CDG CARE

NGLY1.org

2018 SBP Rare Disease Day Symposium & CDG/NGLY1 Family Conference

February 23-25, 2018
San Diego, California

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

Retrophin®

Dear Conference Participant,

On behalf of the Conference Organizing Committee, we warmly welcome you to the 2018 SBP Rare Disease Day Symposium & CDG Family Conference. This year's event is co-organized by Sanford Burnham Prebys (SBP) Medical Discovery Institute, CDG CARE and NGLY1.org. We are excited to present a three-day program representing a diverse panel of clinical, investigational, translational (therapeutic), and community experts from around the World.

The objective of this year's event is to facilitate the meeting and collaboration of medical professionals and families in order to share knowledge and experience and discuss common issues of patients living with CDG. Through this unique organizational partnership, an inclusive and collaborative community infrastructure will be developed to enable medical professionals and families to unite and enhance CDG research, knowledge, therapeutic, and alternative treatment strategies.

The Conference Committee wishes to thank and acknowledge all of our talented speakers who kindly accepted our invitation to participate in this unique and educational program. Without their willingness to share their time and expertise, this conference would not have been possible.

The Conference is organized and run by members of the founding Institutes and volunteers, and is funded by donations, grants, corporate and industry sponsorships, and a nominal participant registration fee. We are especially grateful to Presenting Sponsor Retrophin, whose generous donation made this event possible. We are also proud to be an honored recipient of the 2018 Global Genes RARE Patient Impact Grant Program.

We are indebted to and sincerely thank all of our conference donors, volunteers and contributors for making the organization of this significant event possible and look forward to continued collaboration in the future.

Together, we can make a difference in the fight against Congenital Disorders of Glycosylation (CDG). On behalf of our community, thank you for joining us.

Sincerely,

Hudson Freeze, PhD
2018 Symposium Chair
Sanford Burnham Prebys Medical Discovery Institute (SBP)

Carrie Ostrea
Executive Director
NGLY1.org

Andrea Berarducci, JD
President
CDG CARE

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2018 SBP Rare Disease Day Symposium
& CDG Family Conference
February 23 – 25, 2018

We proudly welcome you to the 2018 SBP Rare Disease Day Symposium & CDG Family Conference. This year's event is co-organized by Sanford Burnham Prebys (SBP) Medical Discovery Institute, CDG CARE and NGLY1.org.

SBP's scientific symposium will be held **Friday, February 23** and **Saturday, February 24, 2018**. We will kick off with keynote presentation "21st Century Medicine Will Transform Healthcare" from scientific luminary **Leroy Hood, MD, PhD**, the President and Co-Founder of the Institute for Systems Biology. In total, 22 noted CDG and NGLY1-focused scientists and clinicians will speak on the following topics: *NGLY1: A Disorder of Glycosylation*, *PMM2-CDG: Overview, Models and Therapies*, and *Other Glycosylation Disorders: Models and Therapies*. We are also excited to offer the symposium's first **Poster Session** the evening of **Friday, February 23**—a new opportunity to present emerging science in glycobiology.

The three-day event will conclude on **Sunday, February 25** with the CDG CARE/NGLY1.org CDG Family Conference. Family topics include education on the science behind CDG, resources available for patients and families, and interventional and therapeutic approaches. The family meeting is more practical in approach about daily living but maintains a significant scientific element.

For questions or more information, please contact:
* SBP – Amy Zimmon, azimmon@sbpdiscovery.org
* CDG CARE – Andrea Berarducci, conference@cdgcare.com
* NGLY1.org – Carrie Ostrea, carrie@ngly1.org

