GARROD SYMPOSIUM

IEM: A FORTRESS TO CONQUER/ EIM: UNE FORTERESSE À CONQUÉRIR

MAY 4–6, 2023

Scan the QR code with your mobile camera to download the handouts and presentations, or visit garrodsymposium.ca/handouts

Password: Garrod2023

SYMPOSIUM HANDOUT

LOCATION

Meeting Space:
Quebec City Convention Centre
900, boul. René-Lévesque Est, suite 600
Québec (Québec) G1R 2B5

WIFI INFORMATION

Network: Garrod 2024
Password: Ottawa24

TARGET AUDIENCE

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

LEARNING OBJECTIVES

At the end of this activity, participants will be able to:

- Identify the challenges from newborn screening to diagnosis and follow-up of Inborn Errors of Metabolism.
- Discuss different practices across Canada in the continuum of care of patients affected by Inborn Errors of Metabolism.
- Recognize the challenges that patients and parents of young patients with an Inborn Error of Metabolism are facing in everyday life.
- Discuss progress and new approaches to improve the care of patients with Inborn Errors of Metabolism.

Visit the website www.garrodsymposium.ca

Thursday, May 4, 2023

12:00–1:00

Registration Open **(Hall 310)**

ALL SESSIONS ARE LOCATED IN ROOM 309 AB

12:45–1:00

Welcome to Symposium

Co-Chairs: Marie-Thérèse Berthier, PhD, and Yves Giguère, MD, PhD CHU de Québec-Uiversité Laval

SESSION 1: OPENING TALK

Moderator: Yves Giguère, CHU de Québec-Université Laval, Québec

1:00–1:45

Krabbe disease: A fortress with layers of moats, walls and secret passage

Keynote: Dietrich Matern, MD, PhD, Mayo Clinic, Minnesota

1:45–2:00

Question and Answer

2:00–2:48

Platform Presentation: Clinical Cases
Moderator: Dietrich Matern, MD, PhD

2:00–2:12

Expanding the phenotype of TUFM-related combined oxidative phosphorylation deficiency-4.

Noemie Villeneuve-Cloutier, University of Ottawa

2:12–2:24

When Fasting Turns Fatal: Eating disorders in patients with medium-chain acyl-CoA dehydrogenase deficiency

Mia Sethna, Western University, London

2:24–2:36

Autosomal dominant Fanconi syndrome due to a heterozygous pathogenic variant in GATM and review of the literature

Crystal Mulik, University of Alberta, Edmonton

2:36–2:48

The value of functional studies and whole genome sequencing in explaining the phenotype in of a patient with two rare genetic diseases

David Rosenblatt, McGill University, Montreal

2022 Garrod Association Trainee Research Award Recipient

2:48–3:00

Management of lipoprotein lipase deficiency with medium-chain triglycerides: a retrospective chart review

Liali Aljouda, The Hospital of Sick Children, Toronto

3:00–3:30

Break **(Hall 310 and Espace Urbain)**

SESSION 2: TYROSINEMIA TYPE 1: MORE THAN 50 YEARS OF EXPERIENCE

Moderator: Guy Parizeault, MD, PhD, Saguenay-Lac-St-Jean

3:30–4:00

Screening for Tyrosinemia type 1 in Québec: Historical perspective

Yves Giguère, MD, PhD, and Marie-Thérèse Berthier, PhD, CHU de Québec-Uiversité Laval

4:00–4:30

Tyrosinemia type I Diagnostics and therapeutics

Grant Mitchell, MD, CHU Ste-Justine, Montréal

4:30–4:45

Correction of hereditary tyrosinemia type I by in vivo genome editing in Fah-/- mice

Jean-Francois Rivest, Laval University, Quebec City

4:45–5:15

Family planning: co-creation of a decision support aid to empower couples carrying the tyrosinemia gene

Marie-Ève Poitras Inf, PhD and Janick Tremblay, Saguenay-Lac-St-Jean

5:15–5:50

Panel Question and Answer

6:00–8:00

Welcome Reception and Poster Judging **(Hall 310 and Espace Urbain)**

7:00–9:00

Board of Directors Meeting **(Room 307)**

Friday, May 5, 2023

8:00–8:30
Breakfast **(Room 308 AB)**

ALL SESSIONS LOCATED IN ROOM 309 AB

SESSION 3: IEMS IN CANADA UPDATE

Moderators: Paula Waters, CHUS–Sherbrooke, Daniela Buhas, McGill University, Montreal

