

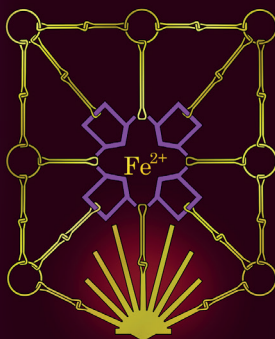
# INTERNATIONAL CONGRESS OF PORPHYRINS AND PORPHYRIAS

---

September 21–25, 2024

Pamplona (Spain)

ICPP 2024



**ipnet**  
INTERNATIONAL PORPHYRIA NETWORK

With the participation of:



Universidad  
de Navarra



Cima  
Universidad  
de Navarra

**ciber** | EHD  
CENTRO DE INVESTIGACIÓN BIOMÉDICA EN RED  
Enfermedades respiratorias y digestivas

**SATURDAY**  
September 21<sup>st</sup>



**CIMA BUILDING**

**15.00 - 18.30**

**Patient's Day**

Disease education  
(in Spanish)

**SUNDAY**  
September 22<sup>nd</sup>



**CIMA BUILDING**

**09.00 - 13.15**

**ICPP Patient and Doctor**

Workshop  
(in English).

**10.30 - 11.00**

**Coffee break**

**13.15 - 14.30**

**Lunch break**



**FACULTY OF SCIENCE  
BUILDING**

**13.30**

**Registration & Documentation  
delivery & Poster Hanging**

**15.00 - 15.15**

**Welcome ceremony and  
opening remarks ICPP**

**15.15 - 17.15**

**SESSION 1**

**Guidelines for  
treatment and follow-up**

**17.15**

**Welcome reception**

**19.00**

**Meet&Greet for young  
clinicians/scientists**

**MONDAY**  
September 23<sup>rd</sup>

**08.30 - 10.25**

**SESSION 2**

**Genetic and molecular  
mechanisms of heme  
synthesis and regulation**

**10.25 - 11.00**

**Coffee break and  
poster visit**

**11.00 - 13.15**

**SESSION 3**

**Heme Metabolism and  
Cellular Energy:**

**Inter-Organ Heme**

**Signaling and Import/Export  
Systems**

**13.15 - 14.30**

**Lunch and poster visit**

**14.30 - 15.00**

**Industry Symposium (\*)**

**15.00 - 16.55**

**SESSION 4**

**Heme in pathophysiology**

**16.55 - 17.25**

**Coffee break and  
poster visit**

**17.25 - 19.55**

**SESSION 5**

**IPNET general assembly**

**TUESDAY**  
September 24<sup>th</sup>

**08.30 - 10.05**

**SESSION 6**

**Current therapies for Acute  
Hepatic Porphyrias (AHP)**

**10.05 - 10.35**

**Industry Symposium (\*)**

**10.35 - 11.00**

**Coffee break and  
poster visit**

**11.00 - 11.30**

**Industry Symposium (\*)**

**11.30 - 13.10**

**SESSION 7**

**New insights and potential  
treatment approaches in  
erythropoietic Porphyrias**

**13.10 - 15.00**

**Lunch and poster visit**

**15.00 - 18.00**

**SESSION 8**

**Emerging therapies for  
Erythropoietic porphyrias**

**15.00 - 15.30**

**Industry**

**Symposium (\*)**

**Industry**

**Symposium (\*)**

**Industry**

**Symposium (\*)**

**Coffee break**

**Industry**

**Symposium (\*)**

**Industry**

**Symposium (\*)**

**20.00**

**Farewell Dinner**

**WEDNESDAY**  
September 25<sup>th</sup>

**08.30 - 10.30**

**SESSION 9**

**Rational approach(es) to  
diagnosis and care in AHPs**

**10.30 - 11.00**

**Coffee break and  
poster visit**

**11.00 - 12.20**

**SESSION 10**

**Rational approach(es) to  
diagnosis and care in  
Cutaneous Porphyrias**

**12.20 - 13.10**

**EXPERT PANEL**

**Diagnostic and Clinical  
conundrums. Critical issues to  
resolve and future directions**

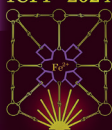
**13.10 - 13.50**

**Posters awards and Congress  
Closing ceremony**

(\*) See Section: Industry  
Sponsored Sessions

# INDEX

<b>DIRECTORS' GREETINGS</b> .....	1
<b>SCIENTIFIC COMMITTEE / LOCAL ORGANIZING COMMITTEE</b> .....	3
<b>FACULTY</b> .....	4
<b>PATIENT'S DAY</b> .....	6
<b>ICPP 2024 CONGRESS</b> .....	10
<b>SESSION 1</b> GUIDELINES FOR TREATMENT AND FOLLOW-UP .....	10
<b>SESSION 2</b> GENETIC AND MOLECULAR MECHANISMS OF HEME SYNTHESIS AND REGULATION .....	14
<b>SESSION 3</b> HEME METABOLISM AND CELLULAR ENERGY: INTER-ORGAN HEME SIGNALING AND IMPORT/EXPORT SYSTEMS.....	15
<b>SESSION 4</b> HEME IN PATHOPHYSIOLOGY .....	17
<b>SESSION 5</b> IPNET GENERAL ASSEMBLY .....	18
<b>SESSION 6</b> CURRENT THERAPIES FOR ACUTE HEPATIC PORPHYRIAS (AHPs) ....	19
<b>SESSION 7</b> NEW INSIGHTS AND POTENTIAL TREATMENT APPROACHES IN ERYTHROPOIETIC PORPHYRIAS .....	21
<b>SESSION 8</b> EMERGING THERAPIES FOR ERYTHROPOIETIC PORPHYRIAS .....	23
<b>SESSION 9</b> RATIONAL APPROACH(ES) TO DIAGNOSIS AND CARE IN AHPs .....	24
<b>SESSION 10</b> RATIONAL APPROACH(ES) TO DIAGNOSIS AND CARE IN CUTANEOUS PORPHYRIAS .....	28
<b>EXPERT PANEL:</b> DIAGNOSTIC AND CLINICAL CONUNDRUMS. CRITICAL ISSUES TO RESOLVE AND FUTURE DIRECTIONS .....	31
<b>INDUSTRY SPONSORED SESSION</b> .....	33
<b>POSTER SESSION</b> .....	41
<b>GENERAL INFORMATION</b> .....	66
<b>ACKNOWLEDGEMENTS</b> .....	68



Dear Colleagues, dear friends,

We are extremely honored to welcome you to The International Congress of Porphyrins and Porphyrias (ICPP 2024) which will take place on September 21-25, 2024 in Pamplona, Spain, bringing together clinicians, researchers, scholars, caregivers, patients and their families for three days networking and learning about the latest research in the porphyria field.

We are preparing an exciting program to learn about and discuss the latest advances in our understanding of the natural history, management and treatment of hepatic and erythropoietic porphyrias.

And, "by the way", once in Pamplona don't miss the opportunity to start walking the Way of Saint James, "El Camino"!

See you all in Pamplona!

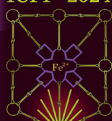
Best regards from,



**Sverre Sandberg**  
Ipnet President



**Antonio Fontanellas**  
President of ICPP 2024



## SCIENTIFIC COMMITTEE FOR ICPP 2024

---

Antonio Fontanellas	CIMA - Navarra University (Pamplona, Spain)
Bruce Wang	University of California (San Francisco, United States)
Jasmin Barman	Triemli City Hospital (Zurich, Switzerland)
Aasne A. Aarsand	Haukeland University Hospital (Bergen, Norway)
Makiko Yasuda	Icahn School of Medicine at Mount Sinai Hospital (New York, US)
Laurent Gouya	Hôpital Louis Mourier (Colombes, France)
Janneke G. Langendonk	Erasmus University Medical Center (Rotterdam, The Netherlands)
Sverre Sandberg	Norwegian Organization for Quality Improvement of Laboratory Examinations (Noklus), Haralds plass Deaconess Hospital (Bergen, Norway)

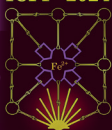
## LOCAL ORGANIZING COMMITTEE FOR ICPP 2024

---

Matias A. Ávila	CIMA-Navarra University (Pamplona, Spain)
Antonio Pineda	CIMA-Navarra University (Pamplona, Spain)
Jordi To-Figueras	University of Barcelona (Barcelona, Spain)
Oscar Millet	CIC bioGUNE (Derio, Spain)
Delia D'Ávola	Navarra University Clinic (Madrid, Spain)
Isabel Solares	Navarra University Clinic (Madrid, Spain)
Fidela Mirón	Spanish Federation of Rare Diseases (FEDER)
Rafael Enríquez de Salamanca	University Hospital 12 de Octubre (Madrid, Spain)
Paula Aguilera	Hospital Clinic, University of Barcelona (Barcelona, Spain)

September  
21-25, 2024

ICPP 2024



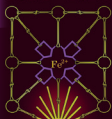
INTERNATIONAL  
CONGRESS OF  
PORPHYRINS AND  
PORPHYRIAS



## FACULTY

---

Aasne Aarsand	Haukeland University Hospital (Bergen, Norway)
Karl Anderson	University of Texas Medical Branch (Galveston, TX, US)
Matías A. Ávila	CIMA-Navarra University (Pamplona, Spain)
Michael Badminton	University Hospital of Wales Cardiff (Cardiff, UK)
Manisha Balwani	Icahn School of Medicine, Mount Sinai (New York, US)
Jasmin Barman	Triemli City Hospital (Zurich, Switzerland)
Sue Burrell	The British Porphyria Association
Maria Domenica Cappellini	University of Milano (Milano, Italy)
Delia D´Avola	Navarra University Clinic (Madrid, Spain)
Robert Desnick	Icahn School of Medicine, Mount Sinai Hospital (New York, US)
Jean Charles Deybach	Hôpital Louis Mourier (Paris, France)
Yonatan Edel	Ben-Gurion University in the Negev (Beer Sheva, Israel)
Veronica Fiorito	University of Turin (Torino, Italy)
Antonio Fontanellas	CIMA-Navarra University, (Pamplona, Spain)
Liz Gill	IPNET Patient Representative
Laurent Gouya	Hôpital Louis Mourier (Colombes, France)
Iqbal Hamza	University of Maryland (Baltimore, US)
Richard J. Hift	University of Kwazulu-Natal (Durban, South Africa)
Aneta Ivanova	St. Ivan Rilski University Hospital (Sofia, Bulgaria)
Louisa Kluijver	Erasmus Medical Center (Rotterdam, The Netherlands)
Eric Lai	Sloan-Kettering Institute (New York, US)
Janneke Langendonk	Erasmus University Medical Center (Rotterdam, The Netherlands)
Mattias Lissing	Karolinska University Hospital (Stockholm, Sweden)
Peter Meissner	University of Cape Town (Cape Town, South Africa)



## FACULTY

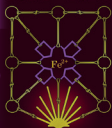
---

Óscar Millet	CIC bioGUNE (Derio, Spain)
Anna E. Minder	Zürich City Hospital (Zürich, Switzerland)
Fidela Mirón	Spanish porphyria assoc. (AEP) & rare diseases (FEDER)
Reem Mustafa	McMaster University (Hamilton, Canada)
Bindu D. Paul	Johns Hopkins University (Baltimore, US)
Petro Petrides	Ludwig Maximilians University (Munich, Germany)
John Phillips	University of Utah (Salt Lake City, US)
Patricia Renard	University of Namur (Namur, Belgium)
Eliane Sardh	Karolinska University Hospital (Stockholm, Sweden)
Penelope E. Stein	King's College Hospital (London, UK)
Miguel P. Soares	Instituto Gulbenkian de Ciência (Oeiras, Portugal)
Isabel Soares	Navarra University Clinic, (Madrid, Spain)
Mark Sonderup	University of Cape Town (Observatory, Cape Town, South Africa)
Ulrich Stölzel	Klinikum Chemnitz (Chemnitz, Germany)
Sverre Sandberg	Norwegian Organization for Quality Improvement of Laboratory Examinations (Noklus), Haraldsplass Deaconess Hospital (Bergen, Norway)
Jordi To-Figueras	University of Barcelona (Barcelona, Spain)
Emanuela Tolosano	University of Torino (Torino, Italy)
Iñaki F. Troconiz	Navarra University (Pamplona, Spain)
Paolo Ventura	University of Modena and Reggio Emilia (Modena, Italy)
Staffan Wahlin	Karolinska University Hospital (Stockholm, Sweden)
Bruce Wang	University of California (San Francisco, US)
Makiko Yasuda	Icahn School of Medicine, Mount Sinai Hospital (New York, US)
Jessica Zucman-Rossi	INSERM, University of Paris (Paris, France)



**SATURDAY**  
**September 21<sup>st</sup>**  
**2024**

ICPP 2024



INTERNATIONAL  
CONGRESS OF  
PORPHYRINS AND  
PORPHYRIAS



## PATIENT'S DAY

**CIMA**  
(Centro de Investigación  
Médica Aplicada)

Av. de Pío XII, 55  
31008 Pamplona, Navarra

[ASSEMBLY ROOM CIMA BUILDING]



15.00 - 18.30

### PATIENT'S DAY IN SPANISH

CHAIRS:

*Isabel Solares (Madrid, Spain)*

*Fidela Mirón (Spanish Porphyria Association)*

Disease education for patients in Spanish: interactive workshops for patients with porphyria.

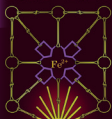
### 15.00 - 17.00 Taller de Emoción y Arte en porfiria

Sesión de restauración creativa: La técnica del collage convierte la incertidumbre en creatividad

*Nuria Adeva (Restauradora de arte)*

### 17.00 - 17.15 PAUSA CAFÉ





## PATIENT'S DAY

**17.15 - 17.40 Taller de Nutrición**

*Isabel Palazón (Nutricionista especializada en porfiria y cáncer)*

**17.40 - 18.05 Técnicas de Respiración y Relajación**

*Kika Shabot (Fundadora de la Asociación Mexicana para Porfiria)*

**18.05 - 18.30 Taller de Fitness**

Fortaleza y Movilidad

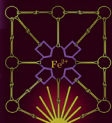
*Francisco Javier Plaza (Entrenador personal)*

Art exhibit with pieces provided by attendees as a creative way to raise awareness about the rare disease and the impact it has on patients and their families.

Exposición de arte con piezas aportadas por los asistentes, una iniciativa creativa para concienciar sobre las enfermedades raras y su impacto en los pacientes y sus familias.

