

# 2017 CIS SUMMER SCHOOL

*Second Edition: Cases and Controversies*

March 23, 2017 • The Westin Seattle • Seattle, Washington





CIS National Office  
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## PROGRAM COMMITTEE

### Co-Chairs:

**Charlotte Cunningham-Rundles, MD, PhD**

Mt. Sinai Medical Center

**Kathleen E. Sullivan, MD, PhD**

Children's Hospital of Philadelphia

### Faculty:

**Thomas Fleisher, MD**

NIH Clinical Center

**Elie Haddad, MD, PhD**

CHU Ste-Justine, University of Montreal

**Steven M. Holland, MD**

NIAID, NIH

**Rebecca A. Marsh, MD**

Cincinnati Children's Hospital

**Luigi Notarangelo, MD**

NIAID, NIH

**Hans D. Ochs, MD**

University of Washington

**Jordan S. Orange, MD, PhD**

Baylor College of Medicine  
Texas Children's Hospital

**Troy R. Torgerson, MD, PhD**

University of Washington

## JOIN US



## 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 be 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)<sup>™</sup>. 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.

### **Charlotte Cunningham-Rundles, MD, PhD**

MOUNT SINAI MEDICAL CENTER

Momenta – Consultant receiving consulting fee

Biotest – Consultant receiving consulting fee

ADMA – Consultant receiving consulting fee

### **Elie Haddad, MD, PhD**

CHU SAINTE-JUSTINE, UNIVERSITY OF MONTREAL

CSL Behring - Investigator Initiated Grant - Principal Investigator

Sigma-Tau - Consultant receiving consulting fee

### **Emma Westermann-Clark, MD**

UNIVERSITY OF SOUTH FLORIDA

Celgene (spouse) – Speaker received speaker fee

### **Jordan Orange, MD, PhD**

BAYLOR COLLEGE OF MEDICINE

ADMA - Membership on Advisory Committee or Review Committee;

Consultant receiving honoraria

CSL Behring - Consultant receiving honoraria

Shire - Consultant receiving honoraria

Grifols - Consultant receiving consulting fee

### **Lisa Forbes, MD**

BAYLOR COLLEGE OF MEDICINE

Baxalta/Shire - Consultant receiving consulting fee

Prometic - Consultant receiving consulting fee

### **Thomas Fleisher, MD**

NIH CLINICAL CENTER

Elsevier – Independent Contractor receiving royalty

### **Tiphonie Vogel, MD, PhD**

BAYLOR COLLEGE OF MEDICINE

AB2Bio - Consultant receiving consulting fee

### **Troy Torgerson, MD, PhD**

UNIVERSITY OF WASHINGTON AND SEATTLE CHILDREN'S RESEARCH INSTITUTE

Baxalta/Shire - Consultant receiving consulting fee

CSL Behring - Consultant receiving consulting fee

ADMA - Consultant receiving consulting fee

aTyr Pharmaceuticals - Consultant receiving consulting fee

# SCHEDULE OF EVENTS

7:00am – 8:00am	<b>Registration &amp; Continental Breakfast</b>	Grand Foyer
8:00am - 8:15am	<b>Welcome &amp; Introduction</b> <b>Charlotte Cunningham-Rundles, MD, PhD</b> , Mt. Sinai Medical Center <b>Jordan Orange, MD, PhD</b> , Baylor College of Medicine and Texas Children's Hospital	
8:15am - 8:40am	<b>Section 1</b> <i>.5 AMA PRA Category 1 Credits™</i> <i>Moderator:</i> <b>Elie Haddad, MD, PhD</b> , CHU Ste-Justine, University of Montreal <i>SPEAKERS:</i> 9 YEAR OLD WITH MOSAIC KRAS VARIANT <b>Nicholas Hartog, MD</b> , Helen DeVos Children's Hospital and Spectrum Health Medical Group AGAMMAGLOBULINEMIA PATIENTS WITH ENTEROVIRUS <b>Analia Gisela Seminario, MD</b> , Ricardo Gutiérrez Children's Hospital REFRACTORY AUTOIMMUNE CYTOPENIA COINCIDES WITH VARICELLA INFECTION IN A CHILD WITH RAG DEFICIENCY <b>Aniko Eva Malik, MD</b> , Semmelweis University	
8:40am - 8:50am	KEY UPDATES ON HSCT AND GENE THERAPY <i>.25 AMA PRA Category 1 Credits™</i> <b>Elie Haddad, MD, PhD</b> , CHU Ste-Justine, University of Montreal	Fifth Avenue Room
8:50am – 9:15am	<b>Section 2</b> <i>.25 AMA PRA Category 1 Credits™</i> <i>Moderator:</i> <b>Charlotte Cunningham Rundles, MD, PhD</b> , Mt. Sinai Medical Center <i>SPEAKERS:</i> A CASE OF BAD CARMA! <b>David Buchbinder, MD</b> , CHOC Children's Hospital - UC Irvine 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 <b>Federica Pulvirenti, MD</b> , Sapienza, University of Rome ANTINEUTROPHILIC CYTOPLASMIC ANTIBODIES (ANCA)-ASSOCIATED VASCULITIS IN COMMON VARIABLE IMMUNODEFICIENCY PATIENT <b>Adeeb A Bulkhi, MD</b> , University of South Florida, Department of Medicine, Division of Allergy and Immunology	
9:15am – 9:25am	KEY UPDATES ON ANTIBODY DEFECTS <b>Charlotte Cunningham-Rundles, MD, PhD</b> , Mt. Sinai Medical Center	

