FIN FABRY EXPERT MEETING 2024

April 19-20, 2024

Prague, CZ

FIN

Fabry International Network

FIN FABRY EXPERT MEETING 2024

Friday April 19

09:30 - 12:30

SHARING BEST PRACTICES*

12:30 - 14:00

LUNCH*

14:00 - 16:30

SHARING BEST PRACTICES*

17:00 - 18:00

FIN AGM*

19:00

WELCOME DINNER*

**For members only*

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FIN FABRY EXPERT MEETING 2024

Saturday April 20

09:00 - 09:15

WELCOME

09:15 - 09:45

THE LATEST ON FABRY AND THE HEART

PROF ALEŠ LINHART

09:45 - 10:15

STANDARD OF CARE

DR UMA RAMASWAMI

10:15 - 10:45

PROM'S AND PREM'S IN FABRY DISEASE

DR BOJAN VUJKOVAC

10:45 - 11:00

BREAK

11:00 - 11:30

FABRY FEMALES: WHAT IS THE SAME AS MALES & WHAT'S NOT

DR ROBERT HOPKIN

11:30 - 12:00

WHAT WE DON'T KNOW ABOUT FABRY YET

DR ALBERTO ORTIZ

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FIN FABRY EXPERT MEETING 2024

Saturday April 20

12:00 - 12:30

PANEL DISCUSSION

12:30 - 13:30

LUNCH

13:30 - 14:00

CLINICAL TRIALS UPDATE SUMMARY

PROF DOMINIQUE GERMAIN

14:00 - 14:30

MENTAL HEALTH, TRANSITIONING TO ADULTHOOD AND THE FABRY FOG

DR NADIA ALI

14:30 - 14:45

CLOSING REMARKS

14:45 - 15:15

BREAK

15:15 - 17:30

WORKSHOPS*

19:00

GROUP DINNER

**For members only*

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Meet the experts!

Prof. Aleš Linhart, DrSc, FESC graduated at Charles University in **Prague, Czech Republic.** Subsequently he received his training in cardiology and vascular medicine at General University Hospital in Prague and Broussais Hospital in Paris, France.

Moving back to the First medical faculty of the Charles University in Prague, he continued his professional career and research focusing mainly on metabolic cardiomyopathies, noninvasive cardiac imaging and atherosclerosis. In 2004 he was appointed professor at Charles University in Prague.

He is currently heading the Department of Cardiovascular Medicine of the First Medical Faculty at Charles University and General University Hospital in Prague . Prof. Linhart authored or co-authored more than 240 papers and reviews, 18 book chapters and 4 monographs. Currently he holds a position of a chairman of the Czech Society of Cardiology.

Dr Uma Ramaswami is a consultant in Inherited Metabolic Disorders with over ten years' experience in the management of metabolic disorders, with a focus on Lysosomal storage disorders. As a consultant at the Lysosomal Disorders Unit at the Royal Free Hospital, her current role includes the management of adolescence and young adults with Lysosomal disorders (LSD)including the transition from paediatric centres to and supporting the clinical trail unit. She trained in paediatric metabolic disorders at Great Ormond Street and Temple Street children's hospital in Dublin.

She is currently the National Clinical lead for the Paediatric Familial Hypercholesterolaemia Register in the **UK**. She has currently secured a European grant to expand it as an international registry, to understand the natural history of familial Hypercholesterolaemia and to asses the outcomes and safety of starting statins in childhood.

She is currently actively involved in teaching at undergraduate and postgraduate level in metabolic medicine. Having completed a Doctorate in Medicine from University College London, she has an active clinical research programme in Lysosomal Storage Disorders – with an emphasis on identification of early disease severity markers, developing severity scoring tools in rare diseases and in translational research on the effects of new therapies, with peer reviewed publications.

Dr Ramaswami feels privileged to have the opportunity to do free clinics in India for children and adults with metabolic disorders, including Lysosomal Storage Disorders.

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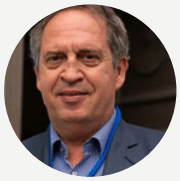
Meet the experts!



Dr Bojan Vujkovac MD, is currently working as a nephrologist at General Hospital Slovenj Gradec in **Slovenia.** He got his medical degree and residency at University Clinical Center in Ljubljana. He started his carrier in 1987 when he took his position at Dept. of Nephrology and Dialysis in the GH Slovenj Gradec and from 1997 to 2021 he was also a head of the department. In 1997, he started working on Fabry disease and in 2004 he founded a national Fabry Center at his hospital where he is still a head of the Center. In 2007 he became a member of European Fabry Registry Advisory Board.

