PROGRAM AT A GLANCE

Thursday, May 29, 2014

SESSION I – ONTARIO NEWBORN SCREENING MEETINGS
* This session is restricted to keynote speakers and members of Ontario newborn screening treatment centres.

7:30-8:45  Registration & Breakfast (Drawing Room Foyer)

8:00-8:45  DERF Workshop for Nurses and Genetic Counsellors (Drawing Room)
           Moderator: Shelley Dougan, Genetic Counsellor, BORN Ontario

8:45-9:00  Welcome & Introductions (Drawing Room)
           Dr. Osama Aldirbashi, Laboratory Head, Newborn Screening Ontario, Chair of Scientific and Planning Committees

9:00-10:00 NSO Overview of 2013 (Drawing Room)
           Dr. Osama Aldirbashi, Laboratory Head, Newborn Screening Ontario
           Sari Zelenietz, Genetic Counsellor, Newborn Screening Ontario

10:00-10:30 Providing access to newborn screening results for health care providers via the Ontario Laboratories Information System (OLIS): A provincial eHealth solution (Drawing Room)
            Christine Davies, Genetic Counsellor, Newborn Screening Ontario
            Peter Wightman, Business Systems Analyst, Newborn Screening Ontario

10:30-10:45 Refreshment Break with Exhibits (Drawing Room Foyer)

10:45-12:30 Provincial Concurrent Disease Specific Workshops
            a) Hematology Workshop (Palladian Room) – Moderator: Christine Davies, Genetic Counsellor, Newborn Screening Ontario
            b) Metabolics Workshop (Renaissance Room)– Moderator: Janet Marcadier, Genetic Counsellor, Newborn Screening Ontario
            c) Cystic Fibrosis Workshop (Burgundy Room) – Moderator: Sari Zelenietz, Genetic Counsellor, Newborn Screening Ontario
            d) Endocrinology Workshop (Drawing Room) – Moderator: Thea Bouwman, RN, Children’s Hospital of Eastern Ontario, Newborn Screening Treatment Centre
            e) Immunology Workshop (Drawing Room) – Moderator: Lauren Higgins, Genetic Counsellor, Newborn Screening Ontario
SESSION II – NEWBORN SCREENING

12:30-1:30 Registration & Lunch (Drawing Room Foyer)

1:30-1:45 Welcome & Introductions (Drawing Room)
Dr. Osama Aldirbashi, Laboratory Head, Newborn Screening Ontario, Chair of Scientific and Planning Committees

1:45-2:30 What’s on the Horizon for Newborn Screening? (Drawing Room)
Dr. Michael Geraghty
Professor, Pediatrics, University of Ottawa
Head, Metabolics, Children’s Hospital Eastern Ontario

2:30-3:15 Platform Presentations (Drawing Room)

2:30-2:45 Newborn Screening Policy in Australia
Speaker: Susannah J Maxwell, Centre for Population Health Research, Faculty of Public Health, Curtin University, Perth, Western Australia

2:45-3:00 The Extended Nationwide Newborn Screening Program in the State of Qatar (in Cooperation with the Neonatal Screening Centre in University Children Hospital Heidelberg-Germany) 10 years outcome
Speaker: Ghassan Abdoh, Department of Pediatrics, Hamad Medical Corporation, Doha, Qatar

3:00-3:15 Exploring Challenges to Newborn Screening Programs in Libya
Speaker: Adel Zeglam, Pediatrics and Child Health, Al-Khadra Hospital, Tripoli, Libya

3:15-3:45 Refreshment Break with Exhibits (Drawing Room Foyer)

3:45-4:45 Platform Presentations (Drawing Room)

3:45-4:00 Glucose 6 Phosphate Dehydrogenase Deficiency: An Urgent Need for Newborn Screening in India
Speaker: Seema Kapoor, Division of Genetics, Department Of Pediatrics, Maulana Azad Medical College, New Delhi, India

4:00-4:15 Mass or High-Risk Screening of Creatine Synthesis and Transport Disorders, Triple H Syndrome and OTC Deficiency using a Multiplex Tandem Mass Spectrometry Methodology
Speaker: Christiane Auray-Blais, Université de Sherbrooke/CRC-CHUS, Sherbrooke, QC, Canada

4:15-4:30 Molecular, Biochemical and Clinical Heterogeneity in Very-long-chain acyl-CoA Dehydrogenase Deficiency in the Canadian Maritime Provinces
Friday, May 30, 2014

SESSION III – EXISTING SCREENING TARGETS: HOW ARE WE DOING?

