



GARROD ASSOCIATION Annual Symposium, 2008

May 2, 2008

Mandarin Ballroom: Metropolitan Hotel, 108 Chestnut St., Toronto

Time	Speaker	Topic
<i>07:30 - 08:00</i>	<i>Continental Breakfast/Registration</i>	<i>Mandarin Foyer</i>
08:00 – 08:10	Welcome and Introduction	Annette Feigenbaum
08:15 - 09:10	Keynote Speaker Dr. Bruce Barshop	Metabolomics for the Biochemical Geneticist
	Short presentations	Moderator: Julian Raiman
09:10 – 09:30	Floyd Snyder	Application of metabolomic principles to the analysis of inherited disorders of: cystinosis, urea cycle defects and adenine phosphoribosyltransferase deficiency
09:30-10:00	Cheryl Greenberg	International Guidelines for Glutaric Aciduria type 1 (GA)
<i>10:00 – 10:30</i>	<i>Break/Poster/exhibits</i>	<i>Mandarin Foyer</i>
	Short presentations	Moderator: Julian Raiman
10:30 – 10:45	Fatma Al-Jasmi	Computer-assisted teaching of MPS by patient management problems
10:45 – 11:00	Christiane Auray-Blais	Mass Urinary Screening Program in the province of Quebec: an overview
11:00 – 11:15	Stephen Goodman	The M405V mutation in GCDH can cause clinically typical GA1, false-negative newborn screens, normal glutaric acid, and variable 3-hydroxyglutaric acid in serum and urine.
11:15 – 12:00	Gustavo Maegawa Ellen Crushell	Chaperones in LSDs: Identification of Ambroxol as a Potential Enzyme Enhancement-Agent for Gaucher Disease Potential Pharmacological Chaperones I GM1 Gangliosidosis
<i>12:00 – 1:00</i>	<i>Lunch</i>	
	Introduction/Moderator	Andreas Schulze
1:15 – 2:10	Keynote Speaker Dr. Ertan Mayatepek	Metabolism and synthesis of leukotrienes – A new group of inborn errors of metabolism
2:10 – 2:25	Anne-Marie Lamhonwah	Expression Patterns of the Organic Cation/Carnitine Transporter Family in Adult Murine Heart: Implications for Cardiac Function
2:25 – 2:50	Michael West	The Canadian Fabry Disease Initiative: a new paradigm for drug evaluation in Canada
2:50 – 3:05	Christiane Auray-Blais	A multiplex urinary Gb3/creatinine analysis using tandem mass spectrometry for Fabry disease
<i>3:05 – 3:30</i>	<i>Break /Posters</i>	<i>Mandarin Foyer</i>
	Moderator	Lianna Kyriakopoulou
3:30 – 3:45	Mariya Kozenko	Abnormal Urine Organic Acid Pattern suggestive of Multiple Biochemical Disorders.
3:45 – 4:00	Chitra Prasad	Primary muscle disorder presenting as a leukodystrophy
4:00 – 4:15	Bruno Maranda	Citrin deficiency is a pan-ethnic disease with apparent founder mutations in the French Canadian population.
4:15 – 4:30	Mohammed Hussain	Efficacy of Sapropterin (6R-BH4) in subjects with phenylketonuria who have elevated phenylalanine levels

Posters: Mandarin Foyer - On display 08:00 – 5:00 pm Dinner at 6:30 for 7

Presenter	Topic
Ellen Crushell	Lebers Hereditary Optic Neuropathy mutation T14484C can cause Leigh – like syndrome.
Mary Maj	Thinking Green: A Scheme to Discover Stimulators of Cytochrome c Oxidase Activity
Fathiya Al-Murshedi	Elevated Propionylcarnitine on Newborn Screening and Vitamin B12 Levels
William Hanley	Finding the Nubile Woman with PKU
David Sinasac	Infantile acute renal failure with hyperuricemia: a newly recognized presenting feature of HPRT deficiency.
Curtis Oleschuk	Late Presentation of Hyperornithinaemia, hyperammonaemia, homocitrullinuria (HHH) syndrome with the F188Δ ORNT1 Mutation in an Adult

Dinner at 6:30 for 7pm Hart House University of Toronto



Garrod Association Annual Symposium, 2008

May 3, 2008

Rooms 1248, 1250 & Main Auditorium: First Floor Elm Wing Hospital for Sick Children, 555 University Ave., Toronto (Downtown)

Time	Speaker	Topic
<i>08:30 - 09:00</i>	<i>Continental Breakfast</i>	<i>Room 1248</i>
09:00 - 11:00	Garrod Association business Meeting-members	Main Auditorium
<i>11:00 - 11:30</i>	<i>Break</i>	<i>1248</i>
11:30 - 2:00	Canadian Urea Cycle Network Members - second Meeting	Room 1250