SUNDAY  
September 22<sup>nd</sup>  
2024

ICPP 2024



INTERNATIONAL  
CONGRESS OF  
PORPHYRINS AND  
PORPHYRIAS



## PATIENT'S DAY

**CIMA**  
(Centro de Investigación  
Médica Aplicada)  
Av. de Pío XII, 55  
31008 Pamplona, Navarra

[ASSEMBLY ROOM CIMA BUILDING]



[Simultaneous Live video-Streaming]

09.00 - 13.15

### ICPP PATIENT AND DOCTOR WORKSHOP

#### CHAIRS:

*Joaquín Montoto* (Vice President, Asociación Española de Porfiria and Past Secretary, GPAC)

*Kika Shabot* (President, Sociedad Mexicana Para Porfiria and Vice President, GPAC)

09.00 - 09.10 **Welcome and overview of porphyria networks**  
**Mission of the porphyria advocacy community**

*Liz Gill*

*Kika Shabot*

09.10 - 10.30 **The science behind the porphyrias**

09.10 - 09.30 Genetics and genomic analysis: challenges and opportunities for diagnosis  
*Ana Patiño* (Navarra University Clinic, Pamplona, Spain)

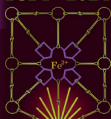
09.30 - 09.50 New therapeutic intervention lines for CEP  
*Oscar Millet* (CIC bioGUNE, Derio, Spain)

09.50 - 10.10 Recent advances and potential treatments on the horizon -  
Acute porphyrias  
*Delia D´Avola* (Navarra University Clinic, Madrid, Spain)

10.10 - 10.30 Emerging therapies in EPP  
*Paula Aguilera* (Hospital Clinic, University of Barcelona, Barcelona, Spain)

10.30 - 11.00 **COFFEE BREAK**





## PATIENT'S DAY

### [Simultaneous Live video-Streaming]

#### 11.00 - 11.20 **The Spanish and global porphyria community**

11.00 - 11.10 Spanish porphyria patient association

*Joaquín Montoto* (Vice President, Asociación Española de Porfiria and Past Secretary, GPAC)

11.10 - 11.20 Update from GPAC and the Global porphyria patient advocacy community

*Sue Burrell* (President, Global Porphyria Advocacy Coalition - GPAC)

#### 11.20 - 12.15 **Real patient and caregiver stories**

11.20 - 11.35 AIP patient perspective

*Eileen Hudson* (AIP), Fundación Porfiria Chile

11.35 - 11.50 AIP patient perspective

*Isabel Palazón* (AIP) Asociación Española de Porfiria

11.50 - 12.05 CEP patient perspective

*Arturo Fierro Monjaraz* (EPP) Sociedad Mexicana Para Porfiria

12.05 - 12.20 CEP patient perspective

*TBC* (CEP)

#### 12.20 - 13.00 **Ask the experts - patient and clinician discussion**

MODERATED BY CHAIRS:

Patients: *Eileen Hudson, Isabel Palazón and Arturo Fierro Monjaraz*

Clinicians: *Ana Patiño, Óscar Millet, Delia D'Ávola and Paula Aguilera*

#### 13.00 - 13.15 **Award presentation**

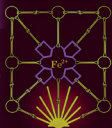
GPAC

#### 13.15 - 14.30 **LUNCH BREAK AND NETWORKING**



SUNDAY  
September 22<sup>nd</sup>  
2024

ICPP 2024



INTERNATIONAL  
CONGRESS OF  
PORPHYRINS AND  
PORPHYRIAS

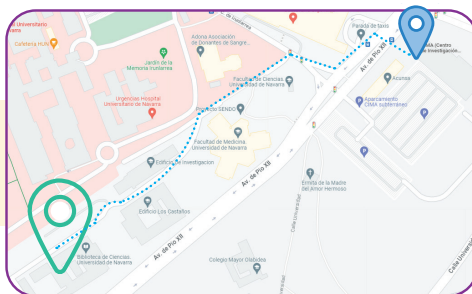
ipnet  
INTERNATIONAL PORPHYRIA NETWORK

## ICPP 2024 CONGRESS

FACULTY OF SCIENCE BUILDING  
(Universidad de Navarra)

C. de Irunlarrea, 1, 31008  
Pamplona, Navarra

[ASSEMBLY ROOM OF  
THE SCIENCE LIBRARY BUILDING]



13.30 REGISTRATION & DOCUMENTATION DELIVERY & POSTER HANGING

15.00 - 15.15 Welcome ceremony and opening remarks ICPP

CHAIRS:

*Sverre Sandberg* (Bergen, Norway)

*Antonio Fontanellas* (Pamplona, Spain)

15.15 - 17.15

SESSION 1 GUIDELINES FOR TREATMENT AND FOLLOW-UP

CHAIRS:

*Sverre Sandberg* (Bergen, Norway)

*Mark Sonderup* (Cape Town, South Africa)

15.15 - 15.55 Development of evidence-based guidelines for acute porphyrias

*Reem Mustafa* (Hamilton, Canada)

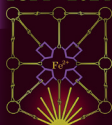
*Yonatan Edel* (Beer Sheva, Israel)

15.55 - 16.15 Basic needs in countries where there is a lack of specialist services

*Penelope E. Stein* (London, UK)

16.15 - 16.35 Long-term complications in acute porphyrias

*Mattias Lissing* (Stockholm, Sweden)



Selected abstracts presentation

**16.35 - 16.45 A PILOT STUDY IN 77 EPP PATIENTS OF THE LIVER FIBROSCAN (“TRANSIENT ELASTOGRAPHY”) TO IDENTIFY PATIENTS DEVELOPING FIBROTIC LIVER DISEASE. IDENTIFICATION OF OBESITY AS A RISK FACTOR**

[Robert P.E. Sarkany](#) (1) | [Oliver D. Tavabie](#) (2) | [Hiva Fassihi](#) (1) | [Adam Fit-yan](#) (1) | [Nisha Arujuna](#) (1) | [Harsha Naik](#) (1) | [John Ferguson](#) (1) | [Varuna Aluvihare](#) (2)

1- Photodermatology Unit, St John’s Institute of Dermatology, Guy’s Hos-  
pital, London, UK | 2- Institute of Liver Studies, King’s College Hospital,  
London, UK

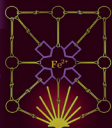
**16.45 - 16.55 LIVER INVOLVEMENT IN A LARGE COHORT OF PATIENTS WITH ERYTHROPOIETIC PROTOPORPHYRIA OR X-LINKED PROTOPORPHYRIA**

[Manisha Balwani](#) (1) | [Hetanshi Naik](#), PhD (2) | [Catherine McDonough](#), M.S (3) | [Mary Freeman](#), MS, CGC (4) | [Siobán B. Keel](#), MD (5) | [Angelika L. Erwin](#), MD, PhD (6) | [Amy K Dickey](#), MD, MS (7) | [Rebecca Karp-Leaf](#), MD (8) | [John Quigley](#) MD (9) | [Marshall Mazepa](#), MD (10) | [Bruce Wang](#), MD (11) | [John Phillips](#), PhD (12) | [Charles Parker](#), MD (12) | [Brendan McGuire](#) MD, MS (13) | [Mohamed Kazamel](#), MD (14) | [Herbert Bonkovsky](#), MD (15) | [Sean Rudnick](#), MD (16) | [Karl E. Anderson](#), MD (17) | [Akshata Moghe](#), MD, PhD (18) | [Manish Thapar](#), MD (19) | [Behnam Saberi](#) MD (20) | [Kristen Wheeden](#), MBA (21,21) | [Robert J Desnick](#) (4,4) | [Cynthia Levy](#), MD (22)

1- Mary Freeman, MS, CGC | 2- Department of Genetics, Stanford Univer-  
sity School of Medicine | 3- Center for Biostatistics, Department of Popu-  
lation Health Science and Policy, Icahn School of Medicine at Mount Sinai  
| 4- Department of Genetics and Genomic Sciences, Icahn School of Medi-  
cine at Mount Sinai, New York, NY | 5- Division of Hematology, University  
of Washington School of Medicine | 6- Center for Personalized Genetic  
Healthcare, Cleveland Clinic Community Care, Cleveland, OH | 7- Division  
of Pulmonary and Critical Care Medicine, Department of Medicine, Mas-  
sachusetts General Hospital, Boston, MA, Harvard Medical School, Boston,

**SUNDAY**  
**September 22<sup>nd</sup>**  
**2024**

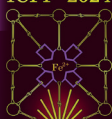
ICPP 2024



INTERNATIONAL  
CONGRESS OF  
PORPHYRINS AND  
PORPHYRIAS



MA | 8- Division of Hematology and Oncology, Department of Medicine, Massachusetts General Hospital, Boston, MA | 9- Division of Hematology/Oncology, Department of Medicine, University of Illinois at Chicago, IL | 10- Division of Hematology and Oncology, University of Minnesota Medical Center, Minneapolis, MN | 11- Department of Medicine, University of California San Francisco Medical Center, San Francisco, CA | 12- Division of Hematology and Hematologic Malignancies, Department of Internal Medicine at the University of Utah | 13- Division of Gastroenterology and Hepatology, University of Alabama at Birmingham, School of Medicine, Birmingham, AL | 14- Department of Neurology, University of Alabama at Birmingham, Birmingham, AL | 15- Department of Internal Medicine, Section on Gastroenterology and Hepatology, Atrium Health Wake Forest Baptist, Winston-Salem, NC. | 16- Department of Internal Medicine, Section on Gastroenterology and Hepatology, Atrium Health Wake Forest Baptist, Winston-Salem, NC | 17- Department of Internal Medicine (Division of Gastroenterology & Hepatology), University of Texas Medical Branch/UTMB Health, Galveston, TX | 18- Division of Gastroenterology, Hepatology, and Nutrition, University of Texas Health Science Center at Houston, TX | 19- Division of Hepatology, Einstein Healthcare Network, Philadelphia, Pennsylvania, United States. | 20- Division of Gastroenterology, Beth Israel Deaconess Medical Center, Boston, MA | 21- United Porphyrins Association | 22- Division of Digestive Health and Liver Diseases, University of Miami Miller School of Medicine, Miami, FL



**16.55 - 17.05 PATIENT DEMOGRAPHICS AND CLINICAL CHARACTERISTICS AT ENROLMENT IN ELEVATE, AN INTERNATIONAL REGISTRY OF ACUTE HEPATIC PORPHYRIA**

[Eliane Sardh](#) (1) | [David Cassiman](#) (2) | [Laurent Gouya](#) (3) | [Bruce Wang](#) (4) | [Weiming Du](#) (5) | [Teresa L Kauf](#) (5) | [Jamie L Weiss](#) (5) | [Manisha Balwani](#) (6)

1- Porphyria Centre Sweden, Centre for Inherited Metabolic Diseases, Karolinska Institutet, Karolinska University Hospital, Stockholm, Sweden | 2- Department of Gastroenterology-Hepatology and Center for Metabolic Diseases, University Hospital Leuven, Leuven, Belgium | 3- Centre Français des Porphyries, Paris, France | 4- University of California, San Francisco, CA, USA | 5- Alnylam Pharmaceuticals, Cambridge, MA, USA | 6- Department of Genetics and Genomic Sciences, Icahn School of Medicine at Mount Sinai, New York, NY, USA

**17.05 - 17.15 USE OF HORMONAL CONTRACEPTIVES AND MENOPAUSAL REPLACEMENT THERAPY IN WOMEN WITH ACUTE PORPHYRIA**

[Linda Eirin Gilleshammer](#) (1) | [Heidi Netland Jensen](#) (1,2) | [Reidun Lisbet Skeide Kjome](#) (2) | [Aasne Karine Aarsand](#) (1,3)

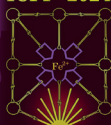
1- Norwegian Porphyria Centre (NAPOS), Department for Medical Biochemistry and Pharmacology, Haukeland University Hospital, Bergen, Norway | 2- Dept of Global Public Health and Primary Care, Centre for Pharmacy, University of Bergen, Bergen, Norway | 3- Norwegian Organization for Quality Improvement of Laboratory Examinations, Haraldsplass Deaconess Hospital, Bergen, Norway

17.15 Welcome reception

19.00 **Meet&Greet for young clinicians/scientists**

[Louisa Kluijver](#) (Rotterdam, The Netherlands)





08.30 - 10.25

**SESSION 2 GENETIC AND MOLECULAR MECHANISMS OF HEME SYNTHESIS AND REGULATION**

CHAIRS:

*John Phillips (Utah, US)*

*Peter Meissner (Cape Town, South Africa)*

08.30 - 09.15 Heme synthesis and succinate in the regulation of pluripotent states transitions

*Patricia Renard (Namur, Belgium)*

09.15 - 09.35 Computational approaches for mapping heme biology

*Iñaki F. Troconiz (Pamplona, Spain)*

09.35 - 09.55 Heme: the missing link in the pathogenesis of severe malaria

*Miguel P. Soares (Oeiras, Portugal)*

Selected abstracts presentation

09.55 - 10.05 **ACUTE INTERMITTENT PORPHYRIA ONSET: BIOINFORMATIC AND GENETIC ANALYSIS OF PORPHYRINOGENIC XENOBIOTICS AND DRUGS TRANSPORTER SYSTEM**

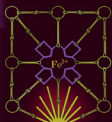
[Priscila Ayelén Pagnotta \(1,2\)](#) | [Viviana Alicia Melito \(1,2\)](#) | [Johanna Romina Zuccoli \(2\)](#) | [Victoria Estela Parera \(2\)](#) | [Ana María Buzaleh \(2\)](#)

1- Dpto. de Química Biológica - Facultad de Ciencias Exactas y Naturales - Universidad de Buenos Aires, Argentina | 2- Centro de Investigaciones sobre Porfirinas y Porfirias (CIPYP), UBA-CONICET, Buenos Aires, Argentina

10.05 - 10.15 **SHAPES AND PATTERNS OF HEME-BINDING MOTIFS IN MAMMALIAN HEME-BINDING PROTEINS**

[Dhruv C. Rathod \(1\)](#) | [Sonali M. Vaidya \(1\)](#) | [Marie-T. Hopp \(2\)](#) | [Toni Kühl \(1\)](#) | [Diana Imhof \(1\)](#)

1- Department of Pharmaceutical Biochemistry and Bioanalytics, Pharmaceutical Institute, University of Bonn, D-53121 Bonn, Germany. | 2- Department of Chemistry, Institute for Integrated Natural Sciences, University of Koblenz, D-56070 Koblenz, Germany



**10.15 - 10.25 GENE EDITING TO MIMIC PORPHYRIA WITH LIGHT: A PROMISING APPROACH TO TREAT PROSTATE CANCER**

Julian Boutin (1) | Jean-Marc Blouin (1) | Emmanuel Richard (1) | Sandrine Dabernat (1) | Aurélie Bedel (1) | [François Moreau-Gaudry \(1\)](#)

1- University of Bordeaux, Inserm, UMR1312, BRIC, BoRdeaux Institute of onCology, 146 Rue Léo Saignat, Bordeaux, F-33076, France

10.25 - 11.00 **Coffee break and poster visit**



11.00 - 13.15

**SESSION 3 HEME METABOLISM AND CELLULAR ENERGY:  
INTER-ORGAN HEME SIGNALING AND IMPORT/EXPORT SYSTEMS**

CHAIRS:

*Bruce Wang (San Francisco, US)*

*Makiko Yasuda (New York, US)*

11.00 - 11.45 Inter-organ heme signaling

*Iqbal Hamza (Baltimore, US)*

11.45 - 12.05 Impact of Dysregulated Heme Synthesis on Metabolism in Liver and Extrahepatic Tissues

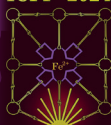
*Emanuela Tolosano (Torino, Italia)*

12.05 - 12.25 Unraveling transcriptomic and metabolomic insights into TCA Cycle and mitochondrial activity in the liver of AIP model in NHPs.

*Antonio Fontanellas (Pamplona, Spain)*

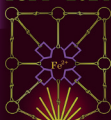
12.25 - 12.45 Non-canonical role of ALAS1 as an inhibitor of RNA interference

*Eric Lai (New York, US)*



Selected abstracts presentation

- 12.45 - 12.55 MYOCARDIAL FUNCTION IN PATIENTS WITH ACUTE INTERMITTENT PORPHYRIA - PRELIMINARY RESULTS OF AIPRACUS STUDY**  
[Krzysztof Jaworski](#) (1) | [Robert Wasilewski](#) (2) | [Jerzy Windyga](#) (2,3) | [Ilona Kowalik](#) (4) | [Ag-nieszka Lipniacka](#) (3) | [Hanna Szwed](#) (1) | [Rafal Dabrowski](#) (1)  
1- Department of Coronary Artery Disease and Cardiac Rehabilitation, National Institute of Cardiology, Warsaw, Poland | 2- Department of Hemostasis Disorders and Internal Medicine, Institute of Hematology and Transfusion Medicine, Warsaw, Poland | 3- Laboratory of Genetics of Hemostasis and Porphyria, Institute of Hematology and Transfusion Medicine, Warsaw, Poland | 4- Clinical Research Support Center, National Institute of Cardiology, Warsaw, Poland
- 12.55 - 13.05 ENHANCED ETB-DEPENDENT CONTRACTION MEDIATES INCREASED ET-1 RESPONSES IN MESENTERIC ARTERIES IN THE AIP MOUSE MODEL**  
[Victor M. Pulgar](#) (1,2) | [Makiko Yasuda](#) (3) | [Lin Gan](#) (3) | [Robert J. Desnick](#) (3) | [Herbert L. Bonkovsky](#) (4)  
1- Department of Pharmaceutical & Clinical Sciences, Campbell University, Buies Creek, NC, USA | 2- Department of Obstetrics & Gynecology, Wake Forest University School of Medicine, Winston-Salem, NC, USASA | 3- Icahn School of Medicine at Mount Sinai, NY, USA. | 4- Wake Forest University School of Medicine, Winston-Salem, NC, USASA
- 13.05 - 13.15 ADRENAL INSUFFICIENCY ASSOCIATED WITH MUTATIONS IN HAEM BIOSYNTHESIS GENES**  
[Ahmed Al-Salihi](#) (1) | [Chris J Smith](#) (1) | [Adam Jackson](#) (2) | [Andreas Janneck](#) (3) | [Elisabeth Steichen](#) (3) | [Denise Darby](#) (2) | [Liesel Griffin](#) (4) | [Siddharth Banka](#) (2) | [Solaf Elsayed](#) (5) | [Neila Talbi](#) (6) | [Laurent Gouya](#) (6) | [Brigitte Delemer](#) (7) | [Li F Chan](#) (1) | [Louise A Metherell](#) (1)  
1- Centre for Endocrinology, WHRI, QMUL, London, UK | 2- University of Manchester, Manchester, UK | 3- University Hospital of Innsbruck, Innsbruck, Austria | 4- Dermatology Centre, Salford Royal Hospital, University of Manchester | 5- Medical Genetics Department, Ain Shams University, Cairo, Egypt | 6- The National Institute of Health and Medical Research (INSERM), Paris, France | 7- Centre Hospitalier Universitaire De Reims, Reims, France



