


12th

INTERNATIONAL
PRIMARY
HYPEROXALURIA

WORKSHOP



Oxalosis &
Hyperoxaluria
Foundation

STEPPING STONES TO A CURE

12th International PH Workshop for Professionals, Patients and Families

July 14 – July 16, 2017

Hotel Las Águilas Conference Center
Puerto de la Cruz, Tenerife, Spain



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Hotel Las Águilas Conference Center

Calle Doctor Barajas, 19, 38400 Puerto de la Cruz, Tenerife, Spain.

Friday, July 14th, 2017

7:00 am **Continental Breakfast**
Conference Center

7:30 am **Registration**
Conference Center

Session One

8:00 am – 10:00 am

PH Mechanism of Disease

Session Chair: Eduardo Salido, MD, PhD

Welcome
Eduardo Salido, MD, PhD
Kim Hollander, OHF Executive Director

Past, Present & Future of PH
Christopher Danpure, PhD

PH Patient Experience Survey
George Tidmore
Josie Tidmore

Update on Oxalate Sources and Metabolism
Ross Holmes, PhD

Assessment of Oxalate Synthesis in PH Patients
Jaap Groothoff, MD, PhD

10:00 am – 10:30 am **Coffee Break/Posters**

10:30 am – 11:30 am Hydroxyproline Metabolism and Oxalate Synthesis on PH
Sonia Fargue MD, PhD

Urinary Oxalate and Glycolate Excretion of Mice Deficient in
Hydroxyproline Dehydrogenase or Glycolate Oxidase
Todd Lowther, PhD

Inflammatory Response to Crystal Deposits
Christian Kurts, MD

11:30 am – 11:40 am **Short Break/Posters**

Session Two

11:40 am – 12:40 pm

Current Approaches to Diagnosis and Treatment

Session Chair: Yaacov Frishberg, MD

Clinical Presentations
Bernd Hoppe, MD

Diagnosis update on PH
Gill Rumsby, PhD

An Update on PH2
Sally Hulton, MD

12:45 pm – 14:00 pm **Buffet Lunch – Conference Center**

14:00 pm – 15:45 pm Urine Proteome Analysis
Craig Langman, MD

Contemporary Lessons from Old Therapies, and Setting the Bar
for the New
Dawn Milliner, MD

Protein Translation Modulation
Ruth Belostosky, PhD

Dissecting the Glyoxylate and Oxalate Metabolic Pathways To
identify novel targets for PH treatment
Bob Brown, PhD, Dicerna Pharmaceuticals

Combined Liver Kidney Transplantation for PH2
John Lieske, MD

15:45 pm – 16:30 pm **Coffee Break**

16:30 pm – 17:20 pm **Poster Session (5 min. oral presentation, outlines)**

Clinical manifestations of systemic oxalosis in Primary
Hyperoxaluria type 1: when do which clinical manifestations
occur
Sanders Garrelfs, MD

Radiological findings (x-ray) in a group of 41 patients with
Primary Hyperoxaluria
Bernd Hoppe, MD

Genotype-Phenotype Correlations in Patients with Primary
Hyperoxaluria Type 3
Mary McIntosh

Descriptive Analysis of Gut Microbiome Alterations in
Hyperoxaluric Patients
Lama Nazzal, MD

Mutational Analysis of AGXT Gene in Libyan Children with Primary Hyperoxaluria type 1 at Tripoli Children Hospital
Naziha Rhuma, MD

Unravelling the Transcriptional Regulation of PH Associated Genes and Glyoxylate Metabolism Through Pathway Analysis
Florian Erger, MD

The NLRP3-Dependent Inflammasome Induces ILC3 Activation and Promotes Local Inflammation in the Kidney
Isis Ludwig-Portugall, PhD

Structural Modulation of Salicylates Reducing Oxalate Production in *Agxt1^{-/-}* Mouse Hepatocytes. Influence of the Linker
Mónica Díaz-Gavilán, PhD

Synthesis of New Potential Lactate Dehydrogenase (LDH) Inhibitors for Treating Primary Hyperoxalurias (PH)
Cristina Cuadrado, BSc

17:30 pm – 18:30 pm

Welcome Cocktail Reception for Professionals & Families
Conference Center

Hotel Las Águilas Conference Center

Saturday, July 15th, 2017

8:00 am

Continental Breakfast
Conference Center

Session Three

9:00 am – 10:20 am

Future Treatments

Session Chair: Marguerite Hatch, PhD

Pharmacological Chaperones Rescue AGT Folding Defects in PH1
Barbara Cellini, PhD

