

14th International Hyperoxaluria Workshop



Oxalosis &
Hyperoxaluria
Foundation

STEPPING STONES TO A CURE

June 23rd –
24th, 2023

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

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Charité – University Medicine

John Knight, PhD
University of Alabama at
Birmingham

W. Todd Lowther, PhD
Wake Forest School of Medicine

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Royal Free Hospital

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New York University

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The Children's Hospital of
Philadelphia

Jonathan Whittamore, PhD
University of Texas Southwestern

Champion Level



Friend Level



14th International Hyperoxaluria Workshop

June 23rd – 24th, 2023 Perugia, Italy



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STEPPING STONES TO A CURE

Friday

23rd

8:00 Registration

8:30 Welcome

Kim Hollander (Oxalosis and Hyperoxaluria Foundation, US)

8:35 Opening Remarks

Prof. Carla Emiliani, Delegate of the Dean & Prof. Vincenzo Nicola Talesa, Head of the Department of Medicine and Surgery (University of Perugia, IT)

8:40 Conference Director Welcome

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

8:45 Keynote Address

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

9:05 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 Grothoff M.D, Peter Harris Ph.D.

Sub-Session One: Clinical Update Presentations

Sub-Chairs: Jaap Grothoff, 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 Controversies in RNAi Therapy

Jaap Grothoff, 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 an
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 Almardnin, 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

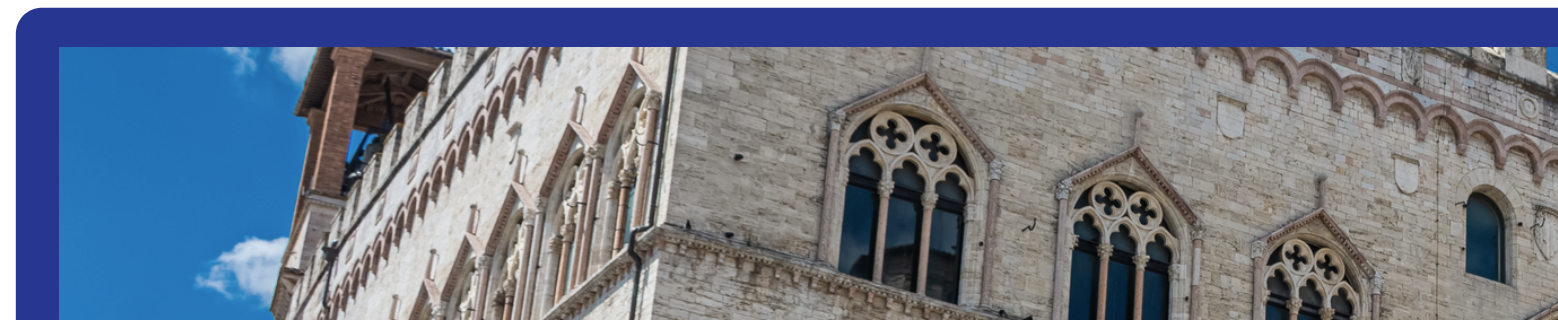
Marta Leporati (AO Ordine Mauriziano di Torino, IT)

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)