13.15 - 14.30 *Lunch and poster visit*



14.30 - 15.00 **Industry Symposium (\*)**

15.00 - 16.55

**SESSION 4 HEME IN PATHOPHYSIOLOGY**

**CHAIRS:**

*Staffan Wahlin (Stockholm, Sweden)*

*Matías Ávila (Pamplona, Spain)*

15.00 - 15.45 Molecular mechanisms in hepatocarcinogenesis

*Jessica Zucman-Rossi (Paris, France)*

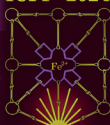
15.45 - 16.05 Inhibiting Heme Synthesis to Modulate Cell Metabolism and Counteract Tumor Initiation and Progression

*Veronica Fiorito (Torino, Italia)*

16.05 - 16.25 Role of the bilirubin/biliverdin reductase axis in neuroprotection

*Bindu D. Paul (Baltimore, US)*

(\*) See Section: Industry Sponsored Sessions



Selected abstracts presentation

**16.25 - 16.35 HEME-DRIVEN PATHOLOGIES IN THE CONTEXT OF INFECTIONS:  
FROM TLR4 SIGNALING TO COVID-19**

[Diana Imhof](#) (1)

1- Pharmaceutical Biochemistry and Bioanalytics, Pharmaceutical Institute, University of Bonn, Bonn, Germany

**16.35 - 16.45 CHARACTERIZATION OF HEME-INDUCED TOXICITY AND  
FERROPTOSIS IN PLACENTAL CELLS**

[Michelle Bedran](#) (1) | [Nicolas Ducrot](#) (1) | [Valeria Fiorentino](#) (1) | [Caroline Schmitt](#) (1,2) | [Laurent Gouya](#) (1,2) | [Katell Peoc'h](#) (1,3) | [Hana Manceau](#) (1,3)

1- Université Paris Cité, Centre de Recherche sur l'Inflammation and INSERM UMR1149, CNRS, ERL 8252, UFR de Médecine Bichat, Paris, France and Laboratory of Excellence GR-Ex | 2- Centre Français des Porphyrines, Hôpital Louis Mourier, Colombes, France | 3- Assistance Publique-Hôpitaux de Paris, Laboratoire de Biochimie Clinique, APHP.Nord, Hôpital Beaujon, Clichy, France

**16.45 - 16.55 ROLE OF HEME AS A MODULATOR OF BLOOD COAGULATION  
PROTEINS**

[Marie-T. Hopp](#) (1)

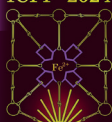
1- Bioorganic Chemistry, Department of Chemistry, Institute for Integrated Natural Sciences, University of Koblenz

**16.55 - 17.25 Coffee break and poster visit**



**17.25 - 19.55**

**SESSION 5 IPNET GENERAL ASSEMBLY**



08.30 - 10.05

**SESSION 6 CURRENT THERAPIES FOR ACUTE HEPATIC PORPHYRIAS (AHPs)**

CHAIRS:

*Paolo Ventura (Modena, Italy)*

*Robert Desnick (New York, US)*

**08.30 - 09.15** Use of Givosiran in acute hepatic porphyrias: World experience

*Eliane Sardh (Stockholm, Sweden)*

*Petro Petrides (Munich, Germany)*

**09:15 - 09.35** Natural history of AHP in US patients

*Bruce Wang (San Francisco, US)*

Selected abstracts presentation

**09.35 - 09.45** **LONG-TERM CLINICAL OUTCOMES OF PATIENTS WITH ACUTE HEPATIC PORPHYRIA WHO WERE NOT ATTACK-FREE AFTER 6 MONTHS OF GIVOSIRAN TREATMENT: A SUBGROUP ANALYSIS OF THE PHASE 3 ENVISION STUDY**

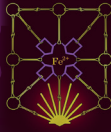
*Paolo Ventura (1) | Encarna Guillen-Navarro (2,3) | Bruce Wang (4) | Weiming Du (5) | Ana Camejo (5) | Manish Thapar (6)*

1- Internal Medicine Unit, University of Modena and Reggio Emilia, Modena, Italy | 2- Medical Genetics Section, Virgen de la Arrixaca University Hospital, IMIB Pascual Parrilla, University of Murcia (UMU), Murcia, Spain | 3- CIBERER-ISCIII, Madrid, Spain | 4- UCSF Health, San Francisco, United States | 5- Alnylam Pharmaceuticals, Cambridge, United States | 6- Thomas Jefferson University, Philadelphia, United States

**09.45 - 09.55** **SINGLE-CENTER REAL-WORLD EXPERIENCE WITH GIVOSIRAN FOR ACUTE INTERMITTENT PORPHYRIA**

*Chanan Stauffer (1) | Luca Fierro (1) | Rachel Evard (1) | Manisha Balwani (1)*

1- Icahn School of Medicine at Mount Sinai



**09.55 - 10.05 TAPERING AND DISCONTINUING GIVOSIRAN TREATMENT IN PREVIOUSLY RECURRENT ACUTE INTERMITTENT PORPHYRIA PATIENTS: AN INTERNATIONAL MULTICENTER CASE SERIES**

[Niels Veldhoen](#) (1) | [Edith Friesema](#) (1) | [Sophie Mijnhout](#) (2) | [Eliane Sardh](#) (3) | [Janneke Langendonk](#) (1)

1- Porphyria Centre Rotterdam, Center for Lysosomal and Metabolic Disease, Department of Internal Medicine, Erasmus MC, University Medical Center Rotterdam, The Netherlands. | 2- Isala Porphyria Centre, Department of Internal Medicine, Isala Clinics, Zwolle, The Netherlands. | 3- Porphyria Centre Sweden, Centre for Inherited Metabolic Diseases, Karolinska Institutet, Karolinska University Hospital, Stockholm, Sweden

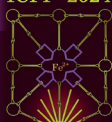
**10.05 - 10.35 Industry Symposium (\*)**

**10.35 - 11.00 Coffee break and poster visit**



**11.00 - 11.30 Industry Symposium (\*)**

(\*) See Section: Industry Sponsored Sessions



11.30 - 13.10

**SESSION 7**

**NEW INSIGHTS AND POTENTIAL TREATMENT  
APPROACHES IN ERYTHROPOIETIC PORPHYRIAS**

**CHAIRS:**

*Janneke Langendonk (Rotterdam, The Netherlands)*

*Ulrich Stölzel (Chemnitz, Germany)*

11.30 - 11.50 Iron metabolism in EPP

*Jasmin Barman (Zurich, Switzerland)*

11.50 - 12.10 Predicting and Improving Osteoporosis in EPP

*Louisa Kluijver (Rotterdam, The Netherlands)*

12.10 - 12.30 Innovative therapies in congenital erythropoietic porphyria (CEP)

*Oscar Millet (Derio, Spain)*

Selected abstracts presentation

12.30 - 12.40 **DRUG REPURPOSING IN ERYTHROPOEITIC PORPHYRIAS:  
HIGH-THROUGHPUT SCREENING FOLLOWED BY HIT-TO LEAD  
STRATEGY TO IDENTIFY ALAS2 INHIBITORS**

Claire Guibet (1,2) | Sarah Terrisse (1) | Magalie Lalanne (1) | Patrick Mercié (1,3) |  
Joris Guyon (1,2) | Jean-Marc Blouin (1,2) | Emmanuel Richard (1,2)

1- University of Bordeaux, INSERM U1312, Bordeaux Research Institute of  
Oncology, Bordeaux, France. | 2- Porphyria Expert Laboratory and Clinical  
Center, Department of Bio-chemistry, CHU de Bordeaux | 3- Porphyria  
Expert Laboratory and Clinical Center, Department of Internal Medicine  
and Clinical Immunology, CHU de Bordeaux

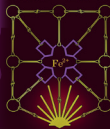
12.40 - 12.50 **RESULTS OF THE AURORA STUDY: A PHASE 2, RANDOMIZED,  
DOUBLE-BLIND, PLACEBO-CONTROLLED STUDY OF BITOPERTIN IN  
ERYTHROPOIETIC PROTOPORPHYRIA**

Amy Dickey, MD, MSc (1) | Sioban Keel, MD (2) | Herbert L. Bonkovsky, MD  
(3) | Karl E. Anderson, MD (4) | Manisha Balwani, MD (5) | Cynthia Levy,  
MD (6) | Manish Thapar, MD (7) | Bruce Wang, MD (8) | Brendan M. McGuire,  
MD (9) | Will Savage, MD, PhD (10)



**TUESDAY**  
**September 24<sup>th</sup>**  
**2024**

ICPP 2024



INTERNATIONAL  
CONGRESS OF  
PORPHYRINS AND  
PORPHYRIAS



1- Harvard Medical School and Massachusetts General Hospital, Boston, MA | 2- University of Washington, Seattle, WA | 3- Wake Forest University School of Medicine and Atrium Health Wake Forest Baptist, Winston-Salem, NC | 4- University of Texas Medical Branch, Galveston, TX | 5- Icahn School of Medicine at Mount Sinai, New York, NY | 6- University of Miami Miller School of Medicine, Miami, FL | 7- Jefferson Center for Genetic and Metabolic Liver Disease, Philadelphia, PA | 8- University of California San Francisco Porphyria Center, San Francisco, CA | 9- University of Alabama at Birmingham, Birmingham, AL | 10- Disc Medicine, Watertown, MA

**12.50 - 13.00 RESULTS FROM THE BEACON TRIAL: A PHASE 2, RANDOMIZED, OPEN-LABEL TRIAL OF BITOPERTIN IN ERYTHROPOIETIC PROTOPORPHYRIA**

[Gayle Ross](#), MD (1) | [Peter Stewart](#), MD (2) | [George Mensing](#), MPH (3) | [Melanie Chin](#), PhD (3) | [Haley Howell](#) (3) | [Heidi Mangus](#) (3) | [Will Savage](#), MD, PhD (3)

1- Royal Melbourne Hospital, Melbourne, Australia | 2- Royal Prince Alfred Hospital, Sydney, Australia | 3- Disc Medicine, Watertown, MA

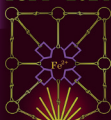
**13.00 - 13.10 PHLEBOTOMY AND AFAMELANOTIDE AS AN EFFICIENT COMBINED TREATMENT OF HEPATOERYTHROPOIETIC PORPHYRIA**

[Luca Barbieri](#) (1) | [Michele De Canio](#) (1) | [Caterina Aurizi](#) (1) | [Miriam Teoli](#) (1)

1- San Gallicano Dermatological Institute, IRCCS, Rome, Italy

**13.10 - 15.00 Lunch and poster visit**





15.00 - 18.00

**SESSION 8** **INDUSTRY SYMPOSIUMS**

15.00 - 15.30 Industry Symposium (\*)

15.30 - 16.00 Industry Symposium (\*)

16.00 - 16.30 Industry Symposium (\*)

16.30 - 17.00 *Coffee break*

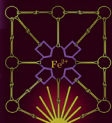


17.00 - 17.30 Industry Symposium (\*)

17.30 - 18.00 Industry Symposium (\*)

**20.00** **FAREWELL DINNER**

(\*) See Section: Industry Sponsored Sessions



08.30 - 10.30

**SESSION 9 RATIONAL APPROACH(ES) TO DIAGNOSIS AND CARE IN AHPs**

**CHAIRS:**

*Aneta Ivanova (Sofia, Bulgaria)*

*Jordi To-Figueras (Barcelona, Spain)*

08.30 - 08.50 Laboratory diagnosis of porphyrias - challenges and potential solutions

*Aasne Aarsand (Bergen, Norway)*

08.50 - 09.10 A modern approach to the determination of drug safety in acute porphyria

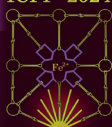
*Richard J. Hift (Durban, South Africa)*

**Selected abstracts presentation**

09.10 - 09.20 **FUNCTIONAL CHANGES IN THE CENTRAL NERVOUS SYSTEM IN A NON-HUMAN PRIMATE MODEL OF AIP CAN BE FULLY RESCUED BY SYSTEMIC MESSENGER RNA REPLACEMENT THERAPY**

[Daniel Jericó](#) (1) | [María Collantes](#) (2,3) | [Lei Jiang](#) (4) | [Karol M. Córdoba](#) (1) | [Francesco Urigo](#) (1) | [Ana Sampedro](#) (1) | [Alberto Rico](#) (5) | [Antonio Martínez de la Cuesta](#) (6) | [Oscar Manzanilla](#) (3,7) | [Estibaliz Alegre](#) (3,8) | [Gemma Quincoces](#) (2,3) | [Jose Luiz Lanciego](#) (5) | [Iván Peñuelas](#) (2,3) | [Paolo G.V. Martini](#) (4) | [Matías A. Ávila](#) (1,3,9) | [Antonio Fontanellas](#) (1,3,9)

1- Hepatology: Porphyrias&Carcinogenesis Lab. Solid Tumors Program, CIMA-University of Navarra (UNAV), Pamplona & CIBERehd, Spain | 2- Translational Molecular Imaging Unit (UNIMTRA), UNAV & Nuclear Medicine-Department, Clínica Universidad de Navarra (CUN), Pamplona, Spain | 3- Instituto de Investigación Sanitaria de Navarra (IdisNA), Pamplona, Spain | 4- Moderna Inc, Cambridge MA, USA | 5- Neurosciences Department, CIMA-University of Navarra, Pamplona & CIBERNed, Spain | 6- Radiology Department, CUN, Pamplona, Spain | 7- Department of Clinical Neurophysiology, CUN, Pamplona, Spain | 8- Service of Biochemistry, CUN, Pamplona, Spain | 9- Cancer Center CUN, Pamplona, Spain



**09.20 - 09.30 POTENTIAL CAUSES OF DYSREGULATED HOMOCYSTEINE METABOLISM IN PATIENTS AFFECTED BY ACUTE INTERMITTENT PORPHYRIA**

[Elena Di Piero](#) (1) | [Lorena Duca](#) (1) | [Francesca Granata](#) (1) | [Francesca Grande](#) (2) | [Valeria Di Stefano](#) (1) | [Giacomo De Luca](#) (3) | [Paolo Ventura](#) (4) | [Giovanna Graziadei](#) (1)

1- Medicine and Metabolic Diseases Unit, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy | 2- Università di Pavia, Italy | 3- Intermediate Care Unit, Ospedale Guglielmo da Saliceto, Piacenza, Italy | 4- Azienda Ospedaliero-Universitaria di Modena-Policlinico, Modena, Italy

**09.30 - 09.40 THE PREVALENCE AND PENETRANCE OF ACUTE INTERMITTENT PORPHYRIA IN THE MASS GENERAL BRIGHAM BIOBANK**

[Amy Dickey](#) (1) | [Lina Rebeiz](#) (1) | [Rebecca K. Leaf](#) (1) | [Emma Perez](#) (1) | [Benny Tran](#) (1) | [Elin Storjord](#) (2) | [Padmini Pillai](#) (3) | [Narmene Bensaber](#) (1) | [Hana Zouk](#) (1) | [Jordan Smoller](#) (1) | [David Christiani](#) (1) | [Karl Anderson](#) (4) | [Nina Gold](#) (1)

1- Massachusetts General Hospital | 2- Nordland Hospital, Norway | 3- Massachusetts Institute of Technology | 4- University of Texas Medical Branch

**09.40 - 09.50 EMBRYO SELECTION IN THE ACUTE HEPATIC PORPHYRIAS: CLINICAL, ETHICAL AND ECONOMIC ISSUES**

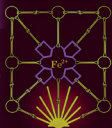
[Cindy Towns](#) (1,2) | [Gisela Kristono](#) (1) | [Leigh Searle](#) (2)

1- University of Otago, New Zealand | 2- Wellington Hospital, New Zealand

**09.50 - 10.00 PREVENTION OF CYCLICAL ATTACKS WITH GNRH-AGONISTS AND HORMONAL CONTRACEPTIVES AMONG PATIENTS WITH ACUTE PORPHYRIA**

[Katrin Baumann](#) (1,2) | [Elena Pischik](#) (3) | [Raili Kauppinen](#) (2)

1- Department of Gynecology and Obstetrics, Helsinki University Central Hospital, Helsinki, Finland. | 2- Department of Medicine, Helsinki University Central Hospital, Helsinki, Finland. | 3- Department of Neurology, Consultative and Diagnostic Centre with Polyclinics, St. Petersburg, Russia



