

Immunodeficiency Canada 7th sCID Symposium

October 24, 2019, 1:30-5:30 pm

Hotel Bonaventure, Montreal, Quebec

October 24, 2019

Time	Duration & Interactivity	Topic
1:30 pm	Moderators	Introduction & Welcome Chaim M. Roifman, CM, MD Richard Thompson, Immunodeficiency Canada
Chair:	<i>Julia Upton, MD, MPH</i>	<i>Hospital for Sick Children, Toronto, ON</i>
1:35 pm	25 min 5 min Q & A	Amit Nahum , MD, PhD Soroka University Medical Center, Beer-Sheva, Israel <i>Defining the biological responses of IL-6 by the study of a novel IL-6 receptor chain (IL6R) immunodeficiency</i>
2:05 pm	25 min 5 min Q & A	Oana Caluseriu , MD University of Alberta, Edmonton, Canada <i>A new form of SCID is underlined by NF-κB signalling abnormalities</i>
2:35 pm	25 min 5 min Q & A	Jolan Walter , MD, PhD John Hopkins All Children's Hospital, St. Petersburg, Florida, United States <i>Cytopenias of Many Kinds: An emerging face of Primary Immunodeficiencies</i>
Lightning Talks		
Chairs:	<i>Reza Alizadehfar, MD</i> <i>Jean Jacques de Bruycker, MD</i>	<i>McGill University, Montreal, QC</i> <i>CHU Sainte-Justine, Montreal, QC</i>
3:05 pm	10 min	Alanna Chomyn , Catherine M. Biggs, Wingfield Rehmus, Jacob Rozmus, Kirk R. Schultz, Stuart E. Turvey, Kyla J. Hildebrand. BC Children's Hospital and University of British Columbia, Vancouver, BC. <i>A case report of Cernunnos deficiency with an evolving phenotype, autoimmunity, and cutaneous granulomas</i>
3:15 pm	10 min	Keelia Farrell ¹ , Rae Brager ² , Thomas Issekutz ¹ , Beata Derfalvi ¹ . ¹ Dalhousie University and IWK Health Center, Halifax, NS; ² McMaster University, Hamilton, ON. <i>Variable spectrum of organ involvement in siblings with LRBA deficiency</i>
3:25 pm	10min	Lucy Duan , Chaim M. Roifman. The Hospital for Sick Children and University of Toronto, Toronto, ON. <i>Long-term PEG-ADA enzyme replacement therapy in a patient with partial adenosine deaminase deficiency</i>
3:35 pm	10 min	Question and Answer
3:45 pm	20 min	Break

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4:05 pm	10 min	Noreen Choe ¹ , Lauren Brick ¹ , Mariya Kozenko ¹ , Pranesh Chakroborty ² , Kristin Kernohan ² , Dennis Bulman ² , Rae Brager ¹ . ¹ McMaster Children's Hospital, Hamilton, ON; ² Newborn Screening Ontario, Ottawa, ON. <i>Positive newborn screen: A case of a novel mutation for Artemis deficiency</i>
4:15 pm	10 min	Ori Scott ¹ , Rae Brager ² , Peter Dent ² , Victoria Siu ³ , Donna Wall ¹ , Brenda Reid ¹ , Vy Hong-Diep Kim ¹ , Chaim M. Roifman ¹ . ¹ The Hospital for Sick Children and University of Toronto, Toronto, ON; ² McMaster University, Hamilton, ON; ³ London Health Sciences Centre, London, ON. <i>Survival of Patients with Severe Combined Immunodeficiency in the Newborn Screening Era: A Single-Centre Experience 2013-2019</i>
4:25 pm	10 min	Shama Sud , Thomas Eiwegger, Julia Upton, Chaim M. Roifman. The Hospital for Sick Children and University of Toronto, Toronto, ON. <i>Intermittent hypereosinophilia in a pediatric patient with X-linked hyper-IgM</i>
4:35 pm	10 min	Question and Answer
4:45 pm	10 min	Elliot James , Catherine M. Biggs, Stuart E. Turvey, Vishal Avinashi, Kyla J. Hildebrand. BC Children's Hospital and University of British Columbia, Vancouver, BC. <i>A case of chronic granulomatous disease due to a rare de novo mutation in the CYBB</i>
4:55 pm	10 min	Amarilla B. Mandola , Nigel Sharfe, Chaim M. Roifman. The Hospital for Sick Children and University of Toronto, Toronto, ON. <i>Novel genetic cause of autoimmune lymphoproliferative syndrome</i>
5:05 pm	10 min	Stephanie Erdle , Kyla J. Hildebrand, Stuart E. Turvey, Catherine M. Biggs. BC Children's Hospital and University of British Columbia, Vancouver, BC. <i>Novel BTK variant in a patient with bronchiectasis and B cell lymphopenia</i>
5:15 pm	10 min	Question and Answer
5:25 pm	Abstract Recognition	Abstract winners (presented by Chaim M. Roifman and Richard Thompson) Best abstract – 2 nd place best abstract – 3 rd place best abstract –
