FABRY
International Network

Amicus Therapeutics

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Fabry international Network
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Fabry International Network

Welcome you to our

6th Fabry Expert Meeting

9th – 10th June 2018

The Vilnius Grand Resort & Conference Centre, Vilnius, Lithuania

The primary aim of the Fabry International Network is to facilitate collaboration between patient organisations around the world to support those affected by Fabry disease

This year the Fabry International Network's Expert Meeting takes place in Vilnius, Lithuania with over 84 registered delegates.

Over the past six years this expert meeting has become an esteemed event for physicians, pharmaceutical companies and Fabry patient organisations. Here, they have the chance to learn the latest state of the art in new therapies, research, clinical trials and many other aspects relating to Fabry Disease. It is an exclusive setting which brings together the leaders of a number of Fabry patient organisations from across the globe with Fabry specialists to provide them with the opportunity to network and share their experiences.

The Fabry International Network Board of Directors would like to thank Amicus, Protalix, Sanofi and Shire for their support in making this Expert Meeting possible once again.

Vilnius Grand Resort, Ežeraičių g. 2, Ežeraičių km., Avižienių sen., Vilniaus raj., LT-14200, Lietuva

Amenities include a lounge, seating space and café. Bedrooms offer free Wi-Fi, flat-screen TVs and seating areas. Room service is available 24/7. Check in at 15.00 and check out by 11.00.

Speakers

Jacqueline Adam MPS Commercial, Amersham, United Kingdom
Jacqueline Adam is the Clinical Communications Lead at MPS Commercial, a wholly owned, not for profit subsidiary of the UK MPS Society. In this role she oversees the development of research surveys and communications on MPS and related disorders. Before joining MPS Commercial, Jacqueline was a medical writer in both the medical communications industry and charity sector. She has a BSc in Pharmacology from the University of Dundee and a PhD in Biochemistry from University College London.

Christopher Armastrong Fabry Patient Alberta, Canada
Chris Armstrong was born and raised in Airdrie, Alberta, Canada. He started displaying signs of Fabry disease around the age of 12 but was not officially diagnosed until the age of 25. He lives with Fabry disease daily however the treatments he follows allows him to function with only minor disruptions to his daily activities. He works for the Bell Helicopter Canadian Supply Centre supporting the Canadian Military's CH146 Griffon Program as an Order Processor. He enjoys time with his family and friends who are there for him when the need arises. Since learning that his family has this disease, and about the symptoms he endures, Chris has been interested in taking tests and helping when he can to assist the specialists find a cure for everyone with genetic deficiencies.

Rebecca Brandon MPS Society, Amersham, United Kingdom
Rebecca Brandon is an Advocacy Support Officer at the MPS Society. She joined the Society in July 2010. Her main areas of responsibility are working with members who suffer with Fabry, MPSII, MLIII/MLIV, Mannosidosis and Fucosidosis. The society has over 450 Fabry members and Rebecca becomes involved with families from diagnosis, assisting within all areas of difficulties encountered dealing with the disease. She has held focus groups for Fabry members to help clinicians and nursing staff gain a better understanding of the impact of the disease on individuals. Rebecca has a background of over 21 years in Local Authority and Private Sector Housing dealing with vulnerable individuals with learning disabilities, mental health and the physically disabled.

Martynas Davidonis Lithuania
Martynas was born in Lithuania. When 4 years old he became unwell and spent much time in hospital. This introduced him to medicine. After graduating from school, Marynas started studying medicine at Vilnius University. During two years of hemodialysis he received his Fabry diagnosis, treatment & a kidney transplant in 2008. Martynas was at that time the only diagnosed Fabry patient in Lithuania now there are more than 10. In 2011 he graduated becoming a medical doctor & later graduated in medicine & forensic medicine. In 2012 he started studying law & hopes for a diploma in July 2018. Martynas has been giving biochemistry lectures for over 5 years at the Vilnius University Faculty of Medicine, attending Fabry patient meetings for 10 years & in October 2016 became a board member of Fabry International Network.

Lut De Baere BOKS,Belguim
is a nurse by formation, but stopped working in 1987, when her two sons were diagnosed with a metabolic disease (MMA). In 1994, Lut was the founder and became president of BOKS - the Belgian umbrella patient organisation for all metabolic diseases. She is member of several committees of the Belgian government: NBS, workgroups of chronic and rare diseases, …. The international landscape and working with all different stakeholders has no secrets for her. Lut is happy to have been given the opportunity to work with other, very skilled people with the same interests and concerns. She is coordinator of the Patient Board of MetabERN. Lut was involved by the foundation and is, at the moment, the longest serving board member of FIN. Her long term -ambition is to optimise the quality of life for the Fabry patients and their families.

