



GARROD ASSOCIATION ANNUAL SYMPOSIUM, 2009

Agenda

Friday, May 1, 2009

Time	Speaker	Topic
<i>08:00-09:00</i>	<i>Breakfast / Registration</i>	
09:00-09:05	Dr. Pranesh Chakraborty	Welcome and opening remarks
09:05-09:50	John Aitchison, Institute for Systems Biology	Systems Approaches to Understand Peroxisome Dynamics and Function
09:50-10:35	Nancy Braverman, McGill University	Challenges and Approaches to the Treatment of Peroxisome Assembly Diseases
<i>10:35-11:00</i>	<i>Coffee Break and Poster viewing</i>	
11:00-11:45	Gerald Raymond, Kennedy Krieger Institute	Adrenoleukodystrophy: An update on treatment and diagnosis
11:45-12:00	Questions for the invited speakers	
<i>12:00-1:00</i>	<i>Lunch</i>	
1:15-2:45	ORAL PRESENTATIONS SESSION 1 – Chair, Dr. Serge Melançon	
	Osama Y. Al-Dirbashi	Novel approach for quantification of fatty acid markers of peroxisomal disorders by LC-MS/MS
	Dione Ng	Universal Assay for Lysosomal Exoglycosidases and Target 4-Methylumbelliferyl Substrates Permit Future Development of a Mass Spectrometry Based Multiplex Assay
	Marie-Thérèse Berthier	Mass spectrometry advances in measurement of cerebrospinal fluid neurotransmitter metabolites.
	Caroline Barr	Fabry disease urinary Gb ₃ biomarker evaluation using tandem mass spectrometry in healthy infants from birth to 6 months
	Lorne Seargeant	Simplified ATP production assay for detection of mitochondrial disorders
	Fabienne Parente	A New Capillary Zone Electrophoresis Method for the Screening of Congenital Disorders of Glycosylation (CDG)
<i>2:45-3:25</i>	<i>Coffee Break and Poster viewing</i>	
3:30-5:00	ORAL PRESENTATIONS SESSION 2 – Chair, Dr. Grant Mitchell	
	Elizabeth Davidson	Raised uracil and thymine: the metabolic clinic and beyond
	Sylvia Stockler	Selective Screening for Pyridoxine Dependent Epilepsy (<i>Antiquitin</i> Deficiency)
	Cheryl Rockman-Greenberg	First Report of Promising Treatment of an Infant with Severe Hypophosphatasia with Bone-Targeted Human Recombinant Alkaline Phosphatase
	David Cole	Molecular diagnostics for hypercalcemic syndromes – Update and evolving guidelines
	S. Mercimek-Mahmutoglu	A female patient with a medically refractory epilepsy and severe X-linked creatine transporter (SLC6A8) deficiency: successful treatment with l-arginine and l-glycine supplementation
	Karen Sappleton	Psychosocial needs of parents of children with X-Linked Adrenoleukodystrophy
<i>6:30-</i>	<i>Dinner</i>	<i>Faculty Club, McGill University</i>



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Saturday, May 2, 2009

Time	Speaker	Topic
<i>08:00 -09:00</i>	<i>Breakfast / Registration</i>	
9:15-10:15	ORAL PRESENTATIONS SESSION 3 – Chair, Dr. Christiane Auray-Blais	
	Anne-Marie Lamhonwah	Upregulation of the Organic Cation/Carnitine Transporter Family in the Pregnant and Lactating Murine Mammary Gland and Implications for the Suckling Infant
	Natascia Anastasio	Spectrum of Mutations in <i>MMACHC</i> : Allelic Expression and Evidence for Genotype-Phenotype Correlations
	Isabelle R. Miousse	Localization of the gene responsible for the <i>cb1F</i> type of vitamin B ₁₂ inborn error of metabolism on chromosome 6 by microcell-mediated chromosome transfer.
	Zarazuela Zlokipli	<i>In vitro</i> fibroblast model to study pathophysiology of SCAD deficiency
<i>10:15-10:30</i>	<i>Coffee Break</i>	
10:30-11:00	Garrod Council Meeting	
11:15-12:00	Annual General Meeting	
12:00-	Adjournment	

Note:

- A booklet of presentation abstracts will be distributed at the meeting.