FINAL PROGRAM

School on Diagnostic Assessment of Immune Phenotyping and Function in Primary Immunodeficiencies

APRIL 10, 2014 • BALTIMORE, MD

CIS Clinical Immunology Society

AMLI

About the School on Diagnostic Assessment of Immune Phenotyping and Function in Primary Immunodeficiences

The School on Diagnostic Assessment of Immune Phenotyping and Function in Primary Immunodeficiencies, co-sponsored by the Clinical Immunology Society (CIS) and the Association of Medical Laboratory Immunologists (AMLI), will cover the latest advances in diagnostic testing for primary immunodeficiencies, the interface between the laboratory and the clinician in diagnosis and management, and future trends in clinical diagnostic immunology.

About the Clinical Immunology Society

The Clinical Immunology Society (CIS), established in 1986, is the key inter-disciplinary organization for the field of clinical immunology and is devoted to fostering developments in the science and practice of clinical immunology. CIS is an international professional organization which includes more than 600 clinicians, investigators, and trainees.

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

The primary objectives and purposes of CIS are to:

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

About the Association of Medical Laboratory Immunologists

The Association of Medical Laboratory Immunologists (AMLI) was organized in 1987 to bring together professional and technical people engaged in the practice and study of medical laboratory immunology.

The organization consists of clinical scientists, physicians, dentists, veterinarians, medical technologists, and other professionals engaged in all areas of current laboratory testing.

The organization holds an annual meeting where information on all aspects of laboratory immunology are presented including new test development, technical and quality control aspects of current testing, the impact of regulatory issues, new developments in basic immunology and their impact on the practice of clinical immunology, clinical case studies, poster sessions, and round table discussions with experts in the field. The organization's mission is to improve the practice of clinical immunology laboratory testing by encouraging education and training of all individuals in the organization, maximizing the quality of test procedures, promoting the exchange of information, supporting clinical research efforts, and improving patient care.

CIS Clinical Immunology Society

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Program Committee

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Troy Torgerson, MD PhD
University of Washington

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Continuing Medical Education

Purpose

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

Target Audience

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

Learning Objectives

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

- Describe and interpret new defects in innate immunity and relevant diagnostic tools for the evaluation of patients;
- Interpret and apply molecular, specifically genetic, techniques in the diagnosis of primary immunodeficiencies;
- State, interpret and employ state-of-the-art tools for newborn screening for selected primary immunodeficiencies;
- Define, interpret and apply information on use of flow cytometric techniques for diagnosis and monitoring of patients with PIDs;
- Recall and explain the clinical and immunological phenotype of radiation sensitivity and DNA repair defects along with application of new methods for diagnosis.

ACCME Accreditation Statement

This activity has been planned and implemented in accordance with the Essential Areas and Policies of the Accreditation Council for Continuing Medical Education (ACCME) through the sponsorship of the Clinical Immunology Society (CIS). The CIS is accredited by the ACCME to provide continuing medical education for physicians.

Designation Statement

The Clinical Immunology Society designates this live activity for a maximum of 8 *AMA PRA Category 1 Credit(s)*™. Physicians should claim only the credit commensurate with the extent of their participation in the activity

Claiming CME

To claim CME credit and print out your CME certificate and/or certificate of attendance, please visit http://www.clinimmsoc.org/cme/pid2014/

Program Committee and Speaker Disclosures

Roshini Sarah Abraham, PhD
Mayo Clinic
There are no relationships to disclose.

Matthew C. Altman, MD
University of Washington
There are no relationships to disclose.

Geraldine Aubert, PhD
British Columbia Cancer Agency
Repeat Diagnostics Inc. company specializing in clinical telomere length measurements using Flow-FISH-employment receiving part time salary

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Alexandra & Steven Cohen Children's Medical Center
CSL-Behring PID Commitee-member advisory committee receiving honorarium
Baxter-speaking and teaching receiving honorarium

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NIH/NIAID
There are no relationships to disclose.

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There are no relationships to disclose.

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There are no relationships to disclose.

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Elsevier-Co-editor of the textbook Clinical Immunology: Principles and Practice receiving Royalty
Georgia Regents University College of Medicine-Teaching program receiving Honorarium

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There were no relationships reported at time of print of the final program.

