

# 14th International Hyperoxaluria Workshop

June 23rd –  
24th, 2023



Oxalosis &  
Hyperoxaluria  
Foundation

STEPPING STONES TO A CURE

Sala dei Notari, Perugia, Italy



## Conference Directors

Barbara Cellini, PhD University of Perugia



David Sas, DO Mayo Clinic

## Steering Committee

Yaacov Frishberg, MD  
Shaare Zedek Medical Center

Jaap Groothoff, MD  
Emma Children's Hospital

Peter C. Harris, PhD  
Mayo Clinic

Felix Knauf, MD  
Charité – University Medicine

John Knight, PhD  
University of Alabama at  
Birmingham

W. Todd Lowther, PhD  
Wake Forest School of Medicine

Shabbir Moochhala, MD  
Royal Free Hospital

Lama Nazzal, MD, MSc  
New York University

Gregory Tasian, MD, MSc, MSCE  
The Children's Hospital of  
Philadelphia

Jonathan Whittamore, PhD  
University of Texas Southwestern

## Champion Level



## Friend Level



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June 23rd – 24th, 2023 Perugia, Italy



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## Friday

23rd

8:00 Registration

8:30 Welcome

University of Perugia

8:35 Conference Director Welcome

David Sas, D.O. (Mayo Clinic, US) & Barbara Cellini,  
Ph.D. (University of Perugia, IT)

8:40 Keynote Address

Nicola Brunetti-Pierri, M.D. (Telethon Institute of  
Genetics and Medicine, IT)

9:00 Stepping Stones to a Cure - Leading the  
Way to End Hyperoxaluria

Kim Hollander (Oxalosis and Hyperoxaluria  
Foundation, US)

### Session One: Primary Hyperoxaluria Type 1

Chairs: Michelle Baum M.D, Jaap  
Groothoff M.D, Peter Harris Ph.D.

Sub-Session One: Clinical Update Presentations

Sub-Chairs: Jaap Groothoff, M.D., Michelle Baum,  
M.D.

9:15 Introduction

David Sas, D.O. (Mayo Clinic, US) & Barbara Cellini,  
Ph.D. (University of Perugia, IT)

9:20 Evaluation and Management in Europe—  
Where Are We Now

Sally Hulton, M.D. (Birmingham Women's and  
Children, UK)

9:30 Current State of PH1 Management in the US

Michael Somers, M.D. (Boston Children's Hospital,  
US)

9:40 Management of End-Stage Kidney Disease  
Before and After Transplantation

Barbara Ruggiero, M.D. (Bambino Gesù Pediatric  
Hospital, IT)

9:55 Current Challenges and Controversies in  
Transplant in PH1

Jaap Groothoff, M.D. (Emma Children's Hospital, ND)

10:05 Discussion on the Current Challenges of  
Treatments for PH1 Patients

Session Chairs

10:45 Break

Sub-Session Two: Research Presentations

Sub-Chair: Peter Harris, Ph.D.

10:55 Effect of Polymorphic and Pathogenic  
Mutations on the Structural Dynamics and  
the Fitness Window of AGT1

Mirco Dindo, Ph.D. (University of Perugia, IT)

11:00 Small-Molecule Approaches Toward  
Pharmacological Treatment for PH1

Monica Diaz-Gavilan, PhD. (University of Granada, SP)

11:10 Biomarkers and Isotope Studies on  
Endogenous Oxalate Production in PH1

Sander Garrelfs, M.D. (Amsterdam University Medical  
Centers, ND)

11:20 Preclinical Development of an In Vivo  
Lentiviral Vector Gene Therapy Approach  
for the Treatment of PH1

Andrea Molinos Vicente (CIEMAT/CIBERER-IIS/FJD,  
SP)

11:30 Generation of AGXT Gene-Corrected  
Induced-Hepatocytes with Restored  
Glyoxylate Metabolic Capacity from PH1  
Patient-Derived Fibroblasts

Virginia Nieto-Romero (CIEMAT/CIBERER-IIS/FJD, SP)

11:40 Summary & Discussion on Next Steps for  
Research in PH1

Session Chairs

Speed Round Poster Presentations

12:10 Evidence that the common AGXT variant  
p.Ile279Thr is pathogenic when associated  
with p.Pro11Leu

Peter Harris, Ph.D. (Mayo Clinic, US)

12:15 Role of SLC26A6 in oxalate crystal clearance  
by macrophages

Ana Clara Najenson, Ph.D. (Charité – Berlin University  
of Medicine, DE)

12:20 Poster Session

13:20 Lunch

## Session Two: Surgery, Genetics, and Ethics in Primary Hyperoxaluria

Chairs: John Lieske, M.D.,  
Shabbir Moochhala, M.D.

