



CIS Clinical
Immunology
Society

2016 Annual Meeting

*Immune Deficiency & Dysregulation
North American Conference*

April 14-17, 2016 • Seaport Hotel • Boston, Massachusetts

Final Program

www.clinimmsoc.org/am16

ABOUT CIS

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 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 among physicians and other investigators who are concerned with immunological/inflammatory diseases;
- promote research on the causes and mechanisms of diseases relating to the immune system and, as a result, to unify concepts of disease pathogenesis;
- encourage investigators and clinicians to share in their knowledge of immunologically active drugs and other interventions;
- promote application and dissemination of recent advances in biomedical science for the prevention, diagnosis and treatment of diseases related to immunity and inflammation; and
- foster excellence in research and medical practice.

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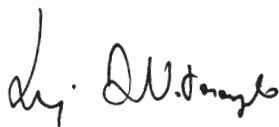
Dear Colleagues,

On behalf of the Clinical Immunology Society (CIS), thank you for participating in the 2016 Annual Meeting: Immune Deficiency & Dysregulation North American Conference taking place April 14-17, 2016 at the Seaport Hotel & World Trade Center in Boston, Massachusetts.

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

The CIS leadership and the members of the Program Committee have jointly defined our goals and have engaged clinicians and researchers from all types of backgrounds to try to identify the best manner of addressing the critical needs. The primary goal of the Annual Meeting is to provide an exciting venue for the presentation of the newest immune deficiency diseases and the pathogenesis of these defects in molecular terms. The substance of the conference will be cutting edge clinical and basic science research. Another goal is to educate clinicians regarding the state of the art clinical care. We recognize that physicians in training who are likely to see these patients need sessions on the pragmatic aspects of diagnosis and clinical care; thus sessions were designed to provide a stimulating discussion of these issues, while delving into unsolved questions. The final major goal of this conference is to begin to grow the community of physicians who feel competent to see patients with primary immune deficiency. We believe that this is the only way to consolidate and build up a community of physicians and scientists who are working in this area. This Annual Meeting is sure to foster enthusiasm and provide advanced training for physicians and researchers working in the field of primary immune deficiency.

The CIS leadership is excited about the 2016 Annual Meeting and we look forward to bringing accomplished clinical researchers together to present what is sure to be outstanding science. We hope you enjoy the me



Luigi D. Notarangelo, MD
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2016 Program Committee Co-Chair



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2016 Program Committee Co-Chair

Submit a Manuscript

The *Journal of Clinical Immunology* accepts manuscripts in the areas of human, basic, and clinical immunology and molecular biology. The areas of basic immunology include (but are not limited to) studies of lymphocytes, antigen presenting cells, neutrophils, natural killer cells, complement components, immunoglobulins, antibodies, cytokines and their receptors, immunoregulation, signal transduction, T-cell receptors, and immunoglobulin genes. Papers on animal models of human diseases are welcome. Articles dealing with molecular biology related to human diseases are accepted. Special Articles are by invitation only. Case reports will only be considered if they are linked to novel new findings/science or are accompanied by an extensive review of the relevant medical literature to the findings of the case report.

Authors, editors and reviewers of *Journal of Clinical Immunology* use our fully web-enabled online manuscript submission and review system. To keep the review time as short as possible, we request authors to submit manuscripts online to the journal's editorial office. Our online manuscript submission and review system offers authors the option to track the progress of the review process of manuscripts in real time.

About the Journal

The *Journal of Clinical Immunology* is a bimonthly international journal that helps researchers and academic clinicians keep current on investigative basic immunology and diseases related to the immune system. This well-established journal publishes articles on basic, translational, and clinical studies in all aspects of immunology, including animal models of human diseases. The OnlineFirst™ feature allows articles to be viewed through the journal's website before they are available in the printed journal. The *Journal of Clinical Immunology* is the official journal of the Clinical Immunology Society.



To submit a manuscript, please visit <http://www.editorialmanager.com/joci/>

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MEETING LOCATION

All educational sessions, posters, exhibits, and registration held as part of the CIS Annual Meeting will take place at the Seaport World Trade Center.

Seaport World Trade Center
200 Seaport Boulevard
Boston, MA 02210
P: (617) 385-5000

Registration Desk

Seaport World Trade Center ~ Commonwealth Hall Lobby

Registration Desk Hours

Wednesday, April 13	4:00pm – 7:00pm
Thursday, April 14	7:00am – 8:00pm
Friday, April 15	7:00am – 6:30pm
Saturday, April 16	7:00am – 7:00pm
Sunday, April 17	7:00am – 10:00am

EXHIBIT HALL

Seaport World Trade Center ~ Commonwealth Hall ABC

Exhibitors provide the latest information on products and services available to physicians, researchers and allied health professionals in the field of immunology. Take this valuable opportunity to meet with companies and organizations specializing in all areas of the immunology field.

Exhibitor Move-In

Thursday, April 14th
8:00am – 6:00pm

Exhibit Hours

Thursday, April 14th
7:00pm – 8:30pm

Friday, April 15th
7:00am – 8:00 am & 12:30pm – 1:45pm

Saturday, April 16th
7:00am – 8:00am & 12:45pm – 1:45pm

Exhibitor Move-Out

Saturday, April 16th
2:00pm – 7:00pm

POSTER SESSIONS

Poster sessions are held in the Commonwealth Hall ABC, and authors will be present with their posters during hall hours. Lunch will be available during the afternoon poster session hours and hors d'oeuvres and an open bar are available during the Opening Reception.

