**APBDRF**

**12th Annual**

**Scientific Advisory Board Meeting**

Monday-Tuesday

**December 5-6, 2016**



Town and Village Synagogue

334 East 14th St., NYC 10003

Gregory Weiss, President

APBD Research Foundation  
2710 Avenue S

Brooklyn, NY 11229

(646) 580-5610

Contact the APBDRF at  **[info@APBDRF.org](mailto:info@APBDRF.org)**

This meeting is dedicated to the memory of LD Coddon.

His commitment and contributions to our mission will not be forgotten.

**8:30 AM Monday Morning: GBE Stabilization**

**Moderator: Dr. Salvatore DiMauro,** Columbia University, New York, New York

Presenters 1 & 2**:** **Dr. Or Kakhlon**, Hadassah-Hebrew University Medical Center, Jerusalem, Israeland

**Dr. Wyatt Yue**, Structural Genomics Consortium, University of Oxford, UK

*“Using recombinant glycogen branching enzyme and structural biology techniques to understand therapeutic opportunities for APBD”* ***1***

Presenter 3: **Dr. Pablo Escribá**, University of the Balearic Islands, Palma, Spain

*“TGMs on the interaction of WT and Y329S GBE1 with membranes: Effects on GBE1 activity and APBD pathophysiology and therapy*” **2**

Monday Late Morning: Genetic Interventions

**Moderator: Dr. Edwin Kolodny,** New York University School of Medicine, NY

Presenter 1**: Erin Chown**, The Hospital for Sick Children, Toronto, Canada

*“Identification of therapeutic targets in an APBD mouse model”* ***3***

Presenter 2: **Dr. Tamar Grossman**, Ionis Pharmaceuticals, Carlsbad, California

*“Antisense Oligonucleotide Therapy for Genetic Disorders”* ***4***

Presenter 3: **Dr. Berge Minassian**, The Hospital for Sick Children, Toronto, Canada

*“Antisense oligonucleotide therapy for APBD”* ***5***

Presenter 4: **Dr. H. Orhan Akman**, Columbia University Medical Center, New York, New York

*“Research Update: Generation of the mouse model of GBE1 intronic insertion/deletion and approaches to treatment”* ***6***

**LUNCH**

Presenter : **Dr. Nicole Schreiber-Agus**, Montefiore Medical Center, Albert Einstein College of Medicine, Bronx, New York

*“A feel-good moment in the realm of APBD awareness”* ***7***

**1PM Monday Early Afternoon**

**Moderator: Dr. Edwin Kolodny**

**Special Presentation by Dr. Priya Kishnani**

Duke University School of Medicine, Durham, North Carolina

*“Summary of Duke Pediatric Medical Genetics Glycogen Storage Disease Type IV (GSD IV) Research Program (2016)”* ***8***

**Monday Mid-Afternoon: An Advanced Approach to Drug Discovery – Triheptanoin Case Study; Clinical Endpoints for Future Clinical Trials**

**Moderator: Dr. Heather A. Lau**, NYU Langone Medical Center, NY

Presenter 1: **Dr. Raphael Schiffmann**, Baylor Research Institute, Dallas, Texas

*“A double-blind placebo-controlled trial of triheptanoin in adult polyglucosan body disease (APBD)”* ***9***

Presenters 2 & 3: **Dr. Or Kakhlon** and **Dr. Alex Lossos**, Hadassah-Hebrew University of Jerusalem, Israel

Introduction of proposed guaiacol trial, followed by a Panel Discussion of clinical endpoints for future clinical trials.**10**

**Panel Discussion: *Clinical Endpoints for Future Clinical Trials***

Dr. Salvatore DiMauro, Columbia University, New York, New York

Dr. Edwin Kolodny, New York University School of Medicine, New York, New York

Dr. Raphael Schiffmann, Baylor Research Institute, Dallas, Texas

Via internet: Dr. Alex Lossos Hadassah-Hebrew University of Jerusalem, Israel

Via internet: Dr. Yoseph Caraco, Hadassah-Hebrew University of Jerusalem, Israel

Dr. Christopher Klein, Mayo Clinic\*

Dr. Brent Fogel, UCLA \*

Dr. Michio Hirano, Columbia University\*

Dr. Marvin Natowicz, Cleveland Clinic, Cleveland, Ohio\*

*\* invited internet panelist*

Presenter 3: **Dr. Or Kakhlon**

*“Combining High Throughput Screening with Image-Based Phenotyping to advance APBD drug discovery”* ***11***

***5:00 – 6:30 PM*** *Cocktail Social Hour and special screening:*

***LIFE THROUGH A LENS****, the award-winning documentary of the life and philosophy of APBD patient Robert Zuckerman*

**9:00 AM Tuesday Morning: Repurposed Drug Initiatives**

**Introductory Speaker and Moderator: Dr. Bruce Bloom,** Cures Within Reach, Skokie, Illinois

*“RarePurposing – The Cures Within Reach Approach”* ***12***

Presenter 2: **Dr. Or Kakhlon**

*“Guaiacol – a novel drug candidate for treating Adult Polyglucosan Body Disease”* ***13***

Presenters 3 & 4: **Dr. Or Kakhlon and Dr. Wyatt Yue**

*“Using recombinant glycogen synthase and structural biology techniques to understand therapeutic opportunities for APBD”* ***14***

Presenter 5**: Dr. H. Orhan Akman**

*“Pharmacological treatment of APBD: Potential for treatment by Ibudilast*® *and/or guaifenesin”* ***15***

Presenter 6: **Dr. Or Kakhlon**

*“Testing pharmacophore-based small molecules for stabilizing GBE-Y329S”* ***16***

**LUNCH**

**Tuesday Afternoon: Essential Groundwork - Patient Registry, Genetic Testing, Natural History Studies, Biobanks**

**Introductory Speaker and Moderator: Lori Ann Correia,** MS,CGC - Founder and President - Allele Consulting, LLC, Boston, MA **17**

**Patient Registry**

Presenter 1: **Dr. Raphael Schiffmann**

*"CAP,****C****olumbia University****A****PBD****P****atient  registry: enrollment update and revised questionnaire"* ***18***

**Genetic Testing**

Presenter 2**: Dr. Ruth Kornreich**, Mt. Sinai School of Medicine, New York, NY

*“Experience with carrier screening for GBE1 mutations associated with Adult Polyglucosan Body Disease/ Glycogen Storage Disease Type IV in the Ashkenazi Jewish and general populations”* ***19***

Presenter 3: **Alana Cecchi**, Counsyl Inc., San Francisco, California

*“Advances in carrier screening: Making the case for APBD”* ***20***

**Natural History Studies**

Presenter 4: **Suzanne Rossov**, National Organization for Rare Disorders (NORD), Danbury, Connecticut

*“The Importance of Natural History Studies for Rare Diseases”* ***21***

Presenter 5**: Dr. Harrison Jones**, Duke Univ. School of Medicine, Durham, NC

*"FDA Orphan Products Natural History Grants Program: An Opportunity for Adult Polyglucosan Body Disease?"* ***22***

Presenter 6: **Dr. Or Kakhlon**

*“A biobank of APBD patient skin fibroblasts”* ***23***

**3 PM End of Conference: Closing and Thanks**

**Harriet Saxe**, APBDRF Board member