## 4th Annual Sanford-Burnham Rare Disease Day Symposium **February 28, 2013**

## Calcification Disorders - from Hardened Arteries to Soft Bones

8:00-9:00	REGISTRATION	
9:00-12:00	Session 1:	Diagnosis and Treatment of Vascular Calcification José Luis Millán, Ph.D.
	9:00-9:05	Introduction and Welcome José Luis Millán, Ph.D., Sanford-Burnham Medical Research Institute
	9:05-9:15	Opening Remarks Kristiina Vuori, Ph.D., President, Sanford-Burnham Medical Research Institute
	9:15-10:15	<b>KEYNOTE ADDRESS William A. Gahl, M.D., Ph.D.</b> , Clinical Director, NHGRI; Director, NIH Undiagnosed Diseases Program  Topic: Whole exome sequencing to identify rare disorders, including the role of NT5E in vascular calcification and PXE.
10:15-10:30	BREAK	
	10:30-11:00	Hervé Kempf, Ph.D., Researcher, Université de Lorraine, France Topic: Development of aortic calcification and MGP deficiency.
	11:00-11:30	<b>Frank Rutsch</b> , <b>M.D.</b> , Professor, Muenster University Children's Hospital, Germany <i>Topic: Clinical experience with patients with GACI</i> .
	11:30-12:00	<b>José Luis Millán</b> , <b>Ph.D.</b> , Professor, Sanford-Burnham Medical Research Institute  Topic: TNAP inhibitors for the treatment of vascular calcification in GACI and Chronic Kidney disease.
12:00-1:00	LUNCH	
1:00-3:00	Session 2:	Pathophysiology and Management of Soft Bones Hudson Freeze, Ph.D.
	1:00-1:30	<b>Michael Whyte</b> , <b>M.D.</b> , Medical-Scientific Director, Center for Metabolic Bone Disease and Molecular Research, Shriners Hospital for Children
	1:30-2:00	Topic: Update on bone-targeted enzyme replacement therapy for hypophosphatasia.  Peter Byers, M.D., Professor, Departments of Pathology and Medicine (Medical Genetics), University of Washington
	2:00-2:30	Topic: Rare forms of Osteogenesis imperfecta.  Kenneth White, Ph.D., Associate Professor of Medical and Molecular Genetics, Indiana University
	2:30-3:00	Topic: Autosomal Dominant Hypophosphatemic Rickets.  Thomas Carpenter, M.D., Professor of Pediatrics (Endocrinology) and of Orthopaedics and Rehabilitation and Clinical Professor of Nursing; Director, Yale Center for X-Linked Hypophosphatemia; Medical Director, Hospital Research Unit Topic: X-Linked Hypophosphatemia.
3:00-3:15	BREAK	
3:15-5:30	Session 3:	Pathophysiology and Management of Soft-Tissue Ossification Yu Yamaguchi, M.D., Ph.D.
	3:15-3:45	<b>Eileen Shore</b> , <b>Ph.D.</b> , Cali and Weldon Research Professor, University of Philadelphia <i>Topic: Fibrodysplasia ossificans progressiva (FOP) and progressive osseous heteroplasia (POH).</i>
	3:45-4:15	<b>Maurizio Pacifici</b> , <b>Ph.D.</b> , Director of Research, Division of Orthopaedic Surgery, The Children's Hospital of Philadelphia <i>Topic: Multiple Hereditary Exotoses (MHE)</i> .
	4:15-5:15	Panel Discussion: Patient Advocacy Groups
	5:15-5:30	Closing Remarks José Luis Millán, Ph.D., Sanford-Burnham Medical Research Institute
5:30-8:00	DINNER FOR SPEAKERS, SPONSORS, AND INVITED GUESTS	

Sanford-Burnham Wireless Access Domain: **SBMRI-Guest**