Doctor-is-in Session
Saturday, February 24, 2018, 1:00 pm – 4:00 pm

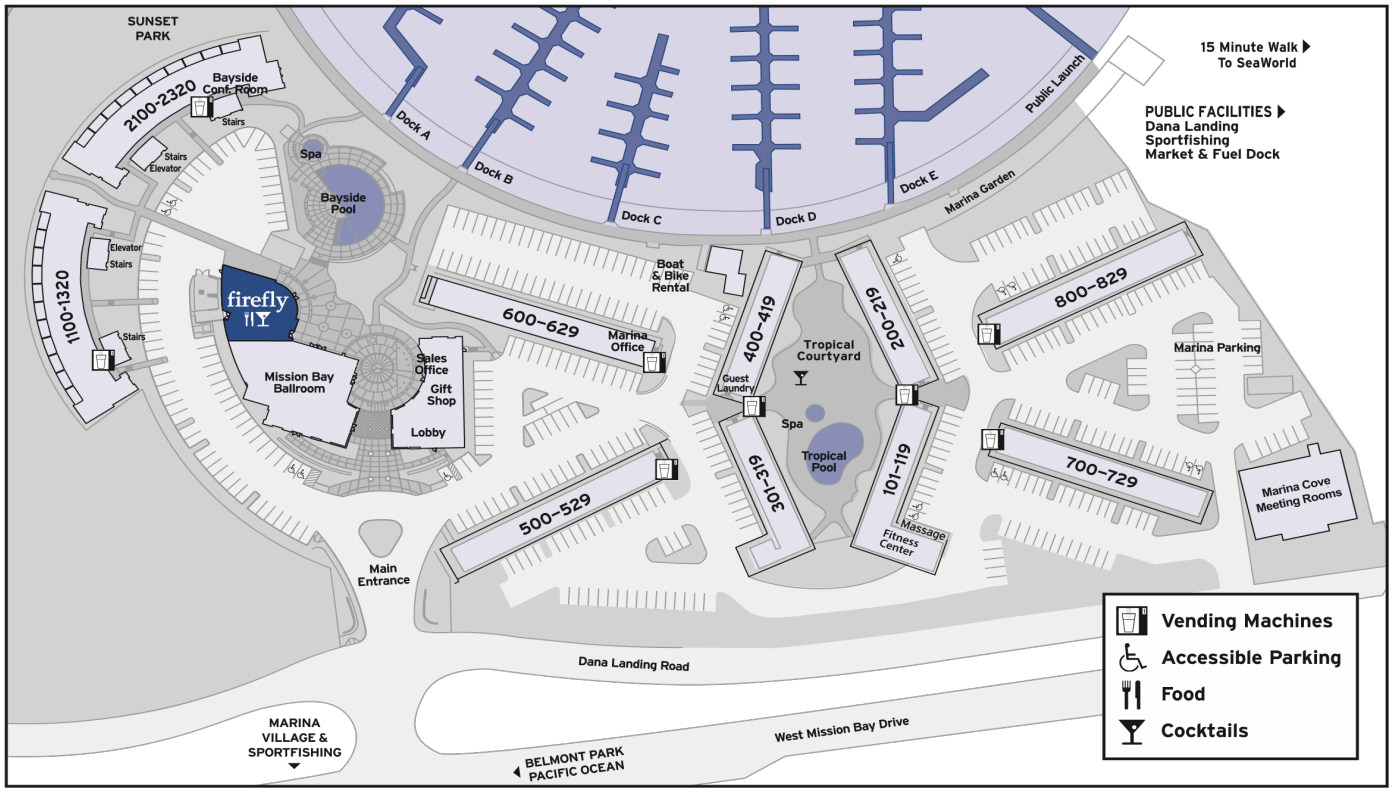
The SBP Rare Disease Day Symposium is a pioneer in *scientist/patient interaction*, opening its doors for CDG families to participate in every aspect of the annual event. Our innovative Doctor-Is-In Session, scheduled for the afternoon of Saturday, February 24, brings together medical researchers, clinicians, advocates, patients and their families for an afternoon of hands-on collaboration in a small-group, round-robin format. For some medical researchers, the doctor-is-in session is their first opportunity to observe and interact with patients in-person. This inclusive, collaborative approach has led to profound experiences and unlikely partnerships, and we are excited to offer the Doctor-Is-In Session again in 2018.

