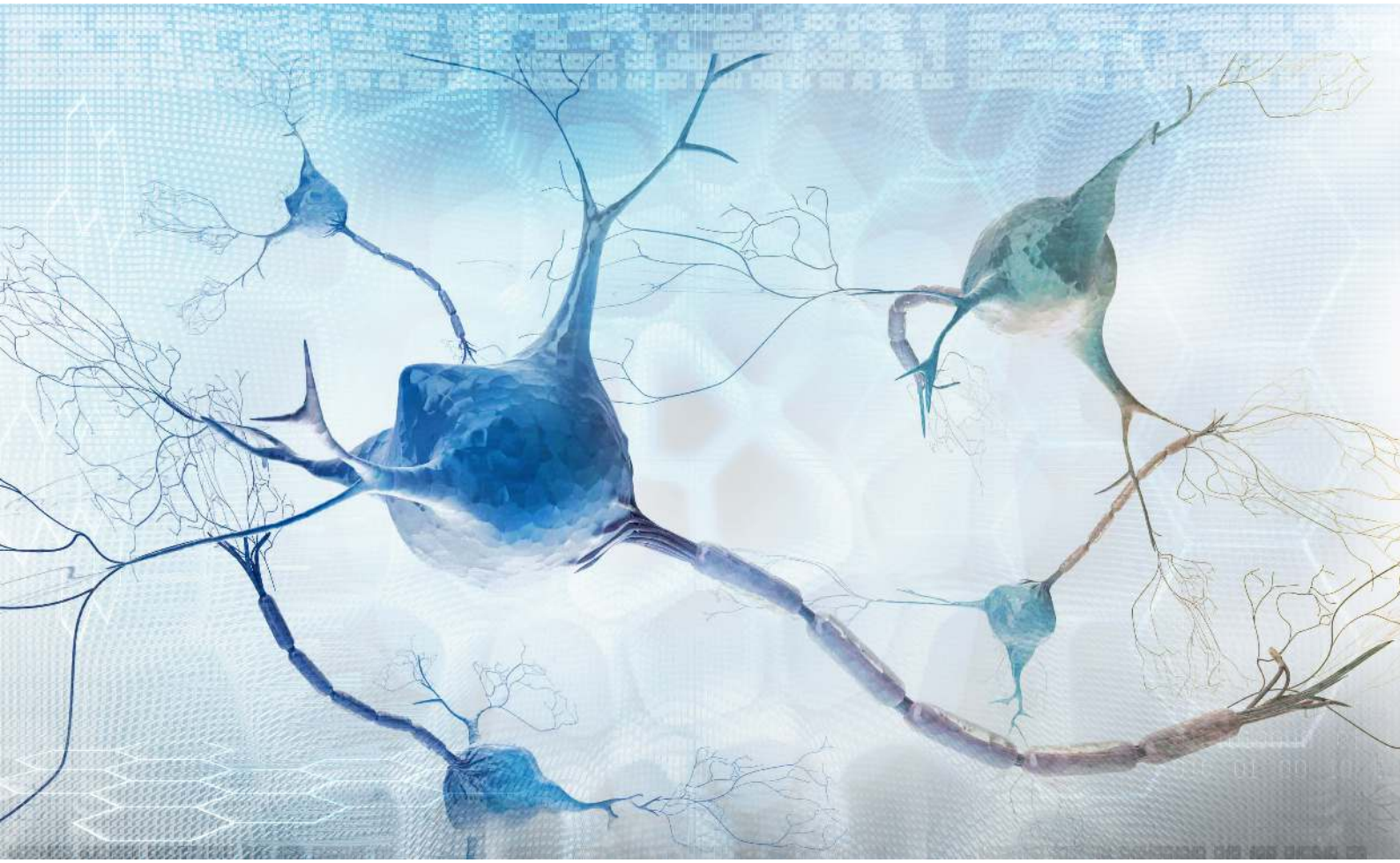
INTERNATIONAL SYMPOSIUM ON NEW DEVELOPMENTS IN NEUROFIBROMATOSES AND RASOPATHIES

Their Management, Diagnosis, Current and Future Therapeutic Targets

27th - 29th November 2017, Crowne Plaza, Kochi, Kerala, South India



THE FIRST INTERNATIONAL SYMPOSIUM ON RASOPATHIES TO BE HELD IN ASIA

RASopathies are a class of developmental disorders with overlapping clinical features as well as genetic mutations. RASopathies are associated with dysregulation of the RAS-MAPK (RAS/mitogen activated protein kinase) signalling pathway, an important pathway in humans as it is related to nine different genetic conditions and in many cancers. The meeting will bring together leading scientists, clinicians, researchers and trainee health care professionals in the field of genetics, cancer, neurology, paediatrics, cardiology, neurosurgery, craniofacial surgery, plastic surgery, dermatology, oncology and biomedical sciences from across the world.

