

Sanford • Burnham • Prebys MEDICAL DISCOVERY INSTITUTE

7th Annual Rare Disease Day Symposium: Human Glycosylation Disorders

February 26-27, 2016

Sanford Burnham Prebys Medical Discovery Institute, La Jolla, CA USA

Preliminary Program

Please note that speakers and presentation titles are subject to change.

Day 1 - February 26, 2016		
Perry Nisen, M.D., Ph.D. and T. Denny Sanford	8:30-8:45	Opening Remarks
Lawrence Tabak, D.D.S, Ph.D.	8:45-9:30	Perspectives from the National Institutes of Health
Leroy Hood, M.D., Ph.D.	9:30-10:15	Systems Biology and Proactive P4 Medicine: Transforming Healthcare with Wellness
William Gahl, M.D., Ph.D.	10:15-11:00	Investigating Rare Diseases Through the NIH Undiagnosed Diseases Program and Network
Break	11:00-11:10	
Shengfang Jin, Ph.D. (Tentative)	11:10-11:35	Therapy for CDG
Richard Steet, Ph.D.	11:35-12:00	New Insight into CDG Pathogenesis from Zebrafish and Stem Cell Models
James Reid Gilmore, Ph.D.	12:00-12:25	Oligosaccharyltransferase Mutations: Role in CDG-1
Hudson Freeze, Ph.D.	12:25-12:50	Emerging CDG Concepts
Lunch	12:50-2:00	
Erik A. Eklund, M.D., Ph.D.	2:00-2:25	Severe skeletal phenotypes in CDG
Raymond Wang, M.D.	2:25-2:50	Mitochondrial GFM1 Deficiency Presenting as a Congenital Disorder of Glycosylation
David Beeson, Ph.D.	2:50-3:15	Myasthenic Syndromes and CDGs
Break	3:15-3:30	
Lance Wells, Ph.D.	3:30-3:55	Dystroglycanopathies Today
Taroh Kinoshita, Ph.D.	3:55-4:20	Molecular Basis of Congenital Deficiencies of GPI Anchors
Day 2 - February 27, 2016		
Matt Might, Ph.D.	9:00-9:15	<i>NGLY1</i> Patient Advocacy
Matt Wilsey, M.B.A	9:15-9:30	2,978 Days & the Race to Cure <i>NGLY1</i>
Lynne Wolfe, M.S., C.N.R.P.	9:30-9:55	NIH CDG Natural History Study
Tadashi Suzuki, D.Sci.	9:55-10:20	ENGase as a Potential Drug Target for <i>NGLY1</i> -deficiency
Hamed Jafar-Nejad, M.D.	10:20-10:45	Using <i>Drosophila</i> to Understand the biology of <i>NGLY1</i>
Break	10:45-11:00	
Nicholas J. Schork, Ph.D.	11:00-11:25	Individualizing Medical Research: The N-of-1 Clinical Trial
Henry I. Miller, M.S., M.D.	11:25-11:50	Let's Not Let Regulations Or Moratoriums Delay New Therapies
Noah Coughlan	11:50-12:10	Run for Rare
Wrap Up	12:10-12:20	Closing Remarks
Lunch	12:20-1:00	
Doctor Is In Session	1:00-4:00	Interactive event for CDG and <i>NGLY1</i> patients and their families. Pre-registration required by November 2, 2015.
Reception	4:00	