FIN FABRY EXPERT MEETING 2023

April 21-23, 2023

Amsterdam, NL

FIN

Fabry International Network

FIN FABRY EXPERT MEETING 2023

Friday April 21

10:00 - 15:00

WORKSHOPS*

17:00-18:00

FIN AGM*

19:00

WELCOME DINNER

**For members only*

Fabry International Network

FIN FABRY EXPERT MEETING 2023

Saturday April 22

09:30-09:45

OPENING & WELCOME
FIN

09:45-10:15

TESTIMONIAL
ANNELIES SWEEB & ERICA VAN DE MHEEN

10:15-10:30

BREAK

10:30-11:15

DR ROBERT HOPKIN

11:15-12:00

FABRY FOG
DR NADIA ALI

12:00-12:30

UPDATE FABRY POLAND
WOJCIECH NADOLSKI

12:30-14:00

LUNCH

**For members only*

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Saturday April 22

14:00-14:45

DISCUSSION ON THE AVAILABILITY AND USE OF BIOMARKERS FOR (EARLY) DETECTION OF CARDIAC DISEASE ASSOCIATED WITH FABRY
PROF FRANCOIS EYSKENS

14:45-15:30

ARRHYTHMIA IN FABRY DISEASE
DR ASHWIN ROY

15:30-16:00

PANEL DISCUSSION DR ROY & PROF EYSKENS

16:00-16:30

BREAK

16:30-18:00

SHARING BEST PRACTICES*

18:45

GROUP PICTURE

19:00

GROUP DINNER

**For members only*

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Sunday April 23

09:30-10:15

THE GLA GENE: FABRY DISEASE AND WOMEN
PROF GERMAIN

10:15-11:00

GENE THERAPY
PROF HUGHES

11:00-11:30

PANEL DISCUSSION PROF HUGHES & PROF GERMAIN

11:30-12:00

D313Y – AN EXAMPLE OF A CONTROVERSIAL FABRY VARIANT
DR BERTHOLD WILDEN

12:00-13:00

LUNCH

13:00-13:45

KIDNEY DISEASE: WHAT IS NEW?
DR ALBERTO ORTIZ

13:45-14:30

FABRY AND BEST NUTRITION PRACTICES
DR SEEMA KANWAL

14:30-14:45

FABRY AUSTRALIA - FABER WITH THE DRAGON
DIANNE WALLYN

14:45-15:00

CLOSING REMARKS
FIN

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Meet the experts!

Annelies Sweeb is 61 years old and Fabry affected. She is the President of FSIGN and mother of two daughters. **Erica van de Mheen** is 66 years old, Fabry affected, she is the Vice President for FSIGN and mother of two daughters. Both are living in **The Netherlands.**

Dr Ashwin Roy is a Cardiology registrar from **Birmingham, UK** undertaking a PhD investigating arrhythmia in Fabry Disease. He is involved in a multi-centre clinical trial assessing the use of implantable loop recorders in Fabry Disease. He is also undertaking laboratory based work using iPSC-derived cardiomyocytes with the N215S GLA mutation. His particular interest is imaging markers of arrhythmia.

Dr Nadia Ali is Director of Psychological Resources for the Emory Genetic Clinical Trials Center in **Atlanta, USA** 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.

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.

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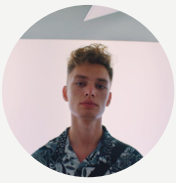
Meet the experts!

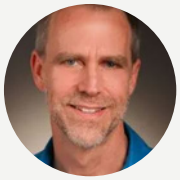
Dr Seema Kanwal has been a practicing naturopathic doctor in Vancouver since 2006 and is the only Naturopathic doctor trained in **Canada** in the Apo E Gene diet. She has extensive experience with and is passionate about working with individuals who have rare genetic diseases and suffer many stress related conditions as a result such as mental health issues, insomnia, and low energy.

She helps to guide people living with rare diseases to increased mental wellness, energy, vitality and an overall higher standard of life – they often didn't know was possible to experience with their condition.

She has 10 years of experience in sport medicine and treating a wide range of conditions such as cancer, hormonal imbalances for women and men, stress related conditions, pre and post natal care, cardiovascular disease, chronic fatigue, autoimmune, thyroid imbalances, digestive issues (IBS, Chron's, Candida), Alzheimer's (through the APOE Gene), sports related injuries and many more.



