



FINAL PROGRAM

2015 Update in Primary Immunodeficiency Diseases

April 9, 2015 | Hilton Americas | Houston, Texas





CIS National Office
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PROGRAM COMMITTEE

Charlotte Cunningham-Rundles, MD, PhD (Co-Chair)

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Kathleen Sullivan, MD, PhD (Co-Chair)

Children's Hospital of Philadelphia

Roshini Sarah Abraham, PhD

Mayo Clinic

Vincent R. Bonagura, MD

Steven & Alexandra Cohen Children's Medical Center of New York

Steven M. Holland, MD

NIAID, NIH

Donald B. Kohn, MD

University of California, Los Angeles

Jordan S. Orange, MD, PhD

Baylor College of Medicine

Troy Torgerson, MD, PhD

University of Washington

ABOUT THE CLINICAL IMMUNOLOGY SOCIETY

The Clinical Immunology Society (CIS), established in 1986, is the key inter-disciplinary 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 600 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.

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 6.5 *AMA PRA Category 1 Credit(s)*[™]. 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/pid2015>

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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 Sarah Abraham, PhD

MAYO CLINIC

There are no relationships to disclose.

Laia Alsina, MD, PhD

HOSPITAL SANT JOAN DE DÉU

There are no relationships to disclose.

Vincent R. Bonagura, MD, FAAAAI

ALEXANDRA & STEVEN COHEN CHILDREN'S MEDICAL CENTER

Baxter – Member on advisory committee receiving Stipend

CSL Behring – Member on advisory committee receiving Stipend

Rebecca Buckley, MD

DUKE UNIVERSITY MEDICAL CENTER

Baxter Healthcare – Principal investigator of Research Grant receiving research grant that pays small percentage of salary at Duke

UCB – Collaborator receiving support for costs of whole exome sequencing and whole genome sequencing

Megan A. Cooper, MD, PhD

WASHINGTON UNIVERSITY SCHOOL OF MEDICINE

There are no relationships to disclose.

Brian Crucian

NASA

There are no relationships to disclose.

Charlotte Cunningham-Rundles, MD, PhD

MT. SINIAL MEDICAL CENTER

ADMA Biologics – Supported Clinical Trials

Baxter – Member on Advisory Committee received Consultant fee

Kedrion – Member on Advisory Committee received Consultant fee

Novartis – Advisor not receiving anything

Regeneron – Consultant not receiving anything

Ivan Fuss, MD

NATIONAL INSTITUTES OF HEALTH

Janssen Pharmaceuticals – Patent issued on use of Anti-IL-12 mAb in the treatment of Crohns disease patients receiving Royalty Payments

Steven M. Holland

NIAID, NIH

There are no relationships to disclose.

Yuval Itan, PhD

THE ROCKEFELLER UNIVERSITY

There are no relationships to disclose.

Michael B. Jordan, MD

CINCINNATI CHILDREN'S HOSPITAL MEDICAL CENTER

There are no relationships to disclose.

Donald B. Kohn, MD

UNIVERSITY OF CALIFORNIA, LOS ANGELES

There are no relationships to disclose.

Tobias Kollmann, MD, PhD

UNIVERSITY OF BRITISH COLUMBIA

There are no relationships to disclose.

Janet Markle, PhD

THE ROCKEFELLER UNIVERSITY

There are no relationships to disclose.

Heardley M. Murdock, BS

NIAID, NIH

There are no relationships to disclose.

Jordan S. Orange, MD, PhD

BAYLOR COLLEGE OF MEDICINE

ADMA Biologics – Consultant receiving Consultant fee

ASD Healthcare – Consultant receiving Consultant fee

Baxter Healthcare – Consultant receiving

Consultant fee; Speaker receiving Speaker fee

CSL Behring – Consultant receiving Consultant

fee; Speaker receiving Speaker fee; Investigator

receiving Research grant

Walgreens – Consultant receiving Consultant fee

Neil Romberg, MD

YALE UNIVERSITY

Baxter – Member on advisory board receiving

Consulting fee

John M. Routes, MD

MEDICAL COLLEGE OF WISCONSIN

There are no relationships to disclose.

Irini Sereti, MD

NATIONAL INSTITUTES OF HEALTH

There are no relationships to disclose.

John H. Stone, MD, MPH

MASACHUSETTS GENERAL HOSPITAL

Genentech – Investigating and Consulting

receiving Grants and Consulting fees

Xencor – Investigating and Consulting receiving

Grants and Consulting fees

Asbjorg Stray-Pedersen, MD

BAYLOR COLLEGE OF MEDICINE

There are no relationships to disclose.

Helen C. Su, MD, PhD

NIAID, NIH

There are no relationships to disclose.