200 PLACES

25+ SPEAKERS

20+ TOPICS

AIMS

TO ENSURE THAT HEALTH PROFESSIONALS AND SCIENTISTS ARE KEPT ABREAST OF ANY NEW DEVELOPMENTS IN CLINICAL PRACTICE

TO IDENTIFY THERAPEUTIC TARGETS FOR SEVERAL OF THESE RARE DISEASES

TOPICS COVERED

- RASopathies: Syndromes of Ras/ MAPK pathway dysregulation
- Neurofibromatosis Type 1
- Legius Syndrome
- Noonan Syndrome
- Noonan Syndrome with multiple lentigines
- Costello Syndrome
- Cardio-Facio-Cutaneous Syndrome
- Capillary Malformation-Arteriovenous Malformation Syndrome
- Natural history, improved diagnosis and genotype phenotype correlation for NF2 and Schwannomatosis
- New developments in mouse modelling of Neurofibromatoses and RASopathies
- Translational/Clinical studies in Children and adults with neurofibromatosesand RASopathies

Professor Meena Upadhyaya, OBE
Cardiff, United Kingdom

Professor Meena Upadhyaya obtained her PhD from Cardiff University and gained Fellowship of the Royal College of Pathologists (FRCPath) from the Royal College of Pathologists, London. Meena Upadhyaya is a distinguished professor of Medical Genetics (Hon) at Cardiff University and was a Consultant Molecular Geneticist and Directorate R&D lead at the University Hospital of Wales, Cardiff. She leads NF1 research in Cardiff. She has made substantial contributions to the molecular understanding of a number of genetic conditions, including neurofibromatosis type 1 (NF1), facioscapulohumeral muscular dystrophy (FSHD), Legius syndrome, Charcot Marie Tooth disease, Duchenne Muscular Dystrophy, Sotos syndrome, Hunters syndrome inter alia. Prof. Upadhyaya has been involved in neurofibromatosis research since 1986, publishing nearly 200 papers in peer reviewed journals and co-editing two books on NF1. She sits on many national and international Committees She has supervised numerous undergraduate, PhD and MD students. Prof. Upadhyaya's research group made significant contributions towards the mapping and cloning of the NF1 gene, molecular diagnosis of NF1, constitutional and somatic mutation analysis, functional analysis, genotype-phenotype correlations and more recently on NF1-associated cancers in order to generate potential therapeutic targets.

Professor Upadhyaya was nominated as a role model by the Cardiff University for the Aurora Leadership Foundation for women in higher education. She has received many awards including the prestigious 2017 Saint David's award in the category of Science, Technology and Innovation. She was also awarded OBE in the January 2016 Queen's Honours list for her contributions to Medical Genetics and the Welsh Asian community and was recently acknowledged amongst the top 12 women in Cardiff University over the last 50 years. She is actively involved with a number of charity and community-based projects and has successfully combined her academic work with public service.

Professor Gareth Evans
Manchester, United Kingdom

Professor Evans has established a national and international reputation in clinical and research aspects of cancer genetics, particularly in neurofibromatosis and breast cancer. He has published 658 peer reviewed research publications; 247 as first or senior author. He has published over 100 reviews and chapters and has had a book published by Oxford University Press on familial cancer. He has an ISI web of knowledge H-index of 90 and google scholar of 123 having only published his first article in 1990. In the last 7 years he has raised over £45 million in grants for multicentre and local studies - approximately £38 million to Manchester. He is Chief Investigator on two NIHR program grants (2009-2014-£1.59 million) (2017-2020-£1million) on breast cancer risk prediction and also has an NIHR RfPB grant as CI (2011). He has led a successful bid for a Nationally funded NF2 service (£7.5 million pa) that started in 2010 and is involved in the national complex NF1 service. He is overall cancer lead (3 themes) and Cancer Prevention Early detection theme lead on the successful all Manchester NIHR Biomedical research centre bid (2017-2022-£28.5million). He is lead clinician on the NICE familial breast cancer guideline group and is a trustee of BCN and the Neuro Foundation.

