Guided Poster Sessions

Thursday, April 10
12:35pm-1:00pm

**Guided Poster Session**

**Group 1**

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 Sao 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**

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
Friday, April 11

7:30am – 8:00am

Guided Poster Session

Group 1

Moderator:
Kathleen E. Sullivan, MD PhD
Children’s Hospital of Philadelphia

Guided Posters:
EARLY DETECTION OF JUVENILE MYELOMONOCYTIC LEUKEMIA (JMML) DUE TO ABSENT TRECS ON NEWBORN SCREENING (NBS) FOR SEVERE COMBINED IMMUNODEFICIENCY (SCID)
Richard L. Wasserman, MD PhD
Medical City Children’s Hospital

KABUKI SYNDROME IS ASSOCIATED WITH IMMUNE DYSFUNCTION FEATURING NOT ONLY IMMUNE DEFICIENCY BUT ALSO AUTOIMMUNITY AND LYMPHOPROLIFERATION
Mark C. Hannibal, MD, PhD
University of Michigan

IMPULSE OSCILLOMETRY (IOS) REVEALS INCREASED PULMONARY IMPEDENCE IN CHILDREN WITH ADENOSINE DEAMINASE-DEFICIENT SEVERE COMBINED IMMUNODEFICIENCY (ADA-SCID)
Robert Sokolic, MD
National Human Genome Research Institute, NIH

FOOD ALLERGY IN PRIMARY IMMUNODEFIENCY DISEASES: PREVALENCE WITHIN THE USIDNET REGISTRY
Karen Thursday S. Tuano, MD
Baylor College of Medicine

DISTURBED B AND T CELL HOMEOSTASIS AND NEOGENESIS IN PATIENTS WITH ATAXIA TELANGIECTASIA
Raz Somech, MD, PhD
Edmond and Lily Safra Children’s Hospital

Group 2

Moderator:
Ralph S. Shapiro, MD
Midwest Immunology Clinic

Guided Posters:
HYPER IGM SYNDROME AND EBV INDEPENDENT HEMATOLOGIC MALIGNANCY ASSOCIATED WITH GAIN OF FUNCTION MUTATIONS IN PIK3CD
Jennifer K. Grossman, MD FRCPC

HIGH THROUGHPUT SEQUENCING OF THE T CELL AND B CELL RECEPTOR REPertoires IN PATIENTS WITH WISKOTT ALDRICH SYNDROME
Amy E. O’Connell, MD, PhD
Boston Children’s Hospital
IL-17 T CELLS’ DEFECTIVE DIFFERENTIATION IN VITRO DESPITE NORMAL RANGE EX VIVO IN CHRONIC MUCOCUTANEOUS CANDIDIASIS DUE TO STAT1 GAIN OF FUNCTION MUTATION
Mohamed-Ridha Barhouche
Institut Pasteur De Tunis and University Tunish El Manar

COEXISTENCE OF CLONAL EXPANDED AUTOLOGOUS AND TRANSPLACENTAL-ACQUIRED MATERNAL T CELLS IN RAG1-DEFICIENT SCID
Atar Lev
Sheba Medical Center

Saturday, April 12
9:30am – 10:00am Guided Poster Sessions

Group 1
Moderator:
Kathleen E. Sullivan, MD PhD
Children's Hospital of Philadelphia

Guided Posters:
IPEX AND ‘IPEX-LIKE’ SYNDROME: FOXP3 AND RELATED PATHWAY
Eleonora Gambineri, MD
University of Florence

FATAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS (HLH) IN A 24 YEAR OLD FEMALE DUE TO PREVIOUSLY UNDIAGNOSED PRF1 P.T450M MUTATION IN THE C2 DOMAIN
Sara Barmettler, MD
Yale-New Haven Hospital

PROTEIN MODELING LED TO THE DISCOVERY THAT MUTATIONS IN THE GENE ENCODING PI(3)K CATALYTIC SUBUNIT p110d (PIK3CD) CAUSE PASLI (p110d ACTIVATING MUTATION CAUSING SENESCENT T CELLS, LYMPHADENOPATHY, AND IMMUNODEFICIENCY) DISEASE
Julie E. Niemela, MS, MLS
NIH

GAIN OF STAT1 SIGNALING ASSOCIATED WITH OVERLAPPING PATHOPHYSIOLOGY IN STAT3, IL21R AND STAT1 MUTANT PATIENTS
Diego B. Lopez

Group 2
Moderator:
Ralph S. Shapiro, MD
Midwest Immunology Clinic

Guided Posters:
ALTERED T CELL RECEPTOR DEVELOPMENT AND DIFFERENTIATION IN X-LINKED AGAMMAGLOBULINEMIA
Manish Ramesh, MBBS PhD
Icahn School of Medicine at Mount Sinai
STAT1 GAIN OF FUNCTION MUTATIONS ARE INFREQUENT IN PULMONARY NONTUBERCULOUS MYCOBACTERIAL INFECTION

**Eva P. Szymanski, BS**
National Institutes of Allergy & Infectious Diseases

INVESTIGATING THE ROLE OF INDUCIBLE T-CELL KINASE IN CD8+ T-LYMPHOCYTE CYTOLYTIC EFFECTOR FUNCTION

**Senta Kapnick**
NHGRI, NIH

SOMATIC REVERSION IN DOCK8 IMMUNODEFICIENCY MODULATES DISEASE PHENOTYPE

**Huie Jing, PhD**
NIAID, NIH