**10.00 - 10.10 REDUCED EXPRESSION OF PPOX, THE VARIEGATE PORPHYRIA GENE, CAUSES INTRINSIC PATHOLOGICAL CHANGES TO KERATINOCYTES**

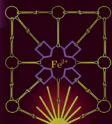
[Dr Chris J Smith](#) (1) | [Prof Li F Chan](#) (1) | [Prof Lou A Metherell](#) (1)

1- William Harvey Research Institute, QMUL, United Kingdom

**10.10 - 10.20 DIETARY INTERVENTIONS TARGETING GLUCOSE METABOLISM AND HYPERINSULINEMIA: A NEW TRANSLATIONAL PERSPECTIVE FOR THE MANAGEMENT OF PATIENTS WITH ACUTE INTERMITTENT PORPHYRIA**

[Miriam Longo](#) (1) | [Teresa Rubio](#) (2) | [Araceli Lamelas](#) (2) | [Daniel Jericó](#) (3) | [Jordi Cervero](#) (2) | [Juan Martínez-Blanch](#) (2) | [Empar Chenoll](#) (2) | [Erika Paolini](#) (1) | [Karol M Córdoba](#) (3) | [Marica Meroni](#) (1) | [José Ignacio Riezu-Boj](#) (4,5) | [Raquel Urtasun](#) (6) | [Isabel Solares](#) (7) | [Ana Sampedro](#) (3) | [Francesco Urigo](#) (3) | [María Collantes](#) (5,8,9) | [Michele Battistin](#) (10) | [Stefano Gatti](#) (10) | [Ivan Peñuelas](#) (5,8,9) | [María Jesús Moreno-Aliaga](#) (4,5,11) | [Matías A Ávila](#) (3,5,12) | [Elena Di Pierro](#) (1) | [Miguel Barajas](#) (6) | [Daniel Ramon](#) (2) | [Fermín I Milagro](#) (4,5,12) | [Paola Dongiovanni](#) (1) | [Antonio Fontanellas](#) (3,5,12)

1- Medicine and Metabolic Diseases, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy. | 2- ADM BIOPOLIS, University of Valencia Science Park (Parc Científic de la Universitat de València), Valencia, Spain. | 3- Hepatology: Porphyrins & Carcino-genesis Lab. Solid Tumors Program. CIMA-University of Navarra, 31008 Pamplona, Spain. | 4- Center for Nutrition Research and Department of Nutrition, Food Sciences and Physiology, Faculty of Pharmacy and Nutrition. University of Navarra, 31008 Pamplona, Spain; | 5- Navarra Institute for Health Research (IdiSNA), 31008 Pamplona, Spain | 6- Biochemistry Area, Department of Health Science, Public University of Navarra, 31008 Pamplona, Spain |



7- Rare disease Unit, Internal Medicine Department. Clinica Universidad de Navarra (CUN), 31008 Pamplona, Spain. | 8- Translational Molecular Imaging Unit (UNIMTRA), University of Navarra, 31008 Pamplona, Spain. | 9- Nuclear Medicine-Department, CUN, 31008 Pamplona, Spain. | 10- Center for Preclinical Research, Fondazione Istituto di Ricovero e Cura a Carattere Scientifico Ca' Granda, Ospedale Maggiore Policlinico, Milan, Italy | 11- Centro de Investigación Biomédica en Red de Fisiopatología de la Obesidad y Nutrición (CIBEROBN), Instituto de Salud Carlos III, 28029 Madrid, Spain. | 12- Centro de Investigación Biomédica en Red de Enfermedades Hepáticas y Digestivas (CIBERehd), Instituto de Salud Carlos III, 28029 Madrid, Spain

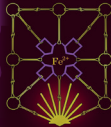
**10.20 - 10.30 NUTRITIONAL ASSESSMENT AND BODY COMPOSITION IN ACUTE INTERMITTENT PORPHYRIA (AIP) PATIENTS**

[Francesca Granata](#) (1) | [Luisella Vigna](#) (2) | [Elena Di Pierro](#) (1) | [Alessandra Piontini](#) (2) | [Lorena Duca](#) (1) | [Giacomo De Luca](#) (3) | [Valeria Di Stefano](#) (1) | [Giovanna Graziadei](#) (1)

1-Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, S.C. Medicina ad Indirizzo Metabolico, Milan, Italy. | 2- Obesity and Work Centre, Occupational Medicine Unit, Clinica del Lavoro L. Devoto, Fondazione IRCCS Ca'Granda Ospedale Maggiore Policlinico, Milan, Italy. | 3- Intermediate Care Unit, Ospedale Guglielmo da Saliceto, Piacenza, Italy

**10.30 - 11.00** *Coffee break and poster visit*





11.00 - 12.20

**SESSION 10 RATIONAL APPROACH(ES) TO DIAGNOSIS AND CARE  
IN CUTANEOUS PORPHYRIAS**

**CHAIRS:**

*Manisha Balwani (New York, US)*

*Oscar Millet (Derio, Spain)*

11.00 - 11.20 Afamelanotide for variegate porphyria related skin symptoms

*Anna E. Minder (Zürich, Switzerland)*

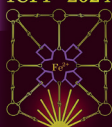
*Chantal Peltenburg (Rotterdam, Netherlands)*

**Selected abstracts presentation**

11.20 - 11.30 **LIVER FUNCTION, IRON STATUS, AND HEMOPOIESIS IN  
ERYTHROPOIETIC PROTOPORPHYRIA (EPP): INSIGHTS INTO A  
COMPLEX INTERPLAY**

*Andrea Ricci (1,2) | Dario Saltini (3) | Camilla Mancini (1) | Tomas Guasconi (3,4) | Giada Di Betto (1) | Cinzia Garuti (2) | Stefano Marchini (1) | Filippo Schepis (3) | Antonello Pietrangelo (1,2) | Elena Corradini (1,2) | Paolo Ventura (1,2)*

1- Regional Reference Centre for Diagnosing and Management of Porphyrins, Internal Medicine Unit, Department of Medical and Surgical Science for Children and Adults, Azienda Ospedaliero-Universitaria Policlinico di Modena, University of Modena and Reggio Emilia, Italy | 2- Internal Medicine Unit and Centre for Hereditary Anemias, ERN-EuroBloodNet Center for Iron Disorders, Azienda Ospedaliero-Universitaria Policlinico di Modena, University of Modena and Reggio Emilia, Italy | 3- Severe Liver Diseases (M.E.C.) Departmental Unit, Department of Medical Specialties, Azienda Ospedaliero-Universitaria of Modena, University of Modena and Reggio Emilia, Modena, Italy | 4- Division of Gastroenterology, Azienda Ospedaliero-Universitaria of Modena, University of Modena and Reggio Emilia, Modena, Italy



**11.30 - 11.40    QUANTITATIVE ANALYSIS OF POLY-UNSATURED FATTY ACIDS PROFILES IN ERYTHROPOIETIC PROTOPORPHYRIA**

[Michele De Canio](#) (1) | [Stefania Briganti](#) (1) | [Caterina Aurizi](#) (1) | [Miriam Teoli](#) (1) | [Anna Di Nardo](#) (1) | [Luca Barbieri](#) (1)

1- San Gallicano Dermatological Institute IRCCS, Rome, Italy

**11.40 - 11.50    BASELINE CHARACTERISTICS OF A PEDIATRIC COHORT OF PATIENTS WITH ERYTHROPOIETIC PROTOPORPHYRIA AND X-LINKED PROTOPORPHYRIA**

[Luca Fierro](#), MS, CGC (1) | [Chanan Stauffer](#), MS, RN, NP-C (1) | [Mary Freeman](#), MS, CGC (1) | [Catherine McDonough](#), MS (1) | [Robert J. Desnick](#), PhD, MD (1) | [Manisha Balwani](#), MD, MS, FACMG (1) | [Hetanshi Naik](#), PhD, MS, CGC (1)

1- Icahn School of Medicine at Mount Sinai

**11.50 - 12.00    QUALITY OF CARE AND RISK OF RELAPSE IN PORPHYRIA CUTANEA TARDA: A STUDY FROM THE NORWEGIAN PORPHYRIA REGISTRY**

[Åshild Rostad Enes](#) (1) | [Jørild Haugen Villanger](#) (1) | [Aasne Karine Aarsand](#) (1)

1- The Norwegian Porphyria Centre, Haukeland University Hospital

**12.00 - 12.10    PORPHYRIA PATIENTS AND THE ROLE OF UV AND VISIBLE SUNSCREENS IN THEIR PHOTO-PROTECTION**

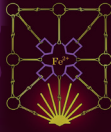
[David Bajek](#) (1) | [Ewan Eadie](#) (2) | [Robert S Dawe](#) (3) | [Sally H Ibbotson](#) (2) | [Victoria A McGuire](#) (3)

1- Photobiology Unit, University of Dundee , Ninewells Hospital & Medical School, Dundee, UK. | 2- Scottish Photobiology Service, Photobiology Unit, University of Dundee & NHS Tayside, Ninewells Hospital & Medical School, Dundee, UK. | 3- Scottish Cutaneous Porphyria Service, Photobiology Unit, University of Dundee & NHS Tayside, Ninewells Hospital & Medical School, Dundee, UK



**WEDNESDAY**  
**September 25<sup>th</sup>**  
**2024**

ICPP 2024



INTERNATIONAL  
CONGRESS OF  
PORPHYRINS AND  
PORPHYRIAS



**12.10 - 12.20 QUALITY OF LIFE IN PATIENTS WITH PORPHYRIAS - FIRST DATA FROM THE GERMAN PORPHYRIA REGISTRY (PoReGer)**

Eva Diehl-Wiesenecker (1) | Perau Said (1) | Nils Wohmann (2) | Ilja Kubisch (2) | Mona Mainert (1) | Ulrich Stölzel (2) | Rajan Somasundaram (1) | [Lea Gerischer](#) (3,4)

1- Charité - Universitätsmedizin Berlin, corporate member of Freie Universität Berlin and Humboldt-Universität zu Berlin, Department of Emergency Medicine and Porphyria Clinic, Berlin, Germany | 2- Department of Internal Medicine II, Saxony Porphyria Center, Klinikum Chemnitz gGmbH, Chemnitz, Germany | 3- Charité - Universitätsmedizin Berlin, corporate member of Freie Universität Berlin and Humboldt-Universität zu Berlin, Department of Neurology, Berlin, Germany | 4- Charité - Universitätsmedizin Berlin, corporate member of Freie Universität Berlin and Humboldt-Universität zu Berlin, Neuroscience Clinical Research Center, Berlin, Germany



12.20 - 13.00

**EXPERT PANEL: DIAGNOSTIC AND CLINICAL CONUNDRUMS.  
CRITICAL ISSUES TO RESOLVE AND FUTURE DIRECTIONS**

CHAIRS:

*Sverre Sandberg (Bergen, Norway)*

**12.20 - 12.30 C-TYPE LECTIN 10A AS A POTENTIAL BIOMARKER IN ACUTE  
INTERMITTENT PORPHYRIA**

*Elin Storjord, MD, PhD (1) | Bård Ove Karlsen, PhD (1,2) | Ole-Lars Brekke,  
Prof. MD, PhD (1,3)*

1- Department of Laboratory Medicine, Nordland Hospital Trust, 8092 Bodø, Norway. | 2- Research Laboratory, Nordland Hospital Trust, 8092 Bodø, Norway. | 3- Department of Clinical Medicine, UiT-The Arctic University of Norway, 9019 Tromsø, Norway

**12.30 - 12.40 STABILITY OF PORPHYRINS AND PORPHYRIN PRECURSORS IN  
URINE AND PLASMA SAMPLES; IMPLICATIONS FOR SAMPLE  
HANDLING AND STORAGE**

*Claire Gallagher (1,2) | Lucy-Anne Bentley (1,2) | Rhiannon Challenger (1,2)  
| Martyn Jones (1,2) | Danja Schulenburg-Brand (1,3)*

1- Cardiff Porphyria Service, University Hospital of Wales, Cardiff, UK | 2- Department of Medical Biochemistry and Immunology, University Hospital of Wales, Cardiff, UK | 3- Department of Haematology, Immunology and Metabolic Medicine, University Hospital of Wales, Cardiff, UK

**12.40 - 12.50 51-YEAR-FOLLOW-UP OF A PATIENT WITH DOSS-PORPHYRIA IN  
TIMES OF GIVOSIRAN**

*Ilja Kubisch (1) | Nils Wohmann (1) | Thomas Stauch (2) | Margareta Doss  
(3) | Till Wissniowski (1) | Ulrich Stölzel (1)*

1- Porphyria Center, Klinikum Chemnitz, Chemnitz, Germany | 2- Labor Volkmann, Karlsruhe, Germany | 3- MVZ Innere Medizin Marburg, Germany

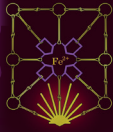
**12.50 - 13.00 MENTAL HEALTH IN THE PORPHYRIAS**

*Kristen Wheeden (1) | Liz Allan (1)*

1- United Porphyrias Association

**WEDNESDAY**  
**September 25<sup>th</sup>**  
**2024**

ICPP 2024



INTERNATIONAL  
CONGRESS OF  
PORPHYRINS AND  
PORPHYRIAS



13:00 - 13.50

### POSTERS AWARDS AND CONGRESS CLOSING CEREMONY

*María Iraburu (Rector of the University of Navarra, Pamplona, Spain)*

*Sverre Sandberg (Bergen, Norway)*

*Matías Ávila (Pamplona, Spain)*

*Antonio Fontanellas (Pamplona, Spain)*

---

# INDUSTRY SPONSORED SESSIONS

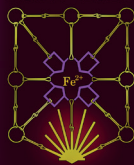
---

## INTERNATIONAL CONGRESS OF PORPHYRINS AND PORPHYRIAS

September 21-25, 2024

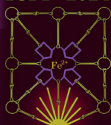
Pamplona (Spain)

ICPP 2024



**ip.net**  
INTERNATIONAL PORPHYRY ASSOCIATION





# INDUSTRY SPONSORED SESSION

## MONDAY September 23<sup>rd</sup>, 2024

### 14.30 - 15.00 Industry Symposium

CHAIRS:

*Karl Anderson (Galveston, TX, US)*

Sponsored by:



Welcome & introduction

*Jordi To-Figueras (Barcelona, Spain)*

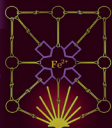
Secondary Induced Heme Deficiency & Acute Attacks of Porphyria

*Amy Dickey (Massachusetts, US)*

Q&A Open Discussion

September  
21-25, 2024

ICPP 2024



INTERNATIONAL  
CONGRESS OF  
PORPHYRINS AND  
PORPHYRIAS



# INDUSTRY SPONSORED SESSION

**TUESDAY**  
**September 24<sup>th</sup>, 2024**

10.05 - 10.35 **Industry Symposium**

CHAIRS:

*Paolo Ventura (Modena, Italy)*

*Robert Desnick (New York, US)*

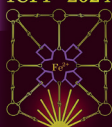
**Sponsored by:**



DEVELOPMENT OF AN IN VIVO CRISPR-CAS9 GENE EDITING TREATMENT FOR AIP

*Sandeep Soni (Executive Director at CRISPR Therapeutics, Inc., Atlanta, US)*

*Mary-Lee Dequeant (Senior Director, Head of in vivo Research at CRISPR Therapeutics, Boston, US)*



# INDUSTRY SPONSORED SESSION

**TUESDAY**

**September 24<sup>th</sup>, 2024**

## 11.00 - 11.30 Industry Symposium

CHAIRS:

*Delia D´Avola (Madrid, Spain)*

*Maria Domenica Cappellini (Milano, Italy)*

**Sponsored by:**



ADVANCING AHP CARE: BRIDGING THE GAP BETWEEN THEORY AND PRACTICE

### 11.00 - 11.05 Welcome & introduction

*Paula Aguilera (Barcelona, Spain)*

### 11.05 - 11.20 From Clinical Studies to Clinical Experience - Exploring the Impact on Long-Term Outcomes and Quality of Life

*Nils Wohmann (Chemnitz, Germany)*

### 11.20 - 11.30 Interactive Discussion on Improving AHP Management

*Paula Aguilera (Barcelona, Spain)*

*Nils Wohmann (Chemnitz, Germany)*

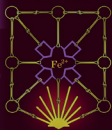
### 11.30 Closing remarks

*Paula Aguilera (Barcelona, Spain)*



September  
21-25, 2024

ICPP 2024



INTERNATIONAL  
CONGRESS OF  
PORPHYRINS AND  
PORPHYRIAS



# INDUSTRY SPONSORED SESSION

**TUESDAY**  
**September 24<sup>th</sup>, 2024**

15.00 - 18.00

## SESSION 8 EMERGING THERAPIES FOR ERYTHROPOIETIC PORPHYRIAS

### CHAIRS:

*Michael Badminton (Wales, UK)*

*Jean Charles Deybach (Paris, France)*

15.00 - 15.30 Industry Symposium

### Sponsored by:



Mitsubishi Tanabe Pharma America

### DERSIMELAGON IN ADULTS AND ADOLESCENTS WITH EPP/XLP

*Kirstine Belongie (Washington, US)*

15.30 - 16.00 Industry Symposium

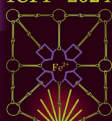
### Sponsored by:



