



# GARROD SYMPOSIUM

Precision therapy and biomarkers in IMDs,  
from cellular models to clinical trials

MAY 8-10, 2025



**Thank You For Attending This In-Person CPD Conference**

## Target Audience

Physicians, Laboratorians,  
Researchers, Registered Nurses,  
Genetic Counsellors, Registered  
Dietitians, Clinical Specialists,  
Laboratory Technicians and Trainees.



**Health Sciences**  
Continuing Professional  
Development

## For Information

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Development Office (CPD)

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Visit the website [garrodsymposium.ca](http://garrodsymposium.ca)



# GARROD SYMPOSIUM

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## Thursday, May 8, 2025



Breakfast, Breaks, and Lunches - Great Hall SOUTH  
All plenary programming - Plenary Room - Great Hall NORTH

11:00 AM – 1:00 PM  
**Registration Open**

12:00 PM – 1:30 PM  
**Lunch**

12:20 PM – 12:25 PM  
**Welcome to the Symposium**  
*Gabriella Horvath, Conference Chair*

12:30 PM – 1:15 PM  
**Lightning Talks**

1:15 PM – 1:30 PM  
**Break**

**Session 1: Understanding Cellular Models in Preclinical Stages of Drug Design, Orphan Drug Development, and Precision Therapy in Rare Disorders**  
*Moderator: Anna Lehman*

1:30 PM – 2:10 PM  
**Precision Therapy in Rare Disorders: Arginase 1 Deficiency Treatment**  
*Speaker: Andreas Schulze*  
30-minute talk + 10-minute Q&A

2:10 PM – 2:50 PM  
**Biallelic Variants in BBOX1 Cause L-Carnitine Deficiency and Elevated  $\gamma$ -Butyrobetaine**  
*Speaker: Maja Tarailo-Graovac*  
30-minute talk + 10-minute Q&A

2:50 PM – 3:30 PM  
**Cellular Models in Precision Therapy in IMD (iPSC): Mouse Model and iPSCs in PDE Deficiency**  
*Speaker: Blair R. Leavitt*  
30-minute talk + 10-minute Q&A

3:30 PM – 4:00 PM  
**Afternoon Break | Visit Posterboards**

**Session 2: Biomarker Development for Clinical Trials – Limitations and Challenges**  
*Moderator: Graham Sinclair*

4:00 PM – 4:40 PM  
**Protein-Based Biomarkers: Emerging Blood Biomarkers of Neuronal Injury and Inflammation**  
*Speaker: Yara Alkhodair*  
30-minute talk + 10-minute Q&A

4:40 PM – 4:55 PM – **Abstract submission**  
**Optimization and Validation of the Kairos Amino Acid Kit For Plasma Amino Acid Monitoring in Inherited Metabolic Disorder Patients**  
*Speaker: Kiara Theron*  
15-minute talk

4:55 PM – 5:10 PM – **Abstract submission**  
**Mitochondrial Complex V Deficiency Due to a Homozygous Splice Variant in the ATP5PO Gene**  
*Speaker: Zainab Al Masseri*  
15-minute talk

5:10 PM – 5:25 PM – **Abstract submission**  
**A Rare Case of VARS-Related Neurodevelopmental Disorder With Dystonic Storm and Oculogyric Crisis**  
*Speaker: Sharmeen Nasir*  
15-minute talk

5:25 PM – 6:00 PM  
**In-Vivo Stable Isotope-Based Functional Biomarkers for Improving Nutritional Therapies in Inborn Errors of Metabolism**  
*Speakers: Rajavel Elango and Sylvia Stockler-Ipsiroglu*  
30-minute talk + 5-minute Q&A

6:00 PM – 8:00 PM  
**Welcome Reception and Poster Judging**  
Location: Foyer and Great Hall South

7:15 PM – 9:15 PM  
**Board of Directors Meeting (By invitation only)**  
Location: Room 2301



# GARROD SYMPOSIUM

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**Friday, May 9, 2025**



Breakfast, Breaks, and Lunches - Great Hall SOUTH  
All plenary programming - Plenary Room - Great Hall NORTH

