Programme

The primary aim of The Fabry International Network (FIN) is to facilitate collaboration between Patient Organisations to support those affected by Fabry Disease. It seeks to do this primarily through enabling communication, promoting good practices and acting as an independent forum for Fabry associations.

Welcome to the 4th FIN Fabry Expert Meeting

20th – 22nd May 2016

The principle foundation of FIN is to be neutral and independent in all of its communication, actions, and decisions worldwide. The Fabry International Network seeks to see the implementation of a Global International Fabry Conference that allows a platform where all relevant aspects and information of Fabry disease, current or future drugs and therapies can be shared in a non-biased environment.

The venue

The Prince Park Tower Hotel, Tokyo, 4-8-1 Shibakoen, Minato, Tokyo 105-8563, Japan

A 4 star hotel in a glass and steel tower, this cosmopolitan high-end hotel is located in the heart of Tokyo and an 8-minute walk from the Tokyo Tower and within a 7 -minute walk from 2 train stations. The polished rooms come with free Wi-Fi, flat-screen TVs, minifridges and whirlpool tubs. Room service is offered 24/7. Amenities include free shuttles from JR Hamamatsucho Station, a fitness centre and an indoor lap pool, Spa & fitness along with a bowling alley, 7 restaurants and a bathhouse with natural hot spring water. A swanky sky bar overlooks the Tokyo Tower. Check in time 3pm, check out 12 noon.

Places of Interest

Speakers

Professor Yoshikatsu Eto, M..D. PhD

Prof. Eto is currently a Director of Advanced Clinical Research Center, Institute of Neurological Diseases and Professor Emeritus , Jikei University School of Medicine. He served as a Professor and chairman, Department of Pediatrics, Director of DNA Institute for Medical Science and also Vice President, University Hospital, the Jikei University School of Medicine. Prof. Eto studied for more than 40 years in the field of lysosomal storage diseases and has left many achievements. He also served as a President of Japan Pediatric Society for four years and also as a President of Japan Society of Inherited Metabolic Disorders for 7 years. He has been leading the research in Pediatrics in Japan. Internationally, he also serves as a Standing Committee Member of International Pediatric Association for 6 years and as a President of the 10th International Congress of Inborn Error of Metabolism, 2006, Tokyo. He contributed to the establishments of Asian Society of Pediatric Research and also Japan Society of Gene Therapy, and is also a founder of Japan Society of LSD. He published more than 350 English written papers. Currently, he has studying about clinical and basic research in LSD, particularly, Fabry disease, Pompe disease and genetic leukodystrophy, and also iPS research in LSD, as well.

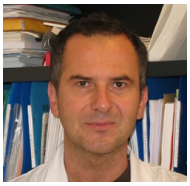
Professor Atul Mehta, MA, MD, FRCP, FRCPath

Professor Mehta is a Consultant Haematologist and Physician at the Royal Free Hospital in London, part of the Department of Haematology of University College London School of Medicine. He completed his undergraduate training at Cambridge University and King's College Hospital. He then trained 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 and 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, and he works with a team of nurses, physicians and researchers who are committed to developing and delivering care to patients and families. He is also the Head of the Myeloma service in the largest hospital in London who provide high quality clinical services as well as running clinical and laboratory research. He is a senior examiner in Haematology and Medicine at the University of London and with the Royal College of Pathologists. He is the author of 3 books, more than 40 Chapters and over 300 publications in peer reviewed journals.

Christine Lavery MBE

Christine Lavery was appointed Chief Executive of the Society for Mucopolysaccharide and Related Diseases (MPS) in 1993. During her time at MPS she has taken the charity to new heights, employing 21 members of staff, managing a substantial research budget and a unique UK-wide advocacy service providing needs led support to nearly 1300 children and adult sufferers, their families and professionals in areas of home adaptations, special educational needs, access to new therapies, respite care, palliative care and pre- and post-bereavement support. Christine has served on the Department of health Advisory Board on Genetic Testing and is currently a patient representative on the NHS England LSD Specialised Commissioning Advisory Group. Christine was awarded the Member of the British Empire for her services to Metabolic diseases by the Queen in the New Year's Honours List for 2002 and at the 2006 International Symposium on Mucopolysaccharide and related diseases received 'A Life Time Award' from the International MPS Community. Christine continues to promote the needs of MPS, Fabry and related Lysosomal Storage Diseases across nations and working closely with the MPS International Network.

