# Immunodeficiency Canada 11th Annual PID Symposium
# November 7th, 2024, 1:00-4:00pm
# Banff, Alberta

This program was co-developed with Immunodeficiency Canada and was planned to achieve scientific integrity, objectivity and balance. 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, Chair of the Jeffrey Modell Foundation Network of Centers, and Chair of DSMB Kedrion KB070 study. BD reports consulting activities for Takeda and Pharming. Other members report no conflicts of interest.

Moderators Disclosures
No conflicts of interest to report.

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**Thursday November 7th, 2024**

| Time | Duration & Interactivity | Topic |
|---|---|---|
| 1:00 pm | 2 min | **Introduction & Welcome**<br>Linda Vong, PhD, Immunodeficiency Canada |
| | | **Lightning Session Moderators**<br>Yael Dinur Schejter, MD, Assistant Professor, University of Alberta, Edmonton, AB<br>Jenny Garkaby, MD, Assistant Professor, McMaster University, Hamilton, ON |
| 1:02 pm | 9 min | **Kristine Jeganathan**[1], Adam Byrne[2], Gabrielle White[2], Elie Haddad[3], Anne Pham-Huy[2]. [1]Department of Pediatrics, Children's Hospital of Eastern Ontario, University of Ottawa, Ottawa, ON. [2]Division of Allergy, Immunology and Infectious Diseases, Children's Hospital of Eastern Ontario, University of Ottawa, Ottawa, ON. [3]Department of Pediatrics, Department of Microbiology, Immunology and Infectious Diseases, University of Montreal, CHU Sainte-Justine, Montreal, QC.<br>***Standard newborn screening does not identify all cases of ADA deficiency. Is this avoidable?***<br>Learning Objectives:<br>1. Recognize the limitations of current newborn screening methods in detecting delayed-onset ADA.<br>2. Describe the clinical outcomes of delayed diagnosis of ADA deficiency and discuss strategies for improving early detection through expanded newborn screening approaches.<br>3. Introduce the potential advantages and drawbacks of incorporating purine profiling by tandem mass spectrometry (TMS) in first-tier SCID newborn screening to enhance the detection of ADA deficiency. |
| | 3 min | Q&A |
| 1:14 pm | 9 min | **Arun Govindapillai**[1], Sneha Suresh[2,3], Minakshi Taparia[4], Malcolm Wells[5], Jean Jacques De Bruycker[6], Adil Adatia[2].[1]Department of Pediatrics, Stollery Children's Hospital, University of Alberta, Edmonton, AB. [2]Division of Pulmonary Medicine, University of Alberta, Edmonton, AB. [3]Division of Pediatric Infectious Disease, Stollery Children's Hospital, University of Alberta, Edmonton, AB. [4]Division of Hematology, University of Alberta, Edmonton, AB. [5]Division of Gastroenterology, University of Alberta, Edmonton, AB. [6]Division of Pediatric Immunology and Rheumatology, Centre Hospitalier Universitaire Sainte-Justine, Department of Pediatrics, University of Montreal, Montreal, QC.<br>***Aggressive systemic mastocytosis and hepatic veno-occlusive disease in a patient with IKAROS deficiency***<br>Learning Objectives:<br>1. Characterize the immunological phenotype associated with a dominant negative mutation in IKAROS (IKZF1).<br>2. Recognize the clinical significance of aggressive systemic mastocytosis in IKAROS (IKZF1) deficiency in a dominant negative mutation. |
| | 3 min | Q&A |