Day Care (Hours, Info)

As a recipient of the 2018 Rare Patient Impact Grant Program offered by Global Genes, we will be providing a 2 ½-day Therapeutic Daycare Program for the CDG-affected children and siblings who will be traveling with their families for this bi-annual event. This year, we have 44 children who are signed up to participate in a full array of sensory and therapeutic activities. The Program will consist of qualified clinical and/or therapeutic volunteers and include special interest activities in the areas of specific need for CDG children, including music and art therapy, physical interaction and exploration, and a full-day of the innovative *Movement, Muscles and Messes Program*, offered again this year by the University of St. Augustine Pediatric Occupational Therapy Class. For a full listing of program activities and operating hours, please refer to the Daycare Program Agenda on page 9.

The check-in for the Daycare Program will be at the Daycare/Volunteer Registration Table in front of the Bayside Conference Room.

The Dana on Mission Bay Property Map



<u>Wi-Fi Conference Code</u>

Access: **Dana Meeting**; Voucher: **CDG18**

SBP Scientific Meeting and Poster Session Friday, February 23, 2018		
Time	**Activity**	**Speaker**
OPENING SESSION		
9:00 am – 9:20 am	Introduction and Welcome	**Malin Burnham and T. Denny Sanford** Philanthropists and Honorary Trustees, Sanford Burnham Prebys Medical Discovery Institute (SBP)
9:20 am – 10:00 am	**Keynote address**: 21st Century Medicine Will Transform Healthcare	**Leroy Hood, MD, PhD** President & Co-Founder, Institute for Systems Biology; Senior Vice President & Chief Science Officer, Providence Health & Services
PMM2-CDG: OVERVIEW, MODELS AND THERAPIES **Chair: Marc Patterson, MD**, Professor of Neurology, Pediatrics and Medical Genetics, and Chair of the Division of Child and Adolescent Neurology, Mayo Clinic		
10:00 am – 10:25 am	Natural History of PMM2-CDG	**Lynne Wolfe, MS, CNRP** Senior Nurse Practitioner, NIH/NHGRI, NIH Site Coordinator, Undiagnosed Diseases Program, Study Coordinator, Epi-743 MitoWorks Study and Congenital Disorders of Glycosylation Protocol
10:25 am – 10:50 am	Coordinated movement, neuromuscular synaptogenesis and *trans*-synaptic signaling defects in a new *Drosophila* PMM2-CDG disease model	**Kendal S. Broadie, PhD** Stevenson Professor of Neurobiology, Professor of Biological Science, Cell & Developmental Biology and Pharmacology, Vanderbilt University and Medical Center
10:50 am – 11:00 am	BREAK	
11:00 am – 11:25 am	Lineage-dependent variations in glycosylation: insights from CDG iPSC models	**Richard Steet, PhD** Professor of Biochemistry and Molecular Biology Complex Carbohydrate Research Center University of Georgia
11:25 am – 11:50 am	Lipo-M1P as a potential therapy for PMM2-CDG	**Patrice Rioux, MD, PhD** Chief Medical Officer, Glycomine Inc.
11:50 am – 12:15 am	Perspectives on ALG9-CDG	**Duncan Webster, MD** CDG Parent and Advocate; Associate Professor, Faculty of Medicine, Dalhousie University; Internal Medicine & Infectious Diseases Consultant and Medical Microbiologist, Saint John Regional Hospital, Saint John, New Brunswick