8:30–9:45
Strategies from Coast to Coast: A lifetime journey
Alberta: Saadet Andrews and Lauren MacNeil
British Columbia: Bojana Rakic
Manitoba: Cheryl Rockman-Greenberg and Marie-Claude Dery

9:45–10:00
Morning Break **(Hall 310 and Espace Urbain)**

10:00–11:15
Strategies from Coast to Coast: A lifetime journey
Maritimes: Zaipling Liu and Sarah Dyack
Ontario: Kristin Kernohan and Pranesh Chakraborty
Quebec: Marie-Thérèse Berthier and Yves Giguère

11:15–12:00
Discussion Panel

12:00–12:45
Lunch **(Room 308 AB)**

12:45–1:30
Poster Judging

SESSION 4: WHAT'S NEW IN IEM?

Moderator: Nathalie Lepage, CHEO, Ottawa

1:30–3:00
Platform Presentations

1:30–1:45
Parent and healthcare personnel perspectives on challenges to family-centred care for children with inherited metabolic diseases: a qualitative analysis of interview data
Andrea Chow, University of Ottawa, Ottawa

1:45–2:00
Mass spectrometry assays for better management of patients with mucopolysaccharidoses
Pamela Lavoie, Université de Sherbrooke, Sherbrooke

2:00–2:15
Genetic landscape of primary mitochondrial disorders due to pathogenic variants in the mitochondrial and nuclear genome
Anastasia Ambrose, University of Alberta, Edmonton

2:15–2:30
Evaluating the "OMICS First" approach to the diagnosis of Inborn Errors of Metabolism: Preliminary analysis of outcome measures comparing next-generation sequencing to the traditional approach to diagnosis
James Elmore, University of Manitoba, Winnipeg

2:30–2:45
Validation and implementation of a multiplexed UPLC-MS/MS method for detecting six lysosomal storage disorders in dried blood spots with separation of interferences.
Joshua Dubland, University of British Columbia, Vancouver

2:45–3:00
Expression and function of guanidinoacetate methyltransferase (GAMT) is restored in cellular and murine models of GAMT creatine deficiency following treatment with scAAV9.hGAMT
Robyn Binsfeld, Queens University, Kingston

3:00–3:15
Tribute to Dr. Charles Scriver

3:15–3:30
Break **(Hall 310 and Espace Urbain)**

SESSION 5: HOW TO BEAT THE EVERYDAY WITH AN IEM

Moderators and Facilitators: Mehdi Yeganeh, CHU de Quebec, Quebec and Ivan Shelihan, MUHC, Montreal

3:30–5:00
Presenters/testimony and discussion with patients and parents of patients on their experience in the continuum of care

5:00–5:30
Question and Answers

7:30–11:30
Gala Awards Dinner Musée national des beaux-arts du Québec
(20 minute walk 1.5 km) Directions: bit.ly/garrod-gala-awards-23

7:30–8:30
Cocktail hour
During the cocktail hour you are welcome visit two exhibits:
Decorative Arts and Inuit Art for 1 hour

8:40–11:30
Dinner, Awards Announcements, and Music

Saturday, May 6, 2023

8:00–8:30
Breakfast **(Room 308 AB)**

ALL SESSIONS LOCATED IN ROOM 309 AB

SESSION 6: ALLIED HEALTH CONFERENCE: TREATMENT OF IEM

Moderator: Marie-Helene Bourdages, CHU de Quebec, Quebec

8:30–9:15
Glycerol phenylbutyrate in the treatment of urea cycle disorders with Testimony/exchange of a patient treated during pregnancy
Barbara Burton, MD, Lurie-Children's Hospital, Chicago

9:15–9:30
Question and Answer

9:30–10:15
Pegvaliase in the treatment of phenylketonuria with Testimony/exchange of a patient
John Mitchell, MD, McGill University, Montreal

10:15–10:30
Question and Answer

10:30–10:45
Break Hall **(310 and Espace Urbain)**

10:45–11:10
Inborn errors of acyl-CoA metabolism
Grant Mitchell, MD, CHU Ste-Justine, Montreal

11:10–11:20
Question and Answer

11:20–11:30
Closing Remarks
Paula Waters, President of Garrod Association

11:30–12:55
Garrod Association AGM
All Members Welcome

13:00
Lunch with a view at Hilton Quebec
Plaines room located 23rd floor

Contact

If you have any general enquiries about the conference, please contact:

Carol Stewart, CMP, Program Coordinator
McMaster University, Continuing Professional Development Office (CPD)
castewa@mcmaster.ca