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| 1:26 pm | 9 min | **Erin Joy Heifetz**[1], Anahita Dehmoobad Sharifabadi[2], Rae Brager[2], Jenny Garkaby[2]. [1]Department of Medicine, McMaster University, Hamilton, ON. [2]Department of Pediatrics, McMaster University, Hamilton, ON.<br><br>***Novel variant in IGMH gene in a patient with agammaglobulinemia: A case report of a preschool child presenting with recurrent pneumonia.***<br><br>Learning Objectives:<br>1. Discuss differential and workup for atypically presenting immunodeficiency |
| | 3 min | Q&A |
| 1:38 pm | 9 min | **Payam Salimi**[1], Patrick Frosk[2], Tom Le Voyer[3,4,5], Jean-Laurent Casanova[3,4,6,7,8], Anne Puel[3,4,6], Tamar Rubin[9]. [1]Department of Internal Medicine, University of Manitoba, Winnipeg, MB. [2]Section of Genetics and Metabolism, Department of Pediatrics & Child Health, University of Manitoba, Winnipeg, MB. [3]Laboratory of Human Genetics of Infectious Diseases, Necker Branch, INSERM UMR 1163, Paris, France. [4]Paris Cité University, Imagine Institute, Paris, France. [5]Clinical Immunology Department, Assistance Publique Hôpitaux de Paris (AP-HP), Saint Louis Hospital, Paris, France. [6]St. Giles Laboratory of Human Genetics of Infectious Diseases, Rockefeller Branch, Rockefeller University, New York, NY, USA. [7]Pediatric Hematology-Immunology Unit, Necker Hospital for Sick Children, Paris, France. [8]Howard Hughes Medical Institute, New York, NY, USA. [9]Section of Clinical Immunology and Allergy, Department of Pediatrics & Child Health, University of Manitoba, Winnipeg, MB.<br><br>***From clinical complexity to genetic clarity: A collaborative three-decade odyssey to diagnose inherited RelB deficiency in a patient with combined immunodeficiency syndrome***<br><br>Learning Objectives:<br>1. Identify clinical and immunological manifestations of inherited RelB deficiency.<br>2. Understand the impact of collaborative research between clinicians and researchers in diagnosing rare inborn errors of immunity. |
| | 3 min | Q&A |
| 1:50 pm | 9 min | **Abrar Al-Ahmadi**[1], Fatemah AlYaqout[1], Noha Benharira[2], Adnan Ali[1], Christian Sirois[3], Christos Tsoukas[1]. [1]Division of Clinical Immunology and Allergy, Department of Medicine, McGill University Health Center, Montreal, QC. [2]Faculty of Medicine and Health Sciences, McGill University, Montreal, QC. [3]Division of Thoracic and Upper Gastrointestinal Surgery , McGill University Health Centre, Montreal, QC.<br><br>***Piezo-1 gene mutations and immune dysregulation: A case report***<br><br>Learning Objectives:<br>1. Assess the impact of PIEZO1 gene mutations on lymphatic system function and related disorders.<br>2. Identify clinical manifestations and outcomes associated with PIEZO1 mutations.<br>3. Evaluate diagnostic techniques for detecting PIEZO1 mutations in clinical practice.<br>4. Explore the influence of PIEZO1 mutations on immune responses, and consider future research approaches to better understand this relationship and develop targeted interventions. |
| | 3 min | Q&A |