Professor Dominique P. Germain



Doctor Dominique P. Germain is Professor of Medical Genetics at the University of Versailles - St Quentin en Yvelines (UVSQ), and head of the Division of Medical Genetics at the Raymond Poincaré Hospital (AP-HP) in Garches, France. Professor Germain graduated in medicine, from the University of Nancy I, France, and went on to specialize in dermatology at the University of Nancy and in human genetics at the University of Paris V (René Descartes), France. In 2007, Professor Germain has been appointed Professor at the University of Versailles (UVSQ), France. Throughout hiscareer, Professor Germain has written extensively on the topics of Fabry disease and hereditary diseases of connective tissue. He has written several book chapters and has published over 100 peer-reviewed papers in medical journals.

Ichiei Narita, M.D.

Dr. Narita is the chief Professor of Medicine, Nephrology and Rheumatology at Niigata University Medical School, Niigata, Japan. He has graduated from Niigata University Medical School in 1983. He has been recognized for his contributions to research and clinical managements of kidney diseases, including primary and secondary glomerulonephritis, diabetic kidney diseases, drug-induced kidney injury, as well as inherited kidney diseases. Recent his major research interests are in genetic study to clarify the mechanism of initiation and progression of IgA nephropathy, molecular genetics and cell biology of primary glomerulonephritis, bone and calcium metabolism in chronic kidney failure, clinical and genetic analysis of autoimmune diseases, complications in ESRD patients, and clinical management of Fabry disease patients. He is now a board certified member of the Japanese Society of Internal Medicine, and a member of board directors of the Japanese Society of Nephorology (JSN) and the Editor in chief of the official journal of JSN, Clinical and Experimental Nephrology.

Dr Dau-Ming Niu M.D. PhD

Dr. Dau-Ming Niu is a professor in Institute of Clinical Medicine at National Yang-Ming University and director of Genetic and Endocrinology Division, Research and Treatment Center of Rare Disease at Taipei Veterans General Hospital. Dr. Niu received his M.D. from Kaohsiung Medical University and was a clinical and research fellow at National Taiwan University Hospital. He had advanced studies in Professor Yuan-Tsong Chen's laboratory in medical genetics at Duke University. After coming back to Taiwan, Dr. Niu received his PhD degree from National Yang-Ming University of Clinical medicine. Dr. Niu's research interests include medical genetics and endocrinology. His research in 6-pyruvoyl-tetrahydropterin synthase deficiency has the most outstanding treatment outcome ranking No. 1 in the world so far. In addition, his team found that Taiwanese aboriginals have the highest prevalence of homocysteinuria and inherited Retinitis Pigmentosa across the globe. In recent years, Dr. Niu focused his studies intensively on lysosomal storage diseases and his team is the first one to discover that Taiwanese have the highest prevalence of Fabry disease in the world. At the same time, a research and treatment center of rare disease was also established, which is the first center integrating research and medical expertise of different areas in the rare disease field in Asia.

Speakers continued

Megan Fookes OAM

Megan is a mother of 2, one who is diagnosed with Fabry disease as well as herself, a former educator, a voluntary Director of Fabry Australia and the Advisor Policy & Stakeholder Relations at Rare Voices Australia; a Not for Profit National Alliance for Rare Diseases in Australia. Her professional association with Rare Diseases stems from a very personal connection. Her late father waited 48 years to receive a diagnosis of Fabry disease. Her parents who were very keen to learn more formed Fabry Australia 22 years ago. She has been working in the rare disease sector since 1999. Fabry Australia successfully advocated on behalf of the Australian Fabry community to receive two Commonwealth funded therapies back in 2004 under the Life Saving Drugs Program. Megan sits on many other Committees and Advisories and served on the FIN Board for 3 years. Megan is keen to utilise her personal and professional experience to make a real difference and advocating for a National Plan and Rare Disease Policy ensuring people impacted by rare diseases have better health incomes in the future. Megan Fookes, was recently awarded the Medal of the Order (OAM) on Australia Day. Megan has dedicated the award to her late father who lived with Fabry Disease and to all adults, children and families who are doing their very best living with rare diseases.