Dr Joshi George
Manchester, United Kingdom

Mr. Joshi George did his Medical Schooling in Kerala, India. He subsequently went to Belfast to learn Trauma Surgery. He was awarded a MD from the University of London for his research on spinal cord injury. Having completed his FRCS Neurosurgery in Birmingham, he went on to complete a fellowship in spine surgery at the University of Toronto. He currently works as a Consultant Neurosurgeon at Salford Royal Foundation Trust in Manchester, which is the largest spine unit in Europe. He is a Fellow of the European Board of Neurological surgery and is one of the two specialist spine surgeons for neurofibromatosis in the U.K.

Professor Ashok Pillai
Kochi, Kerala, India

Dr. Ashok Pillai is a professor of neurosurgery and neuroscience associated with Amrita Institute of Medical Sciences, Kochi. After completing his initial college education in the neurosciences, medical college and surgical training in the United States, he migrated back to India to join AIMS, Kochi in 1999. After completion of his neurosurgical training, he pursued further advanced training in Canada and the United States in the field of epilepsy surgery and other areas of functional neurosurgery.

In the field of neurooncology, Dr. Ashok Pillai has maintained a dedicated clinical and research interest in familial tumor syndromes - namely von Hippel Lindau and the neurofibromatosis. Presently, Dr. Ashok Pillai heads the epilepsy surgery program, neurooncology program (dealing with adult and paediatric brain tumors), and the peripheral neurosurgery program at AIMS, Kochi. He excels in stereotactic neurosurgery and is a pioneer in the upcoming field of robotic neurosurgery. He also has a dedicated interest in pediatric neurosurgery.

Professor Luis F. Parada
New York, USA

Luis F. Parada obtained a BS from the University of Wisconsin and a Ph.D. in Biology from MIT, identifying oncogenes in human cancer. He was a Damon Runyon and Helen Hay Whitney Postdoctoral Fellow at the Pasteur Institute. He headed the Molecular Embryology Section at the National Cancer Institute in Frederick, MD from 1988 to 1994 when he moved to the University of Texas Southwestern Medical Center at Dallas as the inaugurating Diana and Richard C. Strauss Distinguished Chair in Developmental Biology, and was Director of the Kent Waldrep Foundation Center for Basic Neuroscience Research. During his time in Dallas, Dr. Parada advanced his studies of nerve cell survival and regeneration, mood disorders, and renewed his attention on cancer with emphasis on the nervous system. His laboratory uses genetic mouse models to study human disease including Neurofibromatosis, cancers of the nervous system, autism, and neural development. In 2015 Dr. Parada moved his laboratory to Memorial Sloan Kettering Cancer Center to assume leadership of the interdisciplinary Brain Tumor Center. In addition Dr. Parada holds the Albert C. Foster Chair and is Professor of Cancer Biology and Genetics. In recognition of his contributions to science, Dr. Parada has been elected to: The National Academy of Sciences; The American Academy of Arts and Sciences: The Institute of Medicine - National Academy of Sciences (National Academy of Medicine); The American Association for the Advancement of Science (AAAS), The Academy of Medicine, Engineering and Science of Texas TAMEST); and is a life-time American Cancer Society Professor.

Professor Katherine (Kate) Rauen
California, USA

Katherine (Kate) Rauen, MD, PhD is a Professor in the Department of Pediatrics, Division of Genomic Medicine at the UC Davis where she currently serves as the Chief of Genomic Medicine and holds the Albert Holmes Rowe Endowed Chair in Human Genetics. She received a MS in Human Physiology and a PhD in Genetics from UC Davis doing research on gene dosage compensation and genetic evolution. She obtained her MD at UC Irvine where she also did research in cancer genetics. Dr. Rauen did her residency training in Pediatrics and fellowship in Medical Genetics at UC San Francisco.

Dr. Rauen is internationally known for her pioneering work in the early application of microarray technology in clinical genetics and as a leader and major contributor to the understanding of the "RASopathies", the Ras/MAPK pathway genetics syndromes. Her research program involves the clinical and basic science study of cancer syndromes with effort to identify underlying genetic abnormalities affecting common developmental and cancer pathways. Dr. Rauen led the research team, including the CFC International Family Support Group that discovered the genetic cause of cardio-facio-cutaneous syndrome and independently identified the genetic cause of Costello syndrome.