7:30 AM – 8:30 AM  
**Breakfast**

8:30 AM – 9:30 AM  
**Scriver Keynote Lectureship: Biomarkers for Clinical Trials in Mitochondrial Disorders**  
*Speaker: Shamima Rahman*  
50-minute talk + 10-minute Q&A

**Session 3: Drivers of Change in Newborn Screening: Novel Therapies, New Screening Technologies, Patient Advocacy, New Biomarker Discovery**  
*Moderator: Bojana Rakic*

9:30 AM – 10:00 AM  
**Efforts Towards National Coordination of Newborn Screening Programs**  
*Speaker: Hilary Vallance*  
25-minute talk + 5-minute Q&A

10:00 AM – 10:30 AM  
**Biomarkers in Expanded Newborn Screening: Biomarker and Multiplex Enzyme Assay Development for LSD Newborn Screening**  
*Speaker: Michael Gelb*  
25-minute talk + 5-minute Q&A

10:30 AM – 11:00 AM  
**Morning Break | Visit Posterboards**

**Session 4: Biomarker Development (continued)**  
*Moderator: Gabriella Horvath*

11:00 AM – 11:45 AM  
**New Biomarkers in Leukodystrophies: Biomarker Development for Clinical Trials in Metabolic Leukodystrophies**  
*Speaker: Fanny Mochel (virtual)*  
30-minute talk + 10-minute Q&A

11:45 AM – 12:00 PM – **Abstract submission**  
**Impact of regular physiotherapy intervention on the function and quality of life of pediatric patients diagnosed with Mucopolysaccharidosis**  
*Jian Dealy*  
15-minute talk

12:00 PM – 12:15 PM – **Abstract submission**  
**Utilization of a <sup>13</sup>C-Glucose Breath Test to examine glucose metabolism in patients with metabolic myopathies after physiotherapy intervention**  
*Catherine Brunel-Guitton*  
15-minute talk

12:15 PM – 12:30 PM – **Abstract submission**  
**Transforming inherited metabolic disease care: key lessons from Ontario**  
*Syed A. Ahmed*  
15-minute talk

12:30 PM – 1:30 PM  
**Lunch | Visit Posterboards**

*Continued...*



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## Friday, May 9, 2025



Breakfast, Breaks, and Lunches - Great Hall SOUTH  
All plenary programming - Plenary Room - Great Hall NORTH

*Continued...*

### **Session 5: Artificial Intelligence in Precision Treatment in IMD**

Moderator: Gabriella Horvath

1:30 PM – 2:10 PM

#### **AI in Development of Clinical Severity Scores as Biomarkers for Precision Treatment**

Speaker: Xavier Vasquez (IBM Paris) (virtual)

30-minute talk + 10-minute Q&A

2:10 PM – 2:50 PM

#### **AI in Precision Diagnosis: AI in Clinical Diagnostics of IMD**

Speaker: Wyeth Wasserman

30-minute talk + 10-minute Q&A

2:50 PM – 3:05 PM

#### **Creatine Supplementation for CBS-related Homocystinuria – A Proof-of-Concept Study and N-of-1 Clinical Trial - An update**

Speaker: Deepa Subramonian

15-minutes talk

3:05 PM – 3:20 PM

**Afternoon Break | Visit Posterboards**

### **Session 6: Novel Clinical Trial Designs, Advances, and Failures (New Ways of Thinking)**

Moderator: Ramona Salvarinova

3:20 PM – 4:00 PM

#### **Clinical Trial Designs for Rare Diseases**

Speaker: Beth Potter

30-minute talk + 10-minute Q&A

4:00 PM – 4:40 PM

#### **National/International Registries, Big Databases: Using Registry Data and Natural History Studies, Databases to Have Control Group in Clinical Trials**

Speaker: Saadet Andrews

30-minute talk + 10-minute Q&A

4:40 PM – 5:20 PM

#### **Partnership Between Health Care Providers, Researchers, and Patient Advocacy Groups (CRDN Pillar 3)**

Speaker: Ian Stedman

30-minute talk + 10-minute Q&A

5:30 PM – 6:45 PM

#### **The Nitobe Gardens, UBC Campus**

Opened privately for Garrod Symposium Participants' walking pleasure

7:00 PM – 11:00 PM

#### **Awards Dinner**

Sage Restaurant

The University Centre, UBC, 6331 Crescent Rd.

