2017 CIS SUMMER SCHOOL

Second Edition: Cases and Controversies

March 23, 2017 • The Westin Seattle • Seattle, Washington
CONTINUING MEDICAL EDUCATION

PURPOSE
Provide an opportunity for physicians to update their knowledge of primary immune deficiency and have practical experience with real-life cases.

CONTENT AREA
The content will cover practical, cutting edge and theoretical issues related to patient care. Many types of immune deficiencies will be covered to provide the maximal educational experience.

TARGET AUDIENCE
Physicians, scientists and nurses who are involved in the evaluation and/or care of patients with primary immune deficiencies. Participants will pediatricians, internists, or scientists committed to the clinical management of patients with primary immune deficiency diseases. These candidates may have backgrounds in immunology, adult medicine, rheumatology, hematology, laboratory immunology, or allergy/immunology. This specific learning experience is targeted to those with significant clinical experience in the care of patients with primary immune deficiencies. Lab directors or others involved in diagnostic approaches would represent a key target group.

LEARNING OBJECTIVES
Provide advanced training for physicians and researchers working in the field of primary immune deficiency that covers diagnosis, therapy and future trends related to the care of patients with primary immune deficiency.

- Facilitate interactions among clinicians of diverse training to promote multicenter clinical trials and to coordinate participation in existing clinical trials.
- Improve recognition of primary immune deficiencies.
- Improve understanding of the diagnostic issues and the pathophysiology of primary immune deficiencies.
- Improve understanding of the appropriate interventions for these diseases.

ACCME ACCREDITATION STATEMENT
This activity has been planned and implemented in accordance with the accreditation requirements and policies of the Accreditation Council for Continuing Medical Education (ACCME) through the sponsorship of the Clinical Immunology Society (CIS). The CIS is accredited by the ACCME to provide continuing medical education for physicians.

DESIGNATION STATEMENT
The Clinical Immunology Society designates this live activity for a maximum of 5.75 AMA PRA Category 1 Credit(s)™. Physicians should claim only the credit commensurate with the extent of their participation in the activity.
# PROGRAM COMMITTEE AND SPEAKER DISCLOSURES

## DISCLOSURES OF CONFLICT OF INTEREST

The Clinical Immunology Society (CIS) require instructors, planners, managers and other individuals who are in a position to control the content of this activity to disclose any real or apparent conflict of interest (COI) they may have as related to the content of this activity. All identified COI are thoroughly vetted and resolved. The existence or absence of COI for everyone in a position to control content appears below.

<table>
<thead>
<tr>
<th>Name</th>
<th>Institution</th>
<th>Disclosures</th>
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<tbody>
<tr>
<td>Charlotte Cunningham-Rundles, MD, PhD</td>
<td>MOUNT SINAI MEDICAL CENTER</td>
<td>Momenta – Consultant receiving consulting fee</td>
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<td>Biotest – Consultant receiving consulting fee</td>
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<td>ADMA – Consultant receiving consulting fee</td>
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<tr>
<td>Elie Haddad, MD, PhD</td>
<td>CHU SAINTE-JUSTINE, UNIVERSITY OF MONTREAL</td>
<td>CSL Behring - Investigator Initiated Grant - Principal Investigator</td>
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<td>Sigma-Tau - Consultant receiving consulting fee</td>
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<tr>
<td>Emma Westermann-Clark, MD</td>
<td>UNIVERSITY OF SOUTH FLORIDA</td>
<td>Celgene (spouse) – Speaker received speaker fee</td>
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<td>Jordan Orange, MD, PhD</td>
<td>BAYLOR COLLEGE OF MEDICINE</td>
<td>ADMA - Membership on Advisory Committee or Review Committee;</td>
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<tr>
<td>Lisa Forbes, MD</td>
<td>BAYLOR COLLEGE OF MEDICINE</td>
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<td>Thomas Fleisher, MD</td>
<td>NIH CLINICAL CENTER</td>
<td>Elsevier – Independent Contractor receiving royalty</td>
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<td>Tiphanie Vogel, MD, PhD</td>
<td>BAYLOR COLLEGE OF MEDICINE</td>
<td>AB2Bio - Consultant receiving consulting fee</td>
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<tr>
<td>Troy Torgerson, MD, PhD</td>
<td>UNIVERSITY OF WASHINGTON AND SEATTLE CHILDREN’S RESEARCH INSTITUTE</td>
<td>Baxalta/Shire - Consultant receiving consulting fee</td>
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## SCHEDULE OF EVENTS

