Overall Learning Objectives

1. Recognize typical and atypical presentation of primary immunodeficiency in pediatric and adult cases
2. Understand the pathophysiology and underlying genetic aberrations leading to primary immunodeficiency
3. Review management options, challenges, and outcomes of patients with primary immunodeficiency

Immunodeficiency Canada 8th Annual SCID Symposium
October 22, 2020, 1:30-4:30 pm
Virtual Conference

Thursday October 22, 2020

Time	Duration & Interactivity	Topic
1:30 pm	Moderators	**Introduction & Welcome** Chaim M. Roifman, CM, MD, Hospital for Sick Children, Toronto, ON Linda Vong, PhD, Immunodeficiency Canada
1:35 pm	10 min	**Joshua Hochman**[1], Akash Gupta[2], Michelle Zeller[3], Rae Brager[1]. [1]Div. of Clinical Immunology & Allergy, [2]Dept. of Pathology and Molecular Medicine,[3]Div. of Hematology and Thromboembolism, Dept. of Medicine, McMaster University, Hamilton, ON. *Leukocyte adhesion deficiency type II: A rare case of primary immune deficiency with unique finding of para-Bombay phenotype* Learning Objectives: 1. Review leukocyte adhesions deficiency type 2 (LAD-2), including basic immunology, pathophysiology, and clinical manifestations 2. Describe the Bombay blood phenotype and why it is often found in LAD-2 3. Discuss clinical implications of a novel finding of para-Bombay phenotype in our LAD-2 patient
1:45 pm	10 min	**Ori Scott**[1], Amarilla Mandola[1,2], Yehonatan Pasternak[1,2], Yael Dinur-Schejter[1,2], Brenda Reid[1], Vy Hong-Diep Kim[1], Chaim Roifman[1,2]. [1]Div. of Immunology & Allergy, Dept. of Pediatrics, The Hospital for Sick Children and the University of Toronto, Toronto, ON. [2]Canadian Centre for Primary Immunodeficiency, Toronto, ON. *Heterozygous variants in FOXN1 in patients with abnormal newborn screen and T-lymphopenia: a single-centre experience* Learning Objectives: 1. Raise awareness of heterozygous FOXN1 mutations as an increasingly identified cause of lymphopenia and abnormal newborn screen 2. Outline the clinical and laboratory spectrum associated with heterozygous FOXN1 mutations 3. Highlight management challenges in patients with FOXN1 mutations
1:55 pm	10 min	**Nora Alrumayyan**[1], Sarah McAlpine[1], Thomas Issekutz[1], Drew Slauenwhite[1], Adam M Huber[2], Zaiping Liu[3], Beata Derfalvi[1]. [1]Div. of Immunology, [2]Div. of Rheumatology, Dept. of Pediatrics, [3]Div. of Clinical Biochemistry & Maritime Newborn Screening, Dept. of Pathology and Laboratory Medicine, Dalhousie University, IWK Health Centre, Halifax, NS. *Prolidase deficiency: description of its presentation and detailed immunological assessment* Learning Objectives; 1. To describe the clinical presentation and genetic mutations in two patients with prolidase deficiency. 2. To review the pathophysiology of prolidase deficiency as an inborn error of metabolism and immunity. 3. To present the results of an extended immunological assessment toward explaining the immune dysregulation in prolidase deficiency.
2:05 pm	**10 min**	**Question and Answer**