Wojciech Nadolski is a representative of the Association of Families with Fabry Disease in **Poland**, actively working to improve the situation of patients with Fabry disease. Wojciech has been affected by Fabry since his birth. He is a passionate photographer and traveller.



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!

Prof. Derralynn Hughes is Professor of Experimental Haematology at the University College **London**, Clinical Director of Research and Innovation at the Royal Free London NHS Foundation Trust, and Co-Clinical Director of the NCL cancer Alliance. She is also chair of the European Working Group on Gaucher Disease.

She has clinical responsibilities in the area of Haematology and Lysosomal Storage Disorders and is chair of the anaemia clinical practice group. She directs the research programme in the LSD unit where interests include understanding the pathophysiology of phenotypic heterogeneity in Fabry Disease and bone related pathology in Gaucher disease and malignancy.

Prof Hughes is Principal Investigator of a number of clinical trials examining the efficacy of Enzyme, Chaperone and gene therapies and other new agents in the treatment of Gaucher, Fabry, Pompe and MPS disorders. A particular interest relates to the clinical and biological effects of bone disease and malignancy in Gaucher disease. She is an author of over 150 papers in the area of macrophage biology and lysosomal Storage Disorders.

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.

Dianne Wallyn received the unexpected Fabry diagnosis in 2009 after her sister pursued a diagnosis to heart problems. A Brisbane cardiologist first suggested Fabry disease but was obliged to eliminate all other possibilities before requesting the genetic test. After her sister's positive diagnosis, Dianne was told she had a 50 percent chance of having the disease. Ever the optimist she assumed the best, however several weeks later the call came in to say that she had also tested positive.

Dianne is incredibly grateful for the support she received from Fabry **Australia** in those early days and is pleased to serve on the board to assist others with this disease. She calls the Sunshine Coast home and enjoys a laidback country lifestyle and she shares this with her husband and two very spoilt dogs.

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Meet the experts!

Prof François J. M. Eyskens, MD, PhD is the head of the department for the Metabolic Disorders in children (University Hospital of Antwerp, BE). Center of Inherited Metabolic Diseases, Metabolic Lab (PCMA vzw). He did his medical studies at the University of Antwerp. After his training in paediatrics at the Kinderziekenhuis Antwerpen, he became a pediatrician in 1989. His training in metabolic diseases took place in the Wilhelmina Kinderziekenhuis, Utrecht, The Netherlands. From 1987 till now he is involved in the neonatal mass screening program in Antwerp at the Provinciaal Centrum voor Metabole Aandoeningen (PCMA).

In 1997 he obtained his Degree of Doctor in Medical Sciences (PhD) defending his thesis "Neonatal Screening. The Experience in Antwerp". Actually he is working as a chief of clinics in Paediatrics at the University Hospital of Antwerp (UZA) and coordinates the Center of Metabolic Diseases (CEMA) located at the sites of UZA and ZNA.

He is the executive director of the PCMA vzw, the metabolic laboratory, where the analysis of different metabolic compounds is performed. He is an associate professor at the University of Antwerp (Dept of Medicine; Laboratory of Experimental Medicine and Pediatrics). Specific domains of interest and research: creating awareness and screening for inherited metabolic diseases in the neonatal period and in populations at risk ; organic acidurias; lysosomal storage diseases (innovative therapies); learning and cognitive disturbances due to inherited metabolic diseases; psychiatric disturbances as first clinical presentation of inherited metabolic diseases.

Dr Berthold Wilden was diagnosed with Fabry in 2013 and has been a dialysis patient since 2017. He is the President of Morbus Fabry Selbsthilfegruppe (MFSH), i.e. the **German** Fabry Disease Patient Organization, since 2020. However, he is actively working in the group in various positions since 2015. After school, Berthold worked as a chemical lab technician before starting to study chemistry. After his first exams, he switched to biochemistry, where he also earned his PhD. He then worked in the field of clinical and later preclinical research, mainly in quality assurance. Since 2020, Berthold has no longer worked and is now an early retiree.

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Meeting Notes

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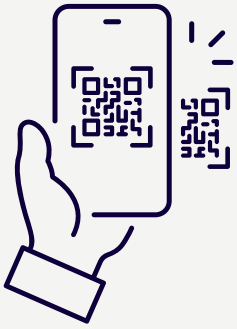
Thank you for participating in the meeting – your feedback is important to us!

Please take a few moments to complete this survey to make sure future meetings can be improved!



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Thank you to our partners!

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