PORT-77, A NOVEL SMALL MOLECULE INHIBITOR OF ABCG2, PROTECTS AGAINST SKIN PHOTOTOXICITY AND LIVER DAMAGE IN A MOUSE MODEL OF EPP

*Pete C. Schmidt (MD MSc)*

*Bhavik Shah (PhD)*



# INDUSTRY SPONSORED SESSION

**TUESDAY**  
**September 24<sup>th</sup>, 2024**

16.00 - 16.30 Industry Symposium

Sponsored by:

CLINUVEL

LONG-TERM SAFETY OF SCENESSE® (AFAMELANOTIDE 16MG) IN EPP:  
LATEST UPDATES FROM THE PASS STUDY

*William Burch (Medical Manager at CLINUVEL, London, UK)*

16.30 - 17.00 *Coffee break*

17.00 - 17.30 Industry Symposium

Sponsored by:

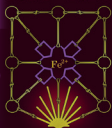
Usense

USE OF AN INNOVATIVE IN VITRO MEDICAL DEVICE FOR THE MEASURING  
OF PORPHOBILINOGEN AND TOTAL URINARY PORPHYRINS IN THE  
DETECTION OF PORPHYRIAS

*Guillaume Lemetais (Co-founder & CEO atvUsense, Boulogne-Billancourt,  
France)*

September  
21-25, 2024

ICPP 2024



INTERNATIONAL  
CONGRESS OF  
PORPHYRINS AND  
PORPHYRIAS



# INDUSTRY SPONSORED SESSION

**TUESDAY**  
**September 24<sup>th</sup>, 2024**

17.30 - 18.00 Industry Symposium

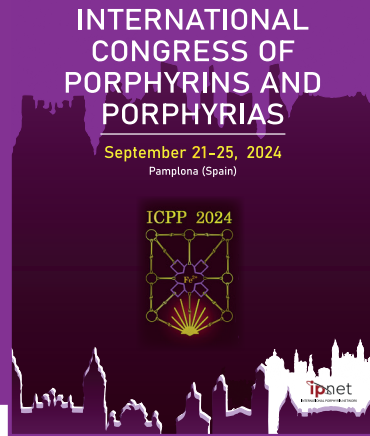
Sponsored by:

**disc**medicine

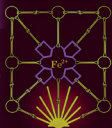
MAKING CHANGES IN EPP

*Will Savage (Chief Medical Office at Disc Medicine, Cambridge, MA, US)*

# POSTER SESSION



THEMATIC AREA	POSTERS
CURRENT AND EMERGING THERAPIES FOR PORPHYRIAS	1 - 14
DIAGNOSIS AND CARE IN PORPHYRIAS	15 - 62 [71 - 72]
HEME SYNTHESIS AND REGULATION	63 - 65
RECENT ADVANCES IN THE PATHOPHYSIOLOGY OF PORPHYRIAS	66 - 70



## POSTER 1 - 14

### Poster 1

#### **INHIBITION OF GLYCINE TRANSPORTER 1 REDUCES EPP PHOTOTOXICITY IN A MOUSE MODEL OF ERYTHROPOIETIC PROTOPORPHYRIA (EPP)**

Yi Xiang (1) | Yue Chen (1) | Scott Reisman (1) | Srikanth Venkatraman (1) | Hua Yang (1) | Brian MacDonald (1) | Min Wu (1)

1- Disc Medicine

### Poster 2

#### **AFAMELANOTIDE FOR THE TREATMENT OF CUTANEOUS PHOTOTOXICITY OF ERYTHROPOIETIC PROTOPORPHYRIA: THE SCOTTISH EXPERIENCE**

Robert S Dawe (1) | Alastair Kerr (2) | Marese O'Reilly (3) | Ewan Eadie (3) | Victoria A McGuire (1)

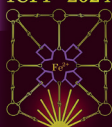
1- Scottish Cutaneous Porphyria Service, Photobiology Unit, University of Dundee & NHS Tayside, Ninewells Hospital & Medical School, Dundee, UK. | 2- Department of Dermatology, Queen Elizabeth University Hospital, NHS Greater Glasgow & Clyde, Glasgow, UK | 3- Scottish Photobiology Service, Photobiology Unit, University of Dundee & NHS Tayside, Ninewells Hospital & Medical School, Dundee, UK.

### Poster 3

#### **METHIONINE CYCLE, POLYAMINE SYNTHESIS, AND TRANSULFURATION PATHWAY IN THE LIVER OF A NOVEL AIP MODEL IN NON-HUMAN PRIMATES AND THE EFFECT OF SYSTEMIC MRNA THERAPY**

Francesco Urigo (1) | Daniel Jericó (1) | Lei Jiang (2) | Karol M. Córdoba (1) | Ana Sampedro (1) | Juan Manuel Falcón (3,4) | Diana Cabrera (3,4) | Sebastiaan Martijn Van Liempd (3,4) | Alfonso Martínez de la Cruz (3,4) | Estíbaliz Alegre (5,6) | Pedro Berraondo (7) | Paolo G.V. Martini (2) | Matías A. Ávila (1,4,6,8) | Antonio Fontanellas (1,4,6,8)

1- Hepatology: Porphyrias&Carcinogenesis Lab. Solid Tumors Program. CIMA-University of Navarra, Pamplona | 2- Moderna Inc, Cambridge MA, USA | 3- Exosomes Lab. & Metabolomics Platform, CIC bioGUNE, Bizkaia Technology Park, Derio, Spain | 4- CIBERehd, Instituto de Salud Carlos III, Spain | 5- Service of Biochemistry. Clínica Universidad de Navarra, Pamplona, Spain | 6- Instituto de Investigación Sanitaria de Navarra, Spain | 7- Program of Immunology and Immunotherapy, CIMA-University of Navarra, and CIBERONC) Instituto de Salud Carlos III, Madrid, Spain | 8- Cancer Center CUN, Pamplona, Spain



#### Poster 4

**FAIRNESS OF FUNDING DECISIONS OF INNOVATIVE THERAPIES FOR THE PORPHYRIAS IN ENGLAND AND WALES: COMPARISON OF THE PARAMETERS FOR THE CALCULATION OF QUALITY-ADJUSTED LIFE YEARS ACCEPTED BY THE NATIONAL INSTITUTE FOR HEALTH AND CARE EXCELLENCE**

Jasmin Barman-Aksözen (1,2) | Nicole Hentschel (3) | Mårten Pettersson (1) | Eva Schupp (1) | Francesca Granata (1,4) | Cornelia Dechant (1) | Mehmet Hakan Aksözen (1) | Rocco Falchetto (1)

1- International Porphyria Patient Network (IPPN), Hegarstrasse 3, 8032 Zurich, Switzerland | 2- University of Zurich | 3- Independent researcher | 4- Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, S.C Medicina ad Indirizzo Metabolico, 20122 Milano, Italy

#### Poster 5

**ASSESSING DISEASE IMPACT AND QUALITY OF LIFE: INSIGHTS FROM A PATIENT SUPPORT PROGRAM FOR ACUTE HEPATIC PORPHYRIA**

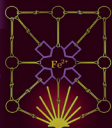
Francesca Brambillasca (1) | Paolo Ventura (2) | Annamaria Porreca (3) | Carlo Poci (4) | Annalisa Crisetti (5) | Antonina Giammanco (6) | Matteo Marcacci (2)

1- PHDIlifescience, Italy. | 2- Internal Medicine Unit and Centre for diagnosing and management of Porphyrias, University Hospital Policlinico of Modena, University of Modena and Reggio Emilia, Italy. | 3- Department of Medical, Oral and Biotechnological Sciences, Laboratory of Biostatistics, D'Annunzio University of Chieti-Pescara, Via dei Vestini 31, 66100 Chieti, Italy. | 4- Department of Medicine, Padova University Hospital, Padova, Italy. | 5- Division of Nephrology, Casa Sollievo della sofferenza, San Giovanni Rotondo, Foggia, Italy. | 6- A.O.U.P. "Paolo Giaccone di Palermo", Palermo, Italy.

#### Poster 6

**VALIDATION OF ANATOMICAL AND BRAIN PERFUSION DYNAMICS IN AIP NON-HUMAN PRIMATE MODEL USING NON-INVASIVE MRI, AND ASSESSMENT OF EFFICACY WITH HPBGD MRNA THERAPY**

Daniel Jericó (1) | Leyre García-Ruiz (2,3) | Verónica Aramendía-Vidaurreta (2,3) | Lei Jiang (4) | Karol M. Córdoba (1) | Maite Aznarez (2,3) | Francesco Urigo (1) | Ana Sampedro (1) | Alberto Rico (5) | Iñigo Insausti (2) | Manuel Alegre (6) | Azucena Aldaz (7) | Carmen Unzu (8) | Jose Luis Lanciego (5) | María A. Fernández-Seara (2,3) | Paolo G.V. Martini (4) | Matías A. Ávila (1,3,9) | Antonio Fontanellas (1,3,9)



## POSTER 1 - 14

1- Hepatology: Porphyrins&Carcinogenesis Lab. Solid Tumors Program, CIMA-University of Navarra (UNAV), Pamplona & CIBERehd, Spain | 2- Radiology Department, Clínica Universidad de Navarra (CUN), Pamplona, Spain | 3- Instituto de Investigación Sanitaria de Navarra (IdisNA), Spain | 4- Moderna Inc, Cambridge MA, USA | 5- Neurosciences Department, CIMA-UNAV, Pamplona & CIBERNed, Spain | 6- Department of Clinical Neurophysiology, CUN, Pamplona, Spain | 7- Service of Pharmacy, CUN, Pamplona, Spain | 8- Gene Therapy and Regulation of Gene Expression Program, CIMA-UNAV, Pamplona, Spain | 9- Cancer Center CUN, Pamplona, Spain

### Poster 7

#### HEMIN TREATMENT DURING ATTACKS IN ARGENTINEAN PATIENTS WITH ACUTE PORPHYRIAS

Viviana Alicia Melito (1,2) | Laura Sabina Varela (1) | María Del Carmen Martínez (1,2) | Verónica Goñi (1) | Claudio Orquera (3) | Fernando Baccaro (4) | Sergio Herrando (5) | Guido Merlo (7) | Diego Sánchez Gelós (1) | María Pasman (6) | Ana María Buzaleh (1) | Lucía Tomassi (6) | Victoria Estela Parera (1)

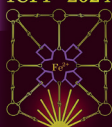
1- Centro de Investigaciones sobre Porphirinas y Porphirias (CIPYP), UBA-CONICET, Buenos Aires, Argentina | 2- Dpto. de Química Biológica, Facultad de Ciencias Exactas y Naturales, Universidad de Buenos Aires, Argentina | 3- Sanatorio Mitre, Buenos Aires, Argentina | 4- Hospital Fernández, Buenos Aires, Argentina | 5- Sanatorio Sagrado Corazón, Buenos Aires, Argentina | 6- Hospital Ramos Mejía, Buenos Aires, Argentina | 7- Hospital de Clínicas, Buenos Aires, Argentina

### Poster 8

#### PATERNAL SPLIT-LIVER TRANSPLANTATION FOLLOWED BY HAPLOIDENTICAL HEMATOPOIETIC CELL TRANSPLANTATION IN AN ADULT PATIENT WITH PROTOPORPHYRIA-INDUCED LIVER FAILURE

Stölzel Ulrich (1) | Jost Lasse (2) | Daniel Seehofer (3) | Egger-Heidrich Katharina (4) | Scheuermann Uwe (3) | Hölig Kristina (4) | Stauch Thomas (5) | Kunadt Desiree (4) | Detlef Schuppan (6) | Schetelig Johannes (4) | Wohmann Nils (1) | Bornhäuser Martin (4) | Stölzel Friedrich (2,4)

1- Department of Internal Medicine II and Porphyria Center, Klinikum Chemnitz, Chemnitz, Germany | 2- Division of Stem Cell Transplantation and Cellular Immunotherapies, Depart-



ment of Internal Medicine II, University Hospital Schleswig Holstein Kiel, Kiel University, Germany | 3- Department of Visceral, Transplantation, Vascular and Thoracic Surgery, University Hospital of Leipzig, Leipzig, Germany | 4- Department of Internal Medicine I, University Hospital Carl Gustav Carus, TU Dresden, Dresden, Germany | 5- Deutsches Kompetenz-Zentrum für Porphyriadiagnostik und Konsultation, MVZ Labor Volkmann und Kollegen GbR, Karlsruhe, Germany | 6- Institute of Translational Immunology, Johannes Gutenberg University Mainz, Mainz, Germany

### Poster 9

#### EFFECT OF GIVOSIRAN FOR HEREDITARY COPROPORPHYRIA: 2 CASES REPORT

Tomohide Adachi (1) | Nobuaki Ozaki (2)

1- Department of General Internal medicine and Neurology, Tokyo Saiseikai Central Hospital | 2- Department of Endocrinology, Japanese Red Cross Aichi Medical Center Nagoya Daiichi Hospital

### Poster 10

#### RECURRENT ACUTE INTERMITTENT PORPHYRIA ATTACKS AFTER NORMALIZATION OF PORPHOBILINOGEN ON GIVOSIRAN PROPHYLAXIS

Akshata Moghe, MD PhD (1,2) | Csilla K. Hallberg, MD (2,3) | Ruksana Huda, PhD (2,3) | Shalonda Turner (2,3) | Rochelle Simmons (2,3) | V.M. Sadagopa Ramanujam, PhD (2,3) | Karl E. Anderson, MD (2,3)

1- University of Texas Health Science Center at Houston | 2- UTMB Porphyria Center and Laboratory | 3- University of Texas Medical Branch

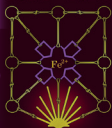
### Poster 11

#### ADMINISTRATION OF HEMIN AND TIME-RELATED EFFECTS ON FERRITIN IN ACUTE HEPATIC PORPHYRIA

Akshata Moghe, MD PhD (1,2) | Garrett T. Coleman (3) | Csilla K. Hallberg, MD (2,3) | Ruksana Huda, PhD (2,3) | Shalonda Turner (2,3) | Rochelle Simmons (2,3) | V.M. Sadagopa Ramanujam, PhD (2,3) | Karl E. Anderson, MD (2,3)

1- University of Texas Health Science Center at Houston | 2- UTMB Porphyria Center and Laboratory | 3- University of Texas Medical Branch





## POSTER 1 - 14

### Poster 12

#### **SUCCESSFUL PREGNANCY AFTER COMBINED LIVER AND KIDNEY TRANSPLANTATION IN A PATIENT WITH ACUTE INTERMITTENT PORPHYRIA**

Petro E.Petrides (1,2) | Joachim Andrassy (3) | Markus Guba (3) | Manfred Stangl (3) | Olivia Bronisch (1) | Maria K.Beykirch (1)

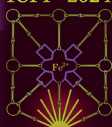
1- IPNET Center Munich | 2- LMU, University of Munich, School of Medicine | 3- Transplantation Unit, LMU University of Munich Grosshadern

### Poster 14

#### **CLINICAL AND BIOCHEMICAL EVOLUTION OF EIGHT ACUTE HEPATIC PORPHYRIA PATIENTS UNDER GIVOSIRAN TREATMENT**

C. Puente Fernández (1), I. Solares (1), F.J. Castellbón (1), I. Liria Fernandez (2), S. Diaz-Diaz (2), D.Bellido (3), J. Ros (4), P.M Alcazar-Carmona (4), R. Enríquez de Salamanca (1), A. Fontanellas (5), M. Morales-Conejo (1) - (1,2,3,4,5)

1- Department of Internal Medicine, Hospital Universitario 12 de Octubre, Madrid, Spain; CIBERER. | 2- Biochemistry service, Hospital Universitario 12 de Octubre, Madrid, Spain; CIBERER. | 3- Department of Internal Medicine, Hospital General Universitario de Ciudad Real, Ciudad Real, Spain | 4- Department of Internal Medicine, Hospital La Mancha Centro, Albacete, Spain. | 5- Center for Applied Medical Research (CIMA)-University of Navarra, Pamplona, Spain. CIBERehd



