



# 2016 Update on Diagnostic Advances in Primary Immunodeficiencies

April 14, 2016 • Seaport Hotel • Boston, Massachusetts

## Final Program





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#### PROGRAM COMMITTEE

**Roshini S. Abraham, PhD (Co-Chair)**

Mayo Clinic

**Troy R. Torgerson, MD PhD (Co-Chair)**

University of Washington

**Jacob Bleesing, MD PhD**

Cincinnati Children's Hospital

**Vijaya Knight, MD PhD**

National Jewish Health

**Sara Nandiwada, PhD D(ABMLI)**

Baylor College of Medicine

**Maurice O'Gorman, PhD MBA MSc D(ABMLI)**

University of Southern California

**Sergio D. Rosenzweig, MD PhD**

NIAID, NIH

**James Verbsky, MD PhD**

Medical College of Wisconsin

## CONTINUING MEDICAL EDUCATION

#### PURPOSE

The purpose is to provide an intellectual forum for all clinicians and researchers from around the world to discuss diagnosis, pathogenesis, and management of the regulation and dysregulation of immunity.

#### TARGET AUDIENCE

The target audience are clinical allergists/immunologists, clinical immunologists, rheumatologists, infectious disease and hematologists who specifically focus in the care and treatment of patients with primary immunodeficiencies (PIDs). Additionally, the target audience includes clinical laboratory immunologists, other PhD level immunologists and scientists along with fellows in training in the above-mentioned clinical disciplines.

#### EXPECTED RESULTS

At the completion of the activity participants should be able to:

- 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 Essential Areas 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 8 *AMA PRA Category 1 Credit(s)*<sup>™</sup>. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

#### CLAIMING CME

To claim CME credit and print out your CME certificate and/or certificate of attendance, please visit <http://www.clinimmsoc.org/cme/pid2016>.

## JOIN US



# 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.

### **Roshini Abraham, PhD**

MAYO CLINIC

No relationships to disclose.

### **Mary Armanios, MD**

JOHNS HOPKINS HOSPITAL

No relationships to disclose.

### **Zuhair K Ballas, MD**

UNIVERSITY OF IOWA HOSPITALS AND CLINICS

Immune Deficiency Foundation – Consultant receiving consulting fee

### **Jack J. Bleesing, MD, PhD**

CINCINNATI CHILDREN'S HOSPITAL MEDICAL CENTER

No relationships to disclose.

### **Francisco Bonilla, MD, PhD**

CHILDREN'S HOSPITAL OF BOSTON

ADMA Biologics – Consultant receiving consulting fee

The Cowen Group – Consultant receiving consulting fee

CSL Behring – Consultant receiving consulting fee; research grant recipient

Food and Drug Administration – Consultant receiving consulting fee

Finger Lakes Allergy Society – Speaking and teaching with honoraria

Gerson-Lehrman Group – Consultant receiving consulting fee

Grand Rounds Health – Consultant receiving consulting fee

Grifols – Consultant receiving consulting fee

Immune Deficiency Foundation – Consultant receiving consulting fee

University of Rochester – Speaking and teaching with honoraria, UptoDate in Medicine- Independent contractor with royalties

### **Kerstin Felgentreff, MD**

UNIVERSITY OF ULM

No relationships to disclose.

### **Thomas Fleisher, MD**

NATIONAL INSTITUTES OF HEALTH

NIH – Employment with salary

Elsevier – Independent contractor with royalties

### **David Hagin, MD, PhD**

UNIVERSITY OF WASHINGTON / SEATTLE

CHILDREN'S HOSPITAL

No relationships to disclose.

### **Elena (Wen-Yuan) Hsieh, MD**

UNIVERSITY OF COLORADO AT DENVER

No relationships to disclose.

### **Vijaya Knight, MD, PhD**

NATIONAL JEWISH HEALTH

No relationships to disclose.

### **Yu Nee Lee, PhD**

BOSTON CHILDREN'S HOSPITAL, HARVARD MEDICAL SCHOOL

No relationships to disclose.

### **Emily M. Mace, PhD**

BAYLOR COLLEGE OF MEDICINE

No relationships to disclose.

### **Rebecca Marsh, MD**

CINCINNATI CHILDREN'S HOSPITAL MEDICAL CENTER

No relationships to disclose.

### **Sara Nandiwada, PhD(ABMLI)**

BAYLOR COLLEGE OF MEDICINE

No relationships to disclose.

