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

Jaap Groothoff, MD
Emma Children’s Hospital

Peter C. Harris, PhD
Mayo Clinic

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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

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)

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**Session One: Primary Hyperoxaluria Type 1**

**Sub-Session One: Clinical Update Presentations**

**Sub-Chairs:** Jaap Groorthooff, 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 Groorthooff, M.D. (Emma Children's Hospital, ND)

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**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 ACT1

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

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**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 HOGAT: 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 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)
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)
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

16:05 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

15:50 Intestinal Oxalate Transport
Jonathan Whittamore, Ph.D. (University of Texas Southwestern, US)