Kathleen E. Sullivan, MD, PhD

CHILDREN'S HOSPITAL OF PHILADELPHIA

Baxter – Primary investigator receiving Grant

Immune Deficiency Foundation – Primary

investigator receiving Grant; Consulting

immunologist receiving Consultant fee

Troy R. Torgerson, MD, PhD

UNIVERSITY OF WASHINGTON

aTyr Pharma – Consultant

Baxter – Consultant, Speaking, and a Member on

Advisory Committee receiving Research grant

CSL Behring – Consultant receiving Research grant

Infant Bacterial Therapeutics - Consultant

QuestCor – Consultant

Stuart E. Turvey, MD, PhD

BRITISH COLUMBIA CHILDREN'S HOSPITAL

There are no relationships to disclose.

David Tweardy, MD

BAYLOR COLLEGE OF MEDICINE

StemMed, Lts. – President and CEO royalty,

intellectual property rights and receiving

Ownership interest

Gulbu Uzel, MD

NIH / NIAID

There are no relationships to disclose.

SCHEDULE OF EVENTS

THURSDAY, APRIL 9

7:00am – 8:00am	Breakfast	Ballroom of the Americas Foyer
8:00am – 10:00am	Welcome 2 AMA PRA Category 1 Credits™	Ballroom of the Americas D&E
8:00am – 8:10am	Charlotte Cunningham-Rundles, MD, PhD, Mt Sinai Medical Center	
	New Diseases	
	<i>Moderator:</i>	
	Troy Torgerson, MD, PhD, University of Washington	
	<i>Speakers:</i>	
8:10am – 8:35am	CTLA4 Haploinsufficiency and Immunodysregulation Gulbu Uzel, MD, NIH	
8:35am – 9:00am	STAT3 Gain-of-Function: A New Syndrome of Early-Onset Autoimmunity Megan Cooper, MD, PhD, Washington University	
9:00am – 9:25am	Primary NLR4-Mediated Disorders Neil Romberg, MD, Yale University	
	<i>Oral Abstracts:</i>	
9:25am – 9:35am	INHERITED IL23R DEFICIENCY CAUSES MYCOBACTERIAL DISEASE Janet Markle, PhD, The Rockefeller University	
9:35am – 9:45am	TRIPEPTIDYL PEPTIDASE II MUTATIONS CAUSE A HUMAN METABOLIC IMMUNODEFICIENCY Helen C. Su, MD, PhD, NIAID, NIH	
9:45am – 10:00am	COMBINED IMMUNODEFICIENCY ASSOCIATED WITH HOMOZYGOUS MALT1 MUTATIONS Stuart E. Turvey, MD, PhD, British Columbia Children's Hospital	
10:05am – 10:30am	Morning Break/Discussion	Ballroom of the Americas Foyer
10:30am – 11:50am	Focus On Therapeutics 1 AMA PRA Category 1 Credit™ <i>Moderator:</i> Donald B. Kohn, MD, University of California, Los Angeles <i>Speakers:</i>	Ballroom of the Americas D&E
10:30am – 11:00am	Modulating STAT Proteostasis to Treat Immunodeficiency Disorders David Twardy, MD, Baylor College of Medicine	
11:00am – 11:20am	IL-7 Immunotherapy in Idiopathic CD4 Lymphopenia Irini Sereti, MD, NIAID, NIH	
11:20am – 11:30am	CVID Therapeutics John Routes, MD, Medical College of Wisconsin	
11:30am – 11:40am	IL-12 Mediated Gastrointestinal Disorder in Common Variable Immune Deficiency Ivan Fuss, MD, NIH	
	<i>Oral Abstract:</i>	
11:40am – 11:50am	GENE THERAPY FOR ADENOSINE DEAMINASE- DEFICIENT SEVERE COMBINED IMMUNE DEFICIENCY (ADA-SCID) Donald B. Kohn, MD, University of California, Los Angeles	