14:20 Introduction

David Sas, D.O. (Mayo Clinic, US) & Barbara Cellini,  
Ph.D. (University of Perugia, IT)

14:35 Surgical Challenges in PH and Surgical  
Innovations

Kyle Wood, M.D. (University of Alabama,  
Birmingham, US)

14:50 Challenges in Peri-Transplant Management  
in PH

Michael Somers, M.D. (Boston Children's Hospital,  
US), Sally Hulton, M.D. (Birmingham Women's and  
Children, UK) & Timucin Taner, M.D., Ph.D.  
(Mayo Clinic, US)

**Audience Polling and Panel Discussion**

15:10 Summary & Discussion on Surgical  
Challenges in PH

Session Chairs

15:40 Update on the Expected Prevalence of PH  
Types

Gill Rumsby, Ph.D. (London, UK)

15:50 Global PH Epidemiology and Genotype-  
Phenotype Correlation

Giorgia Mandrile, M.D. (San Luigi University Hospital,  
IT)

16:00 Break

16:10 Beyond AGXT, GRHPR, and HOGA1: Other  
Genetic Contributors to PH

Peter Harris, Ph.D. (Mayo Clinic, US)

16:20 Intrafamilial Phenotype Variability in PH

Lisa Deesker, M.D. (Amsterdam University Medical  
Centers, ND)

16:30 Discussion on the Genetics of PH and  
Unidentified Forms of PH

Session Chairs

17:00 Diagnosis and Management of PH in  
Pakistan

Imran Khan Jalbani, Ph.D. (Aga Khan University, PK)

17:10 PH Experience in China

Xiaojing Tang, M.D. (Shanghai Changzheng Hospital,  
CN)

17:20 Perspectives on PH in Africa

Neveen Soliman, M.D., Ph.D. (Kasr Al Ainy Medical  
School, Cairo University, EG)

17:35 Challenges in Diagnosis and Management  
of PH in the Middle East

Reham Almardn, M.D. (Princes Rahma Children  
Teaching Hospital, Applied Balqa University- Medical  
School, JO)

17:50 Discussion

Session Chairs

18:20 Patient Advocate Updates from Nepal

Govinda Regmi (Oxalosis and Hyperoxaluria  
Foundation Ambassador, NP)

18:30 Closing

David Sas, D.O. (Mayo Clinic, US) & Barbara Cellini,  
Ph.D. (University of Perugia, IT)



# Saturday

24th

8:00 Registration

8:30 Update from the Association of Patients of Hyperoxaluria in Spain

Ana Prado (Association of Patients of Hyperoxaluria in Spain, SP)

## Session Three: Primary Hyperoxaluria Type 2 and Primary Hyperoxaluria Type 3

Chairs: Sally Hulton, M.D., Yaacov Frishberg, M.D., John Knight, Ph.D., Todd Lowther, Ph.D.

Sub-Session One: Clinical Update Presentations

Sub-Chairs: Sally Hulton, M.D., Yaacov Frishberg, M.D.

8:40 Introduction

David Sas, D.O. (Mayo Clinic, US) & Barbara Cellini, Ph.D. (University of Perugia, IT)

8:45 Historic Perspectives

Dawn Milliner, M.D. (Mayo Clinic, US)

9:00 Diagnosis, Management and Outcome of PH2: Where Are We Now?

Sander Garrelfs, Ph.D. (Amsterdam University Medical Centers, ND)

9:15 Diagnosis, Management, and Outcome of PH3: Where Are We Now?