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<tr>
<th>Time</th>
<th>Event</th>
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<tbody>
<tr>
<td>7:00am – 8:00am</td>
<td>Registration &amp; Continental Breakfast</td>
<td>Grand Foyer</td>
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<tr>
<td>8:00am – 8:15am</td>
<td><strong>Welcome &amp; Introduction</strong>&lt;br&gt;Charlotte Cunningham-Rundles, MD, PhD, Mt. Sinai Medical Center&lt;br&gt;Orange, MD, PhD, Baylor College of Medicine and Texas Children’s Hospital</td>
<td>Grand Foyer</td>
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<tr>
<td>8:15am – 8:40am</td>
<td><strong>Section 1</strong>&lt;br&gt;AMA PRA Category 1 Credits™&lt;br&gt;Moderator:&lt;br&gt;Elie Haddad, MD, PhD, CHU Ste-Justine, University of Montreal&lt;br&gt;<strong>SPEAKERS:</strong>&lt;br&gt;9 YEAR OLD WITH MOSAIC KRAS VARIANT&lt;br&gt;Nicholas Hartog, MD, Helen DeVos Children’s Hospital and Spectrum Health Medical Group&lt;br&gt;AGAMMAGLOBULINEMIA PATIENTS WITH ENTEROVIRUS&lt;br&gt;Analía Gisela Seminario, MD, Ricardo Gutiérrez Children’s Hospital&lt;br&gt;REFRACTORY AUTOIMMUNE CYTOPENIA COINCIDES WITH VARICELLA INFECTION IN A CHILD WITH RAG DEFICIENCY&lt;br&gt;Aniko Eva Malik, MD, Semmelweis University</td>
<td>Fifth Avenue Room</td>
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<td>8:40am – 8:50am</td>
<td><strong>KEY UPDATES ON HSCT AND GENE THERAPY</strong>&lt;br&gt;AMA PRA Category 1 Credits™&lt;br&gt;Elie Haddad, MD, PhD, CHU Ste-Justine, University of Montreal</td>
<td>Fifth Avenue Room</td>
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<tr>
<td>8:50am – 9:15am</td>
<td><strong>Section 2</strong>&lt;br&gt;AMA PRA Category 1 Credits™&lt;br&gt;Moderator:&lt;br&gt;Charlotte Cunningham Rundles, MD, PhD, Mt. Sinai Medical Center&lt;br&gt;<strong>SPEAKERS:</strong>&lt;br&gt;A CASE OF BAD CARMA!&lt;br&gt;David Buchbinder, MD, CHOC Children’s Hospital - UC Irvine&lt;br&gt;A CASE OF ATYPICAL SYNDROMIC FORM OF CONGENITAL SIDEROBLASTIC ANEMIAS ASSOCIATED WITH B-CELL IMMUNODEFICIENCY, PERIODIC FEVERS, FEVERS, AND DEVELOPMENTAL DELAY COMPLICATED BY T-CELL LYMPHOMAS&lt;br&gt;Federica Pulvirenti, MD, Sapienza, University of Rome&lt;br&gt;ANTINEUTROPHILIC CYTOPLASMIC ANTIBODIES (ANCA)-ASSOCIATED VASCULITIS IN COMMON VARIABLE IMMUNODEFICIENCY PATIENT&lt;br&gt;Adeeb A Bulkhi, MD, University of South Florida, Department of Medicine, Division of Allergy and Immunology</td>
<td>Fifth Avenue Room</td>
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<td>9:15am – 9:25am</td>
<td><strong>KEY UPDATES ON ANTIBODY DEFECTS</strong>&lt;br&gt;Charlotte Cunningham-Rundles, MD, PhD, Mt. Sinai Medical Center</td>
<td>Fifth Avenue Room</td>
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### SCHEDULE OF EVENTS

