This event 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, approved by Canadian Society of Allergy and Clinical Immunology

**Overall Learning Objectives** 

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

#### Thursday October 28, 2021

Time	Duration & Interactivity	Торіс
10:30 am	Moderators	Introduction & Welcome Chaim M. Roifman, CM, MD, The Hospital for Sick Children, Toronto, Ontario Linda Vong, PhD, Immunodeficiency Canada
10:32 am	9 min	<b>Adil Adatia</b> <sup>1</sup> , Vicky Breakey <sup>2</sup> , Jennifer Mackenzie <sup>3</sup> , Yogi Chopra <sup>4</sup> , Rae Brager <sup>5</sup> . <sup>1</sup> Div. of Clinical Immunology & Allergy, Dept. of Medicine, <sup>2</sup> Div. of Pediatric Hematology and Oncology, <sup>3</sup> Div. of Genetics, <sup>4</sup> Div. of Haematology/Oncology, <sup>5</sup> Div. of Rheumatology, Immunology, and Allergy, Dept. of Pediatrics, McMaster Children's Hospital, McMaster University, Hamilton, Ontario.
		Griscelli syndrome type 2 treated with hematopoietic stem cell transplantation using an unrelated cord blood donor
		Learning Objectives: 1. Review the diagnostic considerations in Griscelli syndrome type 2 2. Discuss the pre-transplant management of Griscelli syndrome type 2 3. Discuss the evidence of HSCT in Griscelli syndrome type 2
	3 min	Q&A
10:44 am 10.56 am	9 min	<b>Rongbo Zhu</b> <sup>1</sup> , Chaim M. Roifman <sup>2,3</sup> . <sup>1</sup> Div. of Clinical Immunology & Allergy, Dept of Medicine, Schulich School of Medicine & Dentistry, London, Ontario, <sup>2</sup> Div. of Immunology & Allergy, Dept. of Pediatrics, The Hospital for Sick Children and the University of Toronto, Toronto, Ontario. <sup>2</sup> Canadian Centre for Primary Immunodeficiency, Toronto, Ontario.
		Compound heterozygosity with a novel missense variant in PRF1 in a patient with hemophagocytic lymphohistiocytosis (HLH)
		Learning Objectives: 1. Recognize symptoms of hemophagocytic lymphohistiocytosis (HLH) 2. Appreciate the diversity in patients with inborn errors of immunity
	3 min	Q&A
	9 min	<b>Lisa Liang</b> <sup>1</sup> , Marianne Miguel <sup>1</sup> , Patrick Frosk <sup>2,3</sup> , Janet Chou <sup>4</sup> , Abduarahman Almutairi <sup>4</sup> , Craig Platt <sup>4</sup> , Raif Geha <sup>4</sup> , Tamar Rubin <sup>1</sup> . <sup>1</sup> Div. of Clinical Immunology and Allergy, <sup>2</sup> Div. of Biochemistry and Medical Genetics, Dept. of Pediatrics, University of Manitoba, Winnipeg, <sup>3</sup> Children's Hospital Research Institute of Manitoba, Winnipeg, <sup>4</sup> Div. of Clinical Immunology and Allergy, Boston Children's Hospital, Harvard Medical School, Boston, U.S.
		Virally triggered multisystem inflammatory disease associated with a pathogenic mutation in a novel gene
		Learning Objectives: 1. Summarize the clinical presentation, immunological features and potential treatments for patients with this novel gene mutation 2. Describe an approach to investigating patients with suspected PIDs who remain without a genetic diagnosis after undergoing standard of care clinical investigations
	3 min	Q&A

#### Thursday October 28, 2021 (cont.)

Time	Duration & Interactivity	Торіс
11.08 am	9 min	<b>Jenny Garkaby</b> <sup>1</sup> , Jessica Willett-Pachul <sup>1</sup> , Brenda Reid <sup>1,2</sup> , Chaim M. Roifman <sup>1,2</sup> . <sup>1</sup> Div. of Immunology & Allergy, Dept. of Pediatrics, The Hospital for Sick Children and the University of Toronto, Toronto, ON. <sup>2</sup> Canadian Centre for Primary Immunodeficiency, Toronto, Ontario.
		Atopy and autoimmunity in Roifman Syndrome patients
		Learning Objectives: 1. Identify key features of Roifman Syndrome 2. Describe atopy and autoimmunity in Roifman Syndrome patients
	3 min	Q&A
11.20 am	9 min	<b>Keely Loewen</b> <sup>1</sup> , Lily Siok Hoon Lim <sup>2</sup> , Tamar Rubin <sup>1</sup> . <sup>1</sup> Dept. of Pediatrics & Child Health, University of Manitoba, Section of Clinical Immunology and Allergy, Winnipeg, Manitoba, <sup>2</sup> Dept. of Pediatrics & Child Health, University of Manitoba, Section of Pediatric Rheumatology, Winnipeg, Manitoba
		Haploinsufficiency of A20 in a patient with 6q23.2-23.3 deletion
		Learning Objectives: 1. Review the clinical and immunologic features of a rare inborn error of immunity. 2. Discuss importance of immune evaluation in patients with known chromosomal deletions. 3. Understand the value of revisiting previously performed genetic testing in patients with unexplained clinical features.
	3 min	Q&A
11.32 am	9 min	<b>Laura Abrego Fuentes</b> <sup>1</sup> , Amarilla B. Mandola <sup>2</sup> , Bo Ngan <sup>3</sup> , Chaim M. Roifman <sup>1,4</sup> . <sup>1</sup> Div. of Immunology & Allergy, Dept. of Pediatrics, The Hospital for Sick Children and the University of Toronto, Toronto, Ontario. <sup>2</sup> Pediatric Department A, Soroka University Medical Center, Ben-Gurion University of the Negev, Beer-Sheva, Israel. <sup>3</sup> Dept of Paediatric Laboratory Medicine, The Hospital for Sick Children, Toronto, Ontario, <sup>4</sup> Canadian Centre for Primary Immunodeficiency, Toronto, Ontario.
		Immuno-histopathology evaluation of an X-MAID patient with novel mutation in MSN
		Learning Objectives: 1. Recognize the role of cytoskeleton cell proteins in the development of a primary immunodeficiency. 2. Demonstrate the clinical characteristic features of a novel mutation in the moesin gene ( <i>MSN</i> ). 3. Identify the immuno-histopathology findings in the thymus and lung of an X-MAID patient.
	3 min	Q&A

#### Thursday October 28, 2021 (cont.)

Time	Duration & Interactivity	Торіс
11.44 am	9 min	<b>Meriem Latrous</b> <sup>1</sup> , Elliot James <sup>1</sup> , Catherine M. Biggs <sup>1,2</sup> , Stuart E. Turvey <sup>1,2</sup> , Kyla J. Hildebrand <sup>1,2</sup> . <sup>1</sup> Div. of Allergy and Immunology, Dept of Paediatrics, BC Children's Hospital, The University of British Columbia, <sup>2</sup> British Columbia Children's Hospital Research Institute; Vancouver, British Columbia.
		A case of Trichohepatoenteric Syndrome due to biallelic damaging variants in TTC37A
		Learning Objectives: 1. Describe the clinical features of Trichohepatoenteric Syndrome (THES) 2. List the genes in which pathogenic variants cause THES 3. Compare management of THES to Severe Combined Immunodeficiency
	3 min	Q&A
11.56 am	Abstract Recognition	Abstract winners (presented by Chaim M. Roifman)
		Best abstract – 2 <sup>nd</sup> place best abstract –
		3 <sup>rd</sup> place best abstract –