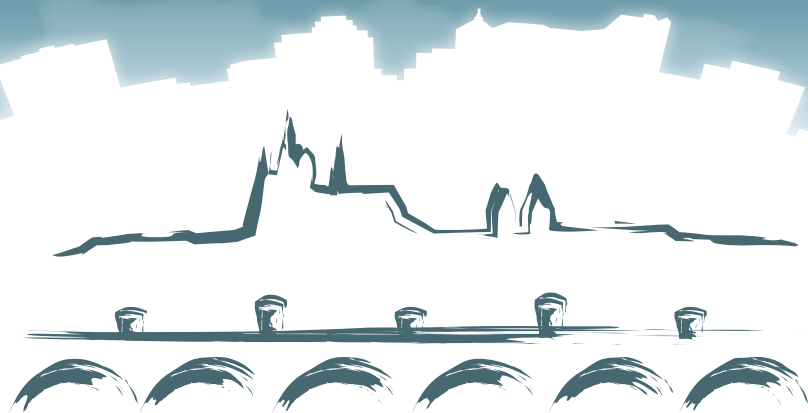


"SOMETHING NEW IS ON THE HORIZON"



# NEW HORIZONS IN FABRY DISEASE

INTERNATIONAL CONFERENCE ON ADVANCES  
IN THE TREATMENT OF FABRY DISEASE

PRAGUE, CZECH REPUBLIC  
MAY 16-17, 2025

HELD UNDER THE AUSPICES OF  
THE CZECH MEDICAL ACADEMY



ENDORSED BY



[WWW. HORIZONS-FABRY.COM](http://WWW.HORIZONS-FABRY.COM)

Organized by GALÉN-SYMPOSION  
under the auspices of the Czech Medical Academy



### Endorsed by

The Czech Society of Cardiology  
The International Cardiomyopathy Network (ICoN)



### Scientific and Organizing Committee

Professor Aleš Linhart, MD DSc., FESC  
(General University Hospital and First Faculty of Medicine,  
Charles University Prague, Czech Republic)

Professor Dominique P. Germain, MD PhD  
(French Referral Centre for Fabry Disease, University of Versailles, France)

Professor Perry M. Elliott, FESC  
(Institute of Cardiovascular Science, University College London,  
London, United Kingdom)

### General information

#### Conference Venue

hotel Diplomat Prague Vienna House by Wyndham  
Evropská 15, Prague, Czech Republic

#### Conference Dates

Friday May 16<sup>th</sup> & Saturday May 17<sup>th</sup>, 2025

#### Organizing agency

GALÉN-SYMPOSION s.r.o.  
Břežanská 10, 100 00 - Prague 10  
Czech Republic  
phone: +420 222 518 535  
www.gsymposion.com

## SCIENTIFIC PROGRAMME

### Friday May 16<sup>th</sup>, 2025

9:00 – 9:05

#### Welcome address

9:05 – 10:00

#### SESSION 1

##### Fabry disease in the context of cardiomyopathies

Perry M. Elliott:

*Differential diagnosis of a hypertrophied heart*

Pablo Garcia-Pavia:

*Cardiac amyloidosis – differential diagnosis and treatments*

Iacopo Olivetto:

*Hypertrophic cardiomyopathy management in the era of myosin inhibitors*

10:20 – 11:40

#### SESSION 2

##### Fabry cardiomyopathy – advances in diagnosis and management

James Moon:

*Contributions of cardiac MRI to the understanding of Fabry disease*

Elena Biagini:

*Novel insights from imaging studies*

Josef Marek:

*Obstructive cardiomyopathy in Fabry disease*

Mehdi Namdar:

*ECG in Fabry cardiomyopathy – a forgotten method?*

12:00 – 13:00

#### INDUSTRY SYMPOSIA

12:00 – 12:30

##### Symposium supported by SANOFI

**Practical recommendations for patients under ERT**  
*presented by Andrea Matucci*

12:30 – 13:00

##### Symposium supported by TAKEDA

**Two Decades of Experience from the Fabry Outcome Survey**

Fabian Braun:

*Welcome and overview of objectives*

Uma Ramaswami:

*Background and structure of the FOS registry*

Uma Ramaswami:

*Effects of long-term agalsidase treatment on renal function*

|               |   |
|---------------|---|
|               | Uma Ramaswami:<br><i>Effects of long-term agalsidase treatment on cardiac structure</i>   |
|               | Uma Ramaswami:<br><i>Effects of long-term agalsidase treatment on morbidity and mortality</i>   |
|               | Fabian Braun:<br><i>Closing remarks and conclusion</i>  |
| 13:00 – 14:00 | LUNCH   |
| 14:00 – 15:20 | <b>SESSION 3</b>  |
|               | <b>Fabry disease and the kidney</b>   |
|               | Camilla Tøndel:<br><i>The role of renal biopsy in the understanding of pathophysiology and treatment effects</i>  |
|               | Christoph Wanner:<br><i>Can we improve outcomes by SGLT2 inhibitors RAAS blockers?</i>  |
|               | David Warnock:<br><i>Monitoring kidney disease – is serum creatinine and eGFR just enough?</i>  |
|               | Sima Canaan-Kühl:<br><i>End-stage kidney disease – what next? How do patients do on Haemodialysis and after kidney transplantation. Does the immune response to ERT differ?</i> |
|               | Alberto Ortiz:<br><i>The role of podocytes and treatment effects</i>  |
| 15:40 – 17:20 | <b>SESSION 4</b>  |
|               | <b>Advancements in understanding mechanisms of the disease – beyond the storage</b>   |
|               | Fabian Braun:<br><i>The role of alpha-synuclein in the pathogenesis of Fabry Nephropathy</i>  |
|               | Guido Iaccarino:<br><i>The role of mitochondrial dysfunction and muscular abnormalities in Fabry disease</i>  |
|               | Moran Dvela Levitt:<br><i>Endoplasmic reticulum and its role in cellular function</i>   |
|               | Martina Živná:<br><i>Agalopathy – the role of ER stress in pathophysiology of Fabry disease</i>   |

|               |   |
|---------------|---|
|               | Maurizio Pieroni:<br><i>The role of inflammation in pathogenesis of cardiac involvement</i> |
| 17:20 – 18:00 | <b>SESSION 5</b>  |
|               | <b>Setting-up a multidisciplinary team of a Fabry center</b>                                |
|               | Federico Pieruzzi:<br><i>Paediatric Fabry disease – the transitional care model issue</i>   |
|               | Bojan Vujkovic:<br><i>Organization of a multidisciplinary Fabry center team</i>             |
| 18:00 – 18:05 | <b>DAY 1 ADJOURN</b>  |
|               | Aleš Linhart  |
| 19:00 – 22:00 | Dinner with expert discussion & networking  |

## Saturday May 17<sup>th</sup>, 2025

|               |   |
|---------------|---|
| 8:40 – 10:00  | <b>SESSION 6</b>  |
|               | <b>Current and novel therapies – where do we stand?</b>   |
|               | Antonio Pisani:<br><i>Switch from agalsidase beta to migalastat – registry data</i>                                     |
|               | Daniel Bichet:<br><i>Migalastat – analysis of registry data, amenability testing, clinical and biomarker monitoring</i> |
|               | Dominique Germain:<br><i>Substrate reduction therapies – where do we stand?</i>   |
|               | Raphael Schiffmann:<br><i>Update on gene therapies in Fabry disease</i>   |
|               | Aleš Linhart:<br><i>Enzyme-replacement therapies – long term data</i>   |
| 10:20 – 11:40 | <b>SESSION 7</b>  |
|               | <b>Monitoring of the disease and treatments impacts</b>   |
|               | Uma Ramaswami:<br><i>Monitoring PROs in Fabry disease</i>   |
|               | Malte Lenders:<br><i>Measurements of migalastat levels and antibody responses</i>                                       |