SBP Scientific Meeting and Poster Session - Friday, February 23, 2018		
Time	**Activity**	**Speaker**
12:15 pm – 1:15 pm	LUNCH	
OTHER GLYCOSYLATION DISORDERS: NOVEL APPROACHES **Chair: Hudson Freeze, PhD**, Professor of Glycobiology and Director, Human Genetics Program, Sanford Children's Health Research Center, SBP		
1:15 pm – 1:40 pm	Novel approaches to identify and validate rare disease genes	**Josef Penninger, MD, PhD** Scientific Director, IMBA, Institute for Molecular Biotechnology of the Austrian Academy of Sciences, Vienna. Austria
1:40 pm – 2:05 pm	Overview of current monosaccharide therapy trial in congenital disorders of glycosylation	**Eva Morava, MD, PhD** Professor of Pediatrics, Clinical Biochemical Geneticist, Senior Associate Consultant, Department of Clinical Genomics, Mayo Clinic *Editor in Chief Journal of Inherited Metabolic Disease*
2:05 pm – 2:30 pm	Peters Plus Syndrome: A congenital disorder of glycosylation	**Robert S. Haltiwanger, PhD** GRA Eminent Scholar in Biomedical Glycosciences Editor-in-Chief, Glycobiology Professor of Biochemistry and Molecular Biology Complex Carbohydrate Research Center University of Georgia
2:30 pm – 2:55 pm	Mechanistic insights and therapeutic approaches for O-glycosylation-deficient muscular dystrophy	**Kevin P. Campbell, PhD** Investigator, Howard Hughes Medical Institute, Roy J. Carver Biomedical Research Chair in Molecular Physiology and Biophysics, Executive Officer of the Dept of Molecular Physiology and Biophysics Director, Senator Paul D. Wellstone Muscular Dystrophy Cooperative Research Center at the Carver College of Medicine, University of Iowa
2:55 pm – 3:10 pm	BREAK	
3:10 pm – 3:35 pm	Genetic knockout screens reveal a critical role for glycosylation in infectious disease	**Jan Carette, PhD** Assistant Professor, Department of Microbiology and Immunology, Stanford University
3:35 pm – 4:00 pm	Novel glycosylation disorders	**Hudson Freeze, PhD** Professor of Glycobiology and Director, Human Genetics Program, Sanford Children's Health Research Center, SBP
4:00 pm – 5:00 pm	Question and Answer Session: CDG	**Moderator: Hudson Freeze, PhD**
5:00 pm – 6:30 pm	POSTER SESSION	

SBP Scientific Meeting and Doctor-Is-In Session

Saturday, February 24, 2018

Time	Activity	Speaker
NGLY1: A DISORDER OF DE-GLYCOSYLATION **Chair: Randal Kaufman, PhD**, Director and Professor, Degenerative Diseases Program, Neuroscience and Aging Research Center, SBP		
9:00 am – 9:25 am	Precision medicine and NGLY1	**Matthew Might, PhD** Director, Hugh Kaul Personalized Medicine Institute, UAB School of Medicine
9:25 am – 9:50 am	Discovering NGLY1 therapeutics: A research collaboration between NCATS, NGLY1.org and Retrophin	**Steve Rodems, PhD** Senior Director, Discovery Biology, Retrophin **Wei Zheng, PhD** Group Leader, Biology, Therapeutics for Rare and Neglected Diseases, Division of Pre-Clinical Innovation, National Center for Advancing Translational Sciences (NCATS), National Institutes of Health (NIH)
9:50 am – 10:15 am	Genetic analysis of NGLY1 action in proteasome biology	**Gary B. Ruvkun, PhD** Department of Molecular Biology, Massachusetts General Hospital Professor, Harvard Medical School
10:15 am – 10:30 am	BREAK	
10:30 am – 10:55 am	Can basic science contribute to curing human genetic disorders?	**Tadashi Suzuki, DSci** Team Leader, Glycometabolome Team, RIKEN Global Research Cluster
10:55 am – 11:20 am	Genetic and environmental modulation of NGLY1 deficiency	**Clement Chow, PhD** Assistant Professor, Department of Human Genetics, University of Utah
11:20 am – 11:45 am	Role of NGLY1 in *Drosophila* development	**Hamed Jafar-Nejad, MD** Associate Professor, Department of Molecular and Human Genetics, Program in Developmental Biology, Baylor College of Medicine
11:45 am – 12:15 pm	Question and Answer Session: NGLY1	**Moderator: Matthew Might, PhD**
12:15 pm – 1:45 pm	LUNCH	