PUBLISHING OF ABSTRACTS

The abstracts submitted to the 2016 Annual Meeting are published in the April issue of *Journal of Clinical Immunology*, the official journal of the Clinical Immunology Society. Copies are available at the CIS registration desk.

ACCESSIBILITY

If you require special arrangements in order to fully participate in the CIS Annual Meeting, please speak with a CIS staff member at the registration desk.

NAME BADGES

All registered attendees at the CIS Annual Meeting will receive a name badge as part of their registration packet. These badges should be worn at all times as they will be used to control access to sessions and activities.

SMOKING

Smoking is prohibited at all CIS Annual Meeting sessions and events.

EVALUATIONS

Delegates will have the opportunity to provide feedback on their experience at the 2016 CIS Annual Meeting. After the meeting all registered delegates will receive an online evaluation to complete. Evaluations are an important part in helping us to improve our educational sessions so please take a moment to complete it!

LISTSERV

CIS-PIDD List Service & ECI List Service

The E-mail CIS-PIDD service has been set up to provide an easy way for CIS members to discuss issues of common interest. The E-mail ECI List Service has been set up to provide fellows and junior faculty a dedicated place to discuss issues with each other; it is limited to Fellows-in-Training, Junior Faculty and a few select Senior Faculty members for mentor purposes. The server group may also be used to look for collaborators or new faculty members. These List Services are set up as a communication tool to help you interact with your colleagues. It is closed to the general public. Visit www.clinimmsoc.org to sign up.

CONTINUING MEDICAL EDUCATION INFORMATION

PURPOSE

Provide an opportunity for physicians to receive updated information and solidify their knowledge of primary immune deficiency.

CONTENT AREA

The content will cover practical, cutting edge and theoretical issues related to patient care. The majority of the 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 should be pediatricians, internists, or scientists committed to the clinical management of patients with primary immunodeficiency diseases. These candidates may have backgrounds in immunology, adult medicine, rheumatology, hematology, laboratory immunology, or allergy/immunology.

LEARNING OBJECTIVES

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

- Recognize the value of new research techniques;
- Describe new research findings in immunology;
- Explain the diagnosis and management of diseases and disorders across various subspecialties of medicine.

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 21.75 *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/16>.



DISCLOSURES OF CONFLICT OF INTEREST

2016 ANNUAL MEETING PROGRAM COMMITTEE AND SPEAKER DISCLOSURES

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.

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Gerson–Lehrman Group – Consultant receiving consulting fee

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royalties

BOSTON VISITOR INFORMATION

THINGS TO DO:

The Seaport Hotel is located in the South Boston Waterfront neighborhood, where the city meets its harbor. Check out the many nearby attractions, local bars, tasty eats, and boutique shopping.

The Institute of Contemporary Art/Boston

This museum was founded in 1936 as the Boston Museum of Modern Art and boasts striking contemporary art and performance space with glass filled walls overlooking the Harbor.

Boston Tea Party Ships and Museum

Relive the most important event leading up to the American Revolution. The museum experience will transport you on an incredible journey back in time as you take part in the famous event – meet the colonists, explore the ships and dump tea overboard!

Boston Common

Visit the nearly 50-acre central public park in downtown Boston that hosted British troops during the American Revolution. Dating from 1634, it is the oldest city park in the United States.

Old State House

The original seat of colonial government and later state capitol, this is a National Historic Treasure and the location of the Boston Massacre which occurred on March 5, 1770.

New England Aquarium

With over 2,000 aquatic animals in the four-story Giant Ocean Tank, this aquarium is sure to not disappoint. Mingle with the turtles, seals, penguins, and balloonfish in the various exhibits they have to offer.

Copley Square

Named for the painter John Singleton Copley, this public square in Boston's Back Bay neighborhood features lawns, a fountain, and is surrounded by the Copley Plaza with high-end shopping in the Shops at Prudential Center.

Fenway Park

People say, no matter which team you are a fan of, there is no better place to watch a baseball game than Fenway Park. With iconic features like the Green Monster, "Pesky's Pole", The Lone Red Seat, and "Duffy's Cliff," it's a must see for sports fanatics.

Museum of Science (MoS)

Located on a plot in Science Park (a plot land spanning the Charles River), this museum has over 700 interactive exhibits and several live presentations every day.

The Freedom Trail

Go on a historical adventure! Follow the 2.5-mile, red-lined route that leads you to 16 historically significant sites. Join one of the daily guided tours or walk it out on your own.

Spirit of Boston

Cruise around the harbor and enjoy the perfect mix of dining, dancing, entertainment and views. The ship was recently treated to a \$2.5 million revolution to give you a whole new experience to the city!

PLACES TO DINE:

Morton's The Steakhouse (0.0 miles)

P: (617) 526-0410

Steakhouse

Open for lunch and dinner

Upscale chain for aged prime beef, seafood and other traditional steakhouse fare in a clubby space.

Temazcal Tequila Cantina (0.1 miles)

P: (617) 439-3502

Mexican

Open for lunch and dinner

Elegantly chic restaurant offering elevated Mexican cuisine along with margaritas and 250+ tequilas.

The Whiskey Priest (0.1 miles)

P: (617) 426-8111

American

Open for lunch and dinner

Big, boisterous hangout with spectacular views starring mega whiskey selection and pub eats.

Legal Test Kitchen (0.1 miles)

P: (617) 330-7430

Seafood

Open for lunch and dinner

Legal Sea Foods offshoot offering inventive, eclectic surf'n'turf in a chic atmosphere.

