



THURSDAY, APRIL 3

**AM SESSION**

7:30 AM **Breakfast**

8:15 AM **Welcome**

Cystinosis Research Foundation

8:20 AM **Keynote**

History of Cystinosis in the United States

Jerry Schneider, MD

University of California, San Diego

**Studies**

AM Moderator:

William Rizzo, MD

University of Nebraska

9:00 AM

Mitochondrial Dysfunction in  
Cystinosis Myopathy

Doris Trauner, MD

University of California, San Diego

9:30AM

Identification and Characterization of the  
Lysosomal Transporter Involved in  
Cysteamine Mediated Cystine Efflux

Bruno Gasnier, PhD

Institut de Biologie

Physico-Chimique, Paris

Ellen Closs, PhD

Johannes Gutenberg U., Mainz

10:00 AM **Coffee**

10:30 AM

Characterization of cystinosis  
intracellular trafficking

Corinne Antignac, MD, PhD

Hôpital Necker-Enfants Malades,

Paris

11:00 AM

Molecular Mechanisms of  
Hypopigmentation in Cystinosis

Christine Chiaverini, MD (trainee)

\*Robert Ballotti, PhD (mentor)

\*presenting

faculté de Médecine, and

Dermatology, hospital Archet 2

Nice

11:30 AM

Yeast Model for Cystinosis

Seasson Phillips, PhD (trainee)

David Pearce, PhD (mentor)

University of Rochester

12 Noon **Lunch**



THURSDAY, APRIL 3

**PM SESSION**

1:10 PM

**Keynote**

History of Cysteamine in Cystinosis

Jess Thoene, MD  
University of Michigan

**Studies**

PM Moderator:

Adam Jonas, MD  
University of California Los Angeles

1:50 PM

Cystinosis: Insights into an Adult  
Lysosomal Storage Disorder

Robert Kleta, MD, PhD  
University College London

2:15 PM

Cognitive Domain of Executive  
Functioning in Cystinosis

Angela Ballantyne, PhD  
University of California, San Diego

2:40 PM

Methodology in Clinical Cystinosis  
Studies

Meredith Fidler, PhD  
University of California, San Diego

3:00 PM

**Coffee**

3:30 PM

Tandem Mass Spectrometry Cystine  
Determination

Jon Gangoiti, PhD  
Bruce Barshop, MD  
University of California, San Diego

4:00 PM

Molecular and Pathogenic Study  
of Cystinosis

Sha Tang, PhD (trainee)  
Taosheng Huang, MD, PhD (mentor)  
University of California, Irvine

4:30 PM

Enteric-Coated Cysteamine in Cystinosis

Ranjan Dohil, MD  
University of California, San Diego



FRIDAY, APRIL 4

**AM SESSION**

7:30 AM **Breakfast**

8:20 AM **Keynote**

History of Cystinosis in Europe.  
Experience at Necker Hospital:  
Outcome after age 20y

Michel Broyer, MD  
Hôpital Necker-Enfants Malades  
Paris

**Studies**

AM Moderator:

Julie Ingelfinger, MD  
Harvard Medical School

9:00 AM

Treatment of Cystinosis Nephropathy with  
Adult Stem Cells in Murine Cystinosis  
Model

Stephanie Cherqui, PhD  
Daniel Salomon, MD  
The Scripps Research Institute

9:30 AM

Gene Transfer Studies for Cystinosis

Claire Hippert (Trainee)  
Vasiliki Kalatzis, PhD (mentor)  
Eric J. Kremer, PhD (mentor)  
Institut Génétique Moléculaire  
de Montpellier

10:00 AM

In Vivo Confocal Microscopy to Detect  
Cystine Crystals in the *Cnts* <sup>-/-</sup> Mouse.

James V. Jester, PhD  
University of California, Irvine

10:10 AM **Coffee**

10:30 AM

Parthenogenetic Embryonic Stem Cells  
as a Source of Immunocompatible Renal  
Progenitor Cells for Therapy of  
Nephropathic Cystinosis

Holger Willenbring, MD  
University of California,  
San Francisco

11:00 AM

Studying the Pathogenesis of Renal Disease  
in Cystinosis

Elena Levtchenko, MD, PhD  
University Hospital Leuven Belgium  
Radboud University Nijmegen

11:30 AM

Cysteamine Effects on Extracellular Matrix  
Accumulation in Chronic Kidney Disease

Allison Eddy, MD  
University of Washington

12 Noon **Lunch**



FRIDAY, APRIL 4

**PM SESSION**

1:10 PM

**Keynote**

The Biology and Genetics of Mitochondrial Disease and Their Relationship to Cystinosis

Douglas Wallace, PhD  
University of California, Irvine

**Studies**

PM Moderator:

Michel Baum, MD  
University of Texas Southwestern Medical Center

1:50 PM

Lysosomal Cystine Enhanced Apoptosis in Cultured Human Cells

Jess Thoene, MD  
University of Michigan

2:20 PM

Auditory and Visual Processing in Cystinosis

Rita Ceponiene, MD, PhD  
University of California, San Diego

2:50 PM

**Coffee**

3:20 PM

Complex Genetic Approaches to Monogenic Disease: Genomic and Transcriptomic Dissection of Normal Expression of *CTNS*

Katy Freed, PhD (trainee)  
Eric Moses, PhD (mentor)  
Southwest Foundation for Biomedical Research, San Antonio

3:50 PM

Transcriptional and Post-transcriptional Regulation of the *CTNS* Gene

Francesco Emma, MD  
Anna Taranta, PhD  
Bambino Gesù Children's Hospital and Research Institute, Rome  
Elena Levtchenko, MD, PhD  
Radboud University Nijmegen

4:20 PM

Targeted Cell Fusion for the Correction of Tubulopathy Due to Cystinosis

Silvia Espejel Carbajal, PhD (trainee)  
Holger Willenbring, MD (mentor)  
University of California, San Francisco