Dr. Rauen is committed to academic medicine, medical education, and advancing best practices for patients with RASopathies. She has successfully obtained both intramural and extramural funding for her research activities, and currently holds a 5-year NIH grant studying skeletal myogenesis in Costello syndrome and CFC. She is the innovator of the world-renowned NF/Ras Pathway Clinic which sheinitiated in 2007and this clinic has now been emulated around the globe. She serves on the medical advisory board of CFC International,is a Co-Director for the Costello Syndrome Family Network, and serves on the advisory broads for RASopathies Network USA and Global Genes.

Dr. Rauen was recently awarded the Presidential Early Career Award for Scientists and Engineers (PECASE) on her work for CFC and Costello syndrome. This Award is the highest honor bestowed by the United States Government on science and engineering professionals in the early stages of their independent research careers. This Presidential Award is awarded for innovative and far-reaching developments in science and technology, in an effort to increase awareness of careers in science and engineering, give recognition to the scientific missions of participating agencies, enhance connections between fundamental research and national goals, and highlight the importance of science and technology for the nation's future. Additionally in June 2014, Dr. Rauen won an International Advocacy Award, the "Global Genes RARE Champion of Hope in Science".

Professor Bruce R. Korf
Birmingham, USA

Dr. Korf is Wayne H. and Sara Crews Finley Chair in Medical Genetics, Professor and Chair of the Department of Genetics, Director of the Heflin Center for Genomic Sciences at UAB, and Co-Director of the UAB-HudsonAlpha Center for Genomic Medicine. He is a medical geneticist, pediatrician, and child neurologist, certified by the American Board of Medical Genetics (clinical genetics, clinical cytogenetics, clinical molecular genetics), American Board of Pediatrics, and American Board of Psychiatry and Neurology (child neurology). Dr. Korf is past president of the Association of Professors of Human and Medical Genetics, past president of the American College of Medical Genetics and Genomics, and current president of the ACMG Foundation for Genetic and Genomic Medicine. He has served on the Board of Scientific Counselors of the National Cancer Institute and the National Human Genome Research Institute at the NIH. His major research interests are molecular diagnosis of genetic disorders and the natural history, genetics, and treatment of neurofibromatosis. He serves as principal investigator of the Department of Defense funded Neurofibromatosis Clinical Trials Consortium. He is co-author of Human Genetics and Genomics (medical student textbook, now in fourth edition), Medical Genetics at a Glance (medical student textbook, now in third edition), Emery and Rimoin's Principles and Practice of Medical Genetics (now in 6th edition), and Current Protocols in Human Genetics.

Professor Brigitte Widemann
Bethesda, USA

Dr. Widemann is the Chief of NCI's Pediatric Oncology Branch, a Deputy Director of NCI's Center for Cancer Research, and head of the Pharmacology and Experimental Therapeutics (PET) Section at the NCI. After obtaining her M.D., from the University of Cologne (Cologne, Germany) in 1986 and completing pediatric residency in 1992 shecompletedher pediatric hematology/oncology fellowship training at the NCI Pediatric Oncology Branch from 1992 to 1995. She subsequently joined the PET Section and received tenure at NCI in 2009. Her primary research interests are in the development of effective therapies for children and young adults with refractory cancers and genetic tumor predisposition syndromes such as neurofibromatosis type 1 (NF1), and multiple endocrine neoplasia types 2A (MEN2A) and 2B (MEN2B). Dr. Widemann and her colleagues have developed novel imaging methods for NF1-related plexiform neurofibromas, are conducting a longitudinal NF1 natural history study, and pioneered the development of early phase drug treatments for this patient population. Dr. Widemann leads multiple clinical trials of new investigational agents in pediatric refractory cancers and NF1 related plexiform neurofibromas and malignant peripheral nerve sheath tumors. She and her team recentlyidentified substantial clinical activity of the MEK inhibitor selumetinib in children with NF1 and inoperable plexiform neurofibromas. Dr. Widemann also serves as the NCI Pediatric Oncology Branch principal investigator of the Children's Oncology Group Phase I Consortium and of the Department of Defense-sponsored Neurofibromatosis Consortium.