Salvatore's (0.2 miles)

P: (617) 737-5454

Italian

Open for lunch and dinner

Contemporary Italian chain serving casual eats, including hand-tossed pizza, in sophisticated digs.

No Name Restaurant (0.2 miles)

P: (617) 423-2705

Seafood

Open for lunch and dinner

Popular, rustic landmark serving New England seafood classics on historic Fish Pier since 1917.

2016 CIS ANNUAL MEETING SCHEDULE AT-A-GLANCE

	Congress Board Room	Show Office 2	Commonwealth Hall ABC	Beacon Hill	Commonwealth Hall DE	Harborview Ballroom	Commonwealth Hall Lobby	Amphitheater	Amphitheater Lobby	
Wednesday, April 13										
4:00 pm - 8:00pm		Speaker ReadyRoom								
4:00pm - 7:00pm							Registration Open			
5:00pm - 7:00pm										CIS Council Meeting
Thursday, April 14										
8:00am - 11:30am			Exhibitor Move-in							
7:00am - 7:30pm		Speaker Ready Room								
7:00am - 8:00pm							Registration Open			
7:00am - 7:40am	Breakfast									
7:40am - 5:15pm							Update in PID			
9:45am-10:00am	Morning Break									
10:00am-12:00pm							Genomics in PIDs			
12:00pm - 1:00pm						Sponsored Lunch Symposium				
1:00pm-3:00pm							Flow Cytometry as a Tool for Diagnosis of PIDs			
3:00pm-3:15pm	Afternoon Break									
3:15pm-5:15pm							Other Laboratory Tools for Diagnosis of PIDs			
6:00pm-6:10pm							Welcome from CIS President			
6:10pm-7:00pm							Robert A. Good Lecture			
7:00pm-8:30pm			Opening Reception							

2016 CIS ANNUAL MEETING SCHEDULE AT-A-GLANCE

	Congress Board Room	Show Office 2	Commonwealth Hall ABC	Beacon Hill	Commonwealth Hall DE	Harborview Ballroom	Commonwealth Hall Lobby	Amphitheater	Amphitheater Lobby	
Friday, April 15										
7:00am - 6:30pm		Speaker Ready Room								
7:00am - 8:00am					Breakfast					
7:00am - 6:30pm							Registration Open			
8:00am - 10:00am					Immune Dysregulation					
10:00am - 11:00am						Morning Break & Guided Poster Session				
11:00am-12:30pm					Tales from the Listserv					
12:30pm-1:45pm										Lunch/Poster Viewing/Exhibit Hall Open
1:45pm-3:45pm					Cell Perturbation and Plasticity in the Immune System					
3:45pm-4:15pm					Afternoon Break					
4:15pm-5:45pm					Gene Therapy/ Editing					
5:45pm-6:00pm					Late Afternoon Break					
6:00pm-7:00pm					The CIS Fahey/ Rose Founders Lecture					
7:15pm-8:45pm							Measuring Quality, Patient Centered Outcomes and Special Populations in Primary Immunodeficiencies			

2016 CIS ANNUAL MEETING SCHEDULE AT-A-GLANCE

	Congress Board Room	Show Office 2	Commonwealth Hall ABC	Beacon Hill	Commonwealth Hall DE	Harborview Ballroom	Commonwealth Hall Lobby	Amphitheater	Amphitheater Lobby	
Saturday, April 16										
7:00am - 7:00pm		Speaker Ready Room								
7:00am-8:00am					Breakfast					
7:00am - 7:00pm							Registration Open			
8:00am-9:30am					Plenary HCT in PID-Old and New Indications					
9:30am-10:15am					CIS Business Meeting & Award Presentation					
10:15am-11:15am						Morning Break & Guided Poster Session				
11:15am-12:45pm					Antibody Deficiencies					
12:45pm-1:45pm										Lunch/Poster Viewing/Exhibit Hall Open
1:45pm-3:15pm					Immunoglobulin: What it Can and Can't Do					
2:00pm-7:00pm										Exhibit Hall Tear Down
3:15pm-3:45pm					Afternoon Break					
3:45pm-5:45pm					T Cell Ontogeny/ Development in Health and Disease					
5:45pm-6:45pm					Richard Schiff Lecture					
6:45pm-8:30pm						CIS Closing Reception				
Sunday, April 17										
7:00am - 12:00pm		Speaker Ready Room								
7:00am - 8:00am					Breakfast					
7:00am - 10:00am							Registration Open			
8:00am - 10:00am					Hot New Defects					
10:00am-12:00pm					Hot New Treatments					

SCHEDULE OF EVENTS

THURSDAY, APRIL 14

6:00pm - 6:10pm	Welcome from CIS President Luigi D. Notarangelo, MD Children's Hospital Boston	Amphitheater
6:10pm - 7:00pm	Robert A. Good Lecture .75 AMA PRA Category 1 Credits™ <i>Moderator:</i> Luigi D. Notarangelo, MD , Children's Hospital Boston <i>Speaker:</i> Immune Response Genes and Human Diseases Alain Fischer, MD, PhD , The Imagine Institute	Amphitheater
7:00pm - 8:30pm	Opening Reception	Commonwealth Hall ABC