SBP Scientific Meeting and Doctor-Is-In Session - Saturday, February 24, 2018		
Time	**Activity**	**Speaker**
DOCTOR-IS-IN SESSION		
1:45 pm – 2:00 pm	Doctor-Is-In Session Orientation	**Hudson Freeze, PhD** Professor of Glycobiology and Director, Human Genetics Program, Sanford Children's Health Research Center, SBP
2:00 pm – 4:00 pm	DOCTOR-IS-IN SESSION	
4:00 pm – 6:00 pm	EVENING RECEPTION	

CDG/NGLY1 Family Conference Sunday, February 25, 2018		
Time	**Activity**	**Speaker**
8:15 am – 8:30 am	Welcome	**Andrea Berarducci, JD, MHA** President, CDG CARE
8:30 am – 9:00 am	CDG Neurologic Overview and Updates	**Marc Patterson, MD** Professor of Neurology, Pediatrics and Medical Genetics, and Chair of the Division of Child and Adolescent Neurology, Mayo Clinic
9:00 am – 9:30 am	Genetic Counseling for CDG	**Carolyn Brown, MSc, CGC, CCGC** Genetic Counselor, Illumina Clinical Services Laboratory
9:30 am – 10:00 am	Puberty and Bone Health in CDG Patients	**Bradley S. Miller, MD, PhD** Associate Professor, Pediatric Endocrinology, University of Minnesota Masonic Children's Hospital
10:00 am – 10:30 am	BREAK	
10:30 am – 11:00 am	Personalized medicine in CDG, an overview of potential therapies	**Eva Morava, MD, PhD** Professor of Pediatrics, Clinical Biochemical Geneticist, Senior Associate Consultant, Department of Clinical Genomics, Mayo Clinic *Editor in Chief Journal of Inherited Metabolic Disease*

CDG/NGLY1 Family Conference - Sunday, February 25, 2018		
Time	**Activity**	**Speaker**
11:00 am – 11:30 am	What is the liver? What is liver disease? And what to expect in CDG?	**David Cassiman, MD, PhD** Professor, Metabolic Center, Dept. of Gastroenterology-Hepatology, University of Leuven, Belgium Professor, University of Hasselt
11:30 am – 12:00 pm	Genetics and CDG	**Christina Lam, MD** Attending physician, Seattle Children's Hospital (SCH) Assistant Professor, Department of Pediatrics, University of Washington School of Medicine (UW)
12:00 pm – 1:00 pm	LUNCH	
1:00 pm – 1:30 pm	CDG Updates and Perspectives	**Hudson Freeze, PhD** Professor of Glycobiology and Director, Human Genetics Program, Sanford Burnham Prebys Medical Discovery Institute (SBP)
1:30 pm – 2:00 pm	Sensory Processing and Behavior	**Becki Cohill, OTD, OTR** University of St. Augustine
2:00 pm – 2:30 pm	Becoming an Advocate for the CDG Community	**Carrie Ostrea** Executive Director, NGLY1.org
2:30 pm – 3:00 pm	BREAK	
3:00 pm – 3:30 pm	Cutting Edge Treatments and Therapies	**Lynette LaScala** Founder and CEO, NAPA Center
3:30 pm – 4:00 pm	Developing Your Child's IEP: Your Role in the Process	**Allyssa Harpst, MEd** CDG Parent, Teacher for Students with Visual Impairments, Urbana School #116
4:00 pm - 4:30 pm	Researching What Matters for the CDG Community by a CDG Sibling	**Vanessa R Ferreira** President, APCDG & CDG Sibling **Rita Francisco** CDG PhD Student and Community Advocate
4:30 pm	CLOSING	**Andrea Berarducci, JD, MHA** President, CDG CARE

2018 CDG Family Conference Daycare Program
Bayside Conference Room

***The check-in for the Daycare Program will be at the Daycare/Volunteer Registration Table in front of the Bayside Conference Room.**

