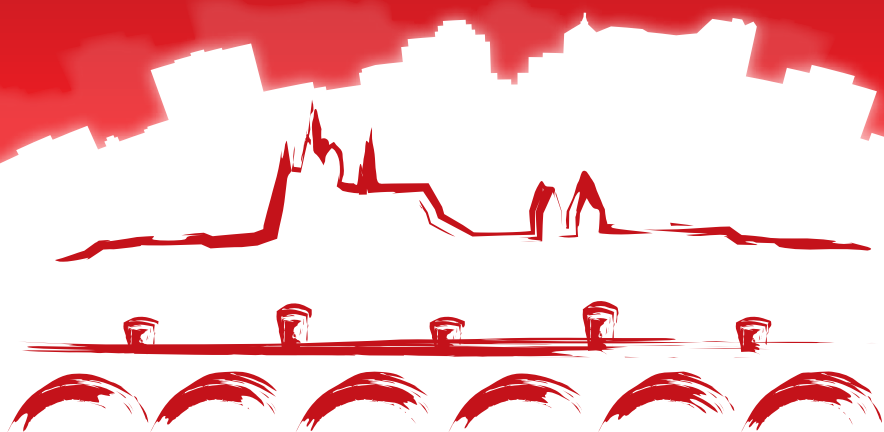


NEW HORIZONS IN FABRY DISEASE

INTERNATIONAL CONFERENCE ON ADVANCES
IN THE TREATMENT OF FABRY DISEASE



PRAGUE, CZECH REPUBLIC
MAY 26–27, 2023

HELD UNDER THE AUSPICES OF
THE CZECH MEDICAL ACADEMY



ENDORSED BY

ICON
International Cardiomyopathy
Network

WWW.HORIZONS-FABRY.COM

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



Endorsed by

Centre for Fabry Disease
General University Hospital
1st Faculty of Medicine,
Charles University
Prague, Czech Republic

French Reference Center
for Fabry Disease (Geneo)
University of Versailles
France



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
(University College of London, London, United Kingdom)



European
Reference
Networks

SCIENTIFIC PROGRAMME

Friday May 26, 2023

8:00 – 9:00 REGISTRATION

9:00 – 9:10 WELCOME AND INTRODUCTION

Aleš Linhart (CZ), Dominique P. Germain (FR), Perry M. Elliott (GB)

9:10 – 9:40 **KEYNOTE LECTURE**

Chair: Aleš Linhart (CZ), Dominique P. Germain (FR), Perry M. Elliott (GB)

Looking back on 20 years of clinical registries in Fabry disease

Christoph Wanner (DE) 30'

9:40 – 10:00 BREAK

10:00 – 11:40 **SESSION 1**

ESSENTIAL AND OVELOOKED INVOLVEMENT IN FABRY DISEASE

Chair: Alessandro Burlina (IT), Fran Borovečki (HR)

Brain in Fabry disease

Alessandro Burlina (IT) 20'

Peripheral neuropathy

Vincenzo Donadio (IT) 20'

Gastrointestinal involvement in Fabry disease

Juan Manuel Politei (AR) 20'

Microbiome in Fabry disease

Alberto Ortiz (ES) 20'

Q & A 20'

11:40 – 12:00 BREAK

12:00 – 13:20 **SESSION 2**

CURRENT DIAGNOSTIC TOOLS AND CONCEPTS – PART I

Chair: Robert Hopkin (US), Juan Manuel Politei (AR),

Alberto Ortiz (ES)

Pathogenicity of GLA mutations and variants

Dominique Germain (FR) 20'

X chromosome inactivation

Fran Borovečki (HR) 20'

Critical points in understanding pathogenicity and X chromosome inactivation

João Paulo Oliveira (PT) 20'

Panel discussion

Q & A 20'

13:20 – 14:20 LUNCH

14:20 – 16:00 SESSION 3

CURRENT DIAGNOSTIC TOOLS AND CONCEPTS – PART II

Chair: Bojan Vujkovic (SI), Aleš Linhart (CZ), Gheona Altarescu (IL)

Lyso Gb3 as a diagnostic tool and for disease monitoring

David Kasper (AT) 20'

Lyso Gb3 for disease severity staging

Albina Nowak (CH)..... 20'

Kidney biopsy

Camilla Tøndel (NO) 20'

Endomyocardial biopsy

Cristina Basso (IT) 20'

Panel discussion

Q & A 20'

16:00 – 16:20 BREAK

16:20 – 18:00 SESSION 4

CURRENT DIAGNOSTIC TOOLS AND CONCEPTS – PART III

Chair: João Paulo Oliveira (PT), Alberto Ortiz (ES),

Sima Canaan-Kühl (DE)

Glomerular filtration as the measure of treatment efficacy

Bojan Vujkovic (SI)..... 20'

Whole blood transcriptomic profiling in Fabry disease

Gheona Altarescu (IL) 20'

Fabry is a pediatric disease

Robert Hopkin (US)..... 20'

Screening programs in Fabry disease

Roberto Giugliani (BR) 20'