Current challenges in Gene Therapy
Gloria González-Asequinolaza, PhD

Preclinical Studies with Gene Editing
Juan R. Rodríguez-Madoz, PhD

siRNA Therapies
John Knight, PhD

10:20 am – 11:00 am

Coffee Break

11:00 am – 12:00 pm	<p>Small Molecule Modulators to Correct AGT Trafficking Carla Koehler, PhD</p> <p>A Probiotic Treatment for PH and Kidney Stone Disease Marguerite Hatch, PhD</p> <p>Novel Crystal Growth Inhibitors Jeff Wesson, MD PhD</p>
12:00 pm – 12:30 pm	<p>Poster session (5 min. oral presentation, outlines)</p> <p>Erythrocytes as Carriers of Oxalate decarboxylase: An Innovative approach for the Treatment of Hyperoxaluria. Carolina Conter, PhD</p> <p>New Strategies for Cell and Gene Therapy in Primary Hyperoxaluria Type 1 María García-Bravo, PhD</p> <p>Diversion Towards Non-Toxic Metabolites for Therapy of Primary Hyperoxaluria Type 1 Nicola Brunetti-Pierri, MD</p> <p>CRISPR/Cas9-Mediated <i>In Vivo</i> Substrate Reduction Therapy Rescues Primary Hyperoxaluria Type I Phenotype Nerea Zabaleta Lasarte, PhD</p> <p>The Patient Voice</p>
12:45 pm – 14:00 pm	Buffet Lunch – Conference Center
Session Four 14:00 pm – 16:00	<p>Industry Pipeline Session Chair: Dawn Milliner, MD</p> <p>Alnylam Clinical Studies Tracy McGregor MD, Alnylam Pharmaceuticals</p> <p>DCR-PHXC: An Optimal Treatment Approach for Primary Hyperoxaluria Doug Fambrough, PhD, Dicerna Pharmaceuticals</p> <p>OxaBact Clinical Studies Elisabeth Lindner, OxThera CEO</p> <p>Oral Enzyme Therapy Danica Grujic, PhD, Allena Pharmaceuticals</p>

Glycolate Oxidase Inhibitors
Miguel Fernandes, PhD, Orfan Biotech

Summary of 12th PH Workshop & Future 13th PH Workshop
Eduardo Salido, MD, PhD

16:00 pm - 17:00 pm **FDA White Paper**
John Lieske, MD

17:30 pm – 12:30 am
(+1day) **Volcano Teide and Dinner Tour**
Meet in Hotel Las Águilas Lobby for Transportation

Sunday, July 16th, 2017

9:30 am – 11:30 am Walk for Kidneys For Professionals & Families

Breakfast on Your Own

The OHF Walk for Kidneys is the rallying place for the OHF community where friends and family come together to raise funds and celebrate hope for the future for those affected by Primary Hyperoxaluria. Please join us for a fun, family-friendly walk.

Meeting point: Playa Martiánez, Puerto de la Cruz (in front of Fund Grube Shop) at 9:30 am. For those staying in Hotel Las Águilas, meet at the lobby to get in the bus at 9:00 am.

Abstracts selected for Poster

Structure Activity Study of New Inhibitors of Lactate Dehydrogenase (LDH) with a 2,8-Dioxabicyclo(3,3,1) Nonane Core
Alfonso Alejo Armijo, PhD

Dissecting the Glyoxylate and Oxalate Metabolic Pathways Using RNA Interference
Bob Brown, PhD

Pharmacological Chaperones rescue Folding Defects of Human Alanine: Glyoxylate Aminotransferase Leading to Primary Hyperoxaluria Type 1
Barbara Cellini, PhD

Erythrocytes as Carriers of Oxalate Decarboxylase: An Innovative Approach for the Treatment of Hyperoxaluria

Carolina Conter, PhD

Inhibition and Regulation of Lactate Dehydrogenase (LDH) by New Antioxidants with a 2,8-Dioxabicyclo (3,3,1) Nonane Core

Cristina Cuadrado García

Structural Modulation of Salicylates Reducing Oxalate Production in *Agxt1*^{-/-} Mouse Hepatocytes. Influence of the Linker

Mónica Díaz-Gavilán, PhD

Unraveling the Transcriptional Regulation of PH Associated Genes and Glyoxylate Metabolism through Pathway Analysis

