



Oxalosis &  
Hyperoxaluria  
Foundation

STEPPING STONES TO A CURE

# 14<sup>th</sup> International Hyperoxaluria Workshop

Conference Directors



**David Sas, DO**  
Mayo Clinic



**Barbara Cellini, PhD**  
University of Perugia

**Perugia**  
**June 23<sup>rd</sup>-24<sup>th</sup>**  
**2023**

Sala dei Notari



# 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



08.00 **Registration**

08.30 **Welcome**

*Kim Hollander (Oxalosis and Hyperoxaluria Foundation, US)*

08.35 **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)*

08.40 **Conference Director Welcome**

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

08.45 **Keynote Address**

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

09.05 **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 Groothoff M.D.

Peter Harris Ph.D.

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

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

09.15 **Introduction**

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

09.20 **Evaluation and Management in Europe - Where Are We Now**

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

09.30 **Current State of PH1 Management in the US**

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

09.40 **Management of End-Stage Kidney Disease Before and After Transplantation**

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

09.55 **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 AGTI**  
*Mirco Dindo, Ph.D. (University of Perugia, IT)*
- 11.00 **Small-Molecule Approaches Toward Pharmacological Treatment for PH1**  
*Monica Diaz-Gavilan, Ph.D. (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 Form 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 Almadnin, M.D. (Princess Rahama 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 24<sup>th</sup>

08.30 **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 AN 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.**

08.40 **Introduction**

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

08.45 **Historic Perspectives**

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

09.00 **Diagnosis, Management and Outcome of PH2: Where Are We Now?**

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

09.15 **Diagnosis, Management and Outcome of PH3: Where Are We Now?**

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

09.30 **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 Loporati (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)*

## CHAMPION LEVEL



## FRIEND LEVEL





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**ORGANISING SECRETARIAT**



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