#### 9:25am - 9:50am
**Section 3**

0.5 AMA PRA Category 1 Credits™

**Moderator:**
Troy Torgerson, MD, PhD, Seattle Children's Hospital and The University of Washington

**SPEAKERS:**
- **Harnessing Pharmacogenomics to Guide the Treatment of LRBA Deficiency**
  Elizabeth L Wisner, MD, LSU Health Sciences Center
- **Elevated IgE, EBV Disease, and Failure to Thrive: Expanding the Phenotype of CTLA4 Haploinsufficiency**
  Sarah Kogan Nicholas, MD, Baylor College of Medicine
- **Hemophagocytic Lymphohistiocytosis (HLH) in a Patient with a Novel STAT3GOF Mutation**
  Lisa R Forbes, MD, Baylor College of Medicine and Texas Children's Hospital

#### 9:50am – 10:00am
**Key Updates on Immune Dysregulatory Disorders**
Troy Torgerson, MD, PhD, Seattle Children's Hospital and The University of Washington

#### 10:00am – 10:10am
**Lightning Round #1**

0.25 AMA PRA Category 1 Credits™

**Panelists:**
- **Secondary Immune Deficiencies**
  Elie Haddad, MD, PhD, CHU Ste-Justine, University of Montreal
- **When the Genetic Shoe Doesn’t Fit**
  Charlotte Cunningham Rundles, MD, PhD, Mt. Sinai Medical Center
- **Complement**
  Troy Torgerson, MD, PhD, Seattle Children's Hospital

#### 10:25am – 10:50pm
**Section 4**

0.5 AMA PRA Category 1 Credits™

**Moderator:**
Hans Ochs, MD, University of Washington

**SPEAKERS:**
- **Human CD40 Ligand Deficiency Dysregulates Neutrophil Ontogeny That is Improved by Recombinant IFN-γ**
  Antonio Condino-Neto, MD, PhD, University of Sao Paulo
- **All in the Family**
  Geeta Madathil Govindaraj, MD, Government Medical College, Kozhikode, Kerala
- **Clinical Presentations of Chromosomal Instability Syndromes**
  Mirinda Gillespie, ScM, MD, Johns Hopkins All Children’s Hospital

#### 10:50am – 11:00am
**Key Updates on Wiskott-Aldrich Syndrome, XLT, & XLN**
Hans Ochs, MD, University of Washington
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<th>Speaker/Panelists</th>
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| 11:00am – 11:35 am | **Section 5**  
 .5 AMA PRA Category 1 Credits™  
 **Moderator:** Luigi Notarangelo, MD, National Institute of Allergy and Infectious Diseases, NIH  
 **SPEAKERS:**  
 Sarah Emily Henrickson, MD, PhD, Children’s Hospital of Philadelphia  
 Filiz O. Seeborg, MD, MPH, Baylor College of Medicine and Texas Children’s Hospital  
 Sara Barmettler, MD, Massachusetts General Hospital  
 Yasmin Khan, MD, Vanderbilt University, Vanderbilt Children’s Hospital |  

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| 11:35am – 11:45am | **KEY UPDATES IN SCID AND CID**  
 Luigi Notarangelo, MD, National Institute of Allergy and Infectious Diseases, NIH |  

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| 11:45am – 12:10 pm | **Section 6**  
 .5 AMA PRA Category 1 Credits™  
 **Moderator:** Thomas Fleisher, MD, NIH Clinical Center  
 **SPEAKERS:**  
 Keith Sacco, MD, Mayo Clinic Jacksonville  
 Joud Hajjar, MD, Texas Children’s Hospital  
 Donna S. Hummell, MD, Vanderbilt University, Vanderbilt Children’s Hospital |  

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<th>Speaker/Panelists</th>
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| 12:10pm – 12:20pm | **KEY UPDATES IN NEW DIAGNOSTIC APPROACHES**  
 Thomas Fleisher, MD, NIH Clinical Center |  