### Poster 15

#### PORPHYRIA REGISTRY PADOVA - START UP

Annamaria Nicolli (1,2) | Mauro Alaibac (2,3) | Cinzia Bertolin (2) | Isabella Bortoletti (2) | Alberto Gambalunga (1,2) | Angelo Moretto (1,2) | Fabiola Pasqualato (1,2) | Leonardo Salviati (2,4) | Paolo Simioni (2,3) | Andrea Trevisan (1,2) | Carlo Poci (2)

1- Department of Cardiac Thoracic Vascular Sciences and Public Health, University of Padova, Padova, Italy | 2- Padova University Hospital, Padova, Italy | 3- Department of Medicine, University of Padova, Padova, Italy | 4- Department of Woman and Child Health, University of Padova, Padova, Italy

### Poster 16

#### HEREDITARY COPROPORPHYRIA WITH CEREBRAL ANEURYSMS: SUCCESSFUL DIAGNOSIS AND TREATMENT WITH GIVOSIRAN

Aurora del Mar Pérez Ys (1) | Annalisa Crisetti (1) | Maria Grazia Savino (2) | Maria Nardella (1) | Francesco Aucella (1) | Claudio Carmine Guida (1) | Filippo Aucella (1)

1- Nephrology and Dialysis Unit, Casa Sollievo della Sofferenza, San Giovanni Rotondo | 2- Immunohematology and Transfusion Medicine Service, Casa Sollievo della Sofferenza, San Giovanni Rotondo, Italy

### Poster 17

#### AN UNUSUAL CASE OF VARIEGATE PORPHYRIA WITH NERVE ENLARGEMENT, CONDUCTION BLOCK AND IDIOPATHIC INTRACRANIAL HYPERTENSION

Michelle Abdo Paiva (1) | Andre Macedo Serafim Silva (1) | Rodrigo de Holanda Mendonça (1) | Ana Lucila Moreira (1) | José Pedro Soares Baima (1) | Nícolas Ruan dos Santos Cavalcante (1) | Carlos Otto Heise (1) | Edmar Zanoteli (1)

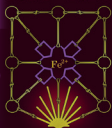
1- Department of Neurology, Faculty of Medicine of University of São Paulo, São Paulo, Brazil

### Poster 18

#### THREE YEARS LATER: HOW THE PREVIOUSLY DEFINED VISUAL THRESHOLD OF THE HOESCH TEST WORKS

Galina Zemtsovskaja (1,2) | Siim Iskül (1,2) | Küllike Palk (2,3)

1- Clinical Chemistry and Haematology Department of the Laboratory, North Estonia Medical Centre Foundation, Tallinn, Estonia | 2- Estonian Porphyria Centre, Tallinn, Estonia | 3- Gastroenterology Centre of the Internal Medicine Clinic, North Estonia Medical Centre Foundation



## POSTER 15 - 62 [71 - 72]

### Poster 19

#### ACUTE PORPHYRIA SCREENING IN EMERGENCY DEPARTMENTS: AN OPPORTUNITY FOR FAST PATIENT TRACKING

Sonia Moreira (1) | Filipa Ferreira (2) | Arlindo Guimas (3) | Anabela Oliveira (4)

1- Internal Medicine Department, ULS Coimbra, EPE | Coimbra Healthcare Integrated delivery system, Coimbra, Portugal | 2- Newborn Screening, Metabolic and Genetics Unit, Department of Human Genetics, National Institute of Health Dr Ricardo Jorge, 4000-055 Porto, Portugal | 3- Internal Medicine Department, Unidade Local de Saúde Santo António, Porto, Portugal | 4- Internal Medicine Department, Unidade Local de Saúde de Santa Maria, Lisboa, Portugal

### Poster 20

#### HORMONAL TREATMENTS IN ACUTE HEPATIC PORPHYRIA

Gayle Ross (1)

1- Royal Melbourne Hospital, Melbourne, Australia

### Poster 21

#### BRIDGING THE GAP: ENHANCING PATIENT ENGAGEMENT THROUGH ACCESSIBLE MEDICAL INFORMATION IN THE PORPHYRIAS. A CASE STUDY IN ERYTHROPOIETIC PROTOPORPHYRIA

Anna Mann (1) | Liz Allan (1) | Kristen Wheeden (1) | Amy Dickey MD (2) | Cythnia Levy, MD (3)

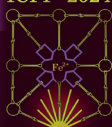
1- United Porphyrias Association | 2- Massachusetts General Hospital | 3- Shiff Cancer Center, University of Miami

### Poster 22

#### THE EPP LIGHT STUDY: UNDERSTANDING THE BURDEN OF ERYTHROPOIETIC PROTOPORPHYRIA (EPP) FROM THE PATIENT PERSPECTIVE

Hetanshi Naik (1) | Kristen Wheeden (2) | Susan D Mathias (3) | Chelsea Norregaard (4) | Hilary H Colwell (3) | Melanie Chin (4) | William Savage (4) | Manisha Balwani (5)

1- Dept of Genetics, Stanford University School of Medicine, Palo Alto, CA | 2- United Porphyrias Association, Bethesda, MD | 3- Health Outcomes Solutions, Palm Beach Gardens, FL | 4- Disc Medicine, Watertown, MA | 5- Departments of Genetics and Genomic Sciences, Icahn School of Medicine at Mount Sinai, New York, NY



### Poster 23

#### **PORPHYRIA CUTANEA TARDA IN SCOTLAND: UNDERLYING ASSOCIATIONS AND TREATMENT APPROACHES**

Chayada Chaiyabutr (1,2) | Robert S Dawe (3) | Sally H Ibbotson (2) | Tara Clarke (2) | Victoria A McGuire (3)

1- Department of Dermatology, Faculty of Medicine Siriraj Hospital, Mahidol University, Bangkok, Thailand. | 2- Scottish Photobiology Service, Photobiology Unit, University of Dundee & NHS Tayside, Ninewells Hospital & Medical School, Dundee, UK. | 3- Scottish Cutaneous Porphyria Service, Photobiology Unit, University of Dundee & NHS Tayside, Ninewells Hospital & Medical School, Dundee, UK.

### Poster 24

#### **MENSTRUAL CYCLE-DEPENDENT SYMPTOMS IN PATIENTS WITH ACUTE HEPATIC PORPHYRIA - DATA FROM THE GERMAN PORPHYRIA REGISTRY (POREGER)**

Mona Mainert (1) | Lea Gerischer (2,3) | Rajan Somasundaram (1) | Sylvia Mechsner (4) | Eva Diehl-Wiesenecker (1)

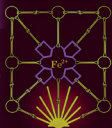
1- Charité - Universitätsmedizin Berlin, corporate member of Freie Universität Berlin and Humboldt-Universität zu Berlin, Department of Emergency Medicine and Porphyria Clinic, Berlin, Germany | 2- Charité - Universitätsmedizin Berlin, corporate member of Freie Universität Berlin and Humboldt-Universität zu Berlin, Department of Neurology, Berlin, Germany | 3- Charité - Universitätsmedizin Berlin, corporate member of Freie Universität Berlin and Humboldt-Universität zu Berlin, Neuroscience Clinical Research Center, Berlin, Germany | 4- Charité - Universitätsmedizin Berlin, corporate member of Freie Universität Berlin and Humboldt-Universität zu Berlin, Department of Obstetrics and Gynaekology, Berlin, Germany

### Poster 25

#### **VARIEGATE PORPHYRIA CASES WITH FAECAL ISOCOPROPORPHYRIN - NOT DUAL PORPHYRIA**

Gayle Ross (1,2) | Joel Smith (1) | Virginia Cronin (1)

1- Royal Melbourne Hospital, Melbourne, Australia | 2- University of Melbourne



## POSTER 15 - 62 [71 - 72]

### Poster 26

#### THE FIRST CASES OF $\delta$ -AMINOLEVULINATE DEHYDRATASE PORPHYRIA (ADP/DOSS PORPHYRIA) IN ITALY

Carlo Poci (1) | Mauro Alaibac (1,2) | Marta Balzarin (3) | Cinzia Bertolin (1) | Isabella Bortoletti (1) | Alessandra Brugiolo (3) | Federica Rachele Danti (4) | Elena Di Pierro (5) | Angelo Moretto (1,6) | Fabiola Pasqualato (1,6) | Leonardo Salviati (1,7) | Paolo Simioni (1,2) | Andrea Trevisan (1,6) | Paolo Ventura (8) | Anna Zanin (9) | Annamaria Nicolli (1,6)

1- Padova University Hospital, Padova, Italy | 2- Department of Medicine, University of Padova, Padova, Italy | 3- Department of Women and Child's Health, San Bortolo Hospital, Vicenza, Italy | 4- Department of Pediatric Neurosciences, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano, Italy | 5- Medicine and metabolic diseases unit, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy | 6- Department of Cardiac Thoracic Vascular Sciences and Public Health, University of Padova, Padova, Italy | 7- Department of Woman and Child Health, University of Padova, Padova, Italy | 8- Internal Medicine Unit, University of Modena and Reggio Emilia, Modena, Italy | 9- Department of Women and Children's Health, Paediatric Palliative Care, Pain Service, University of Padova, Padova, Italy

### Poster 27

#### DUAL PORPHYRIA: INITIAL PORPHYRIA CUTANEA TARDA IN A PATIENT WITH VARIEGATE PORPHYRIA

Marcelo Salazar (1) | Akshata Moghe (1,2) | Csilla K. Hallberg, MD (1) | Ruksana Huda, PhD (1) | Shalonda Turner (1) | Rochelle Simmons (1) | V.M. Sadagopa Ramanujam, PhD (1) | Karl E. Anderson, MD (1)

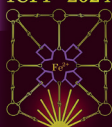
1- Porphyria Center and Laboratory, University of Texas Medical Branch, USA | 2- University of Texas Health Science Center, Houston, Texas, USA

### Poster 28

#### EVALUATING THE IMPACT OF A FAMILY CAMP FOR ERYTHROPOIETIC PROTOPORPHYRIA (EPP) AND OTHER RARE SUN DISORDERS

Kristen Wheeden (1) | Craig Leppert (2) | Liz Allan (1) | Hetanshi Naik (3)

1- United Porphyrins Association | 2- Shadow Jumpers | 3- Stanford University School of Medicine



### Poster 29

#### UNDERSTANDING VARIEGATE PORPHYRIA IN AN HIV PATIENT: A DETAILED CASE STUDY

Laura Sabina Varela (1) | Viviana Alicia Melito (1) | Fabiana Alejandra Caballero (1) | Marcelo Guolo (1) | Verónica Goñi (1) | Pablo Winitzky (1) | Mariana Agosta (2) | María Pasman (3) | Ana María Buzaleh (1) | Lucía Tomassi (3) | Victoria Estela Parera (1)

1- Centro de Investigaciones sobre Porfirinas y Porfirias (CIPYP), UBA-CONICET, Buenos Aires, Argentina | 2- Hospital Rocca, Buenos Aires, Argentina | 3- Hospital Ramos Mejía, Buenos Aires, Argentina

### Poster 30

#### MANAGEMENT OF HEREDITARY COPROPORPHYRIA: A CASE REPORT OF A NEW PATHOGENIC VARIANT, HIGHLIGHTING THE SIGNIFICANCE OF HEMIN THERAPY AND MAINTENANCE TREATMENT COMPLIANCE

Laura Sabina Varela (1) | Viviana Alicia Melito (1,2) | Ana María Buzaleh (1) | Victoria Estela Parera (1) | Elena Di Pierro (3)

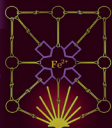
1- Centro de Investigaciones sobre Porfirinas y Porfirias (CIPYP), UBA-CONICET, Buenos Aires, Argentina | 2- Dpto. de Química Biológica, Facultad de Ciencias Exactas y Naturales, Universidad de Buenos Aires, Argentina | 3- Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan, Italia

### Poster 31

#### A CHALLENGING DIAGNOSIS FOR A POTENTIALLY FATAL DISEASE: 2 YEARS OF PORTUGUESE EXPERIENCE IN BIOCHEMICAL AND MOLECULAR DIAGNOSIS OF PORPHYRIAS

Filipa Ferreira (1) | Célia Carmona (1) | Célia Nogueira (2) | Cristina Pereira (2) | Altina Lopes (2) | Laura Vilarinho (1,2)

1- Newborn Screening, Metabolism and Genetics Unit, Human Genetics, Department, National Institute of Health Dr Ricardo Jorge, Rua Alexandre Herculano, 321, 4000-055 Porto, Portugal | 2- Research and Development Unit, Department of Human Genetics, National Institute of Health Dr Ricardo Jorge, Portugal



## POSTER 15 - 62 [71 - 72]

### Poster 32

#### ACQUIRED CONGENITAL ERYTHROPOIETIC PORPHYRIA SECONDARY TO TREATMENT FOR HAEMATOLOGICAL MALIGNANCY

John Gilman (1) | Amira Ibrahim (1) | Saliha Haji (1) | Liezel Griffin (1,2) | Denise Darby (1)

1- Department of Biochemistry, Salford Royal Care Organisation, Northern Care Alliance NHS Trust | 2- Department of Dermatology, Salford Royal Care Organisation, Northern Care Alliance NHS Trust

### Poster 33

#### VERIFICATION AND IMPLEMENTATION OF CHROMSYSTEMS HPLC KIT FOR URINARY PORPHYRINS AT NORTH ESTONIA MEDICAL CENTRE

Siim Iskül (1)

1- North Estonia Medical Centre

### Poster 34

#### HAEMATOPOIETIC STEM CELL TRANSPLANT FOR SEVERE "DOUBLY HOMOZYGOUS" ERYTHROPOIETIC PROTOPORPHYRIA: EARLY OUTCOME

Robert S Dawe (1) | Shahzya Chaudhury (2) | Victoria A McGuire (1)

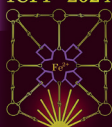
1- Scottish Cutaneous Porphyria Service, Photobiology Unit, University of Dundee & NHS Tayside, Ninewells Hospital & Medical School, Dundee, UK. | 2- Royal Hospital for Children, Queen Elizabeth University Hospital, NHS Greater Glasgow and Clyde, Glasgow, UK

### Poster 35

#### THE GENETIC LANDSCAPE OF PORPHYRIA IN IRELAND

Sarah Savage (1) | Micheál Mac Aogáin (1,2) | Brendan Lawlor (3,4) | Craig Maher (1) | Niamh Stein (1) | Aoife McConnon (1) | Ruth Brennan (1) | Nadia Brazil (1) | Vivion E.F. Crowley (1,2)

1- Department of Biochemistry, St James's Hospital, Dublin, Ireland. | 2- School of Medicine, Trinity College Dublin, Ireland. | 3- Department of Computer Science, Munster Technological University, Cork, Ireland | 4- Department of Biological Sciences, Munster Technological University, Cork, Ireland



### Poster 36

#### CASSANDRA CURSE" OR A MODERN "SISYPHUS BURDEN"? RECURRENT ATTACKS IN A COHORT OF BRAZILIAN PATIENTS WITH ACUTE HEPATIC PORPHYRIAS

Amadeu Queiroz (1) | Jacqueline Harouche Rodrigues Da Fonseca (2) | Regina Albuquerque (1) | Maria Da Penha Ananias Morita (1) | Erica Coelho (1) | Eduardo Estephan (3) | Ieda Bussmann (4) | Charles Marques Lourenco (1)

1- National Reference Center for Rare Diseases - Faculty of Medicine of São José do Rio Preto, São José do Rio Preto - São Paulo, Brazil | 2- DLE - Bioquímica, Rio de Janeiro - RJ, Brazil | 3- Neurology, Faculty of Medicine of São José do Rio Preto, São José do Rio Preto - São Paulo, Brazil | 4- BRAZILIAN PORPHYRIA ASSOCIATION - ABRAPO, Curitiba - Paraná, Brazil

### Poster 37

#### WHOLE EXOME DIAGNOSIS FOR CLARIFIES THE CONCOMITANT PATHOLOGIES OF A PATIENT WITH PORPHYRIA

Hada C. Macher (1,2) | Salvador García-Morillo (3) | Jose Luis GarcíadeVeas-Silva (1) | Ana Rodriguez-Rodriguez (1) | Carmen Delgado-Pecellín (1,2)

1- 1. Hospital Universitario Virgen del Rocío de Sevilla, departamento de Bioquímica Clínica | 2- Instituto de Biomedicina de Sevilla (IBiS) | 3- 2. Hospital Universitario Virgen del Rocío de Sevilla, departamento de Medicina Interna (Enfermedades Minoritarias)

### Poster 38

#### THE INCIDENCE OF TYPES OF PORPHYRIA AND THEIR SYMPTOMS IN TURKEY

Gülbüz Sezgin (1) | Yusuf Emirhan Erkan (1) | Hüseyin Şimşek (1) | Ecehan Uzun (1) | Çağdaş Kaya (1) | Ayli Heydari (1) | Gizem Tatar (2)

1- Maltepe University Faculty of Medicine | 2- Malatya State Hospital

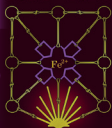
### Poster 39

#### QUALITY OF LIFE IN CHILDREN WITH ERYTHROPOIETIC PROTOPORPHYRIA; A CASE-CONTROL STUDY

Louisa Kluijver (1)

1- Porphyria Center Rotterdam, Center for Lysosomal and Metabolic Diseases, Department of Internal Medicine, Erasmus MC, University Medical Center, Rotterdam, the Netherland.