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There are no relationships to disclose.

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There are no relationships to disclose.

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There are no relationships to disclose.

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There are no relationships to disclose.

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There are no relationships to disclose.

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There are no relationships to disclose.

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There were no relationships reported at time of print of the final program.

Rebecca A. Marsh, MD
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There are no relationships to disclose.

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There are no relationships to disclose.

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Harlan Robins, PhD
Fred Hutchinson Cancer Research Center
Adaptive Biotechnologies Corp.-Consultant receiving Consulting Fee and Ownership Interest

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There are no relationships to disclose.

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There are no relationships to disclose.

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There are no relationships to disclose.

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CSL Behring-Research fund recipient receiving Research funds

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Mirjam Van Der Burg, PhD
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There are no relationships to disclose.

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There are no relationships to disclose.

Keijan Zhang, MD MBA
Cincinnati Children's Hospital
There are no relationships to disclose.

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Preliminary Agenda

Each session will conclude with a panel discussion

7:00am – 7:45am	**Registration & Continetnal Breakfast**	*Key Foyer South*
7:45am – 7:50am	**Welcome** **Roshini S. Abraham, PhD** – *Mayo Clinic*	*Key 6 Ballroom*
7:50am – 9:40am	**Innate Immunity** 2 *AMA PRA Category 1 Credits™* *Moderators:* **Vijaya Knight, MD PhD** – *National Jewish Health* **Sergio D. Rosenzweig, MD PhD** – *NIAID, NIH*	
	Speakers:	
7:50am – 8:10am	Introduction to Concept in Innate Immunity **Steven Holland, MD** – *NIAID, NIH*	
8:10am – 8:30am	Clinical Laboratory Assessment and Interpretation of Innate Immune Defects **Adi V. Gundlapalli, MD PhD** – *University of Utah*	
8:30am – 8:45am	Role of Anti-Cytokine Antibodies in Innate Immune Defects **Sarah Browne, MD** – *NIAID, NIH*	
8:45am – 9:05am	Systems Immunology Approaches for Assessing Immunity **Damien Chaussabel, PhD** – *Benaroya Research Institute*	
9:05am – 9:15am	**Oral Abstracts:** DISTINCT ANTI-CYTOKINE AUTOANTIBODY PROFILE IS LIKELY DRIVEN BY INNATE HYPERINFLAMMATION AMONG PATIENTS WITH PRIMARY IMMUNODEFICIENCY SECONDARY TO RAG MUTATIONS **Jolan E. Walter, MD PhD** – *Massachusetts General Hospital, Harvard Medical School*	
9:15am – 9:25am	HIGH DIMENSIONAL SINGLE CELL MASS CYTOMETRY DEMONSTRATES CONSERVED HUMAN TOLL LIKE RECEPTOR ACTIVATION SIGNATURES **Elena (Wen-Yuan) Hsieh, MD** – *Stanford University*	
9:25am – 9:40am	**Panel Discussion**	
9:35am – 10:40am	**Dried Blood Spots: Newborn Screening and Beyond!** 1 *AMA PRA Category 1 Credits™* *Moderators:* **Roshini S. Abraham, PhD** – *Mayo Clinic* **Lisa Kobrynski, MD MPH** – *Emory University*	*Key 6 Ballroom*
	Speakers:	
9:40am – 9:55am	TREC for SCID and T Cell Lymphopenia **Antonia Kwan, PhD** – *University of California San Francisco*	
9:55am – 10:05am	Mass Spectrometry for DiGeorge Syndrome **Lisa Kobrynski, MD MPH** – *Emory University*	
10:05am – 10:15am	KREC for B Cell Immunodeficiencies **Vincent R. Bonagura, MD** – *Alexandra & Steven Cohen Children's Medical Center*	
10:15am – 10:25am	**Oral Abstract:** TRACKING THE DEVELOPMENT OF THE HUMAN FETAL T AND B CELL IMMUNE SYSTEM **Raz Somech, MD PhD** – *Safra Children's Hospital, Tel-Hashomer, Israel*	
10:25am – 10:40am	**Panel Discussion**	
10:40am – 11:00am	**Morning Break**	*Key Foyer South*