Dr Derralynn Hughes

Dr Derralynn Hughes is Senior Lecturer in Haematology at the University College London, UK and consultant haematologist at the Royal Free London NHS Foundation Trust. She has clinical responsibilities in the area of Haematology and Lysosomal Storage Disorders (LSD). She also directs the research programme in the LSD unit research laboratory where interests include understanding phenotypic heterogeneity in Fabry Disease and the pathophysiology of bone related pathology and malignancy in Gaucher disease. Dr Hughes is Principle Investigator of a number of clinical trials examining the efficacy of Enzyme Replacement Therapy and other new agents in the treatment of Gaucher, Fabry, Pompe and mucopolysaccharidosis (MPS) disorders. She is an author of over 120 papers in the area of macrophage biology and Lysosomal Storage Disorders.

Toya Ohashi M.D.

Dr. Ohashi was graduated from The Jikei University School of Medicine in 1981. After 2 years residency of pediatrics, he joined department of pediatrics at The Jikei University School of Medicine and started study for lysosomal storage disease under direction of Prof. Eto. He spent 3 and half years in US to study molecular genetics of lysosomal storage disease. During his stay in US, he started to study gene therapy for lysosomal storage. From 1995, he moved to Research center for Medical Sciences in The Jikei University School of Medicine and continue to study novel therapeutic approach to lysosomal storage diseases including gene therapy. He was involved in clinical trial of enzyme replacement therapy for Fabry disease in Japan. He was now director of Research Center for Medical Sciences in The Jikei University School of Medicine. His major interest is development of novel therapy, especially gene therapy, for lysosomal storage disease. He is still working as a clinician and taking care of more than 30 Fabry patients in outpatient clinic.

Professor Christoph Kampmann

Christoph Kampmann, MD, PhD, is Professor of Paediatric Cardiology in the Centre for Diseases in Childhood and Adolescence at the University Medical Centre of Johannes Gutenberg-University of Mainz in Germany. He is also Director of the Department of Congenital Heart Diseases at the University Children's Hospital in Mainz, Germany. Dr Kampmann earned his medical degree from the Heinrich-Heine-University of Düsseldorf in Germany. He furthered his training at the Department of Paediatric Cardiology at the German Heart Institute in Berlin; Department of Paediatrics, Neonatology, and Cardiology at the Charité University Hospital in Berlin; and at the Texas Heart Institute in Houston, Texas, USA. Dr Kampmann completed his PhD research on the cardiac manifestations of Fabry disease and the effects of enzyme replacement therapy in 2003. With nearly 2 decades of experience, Dr Kampmann is an expert on lysosomal storage diseases (LSDs), specialising in their impact on the heart and cardiovascular system, and has helped managed various LSDs in more than 700 patients. He has been actively involved in multiple research collaborations and publications with international LSD and research centres, with more than 70 peer-reviewed publications to his credit.

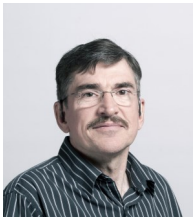
Professor James Moon

James Moon is a Professor of Cardiology UCL, London and the Clinical Director of Imaging at the Barts Heart Centre (BHC). Formerly, he set up the the Heart Hospital Imaging Centre, a dedicated cardiac MRI department performing 3500 scans a year with a triple focus on clinical scanning, research and education. The new Barts Heart Centre has 3 dedicated CMR scanners (2 1.5T Aeras and a 3T Prisma). The centre is at scale – estimates of annual activity are 8,000 CMR scans, 3000 nuclear scans, 3000 CT scans and 40,000 echos. Trained at Cambridge and Oxford, his MD at Imperial was 'myocardial tissue characterization by CMR'. His current research group of 8 fellows have particular interests in measuring diffuse myocardial fibrosis, iron, fat infiltration (Fabrys) and amyloid using "T1 mapping". He has published more than 180 papers and works in partnership to develop surrogate endpoints for clinical trials including first in man studies. Another interest is in making scans faster/cheaper and in improving/maintaining quality as CMR expands. He set up and leads the international T1 mapping development group aiming to "coordinate activity to the goal of being able to change therapy using CMR endpoints". He is a former board member of SCMR, the current president elect of the British equivalent, BSCMR. He is a former member of NICE. Education interests are key to the group and significant successes include the setting up of web resources (SCMR video-on-demand, case of the week, online Moodle training, as well as local hands-on training courses.