2:15 pm	10 min	**Lucy Dong Xuan Li**[1], Julia Upton[1]. [1]Div. of Immunology & Allergy, Dept. of Pediatrics, The Hospital for Sick Children, University of Toronto, Toronto, ON. *Novel mutation in the FLG gene and its relationship with severe atopic dermatitis: A case report* Learning Objectives: 1. To be able to explain the role of FLG on skin barrier function and its effect on atopic dermatitis 2. To be able to list primary immunodeficiencies associated with atopic dermatitis
2:25 pm	10 min	**Lisa Liang**[1], Marianne Miguel[1], Geoff Cuvelier[2], Tamar Rubin[1]. [1]Children's Hospital Winnipeg, Div. Clinical Immunology & Allergy, Dept. Pediatrics and Child Health [2] Manitoba Blood and Marrow Transplant Program, CancerCare Manitoba, Div. of Pediatric Hematology-Oncology-BMT, University of Manitoba, Winnipeg, MB. *Post-hematopoietic stem cell transplant infections in IKBKB deficiency* Learning Objectives: 1. Describe the underlying genetics and pathogenesis of IKBKB deficiency 2. Summarize the clinical presentation, management and outcome of known cases of IKBKB deficiency in Manitoba 3. Recognize the limitations of hematopoietic stem cell transplant as a treatment for IKBKB deficiency
2:35 pm	10 min	**Mei Xu**[1], Brenda Reid[1], Chaim M. Roifman[1,2]. [1]Div. of Immunology & Allergy, Dept. of Pediatrics, The Hospital for Sick Children and the University of Toronto, Toronto, ON. [2]Canadian Centre for Primary Immunodeficiency, Toronto, ON. *A case of RAG1 mutation presenting with disseminated vaccine-strain induced varicella and vasculitis in a previously well infant* Learning Objectives: 1. Understand the role of RAG1 2. Review a case of vaccine-strain varicella 3. Briefly discuss RAG1 mutations and their phenotypes
2:45 pm	**10 min**	**Question and Answer**

2:55pm	**20 min**	**Break**

3:15 pm	10 min	**Annam Bhatti**[1], Rae Brager[1], Andrew Latchman[2], Mariya Kozenkjo[3] [1]Dept. of Clinical Allergy and Immunology, [2]Dept. of Pediatrics, [3]Dept. of Genetics and Metabolics, McMaster University, Hamilton, ON. *Case Report: Delayed presentation of Ataxia-Telangiectasia* Learning Objectives: 1. To highlight an atypical presentation of PID 2. Understanding the importance of early identification of Ataxia Telangiectasia 3. The importance of raising interdisciplinary awareness of PID
3:25 pm	10 min	**Meriem Latrous**[1], Catherine M. Biggs[1,2], Stuart E. Turvey[1,2], Kyla J. Hildebrand[1,2] [1]Div. of Allergy & Immunology, Dept. of Paediatrics, BC Children's Hospital, The University of British Columbia, Vancouver, BC. [2]British Columbia Children's Hospital Research Institute, Vancouver, BC. *A case of failure to thrive, neutropenia, and rotavirus gastroenteritis in an infant* Learning Objectives; 1. Identify a differential diagnosis for neutropenia 2. Recognize Schwachman-Diamond Syndrome as a differential diagnosis for infants presenting with failure to thrive, neutropenia and diarrhea
3:35 pm	10 min	**Yehonatan Pasternak**[1,2], Amarilla Mandola[1,2], Ori Scott[1], Brenda Reid[1], Chaim M. Roifman[1,2]. [1]Div. of Immunology & Allergy, Dept. of Pediatrics, The Hospital for Sick Children and the University of Toronto, Toronto, ON. [2]Canadian Centre for Primary Immunodeficiency, Toronto, ON. *Not just SCID: cohort of non-SCID NBS positive infants with low TRECs* Learning Objectives: 1. Describe possible etiologies of non-SCID positive NBS 2. Understand the significance of long-term lymphopenia in the context of positive NBS
3:45 pm	10 min	**Erika Yue Lee**[1,2], Julia Upton[1,2,3], Michelle Sholzberg[1,4], Luke Reynolds[5], Rola Saleeb[1,6], Dory Abosh[1,4], Stephen Betschel[1,2]. [1]Dept of Medicine, University of Toronto, Toronto, ON. [2]Div. of Clinical Immunology & Allergy, Dept. of Medicine, St. Michael's Hospital, Toronto, ON. [3]Clinical Immunology & Allergy, Dept. of Pediatrics, Hospital for Sick Children, Toronto, ON. [4]Div. of Hematology, St. Michael's Hospital, Toronto, ON. [5]Section of Surgery, Dept. of Urology, University of Chicago, Chicago, Illinois, USA. [6]Dept. of Pathology, St. Michael's Hospital, Toronto, ON. *Non-infectious cystitis in an adult with STAT3 Gain-Of-Function (GOF) Mutation* Learning Objectives: 1. To recognize common presentations in patients with $STAT_3$ GOF mutation 2. To develop an approach to investigating and managing uncommon presentations in patients with $STAT_3$ GOF
3:55 pm	**10 min**	**Question and Answer**
4:10 pm	Abstract Recognition	Abstract winners (presented by Chaim M. Roifman) Best abstract – 2nd place best abstract – 3rd place best abstract –
		Closing Statements