Maud Janssens:  
*Antibody response to enzyme replacement therapies*  
Albina Nowak:  
*Lyso Gb3 and novel biomarkers*

12:00 – 13:00

## **INDUSTRY SYMPOSIA**

*12:00 – 12:30*

### **Symposium supported by CHIESI • Global Rare Diseases**

**From silent damage to prompt intervention:  
enhancing monitoring in Fabry disease**

*chaired by Robert Hopkin*

Robert Hopkin:  
*Welcome and introduction*

Christine Kurschat:  
*Silent kidney progression in Fabry disease: the role of monitoring  
and the prompt intervention*

Christine Kurschat:  
*Main Pegunigalsidase alfa renal data*

Q&A

*12:30 – 13:00*

### **Symposium supported by AMICUS Therapeutics Advancing Patient-Centered Care in Fabry Disease: Real-World Insights and the Impact of Migalastat Therapy**

Robert Hopkin:  
*Patient experience with Fabry disease and Migalastat*

Uma Ramaswami:  
*UK experience of managing patients with Fabry disease with  
Migalastat*

*13:00 – 14:00*

LUNCH

*14:00 – 14:50*

## **SPLIT-OUT ABSTRACT SESSIONS**

15:00 – 16:20

## **SESSION 8**

**Advancements in understanding mechanisms  
of the disease and treatment effects**

João Paulo Oliveira:  
*Peroxidasin (PXDN) expression in kidney tissue of patients carrying  
GLA gene variants, with or without Fabry*

Alessandro Burlina:  
*Cerebrovascular events in Fabry patients - clinical course and  
outcomes*

Juan Manuel Politei:  
*Gastrointestinal complications in Fabry patients*

Gabriela Dostálová:  
*“Once upon a time in Prague... there was a...?”*

16:40 – 18:00

## **SESSION 9**

**Genetics, late onset variants, disease management  
and outcomes**

Gheona Altarescu:  
*Novel insights in Fabry genetics*

Stanislav Kmoch:  
*How to read genetic test results conflicting with clinical evidence*

