

CIS 2019 ANNUAL MEETING AND PRE-CONFERENCE EDUCATION DAY

ATLANTA

APRIL 4-7, 2019 MARRIOTT MARQUIS

FINAL PROGRAM



CIS Clinical
Immunology
Society



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 800 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 immunological/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.

CIS NATIONAL OFFICE

555 East Wells Street, Suite 1100
Milwaukee, WI 53202
Phone: 414.224.8095
Email: info@clinimmsoc.org
www.clinimmsoc.org

CIS-0318-599

TABLE OF CONTENTS

About CIS	Left
General Meeting Information	3
Continuing Medical Education Information	4
Speaker Disclosures	5
Atlanta Visitor Information	8
Schedule at-a-Glance	9
Pre-Conference Program	9
Annual Meeting Program	11
Exhibit and Poster Hall Floor Plan	20
Exhibitor Listing	21
Friday Posters	24
Saturday Posters	29
2019 Annual Meeting Sponsors	34

2019 ANNUAL MEETING AND PRE-CONFERENCE EDUCATION DAY PROGRAM COMMITTEE

Co-Chairs:

Roshini Sarah Abraham, PhD
Nationwide Children's Hospital
Columbus, OH

Sergio Rosenzweig, MD, PhD
NIH
Bethesda, MD

Committee:

Manish Butte, MD, PhD
UCLA
Los Angeles, CA

Kiran Patel, MD
Emory University
Atlanta, GA

Shanmuganathan Chandrakasan, MD
Emory University School of Medicine,
Children's Healthcare of Atlanta
Atlanta, GA

Neil Romberg, MD
Children's Hospital of Philadelphia
Philadelphia, PA

Sarah Henrickson, MD, PhD
Children's Hospital of Philadelphia
Philadelphia, PA

John Routes, MD
Medical College of Wisconsin
Milwaukee, WI

Lisa Kobrynski, MD, MPH
Emory University
Atlanta, GA

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

Jolan Walter, MD, PhD
University of South Florida

CONNECT WITH YOUR COLLEAGUES!

"LIKE" CIS on Facebook

Follow us on Twitter @CLINIMMSOC #CIS2019 #CISinAtlanta

Join our Group on LinkedIn



National Office

555 East Wells Street
Suite 1100
Milwaukee, WI 53202-3823 USA
Telephone 414.224.8095
Fax 414.272.6070
Email info@clinimmsoc.org
www.clinimmsoc.org

Dear Colleagues,

On behalf of the Clinical Immunology Society (CIS), thank you for participating in the 2019 Annual Meeting: Immune Deficiency & Dysregulation North American Conference taking place April 4-7, 2019 at the Marriott Marquis in Atlanta, Georgia.

CIS is the key multidisciplinary 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 a variety of backgrounds to identify the optimal approach to addressing critical educational needs in the field of clinical immunology. The primary goal of the Annual Meeting is to provide a stimulating and lively forum for presentation and discussion of the latest advances in clinical immunology, including primary immunodeficiencies and immune dysregulatory diseases, and related areas, including molecular pathogenesis, and novel therapeutic approaches.

The conference will focus on cutting edge clinical and basic science research, as well as practical applications of these in patient care. The plenary sessions of the conference are specifically designed to tackle key issues facing physicians and scientists in the field of clinical immunology, while also exploring its inter-connectedness to other medical disciplines and healthcare in general, and addressing the challenges and benefits of new diagnostic tools, and therapeutic options currently available for personalized treatment of patients with rare immunological disorders. The sessions will also provide a venue for lively dialogue on the growing field of secondary immunodeficiencies, and the future of immunomodulatory therapies.

The ultimate goal of the Annual CIS meeting is to engage, educate and generate a large and collaborative network of clinical immunology physicians and scientists, in a manner that is consistent with the overarching aims of the Clinical Immunology Society, and equip them for clinical practice and research in this field.

The CIS leadership is delighted to announce the 2019 Annual Meeting, and we look forward to bringing together outstanding clinicians and scientists to provide the latest research and clinical advances for the benefit of the participants and our patients. We hope you enjoy this meeting, and take full advantage of the opportunity to learn and network with colleagues.

Regards,

Roshini S. Abraham

Roshini Sarah Abraham, PhD
Nationwide Children's Hospital
Columbus, OH
2019 Program Committee Co-Chair

[Signature]

Sergio Rosenzweig, MD, PhD
National Institutes of Health
Bethesda, MD
2019 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/>

Editor-in-Chief:

Vincent R. Bonagura
The Feinstein Institute for
Medical Research
Hofstra North Shore-LIJ
School of Medicine
Manhasset, NY, USA

Editor-in-Chief:

Jean-Laurent Casanova
The Rockefeller University
New York, NY, USA

Associate Editors:

Steven M. Holland
National Institute of Allergy
and Infectious Diseases
Bethesda, MD, USA

Christoph Klein
Ludwig Maximilians
University Munich
Munich, Germany

László Maródi
University of Debrecen
Debrecen, Hungary

Luigi D. Notarangelo
National Institutes of Health
Bethesda, MD

Lawrence Schwartz
Virginia Commonwealth
University
Richmond, VA, USA

Ricardo Sorensen
Louisiana State University
Health Science Center
New Orleans, LA, USA

Helen C. Su
National Institute of Allergy
and Infectious Disease
Rockville, MD, USA

Stuart Tangye
Garvan Institute of Medical
Research
Sydney, NSW, Australia

Troy R. Torgerson

Seattle Children's Hospital
Seattle, WA, USA

Editorial Board:

Daniel Adelman
University of California at San
Francisco
San Francisco, CA, USA

Waleed Al-Herz
Kuwait University
Kuwait

Hamoud Al-Moussa
King Faisal Specialist Hospital
and Research Center
Riyadh, Saudi Arabia

Saleh Al-Muhsen
King Saud University
Riyadh, Saudi Arabia

Basel K. Al-Ramadi
United Arab Emirates
University
United Arab Emirates

Mohamed Ridha Barbouche
Institute Pasteur de Tunis
Tunis, Tunisia

Mel Berger
CSL Behring
Cleveland, OH, USA

Francisco Bonilla
Harvard University Children's
Hospital
Boston, MA, USA

Aziz Bousfiha
King Hassan II University
Casablanca, Morocco

Kaan Boztug
Medical University of Vienna
Vienna, Austria

Yenan Bryceson
Karolinska Institutet
Stockholm, Sweden

Yildiz Camcioglu

Istanbul University
Istanbul, Turkey

Regis Campos
Federal University of Bahia
Medical School
Salvador, Brazil

Fabio Candotti
National Human Genome
Research Institute
Bethesda, MD, USA

Andrew Cant
Newcastle University
Newcastle upon Tyne, United
Kingdom

Talal Chatila
University of California at Los
Angeles
Los Angeles, CA, USA

Antonio Condino-Neto
University of São Paulo
São Paulo, Brazil

Beatriz Tavares Costa-
Carvalho
Federal University of São
Paulo

São Paulo, Brazil
Charlotte Cunningham-
Rundles
Mount Sinai Medical Center
New York, NY, USA

Silvia Danielian
Hospital de Pediatria Pr Dr
Juan P Garrahan
Buenos Aires, Argentina

Stephan Ehl
University of Freiburg
Freiburg, Germany

Francisco Espinosa
Instituto Nacional de Pediatría
Mexico

Amos Etzioni

Ruth Rappaport Children's
Hospital
Haifa, Israel

Lisa Filipovich
Cincinnati Children's Hospital
Medical Center
Cincinnati, Ohio, USA

Jose Luis Franco
Universidad de Antioquia
Medellin, Colombia

Benzi Garty
Schneider Children's Medical
Center of Israel and Tel Aviv
University School of Medicine
Tel Aviv, Israel

Bobby Gaspar
University College London
London, United Kingdom

Andrew Gennery
Newcastle University
Newcastle upon Tyne, United
Kingdom

Anete Grumach
Alameda Santos
São Paulo, Brazil

Sophie Hambleton
Newcastle University
Newcastle upon Tyne, United
Kingdom

Lennart Hammarstrom
Karolinska Institute
Stockholm, Sweden

Hal Hoffman
University of California at
San Diego
San Diego, CA, USA

Jamie Inostroza
University of La Frontera
Temuco, Chile

Stephen Jolles
Cardiff University
Cardiff, United Kingdom

Emmanuelle Jouanguy

INSERM
Paris, France

Carl June
University of Pennsylvania
School of Medicine
Philadelphia, PA, USA

Donald Kohn
University of California at Los
Angeles
Los Angeles, CA, USA

Necil Kütükçüler
Ege University Medical School
Ege, Turkey

Harry Malech
National Institute of Allergy
and Infectious Diseases
Bethesda, MD, USA

Tim Nihues
HELIOS Klinikum Krefeld
Krefeld, Germany

Joao Bosco Oliveira
National Institute of Health
Bethesda, MD, USA

Jordan Orange
Texas Children's Hospital
Houston, TX, USA

Qiang Pan-Hammarstrom
Karolinska Institutet
Stockholm, Sweden

Rebeca Perez de Diego
IdiPAZ
La Paz, Spain

Alessandro Plebani
Università degli studi de
Brescia
Brescia, Italy

Jennifer Puck
University of California at San
Francisco
San Francisco, CA, USA

Anne Puel

INSERM
Paris, France

Jose R. Regueiro
Universidad Complutense
Madrid
Madrid, Spain

Janine Reichenbach
University Children's Hospital
Zurich
Zurich, Switzerland

John Routes
Medical College of Wisconsin
Milwaukee, WI, USA

Reinhard Seger
University Children's Hospital
Zurich
Zurich, Switzerland

John W. Sleasman
Duke University School of
Medicine
Durham, NC, USA

Mirjam van der Burg
Erasmus
Rotterdam, The Netherlands

Dewton de Moraes
Vasconcelos
University of São Paulo
Medical School
São Paulo, Brazil

Anna Villa
Institute of Genetic and
Biomedical Research
Italy

Horst von Bernuth
University of Berlin
Berlin, Germany

Founding Editor:
Sudhir Gupta
University of California at
Irvine
Irvine, CA, USA

MEETING LOCATION

All educational sessions, posters, exhibits, and registration held as part of the CIS Annual Meeting will take place at the Marriott Marquis

Atlanta Marriott Marquis
265 Peachtree Center Avenue
Atlanta, Georgia 30303 USA

Exhibit Hall

Marriott Marquis ~ Atrium B & C Ballroom

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 4, 8:00am – 5:00pm

Exhibit Hours

Friday, April 5

11:30am – 1:30pm

Saturday, April 6

11:30am – 1:30pm

5:30pm – 7:00pm

Exhibitor Move-Out

Saturday, April 6

7:30pm – 10:00pm

Sunday, April 7

7:00am – 11:00am

Poster Sessions

Poster sessions are held in the Poster and Exhibit Hall, Atrium B & C Ballroom. There are two separate poster sessions for the 2019 Annual Meeting.

- Poster Session I: Friday, April 5, 12:30pm – 1:30pm
- Poster Session II: Saturday, April 6, 12:30pm – 1:30pm

A listing of all posters, titles, and authors is available at the back of the program.

Publishing of Abstracts

The abstracts submitted to the 2019 Annual Meeting are published in the March issue of *Journal of Clinical Immunology*, the official journal of the Clinical Immunology Society. Copies are available on the CIS website.

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. By registering for the CIS Annual Meeting, you give consent to be photographed by CIS Staff for purposes of advertising and public display.

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 2019 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 & NIT-FIT LIST SERVICE

The CIS-PIDD listserv has been set up to provide an easy way for individuals to discuss cases and provide feedback to colleagues. The NIT-FIT listserv 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. These Listservs are set up as a communication tool to help you interact with your colleagues. You do not have to be a member of CIS to participate on the listservs. To join, please apply on the CIS website, www.clinimmsoc.org.

VUSserve

The purpose of the VUSserve is two-fold: (a) capture information on genetic variants of uncertain significance in immunologically relevant genes, from the larger PID community along with relevant clinical phenotype for purposes of improving our understanding of the genetic etiology of these diseases. (b) To provide a resource for CIS members and the PID community by connecting those who have identified VUS in relevant genes with those who have the capability to do functional analyses for those genes or pathways to ascertain the pathogenic relevance of these variants.

REGISTRATION DESK

Marriott Marquis ~ Atrium Foyer

Registration Desk Hours

Wednesday, April 3	4:00pm – 7:00pm
Thursday, April 4	7:00am – 7:00pm
Friday, April 5	7:00am – 7:00pm
Saturday, April 6	7:00am – 7:00pm
Sunday, April 7	7:00am – 10:30am

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 28 *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://cis.clinimmsoc.org/cme/am19/>



DISCLOSURES OF CONFLICT OF INTEREST

2019 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.