Panel discussion

Q & A 20'

Saturday May 27, 2023

9:00 – 10:40

SESSION 5

KEY CLINICAL ISSUES IN CURRENT THERAPIES

Chair: Derralyann Hughes (GB), Albina Nowak (CH), Dominique P. Germain (FR)

Long-term experience with enzyme replacement therapies

Dominique P. Germain (FR) 20'

Anti-drug antibodies

Malte Lenders (DE) 20'

Treatment choices – ERT vs. Chaperones, long-term experience with chaperone therapy

Derralyann Hughes (GB)..... 20'

Amenability testing

Malte Lenders (DE) 20'

Panel discussion

Q & A 20'

10:40 – 11:00

BREAK

11:00 – 12:00

SESSION 6

NEW CONCEPTS IN FABRY PATHOPHYSIOLOGY

Chair: Perry Elliott (GB), Stanislav Kmoch (CZ), Malte Lenders (DE)

Endoplasmic reticulum stress as a disease-causing mechanism

Moran Dvela-Levitt (IL) 20'

Agalopathy – a novel disease-causing mechanisms in Fabry disease

Martina Živná (CZ) 20'

Panel discussion

Q & A 20'

12:00 – 13:20

LUNCH

13:20 – 15:00

SESSION 7

NOVEL THERAPIES

Chair: Derralyynn Hughes (GB), Alberto Ortiz (ES), Raphael Schiffmann (US)

Pegunigalsidase alfa

Aleš Linhart (CZ)..... 20'

Substrate reduction therapies

Dominique P. Germain (FR) 20'

Gene therapy

Raphael Schiffmann (US)..... 20'

Chaperone development in Fabry disease

Saida Ortolano (ES) 20'

Panel discussion

Q & A..... 20'

15:00 – 15:20

BREAK

15:20 – 17:00

SESSION 8

THE HEART IN FABRY DISEASE

Chair: Ilkka Kantola (FI), Aleš Linhart (CZ), Mehdi Namdar (CH)

Heart failure – the key player in Fabry disease morbidity and mortality

Josef Marek (CZ) 20'

Arrhythmias – diagnosis and management

Mehdi Namdar (CH)..... 20'

Imaging in Fabry disease

Ruxandra Jurcut (RO) 20'

Differential diagnosis of Fabry cardiomyopathy

Perry Elliott (GB)..... 20'

Panel discussion

Q & A..... 20'

17:00 – 17:10

WRAP-UP AND ADJOURN

POSTERS

- 1) **Head-To-Head Trial of Pegunigalsidase Alfa vs. Agalsidase Beta in Fabry Disease: Phase 3 Randomized, Double-Blind, BALANCE Study 2-Year Results**
Derrallynn Hughes, John Bernat, Eric L. Wallace, Ozlem Goker-Alpan, William R. Wilcox, Myrl Holida, Nicola Longo, Pilar Giraldo, Maria Judit Molnar, Damara Ortiz, Robert J. Hopkin, Camilla Tøndel, Aleš Linhart, Patrick Deegan, Ana Jovanovic, Michael Muriello, Bruce A. Barshop, Virginia Kimonis, Bojan Vujkovic, Albina Nowak, Tarekegn G. Hiwot, Antonio Pisani, Dominique P. Germain, Ilkka Kantola, Jasmine Knoll, Ankit Mehta, Stephen Waldek, Amel Karaa, Einat Almon, Sari Alon, Raul Chertkoff, Rossana Rocco, David G. Warnock
- 2) **Long-Term Safety and Efficacy of Pegunigalsidase Alfa Administered Every 4 Weeks in Fabry Disease: 2-Year Interim Results from BRIGHT51**
Camilla Tøndel, John Bernat, Myrl Holida, Nicola Longo, Ozlem Goker-Alpan, Eric Wallace, Patrick Deegan, François Eyskens, Ulla Feldt-Rasmussen, Derrallynn Hughes, Antonio Pisani, Ales Linhart, Rossana Rocco, Einat Brill Almon, Sari Alon, Raul Chertkoff, David G. Warnock, Stephen Waldek, William R. Wilcox
- 3) **FollowME Fabry Pathfinders Registry: multisystemic effectiveness in a multinational, multicenter cohort of patients on migalastat treatment for at least 3 years**
Ana Jovanovic, Derrallynn Hughes, Gere Sunder-Plassmann, Eva Brand, Michael L. West, Daniel G. Bichet, Antonio Pisani, Albina Nowak, Roser Torra, Aneal Khan, Olga Azevedo, Anna Lehman, Aleš Linhart, Jasmine Rutecki, Joseph D. Giuliano, Eva Krusinska, Peter Nordbeck
- 4) **Clinical characteristics of female patients enrolled in the FollowME Fabry Pathfinders registry**
Ana Jovanovic, Peter Nordbeck, Antonio Pisani, Albina Nowak, Ulla Feldt-Rasmussen, Eva Brand, Derrallynn Hughes, Daniel G. Bichet, Michael L. West, Kathleen Nicholls, Robert J. Hopkin, Roberto Giugliani, Juan Politei, Biliانا O. Veleva-Rotse, Jasmine Rutecki, Joseph D. Giuliano, Eva Krusinska, Gere Sunder-Plassmann
- 5) **Estimation of arrhythmia risk in patients with Fabry disease using a machine learning model**
Patrício Aguiar, John L. Jefferies, Giorgio P. Biondetti, David Warnock, Staci Kallish, Matthew W. Nelson, Joseph D. Giuliano, Joseph W. Zabinski, Costas Boussios, Gary Curhan, Jigar N. Bandaria, Richard E. Gliklich
- 6) **Estimation of stroke risk in patients with Fabry disease using a machine learning model**
Patrício Aguiar, John L. Jefferies, Staci Kallish, Giorgio P. Biondetti, Matthew W. Nelson, Joseph D. Giuliano, Joseph W. Zabinski, Costas Boussios, Gary Curhan, Jigar N. Bandaria, Richard E. Gliklich, David Warnock

