospital for Sick Children, Toronto, ON

**Jonathan Kronick**, MD, PhD, FRCPC, Professor Division of Clinical and Metabolic Genetics, Department of Pediatrics, University of Toronto, The Hospital for Sick Children, Toronto, ON

**Lauren MacNeil**, PhD, FCCMG, Metabolic Disease Laboratory, Department of Pediatric Laboratory Medicine, University of Toronto, The Hospital for Sick Children, Toronto, ON

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

**Neal Sondheimer**, MD, PhD, Associate Professor of Paediatrics and Molecular Genetics, The University of Toronto, Toronto, ON

## Garrod 2019 Symposium Organizer

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

## Garrod Association Executive Committee

**Pranesh Chakraborty**, President of Garrod Association, MD, FRCPC, FCCMG, Associate Professor, Pediatrics and Biochemistry/Microbiology/Immunology, University of Ottawa; Medical Director, Newborn Screening Ontario; Metabolic Physician, Department of Pediatrics, Children's Hospital of Eastern Ontario, Ottawa, ON

**Andreas Schulze**, Vice 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





# THE GARROD ASSOCIATION

REGISTRATION FORM | IN9580

THE GARROD ASSOCIATION

THE GARROD SYMPOSIUM 2019: TORONTO, ON | MAY 9 - 11, 2019

## 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 29, 2019	After March 29, 2019 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
Additional* Guest for Welcome Reception (Thursday)		<input type="checkbox"/> \$50
*The welcome reception is included for Garrod registrants.		
Gala Dinner at Hockey Hall of Fame (Friday)		
Regular Garrod Attendee		<input type="checkbox"/> \$80
Additional guest(s)		<input type="checkbox"/> \$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