Professor Nancy Ratner
Cincinnati, USA

Dr. Ratner is interested in peripheral nerve tumors that occur in the Neurofibromatoses, NF1 and NF2, and studies the brain in Neurofibromatosis type 1. She uses genomics to study neurofibroma formation and carries out neurofibroma and MPNST preclinical therapeutics. Ratner received her bachelor's degree from Brown University, her doctorate from Indiana University, Bloomington (during which time she was a student in the Neurobiology Course at the Marine Biological Laboratory), and was a postdoctoral fellow at Washington University in St. Louis where she studied Schwann cells in nerve development under Richard Bunge and Luis Glaser. A member of the faculty at the University of Cincinnati 1987 - 2004, she is currently a Professor in the Department of Pediatrics, Cincinnati Children's Hospital, University of Cincinnati, where she holds the Beatrice C. Lampkin Endowed Chair in Cancer Biology. She co-leads the Rasopathy Program and serves as the Program Leader for the Cancer Biology and Neural Tumors Program in the Cancer and Blood Disorders Institute. She has served on numerous national and international review panels and authored over 100 peer-reviewed manuscripts and 30 reviews. She was awarded the von Recklinghausen Award in 2010, and a Jacob K. Javits Neuroscience Investigator Award (NIH-NINDS Merit Award) in 2014. She has been an active member of the NFTC since its inception.

Professor Eric Legius
Leuven, Belgium

Eric Legius is a clinician scientist with important clinical responsibilities. He was the head of Human Genetics Department of the university of Leuven till 2016 and he is currently the clinical director of the Center for Human Genetics of the University Hospital of Leuven. His research is targeted towards neurofibromatosis type 1 and related conditions. The research group contributed successfully towards the understanding of the molecular aetiology of a number of tumours in NF1 such as benign neurofibromas, gastrointestinal stromal tumours (GIST), and glomus tumours of the digits. The group was also involved in the molecular and cognitive characterization of the NF1 microdeletion region. Other projects are NF1-related oncogenesis (molecular study of malignant peripheral nerve sheath tumours and atypical neurofibromas) and a clinical trial to improve cognitive aspects in children with NF1 using Simvastatin treatment (NF1-SIMCODA trial together with Erasmus University Rotterdam). The research group is now participating in a clinical trial with Lamotrigine for the treatment of cognitive problems in children with NF1 (NF1-EXCEL together with Erasmus University Rotterdam). We are also involved in behavioural research in NF1 regarding autism spectrum disorder. The research group recently initiated a study on bone abnormalities in children with NF1 with a specific emphasis on the biological mechanism of congenital tibial bowing resulting in pseudarthrosis. In 2007 the research team identified a new condition resembling neurofibromatosis type 1, now known as Legius syndrome (autosomal dominant condition caused by a heterozygous mutation in SPRED1). The group is using animal models to gain insight in the importance of SPRED1 and the RAS-MAPK pathway for cognition.

Awards:

1990-1991: Collen Research Foundation Fellowship (Belgian-American Exchange Foundation)
1995: Blackwell Public Service Award for neurofibromatosis (NNFF)
1997: Scientific Prize NFKONTAKT
2002: NF-Holland Award
2010: "Sidmar prize" Three yearly prize for medical research of a chronic disorder, Royal Academy of Medicine Belgium.
2012: Theodor Schwann award for research of neurofibromatosis type 1 and related syndromes
2015: Friedrich von Recklinghausen Award, CTF USA.

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Professor Ludwine Messaen
Birmingham, USA

Professor LudwineMessiaen holds aPhD in Cancer Biology from the University Ghent, Belgium, where she was on facultytill 2003.

In 2003, she joined the University of Alabama in Birmingham asa Professor in Genetics and Director of the Medical Genomics Laboratory, specializing in clinical testing for all rasopathies, with focus onallneurofibromatoses.

She is interested to fully explore the spectrum of NF1 mutations, including mutations not readily identified by DNA-based analysis of the coding regions (e.g. deep-intronic splice mutations or retrotransposon-mediated mutations).

Her group described the presence of 2 NF1 hits in the melanocytes of CALMs, but not in the keratinocytes or fibroblasts.

Professor Messiaen exploresNF1 genotype-phenotype correlations using several complementary approaches. She developed and curates a patient database with de-identified genotypes and phenotypes of over 8,500 unrelated NF1-mutation-positive individuals.

Her lab actively engages in the identification of novel genes contributing to the neurofibromatoses. This includes involvement in the identification of Legius syndrome. After SPRED1gene discovery, she substantially contributed to the quick delineation of the full breath of phenotypes associated with this novel rare disorder. Furthermore, her research on a well-characterized patient cohort recently led to the identification of LZTR1 as a novel major schwannomatosis-predisposing gene.