Vancouver, BC V6T 1Z1



# GARROD SYMPOSIUM

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## Saturday, May 10, 2025



Breakfast, Breaks, and Lunches - Great Hall SOUTH  
All plenary programming - Plenary Room - Great Hall NORTH

7:30 AM – 8:30 AM  
**Breakfast**

### **Session 7: Financial, Ethical, Legal, and Social Issues**

Moderators: Hilary Vallance, Sandra Sirrs

9:00 AM – 9:45 AM  
**Health Technology Assessments, Pricing Negotiations, and Provincial Listing Decisions for Rare Disease Drugs in Canada: Challenges and Opportunities (Drug pricing negotiations, government policies, negotiations)**

Speaker: Trish Caetano

35-minute talk + 10-minute Q&A

9:45 AM – 10:30 AM  
**Planetary Health Is Medicine – Bringing the ABCs of the 21st Century (Air Purity, Biodiversity Preservation, Curbing Carbon Emissions) to Precision Medicine**

Speaker: Andre Mattman

35-minute talk + 10-minute Q&A

10:30 AM – 10:45 AM  
**Morning Coffee Break | Visit Posterboards**

10:45 AM – 12:00 PM  
**Debate and Discussion: Surrogate Endpoints as Markers for Drug Efficacy**

Presenters: Michael West and Shirin Rizzardo

Moderator: Sandra Sirrs

12:00 PM – 12:05 PM  
**Closing Remarks**

Speaker: Aziz Mhanni, President of Garrod Association

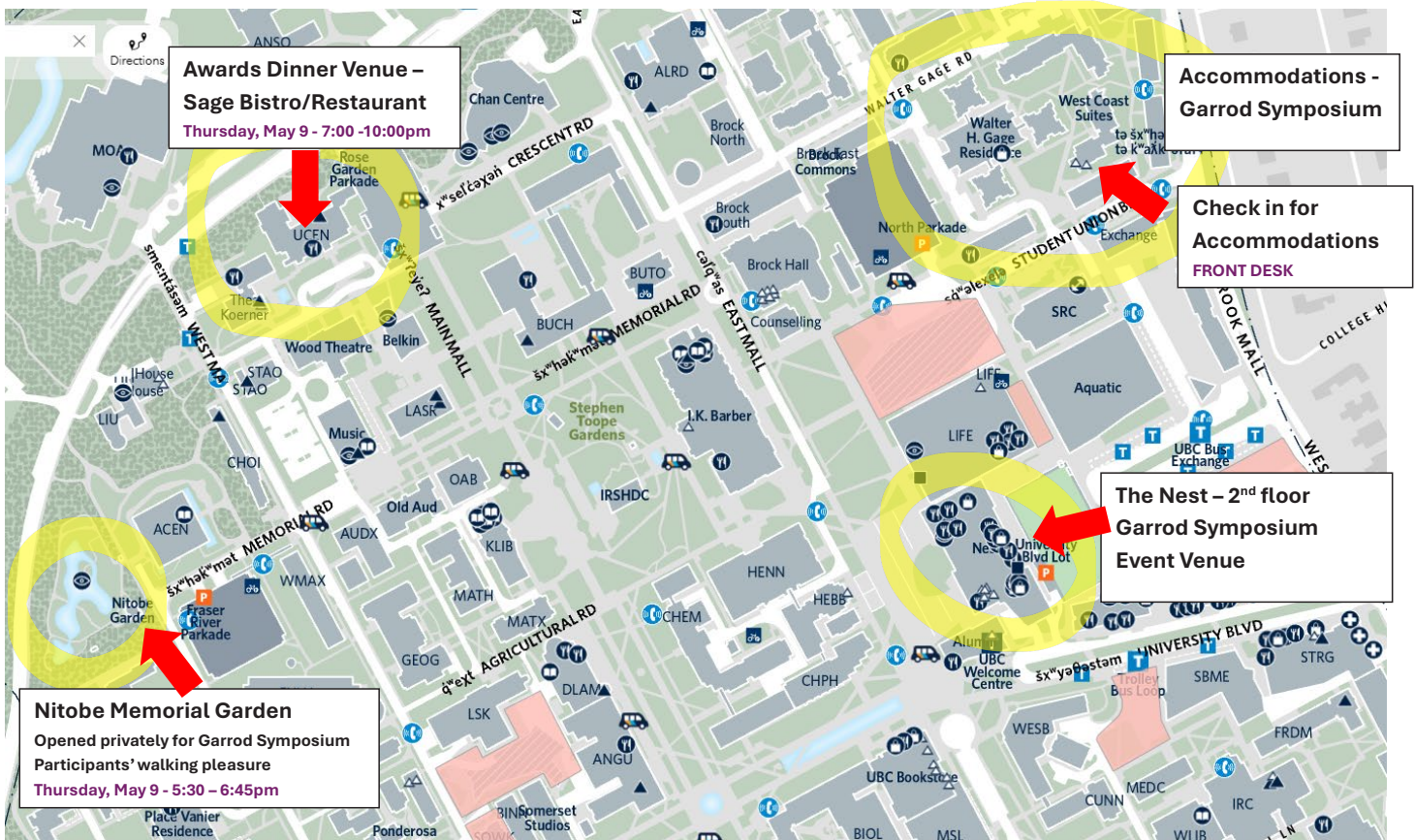
12:05 PM – 1:30 PM  
**Garrod Association AGM with Lunch**  
All Members Welcome