Prof. Adrian Thrasher, MBBS PhD

University College London, London, England

BOARD MEMBER/ADVISORY PANEL MEMBER: 4BioCapital (Self) (Status: Ongoing); Generation Bio (Self) (Status: Ongoing); Orchard Therapeutics (Self) (Status: Ongoing); Rocket Pharmaceuticals (Self) (Status: Ongoing)
CONSULTANT/ADVISORY BOARD MEMBER: Generation Bio (Self) (Status: Ongoing); Orchard Therapeutics (Self) (Status: Ongoing); Rocket Pharmaceuticals (Self) (Status: Ongoing)

Donald Kohn, MD

University of California, Los Angeles

CONSULTANT/ADVISORY BOARD: Orchard Therapeutics (Self) (Status: Ongoing)
RESEARCH GRANT: Orchard Therapeutics (Self) (Status: Ongoing)

Harry Malech, MD

Chief, Genetic Immunotherapy Section, Laboratory of Clinical Immunology and Microbiology, IDGS, DIR, NIAID, NIH

ADVISORY COMMITTEE/BOARD MEMBER: Orchard Therapeutics (Self) (Status: Ongoing)
OTHER FINANCIAL OR MATERIAL SUPPORT: Orchard Therapeutics (Self) (Status: Ongoing) Unpaid, uncompensated member of Scientific Advisory Board of Orchard Therapeutics No payments received no stock nor any other financial stake in Orchard Therapeutics or the study results to be presented

Manish Butte, MD, PhD

University of California, Los Angeles

CO-FOUNDER/NO OPERATIONAL ROLE WITH SHARES: Allertope (self); Microvolution (self); EpikaBio (self)
SCIENTIFIC ADVISORY BOARD/NO OPERATIONAL ROLE WITH SHARES: Alpine Immune Sciences (self); Chimera Bio (self)
SPEAKER FEE: CSL Behring (self)
CONSULTANT RECEIVING CONSULTING FEE: Horizon Pharma (self)

Prof. Andrew Cant, BsC, MD, FRCP, FRCPC

Consultant in Paediatric Immunology & Infectious Diseases, Royal Victoria Infirmary, Newcastle upon Tyne, England

CONSULTANT/ADVISORY BOARD MEMBER: Baxter / Shire (Self) (Status: Terminated – January 2018); Novartis (Self) (Status: Terminated – December 2018)

Morton Cowan, MD

University of California San Francisco, San Francisco, California

ADVISORY COMMITTEE/BOARD MEMBER: Homology Medicine Inc (Self) (Status: Ongoing)
BOARD MEMBER/ADVISORY PANEL MEMBER: Exogen Bio (Self) (Status: Ongoing)
CHAPTER AUTHOR: UpToDate (Self) (Status: Ongoing)
CLINICAL TRIALS GRANTS: California Institute of Regenerative Medicine (Self) (Status: Ongoing)
CONSULTANT/ADVISORY BOARD MEMBER: Bluebird Bio (Self) (Status: Ongoing)
DATA SAFETY MONITORING BOARD: Bluebird Bio (Self) (Status: Ongoing)
OTHER FINANCIAL OR MATERIAL SUPPORT: UpToDate (Self) (Status: Ongoing)
RESEARCH GRANT: California Institute of Regenerative Medicine (Self) (Status: Ongoing); NIH - NIAID (Self) (Status: Terminated – June 2018)
SCIENTIFIC ADVISORY BOARD: Exogen Bio (Self) (Status: Ongoing); Homology Medicine Inc (Self) (Status: Ongoing); U54-Consortium: NIH - NIAID (Self) (Status: Terminated – June 2018)

Professor Bodo Grimbacher, MD

Centre of Chronic Immunodeficiency, Universitätsklinikum Freiburg, Germany

RESEARCH GRANT: BAXTER / SHIRE (SELF) (STATUS: ONGOING); BMBF (Self) (Status: Ongoing); BMS (Self) (Status: Ongoing); CSL Behring (Self) (Status: Ongoing); DFG (Self) (Status: Ongoing); Fritz Thyssen Stiftung (Self) (Status: Ongoing); Merck (Self) (Status: Ongoing)
SPEAKER/HONORARIA: Janssen Cilag GmbH (Self) (Status: Ongoing); Novartis (Self) (Status: Ongoing)

Sarah Henrickson, MD, PhD

The Children's Hospital of Philadelphia

CONSULTANT/ADVISORY BOARD MEMBER: Horizon Pharma

Peter Hotez, MD, PhD

Baylor College of Medicine

Patent holder for various neglected tropical diseases with no financial benefit gained

Stacy A. Kahn, MD

Boston Children's Hospital

RESEARCH COLLABORATOR: OpenBlome (non-profit)
CONSULTANT: AbbVie, No financial support in past 12 months

Lisa Kobrynski, MD

Emory University

SPONSORED RESEARCH AS INVESTIGATOR: Takeda (Self)

DISCLOSURES OF CONFLICT OF INTEREST

David Nguyen, MD, PhD

University of California, San Francisco

OTHER FINANCIAL OR MATERIAL SUPPORT: Alnylam (Self) (Status: Ongoing); AMAG Pharma (Self) (Status: Ongoing); Dynavax (Self) (Status: Ongoing), Inovio Pharma (Self) (Status: Ongoing); Momenta Pharma (Self) (Status: Ongoing); Nektar Therapeutics (Self) (Status: Ongoing); Novavax (Self) (Status: Ongoing)

STOCK: Alnylam (Self) (Status: Ongoing); AMAG Pharma (Self) (Status: Ongoing); Dynavax (Self) (Status: Ongoing); Inovio Pharma (Self) (Status: Ongoing); Momenta Pharma (Self) (Status: Ongoing), Nektar Therapeutics (Self) (Status: Ongoing); Novavax (Self) (Status: Ongoing)

Helen Su, MD, PhD

Laboratory of Clinical Immunology and Microbiology, NIAID, NIH

OTHER FINANCIAL OR MATERIAL SUPPORT: Amgen (Self) (Status: Ongoing); Bristol Myers Squibb (Self) (Status: Ongoing); Eli Lilly (Self) (Status: Ongoing), Johnson & Johnson (Self) (Status: Ongoing); Pfizer (Self) (Status: Ongoing)

OTHER RESEARCH SUPPORT: Merck (Self) (Status: Ongoing); Regeneron (Self) (Status: Ongoing)

STOCK HOLDINGS: Amgen (Self) (Status: Ongoing); Bristol Myers Squibb (Self) (Status: Ongoing); Eli Lilly (Self) (Status: Ongoing); Johnson & Johnson (Self) (Status: Ongoing); Pfizer (Self) (Status: Ongoing)

Kathleen Sullivan, MD, PhD

Children's Hospital of Philadelphia

Immune Deficiency Foundation — Consultant receiving consulting fee
NIH — Grant Recipient, Principal Investigator
Elsevier — Royalties received as book editor
Up to Date — Royalties received as book editor

Troy Torgerson, MD, PhD

University Of Washington and Seattle Children's Research Institute

Shire - Consultant receiving consulting fee and member of advisory board
CSL Behring - Consultant receiving consulting fee and member of advisory board
ADMA - Consultant receiving consulting fee and member of advisory board
Grifols - Consultant receiving consulting fee and member of advisory board

Bruce Trapnell, MD

Translational Pulmonary Science Center, Cincinnati Children's Hospital Medical Center, Cincinnati

CONSULTANT: Savara (Self) (Status: Ongoing)
RESEARCH GRANT: Savara (Self) (Status: Ongoing)

Aaron Wheeler, PhD

University of Toronto, Toronto, ON

BOARD MEMBER/ADVISORY PANEL: Sci-Bots, Inc. (Self) (Status: Ongoing)
RESEARCH GRANT: Abbott Laboratories (Self) (Status: Ongoing)

Jonathan Peled, MD, PhD

Memorial Sloan Kettering Cancer Center

OTHER FINANCIAL OR MATERIAL SUPPORT: Seres Therapeutics (Self) (Status: Ongoing)
RESEARCH FUNDING: Seres Therapeutics (Self) (Status: Ongoing)

The following individuals had no financial relationships, as defined by the ACCME, to disclose:

Roshini Abraham, PhD

Nationwide Children's Hospital

Danielle Arnold, MD

Children's Hospital of Philadelphia

Vivien Béziat, PhD

INSERM U1163

Stephanie Boisson-Dupuis, PhD

The Rockefeller University

David Buchbinder, MD, MSHS

Department of Hematology, Children's Hospital of Orange County, Orange, CA, Department of Pediatrics, University of California at Irvine, Orange, CA

Janet Chou, MD

Boston Children's Hospital

Christopher J. Collins, PhD

Seattle Children's Hospital

Megan Cooper, MD, PhD

Washington University in St. Louis

Emilia Falcone, MD, PhD

IRCM-Montreal Clinical Research Institute

Lisa Forbes, MD

Baylor College of Medicine

William Gahl, MD, PhD

National Institutes of Health

Shubham Goel, PhD

Department of Laboratory Medicine, Clinical Centre, NIH

Sarah Henrickson, MD, PhD

The Children's Hospital of Philadelphia

Gloria Hong, BA

National Institute of Allergy and Infectious Diseases, National Institutes of Health

Shancy Jacob, PhD

University of Utah

Michail Lionakis, MD, ScD

Laboratory of Clinical Immunology & Microbiology (LCIM), National Institute of Allergy & Infectious Diseases (NIAID), NIH

Jeffrey Lo, MD

Boston Children's Hospital/Harvard Medical School

Rebecca Marsh, MD

Cincinnati Children's Hospital Medical Center

DISCLOSURES OF CONFLICT OF INTEREST

Joshua Milner, MD
NIH

Bahar Miraghazadeh, PhD
John Curtin School Medical Research and Translational Research Unit

Luigi Notarangelo, MD, PhD
Laboratory of Clinical Immunology and Microbiology, IDGS, DIR, NIAID, NIH

Kiran Patel, MD
Emory University

Ludmila Perelygina, PhD
Division of Viral Diseases, Centers for Disease Control and Prevention

Neil Romberg, MD
Children's Hospital of Philadelphia

Sergio Rosenzweig, MD, PhD
NIH

John Routes, MD
Medical College of Wisconsin

Sara Seghezzo, MD
University of California, San Francisco

Julie Segre, PhD
National Institutes of Health

Lucia Sereni, PhD
San Raffaele Telethon Institute for Gene Therapy (SR-Tiget), Division of Regenerative Medicine, Stem Cells and Gene Therapy, San Raffaele Scientific Institute, Milan, Italy

Andrew Snow, PhD
Uniform Services University

Maureen Su, MD
University of California, Los Angeles

Sneha Suresh, MD
University of Alberta

Stuart Tangye, PhD
Garvan Institute

Svetlana Vakkilainen, MD
Children's Hospital, Pediatric Research Center, University of Helsinki and HUS Helsinki University Hospital, and Folkhälsan Institute of Genetics, Helsinki, Finland

Nicolai van Oers, PhD
UT Southwestern Medical Center

Ajit Varki, MBBS
University of California, San Diego

Kelly Walkovich, MD
University of Michigan Medical School

Tyler Yates, MD
Duke University

ATLANTA VISITOR INFORMATION

Local Attractions

Centennial Olympic Park	265 Luckie St. 0.7 miles NW from Hotel
College Football Hall of Fame	250 Marietta St NW 0.5 miles from hotel
Center for Civil and Human Rights	100 Ivan Allen Jr Blvd 0.6 miles SW from Hotel
Georgia Aquarium	225 Baker Street 0.5 miles W from Hotel
World of Coca Cola	121 Baker Street 0.4 miles NW from Hotel
Zoo Atlanta	800 Cherokee Ave. SE 3.1 miles SE from Hotel
CNN Center	CNN Center, 100 Techwood Drive 1.5 miles SW from Hotel
Carter Center	1 Copenhill Avenue 2.7 miles NE from Hotel
Six Flags over Georgia	275 Riverside Parkway SW 14.4 miles W from Hotel
Suntrust Park	755 Battery Avenue Southeast 12 miles NE from Hotel
Children's Museum of Atlanta	0.4 miles from hotel

Places to Eat

IN THE MARRIOTT MARQUIS:

High Velocity

Pulse

Sear Bar

Starbucks®

M Store

RESTAURANTS NEARBY:

Agatha's Dinner Theater (5 Course Continental Cuisine)	161 Peachtree Center Ave
Alma Cocina (Modern Mexican)	191 Peachtree St Ne
Big Kahuna (Californian/Mexican)	303 Peachtree Center Ave
Cuts (Casual Steakhouse)	60 Andrew Young International Blvd NE
Der Biergarten (German)	300 Marietta St at Baker St
Gibney's Pub (Irish)	231 Peachtree Center Ave
Gus's World Famous Fried Chicken	Located on bottom level of the Peachtree Center Mall
Herban Fix (PanAsian/Vegan)	565 Peachtree Street NE
Hsu's (Gourmet Chinese)	192 Peachtree Center Ave
Max Lager's (WoodFired Grill and Brewery)	320 Peachtree St
Meehan's Public House (Irish)	200 Peachtree St Suite 1
Mellow Mushroom (Pizzeria)	400 W Peachtree St
Metro Café Diner (American)	229 Peachtree St
Pacific Rim Bistro (PanAsian Cuisine)	303 Peachtree Center Ave
Pitty Pat's Porch (Southern)	25 Andrew Young International Blvd
Ray's In the City (Seafood/Steak/Sushi)	240 Peachtree St
Sweet Georgia's Juke Joint (Southern)	200 Peachtree St
Terrace on Peachtree (Organic; Farm-to-Table)	176 Peachtree St
Tin Lizzy's Cantina (Tex —Mex)	229 Peachtree St
Truva (Mediterranean)	60 Andrew Young International Blvd
White Oak Kitchen & Cocktails (Contemporary Southern)	270 Peachtree St at Baker St

PRE-CONFERENCE EDUCATION DAY AGENDA

PID & ID Diagnosis, Management and Publishing Case Reports
THURSDAY, APRIL 4 | MARRIOTT MARQUIS ATLANTA | SESSION: ATRIUM A BALLROOM | MEALS: A703 & 704

7:00am – 8:00am	Registration and Continental Breakfast
8:00am – 8:05am	Welcome and Introduction John Routes, MD, Medical College of Wisconsin
8:05am – 8:30am	Master Class In Humoral Immunodeficiencies .5 AMA PRA Category 1 Credits™ Speakers: Lisa Kobrynski, MD, Emory University John Routes, MD, Medical College of Wisconsin
8:30am – 8:50am	Case Presentation .5 AMA PRA Category 1 Credits™ Human NF-kappaB2 Defect Results in Defective Intrinsic B-cell Differentiation, Function and Class Switching Shancy Jacob, PhD, University of Utah
8:50am – 9:10am	Case Presentation .5 AMA PRA Category 1 Credits™ Subcutaneous Fat Loss in a Patient with CTLA4 Haploinsufficiency and in a Patient Treated with PD-1 Inhibition: Implication for Immune Dysregulation Resulting in Acquired Generalized Lipodystrophy Kelly Walkovich, MD, University of Michigan
9:10am – 9:30am	Case Presentation .5 AMA PRA Category 1 Credits™ Severe Inflammatory Episodes Associated with COG4-Congenital Disorder of Glycosylation (CDG-IIj) Presenting as Hemophagocytic Lymphohistiocytosis (HLH) Jeffrey Lo, MD, Boston Children's Hospital/Harvard Medical School
9:30am – 10:00am	Master Class In Combined Immunodeficiencies .5 AMA PRA Category 1 Credits™ Speakers: Jennifer Heimall, MD, Children's Hospital of Philadelphia Rebecca Marsh, MD, Cincinnati Children's
10:00am – 10:30am	Morning Break
10:30am – 10:50am	Case Presentation .5 AMA PRA Category 1 Credits™ Gain-of-function IKBKB Mutation Causes Human Combined Immune Deficiency Bahar Miraghazadeh, PhD, John Curtin School Medical Research and Translational Research Unit