Florian Erger, MD

DCR-PHXC: An Optimal Treatment Approach for Primary Hyperoxaluria

Doug Fambrough, PhD

Hydroxyproline Metabolism and Oxalate Synthesis in Primary Hyperoxaluria

Sonia Fargue, MD, PhD

New Strategies for Cell and Gene Therapy in Primary Hyperoxaluria Type 1

María García-Bravo, PhD

Clinical Manifestations of Systemic Oxalosis in Primary Hyperoxaluria Type 1: When do Which Clinical Manifestations Occur

Sander Garrelfs, MD

Clinical Characterization of Primary Hyperoxaluria by Using the OxalEurope Collaborative Database

Sander Garrelfs, MD

Structural Modulation of Salicylates Reducing Oxalate Production in *Agxt1*^{-/-} Mouse Hepatocytes. Influence of the Hydrophobic Moeity

José Antonio Gómez-Vidal, PhD

Development of a Novel Oral Enzyme Therapy for Hyperoxaluria: From Mice to Man

Danica Grujic, PhD

Radiological findings (Xray) in a Group of 41 Patients with Primary Hyperoxaluria

Bernd Hoppe, MD

PHYOS: An Observational Study of Patients with Primary Hyperoxaluria Type 1

Bernd Hoppe, PhD

Small Molecule Modulators to Correct AGT Trafficking from Mitochondria to Peroxisomes

Carla Koehler, PhD

Urinary Oxalate Excretion Predicts Kidney Stone Events in Enteric Hyperoxaluria

John Lieske, MD

Oxalosis Associated with High Dose Vitamin C Ingestion in a Peritoneal Dialysis Patient

John Lieske, MD

Combined Liver Kidney Transplantation for Primary Hyperoxaluria Type 2
John Lieske, MD

OxaBact® Clinical Studies to Date and Future Plans
Elisabeth Lindner, MSc MBA

Urinary Oxalate and Glycolate Excretion of Mice Deficient in Hydroxyproline Dehydrogenase or Glycolate Oxidase
Todd Lowther, PhD

The NLRP3-Dependent Inflammasome Induces ILC3 Activation and Promotes Local Inflammation in the Kidney
Isis Ludwig-Portugall, PhD

Diversion Towards Non-Toxic Metabolites for Therapy of Primary Hyperoxaluria Type 1
Nicola Brunetti-Pierri, PhD

Genotype-Phenotype Correlations in Patients with Primary Hyperoxaluria Type 3
Mary McIntosh

APRT Deficiency Misdiagnosis as Primary Hyperoxaluria in Patients with ESRD
Mary McIntosh

Synthesis and Biological Evaluation of Salicylate Derivatives as Efficient Agents Reducing Oxalate Production
María Dolores Moya-Garzón

Clinical Presentation and Mutations in AGXT, GRHPR and HOGA1 in Native and Migrant Pakistanis - A literature and PH Registry Survey
Saara (Ahmad) Muddasir Khan, PhD

Descriptive Analysis of Gut Microbiome Alterations in Hyperoxaluric Patients
Lama Nazzal, MD

Development of the Talati-Saqib Score Card to Alert Physicians to the Possibility of Primary Hyperoxaluria
Saqib Hamid Qazi, MD

Mutational Analysis of AGXT Gene in Libyan Children with Primary Hyperoxaluria Type 1 at Tripoli Children Hospital
Naziha Rhuma, MD

Cell Reprogramming for Disease Modelling of Primary Hyperoxaluria Type 1
Juan Roberto Rodríguez-Madoz, PhD

Safety of Long-Term Vitamin B6 in Primary Hyperoxaluria, Type 1
Hrafnhildur Runolfssdottir, MD

Virtual Screening of New Potential Lactate Dehydrogenase (LDH) Inhibitors with a 2,8-Dioxabicyclo(3,3,1)Nonane Scaffold
Sofía Salido, PhD

Preemptive Liver Transplantation Preserves Renal Function in Primary Hyperoxaluria Type 1
Hadas Shasha - Lavsky, MD

Primary Hyperoxaluria: Patient Experience Survey
George Tidmore and Josie Tidmore

CRISPR/Cas9-mediated *In Vivo* Substrate Reduction Therapy Rescues Primary Hyperoxaluria
Type I Phenotype
Nerea Zabaleta-Lasarte