Planning Committee

Yves Giguère, MD, PhD, FRCPC, Medical Biochemist – Co-Chair
Quebec Newborn Screening Program
Dept of Laboratory medicine
CHU de Québec-Uiversité Laval

Marie-Thérèse Berthier, PhD, DEPD, CSPQ, FCACB, Clinical Biochemist – Co-Chair
Quebec Newborn Screening Program
Dept of Laboratory medicine
CHU de Québec-Uiversité Laval

Julie Arsenau-Deslauriers, BSc
Nurse Clinician
Quebec Newborn Screening Program
CHU de Québec-Uiversité Laval

Marie-Hélène Bourdages, RN
Metabolic Nutritionst
Clinical Nutrition Service
Department of Multidisciplinary Services

Claudia Drolet, BSc
Nurse Clinician
Medical Genetic Service
Department of Pediatrics
CHU de Quebec-Université-Laval

Annie Durand, BSc
Nurse Clinician
Quebec Newborn Screening Program
CHU de Québec-Uiversité Laval

Rachel Laframboise, MD, CSPQ, FRCPC, Medical Geneticist
Medical Genetic Service
Department of Pediatrics
CHU de Quebec-Université-Laval

Diane Racine, BSc
Nurse Clinician
Medical Genetic Service
Department of Pediatrics
CHU de Quebec-Université-Laval

Ivan Shelihan, MD, FRCPC, Medical Geneticist
Medical Genetic Service
Department of Pediatrics
CHU de Quebec-Université-Laval
Mehdi Yeganeh, MD, PhD, FRCPC, FCCMG, Medical Geneticist
Medical Genetic Service
Department of Pediatrics
CHU de Quebec-Université-Laval

Mehdi Yeganeh, MD, PhD, FRCPC, FCCMG, Medical Geneticist
Medical Genetic Service
Department of Pediatrics
CHU de Quebec-Université-Laval

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

Garrod Association Executive Committee

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

Aziz Mhanni, MD, FRCPC, FCCMG, FACMG, PhD
Vice President of Garrod Association
Associate Professor Department of Pediatrics and Child Health
Department of Biochemistry and Medical genetics
College of Medicine
Faculty of Health Sciences
University of Manitoba, Winnipeg, MB

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

Speakers

Dietrich Matern, MD, PhD
Professor of Laboratory Medicine
Medical Genetics and Pediatrics
Co-Director of Biochemical Genetics Laboratory
Mayo Clinic, Rochester Minnesota

Yves Giguère, MD, PhD, FRCPC, Medical Biochemist
Quebec Newborn Screening Program
Dept of Laboratory medicine
CHU de Québec-Uiversité Laval

Marie-Thérèse Berthier, PhD, DEPD, CSPQ, FCACB, Clinical Biochemist
Quebec Newborn Screening Program
Dept of Laboratory medicine
CHU de Québec-Uiversité Laval

Grant Mitchell, MD
Professor PTG
Medical Genetics
Department of Pediatrics
Faculty of Medicine
CHU Saint-Justine, Montreal, QC

Jean-Francois Rivest,
Centre Hospitalier Universitaire de Québec Research Center and Faculty of Medicine
Laval University, Quebec City
Université Laval Cancer Research Centre, Québec, QC,

Marie-Eve Poitras, Inf. PhD
Assistant Professeure – Chercheure Junior 1 FRQ-S
Department of Family Medicine and emergency medicine
FMSS-Université de Sherbrooke
Campus Saguenay

Janick Tremblay
Saguenay-Lac St-Jean

Barbara Burton, MD
Professor of Pediatrics
Northwestern University Feinberg School of Medicine
Attending Physician in the Division of Genetics, Birth Defects and Metabolism
Ann and Robert H. Lurie Children's Hospital of Chicago

John Mitchell, MD
Associate Professor
Department of Medicine, McGill University

Moderators

Yves Giguère, MD, PhD, FRCPC, Medical Biochemist
Quebec Newborn Screening Program
Dept of Laboratory medicine
CHU de Québec-Uiversité Laval

Guy Parizeault, MD
Pediatrician
Saguenay- Lac-Saint-Jean

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

Daniela Buhas, MD, FRCPC, FCCMG
Assistant Professor
Department of Human Genetics
McGill University, Montreal

Nathalie Lepage, PhD, FCACB, FCCMG
Professor
Department of Pathology and Laboratory Medicine
University of Ottawa
Laboratory Head of the Inherited Metabolic Disease laboratory,
Division of Newborn Screening Ontario
CHEO Ottawa