Thank you to the supporters of the Pre-Conference Education Day:
PID & ID Diagnosis, Management and Publishing Case Reports

CSL Behring

Grifols

Horizon Pharma

Takeda

PRE-CONFERENCE EDUCATION DAY AGENDA

PID & ID Diagnosis, Management and Publishing Case Reports
THURSDAY, APRIL 4 | MARRIOTT MARQUIS ATLANTA | SESSION: ATRIUM A BALLROOM | MEALS: A703 & 704

10:50am – 11:10am	Case Presentation .5 AMA PRA Category 1 Credits™ Homozygosity for a Novel CARD11 Mutation Causes Severe Combined Immunodeficiency (SCID), Inflammatory Gastrointestinal Disease, and Complete Abrogation of MALT1 Activity Henry Lu, BSc, BC Children's Hospital and University of British Columbia
11:10am – 11:30am	Case Presentation .5 AMA PRA Category 1 Credits™ Compound Heterozygous DOCK8 Mutations in a Patient with pre-B Cell Acute Lymphoblastic Leukemia and EBV-associated Diffuse Large B-cell Lymphoma David Buchbinder, MD, Children's Hospital of Orange County
11:30am – 12:00pm	Master Class in Immune Dysregulation .5 AMA PRA Category 1 Credits™ Speakers: Kathleen Sullivan, MD, PhD, Children's Hospital of Philadelphia Troy Torgerson, MD, PhD, Seattle Children's Research Institute
12:00pm – 1:00pm	Lunch and Discussion on Publishing Case Reports in JOCI .5 AMA PRA Category 1 Credits™ Jennifer Heimall, MD, Children's Hospital of Philadelphia Jolan Walter, MD, PhD, University of South Florida
1:00pm – 1:20pm	Case Presentation .5 AMA PRA Category 1 Credits™ Rescue Therapy with Granulocyte Transfusions (GT) as a Bridge to Hematopoietic Stem Cell Transplant (HSCT) in a Chronic Granulomatous Disease (CGD) Patient with Severe Disseminated Aspergillosis Sneha Suresh, MD, FRCPC, University of Alberta
1:20pm – 1:40pm	Case Presentation 5 AMA PRA Category 1 Credits™ Ataxia Telangiectasia with Chronic Skin Granulomas — Preventable with SCID Newborn Screening? Sara Segehzzo, MD, University of California, San Francisco
1:40pm – 2:00pm	Faculty Lightning Round Presentations .5 AMA PRA Category 1 Credits™
2:00pm	Closing Kathleen Sullivan, MD, PhD, Children's Hospital of Philadelphia

2019 CIS ANNUAL MEETING SCHEDULE AT-A-GLANCE

Thursday, April 4		
8:00am - 2:00pm	Pre-conference: PID & ID Diagnosis, Management and Publishing Case Reports	Atrium A
2:15pm - 2:30pm	Welcome to CIS Annual Meeting Roshini Abraham, PhD, CIS President	Atrium A
2:30pm - 4:30pm	Plenary: Immune Dysregulation	Atrium A
4:30pm - 5:30pm	Robert A. Good Lecture: <i>Refusal of Vaccination: Implications for Immunodeficient Patients In The Community</i> Peter Hotez, MD, PhD, Baylor College of Medicine	Atrium A
5:30pm - 6:50pm	Opening Plenary: Grantee Fellowship Presentations and Notable Abstracts	Atrium A
6:50pm - 7:00pm	Tribute to Dr. William Shearer Jordan Orange, MD, PhD, Columbia University	Atrium A
7:00pm - 7:45pm	Mentor/Mentee Reception	A702
7:45pm - 9:00pm	ECI Reception	M101
Friday, April 5		
7:00am - 8:00am	Continental Breakfast	Atrium Foyer
7:00am - 8:00am	Diagnostic Genomics 101	A704
8:00am - 9:30am	Plenary Session: Cross-Talk: PIDs from the Perspectives of Other Specialties	Atrium A
9:30am - 9:40am	Update on NIH Funding for PID Clinical Trials	Atrium A
9:40am - 10:00am	Morning Break	Atrium Foyer
10:00am - 11:30am	Plenary Session: Cross-Talk: PIDs from the Perspectives of Other Specialties II	Atrium A
11:30am - 1:30pm	Exhibit Hall Open/Lunch	Atrium B & C
12:00pm - 12:30pm	Lightning Posters	A706
12:30pm - 1:30pm	Poster Session I	Atrium B & C
1:30pm - 3:00pm	Plenary: Tales from the Listserv	Atrium A
3:00pm - 3:30pm	Afternoon Break	
3:30pm - 4:30pm	Roundtable Breakouts	
	Transplantation	A704
	Gene Therapy	A706
	Diagnostic Workup of PIDs	Atrium A
4:30pm - 5:30pm	CIS Founders Rose/Fahey Lecture: <i>Intrinsic Genetic Defects of Glycosylation in Humans As A Species, Compared With Other Close Evolutionary Relatives</i> Ajit Varki, MD, University of California, San Diego	Atrium A
5:30pm - 6:00pm	Plenary Session: PID Year in Review	Atrium A
6:30pm - 8:00pm	WCIS Reception: Diversity and Implicit Bias in Academic Medicine Laura Castillo-Page, PhD, Association of American Medical Colleges	A706

2019 CIS ANNUAL MEETING SCHEDULE AT-A-GLANCE

Saturday, April 6		
6:15am - 7:00am	Morning Mindfulness	A706
7:00am - 8:00am	Continental Breakfast	Atrium Foyer
7:00am - 8:00am	Functional Validation 102	A704
8:00am - 9:30am	Plenary Session: Therapeutic and Technological Frontiers	Atrium A
9:30am - 10:30am	Plenary: Infections in PIDs	Atrium A
10:30am - 11:00am	Morning Break	Atrium Foyer
11:00am - 11:30am	Plenary: Presidential Award Lecture: <i>B cells, CD4 T cells and EBV-induced disease - PIDs can reveal all!</i> Stuart Tangye, PhD, Garvan Institute	Atrium A
11:30am - 12:30pm	ECI Luncheon: How to Carve a Niche and Lead	A704
11:30am - 1:30pm	Lunch/Exhibit Hall Open	Atrium B & C
12:00pm - 12:30pm	Lightning Posters	A706
12:30pm - 1:30pm	Poster Session II	Atrium B & C
1:30pm - 3:00pm	Plenary Session: Biome	Atrium A
3:00pm - 4:00pm	CIS Business Meeting	Atrium A
4:00pm - 4:30pm	Afternoon Break	Atrium Foyer
4:30pm - 5:30pm	Richard Schiff Lecture Steve Holland, MD	Atrium A
5:30pm - 7:00pm	Closing Reception	Atrium B & C
7:00pm - 8:30pm	CIS Talent Show to Benefit the CIS Foundation	Atrium A

Sunday, April 7		
7:00am - 8:00am	Continental Breakfast	Atrium Foyer
8:00am - 10:30am	Plenary Session: Hot New Defects in Primary Immunodeficiencies	Atrium A

SCHEDULE OF EVENTS

2019 ANNUAL MEETING: IMMUNE DEFICIENCY & DYSREGULATION NORTH AMERICAN CONFERENCE

Plenary Sessions—Atrium A Ballroom
Poster and Exhibit Hall—Atrium B & C Ballroom
Other sessions as listed in program

THURSDAY, APRIL 4, 2019	
2:15pm - 2:30pm	OPENING AND WELCOME TO THE 2019 CIS ANNUAL MEETING Roshini Sarah Abraham, PhD, CIS President, Nationwide Children's Hospital
2:30pm - 4:30pm	PLENARY SESSION #1: IMMUNE DYSREGULATION 2 AMA PRA Category 1 Credits™ <u>Moderators:</u> Jolan Walter, MD, PhD, University of South Florida Kiran Patel, MD, Emory University <u>Speakers:</u> 2:30pm - 3:10pm <i>Immune Dysregulation Observed in Patients with Hypomorphic RAG Defects</i> Luigi Notarangelo, MD , National Institutes of Health 3:10pm - 3:50pm <i>The Paradox of Immune Deficiency and Autoimmunity due to PIK3CD GOF Mutations</i> Stuart Tangye, PhD , Garvan Institute 3:50pm - 4:10pm <i>Update on CARD11 Defects: From BENTA to Atopy and Beyond</i> Andrew Snow, PhD , Uniformed Services University of the Health Sciences <u>Oral Abstracts</u> 4:10pm - 4:20pm <i>Application of Targeted Proteomics in the Diagnosis and Screening of Primary Immunodeficiency Disorders</i> Christopher Collins, PhD , Seattle Children's Hospital 4:20pm - 4:30pm <i>Heterozygous OAS1 Mutations Cause Spontaneous RNA Cleavage and Apoptosis with Resulting Multisystem Inflammation and Immunodeficiency</i> Jennifer Heimall, MD , Children's Hospital of Philadelphia
4:30pm - 5:30pm	ROBERT A. GOOD LECTURE 1 AMA PRA Category 1 Credits™ <u>Moderator:</u> Jordan Orange, MD, PhD , Columbia University <u>Speaker:</u> <i>Refusal of Vaccination: Implications for Immunodeficient Patients In The Community</i> Peter Hotez, MD, PhD , Baylor College of Medicine
5:30pm - 6:50pm	PLENARY SESSION #2: OPENING PLENARY: FELLOWSHIP PRESENTATIONS & NOTABLE ABSTRACTS 1.5 AMA PRA Category 1 Credits™ <u>Moderators:</u> Linda Griffith, MD, PhD , DAIT, NIAID, NIH Caroline Kuo, MD , UCLA Medical Center <u>Speaker:</u> 5:30pm - 6:10pm <i>Developing CRISPR Cas9 Gene Editing Technologies to Model and Correct Genetic Defects Associated with Primary Immunodeficiency</i> David Nguyen, MD, PhD , University of California, San Francisco

SCHEDULE OF EVENTS

THURSDAY, APRIL 4, 2019, continued	
6:10pm - 6:20pm	Oral Abstracts <i>Lentiviral Vector Gene Therapy for X-linked Chronic Granulomatous Disease Corrects Neutrophil Function</i> Harry Malech, MD, NIH
6:20pm - 6:30pm	<i>Novel Compound Heterozygous Mutations in Forkhead Box N1 (FOXN1) Cause a Severe Immunodeficiency Without Alopecia or Nail Dystrophy</i> Nicolai Van Oers, PhD, UT Southwestern
6:30pm - 6:40pm	<i>Investigational Cultured Thymus Tissue Transplantation (RVT-802) Following Failed Stem Cell Transplantation in Athymic Patients</i> Tyler Yates, MD, Duke University
6:40pm - 6:50pm	<i>Low Dose Azithromycin Prophylaxis Reduces Respiratory Exacerbations in Patients Affected by Primary Antibody Deficiencies : A Multicenter, Double-blind, Placebo-controlled Randomized Clinical Trial</i> Cinzia Miloto, MD, PhD, Sapienza University of Rome
6:50pm - 7:00pm	TRIBUTE TO DR. WILLIAM SHEARER Jordan Orange, MD, PhD, Columbia University
7:00pm - 7:45pm	MENTOR-MENTEE RECEPTION <i>Location: A702</i>
7:45pm - 9:00pm	EARLY CAREER IMMUNOLOGIST RECEPTION <i>Location: M101</i>
FRIDAY, APRIL 5	
7:00am - 8:00am	DIAGNOSTIC GENOMICS 101 1 AMA PRA Category 1 Credits™ <i>Location: A704</i> Janet Chou, MD, Boston Children's Hospital Helen Su, MD, PhD, NIH
8:00am - 9:30am	PLENARY SESSION #3: CROSS-TALK: PIDS FROM THE PERSPECTIVE OF OTHER SPECIALTIES 1.5 AMA PRA Category 1 Credits™ Moderators: Nicholas Rider, DO, Baylor College of Medicine Alexandra Freeman, MD, NIH Speakers:
8:00am - 8:20am	<i>Autoimmune Polyendocrinopathies</i> Maureen Su, MD, UCLA
8:20am - 8:40am	<i>Immunological Aspects of Patients in the NIH Undiagnosed Diseases Program</i> William Gahl, MD, PhD, National Institutes of Health
8:40am - 9:00am	<i>Pulmonary Alveolar Proteinosis: A Primary and Secondary Disease</i> Bruce Trapnell, MS, MD, Cincinnati Children's Hospital Medical Center
9:00am - 9:10am	Oral Abstracts <i>CARD9del11 Gene Dosage: From Mono-allelic Protection to IBD, to Bi-allelic Increased Fungal Infection Susceptibility</i> Shubham Goel, PhD, NIH
9:10am - 9:20am	<i>A 30-year Prospective Study Reveals Risk Factors for Malignancies and Early Death in Cartilage-hair Hypoplasia</i> Svetlana Vakkilainen, MD, University of Helsinki, HUS Helsinki University Hospital, and Folkhälsan Institute of Genetics
9:20am - 9:30am	<i>Natural History of Anti-Interferon-gamma Autoantibody-associated Immunodeficiency Syndrome in Thailand</i> Gloria Hong, BA, NIAID, NIH
9:30am - 9:40am	UPDATE ON NIH FUNDING FOR PID CLINICAL TRIALS Speaker: Frosso Voulgaropoulou, PhD, NIH
9:40am - 10:00am	Morning Break