7:30-9:00 Registration (Drawing Room Foyer)

7:30-8:30 Breakfast & Poster Set-Up (Canadian Room)

8:30-9:15 Existing Screening Targets: How are we Doing?
Dr. Murray Potter, Head – Biochemical Genetics Laboratory, McMaster Children’s Hospital
Janet Marcadier, Genetic Counsellor, Newborn Screening Ontario

9:15-10:00 Newborn Screening for Metabolic Disorders in Germany - Evaluation of Outcome and Impact on Families
Dr. Gwendolyn Gramer, University of Heidelberg, Centre for Paediatric and Adolescent Medicine

10:00-10:30 Refreshment Break with Exhibits (Drawing Room Foyer)

10:30-11:45 Platform Presentations (Drawing Room)

10:30-10:45 Outcome of Infants with Elevated C5OH Acylcarnitine: Seven Years of Newborn Screening Follow-up at the Calgary Metabolic Clinic
Speaker: Joanna Lazier, Department of Medical Genetics, University of Calgary, Calgary, AB, Canada

10:45-11:00 Biotinidase Deficiency: Spectrum of Molecular, Enzymatic and Clinical Information - A Pilot Project
Speaker: Chitra Prasad, Department of Paediatrics London Health Sciences Centre and Western University, London, ON, Canada
11:00-11:15  Newborn Population Screening for Classic Homocystinuria in Qatar
"7 years of success"
Speaker: Tawfeg Ben-Omran, Section of Clinical and Metabollic Genetics, Hamad Medical Corporation, Qatar

11:15-11:30  Five-Year Review of Newborn Screening for Congenital Hypothyroidism (CH) in Alberta
Speaker: Fiona Bamforth, Newborn Metabolic Screening / Biochemical Genetics Laboratory, University of Alberta Hospital, Edmonton, AB, Canada

11:30-11:45  Patient- and Family-Oriented Outcomes for Inborn Errors of Metabolism: A Scoping Review
Speaker: Sara Khangura, Canadian Inherited Metabolic Diseases Research Network, University of Ottawa, Ottawa, Ontario, Canada;

11:45-1:15  Lunch & Poster Viewing (Canadian Room)

SESSION IV – THE NARROWING GAP BETWEEN SCREENING & DIAGNOSIS

1:15-2:00  The Narrowing Gap Between Screening and Diagnosis (Drawing Room)
Dr. Pranesh Chakraborty, Director, Newborn Screening Ontario
Associate Metabolic Physician, Children’s Hospital of Eastern Ontario
Professor, University of Ottawa, Faculty of Medicine
Department of Pathology and Laboratory Medicine
Department of Biochemistry, Microbiology and Immunology

2:00-2:45  The Narrowing Gap Between Screening and Diagnosis: Next Generation Newborn Screening (Drawing Room)
Dr. Cynthia Powell, Division Chief, Pediatric Genetics and Metabolism, The University of North Carolina at Chapel Hill

2:45-3:15  Refreshment Break with Exhibits (Drawing Room Foyer)

3:15-4:15  Platform Presentations (Drawing Room)

3:15-3:30  The Development of a Genetic Newborn Screening Assay for Permanent Hearing Loss Using Blood Spots - A Collaboration Between Newborn Screening Ontario (NSO) and the Infant Hearing Program (IHP)
Speaker: Ed Yeh, Newborn Screening Ontario, Ottawa, ON, Canada

3:30-3:45  Rapid Second-Tier Testing for Newborn Screening and Therapeutic Monitoring of Maple Syrup Urine Disease
Speaker: Graham Sinclair, Biochemical Genetics and Newborn Screening Laboratories, Department of Pathology and Laboratory Medicine, BC Children's Hospital, Vancouver, BC, Canada
3:45-4:00 Mass Spectrometric Determination of Purine Metabolites in DBS; A Novel Second Tier Approach to Detecting Adenosine Deaminase Deficiency in the Screening for Severe Combined Immunodeficiency
Speaker: Nathan McIntosh, Newborn Screening Ontario, Ottawa, ON, Canada