Preliminary Agenda

11:00am – 12:35pm	**Molecular Diagnostics for Primary Immunodeficiencies** 1.5 *AMA PRA Category 1 Credits™* *Moderators:* **Keijan Zhang, MD – Cincinnati Children's Hospital** **Amy Hsu** – *NIH* *Speakers:*	*Key 6 Ballroom*
11:00am – 11:20am	Promises and Pitfalls of NextGen Sequencing and Whole-exome/genome Technologies **João Bosco Oliveira, MD PhD** – *Instituto de Medicina Integral Prof. Fernando Figueira – IMIP*	
11:20am – 11:40am	Bioinformatics for the Non-Informatician **Eric Klee, PhD** – *Mayo Clinic*	
11:40am – 12:00pm	Immuno-Repertoire Analysis for T and B Cells **Harlan Robins, PhD** – *Fred Hutchinson Cancer Research Center*	
12:00pm – 12:10pm	**Oral Abstracts:** COHORT-WIDE AUTOMATED IDENTIFICATION OF MORBID ALLELES WITH THE HUMAN GENE CONNECTOME **Yuval Itan, PhD** – *The Rockefeller University*	
12:10pm – 12:20pm	INTERMEDIATE/HIGH AVIDITY OF ANTI-dsDNA AND ANTI-ssDNA AUTOANTIBODIES IN PATIENTS WITH XLT AND WAS **Elena Crestani, MD MS** – *Children's Hospital Boston*	
12:20pm – 12:35pm	**Panel Discussion**	
12:35pm – 1:30pm	**Lunch/Exhibit Hall/Poster Viewing**	*Key 8-12 Ballroom*
1:00pm – 1:30pm	**Guided Poster Sessions** **Group 1** *Moderator:* **Lisa Kobrynski, MD MPH** – *Emory University*	*Key 8-12 Ballroom*
	LIMITATION OF TREC-BASED NEWBORN SCREENING FOR ZAP70 DEFICIENCY **Serge Grazioli** – *BC Children's Hospital, UBC*	
	NGS ANALYSIS OF A 55 GENE PANEL FOR PIDs DIAGNOSIS **Yenhui Chang, MD PhD** – *All Children's Hospital*	
	ESTABLISHMENT OF NEWBORN SCREENING FOR SCID / SEVERE T LYMPHOCYTOPENIA IN SÃO PAULO **Marilia Kanegae, MSc PhD** – *University of São Paulo*	
	HUMAN SYNDROMES OF IMMUNE DEFICIENCY AND DYSREGULATION ARE CHARACTERIZED BY DISTINCT DEFECTS IN T-CELL RECEPTOR REPERTOIRE DEVELOPMENT **Xiaomin Yu, PhD** – *National Institutes of Allergy & Infectious Diseases (NIAID)*	
	Group 2 *Moderator:* **Sergio D. Rosenzweig, MD PhD** – *NIAID, NIH*	
	USING CYTOF AND SEQUENCING TO IDENTIFY A NOVEL PID CAUSING HYPEREOSINOPHILIA **Manish J. Butte, MD PhD** – *Stanford University*	
	NOVEL CLINICAL PRESENTATION OF COMPLETE MYELOPEROXIDASE (MPO) DEFICIENCY: SHOULD MPO DEFICIENCY BE RE-INSTATED AS A PRIMARY IMMUNODEFICIENCY? **Tamara C. Pozos, MD PhD** – *Children's Hospitals & Clinics of MN*	
	PERFORMANCE OF NEXT GENERATION SEQUENCING-BASED GENE PANELS FOR PRIMARY IMMUNODEFICIENCIES **Attila Kumanovics, MD** – *University of Utah*	
	BEYOND T, B, AND NK FLOW: AN EXTENDED ANALYTICAL PANEL FOR THE FLOW CYTOMETRIC EVALUATION OF PRIMARY IMMUNE DEFICIENCIES **Jennifer L. Stoddard** – *NIH*	