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THURSDAY, APRIL 9

11:50am – 12:30pm	Guided Poster Session <i>Moderator:</i> Roshini Sarah Abraham, PhD, Mayo Clinic Group 1 NEWBORN SCREENING BEYOND TRECs AND KRECs: REACHING OUT FOR PHAGOCYtic DEFECTS, COMPLEMENT AND ANTIBODY DEFICIENCIES AND NEXT-GEN SEQUENCING Stephan Borte, MD, PhD, Translational Centre for Regenerative Medicine Leipzig CYTOMETRY BASED SCREENING FOR GAIN-OF-FUNCTION MUTATION IN SIGNAL TRANSDUCER AND ACTIVATOR OF TRANSCRIPTION 1 (STAT1-GOF) David Hagin, MD, PhD, University of Washington, Seattle Children's Hospital ADULT ONSET MYCOBACTERIAL INFECTION DUE TO NON-CODING MUTATIONS IN IKK β AFFECTING NEMO PROTEIN LEVELS Amy P. Hsu, BA, NIAID PERFORIN-2 MUTATIONS IN PULMONARY NONTUBERCULOUS MYCOBACTERIAL INFECTION Ryan McCormack, BS, University of Miami PRELIMINARY DATA FOR mTOR INHIBITOR TREATMENT IN PATIENTS WITH GAIN-OF-FUNCTION MUTATIONS IN PIK3CD Kelli W. Williams, MD, MPH, NIAID, NIH <i>Moderator:</i> Troy Torgerson, MD, PhD, University of Washington Group 2 PATIENTS WITH CARTILAGE-HAIR HYPOPLASIA DISPLAY TELOMERE LENGTH DEFICIENCY COMPARED TO RMRP MUTATION HETEROZYGOtes AND NORMAL CONTROLS Geraldine Aubert, PhD, BC Cancer Research Centre SMITH MAGENIS SYNDROME: CLINICAL IMMUNOLOGIC FEATURES OF 70 PATIENTS CARRYING A SINGLE TAC1 ALLELE Tiffany Perkins, MD, Yale New Haven Hospital ESTIMATED DISEASE INCIDENCE OF RAG1 MUTATION BY QUERYING THE EXOME AGGREGATION CONSORTIUM Jolan E. Walter, MD, PhD, Boston Children's Hospital TREATMENT OF CHRONIC GRANULOMATOUS DISEASE ASSOCIATED COLITIS WITH ANAKINRA Christa S. Zerbe, MD, NIAID, NIH	Ballroom of the Americas A-C
12:30pm – 1:30pm	Lunch/Poster Viewing/Exhibit Hall Open	Ballroom of the Americas A-C
1:30pm – 2:40pm	Misguided Cells 1 AMA PRA Category 1 Credit™ <i>Moderator:</i> Vincent Bonagura, MD, Steven and Alexandra Cohen Children's Medical Center of New York <i>Speakers:</i> 1:30pm – 1:50pm IgG4-Related Disease: How Did We Miss This for so Long? John Stone, MD, MPH, Massachusetts General Hospital 1:50pm – 2:10pm Troy Torgerson, MD, PhD, University of Washington 2:10pm – 2:30pm Inflammatory Innate Lymphoid Cells in CVID Charlotte Cunningham-Rundles, MD, PhD, Mt Sinai Medical Center <i>Oral Abstract:</i> 2:30pm – 2:40pm DOCK8 REGULATES LYMPHOCYTE MIGRATION AND INFLUENCES THE CYTOSKELETON THROUGH DIVERSE RHO GTPASE FAMILY EFFECTORS Hardley M. Murdock, BS, NIAID, NIH	Ballroom of the Americas D&E

SCHEDULE OF EVENTS

THURSDAY, APRIL 9

2:45pm – 3:00pm	Afternoon Break	Ballroom of the Americas Foyer
3:00pm – 5:00pm	Responding to the World Around Us 2 AMA PRA Category 1 Credits™ <i>Moderator:</i> Steven M. Holland, MD, NIAID, NIH <i>Speakers:</i> 3:00pm – 3:30pm Responses to Bacteria in Patients with MYD88 or IRAK4 Deficiencies Laia Alsina, MD, Hospital Sant Joan de Déu, Barcelona 3:30pm – 4:00pm Off-Target Benefits of Vaccination Tobias Kollmann, MD, PhD, University of British Columbia <i>Oral Abstracts:</i> 4:00pm – 4:10pm THE MUTATIONAL DAMAGE LANDSCAPE ACROSS THE HUMAN GENOME: A GENE-LEVEL APPROACH TO THE SELECTION OF CANDIDATE DISEASE-CAUSING VARIANTS Yuval Itan, PhD, The Rockefeller University 4:10pm – 4:20pm MANIPULATING DNA DAMAGE RESPONSE PATHWAYS FOR THE TREATMENT OF IMMUNE DISEASES Michael B. Jordan, MD, Cincinnati Children's Hospital Medical Center 4:20pm – 4:30pm WHOLE EXOME SEQUENCING DETECTS DUAL MOLECULAR DIAGNOSES AND UNEXPECTED SYNDROMES IN PATIENTS WITH PRIMARY IMMUNODEFICIENCY DISEASES Asbjorg Stray-Pedersen, MD, Baylor College of Medicine <i>Speakers:</i> 4:30pm – 5:00pm Immune System Dysregulation Persists During Long-Duration Spaceflight Brian Crucian, NASA, Johnson Space Center	Ballroom of the Americas D&E
5:00pm – 5:30pm	Richard Schiff Lecture .5 AMA PRA Category 1 Credit™ <i>Moderator:</i> Jordan Orange, MD, PhD, Baylor College of Medicine <i>Speaker:</i> Unlocking Secrets from Cryopreserved Cells Rebecca Buckley, MD, Duke University	Ballroom of the Americas D&E

THANK YOU TO OUR SUPPORTERS!

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CIS 2015 School in Primary Immunodeficiency Diseases

October 15-18, 2015 • Thompson Miami Beach
Miami Beach, Florida



Calling all Fellows!

Submit an application today
Submission Deadline April 30, 2015

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*

Faculty:

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*CHU Ste-Justine, University of Montreal
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Jordan S. Orange, MD, PhD
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Steven M. Holland, MD
*NIAID, NIH
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*National Institutes of Health
Bethesda, MD*

Hans D. Ochs, MD
*University of Washington
Seattle, WA*

Troy R. Torgerson, MD, PhD
*University of Washington
Seattle, WA*

Applications need to be submitted 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;
- 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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