- 7) **New renal and cardiac cellular models of Fabry disease exploiting CRISPR/Cas9 technology**
Maximiliano E. Ormazabal, Eleonora Pavan, Emilio Vaena, Dania Ferino, Maurizio Scarpa, Paula A. Rozenfeld, Andrea Dardis
- 8) **Fabry Nephropathy at the time of diagnosis in Argentina**
Fernando Perretta, Norberto Antongiovanni, Sebastián Jaurretche
- 9) **A Validated Model for the Prediction of Adverse Cardiac Outcome in Patients with Fabry Disease derived from readily available clinical data**
Christopher Orsborne, Joshua Bradley, Laura Bonnett, Thomas Anderson, Nik Abidin, Peter Woolfson, Anna Reid, Matthias Schmitt, Christopher Miller, Ana Jovanovic
- 10) **Validated Model for Prediction of Adverse Cardiac Outcome in Patients With Fabry Disease**
Christopher Orsborne, Joshua Bradley, Laura Bonnett, Luke Pleva, Josephine Naish, David Clark, Nik Abidin, Peter Woolfson, Gaetano Nucifora, Matthias Schmitt, Ana Jovanovic, Christopher Miller, Anna Reid
- 11) **Inflammatory Fabry cardiomyopathy demonstrated using simultaneous hybridised 18F-fluorodeoxyglucose positron emission tomography, cardiovascular magnetic resonance imaging**
Christopher Orsborne, Jose Anton-Rodriguez, Neal Sherratt, Amy Watkins, Maelene Lohezic, David Clark, William Lloyd, Josephine Naish, Peter Woolfson, Anna Reid, Matthias Schmitt, Sivakumar Muthu, Parthiban Arumugam, Ana Jovanovic, Christopher Miller
- 12) **Native myocardial T1 dispersion in Fabry disease: comprehensive evaluation of a novel measurement of cardiovascular disease severity**
Christopher Orsborne, Luke Pleva, Josephine Naish, David Clark, Nik Abidin, Peter Woolfson, Gaetano Nucifora, Matthias Schmitt, Ana Jovanovic, Ana Jovanovic, Anna Reid, Christopher Miller
- 13) **Fabry and Neutrophil Extracellular Traps (NETs) Study of Neutrophil Extracellular Traps (NETs) in the development of thrombotic complications in Fabry disease patients.**
Irene Serrano-Gonzalo, Sonia Roca-Esteve, Laura López de Frutos, Carlos Lahoz, Ralf Köhler, Pilar Giraldo
- 14) **Cardiac manifestations prevalence is underestimated in females with Fabry disease**
Maryam Banikazemi
- 15) **Patient-reported experiences with Fabry disease monitoring and disease burden in the real-world setting: results from a double-blind, cross-sectional survey**
Xavier Lloria, Lisa Berry, Jack Johnson, Julia Alton, Irene Koulinska, Dawn Laney

- 16) High prevalence of cardiac findings in individuals with Fabry disease identified via no-charge sponsored testing programmes

Biliana O. Veleva-Rotse, Joseph D. Giuliano, Katherine L. McPherson, Andrew T. Willcock, Heather M. McLaughlin

GENERAL INFORMATION

Conference Venue

Corinthia Hotel Prague

Kongresová 1655/1 • Prague • Czech Republic

Conference Dates

May 26th & 27th, 2023

Registration

online at www.horizons-fabry.com/registration

Registration fees

	Standard (1. 2. – 24. 5.)	Late / On-site (25. 5. – 27. 5.)
REGULAR registration	500 €	600 €

The Registration fee includes:

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

Accommodation

Corinthia Hotel Prague • Kongresová 1655/1 • Prague • Czech Republic

Room	Price
Superior – SGL	170 €
Superior – DBL	95 €

The prices stated are per person per night, including tax, for room.

The hotel capacity is limited.

Cancellation

Cancellation fees are set according to the hotel's policy.

The accommodation can be cancelled only in writing (e-mail).

PLATINUM PARTNER:

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SILVER PARTNERS:



PARTNER:



ORGANIZING AGENCY:

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