Immunodeficiency Canada 12th Annual PID Symposium
October 16, 2025, 1.00 pm – 4.00 pm
Vancouver, British Columbia

This activity is an Accredited Group Learning Activity (Section 1) as defined by the Maintenance of Certification Program of the Royal College of Physicians and Surgeons of Canada and approved by Canadian Society of Allergy and Clinical Immunology.

Overall Learning Objectives

At the end of this session, participants will be able to:

1. Describe the pathophysiology and underlying genetic aberrations leading to inborn errors of immunity

2. Recognize the detection and outcomes of a spectrum of inborn errors of immunity in pediatric and adult cases

Scientific Planning Committee Disclosures

CMR is Board Chair of Immunodeficiency Canada and Chair of the Jeffrey Modell Foundation Network of centers. LMF reports Advisory Board activities for Encoded Therapeutics, Regenxbio, Merck, BioCryst, and Takeda; honorariums from Grifols and Valeo Pharma. BD reports Advisory Board activities for Takeda; payments from Takeda and Pharming. SA reports no conflicts of interest.

Moderators Disclosures

YDS reports grants/honorariums from CSL Behring and payments from Takeda. Other moderators report no conflicts of interest.

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Thursday October 16, 2025

Time	Duration & Interactivity	Topic
1:00 pm	2 min	**Introduction & Welcome** Linda Vong, PhD, Immunodeficiency Canada
		Plenary Session Moderator Kyla Hildebrand, MD, MScCH, FRCPC, University of British Columbia, Vancouver, BC
1.02 pm	20 min	**Tamar Rubin, MD, FRCPC**. University of Manitoba, Winnipeg, MB, Canada. ***Human RelB Deficiency*** Learning Objectives: 1. Describe the clinical features of reported patients with RelB deficiency 2. Outline the role of defective alternative NF-kB pathway activation and neutralizing autoantibodies against type I IFNs in infection susceptibility in human RelB deficiency
	7 min	Q&A
1.29 pm	20 min	**Catherine Biggs, MD, FRCPC**. BC Children's Hospital, University of British Columbia, Vancouver, BC, Canada. ***Inborn errors of immunity underlying severe immune dysregulation*** Learning Objectives: 1. Recognize the clinical features of recently described inborn errors of immunity associated with immune dysregulation 2. Appreciate the importance of precision medicine in diagnosing and managing inborn errors of immunity
	7 min	Q&A
1.56 pm	2 min	**Lightning Session Moderators** Yael Dinur-Schejter, MD, FRCPC, University of Alberta, Edmonton, AB Andrew Wong-Pack, MD, FRCPC, McMaster University, Hamilton, ON
1.58 pm	10 min	**Jason Z X Chen[1], Devyani Bakshi[2], George Cai[2], Jenny Garkaby[3].** [1]Department of Medicine, McMaster University, Hamilton, ON; [2]Division of Clinical Immunology and Allergy, Department of Medicine, McMaster University, Hamilton, ON; [3]Department of Pediatrics, Division of Rheumatology, Immunology and Allergy, McMaster University, Hamilton, ON, Canada. ***"Not SCID Enough": An IL2RG variant leading to a CVID-like phenotype*** Learning Objectives: 1. Review the role of IL2RG in immune function 2. Explore the pathophysiology of hypomorphic IL2RG variants 3. Discuss an atypical case of an IL2RG VUS
	3 min	Q&A

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2.11 pm	10 min	**Kristine Jeganathan**[1], Simon Hotte[2], NSO/Vienna/CHEO collaborators, Gabrielle White[3], Anne Pham-Huy[3].[1]1Department of Pediatrics, Children's Hospital of Eastern Ontario, University of Ottawa, Ottawa, ON; [2]Division of Allergy and Immunology, Montreal Children's Hospital, McGill University Health Center, Montreal, QC; [3]Division of Allergy, Immunology and Infectious Diseases, Children's Hospital of Eastern Ontario, University of Ottawa, Ottawa, ON, Canada. ***Novel IL6ST Splice-Site Variant Expands the Clinical Spectrum of gp130 Deficiency: Report of Two Siblings*** Learning Objectives: 1. Describe the clinical, immunologic, and genetic features observed in patients with IL6ST splice-site variants 2. Evaluate how these findings expand the recognized clinical spectrum of GP130 deficiency and their implications for diagnosis and patient management 3. Compare and contrast the clinical phenotypes of two siblings carrying the same IL6ST splice-site mutation
	3 min	Q&A
2.24 pm	15 min	**Break**
2.39 pm	10 min	**Benjamin Martinez[1], Alina Pace[2]**, Jenny Garkaby[3], Rae Brager[3].[1]Department of Paediatrics, The Hospital for Sick Children, Toronto, ON; [2]Michael G. DeGroote School of Medicine, McMaster University, Hamilton, ON; [3]Division of Immunology, Allergy, and Dermatology, Department of Pediatrics, McMaster Children's Hospital, McMaster University, Hamilton, ON, Canada. ***Novel BCOR variant and humoral immunodeficiency: A case report*** Learning Objectives: 1. Recognize the clinical features of and initial approach to humoral immunodeficiency 2. Understand the role of BCOR in immune system function and its potential link to humoral immunodeficiency 3. appreciate the implications of investigating and reporting on immune manifestations of rare syndromic disorders
	3 min	Q&A
2.52 pm	10 min	**Maha AlZubedy[1]**, Parni Nijhawan[1], Linda Vong[1], Chaim M. Roifman[1]. [1]Division of Immunology & Allergy, Department of Pediatrics, Hospital for Sick Children and University of Toronto, Toronto, ON, Canada. ***Long-term allogeneic hematopoietic stem cell transplantation outcomes in RelB deficiency*** Learning Objectives: 1. Understand the diversity in clinical presentation of disorders involving the NF-κB signaling pathway, with a focus on RelB deficiency.