Carolyn Ellaway Australia
Carolyn is a Clinical Associate Professor at the University of Sydney. She has been working as a Pediatrician and Clinical Geneticist with the Genetic Metabolic Disorders Service, Children's Hospital, Westmead since 2001 and Sydney Children's Hospital, Randwick, since 2014. She is the Deputy Director of the Western Sydney Genetics Program. Carolyn has been responsible for the care of children with a wide range of genetic metabolic disorders which includes the multidisciplinary management of patients with lysosomal storage disorders (LSD), including Fabry disease and enzyme replacement therapy (ERT) delivery. Carolyn obtained her PhD at Sydney University in 2001 and a medical degree from the University of Melbourne in 1990. She has published over 80 journal articles and 4 book chapters. She has been invited to speak at a number of local and international conferences, and has been a co-investigator of several clinical trials, some of which have explored ERT in patients with LSD.

The FIN Board

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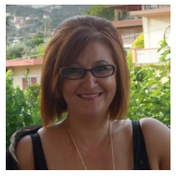
Anne Grimsbo was born in Denmark. Fabry was a part of her life from early childhood, as her father was a Fabry patient and his brother died from Fabry at the age of 32. Anne is married to Rune. They have two adult sons who are not affected by Fabry thanks to prenatal diagnosis. Anne is a social educator and is working full time as an area leader for three preschools and 27 daycare homes. Anne's cousin founded the Fabry Danish Patient Organization in 2002. In 2013 she became chair of the organisation. In 2014 Anne joined the FIN board. Anne has received ERT treatment since the age of 45 and in 2010 she joined the trial of chaperone capsules.

Jack Johnson was born in Missouri, a state in the middle of the U.S. He grew up on cattle ranches and farms across the Midwest and western U.S. Jack has Fabry disease and has traced Fabry back to his grandfather's grandmother. Jack is married to Debra and has two big boys. Jack is the founding member of the Fabry International Network and is now honoured to be the acting president. Jack is one of the founders and executive director of the Fabry Support & Information Group (FSIG) in the U.S. At the annual WORLD Symposium Jack was honoured to receive the Patient Advocate Leader (PAL) Award for 2018. Jack believes that his years of experience with FSIG will help with the management of FIN and hopes to gain beneficial knowledge from greater involvement in the international Fabry community.

Anna Meriluoto holds a Master of political science degree from the University of Helsinki. She has over 15 years of experience as a volunteer in patient advocacy and she has been an active board member of the Finnish Fabry Association since its establishment in 2003. Anna has been a member of the Fabry International Network board of directors for the past 8 years. Through Fabry International Network she keeps in regular contact with EURORDIS and she was trained to become an eligible patient advocate in clinical trials and pharmacovigilance at the EURORDIS Summer School in Barcelona in 2011. She has travelled extensively to speak about Fabry Disease and was one of the founding members of HARSO, the first ever Rare Disease Organisation in Finland in 2012. Anna has been a college lecturer for the past 10 years. Being a Fabry patient herself along with her mother and older brother Anna shares FIN´s vision of a world where every single person affected by Fabry disease has the best quality of life possible through early diagnosis, treatment and cure.



Mary Pavlou was born in Athens, Greece and has been a nurse since 1944, working in several departments such as ER, cardiology and pathology. Currently Mary is chief of staff in pathology – hypatology department. She also holds a Master in healthcare services administration. She is an active member of Greek Lysosomal Association "Solidarity" from 2002 the year of her diagnosis with Fabry disease. In 2018 she was re-elected as vice president. Mary has been on ERT from 2006. In 2012 she joined FIN and became a board member in 2014 and was re-elected in 2017. Her vision for the Fabry community is to gain more knowledge about the condition and to bring together Fabry members from around the world to share their experiences and fears as the first step to overcome obstacles and difficulties that disease brings in patients' every day life.