FRIDAY, APRIL 15

7:00am - 8:00am	Continental Breakfast	Commonwealth Hall E
8:00am - 10:00am	Plenary Session: Immune Dysregulation 2 AMA PRA Category 1 Credits™ <i>Moderators:</i> Troy R. Torgerson, MD, PhD , Seattle Children's Hospital Jolan Walter, MD, PhD , Massachusetts General Hospital <i>Speakers:</i> What's New Up in the Aire Mark Anderson, MD, PhD , University of California, San Francisco APECED in North America: A syndrome highly-enriched for non-endocrine autoimmune manifestations Michail Lionakis, MD, ScD , National Institutes of Health Age-Related Immune Dysregulation Diane Mathis, PhD , Harvard Medical School Metabolic View of the Immune System Talal Chatila, MD, PhD , Children's Hospital Boston <i>Oral Abstract</i> IDENTIFICATION OF TYPE I INTERFERONOPATHIES IN CHILDREN WITH EARLY-ONSET SLE AND VASCULOPATHY Stefano Volpi, PhD , University Hospital of Lausanne	Commonwealth Hall DE
10:00am - 11:00am	Morning Break & Guided Poster Sessions <i>Moderator:</i> Roshini Sarah Abraham, PhD , Mayo Clinic GROUP 1 <i>MYELOID DENDRITIC CELLS DEFECTS CAUSED BY INHERITED SPPL2a DEFICIENCY UNDERLIES MYCOBACTERIAL DISEASE</i> Ruben Martinez-Barricarte , The Rockefeller University <i>ALTERED INVARIANT NKT:B CELL HELP AND DECREASED SAP EXPRESSION IN BLOOD LYMPHOCYTES FROM PATIENTS WITH COMMON VARIABLE IMMUNODEFICIENCY</i> Lucia Erazo Borrás, MSc , University of Antioquia <i>FATAL HYPERINFLAMMATION IN MURINE MODEL OF LEAKY SCID/OMENN SYNDROME: INNATE OVERACTIVATION LOCALIZED IN SMALL AND LARGE INTESTINES</i> Krisztian Csomos, PhD , Massachusetts General Hospital Harvard Medicine School <i>IMPAIRED RASGRF1/ERK-MEDIATED GM-CSF RESPONSE CHARACTERIZES CARD9 DEFICIENCY IN FRENCH-CANADIANS</i> Donald C. Vinh, MD , McGill University Health Centre-Research Institute <i>NOVEL HETEROZYGOUS RAC2 MUTATION PRESENTING AS COMBINED IMMUNODEFICIENCY</i> Amy Hsu , National Institutes of Allergy & Infectious Diseases (NIAID)	Beacon Hill

SCHEDULE OF EVENTS

FRIDAY, APRIL 15, CONTINUED

	<p><i>Moderator:</i> Tamara Pozos, MD, PhD, Children's Hospitals & Clinics of Minnesota GROUP 2 <i>APOPTOSIS RESISTANCE AND INTRINSIC DIFFERENTIATION DEFECTS IN ACTIVATED BENTA PATIENT B CELLS</i> Andrew L. Snow, PhD, Uniformed Services University of Health Sciences <i>INFLUENCE OF BAFF/BAFF-R AXIS UPON COMMON VARIABLE IMMUNODEFICIENCY INTERSTITIAL LUNG DISEASE</i> Paul J. Maglione, MD, PhD, Icahn School of Medicine at Mount Sinai <i>A PATHOGENIC HYPOMORPHIC MUTATION IN LRRC8A IMPAIRS VOLUME REGULATED ANION CHANNEL ACTIVITY BUT SPARES T CELL DEVELOPMENT AND FUNCTION</i> Craig D. Platt, MD, PhD, Boston Children's Hospital, Harvard Medical School <i>NK CELLS FROM PATIENTS WITH RAG AND ARTEMIS DEFICIENCY HAVE AN IMMATURE PHENOTYPE AND DISPLAY INCREASED DEGRANULATION CAPACITY – IMPLICATIONS FOR HEMATOPOIETIC CELL TRANSPLANTATION</i> Luigi D. Notarangelo, MD, Boston Children's Hospital</p>	Beacon Hill
11:00am - 12:30pm	<p>Plenary Session: Tales from the Listserv 1.5 AMA PRA Category 1 Credits™ <i>Moderators:</i> Elie Haddad, MD, PhD, CHU Ste-Justine, University of Montreal Mikko Seppänen, MD, PhD, Helsinki University Central Hospital <i>Speakers:</i> Mystery Case #1 Joshua D. Milner, MD, NIH/NIAID Mystery Case #2 Juan Carlos Aldave, MD, Hospital Nacional Edgardo Rebagliati Martins</p>	Commonwealth Hall DE
12:30pm - 1:45pm	Lunch / Poster Viewing / Exhibit Hall Open	Commonwealth Hall ABC
1:45pm - 3:45pm	<p>Plenary Session: Cell Perturbation & Plasticity in the Immune System 2 AMA PRA Category 1 Credits™ <i>Moderators:</i> Jordan Orange, MD, PhD, Baylor College of Medicine Emily Mace, PhD, Baylor College of Medicine <i>Speakers:</i> Very Early Onset Inflammatory Bowel Disease – From Genes to Function Scott B. Snapper, MD, Children's Hospital Boston Imaging Immunity: Creating a Spatiotemporal Understanding of Host Defense Ronald N. Germain, MD, PhD, National Institutes of Health Dissecting the Heterogeneity of Human Memory T Helper Cells Federica Sallusto, PhD, Institute for Research in Biomedicine Mitophagy Promotes the Generation of Natural Killer Cell Memory Joseph Sun, PhD, Memorial Sloan Kettering Cancer Center</p>	Commonwealth Hall DE
3:45pm - 4:15pm	Afternoon Break	