4:00-4:15 Multiplexed Separations for Rapid Confirmatory Testing of Screen-positive Inborn Errors of Metabolism
Speaker: Alicia DiBattista, McMaster University, Hamilton, ON, Canada

4:15-4:45 Panel Discussion/Question Period (Drawing Room)
Dr. Potter, Dr. Gramer, Dr. Chakraborty, Dr. Powell

4:45 Closing Remarks (Drawing Room)
Dr. Osama Aldirbashi, Laboratory Head, Newborn Screening Ontario, Chair of Scientific and Planning Committees

6:00-10:00 Evening Gala (Canadian Museum of History)

Saturday May 31, 2014

SESSION V – STEM CELL & ORGAN TRANSPLANTATION FOR METABOLIC DISEASE

7:30-8:30 Registration (Drawing Room Foyer)

7:30-8:30 Breakfast & Poster Viewing (Canadian Room)

8:30-8:40 Introductions (Drawing Room)
Dr. Chitra Prasad, Chair/President, the Garrod Association

8:40-9:25 Hematopoietic Stem Cell Transplantation for Inherited Metabolic and Genetic Diseases (Drawing Room)
Dr. Adam Gassas, Associate Professor of Paediatrics, Division of Haematology/Oncology/Blood and Marrow Transplantation, the Hospital for Sick Children, University of Toronto

9:25-10:10 Liver Transplantation for Inborn Errors of Metabolism (Drawing Room)
Dr. Jerry Vockley, Professor of Pediatrics and Human Genetics, Chief of Medical Genetics, University of Pittsburgh School of Medicine and Graduate School of Public Health, Children’s Hospital of Pittsburgh

10:10-10:25 Refreshment Break with Exhibits (Drawing Room Foyer)

10:25-11:55 Platform Presentations (Drawing Room)
10:25-10:40 Liver Cell Transplantation as a “Bridge” Therapy for Urea Cycle Disorders: The Calgary Experience  
Speaker: Aneal Khan, Medical Genetics and Pediatrics, University of Calgary, Alberta Children’s Hospital, Calgary, AB, Canada

10:40-10:55 Towards a Clinical Trial of Lentivirus-Mediated Gene Therapy for Fabry Disease: In Vitro Patient Cell Mobilization and Transduction Outcomes  
Speaker: Tony Rupar, Western University, London, ON, Canada

10:55-11:10 Outcomes in 48 Individuals with Guanidinoacetatemethyltransferase (GAMT) Deficiency and Establishment of a Canadian Longitudinal Database  
Speaker: Sylvia Stockler, Division of Biochemical Diseases, BC Children’s Hospital, University of British Columbia, Vancouver, BC, Canada

11:10-11:25 The Importance of Challenging Phenylalanine (Phe) Tolerance in Patients with Phenylketonuria (PKU)  
Speakers: Julie Nedvidek, Hotel Dieu Hospital, Kingston, ON, Canada and Erica Langley, Department of Metabolics, Children’s Hospital of Eastern Ontario, Ottawa, ON, Canada

11:25-11:40 Metabolic Clinic Atlas: Organization of Care for Pediatric Metabolic Patients in Canada  
Speaker: Jonathan Kronick, Hospital for Sick Children/University of Toronto, Toronto, ON, Canada

11:40-11:55 Psychosis: Clinical Evaluation for Inborn Errors of Metabolism  
Speaker: Yannis Trakadis, Montreal Children’s Hospital-McGill University Health Centre, Montreal, QC, Canada

11:55-12:25 Panel Discussion/Question Period (Drawing Room)  
Dr. Gassas, Dr. Vockley

12:25-12:30 Closing Remarks & Awards (Drawing Room)  
Dr. Osama Aldirbashi, Laboratory Head, Newborn Screening Ontario, Chair of Scientific and Planning Committees

12:30-2:00 GARROD Annual General Meeting (Canadian Room)  
Lunch will be served  
*Restricted to current members of the GARROD Association