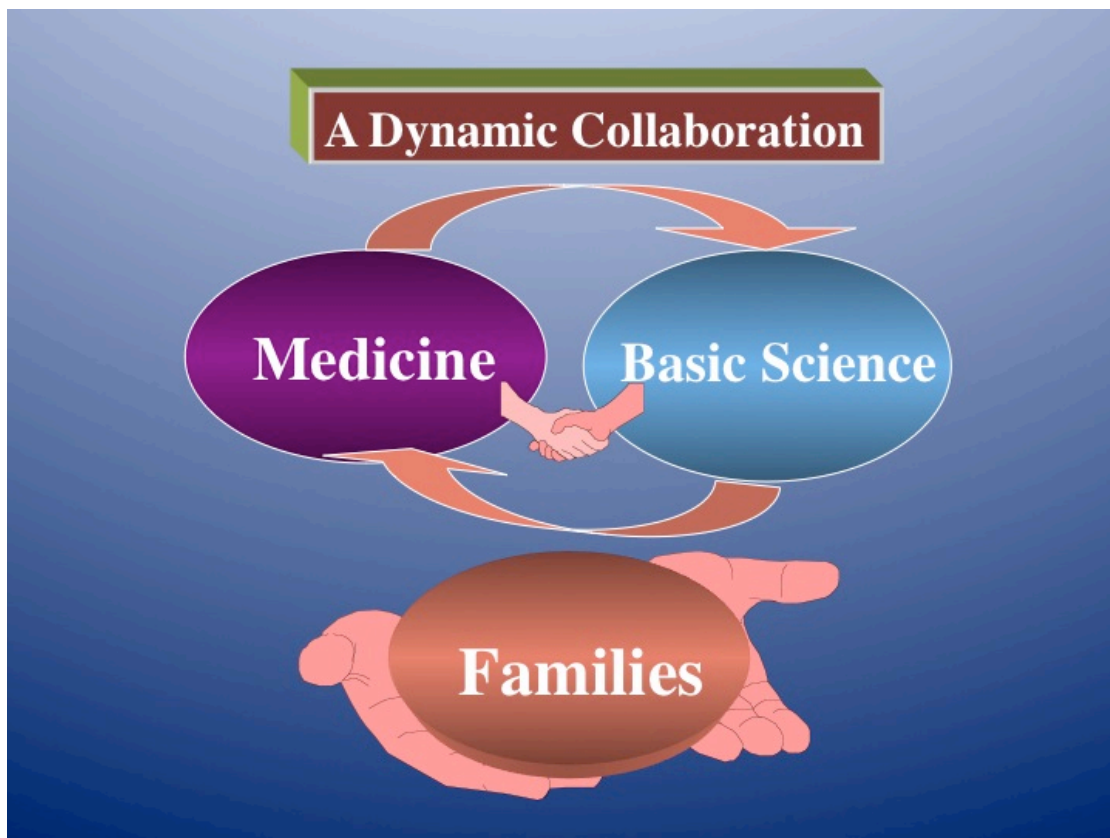


**THE SANFORD CENTER GENETIC DISEASE PROGRAM**  
**The First Burnham Symposium on Rare Diseases:**  
**Collaborations That Drive Discovery, Therapy and Advocacy**

February 28, 2010 is International Rare Disease Day. In a one-day meeting on Feb 26, 2010 we will take this opportunity to focus on the contributions of Sanford-Burnham Faculty Members and their scientific, community, and philanthropic collaborators to understand, treat, and cure Rare Disorders.

In this spirit, we call on each of these communities to collaborate as we focus on the current scientific status of several rare diseases and explore resources for the community of scientists, physicians, and patient family organizations committed to fighting rare diseases.

This meeting will draw on the proven strength of Sanford-Burnham scientists and others, who reached out to funding agencies, physicians, family support groups, and philanthropists to translate our work into treatments. It was these personal, sometimes uncommon, approaches to collaboration that helped drive our discoveries, and they still hold the promise of future therapies. This is the meeting's focus.



# Sanford|Burnham

## Medical Research Institute

*1<sup>st</sup> Annual Sanford-Burnham Rare Disease Symposium*  
*presents*

## **Collaborations that Drive Discovery, Therapy and Advocacy**

### **WELCOME**

8:00 – 8:25

Registration

8:25 – 8:30

Introduction and Welcome by Hudson Freeze, Sanford-Burnham Medical Research Institute

### **BONE DISORDERS**

CHAIR: Hudson Freeze

8:30 – 8:55

Yu Yamaguchi, Sanford-Burnham Medical Research Institute

*Multiple hereditary exostoses: the power of collaboration between patient advocacy groups and research community to reveal the true scope of the disease*

8:55 – 9:20

Jose Luis Millan, Sanford-Burnham Medical Research Institute

*Pathogenesis and treatment of Infantile Hypophosphatasia*

9:20 – 9:40

Craig Eaton, MHE Foundation, New York

*TBD*

### **CONGENITAL DISORDERS OF GLYCOSYLATION**

CHAIR: Yu Yamaguchi

9:40 – 10:05

Hudson Freeze, Sanford-Burnham Medical Research Institute

*CDG Animal Models and Therapy*

10:05 – 10:30

Vincent Cantagrel, University of California, San Diego

*Rare cerebellar disorder pinpoints earliest known step in protein glycosylation*