Olga Azevedo:  
*Fabry disease due to p.F113L mutation*

Ana Jovanovic:  
*Fabry disease due to p.N215S mutation*

*18:00 – 18:10*

## **YOUNG INVESTIGATORS AWARDS**

*18:10 – 18:15*

## **DAY 2 ADJOURN**

Dominique P. Germain

## POSTERS

- 1) Cardiovascular Involvement in Fabry Disease: Association Between Basilar Dolichoectasia and Cardiomyopathy  
*Esteban Calabrese, Guillermo Rodriguez Botta (ARG)*
- 2) Establishment of a patient-specific endothelial cell model to study immune response and endoplasmic reticulum stress in Fabry disease  
*Elisa Rudolph, Malte Lenders, Eva Brand (DEU)*
- 3) Identification of new interaction partners and pathways of  $\alpha$ -galactosidase A to analyse the pathogenesis of Fabry disease  
*Elise Raphaella Menke, Malte Lenders, Eva Brand (DEU)*
- 4) Exploring the journey of patients living with Fabry disease in Poland  
*Agnieszka Brzezinska, Anna Moskal, Chris Wingrove, Jayne Gershkowitz, Adrian Goretzki, Bernadeta Prandzioch-Goretzki, Aleksandra Gladys, Michal Nowicki (GBR)*
- 5) Fabry disease in Argentina: Clinical, biochemical and molecular correlation in all reported GLA variants  
*Juan Politei, Romina Ceci, Domingo Procopio, LUCAS SILVESTROFF, Rita Valdez, Paula Rozenfeld (ARG)*
- 6) Phase III, Open-label, Switch-over Trial of the Efficacy and Safety of Agalsidase Beta Biosidus (Agalzyme®) in Fabry Disease Patients Previously Stabilized with Fabrazyme®  
*Norberto Antongiovanni, Gustavo Cabrera, Alberto A. Fernández, Norberto Guelbert, Juan M. Muraro, Juan M. Politei, Carlos Cusumano, Fernando Gómez Pizarro, Guillermo Guelbert, Sergio Lucca, Erika Nieto, Fernando Perretta, Nicolás M. Antognoni, Sabrina Coppola, Lucía Giménez, Gabriel D. Robbesaul, Gabino Rolandelli, Hugo Sotelo, Viridiana Berstein (ARG)*
- 7) Worse pregnancy and delivery outcomes in children with a pathogenic GLA variant - a post-hoc analysis of the PROFABIA study  
*Christopher Paschen, Natalja Haninger-Vacariu, Constantin Gatterer, Senta Graf, Markus Ponleitner, Paulus Rommer, Raute Sunder-Plassmann, Gere Sunder-Plassmann, Alice Schmidt (AUT)*
- 8) Maternal and Postnatal Outcomes Study (MOS) in patients with Fabry disease receiving pegunigalsidase alfa – study design  
*Joana Lopes de Almeida, Eleonora Riccio, Benedetta Borella, Giovanni Piotti (ITA)*
- 9) Exploring genetic factors of Fabry nephropathy progression via whole-exome sequencing  
*Tina Levstek, Bojan Vujkovic, Albiņa Nowak, João-Paulo Oliveira, Gabriela Dostálová, Aleš Linhart, Markéta Šafaříková, Gheona Altarescu, Katarina Trebušak Podkrajšek (SVN)*
- 10) Electrocardiographic Features Distinguishing Fabry Disease from Sarcomeric Hypertrophic Cardiomyopathy  
*Roberto Giugliani, Fabiano de Oliveira Poswar (BRA)*
- 11) Preliminary Experience of Switching Agalsidase Beta to Pegunigalsidase Alfa in Six Patients in Finland  
*Jukka Saarinen, Päivi Pietilä-Effati, Susanna Kouhi, Minna Kylmälä, Ilkka Kantola (FIN)*
- 12) Quality of life of migalastat-treated adolescents with Fabry disease: results from the ASPIRE study and open-label extension  
*Robert J. Hopkin, Amarilis Sanchez-Valle, Esperanza E. Font-Montgomery, Ozlem Goker-Alpan, Damara Ortiz, Chester B. Whitley, William R. Wilcox, Hai Jiang, Lee Ann Lawson, Jennie Vosk, Haichen Yang, Uma Ramaswami (USA)*
- 13) Clinical assessment of disease severity in patients with Fabry disease treated with pegunigalsidase alfa: an integrated analysis  
*Bojan Vujkovic, Derrallynn Hughes, Aleš Linhart, Eric L. Wallace, William R. Wilcox, Chester B. Whitley, Irene Koulinska, Giovanni Piotti, Raul Chertkoff, Sari Alon, Anat Sakov, Antonio Pisani, John A. Bernat (SVN)*
- 14) Long-term safety and efficacy of migalastat in adolescent patients with Fabry disease: results from the ASPIRE study and open-label extension  
*Uma Ramaswami, Esperanza Font-Montgomery, Ozlem Goker-Alpan, Damara Ortiz, Amarilis Sanchez-Valle, Chester B. Whitley, William R. Wilcox, Hai Jiang, Lee Ann Lawson, Jennie Vosk, Haichen Yang, Robert J. Hopkin (USA)*
- 15) Extending the interval between pegunigalsidase alfa infusions in patients with Fabry disease: five-year interim results from the ongoing BRIGHT51 study  
*Antonio Pisani, John A. Bernat, Myrl Holida, Stephen Waldek, William R. Wilcox, Nicola Longo, Ozlem Goker-Alpan, Eric Wallace, Patrick Deegan, Camilla Tøndel, François Eyskens, Ulla Feldt-Rasmussen, Derrallynn Hughes, Ankit Mehta, Khan Nedd, David G. Warnock, Giovanni Piotti, Meng Wang, Einat Almon-Brill, Sari Alon, Raul Chertkoff, Ales Linhart (ITA)*
- 16) Treatment satisfaction in patients with Fabry disease: patient-reported outcomes from the followME Fabry Pathfinders registry and the SATIS-Fab study  
*Ulla Feldt-Rasmussen, Aleš Linhart, Biliana O. Veleva-Rotse, Joseph D. Giuliano, Hai Jiang, Vipul Jain, Caroline Martinez, Olivier Lidove, on behalf of the followME Fabry Pathfinders registry and the SATIS-Fab study investigators (CZE)*