## POSTER 15 - 62 [71 - 72]

### Poster 40

#### CONCOMITANT AIP AND SYSTEMIC LUPUS ERYTHEMATOSUS (SLE) - COINCIDENCE OR CLINICAL ASSOCIATION?

Mark W. Sonderup (1) | Sandy Mosenye (1) | Peter Meissner (1) | C. Wendy Spearman (1)

1- University of Cape Town, Division of Hepatology

### Poster 41

#### DETECTION AND QUANTIFICATION OF LABILE INTRAVASCULAR HEME IN THE PRESENCE OF HEMOGLOBIN

Marie-T. Hopp (1,2) | Sonali M. Vaidya (1) | Karina M. Grimmig (1) | Xiaojing Yuan (3) | Iqbal Hamza (3) | Diana Imhof (1)

1- Pharmaceutical Biochemistry and Bioanalytics, Pharmaceutical Institute, University of Bonn, Bonn, Germany | 2- Department of Chemistry, Institute for Integrated Natural Sciences, University of Koblenz, Koblenz, Germany | 3- Center for Blood Oxygen Transport and Hemostasis, Department of Pediatrics, University of Maryland School of Medicine, US-21201, Baltimore, MD, USA

### Poster 42

#### RETHINKING ACUTE INTERMITTENT PORPHYRIA NUTRITIONAL GUIDELINES: A NEW APPROACH PROPOSAL

Isabel Palazón Cano (1) | Manuel Palazón Cano (2)

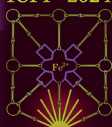
1- Hospital Ruber Internacional | 2- Groupe Hospitalier intercommunal Le Raincy Montfermeil

### Poster 43

#### PREVALENCE OF ACUTE HEPATIC PORPHYRIA IN THE FRENCH POPULATION: FIRST RESULTS OF THE PREVPHA PROSPECTIVE STUDY

Roland Jaussaud (1) | Patrick Mercié (2) | Laurent Alric (3) | Ivan Bertchansky (4) | Claire Douillard (5) | Adrien Bigot (6) | Esther Noel (7) | Bernard Bonnotte (8) | Arnaud Hot (9) | Antoine Poulet (10) | Boris Bienvenu (11) | Camille Lepart (12) | Patrice Cacoub (13)

1- Hôpital Brabois, Nancy, France | 2- Hôpital Saint-André, Bordeaux, France | 3- Hôpital Rangueil, Toulouse, France | 4- Hôpital Saint-Eloi, Montpellier, France | 5- Hôpital Huriez, Lille, France | 6- Hôpital Trousseau, Tours, France | 7- Hôpital Civil, Strasbourg, France | 8- Hôpital Dijon-Bourgogne, Dijon, France | 9- Hôpital Edouard Herriot, Lyon, France | 10- Hôpital Saint-Joseph, Marseille, France | 11- Hôpital National des 15-20, Paris, France | 12- Hôpital Sud, Rennes, France | 13- Hôpital Pitié Salpêtrière, Paris, France



#### Poster 44

### EARLY, QUICK AND ACCURATE IDENTIFICATION OF ACUTE PORPHYRIAS IN HEALTH CENTERS: PBG REAGENT

María Del Carmen Martínez (1,2) | Viviana Alicia Melito (1,2) | Fabiana Alejandra Caballero (1) | Marcelo Guolo (1) | Juan Bautista Lucero (3) | Walter Schlender (3) | Victoria Estela Parera (1) | Ana María Buzaleh (1)

1- Centro de Investigaciones sobre Porfirinas y Porfirias (CIPYP), UBA -CONICET, Buenos Aires, Argentina | 2- Dpto. de Química Biológica, Facultad de Ciencias Exactas y Naturales, Universidad de Buenos Aires, Argentina | 3- Tuteur, Buenos Aires, Argentina

#### Poster 45

### ERYTHROPOIETIC PROTOPORPHYRIA: EXPLAINING THE NORTH-SOUTH GAP

Javier de la Iglesia-Martin (1) | Francisco Javier Castelbon (2) | Jordi To-Figueras (3) | Rafael Enríquez de Salamanca (2) | Celia Badenas (3,4) | Cristina Puente Fernandez (2) | Paula Aguilera Peiro (1,4)

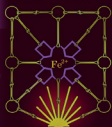
1- Servei de Dermatologia Hospital Clínic de Barcelona. Universitat de Barcelona, Barcelona, Spain | 2- Unidad de Enfermedades Metabólicas Congénitas y otras Enfermedades Minoritarias. Servicio de Medicina Interna Hospital Universitario 12 de Octubre. Universidad Complutense de Madrid. Madrid, Spain | 3- Servei de Bioquímica i genètica Molecular Hospital Clínic de Barcelona. Universitat de Barcelona. Barcelona, Spain | 4- Institut d'Investigacions Biomèdiques August Pi I Sunyer (FRCB-IDIBAPS), Barcelona

#### Poster 46

### FUNCTIONAL STUDY IN AN EUKARYOTIC CELL MODEL OF A NEW HMBS MISSENSE VARIANT IDENTIFIED IN TWO HOMOZYGOUS PATIENTS OF THE SAME FAMILY

Gaël Nicolas (1) | Loréna Chappuis (2) | Hana Manceau (1,3) | Antoine Poli (1,2) | Sophie Scheidecker (4,5) | Louise Porter (6) | Véronique Geoffroy (4) | Jean Muller (4,5) | Esther Noel (7) | Marie-Thérèse Abi-Warde (8) | Corinne Stoetzel (4) | Neila Talbi (2) | Thibaud Lefebvre (1,2) | Héliène Dollfus (4,6) | Laurent Gouya (1,2) | Caroline Schmitt (1,2)

1- Université Paris Cité, Centre de Recherche sur l'Inflammation and INSERM UMR1149, Paris, France | 2- Assistance Publique-Hôpitaux de Paris, Centre Français des Porphyries, Hôpital Louis Mourier, Colombes, France | 3- Assistance Publique-Hôpitaux de Paris, Laboratoire de Biochimie, Hôpital Beaujon, Clichy, France | 4- Laboratoire de Génétique médicale, UMR\_S INSERM U1112, IGMA, Faculté de Médecine FMTS, Université de Strasbourg,



## POSTER 15 - 62 [71 - 72]

Strasbourg, France | 5- Laboratoire de Diagnostic Génétique, Hôpitaux Universitaires de Strasbourg, Strasbourg, France | 6- Centre de Référence pour les affections rares en génétique ophtalmologique, CARGO, Filière SENSGENE, Hôpitaux Universitaires de Strasbourg, Strasbourg, France | 7- Service de Médecine Interne - Diabète et Maladies métaboliques, Hôpitaux Universitaires de Strasbourg, Strasbourg, France | 8- Service de Pédiatrie, Hôpitaux Universitaires de Strasbourg, Strasbourg, France

### Poster 47

#### DRUGS STIMULATING ERYTHROID COMPARTMENT CAN INDUCE HEPATIC DAMAGE IN ERYTHROPOIETIC PROTOPORPHYRIA

Giovanna Graziadei (1) | Valeria Di Stefano (1) | Bruno Fattizzo (2) | Marta Bortolotti (2) | Valentina Brancaleoni (1) | Francesca Granata (1) | Silvia Fustinoni (3) | Elena Di Piero (1)  
1- Medicine and Metabolic Diseases Unit, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy | 2- Hematology Unit, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy | 3- EPIGET Lab, Department of Clinical Sciences and Community Health - University of Milan, Milan, Italy

### Poster 48

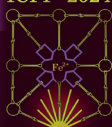
#### INVESTIGATION FOR PORPHYRIA IN DENMARK

Anne Lindegaard Christiansen (1,2) | Henrik Lorentzen (3) | Palle B Nielsen Fruekilde (1) | Jan Frystyk (4)  
1- Clinical Biochemistry, Odense University Hospital, Denmark | 2- Department of Blood test, Biochemistry and Immunology, University Hospital of Southern Denmark, Aabenraa, Soenderborg, Denmark | 3- Department of Dermatology, Odense University Hospital, Denmark | 4- Endocrine Research Unit, Department of Endocrinology, Odense University Hospital, Denmark

### Poster 49

#### FUNCTIONAL CHARACTERIZATION OF NEW PATHOGENIC AND LEAD-POISONING PREDISPOSING VARIANTS IN ALA DEHYDRATASE PORPHYRIA (ADP)

Francisco Javier Castalbón (1,2) | Elena Di Piero (3) | Isabel Solares (4) | Daniel Jericó (5) | Javier Tomás Solera (1) | Antoni Riera-Mestre (6) | María Barreda (2,7) | Annamaria Nicolli (8,9) | Rafael Enríquez de Salamanca (1,2) | Carlo Poci (9) | Marta Gloria Fanlo-Maresma (6,10,11) | Matías A. Ávila (5,12,13) | Matteo Marcacci (14) | Pauline Harper (15) | Encarna Guillén-Navarro (2,7) | Giovanna Graziadei (3) | Bodo Beck (16) | Paolo Ventura (14) | Montserrat Morales-Conejo (1,2,17) | Antonio Fontanellas (5,12,13)



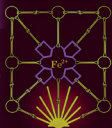
1- Department of Internal Medicine, Research Institute "i+12", University Hospital, 12 de Octubre, Madrid, Spain | 2- Spanish Network for Biomedical Research in Rare Diseases (CIBERER), U723, Spain | 3- Medicine and metabolic diseases unit, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy | 4- Rare disease Unit, Internal Medicine Department. Clinica Universidad de Navarra, 31008 Pamplona, Spain | 5- Hepatology Program, Centre for Applied Medical Research (CIMA), University of Navarra, Pamplona, Spain | 6- Internal Medicine Department, Hospital Universitari de Bellvitge - IDIBELL, L'Hospitalet de Llobregat, Barcelona, Spain; Faculty of Medicine and Health Sciences, Universitat de Barcelona, Barcelona, Spain | 7- Genética Médica, Servicio de Pediatría, Hospital Clínico Universitario Virgen de la Arrixaca, Instituto Murciano de Investigación Biosanitaria (IMIB-Arrixaca), Facultad de Medicina, Universidad de Murcia, Murcia, España | 8- Department of Medicine and public health, University of Padua, Padua, Italy | 9- University Hospital, Padova, Italy | 10- Unitat de Risc Vascular. Medicina Interna. Hospital Universitari de Bellvitge | 11- CIBEROBN Fisiopatología de la Obesidad y Nutrición, Instituto de Salud Carlos III, Madrid, Spain. | 12- Centro de Investigación Biomédica en Red de Enfermedades Hepáticas y Digestivas (CIBEREhd), Instituto de Salud Carlos III, Madrid, Spain | 13- Instituto de Investigación Sanitaria de Navarra (IdiSNA), Pamplona, Spain. | 14- Department of Surgical and Medical Sciences for Children and Adults, Internal Medicine Unit, University of Modena and Reggio Emilia, Modena, Italy | 15- Centre for Inherited Metabolic Diseases (CMMS), Porphyria Centre Sweden, Karolinska University Hospital, Stockholm, Sweden. | 16- Institute of Human Genetics and Center for Molecular Medicine Cologne (CMMC), University of Cologne, Cologne, Germany | 17- National Reference Center for Congenital Errors of Metabolism (CSUR) and European Reference Center for Inherited Metabolic Disease (MetabERN). University Hospital, 12 de Octubre, Madrid, Spain.

## Poster 51

### FATIGUE IN PATIENTS WITH PORPHYRIAS - FIRST DATA FROM THE GERMAN PORPHYRIA REGISTRY (PoReGer)

Lea Gerischer (1,2) | Perau Said (3) | Nils Wohmann (4) | Ilja Kubisch (4) | Mona Mainert (3) | Ulrich Stölzel (4) | Rajan Somasundaram (3) | Andreas Meisel (1,5) | Eva Diehl-Wiese-necker (3)

1- Charité - Universitätsmedizin Berlin, corporate member of Freie Universität Berlin and Humboldt-Universität zu Berlin, Department of Neurology, Berlin, Germany | 2- Charité - Universitätsmedizin Berlin, corporate member of Freie Universität Berlin and Humboldt-Universität zu Berlin, Neuroscience Clinical Research Center, Berlin, Germany | 3-



## POSTER 15 - 62 [71 - 72]

Charité - Universitätsmedizin Berlin, corporate member of Freie Universität Berlin and Humboldt-Universität zu Berlin, Department of Emergency Medicine and Porphyria Clinic, Berlin, Germany | 4- Department of Internal Medicine II, Saxony Porphyria Center, Klinikum Chemnitz | 5- Charité - Universitätsmedizin Berlin, corporate member of Freie Universität Berlin and Humboldt-Universität zu Berlin, Neuroscience Clinical Research Center, Berlin, Germany

### Poster 52

#### A REAL-LIFE OVERVIEW OF DIET PATTERNS AND ACUTE HEPATIC PORPHYRIA

Lucienne Oettel (1) | Ilja Kubisch (1) | Nils Wohmann (1) | Romy Andrae (1) | Jacqueline Uhle (1,1) | Thaddäus Till Wissniowski (2) | Ulrich Stölzel (1)

1- Porphyria Center Chemnitz, Department of Internal II, Klinikum Chemnitz, Flemmingsstraße 2, 09116 Chemnitz | 2- Department of Internal II, Klinikum Chemnitz, Flemmingsstraße 2, 09116 Chemnitz

### Poster 53

#### SLEEP AND STRESS IN PATIENTS WITH ACUTE INTERMITTENT PORPHYRIA

Lucienne Oettel (1) | Ilja Kubisch (1) | Nils Wohmann (1) | Thaddäus Till Wissniowsk (2) | Ulrich Stölzel (1)

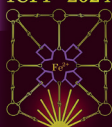
1- Porphyria Center Chemnitz, Department of Internal II, Klinikum Chemnitz, Flemmingsstraße 2, 09116 Chemnitz | 2- Department of Internal II, Klinikum Chemnitz, Flemmingsstraße 2, 09116 Chemnitz

### Poster 54

#### PROFOUND HYPOTONIA IN A NEWBORN WITH BIALLELIC $\delta$ -AMINOLEVULINIC ACID DEHYDRATASE (ALAD) MUTATIONS

Alexis Roach DO (1) | Hannah Barkley, MD (1) | Carissa Rodriguez, MD (1) | T Andrew Burrow, MD (1) | Karl E Anderson MD (2) | Ankita Shukla MD (1)

1- University of Arkansas for Medical Sciences | 2- University of Texas Medical Branch at Galveston



### Poster 55

#### ACUTE PORPHYRIAS AS A PRIME EXAMPLE FOR (UNRECOGNIZED) RARE BUT TREATABLE DISEASE IN THE EMERGENCY DEPARTMENT: A PATIENT SURVEY AND SYSTEMATIZED LITERATURE REVIEW

Sandra Pflock (1) | Hannah Carolina Mücke (1) | Rajan Somasundaram (1) | Eva Diehl-Wiesenecker (1)

1- Charité – Universitätsmedizin Berlin, corporate member of Freie Universität Berlin and Humboldt-Universität zu Berlin, Department of Emergency Medicine and Porphyria Clinic, Berlin, Germany

### Poster 56

#### INCREASED PANCREATIC ENZYMES IN A PATIENT WITH ACUTE INTERMITTENT PORPHYRIA

Valeria Di Stefano (1) | Elena Di Pierro (1) | Francesca Granata (1) | Lorena Duca (1) | Dario Tavazzi (2) | Maria Domenica Cappellini (1) | Giovanna Graziadei (1)

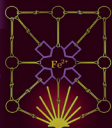
1- Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, S.C. Medicina ad Indirizzo Metabolico, Milan, Italy | 2- Epidemiology, Epigenetics, and Toxicology Lab, Department of Clinical Sciences and Community Health, University of Milan, 20122 Milan, Italy