### **Maurice O'Gorman, PhD**

UNIVERSITY OF SOUTHERN CALIFORNIA AND THE CHILDREN'S HOSPITAL OF LOS ANGELES

BD Biosciences – Research Grant; Consulting fee; Grant Recipient; Consulting

### **Elena Elizabeth Perez, MD, PhD**

UNIVERSITY OF MIAMI SCHOOL OF MEDICINE

Baxter – Consulting; Consulting fee

Grifols – Consulting; Consulting fee

CSL – Consulting; Consulting fee

### **Sergio D. Rosenzweig, MD, PhD**

NATIONAL INSTITUTES OF HEALTH

No relationships to disclose.

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LOUISIANA STATE UNIVERSITY

No relationships to disclose.

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No relationships to disclose.

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CSL Behring – Research grant and honoraria for consulting and research

### **James Verbsky, MD, PhD**

MEDICAL COLLEGE OF WISCONSIN

Baxalta – Speaking and teaching with speaker fee

# SCHEDULE OF EVENTS

## THURSDAY, APRIL 14

7:00am - 7:40am	<b>Registration</b>	Commonwealth Hall Lobby
7:00am - 8:00am	<b>Continental Breakfast</b>	Amphitheater Lobby
7:40am - 7:45am	<b>Welcome &amp; Introduction</b> <b>Roshini S. Abraham, PhD</b> , Mayo Clinic <b>Troy R. Torgerson, MD, PhD</b> , Seattle Children's Hospital	Amphitheater
7:45am - 9:45am	<b>Diagnostic Approaches to PIDs</b> <i>2 AMA PRA Category 1 Credits™</i>  <i>Moderators:</i> <b>Roshini S. Abraham, PhD</b> , Mayo Clinic <b>James Verbsky, MD, PhD</b> , Medical College of Wisconsin  <i>Speakers:</i> Test Utilization: Selecting and Interpreting Laboratory Tests for PIDs: A Clinician's Perspective <b>Troy R. Torgerson, MD, PhD</b> , Seattle Children's Hospital T Cell Functional Assays: Application and Interpretation <b>Thomas Fleisher, MD</b> , National Institutes of Health  <b>Panel Discussion:</b> <b>Francisco Bonilla, MD, PhD</b> , Children's Hospital Boston <b>Zuhari Ballas, MD</b> , University of Iowa <b>Jordan Orange, MD, PhD</b> , Baylor College of Medicine <b>Elena Perez, MD</b> , University of Miami <b>Ricardo Sorensen, MD</b> , Louisiana State University  <i>Oral Abstract</i> THE USE OF SALMONELLA TYPHI VACCINE TO DIAGNOSE ANTIBODY DEFICIENCY <b>Mary T. Bausch-Jurken, PhD</b> , Medical College of Wisconsin	Amphitheater
9:45am - 10:00am	<b>Morning Break</b>	Amphitheater Lobby
10:00am - 12:00pm	<b>Genomics in PIDs</b> <i>2 AMA PRA Category 1 Credits™</i>  <i>Moderators:</i> <b>Troy R. Torgerson, MD, PhD</b> , Seattle Children's Hospital <b>Jack Bleesing, MD, PhD</b> , Cincinnati Children's Hospital Medical Center  <i>Speakers:</i> Genomics 101: Methods and Interpretation <b>Sergio D. Rosenzweig, MD, PhD</b> , NIAID, NIH Pitfalls of Genomic Testing <b>James Verbsky, MD, PhD</b> , Medical College of Wisconsin Functional Validation of Genomics Data: Clinically Available Approaches <b>David Hagin, MD</b> , Seattle Children's Hospital Functional Validation of Genomics Data: RAG Defects <b>Yu Nee Lee, PhD</b> , Children's Hospital Boston  <i>Oral Abstract</i> ACCURATE COPY NUMBER VARIANT (CNV) ANALYSIS OF IMMUNODEFICIENCY GENES BY NEXT-GENERATION SEQUENCING <b>Gesmar Segundo, MD</b> , Seattle Children's Hospital	Amphitheater