Andrew Talbot

Speakers continued

Jack Johnson



Jack Johnson was born in 1963 in Missouri, a state in the middle of the U.S. He grew up on cattle ranches, a dairy and farms across the Midwest and western U.S. Jack has Fabry disease along with many family members. They have 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 vice-president. Jack is one of the founders and executive director of the Fabry Support & Information Group (FSIG) in the U.S. 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 Meriluto

Anna Meriluoto holds a Master of political science degree from the University of Helsinki. She has over 10 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. Coming from a small country Anna has always considered the role of international patient relations as pivotal and she has been a member of the Fabry International Network board of directors for the past 6 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. In her international role she has travelled extensively to speak about Fabry Disease in conferences such as the first All-Russian Conference for Rare Diseases in St. Petersburg in 2011 and the first Eurasian Conference for Rare Diseases in Moscow in 2012. Anna was also one of the founding members of HARSO, the first ever Rare Disease Organisation in Finland in 2012. Her professional background is in travel industry but she has been a college lecturer for the past 7 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.

Anne Grimsbo

PROGRAMME

Board and Industry Meetings

FRIDAY 20th MAY

09.00 —10.15	FIN & Shire Meeting	FIN Board & Shire
10.15 — 10.45	Tea/Coffee Break	All
10.45 — 12.00	FIN & Amicus Meeting	FIN Board & Amicus
12.00 — 13.00	Lunch	All
13.00 — 14.15	FIN & Genzyme Meeting	FIN Board & Genzyme
14.15 — 18.30	Break	All
18.30 — 21.00	Dinner	All

FIN Expert Meeting

SATURDAY 21 MAY

Session 1	Chairs	Professor Atul Mehta & Professor Yoshikatsu Eto	
	09.00—09.15	Welcome & Introductions	Christine Lavery
	09.15—09.45	Opening remarks & an introduction to Fabry disease in Japan	Professor Yoshikatsu Eto
	09.45—10.15	Understanding the natural history of Fabry disease	Professor Atul Mehta
	10.15—10.45	Break	
Session 2	Chairs	Dr Dominique Germain & Dr Ichiei Narita	
	10.45—11.15	Fabry disease in females	Professor Dominique Germain
	11.15—11.45	Fabry disease in children – The need for early intervention	Dr Dau-Ming Niu
	11.45—12.15	Fabry experience for children in Australia	Megan Fookes
	12.15—13.15	Lunch	
Session 3	Chairs	Dr Derralynn Hughes & Dr Toya Ohashi	
	13.15—13.45	Fabry disease and the heart	Professor Christoph Kampmann
	13.45—14.15	Fabry disease and the valve of cardiac imaging	Professor James Moon
	14.15—14.45	Fabry Disease and the kidney	Professor Andrew Talbot
	14.45—15.15	Break	
Session 4	Chairs	Jack Johnson & Anna Meriluto	
	15.15—15.45	The role of ERT and the emerging therapies in Fabry disease	Dr Derralynn Hughes
	15.45—16.15	The Importance of access to medicine for Fabry Disease	Anne Grimsbo
	16.15—16.45	Discussions on patient expectations for current and new therapies	Jack Johnson & Anna Meriluoto
	16.45—17.00	Thank you and End	Jack Johnson
	18.30—19.00	**Pre-dinner drinks in the Green Rattan bar**	*All*
	19.00—22.00	**Dinner at the Green Rattan**	*All*