Rare Disease Day Symposium 2018
SBP Sanford Burnham Prebys MEDICAL DISCOVERY INSTITUTE

NGLY1.org CDG CARE

DAY 1 – FRIDAY, FEBRUARY 23, 2018; SBP SCIENTIFIC MEETING & POSTER SESSION

OPENING SESSION		
Perry Nisen, **MD**, **PhD**, CEO, Sanford Burnham Prebys Medical Discovery Institute (SBP)	Introduction and Welcome	9:00–9:10
T. Denny Sanford, Philanthropist and Honorary Trustee, SBP	My Investment in Children	9:10–9:15
Leroy Hood, **MD**, **PhD**, President and Co-Founder, Institute for Systems Biology	**Keynote address**: Scientific Wellness and Transforming the Healthcare System	9:15–10:00
PMM2-CDG: OVERVIEW, MODELS AND THERAPIES		
Lynne Wolfe, **MS**, **CNRP**, NIH Undiagnosed Diseases Program (UDP)	Natural History of PMM2-CDG	10:00–10:25
Duncan Webster, **MD**, Parent/Advocate/Investigator	Our CDG Journey	10:25–10:50
BREAK		10:50–11:00
Kendal S. Broadie, **PhD**, Vanderbilt University School of Medicine	Fly Model of PMM2	11:00–11:25
Stephen Dalton, **PhD**, University of Georgia	iPS Models of PMM2	11:25–11:50
Agnes Rafalko, **PhD**, CEO and President, Glycomine, Inc.	Therapy for PMM2-CDG	11:50–12:15
LUNCH		12:15–1:15
Josef Penninger, **MD**, **PhD**, Institute of Molecular Biotechnology Austrian Academy of Sciences	Repairable mutagenesis in haploid stem cells for reproducible research	1:15–1:40
Eva Morava, **MD**, **PhD**, Tulane University Medical Center, USA and University of Leuven, Belgium	Overview of current monosaccharide therapy trial in congenital disorders of glycosylation	1:40–2:05
OTHER GLYCOSYLATION DISORDERS: NOVEL APPROACHES		
Robert Haltiwanger, **PhD**, The University of Georgia Complex Carbohydrate Research Center	Peters Plus Syndrome: A Congenital Disorder of Glycosylation	2:05–2:30
Kevin P. Campbell, **PhD**, Howard Hughes Medical Institute, University of Iowa, Carver College of Medicine	Mechanistic Insights and Therapeutic Approaches for O-Glycosylation-Deficient Muscular Dystrophy	2:30–2:55
BREAK		2:55–3:10
Jan Carette, **PhD**, Stanford University	Genetic knockout screens reveal a critical for glycosylation in infectious disease	3:10–3:35
Hudson Freeze, **PhD**, Sanford Burnham Prebys Medical Discovery Institute (SBP)	Novel glycosylation disorders	3:35–4:00
PANEL SESSION (Participants TBD)		4:00–5:00
POSTER SESSION		
Poster Session: An interactive poster session on Friday evening will allow junior investigators and postdocs to show emerging science alongside patient and advocate presenters.		5:00–7:00

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DAY 2 – SATURDAY, FEBRUARY 24, 2018; SBP SCIENTIFIC MEETING & DOCTOR-IS-IN SESSION

NGLY1: A DISORDER OF DEGLYCOSYLATION		
Matthew Might, **PhD**, University of Alabama at Birmingham	Precision medicine and NGLY1	9:00–9:25
Gary B. Ruvkun, **PhD**, Harvard Medical School	Genetic analysis of NGLY1 action in proteasome biology	9:25–9:50
Tadashi Suzuki, **DSci**, RIKEN Global Research Cluster	Can basic science contribute to curing human genetic disorders?	9:50–10:15
Clement Chow, **PhD**, University of Utah	Genetic and environmental modulation of NGLY1 deficiency	10:15–10:40
Hamed Jafar-Nejad, **MD**, Baylor College of Medicine	Role of NGLY1 in *Drosophila* Development	10:40–11:05
BREAK		11:05–11:15
Platform Presentations from Posters (5)		11:15–12:00
LUNCH		12:00–1:00
DOCTOR-IS-IN SESSION		
Hudson Freeze, **PhD**, Sanford Burnham Prebys Medical Discovery Institute (SBP)	Welcome and Introductions	1:00–1:15
Symposium Speakers and Invited Guests (TBD)	**Doctor-is-in Sessions**: Now in its ninth 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**.	1:15–3:45
Hudson Freeze, **PhD**, Sanford Burnham Prebys Medical Discovery Institute (SBP)	**SBP Symposium Closing**	3:45–4:00
NETWORKING TIME		
Family/Scientist Networking Time (Details TBD)		4:00–7:00

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DAY 3 – SUNDAY, FEBRUARY 25, 2018; CDG FAMILY MEETING

AGENDA		
Andrea Berarducci, **JD**, **MHA**, President, CDG CARE	**WELCOME**	8:15–8:30
TBN	*CDG and Neurological Overview*	8:30–9:00
TBN	*Clinical Focus – Gastroenterology, Hormones & Nutrition*	9:00–9:30
Allyssa Harpst, **MEd**, CDG Parent, Teacher for Students with Visual Impairments, Urbana School District #116	Developing Your Child's IEP: Your Role in the Process	9:30–10:00
BREAK		10:00–10:30
Eva Morava, **MD**, **PhD**, Tulane University Medical Center, USA and University of Leuven, Belgium	Personalized Medicine in CDG	10:30–11:00
David Cassiman, **MD**, **PhD**, KU Leuven, Belgium	What is the liver? What is liver disease? And what to expect in CDG?	11:00–11:30
Christina Lam, **MD**, University of Washington	CDG & Genetics Focus / Seattle Children's Hospital	11:30–12:00
LUNCH		12:00–1:00
Hudson Freeze, **PhD**, Sanford Burnham Prebys Medical Discovery Institute (SBP)	CDG Updates and Perspectives	1:00–1:30
Becki Cohill, **OTD**, **OTR**, University of St. Augustine	Sensory Processing and "Behavior"	1:30–2:00
Carrie Ostrea, NGLY1.org	Becoming an Advocate for the CDG Community	2:00–2:30
BREAK		2:30–3:00
Lynette LaScala, Founder and CEO, NAPA Center	Cutting Edge Treatments and Therapies (TBD)	3:00–3:30
TBN	*Medical Equipment Overview*	3:30–4:00
TBN	*Caring for the Aging CDG Child, Continuing Support into Adulthood*	4:00–4:30
Andrea Berarducci, **JD**, **MHA**, President, CDG CARE	**CLOSING**	4:30–5:00