Delegates

Amicus - UK

Amicus - USA

APMF - France

Association of Families with Fabry Disease - Poland

Avrobio - USA

Belgian Organisation for Metabolic Diseases - Belgium

Chiesi Farmaceutici S.p.A. - Belgium

Chiesi Farmaceutici S.p.A. - Italy

China Fabry Community - China

Chinese Organisation for Rare Disorders - China

Danish Fabry Group - Denmark

Fabry Paitentforening Denmark - Denmark

Fabry Russia - Russia

Fabry Support and Information Group - USA

Fabry Swiss - Switzerland

Fabryföreningen i Sverige - Sweden

Finnish Fabry Association - Finland

Freeline - UK

FSIGN - Netherlands

Greek Lysosomal Association Solidarity - Greece

Hungarian Foundationfor patients with Fabry Disease - Hungary

Idorsia - Switzerland

Japan Fabry Disease - Japan

Korea Fabry Patient Group - Korea

Lithuanian Association GYVASTIS - Lithuania

Protalix - Israel

Rare Diseases South Africa - South Africa

Road to life - Russia

Sanofi - Finland

Sanofi - Italy

Sanofi - Lithuania

Sanofi - Spain

Sanofi - UK

Sanofi - USA

Shire - Lithuania

Shire - Switzerland

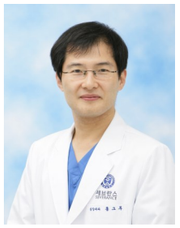
Society for Mucopolysaccharide Diseases - Spain

Society for Mucopolysaccharide Diseases - Turkey

Society for Mucopolysaccharide Diseases - UK

TDFA - Taiwan

Speakers



Geu Ru Hong Korea

Geu Ru Hong is a medical educator and a member of the Korean Society of Echocardiography and an Associate professor at Yeungnam University in Daegu since 2003. He his Director of echocardiography at Yeungnam University Hospital and a Council member of the Korean Society of Echocardiography in Seoul since 2003. Geu Ru was a member of the research committee between 2004-2006. He has also been a member of the board examination committee at the Korean Association of Internal Medicine in Seoul and a member of the public information committee from 2004-2006. He was an associate editor for the Journal of the Korean Society of Echocardiography in Seoul for a year and has been an editor for the Journal of Cardiovasc and Ultrasound since 2006.

Derralynn Hughes Royal Free Hospital and University College London, United Kingdom

Dr Derralynn Hughes is Clinical Director Haematology, Oncology and Palliative Care at the Royal Free London NHS Foundation Trust and Senior Lecturer in Haematology at the University College London, UK. She has clinical responsibilities in the area of Haematology and Lysosomal Storage Disorders. She also directs the research programme in the LSD unit research laboratory where interests include understanding the pathophysiology of phenotypic heterogeneity in Fabry Disease and bone related pathology in Gaucher disease. Dr Hughes is Principle Investigator of a number of clinical trials examining the efficacy of Enzyme Replacement and Chaperone Therapy 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 Fabry disease in women. She is an author of over 120 papers in the area of macrophage biology and lysosomal Storage Disorders.

Aneal Khan Alberta, Canada

The primary goal of Dr. Khan's research is to investigate novel methods of treatment of childhood onset genetic diseases. As a pediatrician, medical geneticist and metabolic diseases specialist, his clinical research has primarily included subjects with inborn errors of metabolism. Currently, he is involved as a principal investigator in 15 clinical trials from novel therapies to next generation sequencing for mitochondrial disorders, bone imaging in metabolic diseases, metabolic cardiomyopathies and rare disease registries. In 2014, his group at the Alberta Children's Hospital was the first in Canada to use liver cell transplant to treat urea cycle disease. In February 2017, as a principal investigator, for a pan-Canadian group, they were the first in the world to treat Fabry disease with *ex vivo* gene therapy and the first group in Canada to treat a human with an inborn error of metabolism with gene therapy at Foothills Medical Centre. In March 2018, he was the first in Canada to treat OTC deficiency using an *in vivo* gene therapy using an AAV8 vector. He is the principal investigator for MITOFIND. He has been nominated for the Alberta Health Services Life Time Achievement Award. He is situated at Alberta Children's Hospital in Calgary and the University of Calgary and cares for both pediatric and adult patients.

Simon Körver Academic Medical Center , Amsterdam

Simon Körver is working as a PhD student and medical doctor in the Academic Medical Center in Amsterdam, under supervision of Carla Hollak and Mirjam Langeveld. He sees patients with Fabry disease in the outpatient clinic and is also engaged in clinical research in this patient group. His main points of focus are the cognitive, psychological and cerebral consequences of Fabry disease. He is interested in the subjective experienced complaints of patients with Fabry disease and strongly believes in the use of patient reported outcomes in medical research.