### Poster 57

#### FIRST REPORT OF CLINICALLY MANIFEST VARIEGATE PORPHYRIA (VP) AFTER MATERNAL SPLIT LIVER TRANSPLANTATION IN A ONE-YEAR-OLD GIRL WITH IDIOPATHIC BILIARY ATRESIA

Stölzel Ulrich (1) | Knoppke Birgit (2) | Melter Michael (2) | von Eichborn Maria Isabel (3) | Bonkovsky Herbert Lloyd (4) | Stauch Thomas (5)

1- Department of Internal Medicine II and Porphyria Center, Klinikum Chemnitz, Chemnitz, Germany | 2- University Children's Hospital Regensburg (KUNO), University Medical Center Regensburg, Regensburg, Germany | 3- Department of Dermatology, University Hospital Regensburg, Regensburg, Germany. | 4- Laboratory for Liver & Metabolic Disorders Wake Forest University School of Medicine/NC Baptist Hospital E-112, NRC, USA | 5- Deutsches Kompetenz-Zentrum für Porphyriadiagnostik und Konsultation, MVZ Labor Volkmann und Kollegen GbR, Karlsruhe, Germany



## POSTER 15 - 62 [71 - 72]

### Poster 58

#### ACQUIRED ERYTHROPOIETIC PROTOPORPHYRIA DUE TO MYELOYDYSPLASTIC SYNDROME

Gayle Ross (1,2)

1- Royal Melbourne Hospital, Melbourne, Australia | 2- University of Melbourne

### Poster 59

#### PORTAL HYPERTENSION IN ADVANCED PROTOPORPHYRIC HEPATOPATHY: A CASE REPORT

Andrea Ricci (1,2) | Dario Saltini (3) | Camilla Mancini (1) | Giada Di Betto (1) | Stefano Marchini (1) | Marcello Bianchini (3) | Filippo Schepis (3) | Antonello Pietrangelo (1,2) | Elena Corradini (1,2) | Paolo Ventura (1,2)

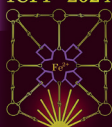
1- Regional Reference Centre for Diagnosing and Management of Porphyrins, Internal Medicine Unit, Department of Medical and Surgical Science for Children and Adults, Azienda Ospedaliero-Universitaria Policlinico di Modena, University of Modena and Reggio Emilia, Italy | 2- Internal Medicine Unit and Centre for Hereditary Anemias, ERN-EuroBloodNet Center for Iron Disorders, Azienda Ospedaliero-Universitaria Policlinico di Modena, University of Modena and Reggio Emilia, Italy | 3- Severe Liver Diseases (M.E.C.) Departmental Unit, Department of Medical Specialties, Azienda Ospedaliero-Universitaria of Modena, University of Modena and Reggio Emilia, Modena, Italy

### Poster 60

#### CHRONIC NEUROLOGICAL MANIFESTATIONS IN ACUTE PORPHYRIA

Elena Pischik (1) | Katrin Baumann (2) | Raili Kauppinen (3)

1- Department of Neurology, Consultative and Diagnostic Centre with Polyclinics, St Petersburg, Russia | 2- Departments of Gynaecology and Obstetrics, and Medicine, Helsinki University Central Hospital, Helsinki, Finland | 3- Department of Medicine, Helsinki University Central Hospital, Helsinki, Finland



### Poster 61

#### THE IMPACT OF ERYTHROPOIETIC PROTOPORPHYRIA (EPP) ON FAMILY QUALITY OF LIFE

Griffin LL (1,2) | Sarkany R (3) | Schulenburg-Brand D (4) | Rowland C (5) | Rhodes LE (1,2) | Farrar MD (1)

1- Division of Musculoskeletal and Dermatological Sciences, School of Biological Sciences, Faculty of Biology, Medicine and Health, The University of Manchester, UK. | 2- Photobiology Unit, Dermatology Centre, Salford Royal Hospital, Northern Care Alliance NHS Foundation Trust, Manchester Academic Health Science Centre, Manchester, UK. | 3- Cutaneous Porphyria Unit, Photodermatology, St John's Institute of Dermatology, Guy's and St Thomas's NHS Foundation Trust, London, UK. | 4- Cardiff Porphyria Service, Department of Haematology, Immunology and Metabolic Medicine, University Hospital of Wales, Cardiff. | 5- Division of Psychology and Mental Health, School of Health Sciences, Faculty of Biology, Medicine and Health, The University of Manchester.

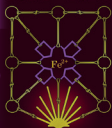
### Poster 62

#### IN-DEPTH BIOCHEMICAL INVESTIGATIONS IN A NEW CASE OF ALAD DEFICIENCY: BETTER UNDERSTANDING FOR BETTER TREATMENT

Thibaud Lefebvre (1,2) | Agathe Molimard (3) | Tristan Mekdade (3) | Caroline Schmitt (1,2) | Jérôme Lamoril (4) | Neila Talbi (1,2) | Nathalie Dessendier (1) | Antoine Poli (1,2) | Andrea Araujo (1) | Lorena Chappuis (1) | Hervé Puy (1,2) | Isabelle Desguerre (3) | Laurent Gouya (1,2)

1- Assistance Publique-Hôpitaux de Paris, Centre Français des Porphyries, Hôpital Louis Mourier, Colombes, France | 2- Université Paris Cité, Centre de Recherche sur l'Inflammation and INSERM UMR1149, Paris, France | 3- Assistance Publique-Hôpitaux de Paris, Service de neurologie pédiatrique, Hôpital Necker-Enfants Malades, Paris, France | 4- Assistance Publique-Hôpitaux de Paris, Département de génétique moléculaire, Hôpital Bichat, Paris, France





## POSTER 15 - 62 [71 - 72]

### Poster 71

#### CAUSAL GENETIC VARIANTS OF ACUTE PORPHYRIA IN COLOMBIA

Julián Ramírez-Cheyne, MD (1) | Mónica Murillo Ortiz (2) | Wilmar Saldarriaga

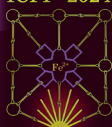
1- Msc, Professor at Universidad del Valle, Cali, Colombi | 2- MD at Hospital Universitario del Valle Evaristo García and E.S.E., and Professor at Universidad del Valle

### Poster 72

#### CLINICAL CHARACTERISTICS AND HEALTH-RELATED QUALITY OF LIFE OF PATIENTS INCLUDED IN THE FIRST NATIONAL REGISTRY OF ACUTE HEPATIC PORPHYRIAS IN COLOMBIA

Julián Ramírez-Cheyne (1) | José Rojas-Suárez | Fernando Arnedo Arteaga | Leydis Lora-Lián | Jose Santacruz-Arias | María Camila Vargas Hernández | Rianna Muñoz-Fuenmayor | Lievi Correa | Javier Barón | Mónica Murillo Ortiz (2)

1- Msc, Professor at Universidad del Valle, Cali, Colombia | 2- MD at Hospital Universitario del Valle Evaristo García and E.S.E., and Professor at Universidad del Valle



### Poster 63

#### THE GOOD, THE BAD AND THE UGLY: GLUCOSE, INSULIN AND HEME DEFICIENCY - IMPAIRMENT OF GLUCOSE METABOLISM IN PATIENTS WITH ACUTE HEPATIC PORPHYRIAS PRESENTING WITH RECURRENT ATTACKS

Amadeu Queiroz (1) | Jacqueline Harouche Rodrigues Da Fonseca (2) | Regina Albuquerque (1) | Maria Da Penha Ananias Morita (1) | Erica Coelho (1) | Eduardo Estephan (3) | Charles Marques Lourenco (1)

1- National Reference Center for Rare Diseases - Faculty of Medicine of São José do Rio Preto, São José do Rio Preto - São Paulo, Brazil | 2- DLE - BIOQUIMICA - RIO DE JANEIRO - RJ, BRAZIL | 3- Faculty of Medicine of São José do Rio Preto, São José do Rio Preto - São Paulo, Brazil

### Poster 64

#### THE RELATIONSHIP BETWEEN PROTOPORPHYRIN IX AND HEMATOLOGIC PARAMETERS IN PATIENTS WITH PROTOPORPHYRIA

Rebecca Karp Leaf (1) | Paul Y. Jiang (1) | Benny Tran (1) | Robert J. Goddu (1) | Narmene Bensaber (1) | Lina Rebeiz (1) | Sarina B. Elmariah (1) | Pamela Hodges (1) | Jennifer Mead (1) | John Trinidad (1) | Behnam Saberi (2) | Mark D. Fleming (3) | Sarah Ducamp (3) | Karl E. Anderson (4) | Staffan Wahlin (5) | Amy K. Dickey (1)

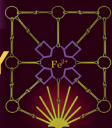
1- Massachusetts General Hospital | 2- Beth Israel Deaconess Medical Center | 3- Boston Children's Hospital | 4- University of Texas, Medical Branch | 5- Karolinska University Hospital

### Poster 65

#### OBSERVED AND PREDICTED EFFECTS OF HMBS VARIANTS - RESULTS FROM THE PREDPOR STUDY

Helene J. Bustad (1) | Marthe S. Christie (1) | Juha P. Kallio (2) | Marte Høvik Hammersland (1) | Åshild Enes Rostad (1) | Janice Andersen (1) | Jørlid Haugen Villanger (1) | Mikko Laitaoja (3) | Janne Jänis (3) | Aurora Martinez (2) | Aasne K. Aarsand (1)

1- Norwegian Porphyria Centre (NAPOS), Department for Medical Biochemistry and Pharmacology, Haukeland University Hospital, Bergen, Norway | 2- Department of Biomedicine, University of Bergen, Norway | 3- Department of Chemistry, University of Eastern Finland, Joensuu, Finland



## POSTER 66 - 70

### Poster 66

#### COEXISTENCE OF WILSON'S DISEASE AND HEREDITARY COPROPORPHYRIA: EFFECT OF COPPER ON THE HEME PATHWAY

Elif bilge atasay (1) | layda altun (1) | Sude çavdaro lu (1) | Gülbüz sezgin (2)

1- Faculty of Medicine, Maltepe University, Istanbul/TURKEY | 2- Department of Internal Medicine, Maltepe University Hospital, Istanbul/TURKEY

### Poster 67

#### BLOOD PRESSURE PROFILES AND LEFT VENTRICULAR HYPERTROPHY IN PATIENTS WITH ACUTE INTERMITTENT PORPHYRIA

Krzysztof Jaworski (1) | Robert Wasilewski (2) | Jerzy Windyga (2,3) | Ilona Kowalik (4) | Agnieszka Lipniacka (3) | Hanna Szwed (1) | Rafal Dabrowski (1)

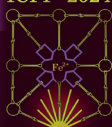
1- Department of Coronary Artery Disease and Cardiac Rehabilitation, National Institute of Cardiology, Warsaw, Poland | 2- Department of Hemostasis Disorders and Internal Medicine, Institute of Hematology and Transfusion Medicine, Warsaw, Poland | 3- Laboratory of Genetics of Hemostasis and Porphyria, Institute of Hematology and Transfusion Medicine, Warsaw, Poland | 4- Clinical Research Support Center, National Institute of Cardiology, Warsaw, Poland

### Poster 68

#### HEPATIC PORPHYRIAS ONSET. THE ROLE OF NR1/2 GENE VARIANTS

Johanna Romina Zuccoli (1) | Viviana Alicia Melito (1,2) | Laura Sabina Varela (1) | Priscila Ayelén Pagnotta (1,2) | Victoria Estela Parera (1) | Ana María Buzaleh (1)

1- Centro de Investigaciones sobre Porfirinas y Porfirias (CIPYP), UBA-CONICET, Buenos Aires, Argentina | 2- Dpto. de Química Biológica, Facultad de Ciencias Exactas y Naturales, Universidad de Buenos Aires, Argentina



### Poster 69

#### SMART METABOLIC METRO-MAP: DYNAMICS OF THE BIOCHEMICAL CHAIN REACTIONS AND ITS CORRELATION TO PATHOPHYSIOLOGY OF METABOLIC DISORDERS

Dr. Farzaneh Taringou (1) | Prof. Dr. Thomas Weiland (2)

1- ACE - Advanced Computational Engineering GmbH | 2- Technical University of Darmstadt

### Poster 70

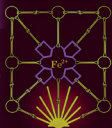
#### A CONGENITAL ERYTHROPOIETIC PORPHYRIA CASE OF UNKNOWN GENETIC MUTATION

Dalal Yehya (1) | Liezel Griffin (2,3) | Denise Darby (1)

1- Department of Biochemistry, Salford Care Organization, Northern Care Alliance NHS Foundation Trust, Manchester, UK | 2- The Dermatology Centre, Salford Care Organization, Northern Care Alliance NHS Foundation Trust, Manchester, UK | 3- The University of Manchester and Salford Royal NHS Foundation Trust, Manchester Academic Health Science Centre, Manchester, UK

**September**  
**21-25, 2024**

ICPP 2024



INTERNATIONAL  
CONGRESS OF  
PORPHYRINS AND  
PORPHYRIAS

**ipnet**  
INTERNATIONAL PORPHYRY NETWORK

## GENERAL INFORMATION

### COURSE DIRECTORS:

Sverre Sandberg  
Ipnet President

Antonio Fontanellas  
President of ICPP 2024

### ORGANIZING SECRETARIAT:

**oasis**  
EVENTS & TRAVEL SOLUTIONS

**Oasis Events & Travel Solutions**

C/ Sor Ángela de la Cruz, 8. 1º A  
28020 - Madrid (Spain)

☎ +34 91 5551119

Email: [congresos@viajesoasis.com](mailto:congresos@viajesoasis.com)

Web site: [www.viajesoasis.com](http://www.viajesoasis.com)

### MEETING VENUE:

**PATIENT´S DAY**

**[ASSEMBLY ROOM CIMA BUILDING]**

**CIMA**

(Centro de Investigación Médica Aplicada)

Av. de Pío XII, 55  
31008 Pamplona, Navarra

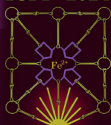
**ICPP 2024 CONGRESS**

**[ASSEMBLY ROOM OF THE SCIENCE LIBRARY BUILDING]**

**FACULTY OF SCIENCE**

(Universidad de Navarra)

C. de Irunlarrea, 1, 31008  
Pamplona, Navarra



## GENERAL INFORMATION

### MEETING DATES:

September 21-25, 2024

### SECRETARIAT HOURS:

Saturday, 21 September 2024 / From 14.00 to 18.30

Sunday, 22 September 2024 / From 08.30 to 19.00

Monday, 23 September 2024 / From 08.00 to 19.30

Tuesday, 24 September 2024 / From 08.00 to 18.00

Wednesday, 25 September 2024 / From 08.00 to 13.30

### FEE INCLUDES:

- Access to all scientific sessions.
- Lunches.
- Coffee and soft drinks in pauses scheduled.
- One full set with Meeting documentation.
- Certificate of attendance

### REGISTRATION PROCEDURES AND GUIDELINES:

Registration can be made as follows:

- Online: Web site: <https://icpp2024.com/>

### BADGE:

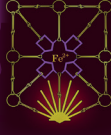
Identification badge will be necessary for admission to the scientific sessions and social events.

### DELIVERY OF MULTIMEDIA MATERIAL:

It will be delivered at least one hour before the start of each session. The meeting room will be equipped with a multimedia projector for computer projection, whose program is PowerPoint. No other software will be accepted. Speakers are kindly requested to refrain from using their laptops. Presenters who need other audiovisual material different from the one provided, must notify the Technical Secretariat sufficiently in advance.

September  
21-25, 2024

ICPP 2024



INTERNATIONAL  
CONGRESS OF  
PORPHYRINS AND  
PORPHYRIAS

**ipnet**  
INTERNATIONAL PORPHYRIA NETWORK

## ACKNOWLEDGEMENTS

The **IPNET** is grateful to the following Sponsors for their continued support of this important educational and scientific activity.



PLATINUM SPONSOR



Mitsubishi Tanabe Pharma America



GOLD SPONSOR



disc)medicine



SILVER SPONSOR

CLINUVEL



RECORDATI



REGULAR SPONSOR



**ciber** | EHD  
CENTRO DE INVESTIGACIÓN BIOMÉDICA EN RED  
Enfermedades Hepáticas y Digestivas

Our special thanks to the **Spanish Porphyria Association (AEP)** for organizing a crowdfunding campaign in support of ICPP Patient and Doctor Workshops.



Asociación  
Española de  
Porfiria



oasis

EVENTS & TRAVEL SOLUTIONS

**TECHNICAL SECRETARY:**

Oasis Events & Travel Solutions

C/ Sor Ángela de la Cruz, 8. 1A.

28020 - Madrid (Spain)

Phone: +34 915551119

Email: [congresos@viajesoasis.com](mailto:congresos@viajesoasis.com)