His primary areas of scientific interest include Fabry disease diagnosis, treatment and management and CKD epidemiology and progression factors. He is currently a Principal Investigator of numerous trials and has authored over 100 original research and review articles in national and international papers and chapters in textbooks and presented his work at several scientific conferences.

He is a first author of Slovenian Fabry guidelines published already in 2006 and preparing an update in 2021. He is also ad hoc reviewer for several international scientific journals. He organized and chaired many scientific events at national and international level. He is an organizer and chair for traditional international annual Fabry School, which is held in GH Slovenj Gradec since 2008. He is a member of several working groups regarding management of rare diseases at the Ministry of Health of Slovenia.

Dr Robert J. Hopkin, is an associate professor of clinical pediatrics at Cincinnati Children's Hospital Medical Center within the University of **Cincinnati** College of Medicine. Dr Hopkin graduated from the University of Nevada Medical School. He completed residency and chief residency in pediatrics at the Phoenix Children's Hospital, Maricopa Medical Center Combined Residency Program. His training in medical genetics was completed at Cincinnati Children's Hospital Medical Center.

The majority of Dr. Hopkin's time is spent in caring for patients with genetic disorders. He participates in clinics from Fetal Care to Adult Genetics. He is also actively involved in education of health care providers regarding the application of genetics for patient care. Dr Hopkin has participated in a number of clinical trials and is a member of American College of Medical Genetics Committee on Therapeutics.

He has participated in natural history studies on Fabry disease, Pompe disease, velocardiofacial syndrome, Pierre Robin sequence, neurofibromatosis type I, and several other genetic conditions. The unifying principle in his research interests is application of scientific knowledge to improve outcomes for patients afflicted with genetic disorders.

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Meet the experts!

Dr Alberto Ortiz is Chief of Nephrology and Hypertension at the Health Research Institute of the Jiménez Díaz Foundation (IIS-FJD UAM), **Madrid, Spain**, and Professor of Medicine at the Autonomous University of Madrid (UAM). Professor Ortiz's research interests include the pathogenesis and treatment of acute kidney injury, diabetic nephropathy and proteinuric chronic kidney disease, vascular injury in kidney disease, Fabry disease, and systems medicine.

He is Editor-in-Chief of the Clinical Kidney Journal, an editorial board member of the Journal of the American Society of Nephrology, a council member of the European Renal Association (ERA-EDTA), a member of European Uremic Toxins (EUTox) Work Group and European Renal Best Practice (ERBP) Organization, coordinator of the Spanish Renal Research Network (REDINREN), ERA-EDTA Award for Research Excellence 2020, Distinguished Fellow of the ERA-EDTA, and corresponding member of the Spanish Royal National Academy of Medicine.

Prof Dominique P. Germain is Professor of Medical Genetics at the University of Versailles (UVSQ), and head of the Division of Medical Genetics at the Raymond Poincaré Hospital (AP-HP) in **Garches, France.** He is also Director of the French Referral Center for Fabry disease in Garches, France. In 2016, Prof Germain was appointed Director of a European Referral Centre for rare disorders (MetabERN: rare metabolic diseases) at the Hôpitaux Universitaires Paris Ile de France Ouest.

In 2017, he also became the Director of a Centre of Expertise for developmental disabilities band rare disabling congenital malformations. He has a strong interest in enzyme replacement therapies, chaperon therapies, the genetics of neuro-muscular diseases, and the use of clinical exome sequencing for deciphering unknown disabling genetic diseases.

Throughout his career, Prof Germain has contributed extensively on the topics of Fabry disease and hereditary diseases of connective tissue. He has written several book chapters and has published over 130 peer-reviewed papers in medical journals.

Dr Nadia Ali is Director of Psychological Resources for the Emory Genetic Clinical Trials Center, where she participates in clinical drug trials for genetic conditions, as well as conducting her own clinical research into neurocognitive and psychological manifestations of lysosomal storage diseases and other metabolic disorders. She is co-author of the book, "Transitions: Managing Your Own Healthcare: What Every Teen with an LSD Needs to Know."

Dr. Ali also serves as Co-Assistant Director of the Emory Genetic Counseling Master's degree training program, where she trains future genetic counselors in the counseling skills necessary for compassionate, effective patient care. Dr. Ali earned her doctorate in Clinical Psychology from the University of South Florida and completed both an internship in Neuropsychology and a post-doctoral fellowship in Health Psychology.

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Empowering people living with Fabry

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