Atul Mehta Royal Free Hospital, London, United Kingdom

Professor Mehta is a Consultant Haematologist and Physician at the Royal Free Hospital in London. He completed his undergraduate training at Cambridge University and King's College Hospital. Before training in General Internal Medicine at King's College Hospital and Hammersmith Hospital in London and in Haematology at the Hammersmith Hospital. He has been in his current post since 1986. He is Clinical Director of one of the nationally designated Lysosomal Storage Disorders Units, which focus on multidisciplinary assessment and treatment of LSDs. This is one of the largest centres for adult patients with inherited metabolic diseases in the UK. . He is the author of 3 books, more than 40 Chapters and over 300 publications in peer reviewed journals.

Gere Sunder-Plassmann Medical University Vienna, Austria

Gere Sunder-Plassmann is Associate Professor of Medicine at the Division of Nephrology and Dialysis, Department of Medicine III, Medical University Vienna, Austria. Born in Vienna in 1960, he is married and father of two sons. He trained in Medicine at the University of Vienna where he earned his MD in 1985. He had his postdoctoral training at the Department of Internal Medicine I, University of Vienna from 1986 to 1991. In 1992 he became a Assistant Professor at the Division of Nephrology and Dialysis at the Department of Medicine III, Medical University Vienna, where he is Associate Professor since 1994. His research focuses on cardiovascular disease risk factors, anemia in renal disaese, hereditary kidney diseases, as well as treatment and outcomes of kidney transplant recipients and he has published more than 300 papers or book chapters.

Rick Steeds United Kingdom

Rick Steeds is a Consultant Cardiologist at University Hospitals in Birmingham, specialising in cardiovascular imaging and in the management of metabolic cardiomyopathy through the Rare Diseases Centre in Birmingham. He is the immediate past President of the British Society of Echocardiography. He is an Honorary Reader at the Institute of Cardiovascular Sciences, University of Birmingham, has published more than 200 peer-reviewed research articles, and is a Strategic Associate Editor for Echo, Research and Practice. With funding from the BHF and commercial sources, he is currently using multiparametric CMR and echocardiography to investigate the role of myocardial infiltration in valvular heart disease and in cardiomyopathy related to Rare diseases.

Friday 8th June

AGM

(All FIN Members are required to attend)

Sonata Meeting Room 5th Floor

15.00-16.00

AGENDA

1. Welcome & Introductions
2. Apologies
3. Minutes of AGM 2017
4. Report on behalf of FIN Board Directors
5. Financial Review
6. Election of Directors
7. Appointment of Officers
8. FIN Audit & Appointment of External Auditor
9. AOB
10. Close

DINNER

Tilia Restaurant, 1st Floor

19.00 — Late

Saturday 9th June

Belvedere 3 Conference Hall , 2nd Floor

09.00	Welcome & Introductions	Jack Johnson

Session 1 chair ***Jack Johnson***

09.10	A personal Experience of Fabry Disease	Martynas Davidonis
09.40	Paediatric Fabry Disease	Prof Carolyn Ellaway
10.10	Cognition in Fabry disease, relations to cerebral imaging and depression	Dr Simon Körver
10.45	*Break*	

Session 2 Chair ***Lut De Baere***

11.00	Cardiac Disease in Children and Adults with Fabry Disease	Prof Gue-Ru Hong
11.30	Fabry Disease; National Audit of cardiac devices	Dr Rick Steeds
12.00	Prevalence of kidney diseases in males & females with Fabry Disease	Dr Gere Sunder-Plassmann

12.30 ***Lunch***

Session 3 Chair ***Mary Pavlou***

13.30	Update on Results of ERT	Prof Atul Mehta
14.00	MetabERN: What's in for the patients	Lut De Baere
14.30	Understanding Fabry in Families—Global Survey Update	Dr Jacqueline Adam
15.00	*Break*	

Session 4 Chair ***Anne Grimsbo***

15.20	Current & Future Treatment strategies in Fabry Disease	Dr Derralynn Hughes
15.50	First Gene Therapy in Fabry Disease	Dr Aneal Khan
16.20	My Experience having Gene Therapy	Christopher Armstrong
16.50	A Tribute to Christine Lavery	Rebecca Brandon
17.20	Thank you & close	Anna Meriluoto
17.30	*Break*	

19.00 ***Dinner - Bora Bora Grill Restaurant & Bar***