Mehdi Yeganeh, MD, PhD, FRCPC, FCCMG, Medical Geneticist
Medical Genetic Service
Department of Pediatrics
CHU de Quebec-Université-Laval

Ivan Shelihan, MD, FRCPC, Medical Geneticist
Medical Genetic Service
Department of Pediatrics
CHU de Quebec-Université-Laval

Marie-Hélène Bourdages, RN
Metabolic Nutritionist
Clinical Nutrition Service
Department of Multidisciplinary Services

Declaration of Conflict of Interest

The following Planning Committee Members and Speakers did not declare a relationship with a FOR-PROFIT and/or NOT-FOR-PROFIT organization over the previous two years:

Julie Arseneau Deslauriers

Marie-Thérèse Berthier

Annie Durand

Yves Giguère

Rachel Laframboise

Aziz Mhanni

Ivan Shelihan

Janick Tremblay

Paula Waters

Mehdi Yeganeh

The following Planning Committee Members and Speakers have or had a relationship with a FOR-PROFIT and/or NOT-FOR-PROFIT organization over the previous two years:

Marie-Helene Bourdages
Has received honoraria from Vitaflo, Nestle, Nutricia, Abbott, Ultragenyx and Horizon Therapeutics.

Barbara Burton
Has received honoraria for speaking engagements and or consulting fee with Aeglea, Agios, Biomarin, Horizon, JCR Pharma, Jnana, Modema, Takeda and Ultragenyx. Speaker bureau with Biomarin, Horizon and Takeda. Clinical trial funding from Biomarin, Denali, JCR Pharma, Homology Medicines, Sangamo and Ultragenyx.

Claudia Drolet
Has received honoraria or in-kind support from Biomarin and Horizon Therapeutics.

Dietrich Matern
Is a Member of the Board of Directors for the American College of Medical Genetics and Genomics. Is a Chapter Author for UpToDate, Inc.

Grant Mitchell
Funding from a private source (M.A Imbeau).

John Mitchell
Honoraria received from Biomarin, Ultragenyx, RegenXbio, Takeda. Advisory board and with Biomarin, Takeda, Sanofi Genzyme and has received funded grants and research trials.

Marie-Ève Poitras
Groupe d'aide aux enfants tyrosinémiques du QuébecFunding received to create the patient decision-aid

Murray Potter
Advisory board/consultant with Ultragenyx Therapeutics and Advisory board member with Horizon Therapeutics.

Diane Racine
Has received honoraria or in-kind support from Biomarin and Horizon Therapeutics.

Jagdeep Walia
Has Licensed the patented technology is and Advisory board member with Taysha Gene Therapies Inc and received research grants from Taysha Gene Therapies Inc, and Azafaros.

General Information

WIFI

Network: Garrod 2024
Password: Ottawa24

PHOTOGRAPHY

Photos will be taken at this activity which may be used in future marketing materials. If you have questions, please visit the front desk.

LANYARDS/ACTIVITY PACKAGES

As the registrant your activity package includes breakfast, nutritional breaks, and lunch. Your activity lanyard must be worn at all times throughout the conference.

CERTIFICATE OF ATTENDANCE

An email will be sent after the activity, confirming your certificate of attendance is ready to download or print.

DISCLOSURE OF CONFLICTS OF INTEREST

In keeping with accreditation requirements and the National Standard for Support, McMaster University, Continuing Health Sciences Education Program requires that all speakers, planning committee members, moderators, facilitators and authors participating in this activity must disclose all relationships with for-profit and not-for-profit organizations over the previous two years. Disclosures must be done in print, verbally, and in writing on a slide prior to the speaker's presentation.

CONFERENCE EVALUATIONS

Your feedback is valuable! Please take a moment to complete the evaluation surveys for this CPD Conference.

Simply scan the QR code below with your mobile camera. Alternatively, you will be emailed a link to complete it after the event.

Day 1 May 4, 2023

Day 2 May 5, 2023

Day 3 May 6, 2023

GARROD SYMPOSIUM

IEM: A FORTRESS TO CONQUER/EIM: UNE FORTERESSE À CONQUÉRIR

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

GOLD LEVEL

BIOMARIN

HORIZON

sanofi

ultragenyx

SILVER LEVEL

Leadiant Biosciences

PTC THERAPEUTICS

RECORDATI RARE DISEASES GROUP

Focused on the Few

BRONZE LEVEL

Amicus Therapeutics

KYOWA KIRIN

Mēdunik Canada

Medunik USA

PerkinElmer For the Better

synlogic

Takeda

Waters