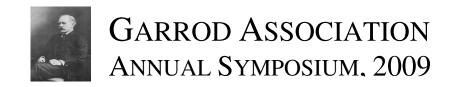


## Agenda

## Friday, May 1, 2009

Time	Speaker	Topic	
08:00-09:00	Breakfast / Registration	1	
09:00-09:05	Dr. Pranesh Chakraborty	Welcome and opening remarks	
09:05-09:50	John Aitchison, Institute for	Systems Approaches to Understand Peroxisome Dynamics	
	Systems Biology	and Function	
09:50-10:35	Nancy Braverman, McGill	Challenges and Approaches to the Treatment of Peroxisome	
	University	Assembly Diseases	
10:35-11:00	Coffee Break and Poster viewing		
11:00-11:45	Gerald Raymond, Kennedy	Adrenoleukodystrophy: An update on treatment and	
	Krieger Institute	diagnosis	
11:45-12:00	Questions for the invited speakers		
12:00-1:00	Lunch		
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	
	Marie-Thérèse Berthier	Assay	
	Marie-Therese Bertiner	Mass spectrometry advances in measurement of	
	Caroline Barr	cerebrospinal fluid neurotransmitter metabolites.  Fabry disease urinary Gb <sub>3</sub> biomarker evaluation using	
	Caronne Barr	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)	
2:45-3:25	Coffee Break and Poster viewing		
3:30-5:00	ORAL PRESENTATIONS SE	SSION 2 – Chair, Dr. Grant Mitchell	
	Elizabeth Davidson	Raised uracil and thymine: the metabolic clinic and beyond	
	Sylvia Stockler	Selective Screening for Pyridoxine Dependent Epilepsy (Antiquitin 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	
6:30-	Dinner	Faculty Club, McGill University	



## Saturday, May 2, 2009

Time	Speaker	Topic
08:00 -09:00	Breakfast / Registration	
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
	N	
	Natascia Anastasio	Spectrum of Mutations in <i>MMACHC</i> : Allelic Expression
	X 1 11 D 16	and Evidence for Genotype-Phenotype Correlations
	Isabelle R. Miousse	Localization of the gene responsible for the <i>cblF</i> type of
		vitamin B <sub>12</sub> 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
10:15-10:30	Coffee Break	
10:30-11:00	Garrod Council Meeting	
11:15-12:00	Annual General Meeting	
12:00-	Adjournement	

## Note:

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