SCHEDULE OF EVENTS

FRIDAY, APRIL 5, continued	
10:00am - 11:30am	PLENARY SESSION #4: CROSS-TALK: PIDS FROM THE PERSPECTIVE OF OTHER SPECIALTIES II 1.5 AMA PRA Category 1 Credits™ <u>Moderators:</u> Surjit Singh, MD , Postgraduate Institute of Medical Education and Research, Chandigarh, India Elena Hsieh, MD , University of Colorado Denver <u>Speakers:</u> 10:00am - 10:30am <i>A NICER Approach to Immunology: Opportunities in Hematology/Oncology</i> Kelly Walkovich, MD , CS Mott's Children's Hospital 10:30am - 11:00am <i>Fecal Microbiome Transplants</i> Stacy Kahn, MD , Boston Children's Hospital <u>Oral Abstracts:</u> 11:00am - 11:10am <i>Immunophenotype and Metabolic Characteristics of EBV-Specific T Cells Generated Using Different Manufacture Approaches</i> Danielle Arnold, MD , Children's Hospital of Philadelphia 11:10am - 11:20am <i>Evaluation of Persistent Hypogammaglobulinemia Post-rituximab in Patients Undergoing HSCT for XLP-1 Reveals Preserved Ability of B-cells to Class-switch and Differentiate to Plasmablasts In-vitro</i> Shanmuganathan Chandrakasan, MD , Children's Healthcare of Atlanta, Emory University 11:20am - 11:30am <i>Emapalumab-Izsg for the Treatment of Pediatric Patients with Primary Hemophagocytic Lymphohistiocytosis</i> Michael Jordan, MD , Cincinnati Children's Hospital Medical Center
11:30am - 1:30pm	LUNCH/EXHIBIT HALL OPEN
12:00pm - 12:30pm	FRIDAY LIGHTNING POSTER PRESENTATIONS <i>Location: A706</i> <u>Moderators:</u> Troy Torgerson, MD, PhD , University of Washington/Seattle Children's Research Institute Rebecca Marsh, MD , Cincinnati Children's Hospital Medical Center <u>Presenters:</u> <i>Epigenetic Immune Cell Quantification for Diagnosis and Monitoring of Patients with Primary Immune Deficiencies and Immune Regulatory Disorders</i> Janika Schulze, MSc , Epimune GmbH <i>Characterization of Gut Inflammation and Autoimmunity in Mice Carrying Rag1 Hypomorphic Mutations</i> Riccardo Castagnoli, MD , NIAID <i>Quercetin Halts Abnormal IL-1beta and IL-18 Production: A Natural Calm for XIAP Deficiency?</i> Rebecca Marsh, MD , Cincinnati Children's Hospital Medical Center <i>Hyperactivated PI3Kd Promotes Self and Commensal Reactivity at the Expense of Optimal Humoral Immunity</i> Silvia Preite, PhD , NHGRI <i>Clinical Phenotyping of a DOCK8 Deficiency Cohort</i> Alexandra Freeman, MD , NIH
12:30pm - 1:30pm	POSTER SESSION I
1:30pm - 3:00pm	PLENARY SESSION #5: TALES FROM THE LISTSERV 1.5 AMA PRA Category 1 Credits™ <u>Moderators:</u> Elie Haddad, MD, PhD , University of Montreal/CHU-Ste. Justine Mikko Seppänen, MD, PhD , Helsinki University Central Hospital Eric Allenspach, MD, PhD , University of Washington & Seattle Children's Hospital

SCHEDULE OF EVENTS

FRIDAY, APRIL 5, continued	
1:30pm - 2:00pm	<p><u>Speakers:</u> Mystery Case #1 David Hagin, MD, PhD, Tel-Aviv Sourasky Medical Center</p>
2:00pm - 2:30pm	<p>Mystery Case #2 Elizabeth Feuille, MD, Weill Cornell Medicine</p>
2:30pm - 3:00pm	<p>Mystery Case #3 Christopher Chang, MD, PhD, MBA, Joe DiMaggio Children's Hospital</p>
3:00pm - 3:30pm	Afternoon Break
3:30pm - 4:30pm	<p>ROUNDTABLE BREAKOUT DISCUSSIONS 1 AMA PRA Category 1 Credits™</p> <p><u>TRANSPLANTATION:</u> <i>Location: A704</i> Andrew Cant, MD, Newcastle General Hospital Mort Cowan, MD, UCSF Benioff Children's Hospital</p> <p><u>GENE THERAPY in PIDs:</u> <i>Location: A706</i> Don Kohn, MD, University of California, Los Angeles Adrian Thrasher, MD, PhD, UCL Great Ormond Street Institute of Child Health</p> <p><u>DIAGNOSTIC WORK-UP of PIDS:</u> <i>Location: Atrium A</i> Sergio Rosenzweig, MD, PhD, NIH Megan Cooper, MD, PhD, Washington University in St. Louis</p>
4:30pm - 5:30pm	<p>CIS ROSE/FAHEY LECTURE 1 AMA PRA Category 1 Credits™</p> <p><u>Moderator:</u> Sergio Rosenzweig, MD, PhD, NIH</p> <p><u>Speaker:</u> <i>Intrinsic Genetic Defects of Glycosylation in Humans As A Species, Compared With Other Close Evolutionary Relatives</i> Ajit Varki, MD, University of California, San Diego</p>
5:30pm - 6:00pm	<p>PLENARY SESSION #6: PID YEAR IN REVIEW .5 AMA PRA Category 1 Credits™</p> <p><u>Moderators:</u> Prescott Atkinson, MD, PhD, FAACAP, University of Alabama at Birmingham/Children's Hospital Joe Church, MD, Children's Hospital of Los Angeles</p> <p><u>Presenters:</u> <i>Genetic and mechanistic diversity in pediatric hemophagocytic lymphohistiocytosis. Blood 2018 132:89-100; doi: https://doi.org/10.1182/blood-2017-11-814244</i> Annaliese Blincoe, MD, Starship Children's Health <i>Inherited p40phox deficiency differs from classic chronic granulomatous disease. J Clin Invest. 2018 Aug 31;128(9):3957-3975. doi: 10.1172/JCI97116. Epub 2018 Aug 6.</i> Karin Chen, MD, University of Utah <i>Lymphocyte Apoptosis and FAS Expression in Patients with 22q11.2 Deletion Syndrome. J Clin Immunol. 2019 Jan;39(1):65-74. doi: 10.1007/s10875-018-0579-7. Epub 2018 Dec 19.</i> Cathleen Collins, MD, PhD, University of California, San Diego <i>T-cell defects in patients with ARPC1B germline mutations account for combined immunodeficiency. Blood. 2018 Nov 29;132(22):2362-2374. doi: 10.1182/blood-2018-07-863431. Epub 2018 Sep 25.</i> Samuel Chiang, PhD, Cincinnati Children's Hospital Medical Center</p>

SCHEDULE OF EVENTS

SATURDAY, APRIL 6, continued	
6:30pm - 8:00pm	WOMEN IN CLINICAL IMMUNOLOGY SCIENCES RECEPTION: DIVERSITY AND IMPLICIT BIAS IN ACADEMIC MEDICINE <i>Sponsored by Horizon Pharma</i> <u>Moderator:</u> Victoria Dimitriadis, MD , University of California, Davis <u>Speaker:</u> Laura Castillo-Page, PhD , Association of American Medical Colleges This reception is open to all Annual Meeting attendees!
SATURDAY, APRIL 6	
6:15am - 7:00am	Morning Mindfulness <i>Location: A706</i>
7:00am - 8:00am	Functional Validation 102 <i>1 AMA PRA Category 1 Credits™</i> <i>Location: A704</i> <u>Moderators:</u> Sarah Henrickson, MD, PhD , Children's Hospital of Philadelphia Manish Butte, MD, PhD , University of California, Los Angeles
8:00am - 9:30am	PLENARY SESSION #7: THERAPEUTIC AND TECHNOLOGICAL FRONTIERS <i>1.5 AMA PRA Category 1 Credits™</i> <u>Moderators:</u> Neil Romberg, MD , Children's Hospital of Philadelphia Tandakha Dieye, PhD , Université Cheikh Anta Diop, Senegal <u>Speakers:</u>
8:00am - 8:40am	<i>Therapeutic Advances in Clinical Immunology/PIDs</i> Bodo Grimbacher, MD , University of Freiburg
8:40am - 9:20am	<i>A Lab On A Chip System To Screen For Infections In Remote Settings</i> Aaron Wheeler, PhD , University of Toronto <u>Oral Abstract:</u>
9:20am - 9:30am	<i>Lentiviral Gene Therapy Corrects Platelet Phenotype and Function in Wiskott-Aldrich Patients</i> Lucia Sereni, PhD , San Raffaele Telethon Institute for Gene Therapy (SR-Tiget)
9:30am - 10:30am	PLENARY SESSION #8: INFECTIONS IN PIDS <i>1 AMA PRA Category 1 Credits™</i> <u>Moderators:</u> Maite de la Morena, MD , Seattle Children's Hospital Francisco Espinosa, MD , Centro de Inmunología Alergia y Pediatría Hospital Ángeles Lomas <u>Speakers:</u>
9:30am - 10:00am	<i>Tissue-specific Antifungal Immune Mechanisms Revealed by Studies in PIDs</i> Michail Lionakis, MD, ScD , NIAID
10:00am - 10:30am	<i>Rubella in Chronic Inflammation</i> Ludmila Perelygina, PhD , CDC
10:30am - 11:00am	Morning Break

SCHEDULE OF EVENTS

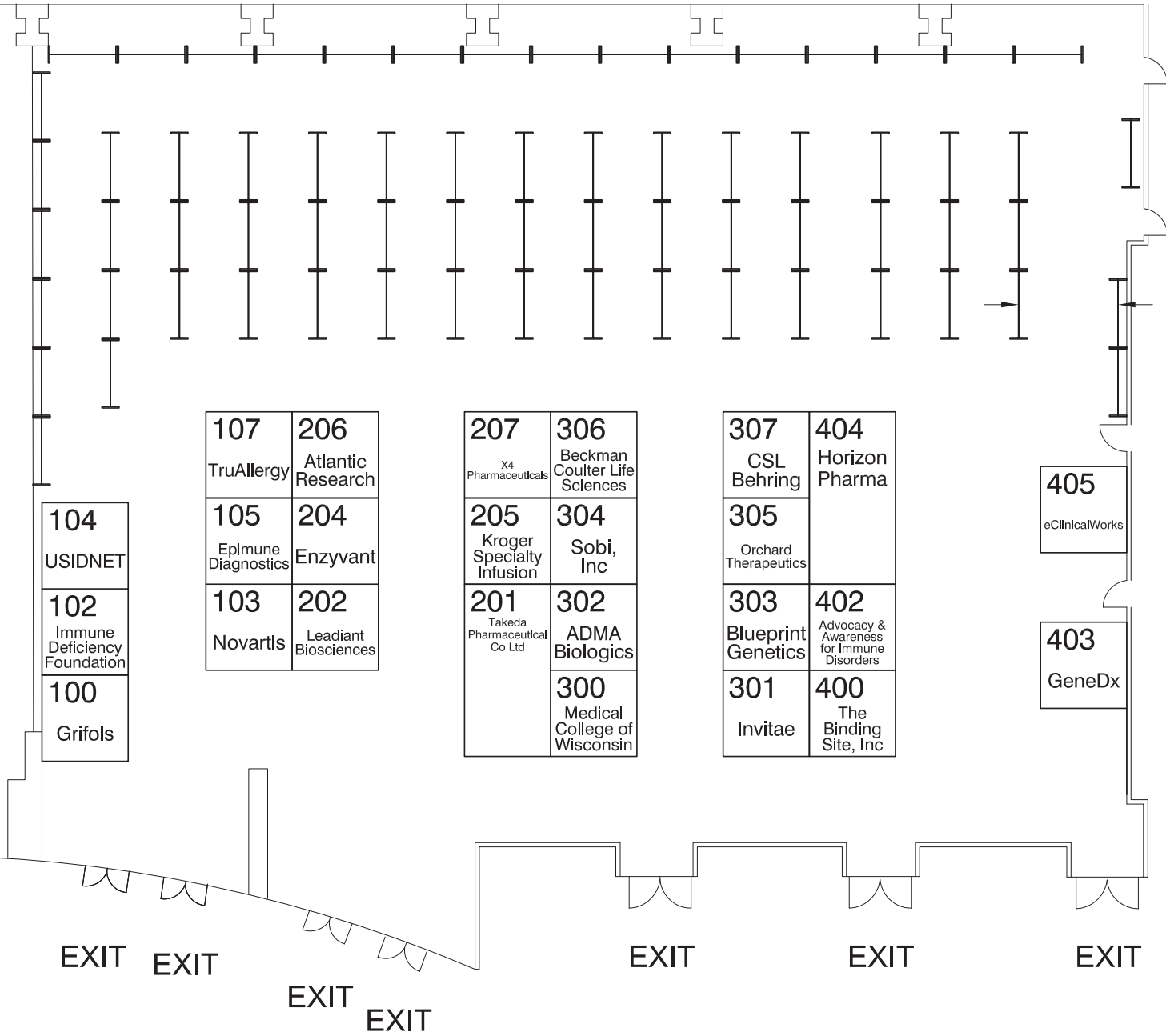
SATURDAY, APRIL 6, continued	
11:00am - 11:30am	PLENARY SESSION #9: PRESIDENTIAL AWARD LECTURE .5 AMA PRA Category 1 Credits™ <u>Moderator:</u> Roshini Abraham, PhD , Nationwide Children's Hospital <u>Speaker:</u> <i>B cells, CD4 T cells and EBV-induced disease - PIDs can reveal all!</i> Stuart Tangye, PhD , Garvan Institute
11:30am - 1:30pm	LUNCH/EXHIBIT HALL OPEN
11:30am - 12:30pm	EARLY CAREER IMMUNOLOGIST LUNCHEON: HOW TO CARVE A NICHE AND LEAD Location: A704 <u>Speakers:</u> <ul style="list-style-type: none"> • Thomas Fleisher, MD, AAAAI • Megan Cooper, MD, PhD, Washington University in St. Louis • Emily Mace, PhD, Baylor College of Medicine • Mark Ballow, MD, FAAAAI, University of South Florida
12:00pm - 12:30pm	LIGHTNING POSTER PRESENTATIONS Location: A706 <u>Moderators:</u> Troy Torgerson, MD, PhD , University of Washington/Seattle Children's Research Institute Eleonora Gambineri, MD , University of Florence <u>Presenters:</u> <i>Prevalence of Hypogammaglobulinemia in Newly Diagnosed Lymphoma</i> Namrata Singh, MD, MSCI, FACP , University of Iowa Hospitals and Clinics <i>Hexaviral-Specific T-cells for Treatment and Prevention of Viral Infections Post Hematopoietic Stem Cell Transplant</i> Michael Keller, MD , Children's National Health System <i>NGS Reveals Repertoire Restriction of Treg Cells in APDS1 Patients</i> Ottavia Delmonte, MD, PhD , NIAID <i>CVID Plasma Promotes Early Commitment to the Follicular Lineage</i> Caroline Khanna , Children's Hospital of Philadelphia <i>Functional Determination of All Possible Disease-associated Variants of a Region in CARD11 Using Saturation Genome Editing</i> Richard James, PhD , Seattle Children's Research Institute
12:30pm - 1:30pm	POSTER SESSION II
1:30pm - 3:00pm	PLENARY SESSION #10: BIOME 1.5 AMA PRA Category 1 Credits™ <u>Moderators:</u> Shanmuganathan Chandrakasan, MD , Emory University School of Medicine, Children's Healthcare of Atlanta Elizabeth Kang, MD , NIH <u>Speaker:</u> <i>Relevance of The Microbiome In HCT, and Differences Between Malignancies and Immunological Disorders</i> Jonathan Peled, MD, PhD , Memorial Sloan Kettering Cancer Center <i>Host-Pathogen Interactions at Mucosal Surfaces: Relevance for Immune Disorders</i> Julie Segre, PhD , NIH <u>Oral Abstract:</u> <i>Patients with Chronic Granulomatous Disease Have Distinct Intestinal Microbiome and Metabolomic Signatures</i> Emilia L. Falcone, MD, PhD , IRCM-Montreal Clinical Research Institute