# SCHEDULE OF EVENTS

9:25am - 9:50am	<p><b>Section 3</b>  <i>.5 AMA PRA Category 1 Credits™</i></p> <p><i>Moderator:</i>  <b>Troy Torgerson, MD, PhD</b>, Seattle Children's Hospital and The University of Washington</p> <p><i>SPEAKERS:</i>  HARNESSING PHARMACOGENOMICS TO GUIDE THE TREATMENT OF LRBA DEFICIENCY  <b>Elizabeth L Wisner, MD</b>, LSU Health Sciences Center  ELEVATED IgE, EBV DISEASE, AND FAILURE TO THRIVE: EXPANDING THE PHENOTYPE OF CTLA4 HAPLOINSUFFICIENCY  <b>Sarah Kogan Nicholas, MD</b>, Baylor College of Medicine  HEMOPHAGOCITIC LYMPHOHISTIOCYTOSIS (HLH) IN A PATIENT WITH A NOVEL STAT3GOF MUTATION  <b>Lisa R Forbes, MD</b>, Baylor College of Medicine and Texas Children's Hospital</p>	Fifth Avenue Room
9:50am – 10:00am	<p>KEY UPDATES ON IMMUNE DYSREGULATORY DISORDERS  <b>Troy Torgerson, MD, PhD</b>, Seattle Children's Hospital and The University of Washington</p>	
10:00am – 10:10 am	<p><b>Lightning Round #1</b>  <i>.25 AMA PRA Category 1 Credits™</i></p> <p><i>Panelists</i>  SECONDARY IMMUNE DEFICIENCIES  <b>Elie Haddad, MD, PhD</b>, CHU Ste-Justine, University of Montreal  WHEN THE GENETIC SHOE DOESN'T FIT  <b>Charlotte Cunningham Rundles, MD, PhD</b>, Mt. Sinai Medical Center  COMPLEMENT  <b>Troy Torgerson, MD, PhD</b> Seattle Children's Hospital</p>	
10:10am – 10:25am	BREAK	Grand Foyer
10:25am - 10:50pm	<p><b>Section 4</b>  <i>.5 AMA PRA Category 1 Credits™</i></p> <p><i>Moderator:</i>  <b>Hans Ochs, MD</b>, University of Washington</p> <p><i>SPEAKERS:</i>  HUMAN CD40 LIGAND DEFICIENCY DYSREGULATES NEUTROPHIL ONTOGENY THAT IS IMPROVED BY RECOMBINANT IFN-<math>\gamma</math>  <b>Antonio Condino-Neto, MD, PhD</b>, University of Sao Paulo  ALL IN THE FAMILY  <b>Geeta Madathil Govindaraj, MD</b>, Government Medical College, Kozhikode, Kerala  CLINICAL PRESENTATIONS OF CHROMOSOMAL INSTABILITY SYNDROMES  <b>Mirinda Gillespie, ScM, MD</b>, Johns Hopkins All Children's Hospital</p>	Fifth Avenue Room
10:50am – 11:00am	<p>KEY UPDATES ON WISKOTT-ALDRICH SYNDROME, XLT, &amp; XLN  <b>Hans Ochs, MD</b>, University of Washington</p>	