10:30 – 10:45

BREAK

10:45 – 11:10

Madurhi Hegde, Emory University

*Update on molecular testing for rare disorders: CMD and CDG*

11:10 – 11:30

Cindy Wren-Gray, CDG Family Network

*Vision and Balance – A Collaborative Opportunity for Rare Pioneers*

### **MUSCLE DISORDERS**

CHAIR: Jose Luis Millan

11:30 – 11:55

Alessandra Sacco, Sanford-Burnham Medical Research Institute

*TBD*

11:55 – 12:20

Lance Wells, University of Georgia

*Structural Analysis of Functional O-Mannose Glycans in  $\alpha$ -Dystroglycan*

12:20 – 12:40

Anne Rutkowski, Cure CMD

*CMD (Congenital Muscular Dystrophy): From Diagnosis to Translation*

### **LUNCH**

12:40 – 1:30

Lunch Break

## **LUNCH**

1:00 – 1:20

Lunchtime Presentation

Eric Little, CTG Executive Consultant, Buffalo, NY

*Using Semantic Technologies for Building User Communities Around Rare Disease: Linking Providers, Researchers and Families*

## **NEEDLES & HAYSTACKS**

CHAIR: David Pearce

1:30 – 1:55

Jeffrey Esko, University of California, San Diego

*Resolving Glycosaminoglycan Accumulation in Lysosomal Storage Disorders*

1:55 – 2:20

Michael Jackson, Sanford-Burnham Medical Research Institute

*TBD*

2:20 – 2:45

Phil Wood, Sanford-Burnham Medical Research Institute

*TBD*

2:45 – 3:00

BREAK

## **WITHIN THE CELL**

CHAIR: Jeffrey Esko

3:00 – 3:25

David Pearce, Sanford Childrens Health Research Center, Sioux Falls, SD

*Juvenile Batten Disease: Bench to Bedside*

3:25 – 3:50

Sarah Courtneidge, Sanford-Burnham Medical Research Institute

*Frank-Ter Haar Syndrome is caused by mutations in the podosome protein Tks4*

3:50 - 4:15

Bruce Barshop, University of California, San Diego

*Lessons from Cystinosis and Lesch-Nyhan Disease*

## **DISCUSSION AND RECEPTION**

CHAIRS: Hudson Freeze & Yu Yamaguchi

4:20 – 5:00

Discussion

5:15 – 5:30

Tour of Conrad Prebys Center for Chemical Genomics

5:30 – 6:30

Reception in Fishman Auditorium

**MULTISYSTEMIC RARE DISEASES  
AN OPPORTUNITY FOR NON-TRADITIONAL COLLABORATIONS BETWEEN  
PHYSICIANS AND SCIENTISTS**

Hudson Freeze PhD  
Sanford Professor of Glycobiology  
Sanford-Burnham Medical Research Institute, La Jolla, CA

In 2005, Ley and Rosenberg predicted that the dwindling supply of funded Physician-Scientists would witness resurgence, thanks to new NIH initiatives in 2002. (*JAMA*. 2005; 294:1343-1351). Physician scientists are indispensable to train the next generation of physicians in the medical specialties. However, the demands and realities of the organ-system based approach to specialized medical education does not encourage specialists to cross their traditional boundaries. For example, where does a parent turn for an explanation of why their child develops protein-losing enteropathy years after his or her Fontan surgery? Is it the pediatric cardiologist or gastroenterologist? Why should a high proportion of patients with hereditary multiple exostosis be on the autistic spectrum? Traditionally trained specialists cannot easily supply the answers. It calls for boundary-crossing collaborations not only among physicians and physician-scientists, but also basic scientists. This last group needs to be actively recruited into these collaborations. Novel perspectives on the two examples above emerged from basic scientists who study rare human glycosylation disorders. They easily cross the traditional medical boundaries and offer different perspectives than the physicians treating the patients with multi-systemic pathologies.

Studies of rare disease inform more prevalent disorders, as well. For instance some features of protein-losing enteropathy provided a molecular metric to distinguish protein-energy malnutrition patients with kwashiorkor vs. marasmus. Programs that bring physicians and their patients together under the umbrella of a scientific discipline offer precedent-setting, non-traditional solutions. This is especially so for those disciplines not yet fully integrated into the medical and scientific gestalt. Glycobiology is an example. A pilot program is needed to encourage and enhance interactions of these basic scientists with academic physicians from different specialties to cross educate each other. This would require the creation of appropriately protected (anonymous) accessible and searchable genetic, biochemical and medical databases, sample (cell, serum, DNA) repositories for analysis by basic scientists. Cross training of postdoctoral fellows from basic science in some of the basic medical aspects would add a new dimension to the medical-science enterprise: The intent is not to train a scientist to care for patients, but to know enough to appreciate and value and integrate medical perspectives. This exchange would empower and inform both groups and narrow the divide between them. This is needed as we all become more interdependent; it will ultimately benefit the patients and families who look to us for the answers and solutions. Collaboration with each other is essential.