Rare Disease Day Symposium 2020
SBP Sanford Burnham Prebys MEDICAL DISCOVERY INSTITUTE

CDG CARE

DAY 1 – FRIDAY, FEBRUARY 28, 2020; SBP SCIENTIFIC MEETING & POSTER SESSION

OPENING SESSION		
Malin Burnham, Sanford Burnham Prebys Medical Discovery Institute (SBP)	Introduction and Welcome	8:30
T. Denny Sanford, Philanthropist and Honorary Trustee, SBP	My Investment in Children	
Debra Turner, Philanthropist and Honorary Trustee, SBP	TBD	
Randy Schekman, PhD UC Berkeley, Nobel Laureate	**Keynote Address:** Sorting and Trafficking of Protein and RNA Mediated by Extracellular Vesicles	
CLASSICAL CDG		
Heather Flanagan-Steet, PhD Greenwood Genetic Center	A PMM2-CDG Zebrafish Model Provides Novel Mechanism Insight Into Disease Pathogenesis	
Peter McWilliams, PhD, Glycomine, Inc.	Mannose-1-P Replacement Therapy – Toward Clinical Trials	
BREAK		
Eva Morava, MD, PhD, Mayo Clinic	The Growing CDG Network	
Mercedes Serrano, MD, University of Barcelona	Clinical Trials in CDG: The Long and Winding Road	
LUNCH		
William Brucker, MD, Harvard Medical School	The Role of Serum Proteins in the Treatment of Acute Illness & Shock in CDGs	
Lynne Wolfe, RN, NHGRI	CDG and the Undiagnosed Disease Program at NIH	
GPI DISORDERS		
Taroh Kinoshita, PhD, Osaka University	Basic Science and Potential Treatment of GPI Disorders	
Clement Chow, PhD, University of Utah	Precision Medicine Approaches in Drosophila to Understanding PIGA Deficiency	
Marjan Huizing, PhD, NIH/NHGRI	Basic Science and the Clinical Results of ManNAc Therapy in Gene Myopathy	
Yin Dong, PhD, Oxford University	Insights into the Gate Keeper of N-glycosylation	
BREAK		
DYSTROGLYCANOPATHIES		
Qi Lu, MD, PhD, Atrium Health	Ribitol Therapy for FKRP Muscular Dystrophy	
Jeffrey Chamberlain, PhD, University of Washington	Gene Therapy for FKRP	
Marjan Huizing, PhD, NHGRI	Basic Science and Clinical Results in ManNAc Therapy in GNE Myopathy	
Yin Dong, PhD, Oxford University	PAGT1 in CDG and Myasthenic Syndromes	
POSTER SESSION		
Poster Session: An interactive poster session/cocktail hour on Friday evening will allow junior investigators and postdocs to show emerging science alongside patient and advocate presenters.		Closing 7:00

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DAY 2 – SATURDAY, FEBRUARY 29, 2020; SPB SCIENTIFIC MEETING & DOCTOR-IS-IN-SESSION

CDG-CENTERED GLYCOBIOLOGY		
Ethan Perlstein, PhD, Christopher and Dana Reeve Foundation	Identification of PMM2 Small Molecule Therapy	8:30 am
Richard Cummings, PhD, Harvard University	National Center for Functional Glycomics: A Home for CDG	
Francois Foulquier, PhD, University of Lille	Biology of Mn^{2+} Transport in CDGs	
BREAK		
Kent Lai, PhD, University of Utah	Mouse Model of PGM1-CDG	
Vladimir Lupashin, PhD, University of Arkansas for Medical Sciences	Biology of the COG Complex	
Hudson Freeze, PhD, Sanford Burnham Prebys Medical Discovery Institute	CDG Models and Therapy	
TENTATIVE: DISCUSSION AND WRAP UP OF IMPORTANT FINDINGS		
LUNCH		
DOCTOR-IS-IN SESSION		
Hudson Freeze, **PhD**, Sanford Burnham Prebys Medical Discovery Institute	Welcome and Introductions	
Symposium Speakers and Invited Guests (TBD)	**Doctor-is-in Sessions**: Now in its 3rd year, the SBP Rare Disease Day Symposium's Doctor-Is-In-Session brings together medical researchers, clinicians, advocates, patients and their families for an afternoon of hands-on collaboration. In this innovative forum, physicians and scientists visit like-minded small family groups in a round-robin format to facilitate bi-directional conversations. **Pre-registration is required**.	
Hudson Freeze, **PhD**, Sanford Burnham Prebys Medical Discovery Institute	**SBP Symposium Closing**	4:00 pm
FAMILY-FRIENDLY ACTIVITY		
Family-Friendly Activity: A fun, family-friendly evening activity for families and scientists to provide a transition to Sunday's meeting. Details will be announced once available.		

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DAY 3 – SUNDAY, MARCH 1, 2020; CDG CARE FAMILY MEETING

FAMILY MEETING

The family meeting combines patient-centered science and real-world strategies for patients and families living with a Congenital Disorder of Glycosylation/De-Glycosylation. The goals of the 2020 meeting are to:

1. Develop an inclusive, collaborative community infrastructure to discuss common issues of patients living with CDG,
2. Promote the exchange of education, resources and therapeutic approaches to maximize the overall quality of life of CDG patients,
3. Introduce alternative, cutting-edge treatment and therapies to improve health outcomes for children, youth and adults affected by CDG.

AGENDA

Andrea Miller, **JD**, **MHA**, President, CDG CARE	**WELCOME**	8:15 am
Bradley Miller, MD, PhD, University of Minn. Masonic Children's Hospital	Hormonal Fluctuations, Puberty, and Bone Density in Adolescents Diagnosed with CDG	
Canice Crerand, PhD, Nationwide Children's Hospital	Navigating Adolescence with CDG: Behavioral Health and Transition Needs	
Christina Lam, MD, Seattle Children's Hospital	CDG Genetics and Introduction to Gene Therapy	
BREAK		
Melissa Merideth, MD, MPH, Obstetrician & Geneticist, NIH/NHGRI	Reproductive Health Issues in Females with CDG	
Hudson Freeze, PhD, Sanford Burnham Prebys Medical Discovery Institute	CDG Updates and Perspectives	
Andrew Edmondson, MD, PhD, Children's Hospital of Philadelphia	Models for CDG Research	
Eva Morava, MD, PhD, Mayo Clinic	CDG Studies, Treatments, and Future Therapies	
LUNCH		
Lynne Wolfe, CRNP, NIH/NHGRI	Updates from the NIH CDG Natural History Study	
Ivan Martinez Duncker, **MD, ScD**, Morelos State Autonomous University	Summary of CDG Cases & Physician/Research Network in Latin America	
Karyn Searcy, MA, CCC, San Diego State University	Beyond the Diagnostic Label	
BREAK		
Becki Cohill, OTD, OTR, University of St. Augustine	Caring for the Caregiver	
Philip P. Lindsley, Principal & Attorney, San Diego Special Needs Law Center	Special Needs Family Planning	
Margaret Perkins, MA, CCC-SLP, ATP, SoCal AAC Therapy	AAC Follow the Map	
Andrea Miller, **JD**, **MHA**, President, CDG CARE	**CLOSING**	4:30 pm