# SCHEDULE OF EVENTS

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11:00am – 11:35 am	<b>Section 5</b> .5 AMA PRA Category 1 Credits™ <i>Moderator:</i> <b>Luigi Notarangelo, MD</b> , National Institute of Allergy and Infectious Diseases, NIH <i>SPEAKERS:</i> NOVEL COMBINED IMMUNODEFICIENCY IDENTIFIED BY NEWBORN SCREEN <b>Sarah Emily Henrickson, MD, PhD</b> , Children's Hospital of Philadelphia UNUSUAL PRESENTATION OF A RARE COMBINED IMMUNODEFICIENCY <b>Filiz O. Seeborg, MD, MPH</b> , Baylor College of Medicine and Texas Children's Hospital FUNCTIONAL DIAGNOSIS OF ATAXIA TELANGIECTASIA IN AN INFANT IDENTIFIED VIA NEWBORN SCREENING FOR SCID (NBS SCID) <b>Sara Barmettler, MD</b> , Massachusetts General Hospital A CASE OF FAMILIAL WHIM SYNDROME IDENTIFIED BY TREC SCREENING <b>Yasmin Khan, MD</b> , Vanderbilt University, Vanderbilt Children's Hospital	
11:35am – 11:45am	KEY UPDATES IN SCID AND CID <b>Luigi Notarangelo, MD</b> , National Institute of Allergy and Infectious Diseases, NIH	
11:45am – 12:10 pm	<b>Section 6</b> .5 AMA PRA Category 1 Credits™ <i>Moderator:</i> <b>Thomas Fleisher, MD</b> , NIH Clinical Center <i>SPEAKERS:</i> DHR FLOW CYTOMETRY IN CHRONIC GRANULOMATOUS DISEASE (CGD): LOOKING AT THE 'LOW FLOW' DHR <b>Keith Sacco, MD</b> , Mayo Clinic Jacksonville VARIANTS OF UNKNOWN SIGNIFICANCE IN A GIRL OF SEVERE ATOPIC DERMATITIS, HYPEREOSINOPHILIA AND FAILURE TO THRIVE <b>Joud Hajjar, MD</b> , Texas Children's Hospital FAMILIAL ALPS WITH VARIABLE PRESENTATION <b>Donna S. Hummell, MD</b> , Vanderbilt University, Vanderbilt Children's Hospital	Fifth Avenue Room
12:10pm – 12:20pm	KEY UPDATES IN NEW DIAGNOSTIC APPROACHES <b>Thomas Fleisher, MD</b> , NIH Clinical Center	
12:20pm – 12:30pm	<b>Lightning Round #2</b> .25 AMA PRA Category 1 Credits™ <i>Panelists</i> CD40L <b>Hans Ochs, MD</b> , University of Washington TTC7A <b>Luigi Notarangelo, MD</b> , National Institute of Allergy and Infectious Diseases, NIH APDS/PALSI <b>Thomas Fleisher, MD</b> , NIH Clinical Center	

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# SCHEDULE OF EVENTS

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

# SCHEDULE OF EVENTS

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2:55pm – 3:30pm	<b>Section 10</b> .5 AMA PRA Category 1 Credits™ <i>Moderator:</i> <b>Kathleen Sullivan, MD, PhD</b> , Children’s Hospital of Philadelphia <i>SPEAKERS:</i> CLINICAL PRESENTATION OF DISSEMINATED MYCOBACTERIA GENAVENSE IN A MOTHER AND HER CHILD DUE TO MENDELIAN SUSCEPTIBILITY TO MYCOBACTERIAL DISEASE (MSMD) <b>Lacey B. Robinson, MD</b> , Massachusetts General Hospital A DOUBLE EDGED SWORD: TREATING HYPOGAMMAGLOBULINEMIA IN A COMPLEMENT DEFICIENCY PATIENT <b>Sara E Sussman, MD</b> , LSU Health Sciences Center-New Orleans DIAGNOSIS OF KABUKI SYNDROME IN A PATIENT WITH IMMUNODEFICIENCY <b>Bhumika Patel, MD</b> , University of South Florida SEVERE COMBINED IMMUNODEFICIENCY (SCID) CAUSED BY THYMIC APLASIA DUE TO FAMILIAL MUTATION IN TBX1 <b>David S Lindsay II, MD</b> , University of South Florida	
3:30pm – 3:40pm	KEY UPDATES ON PIDD ASSOCIATED WITH IBD <b>Kathleen Sullivan, MD, PhD</b> , Children’s Hospital of Philadelphia	Fifth Avenue Room
3:40pm – 3:50pm	<b>Lightning Round #3</b> .25 AMA PRA Category 1 Credits™ <i>Panelists</i> XIAP <b>Rebecca Marsh, MD</b> , Cincinnati Children’s Hospital GATA2 <b>Steve Holland, MD</b> , National Institute of Allergy and Infectious Diseases, NIH NEMO <b>Jordan Orange, MD, PhD</b> , Baylor College of Medicine and Texas Children’s Hospital 22q11.2 DELETION AND DUPLICATION <b>Kathleen Sullivan, MD, PhD</b> , Children’s Hospital of Philadelphia	
3:50pm – 4:00pm	WRAP UP <b>Kathleen Sullivan, MD, PhD</b> , Children’s Hospital of Philadelphia	

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# SAVE THE DATE

## CIS 2018 Annual Meeting:

Immune Deficiency and Dysregulation  
North American Conference

**CIS** Clinical  
Immunology  
Society

April 26-29, 2018 🍁 Sheraton Centre Toronto 🍁 Toronto, Canada

# 2017 CIS School in Primary Immunodeficiency Diseases

GALVESTON, TEXAS • OCTOBER 19-22, 2017



## Save the Date

### CO-CHAIRS:

Charlotte Cunningham-Rundles, MD, PhD  
Mount Sinai Medical Center  
New York, NY

Kathleen E. Sullivan, MD, PhD  
Children's Hospital of Philadelphia  
Philadelphia, PA

Applications now open!  
Apply online via the CIS Website,  
[www.clinimmsoc.org](http://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.







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