# SCHEDULE OF EVENTS

12:00pm - 1:00pm	<p><b>Lunch Symposium</b> Pneumococcal Antibody Responses: Laboratory Assessment and Clinical Correlation</p> <p><i>Speaker:</i> <b>Anthony Parker</b>, The Binding Site</p> <p><i>This session does not offer CME</i></p>	Harborview Ballroom
1:00pm - 3:00pm	<p><b>Flow Cytometry as Tool for Diagnosis of PIDs</b> 2 AMA PRA Category 1 Credits™</p> <p><i>Moderators:</i> <b>Sergio D. Rosenzweig, MD, PhD</b>, NIAID, NIH <b>Vijaya Knight, MD, PhD</b>, National Jewish Health</p> <p><i>Speakers:</i> An overview of applicability of flow cytometry based on assays in the diagnostic evaluation of primary immunodeficiency <b>Maurice O’Gorman, PhD, MBA, MSc, D(ABMLI)</b>, Children’s Hospital Los Angeles Not Red OR Blue but Many Colors: Vote Flow Cytometry for President! <b>Jack Bleesing, MD, PhD</b>, Cincinnati Children’s Hospital Medical Center Pearls, Perils and Pitfalls of NK Cell Functional Testing <b>Emily Mace, PhD</b>, Baylor University, Texas Children’s Hospital Immune Dysregulation Characterized by Mass Cytometry: Potential Clinical Applications <b>Elena Hsieh, MD</b>, University of Colorado, Denver Emerging Diagnostic Markers for HLH <b>Rebecca Marsh, MD</b>, Cincinnati Children’s Hospital Medical Center</p> <p><i>Oral Abstract</i> IN-VITRO NAÏVE B CELL CLASS SWITCH ASSAY FOR EVALUATION OF B CELL AND HELPER T CELL FUNCTION <b>David Hagin, MD, PhD</b>, University of Washington / Seattle Children’s Hospital</p> <p><b>Panel Discussion</b></p>	Amphitheater
3:00pm - 3:15pm	<b>Afternoon Break</b>	Amphitheater Lobby
3:15pm - 5:15pm	<p><b>Other Laboratory Tools for Diagnosis of PIDs</b> 2 AMA PRA Category 1 Credits™</p> <p><i>Moderators:</i> <b>Maurice O’Gorman, MD, PhD, MBA, MSc, D(ABMLI)</b>, Children’s Hospital Los Angeles <b>Sara Nandiwada, PhD</b>, Baylor College of Medicine</p> <p><i>Speakers:</i> Telomere Syndromes and Telomere Testing in Clinical Settings <b>Mary Armanios, MD</b>, Johns Hopkins Hospital Radiation Sensitivity Testing on Lymphocytes. A Diagnostic Tool to Identify Specific DNA Repair Defects <b>Kerstin Felgentreff, MD</b>, University of Ulm Microbiome and Repertoire Analysis <b>Ying Taur, MD, MPH</b>, Memorial Sloan Kettering Cancer Center</p> <p><i>Oral Abstract</i> T CELL IMMUNOSENESCENCE AND PRIMARY IMMUNODEFICIENCY IN TELOMERASE MUTATION CARRIERS <b>Christa L. Wagner</b>, Johns Hopkins University School of Medicine</p> <p><b>Panel Discussion</b></p>	Amphitheater

## THANK YOU TO OUR SUPPORTERS!

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### ABOUT THE ASSOCIATION OF MEDICAL LABORATORY IMMUNOLOGISTS

The Association of Medical Laboratory Immunologists (AMLI) was organized in 1987 to bring together professional and technical people engaged in the practice and study of medical laboratory immunology. The organization consists of clinical scientists, physicians, dentists, veterinarians, medical technologists, and other professionals engaged in all areas of current laboratory testing. The organization holds an annual meeting where information on all aspects of laboratory immunology are presented including new test development, technical and quality control aspects of current testing, the impact of regulatory issues, new developments in basic immunology and their impact on the practice of clinical immunology, clinical case studies, poster sessions, and round table discussions with experts in the field. The organization's mission is to improve the practice of clinical immunology laboratory testing by encouraging education and training of all individuals in the organization, maximizing the quality of test procedures, promoting the exchange of information, supporting clinical research efforts, and improving patient care.



### ABOUT THE CLINICAL IMMUNOLOGY SOCIETY

The Clinical Immunology Society (CIS), established in 1986, is the key interdisciplinary organization for the field of clinical immunology and is devoted to fostering developments in the science and practice of clinical immunology. CIS is an international professional organization which includes more than 900 clinicians, investigators, and trainees.

The mission of CIS is to facilitate education, translational research and novel approaches to therapy in clinical immunology to promote excellence in the care of patients with immunologic/inflammatory disorders.

The primary objectives and purposes of CIS are to:

- facilitate the interchange of ideas and information;
- promote research and recent advances;
- share knowledge; and
- foster excellence.

# 2016 CIS School in Primary Immunodeficiency Diseases

Thompson Miami Beach • Miami Beach, Florida  
October 6-9, 2016



## Save the Date

### CO-CHAIRS:

Charlotte Cunningham-Rundles, MD, PhD  
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Troy R. Torgerson, MD, PhD  
University of Washington  
Department of Pediatrics  
Seattle, WA

Applications need to be submitted 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;
- Junior faculty members.

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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