SCHEDULE OF EVENTS

FRIDAY, APRIL 15, CONTINUED

4:15pm - 5:45pm	Plenary Session: Gene Therapy/Editing 1.5 AMA PRA Category 1 Credits™ Moderators: Caroline Kuo, MD , UCLA Medical Center David Hagin, MD, PhD , University of Washington/Seattle Children's Hospital Speakers: Efficacy and Safety of gene therapy approach to Wiskott-Aldrich Syndrome and X-linked Severe Combined Immunodeficiency David A. Williams, MD , Children's Hospital Boston Donald B. Kohn, MD , University of California, Los Angeles Breaking Bad: Genome-wide Detection of Designer Nuclease Targeting and its Ensuing Collateral Damage Richard Frock, PhD , Children's Hospital Boston Oral Abstracts TARGETED GENE EDITING RESTORES REGULATED CD40L EXPRESSION AND FUNCTION IN X-HIGM T CELLS Nicholas W. Hubbard , Seattle Children's Research Institute REVERSING EPIGENETIC GENE SILENCING IN COMMON VARIABLE IMMUNODEFICIENCY M. Julia B. Felipe, MedVet, MS, PhD, DACVIM , Cornell University College of Veterinary Medicine	Commonwealth Hall DE
5:45pm - 6:00pm	Late Afternoon Break	
6:00pm - 7:00pm	The CIS Fahey/Rose Founders Lecture 1 AMA PRA Category 1 Credits™ Moderator: Luigi D. Notarangelo, MD , Children's Hospital Boston Speaker: Emerging Infectious Diseases Steven M. Holland, MD , NIAID, NIH	Commonwealth Hall DE
7:15pm - 8:45pm	Measuring Quality, Patient-Centered Outcomes and Special Populations in Primary Immunodeficiencies 1.5 AMA PRA Category 1 Credits™ Moderator: Kathleen E. Sullivan, MD, PhD , Children's Hospital of Philadelphia Speakers: Disease-Specific Health-Related Quality of Life Outcomes in Patients with Antibody Immune Deficiency Mark Ballow, MD, FAIAA , University of South Florida Primary Immunodeficiencies in the Old Order Amish & Mennonite Communities Nicholas Rider, MD , Baylor College of Medicine SCID in the Navajo: A Founder Population in the US Jennifer M. Puck, MD , University of California, San Francisco Latin American Challenges with Diagnoses and Treatment of PID Beatriz Costa-Carvalho, MD , Federal University of Sao Paulo, Brazil Supported by an Educational Grant from Baxalta.	Harborview Ballroom
9:00pm - 10:00pm	Early Career Immunologists (ECI) Reception	Tamo Loft Room

SCHEDULE OF EVENTS

SATURDAY, APRIL 16

7:00am - 8:00am	Continental Breakfast	Commonwealth Hall E
8:00am - 9:30am	<p>Plenary Session: HCT in PID - Old & New Indications 1.5 AMA PRA Category 1 Credits™ Moderators: Donald Kohn, MD, University of California, Los Angeles Jennifer Heimall, MD, The Children's Hospital of Philadelphia</p> <p>Speakers: The Primary Immune Deficiency Treatment Consortium: How Can it Improve Definitive Therapy for PID? Morton J. Cowan, MD, University of California, San Francisco Transplanting New Indications: NEMO Deficiency Bertrand Boisson, PhD, The Rockefeller University HSCT for STAT3 and CTLA4 Deficiency – Why, How and When Andrew Gennery, MD, Newcastle University STAT1 GOF Jen Leiding, MD, University of South Florida STAT3 GOF Lisa Forbes, MD, Baylor College of Medicine</p> <p>Oral Abstracts A SINGLE CENTRE COHORT REPORT OF LONG TERM CLINICAL OUTCOME OF SEVERE COMBINED IMMUNODEFICIENCY FOLLOWING HAEMATOPOIETIC STEM CELL TRANSPLANTATION Intan Juliana Abd Hamid, Institute of Cellular Medicine, Newcastle University A PROSPECTIVE ALEMTUZUMAB PHARMAKOKINETIC (PK) STUDY REVEALS THAT PERI-TRANSPLANT ALEMTUZUMAB LEVELS DEPEND ON ABSOLUTE LYMPHOCYTE COUNT AND UNDERLYING PRIMARY IMMUNE DEFICIENCY Rebecca Marsh, MD, Cincinnati Children's Hospital Medical Center</p>	Commonwealth Hall DE
9:30am - 10:15am	CIS Business Meeting & Award Presentations	Commonwealth Hall DE
10:15am - 11:15am	<p>Morning Break & Guided Poster Sessions Moderator: Roshini Sarah Abraham, PhD, Mayo Clinic GROUP 1</p> <p>INHERITED IL-12Rβ2 DEFICIENCY CAUSES MYCOBACTERIAL SUSCEPTIBILITY Ruben Martinez-Barricarte, The Rockefeller University</p> <p>HUMAN T FOLLICULAR HELPER CELLS IN PRIMARY IMMUNODEFICIENCY: QUALITY JUST AS IMPORTANT AS QUANTITY Cindy S. Ma, PhD, Garvan Institute of Medical Research</p> <p>DEFECTS IN HUMAN B-CELL DEVELOPMENT AND DIFFERENTIATION DUE TO DISEASE-CAUSING MUTATIONS IN THE PI3K PATHWAY Elissa Deenick, Garvin Institute of Medical Research</p> <p>NOVEL STAT1 GOF MUTATION AFFECTING PROTEIN SUMOYLATION Elizabeth P. Sampaio, MD, PhD, NIAID/NIH</p> <p>A NOVEL DONOR SPLICE-SITE MUTATION CAUSING CTLA4 HAPLO-INSUFFICIENCY AND SEVERE AUTOIMMUNITY David Hagin, MD, PhD, University of Washington/Seattle Children's Hospital</p>	Beacon Hill