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| 2:02 pm | 9 min | **Tess Robart[1],** Amro Alamro[1], Trinda Hayden[2], Thomas Issekutz[3], Alejandro Palma[3], Beata Derfalvi[3]. [1]Pediatric Clinical Immunology and Allergy, Department of Pediatrics, Dalhousie University, Halifax, NS. [2]Department of Pediatrics, Dalhousie University, Fredericton, NB. [3]Division of Immunology, Department of Pediatrics, Dalhousie University, IWK Health, Halifax, NS.<br><br>***Hyper-IgE syndrome with normal IgE level: A pediatric case of recurrent and severe infection caused by DOCK8 deficiency***<br><br>Learning Objectives:<br>1. Describe an approach and differential diagnosis for suspected combined immunodeficiency<br>2. Highlight diagnostic challenges for patients with Hyper-IgE syndrome<br>3. Describe clinical manifestation, immunologic profiles and immunophenotypes of autosomal recessive DOCK8 deficiency |
| | 3 min | Q&A |
| 2.14 pm | 15 min | **Break** |
| 2:29 pm | 9 min | **Tatiana Kalashnikova[1]**, Greg Guilcher[1], Victor Lewis[1], Ashish Marwaha[1], Luis Murguia-Favela[1], Tamar Rubin[2], Sneha Suresh[3], Brenda Turley[1], Jennifer Leiding[4], Lauri Burroughs[5], Linda M. Griffith[6], Luigi Notarangelo[7], Michael A. Pulsipher[8], Morton J. Cowan[9], Rebecca Marsh[10], Sung-Yun Pai[11], Troy Torgerson[12], Jennifer Puck[9], Christopher Dvorak[9], Elie Haddad[13], Donald B. Kohn[14], Geoff Cuvelier[1†], Nicola AM Wright[1†]. [1]Dept of Pediatrics, University of Calgary, AB. [2]Dept of Clinical Immunology and Allergy, University of Manitoba, Winnipeg, MB.[3]Dept of Pediatrics, University of Alberta, Edmonton, AB. [4]Blood and Marrow Transplant, John Hopkins All Children's Hospital, St. Petersburg, FL. [5]Fred Hutchinson Cancer Research Center, Seattle, WA. [6]Division of Allergy, Immunology, and Transplantation, National Institute of Allergy and Infectious Diseases, National Institutes of Health, Bethesda, MD. [7]Chief Laboratory of Clinical Immunology and Microbiology/National Institutes of Health, National Institute of Allergy and Infectious Diseases. [8]Division of Pediatric Hematology/Oncology, Intermountain Primary Children's Hospital, Huntsman Cancer Institute, Spencer Fox Eccles School of Medicine at the University of Utah. [9]Pediatric Allergy, Immunology, and Blood and Marrow Transplant Division, University of California, San Francisco Benioff Children's Hospital, San Francisco, CA.[10]Division of Bone Marrow Transplantation and Immune Deficiency, Cancer and Blood Diseases Institute, Cincinnati Children's Hospital, Cincinnati, OH. [11]Immune Deficiency Cellular Therapy Program, Center for Cancer Research, National Cancer Institute, Bethesda, MD, USA. [12]Director of Experimental Immunology, Allen Institute for Immunology. [13]Pediatric Immunology and Rheumatology Division, CHU Sainte-Justine, University of Montreal, Montreal, QC. [14]Dept of Microbiology, Immunology & Molecular Genetics, University of California, Los Angeles. [†]Authors contributed equally.<br><br>***CD3δ SCID patients in Canada: Baseline characteristics and outcomes following hematopoietic cell transplant***<br><br>Learning Objectives:<br>1. Assess outcomes and complications of allo-HCT of Canadian infants with CD3δ SCID in a more modern era.<br>2. Analysis of bone marrow transplantation-related challenges to develop new management strategies focused on maximizing engraftment across all cell lines, optimizing immune reconstitution, and preventing complications such as graft-versus-host disease (GVHD), autoimmune disorders, and infections. This data will be utilized as a control group in a gene therapy clinical trial. |
| | 3 min | Q&A |

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| 2:41 pm | 9 min | **Jason Z X Chen[1],** Rae Brager[2], Tania Cellucci[2], Sam Waserman[3], Yue (Jennifer) Du[3], Jenny Garkaby[2]. [1]Michael G. DeGroote School of Medicine, McMaster University, Hamilton, ON. [2]Dept. of Pediatrics, Division of Rheumatology, Immunology and Allergy, McMaster University, Hamilton, ON. [3]Dept. of Medicine, McMaster University, Hamilton, ON.<br><br>***Treatment-resistant vesiculopustular lesions in APLAID patient: a case report***<br><br>Learning Objectives:<br>1. Review etiology and associated pathophysiology of APLAID<br>2. Explore management strategies for APLAID<br>3. Discuss further management options for resistant skin manifestations of APLAID |
| | 3 min | Q&A |
| 2:53 pm | 9 min | **Karan S. Purewal[1]**, Lily Lim[2], Michael S. Salman[3], Edward Leung[3], Aziz Mhanni[4], Sandra Marles[4], Cheryl Rockman Greenberg[4], Angela Krutish[4], Valerie A. Brulé[5], Samantha E. Marin[3], Patrick Frosk[4], Donald Vinh[6], Tamar Rubin[7]. [1]Department of Pediatrics, University of Saskatchewan, Saskatoon, SK. [2]Section of Pediatric Rheumatology, Department of Pediatrics and Child Health, University of Manitoba, Winnipeg, MB. [3]Section of Pediatric Neurology, Department of Pediatrics and Child Health, University of Manitoba, Winnipeg, MB. [4]Program of Genetics and Metabolism, Shared Health and Department of Pediatrics and Child Health, University of Manitoba, Winnipeg, MB. [5]Section of Pediatric Critical Care, Department of Pediatrics and Child Health, University of Manitoba, Winnipeg, MB. [6]Division of Infectious Diseases, Department of Medicine, Department of Medical Microbiology, Research Institute of the McGill University Health Centre, McGill University Health Centre, Montreal, QC. [7]Section of Pediatric Clinical Immunology and Allergy, Department of Pediatrics and Child Health, University of Manitoba, Winnipeg, MB.<br><br>***Two homozygous TREX1 variants in an Inuit pedigree with Aicardi-Goutières Syndrome***<br><br>Learning Objectives:<br>1. Describe the clinical features and spectrum of Aicardi-Goutières Syndrome (AGS) in an Inuit family with homozygous TREX1 variants.<br>2. Review unique features of AGS in our pedigree, including suspected immunodeficiency and interstitial lung disease.<br>3. Discuss emerging therapeutic strategies for AGS |
| | 3 min | Q&A |
| 3:05 pm | 9 min | **Sujen Saravanabavan[1]**, Elliot James[1], Kyla Hildebrand[1], Sarah Lohrenz[1], Audi Setiadi[2], Stuart Turvey[1], Kevin C. Harris[1], Catherine M Biggs[1]. [1]Department of Paediatrics, BC Children's Hospital, The University of British Columbia, Vancouver, BC. [2]Department of Pathology and Laboratory Medicine, The University of British Columbia, Vancouver, BC.<br><br>***A case of protein losing enteropathy post-Fontan procedure with associated hypogammaglobulinemia and T-cell deficiency***<br><br>Learning Objectives:<br>1. Understand proposed pathophysiologic mechanisms that link protein-losing enteropathy (PLE) and immune dysfunction in post-Fontan patients.<br>2. Discuss the presentation including relevant findings on immune work-up in patients with PLE post-Fontan procedure.<br>3. Evaluate the options for immunoprophylaxis in patients with PLE post-Fontan procedure. |
| | 3 min | Q&A |

