14th International Hyperoxaluria Workshop
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

John Knight, PhD
University of Alabama at Birmingham

Jaap Groothoff, MD
Emma Children’s Hospital

W. Todd Lowther, PhD
Wake Forest School of Medicine

Peter C. Harris, PhD
Mayo Clinic

Shabbir Moochhala, MD
Royal Free Hospital

Felix Knauf, MD
Charité – University Medicine

Lama Nazzal, MD, MSc
New York University

Gregory Tesian, MD, MSc, MSCE
The Children’s Hospital of Philadelphia

Jonathan Whittamore, PhD
University of Texas Southwestern

Champion Level

Friend Level

Oxalosis & Hyperoxaluria Foundation | 579 Albany Post Road, New Paltz, NY 12561 | www.ohf.org
Friday

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

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 Gesu Pediatric Hospital, IT)

9:55 Controversies in RNAi Therapy
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 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)

June 23rd – 24th, 2023 Perugia, Italy
Saturday

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

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)