SCHEDULE OF EVENTS

SATURDAY, APRIL 6, continued	
3:00pm - 4:00pm	CIS BUSINESS MEETING
4:00pm - 4:30pm	Afternoon Break
4:30pm - 5:30pm	RICHARD SCHIFF LECTURE 1 AMA PRA Category 1 Credits™ <u>Moderator:</u> Charlotte Cunningham-Rundles, MD, PhD , Mt. Sinai Medical Center <u>Speaker:</u> Steve Holland, MD <i>Supported by an unrestricted educational grant from Takeda.</i>
5:30pm - 7:00pm	CLOSING RECEPTION
7:00pm - 8:30pm	CIS TALENT SHOW TO BENEFIT THE CIS FOUNDATION

SUNDAY, APRIL 7	
8:00am - 10:30am	PLENARY SESSION #11: HOT NEW DEFECTS IN PIDS 2.5 AMA PRA Category 1 Credits™ <u>Moderators:</u> Monica Lawrence, MD , University of Virginia Steve Holland, MD , National Institutes of Health <u>Speakers:</u> 8:00am - 8:30am <i>Monogenic Cause of Atopic Diathesis</i> Josh Milner, MD , NIAID, NIH 8:30am - 9:00am <i>Partners in Crime: A New Class Switch Defect</i> Lisa Forbes Satter, MD , Baylor College of Medicine 9:00am - 9:30am <i>A Novel Inborn Error Of Immunity Underlying T Cell Deficiency In "Tree Man Syndrome"</i> Vivien Béziat, PhD , Institut Imagine 9:30am - 10:00am <i>A Monogenic but Common Genetic Etiology Of Tuberculosis</i> Stephanie Boisson-Dupuis, PhD , The Rockefeller University <u>Oral Abstracts:</u> 10:00am - 10:10am <i>Novel Primary Immunodeficiency with Lymphoproliferative Disease Due to Biallelic Defects in NCKAP1L</i> William (Drew) Comrie, PhD , National Institutes of Health 10:10am - 10:20am <i>F-BAR Domain Only Protein 1 (FCHO1) Deficiency Is A Novel Cause Of Combined Immune Deficiency In Humans</i> Enrica Calzoni, MD , NIAID, NIH 10:20am - 10:30am <i>The bona-fide effect of long-term glucocorticoids on T cells: the endogenous Cushing Syndrome model and the role of IL-21</i> SuJin Hwang, PhD , Immunology Service, Department of Laboratory Medicine, NIH Clinical Center, NIH

EXHIBIT AND POSTER HALL FLOOR PLAN



EXHIBITOR LISTING

BOOTH 302

ADMA Biologics

ADMA Biologics is a US-based vertically integrated plasma products manufacturer committed to developing specialty plasma-derived products for the immune-compromised and other patients at risk for infection. It is our devotion to these underserved populations that fuels us, and our hands-on approach to production and development that sets us apart. Learn more about our products and capabilities at exhibit #302 and visit us at www.admabiologics.com.

BOOTH 402

Advocacy & Awareness for Immune Disorders Association (AAIDA)

AAIDA exists to advocate for patients living with Primary Immunodeficiency (PI), Secondary Immunodeficiency and Autoimmune Diseases, to create awareness and conduct research to find cures for these different groups of diseases.

AAIDA is a 501(c)3, nonprofit whose purpose is to educate patients and providers alike, to advocate for patients living with immune disorders and to create awareness about specific immune disorders, all the while making strides in research to find cures and novel treatment options for these diseases.

BOOTH 206

Atlantic Research Group

ARG is an immunology-focused contract research organization that provides comprehensive clinical program development services ranging from pre-launch consulting to commercialization. Founded in 2004, ARG has experienced consistent growth across the globe, expanding our reach to include drug and device trial management. ARG has remained true to our original vision: every project is highly significant and visible, delivered through strategic collaboration with partners.

ARG has strong relationships with leading investigators and coordinators in multiple strategic areas in immunology. Equally important, our deep experience, rooted in long-tenured project management and clinical operations team members, provides stability when asking questions and solving problems.

BOOTH 306

Beckman Coulter Life Sciences

Beckman Coulter Life Sciences develops, manufactures and markets products that simplify, automate and innovate complex biomedical testing. For more than 80 years, our products have been making a difference in people's lives by improving the productivity of scientists by supplying critical information for improving patient health and delivering trusted solutions for research and discovery. Scientists can optimize their workflow and increase laboratory efficiency with our centrifugation, particle counting, characterization, liquid handling, flow cytometry and custom integration solutions.

Visit www.beckman.com or at booth #306.

BOOTH 400

The Binding Site, Inc.

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 complement proteins and quantify Immunoglobulins, IgG and IgA subclasses. Run on Optilite®, special protein analyzer, 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.us.bindingsite.com

BOOTH 303

Blueprint Genetics

Blueprint Genetics delivers high-quality genetic testing to the global clinical community. Through our innovative laboratory process, latest sequencing technology with AI empowered data crunching tools, and experienced professionals, we are advancing mainstream healthcare. As the most transparent genetic diagnostic company on the market, we offer full access to our methodology, sensitivity, performance data, and analytic validations of our testing platform.

BOOTH 307

CSL Behring

CSL Behring is a global biotherapeutics leader driven by our promise to save lives. We meet patients' need using the latest technologies to develop and deliver innovative biotherapies that are used to treat serious and rare conditions such as immune deficiencies, coagulation disorders, primary hereditary angioedema and inherited respiratory disease.

BOOTH 405

eClinicalWorks

The eClinicalWorks EHR/PM is a complete solution for your healthcare IT needs, including everything providers need for the testing and treatment of allergies. Allergy module contains configurable worksheets for the antigen tests needed to document wheal/flare or impression. Once skin tests are complete, providers can write formulations for the vials to be prepared.

BOOTH 204

Enzyvant

Enzyvant is a biopharmaceutical company focused on developing innovative treatments for patients with rare diseases. Enzyvant has initiated a rolling BLA submission with the FDA for RVT-802, an investigational tissue-based therapy for the treatment of primary immunodeficiency associated with complete DiGeorge Anomaly.

Enzyvant is simultaneously initiating a clinical trial of RVT-801, an investigational enzyme replacement therapy for the treatment of Farber disease. Enzyvant plans to develop treatments for additional rare diseases with high unmet need.

BOOTH 105

Epimune Diagnostics

Epimune was founded in 2017 with the goal to revolutionize immune cell profiling in humans. Using molecular (i.e. epigenetic) techniques, Epimune develops in-vitro diagnostic tests for quantification of immune cells from a drop of blood. This makes Epimune's approach uniquely suited for applications where fresh blood samples of sufficient quality or quantity are limited — e.g. patient monitoring in remote areas, near-patient testing or newborn screening.

EXHIBITOR LISTING

BOOTH 403

GeneDx

GeneDx was founded in 2000 by two scientists from the National Institutes of Health (NIH) to diagnose patients with rare disorders and assist the clinicians responsible for treating these patients. Led by its world-renowned clinical genomics program, GeneDx has cutting-edge diagnostic testing for a majority of inherited genetic disorders. www.genedx.com

BOOTH 304

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 404

Horizon Pharma

Horizon Pharma plc is a biopharmaceutical company focused on improving patients' lives by identifying, developing, acquiring and commercializing differentiated and accessible medicines that address unmet medical needs. We market 11 medicines through our rare disease, rheumatology and primary care business units.

BOOTH 102

Immune Deficiency Foundation

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 301

Invitae

Invitae's mission is to bring comprehensive genetic information into mainstream medical practice to improve the quality of healthcare for billions of people. Our goal is to aggregate most of the world's genetic tests into a single service with higher quality, faster turnaround time and lower prices. Visit www.invitae.com.

BOOTH 205

Kroger Specialty Infusion

Kroger Specialty Infusion 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 comprehensive patient advocacy, support program tailored to the individualized needs of IgG patients and their caregivers.

BOOTH 202

Leadiant Biosciences Inc.

Leadiant Biosciences, Inc. is a rare corporation dedicated to creating novel medicines for the unmet needs of patients with rare diseases. Truly unique in its field, Leadiant Biosciences places its considerable scientific resources behind the discovery of compounds that benefit the few. Simply because it's the right thing to do.

By maintaining an environment based on integrity, commitment, and placing the patient first, Leadiant Biosciences is able to consider what may be a small commercial success nothing less than a human triumph.

BOOTH 300

Medical College of Wisconsin

The Clinical Immunodiagnostic and Research Laboratory (CIRL) and Allergy Immunology Diagnostic Lab Center (AIDLC), at MCW, perform comprehensive testing for the diagnosis of primary immunodeficiencies (PIDs) and allergic disorders. The CIRL performs immunophenotyping and flow cytometric-based functional assays to aid in the screening and diagnosis 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 103

Novartis

Our mission at Novartis is to discover new ways to improve and extend people's lives. We use science-based innovation to address some of society's most challenging healthcare issues and develop breakthrough treatments, and find new ways to deliver them to as many people as possible.

BOOTH 201

Takeda Pharmaceutical Company Limited

Takeda is a global, values-based, R&D-driven biopharmaceutical leader headquartered in Japan, committed to bringing Better Health and a Brighter Future to patients by translating science into highly-innovative medicines. Takeda focuses its R&D efforts on four therapeutic areas: Oncology, Gastroenterology (GI), Neuroscience and Rare Diseases. We also make targeted R&D investments in Plasma-Derived Therapies and Vaccines. www.takeda.com

BOOTH 304

Sobi, Inc.

Sobi, Inc. is an international biopharmaceutical company dedicated to rare diseases. Primary hemophagocytic lymphohistiocytosis (HLH) is an ultra-rare, rapidly progressive, often-fatal syndrome of hyperinflammation in which hyperproduction of interferon gamma may lead to immune system hyperactivation, ultimately leading to multiple organ failures. Diagnosis is challenging due to the variability in signs and symptoms, so increased awareness is important. Treatment options for primary HLH will be discussed. The immediate goal of treatment is to quickly control the hyperinflammation and to prepare for hematopoietic stem cell transplantation.

BOOTH 107

TruAllergy

Misunderstanding Food Allergies and Food Intolerances may lead to unnecessary dietary restrictions. The TruAllergy™ and TruTolerance™ Dried Bloodspots (DBS) Test system in Conjunction with a amplified immune diagnostic monitoring system (AIMS) Smart Report provides clinicians with the tools to clear up the confusion.

EXHIBITOR LISTING

BOOTH 104

USIDNET

The United States Immunodeficiency Network (USIDNET) is an NIH-funded research program of the Immune Deficiency Foundation (IDF), established to advance scientific research in the field of primary immunodeficiency diseases (PI).

BOOTH 207

X4 Pharmaceuticals

X4 Pharmaceuticals, headquartered in Cambridge, MA, is developing novel medicines designed to improve immune cell trafficking and restore healthy immunity in certain rare, genetic primary immunodeficiency diseases, specifically WHIM syndrome and Severe Congenital Neutropenia, as well as cancer. X4's oral small molecule drug candidates antagonize the CXCR4 pathway, which plays a central role in immune surveillance. For more information, please visit www.x4pharma.com.

POSTER SESSION I: FRIDAY, APRIL 5

Posters highlighted in blue will also be presented as lightning posters.

