

**Calcification Disorder Advocacy Group Representatives**  
**Charlene Waldman, Session Chair**

**Becky Mock**  
President,  
*XLH Network*

**Tiffany Ferry**  
President, *Lymphatic  
Malformation Institute*

**Craig Eaton, President**  
**Sarah Ziegler, Founder**  
*MHE Research Foundation*

**Deborah Sittig**  
Founder,  
*Soft Bones Inc.*

**David Heaps**  
Founder, Patient  
*HPP Choose Hope*

**Tracy Smith Hart**  
CEO, *Osteogenesis  
Imperfecta Foundation*

**Jack Kelly**  
President, *Lymphangiomas &  
Gorham's Disease Alliance*

*To learn more about organizations supporting  
calcification disorder research, please see the literature table  
in the building lobby near the registration desk.*

**Sanford|Burnham**  
Medical Research Institute

**Fourth Annual Rare Disease Day Symposium**

**Calcification Disorders:  
from Hardened  
Arteries to Soft Bones**

Sponsored by:



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**Symposium Chair**

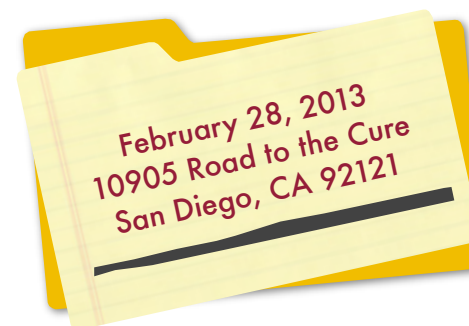
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# Sanford-Burnham Medical Research Institute

## Rare Disease Day Symposium | February 28, 2013

### 8:00-8:30 REGISTRATION

#### Session 1: Diagnosis and Treatment of Vascular Calcification

Chair: José Luis Millán, Ph.D.

#### 8:30-8:35 Introduction and Welcome

José Luis Millán, Ph.D.

#### 8:35-8:45 Opening Remarks

Kristiina Vuori, M.D., Ph.D.

President and Interim CEO,  
Sanford-Burnham Medical Research Institute

### 8:45-9:45 KEYNOTE ADDRESS

#### The NIH Undiagnosed Diseases Program: New insights using genomic methodologies

William A. Gahl, M.D., Ph.D.

Clinical Director, NHGRI

Director, NIH Undiagnosed Diseases Program

### 9:45-10:00 BREAK

#### 10:00-10:30 New insights into the development of aortic calcification and MGP deficiency

Hervé Kempf, Ph.D.

Université de Lorraine, France

#### 10:30-11:00 Generalized arterial calcification of infancy and pseudoxanthoma elasticum: Two faces of the same coin

Frank Rutsch, M.D.,

Muenster University Children's Hospital, Germany

#### 11:00-11:30 Phosphatase inhibitors for the prevention and treatment of medial vascular calcification

José Luis Millán, Ph.D.

Sanford-Burnham Medical Research Institute

Lunch is included with your registration and will be available from  
**11:30 a.m. to 12:30 p.m.**

### Session 2: Pathophysiology and Management of Soft Bones

Chair: Hudson Freeze, Ph.D.

#### 12:30-1:00 Update on bone-targeted enzyme replacement therapy for hypophosphatasia

Michael P. Whyte, M.D.

Shriners Hospital for Children

#### 1:00-1:30 Recessively inherited osteogenesis imperfecta and the biology and medicine of defects in collagen processing

Peter H. Byers, M.D.

University of Washington

#### 1:30-2:00 FGF23: a common denominator in metabolic bone diseases

Kenneth E. White, Ph.D.

Indiana University

#### 2:00-2:30 X-linked hypophosphatemia 2013: a clinical update of the prototype renal phosphate wasting disorder

Thomas O. Carpenter, M.D.

Yale University

### 2:30-2:45 BREAK

### Session 3: Pathophysiology and Management of Soft-Tissue Ossification

Chair: Yu Yamaguchi, M.D., Ph.D.

#### 2:45-3:15 Heterotopic bone formation in fibrodysplasia ossificans progressiva (FOP) and progressive osseous heteroplasia (POH)

Eileen M. Shore, Ph.D.

University of Pennsylvania

#### 3:15-3:45 MHE: Pathogenic mechanisms and prospects for therapies

Maurizio Pacifici, Ph.D.

The Children's Hospital of Philadelphia

#### 3:45-4:15 Genetic skeletal diseases caused by non-heritable mutations, such as Gorham-Stout and CLOVES

Matthew L. Warman, M.D.

Boston Children's Hospital

#### 4:15-5:15 Panel Discussion: Patient Advocacy Groups

Session Chair: Charlene Waldman

#### 5:15-5:30 Closing Remarks