Professor Bronwyn Kerr
Manchester, United Kingdom

Professor Bronwyn Kerr is a consultant clinical geneticist who has worked in Manchester in the UK since 1995. Her principal research interest has been Costello syndrome, and more recently, other disorders of the RAS/MAPK pathway. She is a member of the Medical Advisory Boards of the International Costello syndrome support group, the Costello Family Support Network and the Noonan syndrome association (UK). She has a number of key publications in this area, and is frequently consulted by national and international colleagues for advice on diagnosis and management in this group of disorders. A particular interest is developing an evidence base for management of rare disorders.

She is a member of the steering group of GenIDA (Genetically determined Intellectual Disabilities and Autism Spectrum Disorders), an on-line registry for patients, families and professionals. She is the lead for the Guideline Development work-stream of the European Reference Network project ITHACA (HP-ERN-2016; 739516), a five year project commencing April 2017.

With Dr Emma Burkitt-Wright, she runs a specialist Rasopathy clinic, seeing patients for confirmation of diagnosis and management advice from across the UK.

With Professor Martin Zenker and Marco Tartaglia, she has organised a second yearly European meeting on Rare Disorders of the RAS/MAPK pathway, commencing in 2009. Attracting over 70 attendees, this has been a focus for facilitating European collaboration and research in these conditions

Professor Ype Elgersma
Rotterdam, Netherlands

Prof.Dr. Ype Elgersma received his PhD in 1995 at the University of Amsterdam on the study of peroxisome biogenesis and protein trafficking in yeast. During his post-doc in the US, he switched his interest towards Neuroscience, studying the mechanisms underlying learning and memory at Cold Spring Harbor Laboratories and UCLA. In 2002 Ype Elgersma started his laboratory at Erasmus University Medical Center, Rotterdam. The main focus of the laboratory is to understand the mechanism underlying neurodevelopmental disorders. To translate the findings to the clinic, Ype Elgersma is founder and director of the ENCORE expertise center for neuro-developmental disorders, which includes the national referral center for TSC, Angelman Syndrome, Neurofibromatosis, Costello Syndrome, CFC and Fragile X, as well as a neurogenetics and autism clinic. The center is recognized as a national and international referral center. Ype Elgersma is member of a number of scientific advisory boards and won several prizes including the VIDI and VICI award.

Selected recent Publications

Overwater, I.E., Rietman, A.B., Bindels-de Heus, K., Looman, C.W.N., Rizopoulos, D., Sibindi, T.M., Cherian, P.J., Jansen, F.E., Moll, H.A., Elgersma, Y., de Wit, M.C.Y., (2016). Sirolimus for epilepsy in children with tuberous sclerosis complex: A randomized controlled trial. Neurology. Elgersma, Y. (2015). Neurodevelopmental disease: A molecular tightrope. Nature526, 50-51.

Bruinsma, C.F., Schonewille, M., Gao, Z., Aronica, E.M.A., Judson, M.C., Philpot, B.D., Hoebeek, F.E., van Woerden, G.M., de Zeeuw, C.I., Elgersma, Y (2015). Dissociation of locomotor and cerebellar deficits in a murine Angelman syndrome model. J. Clin Invest. 125, 4305-4315.

Silva-Santos, S., Van Woerden, G. M., Bruinsma, C. F., Mientjes, E., Aghadavoud-Jolfaei, M., Distel, B., Kushner, S.A., Elgersma, Y., (2015). Ube3a reinstatement identifies distinct developmental windows in a murine Angelman syndrome model J. Clin. Invest. 125:2069-2076.

Omrani, A., Van Der Vaart, T., Mientjes, E., Van Woerden, G.M., Hojjati, M.R., Li, K.W., Gutmann, D.H., Levelt, C.N., Smit, A.B., Silva, A.J., Kushner, S.A., Elgersma, Y., (2015).HCN channels are a novel therapeutic target for cognitive dysfunction in Neurofibromatosis type 1 Mol. Psych.20, 1311-1321.

Goorden, S.M.I., Abs, E., Bruinsma, C.F., Riemslagh, F.W., Van Woerden, G.M., Elgersma, Y., (2015). Intact neuronal function in Rheb1 mutant mice: implications for TORC1 based treatments Hum. Mol. Gen. 24:3390-8

Van Der Vaart, T., Overwater, I.E., Oostenbrink, R., Moll, H.A., Elgersma, Y., (2015). Treatment of cognitive deficits in genetic disorders: a systematic review of clinical trials JAMA Neurol.