POSTER	TITLE	PRESENTER	INSTITUTION
1	Alpha Fetoprotein Levels in Ataxia Telangiectasia as Related to Age, Disease Characteristics and Outcomes	Ariela Agress	Westchester Medical Center
2	Human PLCG2 Haploinsufficiency Results in NK Cell Immunodeficiency and Herpesvirus Susceptibility	Joshua Alinger	St. Louis Children's Hospital
3	Immune Dysregulation: Diagnosis of Behcets Disease in an Affected Chronic Granulomatous Disease Carrier	Aba Al-Kaabi	University of Kansas Medical Center
4	Risk of Bacterial Infections Among Patients with Secondary Complement Deficiency	Taha Al-Shaikhly	Division of Allergy & infectious Diseases, University of Washington
5	Benign Reactive Gamma Delta T Cells Proliferation in Spleen – “The Mirage Effect”	Snegha Ananth	UTHSCSA
6	Infective Endocarditis, Osteomyelitis of Skull and Invasive Aspergillosis in a Child with Chronic Granulomatous Disease	Gummadi Anjani	Postgraduate Institute Of Medical Education and Research
7	Fertility, Pregnancy, and Progeny Outcomes in a Large Academic Cohort of Patients with Antibody Deficiencies	Carolyn Baloh	Duke University Medical Center
8	T-cell Receptor Repertoire Clonality in Peripheral Blood and Affected Tissue in Activated PI3 Kinase Delta Syndrome (APDS)	Sara Barmettler	Massachusetts General Hospital
9	Deficiency of Adenosine Deaminase 2: An Expanding Spectrum of Disease	Jenna Bergerson	NIH/NIAID/LCIM
10	A Family with Hypogammaglobulinemia, ACTH Deficiency, Ectodermal Dysplasia and a Novel NFKB2 Mutation	Marc Bienias	Department of Pediatrics, Medizinische Fakultät Carl Gustav Carus, Technische Universität Dresden
11	Isolated Central Nervous System Disease in Familial Hemophagocytic Lymphohistiocytosis – a Multicenter Case Series	Annaliese Blincoe	CHU Sainte-Justine, University of Montreal
12	Artificial Thymic Organoids Represent a Reliable and Quick Tool to Study T Cell Differentiation in Human Bone Marrow Samples from Patients with Severe T Cell Immunodeficiency	Marita Bosticardo	Laboratory of Clinical Immunology and Microbiology, IDGS, DIR, NIAID, NIH
13	A Demonstration of the Diagnostic and Clinical Utility of Genomic Sequencing in Primary Immunodeficiency Diseases in Australia	Vanessa Bryant	Walter & Eliza Hall Institute/Royal Melbourne Hospital
14	American Society of Pediatric Hematology and Oncology (ASPHO) Clinical Immunology Special Interest Group (SIG): Expanding Clinical Immunology Education, Research and Care	David Buchbinder	Children's Hospital of Orange County & University of California at Irvine
15	A Case of Immunodeficiency, Centromeric Instability, and Facial Anomalies Syndrome (ICF) with NK Deficiency and Subsequent EBV-driven Diffuse Large B-cell Lymphoma Treated with Bone Marrow Transplant	Caitlin Burk	UPMC Children's Hospital of Pittsburgh
16	Human STAT5 Deficiency Results in an Increase of Follicular T Cells Leading to Expanded Germinal Center B Cells and Autoimmunity	Maria Caldirola	Grupo de Inmunología Hospital de Niños “R.Gutierrez”- IMIPP-CONICET, Buenos Aires-Argentina
17	Genotype-phenotype Correlation in Human RAG1 Deficiency	Enrica Calzoni	Laboratory of Clinical Immunology and Microbiology, IDGS, DIR, NIAID, NIH
18	Sexual Dimorphism and AIRE-AIRE Interactors-miRNA Coexpression Networks in the Infant Human Thymus	Magda Carneiro-Sampaio	Department of Pediatrics, Faculdade de Medicina da Universidade de Sao Paulo
19	Yellow Fever: Is It Possible to Vaccinate Patients with IgA Deficiency?	Magda Carneiro-Sampaio	Department of Pediatrics, Faculdade de Medicina da Universidade de Sao Paulo
20	Characterization of Gut Inflammation and Autoimmunity in Mice Carrying Rag1 Hypomorphic Mutations	Riccardo Castagnoli	National Institute of Allergy and Infectious Diseases, National Institutes of Health & University of Pavia
21	HSCT Utilizing Related Carrier Donor for CD40 Ligand Deficiency	Shanmuganathan Chandrakasan	Children's Healthcare of Atlanta, Emory University

POSTER SESSION I: FRIDAY, APRIL 5

POSTER	TITLE	PRESENTER	INSTITUTION
22	Longitudinal Follow up of EBV-driven Lymphoproliferative Disease and Combined Immunodeficiency in RASGRP1 Deficiency: Successful Treatment and Allogenic Matched Unrelated Bone Marrow Transplant	Alice Chau	University of Washington
23	T and NK Cell Dysfunction Arising from BCL11B Deficiency	Samuel Chiang	Cincinnati Children's Hospital Medical Center
24	Treatment-resistant Autoimmune Cytopenias as a Sign of Primary Immunodeficiency Disorders (PIDs)	Sara Ciullini Mannurita	University of Florence, Dep. Neurosciences, Psychology, Drug Research and Child Health (NEUROFARBA), Florence, Italy.
25	Multigenicity of the Deficit of the Immune System: Novel Frontiers of Primary Immunodeficiencies (PIDs)	Sara Ciullini Mannurita	University of Florence, Dep. Neurosciences, Psychology, Drug Research and Child Health (NEUROFARBA), Florence, Italy.
26	Specific Functional Gammopathy Underlying Infectious Susceptibility in a Patient with Autoimmune-Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy (APECED)	Gregory Constantine	Fungal Pathogenesis Section, Laboratory of Clinical Immunology & Microbiology (LCIM), National Institute of Allergy & Infectious Diseases (NIAID), NIH
27	Defective B Cell Fitness Impairs Mutation Away from Self and Sustains Red Blood Cell Reactivity in Hypomorphic RAG Deficiency	Krisztian Csomos	University of South Florida
28	A Case of Autoinflammatory Syndrome with Osteoporosis and Specific Antibody Deficiency	Irina Dawson	Children's Research Institute, University of South Florida
29	Anti-cytokine Antibodies Emerge After Viral Infections and Persist in Patients in Partial RAG Deficiency	Irina Dawson	Children's Research Institute, University of South Florida
30	Immunomodulatory Effects of Immunoglobulin Replacement Therapy on T-cells in Patients with Hypogammaglobulinemia	Tri Dinh	University of Ottawa
31	Rapid Identification of Patients with RAG Mutations Using Valpha 7.2 Antibody	Kerry Dobbs	Laboratory of Clinical Immunology and Microbiology, NIAID, NIH
32	Sphingosine-1-phosphate Lyase Deficiency Identified by Newborn T Cell Receptor Excision Circle Screening for Severe Combined Immunodeficiency	Cullen Dutmer	Children's Hospital Colorado - University of Colorado School of Medicine
33	Abnormal B Cell Function in WHIM Syndrome	Maryssa Ellison	University of South Florida
34	Encephalopathy in an Adolescent with CD40-ligand Deficiency	Elizabeth Feuille	Weill Cornell Medicine
35	Clinical Phenotyping of a DOCK8 Deficiency Cohort	Alexandra Freeman	National Institutes of Health
36	Expanding Phenotypes: A Complex Case of an Adult Patient with DNA Repair Defect and Immunodeficiency	Claudia Gaefke	University of South Florida, Division of Allergy and Immunology
37	Novel CDC42 Mutation Causes Severe Autoinflammatory Syndrome Responsive to IL-1 Inhibition	Yael Gernez	Stanford School of Medicine
38	Unexpected Diagnosis in a Family with Autoimmune Multilineage Cytopenia and Hypogammaglobulinemia	Yael Gernez	Stanford School of Medicine
39	Extreme Phenotypes, Identical Mutations: Two Patients with Same Nonsense XLF/Cernunnos Homozygous Mutation	Luis Gonzalez-Granado	Hospital 12 de Octubre
40	Guidance for the Care of Patients Undergoing Cultured Thymus Tissue Transplantation (RVT-802)	Stephanie Gupton	Division of Allergy, Immunology and Pulmonary, Duke University Medical Center
41	Smoldering Hemophagocytic Lymphohistiocytosis Secondary to Compound Heterozygous Variants in SLCA7 Treated with Anakinra	Nicholas Hartog	Helen DeVos Children's Hospital and Michigan State University
42	What We Are Missing with PID Exomes, Including Poorly Covered Exons	Sarah Henrickson	Children's Hospital of Philadelphia
43	Clinical and Laboratory Features of Thymoma and Immunodeficiency (Good's Syndrome): A Report from the USIDNET Registry and the Mount Sinai Hospital Cohort	Hsi-en Ho	Icahn School of Medicine at Mount Sinai

POSTER SESSION I: FRIDAY, APRIL 5

POSTER	TITLE	PRESENTER	INSTITUTION
44	Show Me the Phenotype: The Ordering Clinician's Role in Genetic Variant Interpretation for Primary Immunodeficiency Diseases	Jennifer Holle	Invitae
45	Droplet Digital PCR Analysis of GATA2 Deficiency	Amy Hsu	NIH
46	Missense Variants as a Contributing Cause to DOCK8 Immune Deficiency	Haley Hullfish	National Institute of Allergy and Infectious Disease (NIAID)
47	IgD Class Switched B Cells in Patients with Common Variable Immunodeficiency	Taissa Kasahara	State University of Rio de Janeiro and University of California Irvine
48	Allogeneic Hematopoietic Stem Cell Transplant Outcomes for Patients with Dominant-Negative IKZF1/IKAROS Mutations	Erinn Kellner	Division of Bone Marrow Transplantation and Immune Deficiency, Cincinnati Children's Hospital Medical Center
49	Reduced-intensity, T Cell-replete, Alternative Donor Allogeneic Hematopoietic Cell Transplantation with Post-transplantation Cyclophosphamide Is Safe and Effective for Primary Immune Deficiencies	Orly Klein	Johns Hopkins University School of Medicine
50	Primary Immune Deficiency Disease in Patients over Age 60: An Analysis from a Proprietary Immunology Patient Registry	Roger Kobayashi	Allergy Asthma & Immunology Associates
51	Autologous Ex Vivo Lentiviral Gene Therapy for the Treatment of Severe Combined Immune Deficiency Due to Adenosine Deaminase Deficiency	Donald Kohn	UCLA
52	Autologous Ex Vivo Lentiviral Gene Therapy for the Treatment of Severe Combined Immune Deficiency Due to Adenosine Deaminase Deficiency Improves B Cell Function	Donald Kohn	UCLA
53	Quality of Life in Adult Patients with Chronic Granulomatous Disease	Samantha Kreuzburg	National Institutes of Health/National Institute of Allergy and Infectious Diseases
54	A Novel Germline IKAROS C-terminal Mutation in a Patient with Burkitt Lymphoma, Lymphoproliferation and Cytopenias	Hye Sun Kuehn	Immunology Service, Department of Laboratory Medicine, Clinical Center, NIH
55	Nucleus-retained WASP Is Deleterious to T-cell Development	Carole Le Coz	Children's Hospital of Philadelphia
56	Dosing Ruxolitinib for the Treatment of Immunodysregulation in STAT1-GOF	Jennifer Leiding	University of South Florida
57	Telomeres in Schimke Immuno-Osseous Dysplasia: Comparing Telomere Length in Individuals with Homozygous and Heterozygous SMARCA1 Mutations	Elizabeth Lippner	Stanford University Medicine & Lucile Packard Children's Hospital
58	The Forest and the Trees: Machine Learning to Classify Cases of Suspected Inborn Errors of Immunity Using Decision Tree and Random Forest Algorithms	Saul Lugo Reyes	Immunodeficiencies Research Unit, National Institute of Pediatrics, Mexico City
59	Failing to Make Ends Meet: The Broad Clinical Spectrum of DNA Ligase IV Deficiency Case Series and Review of the Literature	Saul Lugo Reyes	Immunodeficiencies Research Unit, National Institute of Pediatrics, Mexico City
60	Immunological and Genetic Outcomes of Infants with Positive Newborn Screening for Severe Combined Immunodeficiency (SCID)	Vasudha Mantravadi	Department of Pediatrics, Washington University School of Medicine, St. Louis, Missouri, 63110
61	Quercetin Halts Abnormal IL-1 β and IL-18 Production: A Natural Calm for XIAP Deficiency?	Rebecca Marsh	Cincinnati Children's Hospital Medical Center
62	Granulomatous Disease and Lymphoma in a Cohort of 1395 Patients with CVID in the USIDNET Registry	Joao Pedro Matias Lopes	Icahn School of Medicine at Mount Sinai
63	FAS-mediated Apoptosis Assay in Patients with ALPS/ALPS-like Phenotype Carrying CASP10 mutations	Maurizio Miano	IRCCS Istituto Giannina Gaslini
64	Osteomyelitis in Chronic Granulomatous Disease: Experience from a Tertiary Care Centre in North-West India	Johnson Nameirakpam	Allergy and Immunology Unit, Advanced Pediatrics Centre, Postgraduate Institute of Medical Education and Research
65	The Role of Glycosylation Modification on Resistance to Viral Infections	Cristiane Nunes-Santos	Immunology Service, Department of Laboratory Medicine, Clinical Center, NIH

POSTER SESSION I: FRIDAY, APRIL 5

POSTER	TITLE	PRESENTER	INSTITUTION
66	The Effect of Hydroxychloroquine on CTLA4 Expression in Siblings with LRBA (Lipopolysaccharide-responsive and Beige-like Anchor Protein) Deficiency	Nurcicek Padem	Northwestern University
67	Single Cell RNAseq Analysis Reveals Profound Abnormalities in the Distribution and Diversity of Thymic Epithelial Cells in Wild-type and Rag1 Mutant Mice	Francesca Pala	NIAID, NIH
68	Immunodeficiency in Elderly: Data from the USIDNET Registry	Charmi Patel	Donald Barbara Zucker School of Medicine at Hofstra/Northwell
69	Oral Immunoglobulins to Treat Norovirus Gastroenteritis in Patients with Primary and Secondary Immunodeficiency	Matthew Perez	Baylor College of Medicine
70	Two Siblings with a Delayed/Late-Onset Presentation of Combined Immunodeficiency Due to Adenosine Deaminase Deficiency	Tamara Pozos	Children's Hospital Minnesota
71	Hyperactivated PI3Kd Promotes Self and Commensal Reactivity at the Expense of Optimal Humoral Immunity	Silvia Preite	National Institutes of Health
72	Quantitation of T Cell Repertoire Diversity Following Treatment for SCID	Jennifer Puck	University of California, San Francisco
73	Newborn Screening for SCID in Puerto Rico: A Three-year Experience	Cristiana Ramos-Romey	University of Puerto Rico School of Medicine
74	Interim Analysis of Infusion Characteristics and Adverse Events During Facilitated Subcutaneous Immunoglobulin Treatment for Primary Immunodeficiency Diseases: Global Post Authorization Safety Study	Arye Rubinstein	Albert Einstein College of Medicine & Montefiore Medical Center
75	Heterozygous Variants in FOXP1 in Infants with Abnormal Newborn Screening for SCID	Lauren Sanchez	Department of Pediatrics, Division of Allergy, Immunology, and Bone Marrow Transplant, University of California, San Francisco
76	Plasma Metabolomic Signatures in Patients with Chronic Granulomatous Disease	Christopher Santaralas	University of South Florida
77	Indications of Depressive Disorders in Adults with Primary Immunodeficiency	Christopher Scalchunes	Immune Deficiency Foundation
78	Epigenetic Immune Cell Quantification for Diagnosis and Monitoring of Patients with Primary Immune Deficiencies and Immune Regulatory Disorders	Janika Schulze	Department of Pediatrics, Medizinische Fakultät Carl Gustav Carus, Technische Universität Dresden, Dresden, Germany
79	Two Siblings with Autoimmune Polyendocrinopathy-candidiasis-ectodermal Dystrophy-like Phenotype Demonstrating Classic and Atypical Symptoms	Edith Schussler	Weill Cornell Medicine
80	Ex Vivo Generation and Single-Cell Analysis of Human Monoclonal Antibodies from Dengue Virus Infected Patients	Pragati Sharma	International center for genetic engineering and biotechnology, New Delhi, India
81	Enigmas of IL-12R β Deficiency: Contemporary of Two Disease, Mendelian Susceptibility to Mycobacterial Disease and Crohn Disease	Roya Sherkat	Acquired Immunodeficiency Research Center, Isfahan University of Medical Sciences, Isfahan, Iran
82	Cytomegalovirus Specific Cell-mediated Immunity Status in Women with Pre-eclampsia: A Case-control Study	Roya Sherkat	Acquired Immunodeficiency Research Center, Isfahan University of Medical Sciences, Isfahan, Iran
83	The B-cell Subset Milieu of Autoimmune Cytopenias in Primary Immunodeficiency	Travis Sifers	Icahn School of Medicine at Mount Sinai
84	Deficiency of the Non-classical Inhibitor of NF-kappaB, IkappaBNS, Causes a Novel Primary Immunodeficiency Due to Dysregulated NF-kappaB Signaling	Charlotte Slade	Walter & Eliza Hall Institute/Royal Melbourne Hospital
85	Variant Mutation in PLCG2 Associated with Common Variable Immunodeficiency Without Cold Urticaria	Veronica Solivan-Vargas	Hospital Episcopal San Lucas
86	Epidemiology on Primary Immunodeficiencies in Korea: A Systematic Review of Reported Literature and Analysis of Bigdata from National Health Insurance System	Sohee Son	Department of Pediatrics, Samsung Medical Center, Sungkyunkwan University School of Medicine, Seoul, Korea
87	Thinking Outside of Infection: Hemophagocytic Lymphohistiocytosis in a 5-week-old Male with Chronic Granulomatous Disease and Burkholderia Cepacia Sepsis	Jacqueline Squire	USF - John Hopkin's All Children's Hospital

