Southern California Rare Disease Genetics Meeting

Friday, March 8, 2019

8:30 am Breakfast

9:00 am Opening Remarks and Introduction
Virginia Kimonis, MD
Dan Cooper, MD

Moderator: Michael Zaragoza, MD

9:15 am Moving Treatment Forward for children with rare neuromuscular disease (1)
Jerry Mendell, MD - Nationwide Children's Hospital's Center for Gene Therapy

10:15 am Neural regeneration strategies to treat degenerative diseases of the retina (.25)
Henry Klassen – UCI, Department of Ophthalmology

10:30 am Characterization of copy number variants (CNVs) identified by genetic testing of inherited retinal disorders (.25)
Lucia Guidugli, PhD – Blueprint Genetics

10:45 am Coffee Break
Moderator: Moyra Smith, MD, PhD

11:00 am Lesch-Nyhan Disease (.75)
William Nyhan, MD, PhD - UCSD, Department of Pediatrics

11:45 am TK2-related mitochondrial depletion syndrome in a pair of siblings – a diagnostic odyssey leading to possible treatment (.25)
Joseph Shen, MD, PhD - UCSF Fresno, Division of Genetics, Department of Pediatrics

12:00 pm Lunch

12:30 pm Poster Session (Presenting author to be present at posters)
Moderator: John J. Gargus, MD

1:30 pm  The “transcriptomopathy” concept: Cornelia de Lange Syndrome and its relationship to other rare—and not so rare—disorders (.75)
Arthur Lander, MD, PhD - UCI, Developmental & Cell Biology

2:15 pm  The cohesin loader NIPBL interacts with TCOF1 and RNA to regulate pre-ribosomal RNA synthesis (.25)
Kyoko Yokomori, PhD – UCI, Department of Developmental & Cell Biology

2:30 pm  Epithelium mediated mechanisms for soft tissue syndactyly in Van der Woude syndrome (.25)
Ghaidaa Kashgari, 6th Year PhD Candidate – UCI, Department of Biological Chemistry, School of Medicine

2:45 pm  Coffee Break

Moderator: Meredith Cagle – Global Genes

3:15 pm  The Rare and Precious: How Rare Diseases Are Revolutionizing Medicine (.75)
Fowzan Alkuraya, MD - King Faisal Specialist Hospital, Department of Genetics

4:00 pm  Patient Voices: Living with a Rare Disease. Challenges in treating and caring for novel rare diseases (1)
Meredith Cagle – Global Genes

Paul Harmatz, MD

Nathan Peck – Patient and advocate diagnosed with Inclusion Body Myopathy with early-onset Paget Disease of the bone and Frontotemporal Dementia (IBMPFD)

Cristy Spooner – Mother and advocate for patient with an NUBPL mutation

Mark E. Nunes - Division Chief Medical Genetics, Kaiser Permanente, San Diego, CA

Yogesh Bansal – Father of patient with DNM1 Mutation

5:00 pm  Closing Remarks – Virginia Kimonis, MD

6 pm  Happy Hour – Location TBA