Abs, E., Goorden, S.M.I., Schreiber, J., Overwater, I.E., Hoogeveen-Westerveld, M., Bruinsma, C.F., Aganovic, E., Borgesius, N.Z., Nellist, M., and Elgersma, Y. (2013). TORC1-dependent epilepsy caused by acute biallelic Tsc1 deletion in adult mice. Ann Neurol 74, 569-579.

Van Der Vaart, T., Plasschaert, E., Rietman, A.B., Renard, M., Oostenbrink, R., Vogels, A., de Wit, M.C.Y., Descheemaeker, M.-J., Vergouwe, Y., Catsman-Berrevoets, C.E., Legius, E., Elgersma, Y., Moll, H.A., (2013). Simvastatin for cognitive deficits and behavioural problems in patients with neurofibromatosis type 1 (NF1-SIMCODA): a randomised, placebo-controlled trial. Lancet Neurol 12, 1076-1083.

Professor Miikka Vikkula
Brussels, Belgium

Prof Vikkula obtained his M.D. at the University of Helsinki in 1992 and his Ph.D. in molecular genetics, in 1993. He was a Research Associate at Harvard Medical School 1993-1997, during which time he became interested in vascular and lymphatic anomalies. With his wife, Prof Laurence Boon, Plastic Surgeon, Co-ordinator of the Vascular Anomaly Center, Brussels, the couple discovered the gene for familial venous malformation in 1996, and since then many others. They settled in Brussels in 1997, where Dr Vikkula developed his own laboratory. He obtained a "docentship PhD" in 2000, and was nominated Assistant Professor at the Faculty of Medicine in UCL. He is a member of the Directorate of the de Duve Institute since 2004, and a full professor of Human Genetics since 2013. He has received numerous honours and awards; most recently, the Inbev-Baillet Latour Clinical Prize in 2013. He served as president of the Belgian Society of Human Genetics 2004-2008, and as a member of the Scientific Program Committee of the European Society of Human Genetics 2008-2012. He is a Member of the Royal Belgian Academy of Medecine since 2012. Prof Vikkula is well known internationally as a major contributor to the understanding of molecular basis of vascular anomalies and lymphedema with >150 peer-reviewed pulications and numerous chapters in major bio-medical text books.

Professor Partha Majumdar
Calcutta, India

PARTHA P. MAJUMDER is a Professor of Human Genetics in the Indian Statistical Institute, Kolkata, and the founder of the National Institute of Biomedical Genomics, Kalyani, West Bengal, where he is currently a Distinguished Professor. He is also a Sir J.C. Bose National Fellow. He has pioneered the development of many statistical methods in the analysis of human genome diversity and genetic epidemiology. His work on genetics of human populations in India has provided a deep insight into the peopling of the Indian subcontinent. He is currently the national co-ordinator of the Indian project of the International Cancer Genome Consortium. He is an elected Fellow of all the three science academies of India and of TWAS (The World Academy of Sciences, Trieste). He has served on the Board of Directors of the International Genetic Epidemiology Society (IGES), and was the founding Chair of its ELSI Committee. He has worked for the International Bioethics Committee of UNESCO. He is currently a Council Member of the Human Genome Organisation. He is a recipient of TWAS Biology Prize - 2009, G.D. Birla Award for Scientific Research - 2002, Ranbaxy Research Award in Applied Medical Sciences - 2000, and the New Millennium Science Gold Medal, Government of India, 2000.

Dr Michael Fisher
Philadelphia, USA

His research focuses on identifying new treatments and novel biomarkers (particularly using new imaging modalities) and exploring functional outcomes for children with tumors associated with NF1. Dr. Fisher is Chair of the Steering Committee of the Department of Defense NF Clinical Trials Consortium. He is Chair of the Visual Outcomes Committee and member of the Steering Committee for REiNS (Response Evaluation in Neurofibromatosis and Schwannomatosis), an international effort to develop standardized outcome measures for clinical trials. In addition, he serves as co-leader of a CTF-funded, large, international, multi-institutional, prospective longitudinal study of patients with newly diagnosed NF1-associated optic pathway glioma, and is co-PI of the NF1 Low Grade Glioma Synodos project.