SCHEDULE OF EVENTS

SATURDAY, APRIL 16, CONTINUED

	<p><i>Moderator:</i> Tamara Pozos, MD, PhD, Children's Hospitals & Clinics of Minnesota GROUP 2 <i>SELECTIVE DEFICIENCY OF WASP IN TREG CELLS IS SUFFICIENT TO CAUSE AUTOIMMUNITY IN MICE</i> Stefano Volpi, PhD, University Hospital of Lausanne <i>IMMUNODEFICIENCY SYNDROME ASSOCIATED WITH HAPLOINSUFFICIENCY OF NFKB1 SUBUNIT p50: REPORT OF 5 IDENTIFIED CASES</i> Elizabeth Feuille, MD, Icahn School of Medicine at Mount Sinai <i>MULTI-LINEAGE ANALYSIS OF X-INACTIVATION IN 54 FEMALES CARRIER OF GENETIC ALTERATIONS IN THE WISKOTT-ALDRICH SYNDROME PROTEIN (WASP) GENE LOCUS</i> Silvia Giliani, PhD, University of Brescia <i>X-LINKED CARRIERS OF CHRONIC GRANULOMATOUS DISEASE</i> Betty Marciano, MD, NIAID/NIH</p>	
11:15am - 12:45pm	<p>Plenary Session: Antibody Deficiencies 1.5 AMA PRA Category 1 Credits™ <i>Moderators:</i> Vincent Bonagura, MD, Children's Medical Center of New York Joyce Yu, MD, Morgan Stanley Children's Hospital/Columbia University Medical <i>Speakers:</i> The Pathogenesis of IgG4-related disease Shiv Pillai, MD, PhD, Harvard Medical School Shutting off Autoimmunity with anti-CD21 Therapy Michael C. Carroll, MD, PhD, Children's Hospital Boston Defects in Transcription Factors Resulting in Antibody Deficiency Mary Ellen Conley, MD, The Rockefeller University S-6-Kinopathies Bodo Grimbacher, MD, University of Freiburg <i>Oral Abstract</i> RITUXIMAB AND HYPOGAMMAGLOBULINEMIA – COMPARATIVE OUTCOMES AT MASSACHUSETTS GENERAL HOSPITAL Sara Barmettler, MD, Massachusetts General Hospital</p>	Commonwealth Hall DE
12:45pm - 1:45pm	Lunch / Poster Viewing / Exhibit Hall Open	Commonwealth Hall ABC
1:45pm - 3:15pm	<p>Plenary Session: Immunoglobulin: What it Can and Can't Do 1.5 AMA PRA Category 1 Credits™ <i>Moderators:</i> Charlotte Cunningham-Rundles, MD, PhD, Mount Sinai Medical School Kathleen E. Sullivan, MD, PhD, Children's Hospital of Philadelphia <i>Panelists:</i> Zuhair Ballas, MD, University of Iowa Healthcare Francisco Bonilla, MD, PhD, Children's Hospital Boston Kenneth Paris, MD, LSU Health Sciences Center</p>	Commonwealth Hall DE
3:15pm - 3:45pm	Afternoon Break	

SCHEDULE OF EVENTS

SATURDAY, APRIL 16, CONTINUED

3:45pm - 5:45pm	Plenary Session: T Cell Ontogeny/Development in Health and Disease 2 AMA PRA Category 1 Credits™ <i>Moderators:</i> Jennifer Puck, MD , University of California, San Francisco Shanmuganathan Chandrakasan, MD , Emory University School of Medicine, Children's Healthcare of Atlanta <i>Speakers:</i> Development Cues in Thymopoiesis Georg Holländer, MD , University of Basel, Switzerland Long Term Outcomes of Patients with SCID Elie Haddad, MD, PhD , CHU Ste-Justine, University of Montreal Following Infants with Low (T Cell) Lymphocytes: FILL Project John M. Routes, MD , Medical College of Wisconsin A Patient with a CD4 Gene Mutation and Immunodeficiency Jose Luis Franco, MD, PhD , University of Antioquia	Commonwealth Hall DE
5:45pm - 6:45pm	Richard Schiff Lecture 1 AMA PRA Category 1 Credits™ <i>Moderator: Jordan Orange, MD PhD</i> Baylor College of Medicine <i>Speaker:</i> An Approach to Novel Immune Deficiencies Raif Geha, MD , Children's Hospital Boston <i>Supported by an unrestricted educational grant from Baxalta</i>	Commonwealth Hall DE
6:45pm - 8:30pm	CIS Closing Reception	Harborview Ballroom & Balcony