POSTER SESSION I: FRIDAY, APRIL 5

POSTER	TITLE	PRESENTER	INSTITUTION
88	Herpes Simplex Virus Whole Genome Sequencing for Antiviral Resistance in a Child with DOCK8 Deficiency and Chronic Infection	Sean Stout	Children's Mercy Hospital
89	A 29-year-old Woman with History of Hypogammaglobulinemia and Acute Liver Failure	Beth Thielen	University of Minnesota, Departments of Internal Medicine and Pediatrics, Divisions of Infectious Diseases and International Medicine and Pediatric Infectious Diseases and Immunology
90	Miller-Dieker Syndrome May Be Another Syndromic Primary Immunodeficiency	Erika Tsutsui	The University of Tokyo Hospital
91	Patient Education with a Self-Efficacy Focus for Adult Autosomal Dominant Hyper IgE Syndrome Patients	Amanda Urban	Clinical Monitoring Research Program Directorate, Frederick National Laboratory for Cancer Research sponsored by the National Cancer Institute
92	Profiling Serum Antibody Specificities in Healthy Toddlers Reveals a Subgroup with Strong IgG Responses to Autoantigens and Infectious Agents	Nicolai van Oers	UT Southwestern Medical Center
93	Diagnostic Yield of a Next-Generation Sequencing Panel for Primary Immunodeficiencies in a Cohort of Pediatric Patients with Immunohematologic Disorders	Elizabeth Varga	Division of Hematology/Oncology/BMT and the Institute for Genomic Medicine, Nationwide Children's Hospital
94	Evaluation of Novel STAT1 Mutations: Phosphorylation, Luciferase Assay or Both?	Alexander Vargas Hernandez	Department of Pediatrics, Baylor College of Medicine, Houston, TX, USA.
95	Loss of Human ICOSL Results in Combined Immunodeficiency	Don Vinh	Research Institute - McGill University Health Centre
96	Neutralizing anti-IL-6-autoantibodies Are a Risk Factor for Pyogenic Bacterial Infections	Horst von Bernuth	Department of Pediatric Pulmonology, Immunology and Intensive Care Medicine, Charité University Medicine, Berlin, Germany
97	Conventional Treatment versus Stem Cell Transplantation — Outcome in 105 Patients with Chronic Granulomatous Disease	Horst von Bernuth	Department of Pediatric Pulmonology, Immunology and Intensive Care Medicine, Charité University Medicine, Berlin, Germany
98	Centralized Sequencing Initiative at NIAID: Year 1	Magdalena Walkiewicz	National Institute of Allergy and Infectious Diseases (NIAID)
99	Treatment of Adenosine Deaminase Severe Combined Immunodeficiency with Pegylated Recombinant Adenosine Deaminase. a Clinical Trial of Patients Transitioned from Pegademase to Elapegademase-lvr	Joseph Wiley	Leadant Biosciences, Inc.
100	Cure of a Complicated Patient with X-linked Lymphoproliferative Syndrome Type I and Hepatitis C Cirrhosis, Through Combined Living Related Liver and Hematopoietic Stem Cell Transplantation	Christian Wysocki	UT Southwestern Medical Center
101	Transcriptome Analysis Reveals an Important Role for EXTL3 in Human Hematopoietic Cell Differentiation	Yasuhiro Yamazaki	NIAID NIH
102	Different Clinical Manifestations in a Large Cohort of Predominantly Antibody Deficiency Patients with Monogenic Defects	Reza Yazdani	Research Center for Immunodeficiencies, Children's Medical Center, Tehran University of Medical Sciences, Tehran, Iran
103	Infection Rates and Tolerability in Patients with Primary Immunodeficiency Diseases Treated with Three Different Immunoglobulin Administration Modalities	Leman Yel	Allergy Partners of North Texas Research
104	Lymphocyte Radiosensitivity in Cartilage Hair Hypoplasia	Jennifer Yonkof	Division of Allergy and Immunology, Nationwide Children's Hospital
105	Capturing Quality of Life in Patients with Common Variable Immunodeficiency (CVID) Using the Patient-Reported Outcomes Measurement Information System (PROMIS-29) Survey	Shouling Zhang	Department of Pediatrics, Donald and Barbara Zucker School of Medicine at Hofstra/Northwell, New Hyde Park, NY
106	Describing Transient T Cell Lymphopenia in the United States Immunodeficiency Network (USIDNET) Following Infants with Low Lymphocytes (FILL) Program and a Single Referral Center from 2010-2017	Shouling Zhang	Department of Pediatrics, Donald and Barbara Zucker School of Medicine at Hofstra/Northwell, New Hyde Park, NY

POSTER SESSION II: SATURDAY, APRIL 6

Posters highlighted in blue will also be presented as lightning posters.

POSTER	TITLE	PRESENTER	INSTITUTION
1	Visualizing the Effect of Lymphatic Pump Techniques on Immune System in Normal Subjects “ Randomized Control Trial”	Ahmed Abdelfattah	Faculty of Physical Therapy - Cairo University
2	Procedure Related Airway Spasms in GOF PI3K Patients with Airway Nodular Hyperplasia	Anahita Agharahimi	NIH
3	A Case of C6 Complement Deficiency with a Novel Mutation	Hassan Ahmad	Rush University Medical Center
4	Case of WHIM Syndrome with Unique CXCR4 Variant	Hassan Ahmad	Rush University Medical Center
5	Atypical Presentation of Complete DiGeorge Syndrome Without Correlating Genetic Defect: Rescued by State Newborn Screening	Aba Al-Kaabi	University of Kansas Medical Center
6	A New High Concentration Immunoglobulin Product for Subcutaneous Administration (IGSC 20%)	William Alonso	Grifols Bioscience Research Group
7	Hyper IgM2 Diagnosed in a Brazilian Boy	Maine Bardou	Reference Center on Rare Diseases, Faculdade de Medicina ABC
8	PIK3CD, a Rare Autosomal Dominant Disorder of the Immune System: A Reason for the Use of Next Generation Sequencing	Jacob Barish	University of Florida, Department of Internal Medicine
9	Atopic Complications Associated with Elevated IgE in a Subset of Common Variable Immunodeficiency	Sara Barmettler	Massachusetts General Hospital
10	Molecular Study in Children with Chronic Granulomatous Disease (CGD) at a Tertiary Care Center in North India	Dharmagat Bhattarai	Postgraduate Institute of Medical Education and Research, Chandigarh
11	Clinical Description of Rosacea in a Family with STAT 1 GOF Mutation	Lizbeth Blancas Galicia	National Institute of Pediatrics
12	Two-year-old Male with Recurrent Cervical Lymphadenopathy Presenting with Rash	Deborah Bloch	Emory University School of Medicine
13	Variable Phenotypes in Three Patients with Two Novel STAT3 Gain of Function Mutations	Musa Bolkent	University of Florida
14	XLA Presenting as HHV-6 Meningitis and Pseudomonas Meningitis/Bacteremia in a Patient Without History of Recurrent Infections	Melissa Cardenas-Morales	Nicklaus Children’s Hospital
15	Coexistence of Lymphoproliferative Syndrome, Neurofibromatosis, Systemic Lupus Erythematosus and Hyper IgM Syndrome in a Patient with MSH6 Mutation	Sukru Cekic	Uludag University Faculty of Medicine
16	CD27 Deficiency Causes Human NK Cell Deficiency with Specific Loss of the CD56(bright) Subset: A Single Case Report	Natalia Chaimowitz	Texas Children’s Hospital/Baylor College of Medicine
17	Pill Endoscopy as a Diagnostic Tool for an Abdominal Exacerbation in a Pediatric Patient with Hereditary Angioedema: A Case Report	Yatyng Chang	Nicklaus Children’s Hospital
18	Dose and Clinical Outcomes in Patients with CVID and Bronchiectasis Receiving Immunoglobulin Replacement Therapy in the Home	Allyson Checkley	Coram CVS Specialty Infusion Services
19	Common Variable Immunodeficiency (CVID) Associated with Variants in Neuroblastoma Amplified Sequence (NBAS) Gene	Joseph Church	Children’s Hospital Los Angeles and Keck School of Medicine of U.S.C.
20	When Mosquito Bite Allergy Is Treated with Bone Marrow Transplantation (BMT)	Joseph Church	Children’s Hospital Los Angeles and Keck School of Medicine of U.S.C.
21	A Case of Burkholderia Cepacia in a Child with Takayasu Arteritis	Michelle Clark	Nemours/Alfred I. duPont Hospital for Children
22	A Case of Memory B-cell Dysfunction in a Child with Recurrent Otitis Media	Arjola Cosper	Goryeb Children’s Hospital
23	A Case of Complement Factor D Deficiency with Streptococcus Pneumoniae Pneumonia with Associated Lung Abscess and Empyema	Ashleah Courtney	Arkansas Children’s Hospital, University of Arkansas for Medical Sciences
24	Lupus-like Syndrome in a Patient with NOD2-associated Autoinflammatory Disease	Lyda Cuervo-Pardo	University of Florida, Division of Rheumatology & Clinical Immunology, Department of Medicine
25	Bartonella Endocarditis in a Child with Probable ALPS	Keerti Dantuluri	Vanderbilt University Medical Center

POSTER SESSION II: SATURDAY, APRIL 6

POSTER	TITLE	PRESENTER	INSTITUTION
26	Siblings with Copy Number Gain in ATM Results in Variable Clinical Phenotypes and Defects in ATM, SMC1, and H2AX Phosphorylation	Jasmeen Dara	UCSF Benioff Children's Hospital
27	6 Month Old Female with Congenital Onset Indolent Systemic Mastocytosis Successfully Treated with Midostaurin	Christin Deal	UCLA
28	NGS Reveals Repertoire Restriction of Treg Cells in APDS1 Patients	Ottavia Delmonte	National Institutes of Health
29	Patient with Hypohidrotic Ectodermal Dysplasia and Recurrent Infections Mimicking NEMO-Deficiency Syndrome	Roman Deniskin	Baylor College of Medicine/Texas Children's Hospital
30	A Novel Mutation in the Cytotoxic T-lymphocyte Antigen-4 (CTLA-4) Gene with Cytopenias, Interstitial Lung Disease, Hypogammaglobulinemia and Recurrent Bacterial Endocarditis	Victoria Dimitriades	University of California, Davis
31	Recurrent Sinusitis in Heterozygous Hemochromatosis; Is It a Risk Factor?	Elif Dokmeci	University of New Mexico
32	Non-Arteritic Anterior Ischemic Optic Neuropathy in a Patient with XIAP Deficiency: Expanding the Inflammatory Ocular Findings in X-Linked Lymphoproliferative Disorders	Mark Dulchavsky	University of Michigan Medical School
33	An Unexpected Diagnosis in a Premature Infant with Persistent Fever, Respiratory Distress and Significant Neutrophilia: Congenital Tuberculosis	Idil Ezhuthachan	Department of Pediatrics, Zucker School of Medicine at Hofstra Northwell School of Medicine
34	Recurrent Viral Encephalitis in a Child with Several Variants of Uncertain Significance in Primary Immunodeficiency Genes	Idil Ezhuthachan	Department of Pediatrics, Zucker School of Medicine at Hofstra Northwell School of Medicine
35	Late Adaptive Immune Dysfunction 34 Years After Unconditioned Allogeneic Stem Cell Transplant for T-B-NK+ SCID with Novel RAG1 Mutations	Lauren Franzblau	Department of Internal Medicine, UT Southwestern Medical Center
36	Four Patients with Refractory Pericarditis Treated with Concurrent Hyaluronidase-facilitated Subcutaneous Immunoglobulin and Anti-interleukin 1 Therapy	Melissa Gans	Montefiore Medical Center
37	NIH Participation to USIDNET Registry (Poster Submission)	Elizabeth Garabedian	National Genome Research Institute, National Institutes of Health
38	A Case of Disseminated Pneumocystis Jiroveci in a Non-Human Immunodeficiency Virus Infected Patient	Yael Gernez	Division of Allergy and Immunology, Department of Pediatrics, Stanford School of Medicine
39	Combined Immunodeficiency in a Patient with a Heterozygous TNFRSF13B (TACI) Variant	Amanda Grippen Goddard	National Jewish Health
40	Etanercept Use in Refractory Chronic Henoch-Schönlein Purpura	Sana Habib	University of New Mexico
41	Predictors of Fatigue in Common Variable Immunodeficiency	Joud Hajjar	Baylor College of Medicine, 1Texas Children's Hospital Center for Human Immunobiology and Division of Immunology, Allergy and Rheumatology
42	Natural History of X-linked Lymphoproliferative Disease, Lessons Learned from a Long-term Survivor	Joud Hajjar	Baylor College of Medicine, Texas Children's Hospital Center for Human Immunobiology and Division of Immunology, Allergy and Rheumatology
43	Antibody Responses After Vaccination with Prevnar13® in IgG Subclass Deficient Patients	Stephen Harding	The Binding Site
44	Assay Characteristics of an Automated, Liposome-based Assay for the Measurement of CH50 Complement Activity and Comparison with a Haemolytic Method	Stephen Harding	The Binding Site
45	An Assay to Measure the Complement Binding Activities of Anti-dsDNA Antibodies in SLE	Stephen Harding	The Binding Site
46	Functional Determination of All Possible Disease-associated Variants of a Region in CARD11 Using Saturation Genome Editing	Richard James	Seattle Children's Research Institute
47	Genetic Testing Reveals a Homozygous RTEL1 Mutation in a 12 Month Old Female with Pancytopenia, Failure to Thrive and Low Immunoglobulins	Artemio Jongco	Northwell Health