Efrat Ben-Shalom, M.D. (Shaare Zedek Medical Center, IL)

9:30 Clinical Summary and Discussion on PH2 and PH3 Management

Session Chairs

10:00 Break

Sub-Session Two: Research Presentations

Sub-Chairs: John Knight, Ph.D. & Todd Lowther, Ph.D.

10:15 Endogenous Oxalate Synthesis in PH: Unknown and Future Research

Sonia Fargue, Ph.D. (University of Alabama, Birmingham, US)

10:30 Hydroxyproline Metabolism in PH2 and PH3

John Knight, Ph.D. (University of Alabama, Birmingham, US)

10:45 Hydroxyproline Dehydrogenase Inhibition as a Therapeutic Strategy for Treating PH2 and PH3

Todd Lowther, Ph.D. (Wake Forest University School of Medicine, US)

11:00 HOGA1 Biochemical Properties and Effects of Pathogenic Mutations

Kerry M. Loomes, Ph.D. (University of Auckland, NZ)

11:15 Summary and Discussion on the Next Steps for Research in PH2 and PH3

Session Chairs

### Speed Round Poster Presentations

11:45 Stiripentol: mechanisms of action and perspectives in primary hyperoxaluria

Emmanuel Letavernier, M.D., Ph.D. (Sorbonne University/Tenon Hospital, FR)

11:50 Biochemical Diagnosis and Monitoring of Hyperoxaluria Syndromes: A Useful Tool for Prompt Decision Making

Marta Leporati (AO Ordine Mauriziano di Torino, IT)

11:55 Poster Session

12:55 Lunch

13:55 Patient Advocate Updates from Canada

Jennifer Adams, M.D. (Oxalosis and Hyperoxaluria Foundation Ambassador, CA)





## Session Four: Enteric Hyperoxaluria

Chairs: Felix Knauf, M.D., Greg Tasian M.D.,  
Jonathan Whittamore, Ph.D., Lama Nazzal, M.D.

### Sub-Session One: Clinical Update Presentations

Sub-Chairs: Felix Knauf, M.D. & Greg Tasian, M.D.

#### 14:05 Introduction

David Sas, D.O. (Mayo Clinic, US) & Barbara Cellini,  
Ph.D. (University of Perugia, IT)

#### 14:10 Mechanisms of Stone Formation in Enteric Hyperoxaluria

John Lieske, M.D. (Mayo Clinic, US)

#### 14:30 Management of Patients with Enteric Hyperoxaluria

Megan Prochaska, M.D. (The University of Chicago  
Medicine, US)

#### 14:50 Surgical Challenges in Hyperoxaluria

Kevin Koo, M.D. (Mayo Clinic, US)

#### 15:05 Clinical Summary and Discussion on the Challenges in EH Patient Management

Session Chairs

#### 15:35 Break

### Sub-Session Two: Research Presentations

Sub-Chairs: Jonathan Whittamore, Ph.D. & Lama  
Nazzal, M.D.

#### 15:50 Intestinal Oxalate Transport

Jonathan Whittamore, Ph.D. (University of Texas  
Southwestern, US)

#### 16:00 Role of the Microbiome in Oxalate Stone Disease

Lama Nazzal, M.D. (New York University Langone  
Health, US)

#### 16:10 Role of the Urinary Microbiome in Hyperoxaluria

Aaron Miller, Ph.D. (Cleveland Clinic, US)

#### 16:20 Hyperoxaluria and Microbes: Who Does What?

Luigina Romani (University of Perugia, IT)

#### 16:30 Update on the Enteric Hyperoxaluria Mouse Model

Lama Nazzal, M.D. (New York University Langone  
Health, US)

#### 16:40 Progress on the Application of Oxalate-Degrading Enzymes in PH

John Lieske, M.D. (Mayo Clinic, US)

#### 16:50 Role of SLC26A1– From Mouse to Man

Felix Knauf, M.D., Charité (University Medicine Berlin,  
DE)

#### 17:00 Research Summary and Discussion on the Next Steps for Research in EH

Session Chairs

#### 17:30 Closing

David Sas, D.O. (Mayo Clinic, US), Barbara Cellini,  
Ph.D. (University of Perugia, IT) & Kim Hollander  
(Oxalosis and Hyperoxaluria Foundation, US)