SUNDAY, APRIL 17

7:00am - 8:00am	Continental Breakfast	Commonwealth Hall E
8:00am - 10:00am	Plenary Session: Hot New Defects! 2 AMA PRA Category 1 Credits™ <i>Moderator:</i> Magda Carneiro-Sampaio, MD, PhD , Universidade De Sao Paulo <i>Speakers:</i> Sergio D. Rosenzweig, MD , NIAID, NIH Jean-Laurent Casanova, MD, PhD , The Rockefeller University Kaan Boztug, MD , Medical University of Vienna <i>Oral Abstracts</i> NOVEL IMMUNODYSREGULATION DISORDER CAUSED BY LOSS-OF-FUNCTION MUTATIONS IN SBNO2 Huie Jing, PhD , National Institute of Allergy and Infectious Diseases RUBELLA VIRUS IN CUTANEOUS GRANULOMAS IN IMMUNE DEFICIENT PATIENTS Kathleen Sullivan, MD, PhD , Children's Hospital of Philadelphia EXOSTOSIN-LIKE GLYCOSYL TRANSFERASE 3 (EXTL3) GENE MUTATION CAUSES A NOVEL FORM OF IMMUNO-OSSEOUS DYSPLASIA AND UNVEILS A CRITICAL ROLE OF HEPARAN SULFATE IN THYMOPOIESIS Stefano Volpi, PhD , Boston Children's Hospital	Commonwealth Hall DE

SCHEDULE OF EVENTS

SUNDAY, APRIL 17, CONTINUED

10:00am - 12:00pm	<p>Plenary Session: Hot New Treatments! 2 AMA PRA Category 1 Credits™ <i>Moderator:</i> Andrew Gennery, MD, Newcastle University</p> <p><i>Speakers:</i> HLH: Variations in Etiology, Pathophysiology and Manifestations Michael B. Jordan, MD, Cincinnati Children's Hospital Medical Center JAK1 Inhibitors Raphaela Goldbach-Mansky, MD, NIAMS, NIH Understanding Immune Function Through Primary Immunodeficiency Diseases Michael Lenardo, MD, NIH</p> <p><i>Oral Abstracts</i> TREATMENT WITH IL-6 BLOCKADE IN PATIENTS WITH GAIN OF FUNCTION STAT3 MUTATIONS Johana B. Castro-Wagner, MD, University of Florida/All Children's Hospital A NOVEL TARGETED APPROACH TO THE TREATMENT OF HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS (HLH) WITH NI-0501, AN ANTI-INTERFERON GAMMA MONOCLONAL ANTIBODY Michael B. Jordan, MD, Cincinnati Children's Hospital Medical Center CLINICAL SPECTRUM AND OUTCOME OF TREATMENT FOR AUTOIMMUNE CYTOPENIAS IN RAG DEFICIENCY Zsolt Foldvari, MD, University of Debrecen</p>	Commonwealth Hall DE
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EXHIBITOR LISTING

BOOTH #6

Axela Care

AxelaCare, national home infusion provider, taking care of acute and chronic patients across country. With over 30 pharmacy locations, and our extensive team of employed clinicians, our goal is to provide exceptional patient care. Services include anti-infectives, inotropes, parenteral nutrition, immunoglobulin, and other specialty infusions. AxelaCare is a leader in innovation, including CareExchange technology for wireless collection of patient outcomes for therapy optimization, and Transitions program focused on hospital to home site of care. Joint Commission and URAC certified, AxelaCare has extensive local, regional, and national payer contracts to facilitate an easy process to ensure excellent care for your patients.

BOOTH #21

Baxalta – Medical Affairs

The Baxalta Medical Affairs booth will be staffed by Baxalta Medical Affairs representatives who are able to answer medical and scientific questions about Baxalta's immune globulin products. For more information on the Immunology therapeutic area, please visit www.baxalta.com.

BOOTH #9

Baxalta – Commercial

Baxalta Incorporated is a \$6 billion global biopharmaceutical leader developing, manufacturing and commercializing therapies for orphan diseases and underserved conditions in hematology, oncology and immunology. Driven by passion to make a meaningful impact on patients' lives, Baxalta's broad and diverse pipeline includes biologics with novel mechanisms and advanced technology platforms such as gene therapy.

BOOTH #5

Beckman Coulter

BOOTH #3

The Binding Site

Binding Site is committed to developing special protein assays and automated systems for the improvement of patient care. The market leader in immunodiagnostics for Immunodeficiency, Binding Site assays measure specific antibody response to vaccinations and quantify immunoglobulins, IgG and IgA subclasses. Run on the Optilite® analyzer, the future of special protein testing, our wide menu of assays includes Freelite® for the diagnosis and monitoring of myeloma. With our extensive development expertise, patented antibody production technology, and medical educators, backed by clinical practice guidelines, Binding Site provides healthcare professionals with tools to significantly improve diagnosis and management of patients. www.thebindingsite.com

BOOTH #1

Biofusion

Biofusion is a specialty provider of IgG therapy. Our unique service model includes pharmacy expertise that includes review and monitoring of patient adherence to therapy. We possess a national network of infusion nurses specifically trained in IgG administration. Our streamlined care coordination includes reimbursement services and a patient advocacy program tailored to the individualized needs of IgG patients and their caregivers.

BOOTH #12

Bio Products

Bio Products Laboratory (BPL) is a global company with 60 years of history in plasma research, technology, and manufacturing. Originally part of the world-famous Lister Institute, BPL has a full line of products derived from plasma collected at 35 company-owned collection centers across the US. For more information please visit www.bpl-us.com.

BOOTH #13

Cincinnati Children's

Our laboratories offer a wide variety of molecular and cellular diagnostic testing for heritable immunodeficiency and hematology disorders, in addition to expert clinical interpretations, rapid turnaround times, ongoing customer support and competitive pricing. For patients undergoing genetic testing, board certified pediatric immunologists, hematologists, molecular geneticists and genetic counselors compile all available information into a personalized report in order to provide the referring physician with meaningful clinical interpretations and recommendations specific to his or her patient.