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<th>Speaker/Panelists</th>
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| 12:20pm – 12:30pm | **Lightning Round #2**  
 .25 AMA PRA Category 1 Credits™  
 **Panelists**  
 CD40L  
 Hans Ochs, MD, University of Washington  
 TTC7A  
 Luigi Notarangelo, MD, National Institute of Allergy and Infectious Diseases, NIH  
 APDS/PALSI  
 Thomas Fleisher, MD, NIH Clinical Center |  

**Fifth Avenue Room**
## SCHEDULE OF EVENTS

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<tbody>
<tr>
<td>12:30pm – 1:00pm</td>
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| 1:00pm – 1:25pm | **Section 7**  
5 AMA PRA Category 1 Credits™  
**Moderator:**  
**Rebecca Marsh, MD,** Cincinnati Children’s Hospital  
**SPEAKERS:**  
**PARTIAL RAG DEFICIENCY IN A CHILD WITH AUTOIMMUNE CYTOPENIA AND FEATURES OF AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME**  
**Emma Westermann-Clark, MD,** University of South Florida Morsani College of Medicine  
**A NOVEL NLRC4 MUTATION TREATED WITH BONE MARROW TRANSPLANTATION**  
**Amanda V Gripen Goddard, DO,** University of Iowa Hospitals and Clinics  
**INFLAMMASOME MUTATION IN A PATIENT WITH SYSTEMIC LUPUS ERYTHEMATOSUS: UNEXPECTED GENETIC FINDING HARMONIZES WITH CLINICAL HISTORY**  
**Tiphanie Vogel, MD, PhD,** Baylor College of Medicine and Texas Children’s Hospital Center for Human Immunobiology |
| 1:25pm – 1:35pm | KEY UPDATES IN HLH  
**Rebecca Marsh, MD,** Cincinnati Children’s Hospital |
| 1:35pm – 2:00pm | **Section 8**  
5 AMA PRA Category 1 Credits™  
**Moderator:**  
**Steve Holland, MD,** National Institute of Allergy and Infectious Diseases, NIH  
**SPEAKERS:**  
**MULTIPLE BRAIN ABSCESSES CAUSED BY TRICHOSTORON INKIN IN A PATIENT WITH X-LINKED CHRONIC GRANULOMATOUS DISEASE (CGD) SUCCESSFULLY TREATED WITH ANTIFUNGAL THERAPY**  
**Joud Hajjar, MD,** Texas Children’s Hospital  
**USE OF VEDOLIZUMAB IN TREATMENT OF CHRONIC GRANULOMATOUS DISEASE (CGD)-ASSOCIATED COLITIS**  
**Johana B Castro-Wagner, MD,** University of South Florida |
| 2:00pm – 2:10pm | KEY UPDATES IN INNATE IMMUNE DEFICIENCIES  
**Steve Holland, MD,** National Institute of Allergy and Infectious Diseases, NIH |
| 2:10pm – 2:20pm | BREAK |
| 2:20pm – 2:45pm | **Section 9**  
5 AMA PRA Category 1 Credits™  
**Moderator:**  
**Jordan Orange, MD, PhD,** Baylor College of Medicine and Texas Children’s Hospital  
**SPEAKERS:**  
**FINDING A NEEDLE IN A HAYSTACK: NEW APPROACHES TO SELECT DISEASE-CAUSING MUTATIONS IN PATIENTS’ GENOMES**  
**Yuval Itan, PhD,** The Rockefeller University  
**UNIQUE PID PEDIGREE FAMILY TEACHING ABOUT MUTATION ORIGINS**  
**Gesmar Segundo, MD, PhD,** Seattle Children’s Research Institute  
**ACUTE LEUKEMIA, HLH, LYMPHOMATOSIS GRANULOMATOSA AND DISSEMINATED CMV INFECTION IN A PATIENT WITH PIK3R5 MUTATION**  
**Maria Soledad Caldirola, MD,** Ricardo Gutierrez Children’s Hospital |
| 2:45pm – 2:55pm | EMERGING STRATEGIES FOR THE DIAGNOSIS OF PIDD  
**Jordan Orange, MD, PhD,** Baylor College of Medicine and Texas Children’s Hospital |
### SCHEDULE OF EVENTS