## Accreditation Statement

The Continuing Professional Development (CPD) Office at McMaster University is fully accredited by the Committee on Accreditation of Continuing Medical Education (CACME) to provide College of Family Physicians of Canada (CFPC) Mainpro+ and Royal College of Physicians and Surgeons of Canada (RCPSC) Maintenance of Certification (MOC) study credits for Continuing Medical Education.

This program is approved as an Accredited Group Learning Activity (Section 1) as defined by the MOC Program of the RCPSC. **A maximum of 13.5 MOC Section 1 hours has been approved.** Through an agreement between the RCPSC and the American Medical Association (AMA), physicians may convert Royal College MOC credits to AMA PRA Category 1 Credits™.

\*Each attendee should only claim the hours they attended.



<p><b>Front Desk for Accommodations</b>          UBC Conferences and Accommodations          5959 Student Union Blvd          Vancouver, BC V6T 1K2</p> <p><b>UBC Interactive map:</b>  <a href="https://www.maps.ubc.ca/">https://www.maps.ubc.ca/</a></p>	<p><b>Garrod Symposium Venue</b>          Great Hall at the University of British Columbia (UBC)          2nd floor, 6133 University Blvd,          Vancouver, BC, V6T 1Z1</p>	<p><b>Nitobe Memorial Gardens</b>          Thursday, May 9 - 5:30 – 6:45pm          Opened privately for Garrod Symposium          Participants' walking pleasure          1895 Lower Mall,          Vancouver, BC V6T 1Z4</p>	<p><b>Awards Dinner Venue – Sage Bistro/Restaurant</b>          Thursday, May 9 - 7:00 -10:00pm          The University Centre,          6331 Crescent Rd,          Vancouver, BC V6T 1Z1</p>
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### Program Disclaimers

**Confirmation of Registration**

A written acknowledgment of your registration will be sent prior to the activity. Receipts are emailed at the time of registration, and an additional email will be sent after the activity, confirming that your confirmation of attendance is ready to download or print.

**Liability**

Continuing Professional Development Office (CPD) hereby assumes no liability for any claims, personal injury, or damage:

- To any individual participating in this activity.
- That may result from the use of technologies, programs, products and services during this activity.
- That may arise out of or during this activity.

**Disclosure of Conflict of Interest**

Keeping with accreditation requirements and the National Standard for Support, McMaster University, Continuing Professional Development (CPD) requires all speakers, planning committee members, moderators, facilitators and authors participating in this activity to disclose all relationships with for-profit and not-for-profit organizations over the previous two years. The disclosure must be done in print, verbally, and in writing on a slide prior to the speaker's presentation.

**Activity Package**

As a registrant, your activity package includes the welcome reception (May 8), breakfast, lunch (May 9 and 10) and nutritional breaks (May 8, 9, and 10). Your activity lanyard must be worn at all times throughout the day.



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*This program has received an educational grant or in-kind support from*

## PLATINUM LEVEL



## GOLD LEVEL



## BRONZE LEVEL