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		2. Recognize the clinical variability and challenges in HSCT outcomes for patients with RelB deficiency. 3. Appreciate the importance of early diagnosis, timely HSCT, and long-term follow-up in improving survival and quality of life for patients with RelB deficiency.
	3 min	Q&A
3.05 pm	10 min	**Vivian Chen**[1], Andrew Wong-Pack[2], Rae Brager[3], Jenny Garkaby[3].[1]Department of Pediatrics, McMaster University, Hamilton, ON; [2]Department of Medicine, Division of Clinical Immunology and Allergy, McMaster University, Hamilton, ON; [3]Department of Pediatrics, Division of Immunology, Allergy, and Dermatology, McMaster University, Hamilton, ON. ***A heterozygous NPAT variant in a patient with relapsed Hodgkin's lymphoma and persistent hypogammaglobulinemia: A case report and review of the literature*** Learning Objectives: 1. Recognize the association between inborn errors of immunity (IEI) and increased malignancy risk, particularly lymphoma, in pediatric patients. 2. Describe the potential role of novel genetic variants, such as those in the NPAT gene, in predisposing individuals to immune dysfunction and cancer. 3. Discuss the clinical approach to evaluating persistent immune abnormalities following lymphoma treatment, including the role of genetic testing in identifying underlying IEIs or cancer predisposition syndromes.
	3 min	Q&A
3.18 pm	10 min	**Candice Luo**[1], Owsley E[2], Akeno N[2], Cobb C[2], Yang L[2], Marsh RA[2,3], Chiang, SCCC[2,3,¥], Tamar Rubin[4,5,¥].[1]Division of Adult Clinical Immunology and Allergy, Department of Medicine, University of Manitoba, Winnipeg, MB, Canada; [2]Division of Bone Marrow Transplantation and Immune Deficiency, Cincinnati Children's Hospital Medical Center, Cincinnati, OH, United States; [3]Department of Pediatrics, University of Cincinnati, Cincinnati, OH, United States; [4]Division of Pediatric Clinical Immunology and Allergy, Children's Hospital Winnipeg, Winnipeg, MB, Canada; [5]Department of Pediatrics and Child Health, University of Manitoba, Winnipeg, MB, Canada; [¥]Contributed equally ***Immune Deficiency and Dysregulation in a Patient with a Heterozygous IRF8 Variant: A Case Report*** Learning Objectives: 1. Describe a de novo IRF8 variant presenting pathogenically unique in a young child.
	3 min	Q&A

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3.31 am	10 min	**Jacqueline Mincer**[1], Jessica Gu[2], Stacey Hume[3], Jason Ohayon[2,4], Jenny Garkaby[5], Rae Brager[5]. [1]Department of Pediatrics, McMaster Children's Hospital, McMaster University, Hamilton, ON; [2]Michael G. DeGroote School of Medicine, McMaster University, Hamilton, ON; [3]Children's Hospital of Eastern Ontario Research Institute; [4]Hamilton Allergy, Hamilton, ON; [5]Division of Immunology, Allergy, and Dermatology, Department of Pediatrics, McMaster Children's Hospital, McMaster University, Hamilton, ON, Canada. ***Identification of a Novel BTK Variant Associated with X-Linked Agammaglobulinemia: A Case Report*** Learning Objectives: 1. Recognize key clinical "red flags" in a child with recurrent infections that should prompt consideration of XLA. 2. Discuss the implications of identifying novel BTK variants, with emphasis on the role of genetic databases and variant reporting. 3. Explain how this case illustrates the importance of early recognition and timely initiation of therapy in improving outcomes for children with XLA.
	3 min	Q&A
3.44 pm	10 min	**Jason Z X Chen**[1], George Cai[2], Andrew Wong-Pack[2], Jenny Garkaby[3], Rae Brager[3] . [1]Department of Medicine, McMaster University, Hamilton, ON; [2]Division of Clinical Immunology and Allergy, Department of Medicine, McMaster University, Hamilton, ON; [3]Department of Pediatrics, Division of Rheumatology, Immunology and Allergy, McMaster University, Hamilton, ON, Canada. ***Atypical X-linked lymphoproliferative syndrome type 2 presenting with polysaccharide antibody deficiency and recurrent infections in an EBV-naïve adult survivor of infantile Langerhans cell histiocytosis and childhood splenectomy*** Learning Objectives: 1. Review the pathophysiology of XLP-2 2. Explore the phenotypic range of XLP-2 variants 3. Discuss a rare variant in XIAP leading to atypical XLP-2 presentation
	3 min	Q&A
3.57 pm	3 min	**Abstract Winners** Best abstract – 2nd place best abstract – 3rd place best abstract –