BOOTH #10

Context Media

ContextMedia:Health is the largest and most innovative provider of digital education solutions at the point of care. ContextMedia:Health delivers hyper-targeted patient education to waiting and exam rooms, impacting over 12M patient visits monthly. Recently, ContextMedia:Health introduced Patient Wifi and the Digital Exam Room Wallboard, which guarantee patients access to relevant health information during resolution moments of their treatment. The Digital Exam Room Wallboard features an interactive physician mode, which provides technology to educate patients with 3D responsive anatomical diagrams that physicians can mark to illustrate aspects of patient illness, and communicate treatment information to improve retention.

BOOTH #17

CSL Behring

CSL Behring is a leader in the plasma protein therapeutics industry. Committed to saving lives and improving the quality of life for people with rare and serious diseases, the company manufactures and markets a range of plasma-derived and recombinant therapies worldwide.

CSL Behring therapies are used for the treatment of primary immune deficiencies, coagulation disorders including hemophilia and von Willebrand disease, hereditary angioedema and inherited respiratory disease. The company's products are also used in cardiac surgery, organ transplantation, burn treatment and to prevent hemolytic diseases in newborns.

EXHIBITOR LISTING

CSL Behring operates one of the world's largest plasma collection networks, CSL Plasma. CSL Behring is a subsidiary of CSL Limited (ASX:CSL), a biopharmaceutical company headquartered in Melbourne, Australia. For more information, visit www.cslbehring.com.

BOOTH #20

Dana-Farber/Boston Children's Cancer and Blood Disorders Center

Dana-Farber/Boston Children's Cancer and Blood Disorders Center offers the combined strengths of two internationally recognized Harvard-affiliated research and teaching centers. Neighboring institutes connected by pedestrian bridges, Dana-Farber Cancer Institute and Boston Children's Hospital offer seamless, integrated care for pediatric cancer and blood disorders patients. Working together, we provide more specialists, more programs, and more clinical trials than almost any other pediatric center treating cancer and blood disorders in the world.

BOOTH #18

Diplomat

Our dedicated specialty infusion team has many years of clinical experience and customer service. We provide specialty infusion therapies nationwide—including self-administration assistance, education and other patient services—in homes and in other infusion sites of care. Additionally, Diplomat can provide in-home nursing services through our extensive national nursing network. We customize programs and strategies to exceed customer expectations while staying true to our high-touch service model. Our specialty infusion team now includes:

- AHF- A Diplomat Company
- BioRx- A Diplomat Company
- MedPro Rx- A Diplomat Company

BOOTH #4

GeneDx

GeneDx is a genetic testing company founded in 2000 by scientists from the National Institutes of Health (NIH) to address the needs of patients and clinicians concerned with rare inherited disorders. GeneDx offers sequencing and deletion/duplication testing for inherited cardiac disorders, mitochondrial disorders, neurological disorders, inherited cancer disorders, and other rare genetic disorders. Whole exome sequencing, microarray-based testing, targeted variant testing, and prenatal diagnostic

services also offered. At GeneDx, technical services are matched by scientific expertise and customer support. GeneDx employs more than 100 geneticists and genetic counselors specializing in clinical, molecular, and metabolic genetics, and also cytogenetics. Visit www.genedx.com.

BOOTH #19

Grifols USA, LLC

Grifols is a global healthcare company whose mission is to improve the health and well-being of people around the world. We have three primary divisions – Bioscience, Diagnostic and Hospital – that develop, produce and market our innovative products and services to medical professionals in more than 100 countries around the world.

BOOTH #8

Horizon Pharma

Horizon Pharma plc is a specialty biopharmaceutical company focused on improving patients' lives by identifying, developing, acquiring and commercializing differentiated and accessible medicines that address unmet medical needs. The Company markets seven medicines through its orphan, primary care and specialty business units. Horizon's global headquarters are in Dublin, Ireland.

BOOTH #15

IDF

The Immune Deficiency Foundation is the national patient organization dedicated to improving the diagnosis, treatment and quality of life of persons with primary immunodeficiency diseases through advocacy, education and research.

BOOTH #14

Invitae

Invitae's mission is to bring genetic information into mainstream medical practice to improve healthcare for everyone. We offer clinically validated, affordable genetic testing for oncology, cardiology, neurology, pediatrics, and more.

BOOTH #2

Medical College of Wisconsin

MCW, through the Clinical Immunodiagnostic and Research Laboratory (CIRL) and Allergy Immunology Diagnostic Lab Center (AIDLC), performs comprehensive testing for the diagnosis of primary immunodeficiencies (PIDs) and allergic disorders. The CIRL performs flow cytometric-based assays for the detection of PIDs. The AIDLC performs serologic assays for the

diagnosis of hypersensitivity pneumonitis, allergic bronchopulmonary aspergillosis and latex allergy. The AIDLC also performs a *Salmonella Typhi* IgG assay to quantitate specific antibody responses to the killed polysaccharide *S. typhi* vaccine.

BOOTH #16

National Jewish Health

National Jewish Health Advanced Diagnostic Laboratories is a CAP, CLIA and CAP15189SM laboratory with decades of experience developing immunology, complement, infectious disease and molecular diagnostic tests. We provide unparalleled clinical expertise and diagnostic testing in immune deficiency, respiratory disease, allergy and autoimmunity. Through collaborations with our fellow physicians and laboratory scientists, we share our passion for science and develop new therapies and high quality laboratory tests that transform patient lives.

BOOTH #11

St. Jude

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