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| 3:17 pm | 9 min | **Noha Benharira[2]**, Fatemah AlYaqout[1], Michael Aw[2], Hamza Alghamdi[1], Bruce Mazer[1], Reza Alizadehfar[1], Christos Tsoukas[1], Michael Fein[1]. [1]Division of Clinical Immunology and Allergy, Department of Medicine, McGill University Health Center (MUHC), Montreal, QC. [2]McGill University Faculty of Medicine, Montreal, QC.<br><br>***Novel AICDA mutation (c.238T>C, p.Trp80Arg) associated with Hyper-IgM Syndrome: A case study***<br><br>Learning Objectives:<br>1. Demonstrate the diagnostic dilemma of hypomorphic Hyper-IgM Syndrome.<br>2. Recognize c.238T>C, p.Trp80Arg AICDA mutation as a novel variant of Hyper-IgM Syndrome. |
| | 3 min | Q&A |
| 3:29 pm | 9 min | **Shatha Alhamdi[1,2]**, Chaim Roifman[1]. [1]Division of Immunology & Allergy, Department of Pediatrics, Hospital for Sick Children and University of Toronto, Toronto, ON. [2]Allergy and Immunology Division, Department of Pediatrics, King Abdulaziz Medical City, Riyadh, Saudi Arabia.<br><br>***Consideration of multiple genetic findings can improve patient care***<br><br>Learning Objectives:<br>1. Recognize the importance of identifying and reporting multiple genetic findings that have the potential to impact patient care<br>2. To improve communication among healthcare providers and patients regarding multiple genetic findings, promote informed decision-making, and enhance patient engagement in their care |
| | 3 min | Q&A |
| 3:41 pm | 9 min | **Angela Maccan[1],** Catherine M. Biggs[1], Kyla J. Hildebrand[1], Stuart E. Turvey[1], Bruce Carleton[2], Sean Young[3], Elliot James[1]. [1]Division of Clinical Immunology & Allergy, Department of Paediatrics, BC Children's Hospital, The University of British Columbia, BC. [2]Division of Translational Therapeutics, Department of Pediatrics, BC Children's Hospital, University of British Columbia, BC. [3]Cancer Genetics and Genomics Laboratory, Department of Pathology and Laboratory Medicine, BC Cancer, Vancouver, BC.<br><br>***In-utero azathioprine induced severe lymphopenia in a neonate with a positive newborn screen for severe combined immunodeficiency***<br><br>Learning Objectives:<br>1. Review a case of an infant who presented with T, B, and NK-cell lymphopenia after in-utero exposure to azathioprine |
| | 3 min | Q&A |
| 3.53 pm | | **Closing and Abstract Winners**<br><br>*chosen based on submitted abstract content*<br><br>Best abstract –<br>2nd place best abstract –<br>3rd place best abstract – |