- 17) **Additional value of standard ECG to echo staging in prediction on cardiovascular risk in Anderson Fabry disease**  
*Serena Serratore, Maria Alessandra Schiavo, Raffaello Ditaranto, Vanda Parisi, Rosa Lillo, Annamaria Del Franco, Emanuele Monda, Pier Paolo Bocchino, Nevio Taglieri, Claudia Raineri, Chiara Leuzzi, Andrea Barison, Ines Monte, Cinzia Zuchi, Daniele Torella, Francesco Cappelli, Maurizio Pieroni, Francesca Graziani, Iacopo Olivotto, Giuseppe Limongelli, Elena Biagini (ITA)*
- 18) **Evaluating the relationship between infusion-related reactions and antidrug antibody status: results from 109 patients with Fabry disease treated with pegunigalsidase alfa**  
*Robert J. Hopkin, Derrallynn Hughes, John A. Bernat, Aleš Linhart, Nicola Longo, Camilla Tøndel, Bojan Vujkovic, Antonio Pisani, Jasmine Knoll, Irene Koulinska, Giovanni Piotti, Raul Chertkoff, Sari Alon, Anat Sakov, Eric L. Wallace (USA)*
- 19) **Echocardiographic Analysis: Factors Affecting Mitral Regurgitation in Fabry Disease**  
*Alena Vecerova, Gabriela Dostalova, Beata Soltesova, Ales Linhart (CZE)*
- 20) **Uncovering the Fabry Disease treatment experience: a survey of US patients receiving enzyme replacement therapy (ERT)**  
*Robert J. Hopkin, Angel Rosas, Irene Koulinska, Lucas Oliveira (USA)*
- 21) **Real-World Patient Experiences of Enzyme Replacement Therapy (ERT) in Adults with Fabry Disease: A UK Market Research Study**  
*Derrallynn Hughes, Gisela Wilcox, Alison Muir, Tom Kenny, Kamran Iqbal, John Ndikum (GBR)*
- 22) **Novel insights on blood pressure variability impacts on cardiovascular manifestations of Fabry Disease**  
*Jan Pudil, Josef Marek, Gabriela Dostalova, Kristyna Bayerova, Beata Soltesova, Ales Linhart (CZE)*
- 23) **The Price of Not Diagnosing Fabry Disease in Puerto Montt, Chile**  
*Benjamin Solar, Juan Politei, Felipe Mendez (CHL)*
- 24) **Novel Variant in GLA and Classic Fabry Disease: Defining an Aggressive Phenotype in Southern Chile**  
*Benjamin Solar, Juan Politei (CHL)*
- 25) **Trying to Reducing the Burden of Fabry Disease: The Most Southern Chilean Experience**  
*Benjamin Solar, Joaquin Cerda, Ivonne Zamorano, Nicole Rivera, Felipe Mendez (CHL)*

## NOTES

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## **Registration**

online at [www.horizons-fabry.com/registration](http://www.horizons-fabry.com/registration)

| Registration fees                                 | On-site |
|---|---------|
| REGULAR registration                              | 600 €   |
| POSTER 1 <sup>st</sup> Author under the age of 35 | 180 €   |

The Registration fee includes:

- access to all sessions,
- access to the industry exhibition,
- programme booklet,
- all conference meals (coffee breaks and lunches).

## **Accommodation**

Vienna House by Wyndham Diplomat Prague  
Evropská 15 | Prague | Czech Republic



PLATINUM PARTNERS:

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