POSTER SESSION II: SATURDAY, APRIL 6

POSTER	TITLE	PRESENTER	INSTITUTION
48	Infants with Idiopathic Transient and Persistent T Cell Lymphopenia Identified by Newborn Screening — a Single Center's Experience from September 2010 — December 2017	Artemio Jongco	Division of Allergy & Immunology, Donald and Barbara Zucker School of Medicine at Hofstra/Northwell; Center for Health Innovations and Outcomes Research, Feinstein Institute for Medical Research
49	Health Disparities in CVID: a Report of 1,546 Patients from the USIDNET Registry	Avni Joshi	Mayo Clinic College of Medicine
50	Refractory Giardia Infection in a Patient with Common Variable Immune Deficiency	Ekta Kakkar	National Jewish Health and The University of Colorado
51	A Novel Mutation in Zap 70 Leading to an Infant with T+B+NK+ Severe Combined Immunodeficiency	Kelsey Kaman	Baystate Medical Center
52	NBAS Compound Heterozygous Variants as a Cause of Recurrent Acute Liver Failure Triggered by Common Childhood Infections	Suthida Kankirawatana	Division of Allergy and Immunology, Department of Pediatrics, Children's of Alabama
53	Plastic Bronchitis and Secondary T-cell Lymphopenia	Saara Kaviany	Vanderbilt Children's Hospital
54	Hexaviral-Specific T-cells for Treatment and Prevention of Viral Infections Post Hematopoietic Stem Cell Transplant	Michael Keller	Children's National Health System
55	DLCO Is a Reliable Noninvasive Approach for Pulmonary Monitoring in Patients with HIES	Alyssa Kerber	Pediatric and Adolescent Medicine, Mayo Clinic
56	CVID Plasma Promotes Early Commitment to the Follicular Lineage	Caroline Khanna	Children's Hospital of Philadelphia
57	Epidemiology of Anti-epileptic Drug Induced Hypogammaglobulinemia in a Tertiary Care Network	ErinMarie Kimbrough	Mayo Clinic Jacksonville
58	XMEN: MAGT1 Mutation Associated Immunodeficiency. Case Report of an Atypical Presentation	Alejandra King	Universidad de Chile
59	The First Year: Experience from Mayo Clinic Laboratories After Clinical Implementation of Nine Primary Immunodeficiency Next Generation Sequencing Tests	Michelle Kluge	Department of Laboratory Medicine and Pathology, Mayo Clinic
60	A Novel Genetic Etiology for FAS-associated Protein with Death Domain Deficiency	Lisa Kohn	Pediatrics in the Division of Allergy, Immunology, and Rheumatology at UCLA
61	Efficacy, Tolerability and Safety of Cutaquig®, a New Subcutaneous Human Immunglobulin 16.5% in Adult Patients with Primary Immunodeficiencies	Elena Latysheva	National Research Center Institute of Immunology FMBA
62	A Case of Lymphopenia in Rosai Dorfman Disease	Lisa Liang	Memorial University
63	Autoimmune Lymphoproliferative Syndrome with Histopathologic Features of Castleman Disease	Rachelle Lo	Division of Allergy and Immunology, Department of Pediatrics, Stanford University School of Medicine
64	Auto-inflammation and Immunodeficiency — 2 Genes One Presentation	Amarilla Mandola	Canadian Centre for Primary Immunodeficiency, The Roifman Laboratory, Research Institute Division of Immunology and Allergy, Department of Paediatrics, The Hospital for Sick Children, University of Toronto
65	Generating a CYBB-KO THP-1 Cell Line Model for Studying Auto-inflammation in Chronic Granulomatous Disease	Lorie Marchitto	CHU Ste-Justine
66	Ataxia Telangiectasia and Common Variable Immunodeficiency with B-cell Lymphoma in Adolescent	William Marquez	Misericordia Children Hospital, Bogota, Colombia
67	Acute Central Nervous System GvHD After Liver Transplantation	Valérie Massey	Université de Montréal
68	Acquired Immunodeficiency - More Than Meets the BMI (Body Mass Index)	Laura Maurer	Yale New Haven Hospital
69	CARD11 (caspase Recruitment Domain-containing Member 11) Defect: When the Deck Is Stacked	Laura Maurer	Yale New Haven Hospital

POSTER SESSION II: SATURDAY, APRIL 6

POSTER	TITLE	PRESENTER	INSTITUTION
70	Clinical and Laboratory Manifestations of Autoinflammatory Diseases: The Results from the First Iranian Registry	Sahar Memar Montazerin	Acquired Immunodeficiency Research Center, Isfahan University of Medical Sciences, Iran
71	Absolute Neutrophil Counts in Pediatric Duffy Null (FyA-/FyB-) Patients: Assessing Expected Neutrophil Counts in Benign Ethnic Neutropenia	Lauren Merz	University of Michigan Medical School
72	A Novel Form of Partial Recessive IFN-gamma R2 Deficiency Caused by a Mutation of the Initiation Codon Presenting with a Severe Phenotype	Ayse Metin	SBU, Ankara Children's Health and Diseases Hematology Oncology Training and Research Hospital, Turkey
73	Provider Perceptions of Primary Immunodeficiency Disease Patients' Quality of Life, Neurocognition, Physical Well-Being and Psychosocial Health	Thomas Michniacki	University of Michigan
74	The Effects of Adiantum Capillus Hydro Alcoholic Extract on Some Immunological Parameters in Mice	Mehrdad Modaresi	Isfahan (Khorasgan) Branch, Islamic Azad University, Iran
75	Recalcitrant Abdominal Abscesses in a Patient with Hyperimmunoglobulin E Syndrome	Anh Nguyen	University of California Davis Health
76	Novel Mutation in the WAS Gene Causing a Phenotypic Presentation of Wiskott-Aldrich Syndrome	Elisa Ochfeld	Northwestern University
77	Maternal Diabetes Causing Atypical, Complete DiGeorge Syndrome	Richa Panara	Department of Internal Medicine, Emory University School of Medicine
78	Severe Necrotic Reaction to 23-valent Polysaccharide Pneumococcal Vaccine in a Patient with STAT3 Deficiency	Mervin Piñones	Pontificia Universidad Católica de Chile
79	Abatacept for CTLA-4 Haploinsufficiency Presenting with Severe Bone Marrow Aplasia and Septic Shock - A Case Report	Emilie Proulx	CHU de Québec, Université Laval
80	Newborn Infant with Purine Nucleoside Phosphorylase (PNP) Deficiency and Congenital Cytomegalovirus (CMV) Infection	Benjamin Rahoy	University of Nebraska Medical Center
81	T Cell Transcriptome in Chromosome 22q11.2 Deletion Syndrome	Nikita Rajé	University of Missouri Kansas City
82	Two Novel Mutations of Major Histocompatibility Class-II Associated Molecules	Lauren Rigg	University Hospitals Cleveland Medical Center / Case Western Reserve University
83	A Case Review of IgG4 Related Disease	Mario Rodenas	University of Florida
84	A Case Report of Focal Epithelial Hyperplasia (Heck's Disease) with Elevated Tumor Necrosis Factor Alpha	David Rosenthal	Donald and Barbara Zucker School of Medicine at Hofstra/Northwell, Departments of Medicine and Pediatrics, Division of Allergy/Immunology
85	A Rare Case of Helicobacter Bilis Chronic Complicated Osteomyelitis with Pyomyositis and Cellulitis in a Patient with XLA Agammaglobulinemia : Discussion of Challenges in Diagnosis and Management	Candace Rypien	Alberta Children's Hospital
86	Chronic Rhinosinusitis and Nasal Polyposis in Patients with IRAK-4 Deficiency	Sara Seghezzo	Department of Pediatrics, Division of Allergy, Immunology, and Bone Marrow Transplant, University of California, San Francisco
87	A Literature Review on Shared Decision-Making (SDM) to Inform the Development of an SDM Tool in Primary Immunodeficiency Diseases	Ihor Sehinovych	Shire
88	Sepsis as a Sign of Immunodeficiency	Katsiaryna Serhiyenko	Belarussian State Medical University
89	A Clinical Genomic Research Ecosystem Maximizes Data and Improves Patient Care	Morgan Simluk	Molecular Development of the Immune System Section, Laboratory of Immune System Biology, NIAID, National Institutes of Health
90	Clinical Features and Management of Patients with Rheumatoid Arthritis and a Coexisting Immunodeficiency Disorder	Namrata Singh	University of Iowa Hospitals and Clinics
91	Prevalence of Hypogammaglobulinemia in Newly Diagnosed Lymphoma	Namrata Singh	University of Iowa Hospitals and Clinics

POSTER SESSION II: SATURDAY, APRIL 6

POSTER	TITLE	PRESENTER	INSTITUTION
92	Safety and Pharmacokinetics of IGSC 20% in Subjects with Primary Immunodeficiency in an Open-label, Multicenter, Phase 3 Study	John Sleasman	Division of Allergy, Immunology, and Pulmonary Medicine, Duke University School of Medicine
93	Safety of Administration of Rotavirus Vaccine in Infants Born to Mothers Receiving Biologic Therapy During Pregnancy: A Retrospective Case Series	Christina Smith	Levine Children's Hospital, Atrium Health
94	Frequency of Specific Antibody Deficiency (SAD) and Respiratory Allergy in Patients with Recurrent Sinusitis	Charles Song	Ronald Reagan UCLA Medical Center, UCLA Mattel Children's Hospital
95	Combined Immune Deficiency in Association with a Single RAG1 Missense Variant in a 28-year-old Female	Charles Song	Ronald Reagan UCLA Medical Center, UCLA Mattel Children's Hospital
96	Comorbidities, Concomitant Medications, Infusion Parameters, and Tolerability in Advanced Age Patients with Primary Immunodeficiency Diseases Treated with Ig20Gly	Mark Stein	Allergy Section, Good Samaritan Medical Center
97	Expanding the Phenotypic Spectrum for STIM1-related Disorders: A Case Report	Anjali Sura	University of Michigan
98	Persistent Transaminitis in COPA Syndrome	Silpa Thaivalappil	UVA Pediatrics
99	Rapid Response of CVID Skin Granulomatous Disease to Infliximab	Maria Gabriela Torre	Unidad de Alergia, Asma e Inmunologia Clinica; British Hostital; Army Hospital
100	Infusion Parameters and Key Characteristics of Pediatric and Adolescent Patients with Primary Immunodeficiency Initiated on Ig20Gly in a Patient Program	Spiros Tzivelekis	Shire
101	Infusion Parameters of Patients with Primary Immunodeficiency by Previous Immunoglobulin Routes of Administration Among Enrollees in a Patient Program Initiated on Ig20Gly	Spiros Tzivelekis	Shire
102	Hepatic Complications of CVID	Kristine Vanijcharoenkarn	Emory University School of Medicine
103	The Clinical and Genetic Spectrum of RAG Deficiency Including a c.256_257delAA Founder Variant in Slavic Countries	Jolan Walter	Research Department, Belarusian Research Center for Pediatric Oncology, Hematology and Immunology
104	Heterozygous TACI Mutation (TNFRSF13B: A181E) Causing Significant Infections in a Patient with Normal Immunoglobulins	Shan Shan Wu	University Hospitals Cleveland Medical Center, Cleveland, Ohio
105	First Three Years' Experience in the Immunology Outpatient Clinic of a University Hospital in Cali, Colombia	Andres Zea-Vera	Universidad del Valle, Hospital Universitario del Valle
106	Diagnostic Challenges in Hospitalized Patients with Suspected Inborn Errors of Immunity in a Reference Center in the Southwest of Colombia	Andres Zea-Vera	Universidad del Valle, Hospital Universitario del Valle
107	Loss of Donor Chimerism 20 Years After Bone Marrow Transplant for Chronic Granulomatous Disease	Christa Zerbe	Laboratory of Clinical Immunology and Microbiology, NIAID, NIH
108	Body Temperature in Patients with Primary Immunodeficiency	Shouling Zhang	Department of Pediatrics, Donald and Barbara Zucker School of Medicine at Hofstra/Northwell, New Hyde Park, NY
109	A 2-year-old Male with Compound Heterozygous Familial Mediterranean Fever (FMF)	Shouling Zhang	Department of Pediatrics, Donald and Barbara Zucker School of Medicine at Hofstra/Northwell, New Hyde Park, NY

SPONSORS

THANK YOU TO OUR ANNUAL MEETING SUPPORTERS!

PREMIUM

CSL Behring

Takeda Pharmaceutical Company Limited

Grifols

PLATINUM

Enzyvant

Horizon Pharma

SILVER

Immune Deficiency Foundation

Leadiant Biosciences

USIDNET

BRONZE

Jeffrey Modell Foundation

NOTES

This image shows a single sheet of white paper with horizontal ruling lines. The lines are evenly spaced and run across the width of the page. There are no margins, text, or other markings on the paper.

[illegible]

2019 CIS School in Primary Immunodeficiency Diseases

October 24 - 27, 2019
Sonesta Fort Lauderdale Beach | Fort Lauderdale, Florida



CIS 2020 ANNUAL MEETING
IMMUNE DEFICIENCY AND DYSREGULATION NORTH AMERICAN CONFERENCE

DENVER

APRIL 2-5, 2020 HYATT REGENCY



CiS Clinical
Immunology
Society