Rare Disease Day Symposium 2018

SBP Sanford Burnham Prebys MEDICAL DISCOVERY INSTITUTE




Join us for the 9th Annual SBP Rare Disease Day Symposium and Bi-Annual CDG Family Conference

Congenital Disorders of Glycosylation and De-glycosylation

February 23-25, 2018
The Dana on Mission Bay, San Diego, CA

Keynote Speaker

Scientific Wellness and Transforming the Healthcare System

Leroy Hood, MD, PhD
Senior Vice President/Chief Science Officer
Providence St. Joseph Health
President, Institute for Systems Biology

Presented in Partnership with:

CDG CARE

NGLY1.org

PMM2-CDG: OVERVIEW, MODELS AND THERAPIES

Lynne Wolfe, **MS**, **CNRP**, NIH Undiagnosed Diseases Program
Natural History of PMM2-CDG

Duncan Webster, **MD**, Parent/Advocate/Investigator
Our CDG Journey

Kendal S. Broadie, **PhD**, Vanderbilt University School of Medicine
Fly Model of PMM2

Stephen Dalton, **PhD**, University of Georgia
iPS Models of PMM2

Agnes Rafalko, **PhD**, CEO and President, Glycomine, Inc.
Therapy for PMM2-CDG

OTHER GLYCOSYLATION DISORDERS: NOVEL APPROACHES

Josef Penninger, **MD**, **PhD**, Institute of Molecular Biotechnology Austrian Academy of Sciences
Repairable mutagenesis in haploid stem cells for reproducible research

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

Robert Haltiwanger, **PhD**, The University of Georgia
Peters Plus Syndrome: A Congenital Disorder of Glycosylation

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

Jan Carette, **PhD**, Stanford University
Genetic knockout screens reveal a critical for glycosylation in infectious disease

Hudson Freeze, **PhD**, Sanford Burnham Prebys Medical Discovery Institute
Novel glycosylation disorders

NGLY1: A DISORDER OF DE-GLYCOSYLATION

Matthew Might, **PhD**, University of Alabama at Birmingham
Precision medicine and NGLY1

Gary B. Ruvkun, **PhD**, Harvard Medical School
Genetic analysis of NGLY1 action in proteasome biology

Tadashi Suzuki, **DSci**, RIKEN Global Research Cluster
Can basic science contribute to curing human genetic disorders?

Clement Chow, **PhD**, University of Utah
Genetic and environmental modulation of NGLY1 deficiency

Hamed Jafar-Nejad, **MD**, Baylor College of Medicine
Role of NGLY1 in Drosophila Development

CDG FAMILY CONFERENCE

Marc Patterson, **MD**, Mayo Clinic
CDG Neurologic Overview and Updates

Carolyn Brown, **MSc**, **CGC**, **CCGC**, Illumina Clinical Services Laboratory
Genetic Counseling for CDG

Allyssa Harpst, **MEd**, CDG Parent, Teacher for Students with Visual Impairments
Developing Your Child's IEP: Your Role in the Process

Eva Morava, **MD**, **PhD**, Tulane University Medical Center, USA and University of Leuven, Belgium
Personalized Medicine in CDG

David Cassiman, **MD**, **PhD**, KU Leuven, Belgium
What is the liver? What is liver disease? And what to expect in CDG?

Christina Lam, **MD**, University of Washington, Seattle Children's Hospital
CDG & Genetics Focus / Seattle Children's Hospital

Becki Cohill, **OTD**, **OTR**, University of St. Augustine
Sensory Processing and "Behavior"

Carrie Ostrea, NGLY1.org
Becoming an Advocate for the CDG Community

Lynette LaScala, Founder and CEO, NAPA Center
Cutting Edge Treatments and Therapies

LaRae Mercer-Wise, CDG Parent
Caring for CDG Children into Adulthood, Planning for Transition and Continued Support Services

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Registration Closes January 22, 2018