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<th>Time</th>
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| 2:55pm – 3:30pm | **Section 10**  
.5 AMA PRA Category 1 Credits™  
Moderator: Kathleen Sullivan, MD, PhD, Children’s Hospital of Philadelphia  
**SPEAKERS:**  
CLINICAL PRESENTATION OF DISSEMINATED MYCOBACTERIA GENAVENSE IN A MOTHER AND HER CHILD DUE TO MENDELIAN SUSCEPTIBILITY TO MYCOBACTERIAL DISEASE (MSMD)  
Lacey B. Robinson, MD, Massachusetts General Hospital  
A DOUBLE EDGED SWORD: TREATING HYPOGAMMAGLOBULINEMIA IN A COMPLEMENT DEFICIENCY PATIENT  
Sara E Sussman, MD, LSU Health Sciences Center-New Orleans  
DIAGNOSIS OF KABUKI SYNDROME IN A PATIENT WITH IMMUNODEFICIENCY  
Bhumika Patel, MD, University of South Florida  
SEVERE COMBINED IMMUNODEFICIENCY (SCID) CAUSED BY THYMIC APLASIA DUE TO FAMILIAL MUTATION IN Tbx1  
David S Lindsay II, MD, University of South Florida | Fifth Avenue Room                                                                                                                                 |
| 3:30pm – 3:40pm | **KEY UPDATES ON PIDD ASSOCIATED WITH IBD**  
Kathleen Sullivan, MD, PhD, Children’s Hospital of Philadelphia | | |
| 3:40pm – 3:50pm | **Lightning Round #3**  
.25 AMA PRA Category 1 Credits™  
Panelists  
XIAP  
Rebecca Marsh, MD, Cincinnati Children’s Hospital  
GATA2  
Steve Holland, MD, National Institute of Allergy and Infectious Diseases, NIH  
NEMO  
Jordan Orange, MD, PhD, Baylor College of Medicine and Texas Children’s Hospital  
22q11.2 DELETION AND DUPLICATION  
Kathleen Sullivan, MD, PhD, Children’s Hospital of Philadelphia | | |
| 3:50pm – 4:00pm | **WRAP UP**  
Kathleen Sullivan, MD, PhD, Children’s Hospital of Philadelphia | | |
THANK YOU TO OUR SUPPORTERS!
CSL Behring
Grifols
Kedrion Biopharama
Shire
USIDNET

SAVE THE DATE
CIS 2018 Annual Meeting:
Immune Deficiency and Dysregulation
North American Conference

April 26-29, 2018 Sheraton Centre Toronto Toronto, Canada
Applications now open! Apply online via the CIS Website, www.clinimmsoc.org

Applications must be supported by a:
• Letter from the Training Program Director or Department Chair;
• Curriculum Vitae;
• Statement of how the School will be beneficial;
• Abstract of the case or research project. CIS is looking for a case report including those which can be presented as a mystery for discussion purposes but should have a final diagnosis. Alternatively, cases with diagnostic dilemmas which could be discussed are acceptable. In addition, completed research projects germane to the field of primary immune deficiency are also acceptable.

Accepted abstracts will be provided with travel grants to assist with airfare, hotel, and meal expenses. The language of the School will be in English and selected participants are required to become CIS members at the conclusion of the School.

Target Audience:
• Fellows-in-training (within their last two years of training);
• Junior faculty members (within two years of completion of training).

Participants should be pediatricians, internists, or scientists committed to the clinical management of patients with primary immunodeficiency diseases. Candidates may have backgrounds in immunology, infectious diseases, adult medicine, rheumatology, hematology, laboratory immunology, stem cell transplantation, or allergy/immunology.

Objectives:
• Education on the diagnosis, pathogenesis, and treatment of PIDD;
• Attract and develop future scientists in academic medicine;
• Enhance awareness of clinical immunology and its importance in scientific discoveries and clinical applications;
• Stimulate future collaborations between young investigators in different medical centers and countries, and between young investigators and experienced physician/scientists in the field.