Preliminary Agenda

1:30pm – 3:35pm	**Flow Cytometry for the Diagnosis of Primary Immunodeficiences** 2 *AMA PRA Category 1 Credits™* *Moderators:* **Maurice (Mo) O'Gorman, PhD D(ABMLI) MBA MSc** – *Children's Hospital Los Angeles* **John Schmitz, PhD** – *University of North Carolina at Chapel Hill School of Medicine* *Speakers:*	*Key 6 Ballroom*
1:30pm – 1:50pm	B Cell Immunophenotyping in PIDs **Mirjam van der Burg, PhD** – *Erasmus MC, Netherlands*	
1:50pm – 2:10pm	Functional Flow Cytometry for the Diagnosis of HLH and Related Disorders **Rebecca Marsh, MD** – *Cincinnati Children's Hospital*	
2:10pm – 2:30pm	Flow Cytometry Screening for Signaling Pathway Defects **Thomas Fleisher, MD** – *NIH*	
2:30pm – 2:50pm	Clinical Utility and Laboratory Approach to Assessment of Telomere Defects **Geradine Aubert, PhD** – *British Columbia Cancer Agency*	
2:50pm – 3:10pm	Clinical Applications of Flow Cytometry: A Practical Approach **Troy Torgerson, MD PhD** – *University of Washington*	
3:10pm – 3:25pm	**Panel Discussion**	
3:35pm – 3:50pm	**Afternoon Break**	*Key Foyer South*
3:50pm – 5:00pm	**Radiation Sensitivity and DNA Repair Defects** 1.25 *AMA PRA Category 1 Credits™* *Moderators:* **Howard Lederman, MD PhD** – *Johns Hopkins* **Burcin Uygungil, MD MPH** – *Children's National Medical Center*	*Key 6 Ballroom*
3:50pm – 4:10pm	*Speakers:* Introduction and overview of Radiation Sensitivity and DNA Repair Disorders **Richard Gatti, MD** – *University of California, Los Angeles*	
4:10pm – 4:25pm	Alternate Approaches to Fibroblast Cultures for Radiation Sensitivity Testing **Kerstin Felgentreff, MD** – *Children's Hospital Boston*	
4:25pm – 4:45pm	Radiation Sensitivity and DNA Repair Defects: the Present and the Future **Mirjam van der Burg, PhD** – *Erasmus MC, Netherlands*	
4:45pm – 5:00pm	**Panel Discussion**	
5:00pm – 5:15pm	**Wrap Up**	

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CIS 2014 School in Primary Immunodeficiency Diseases

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Co-Chairs:

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Mount Sinai Medical Center
New York, NY

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

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Luigi D. Notarangelo, MD PhD
Children's Hospital Boston
Harvard Medical School
Boston, MA

Troy Torgerson, MD PhD
University of Washington
Seattle, WA

Applications need to be submitted via the CIS website, www.clinimmsoc.org. Applications must be supported by a:

- Letter from the Training Program Director or Department Chair;
- Curriculum Vitae;
- Statement of how the School will be beneficial;
- Abstract of the case or research project. CIS is looking for a case report including those which can be presented as a mystery for discussion purposes but should have a final diagnosis. Alternatively, cases with diagnostic dilemmas which could be discussed are acceptable. In addition, completed research projects germane to the field of primary immune deficiency are also acceptable.

Accepted abstracts will be provided with travel grants to assist with airfare, hotel, and meal expenses. The language of the School will be in English and selected participants are required to become CIS members at the conclusion of the School.

Target Audience:

- Fellows-in-training;
- Junior faculty members.

Participants should be pediatricians, internists, or scientists committed to the clinical management of patients with primary immunodeficiency diseases. Candidates may have backgrounds in immunology, infectious diseases, adult medicine, rheumatology, hematology, laboratory immunology, stem cell transplantation, or allergy/immunology.

Objectives:

- Education on the diagnosis, pathogenesis, and treatment of PIDD;
- Attract and develop future scientists in academic medicine;
- Enhance awareness of clinical immunology and its importance in scientific discoveries and clinical applications;
- Stimulate future collaborations between young investigators in different medical centers and countries, and between young investigators and experienced physician/scientists in the field.

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CIS-0114-705

Notes

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