Friday, February 23, 2018	
8:45 am - 9:00 am	Parents drop off their child/children to assigned daycare room
9:00 am - 10:50 am	Daycare Program - Movement, Muscles and Messes
10:50 am - 11:00 am	Parents check in on their child/children in Daycare and/or bring them to Break
11:00 am - 12:15 pm	Daycare Program - Movement, Muscles and Messes
12:15 pm - 1:15 pm	Parents pick up their child/children for LUNCH Break
1:15 pm - 2:55 pm	Daycare Program - Movement, Muscles and Messes
2:55 pm - 3:10 pm	Parents check in on their child/children in Daycare and/or bring them to Break
3:10 pm - 4:45 pm	Daycare Program - Movement, Muscles and Messes
4:45 pm - 5:00 pm	Parents pick up their child/children from the daycare rooms

Saturday, February 24, 2018	
8:45 am - 9:00 am	Parents drop off their child/children to assigned daycare room
9:00 am - 10:30 am	Daycare Program GROUP A - Pacific Animal Zoo Encounter Daycare Program GROUP B - Disney movie presentation & My Colors Speak
10:15 am - 10:30 am	Parents check in on their child/children in Daycare and/or bring them to Break
10:30 am – 12:15 pm	Daycare Program GROUP A - Disney movie presentation & My Colors Speak Daycare Program GROUP B - Pacific Animal Zoo Encounter
12:15 pm	Parents pick up their child/children from the Daycare rooms

***NOTE – No Saturday Afternoon Daycare Program**

Sunday, February 25, 2018	
8:00 am – 8:15 am	Parents drop off their child/children to assigned daycare room
8:15 am – 10:00 am	Daycare Program - Carnival
10:00 am – 10:30 am	Parents check in on their child/children in Daycare and/or bring them to Break
10:30 am – 12:00 pm	Daycare Program - Music Therapy Group Program
12:00 pm – 1:00 pm	Parents pick up their child/children for LUNCH Break
1:00 pm – 2:30 pm	Daycare Program - Carnival and FRESH mini doughnuts
2:30 pm – 3:00 pm	Parents check in on their child/children in Daycare and/or bring them to Break
3:00 pm – 4:30 pm	Daycare Program - Disney movie presentation
4:30 pm	Parents pick up their child/children from the daycare rooms

Special Acknowledgement to our Therapeutic Daycare Program Service Providers, In-Kind Donors and Volunteers:

About SBP
Sanford Burnham Prebys Medical Discovery Institute (SBP) is an independent nonprofit medical research organization that conducts world-class, collaborative, biological research and translates its discoveries for the benefit of patients. SBP focuses its research on cancer, immunity, neurodegeneration, metabolic disorders and rare children's diseases. The Institute invests in talent, technology and partnerships to accelerate the translation of laboratory discoveries that will have the greatest impact on patients. Recognized for its world-class NCI-designated Cancer Center and the Conrad Prebys Center for Chemical Genomics, SBP employs about 1,000 scientists and staff in San Diego (La Jolla), Calif., and Orlando (Lake Nona), Fla. For more information, visit us at SBPdiscovery.org or on Facebook at facebook.com/SBPdiscovery and on Twitter @SBPdiscovery.

About CDG CARE
CDG CARE is a nonprofit organization founded by parents seeking information and support for a group of disorders known as Congenital Disorders of Glycosylation (CDG). Our mission is to promote greater awareness and understanding of CDG, to provide information and support to families affected by CDG, and to advocate for scientific research to advance the diagnosis and treatment of CDG. The strategy of CDG CARE is to disseminate information through a modern website, publish and distribute a semi-annual e-newsletter, organize a bi-annual North American CDG family conference, and collaborate globally with medical professionals and nonprofit associations worldwide. To learn more about CDG CARE and our programs, like us on the CDG CARE Facebook page and visit our website at cdgcare.com.

About NGLY1.org
NGLY1.org is a nonprofit 501(c)(3) patient support, advocacy and research organization for N-Glycanase (NGLY1) Deficiency. NGLY1.org's mission is to eliminate the challenges of NGLY1 through research, awareness and support. NGLY1.org improves the lives of patients through family support, diagnostic access, clinical and patient education, conferences, and grants. For more information, visit NGLY1.org