Dr Suma Shankar
California, USA

Dr. Suma Shankar is an Associate Professor in the Department of Pediatrics, Division of Genomic Medicine at UC Davis Medical Center. She is the Director of Precision Genomics and will lead the effort in bringing genomics to every day clinical practice. She is working on integrating genomic medicine with multiple specialties in pediatrics and adults for rare Mendelian diseases and complex disorders. Dr. Shankar is a clinician scientistwho received her medical degree from Bangalore Medical College, Bangalore and PhD in Molecular Geneticsfrom University of Iowa, USA. She isan ophthalmologist trained in United Kingdom and is a Fellow of the Royal College of Surgeons (FRCS),Edinburgh and Member of the Royal College of Ophthalmologists (MRCOphth), London. Shealso completed afellowship in Medical Genetics at the University of California, San Francisco and is board certified in Medical Geneticsby the American Board of Medical Genetics (FACMG).She was faculty in the Departments of Human Genetics and Ophthalmology at Emory University, School of Medicine, Atlanta, USA where she initiated Ocular Precision Health Initiative with biobank and genetic studies for inherited eye diseases for the first time in Georgia. She served as Medical Director for Emory Genetics Laboratory and was primary investigator on a number of clinical trials investigating novel therapies for rare genetic disorders. She has won the Western scholar and Henry Christian Award and Certificate of Excellence in Research from the American Federation Medical Research for research in ocular manifestations in RASopathies. She has a number of publications and several book chapters on genetic testing, ocular genomics, novel enzyme replacement, substrate, and chaperone therapies. Her chief research interests are gene discovery projects and translational research for genetic diseases.

A link to her publications can be found at this url.
http://scholar.google.com/citations?hl=en&user=u6FQarEAAAAJ&view_op=list_works&sortby=pubdate

SPEAKER

Dr Vijaya Ramesh
Boston, USA

Dr. Ramesh is an Associate Professor of Neurology (Genetics) at Harvard Medical School and an Associate Neurologist at MGH. She obtained her Ph.D. from University of Madras, India and completed postdoctoral training in the laboratories of James Gusella (MGH) and Dr. Vivian Shih (MGH). She is a member of the Center for Genomic Medicine at MGH. Ramesh laboratory has been investigating the pathophysiology of Neurofibromatosis 2 (NF2) and Tuberous Sclerosis Complex (TSC) for more than two decades. Her lab employs molecular, genetic, cell signaling and biochemistry techniques in disease-relevant human cells to discover pathways and mechanisms of pathophysiology in NF2 and TSC. The work on NF2 in human arachnoidal and meningioma cells discovered that NF2 protein merlin is a novel negative regulator of mTORC1/mTORC2 signaling. This work has been translated into clinical trials for NF2 and sporadic meningiomas. Ramesh lab has also established CRISPR-Cas genome editing technology in human arachanoidal cells, Schwann cells and iPSCs and has used this technique to create/correct mutations in NF1, NF2, TSC1 and TSC2 generating isogenic sets of human lines for drug screening and other research efforts.

SPEAKER

Dr Abeer Alsaegh
Oman

Dr Abeer Alsaegh is the Head of Genetics Department at Sultan Qaboos University and Consultant of Medical Genetics and Cancer Genetics at Sultan Qaboos University Hospital. Her clinical and academic interests includes familial cancer syndromes, developmental disorders and autosomal recessive syndromes associated with intellectual disability. She established the cancer genetics clinic in Oman and is an Oman Medical Specialty Board trainer.

Dr Joanne Ngeow
Singapore

Dr Joanne Ngeow, MBBS, MRCP, MPH is Senior Consultant, Division of Medical Oncology at the National Cancer Centre Singapore. Dr Ngeow currently heads the Cancer Genetics Service at the National Cancer Centre Singapore with an academic interest in hereditary cancer syndromes and translational clinical cancer genetics. She was awarded consecutive fellowships by the National Medical Research Council and the Ambrose Monell Foundation to complete formal clinical and wet bench training in Cancer Genomic Medicine at the Genomic Medicine Institute, Cleveland Clinic, Ohio. Dr Ngeow is an Editorial Board Member for Endocrine Related Cancers. Dr Ngeow was awarded the NMRC Clinician Scientist Award in 2017 aimed at understanding how gene-environmental and gene-gene interactions predisposes to cancer initiation and progression.

SPEAKER

Dr Anant Tambe
Manchester, United Kingdom

Dr Anant Tambe is a complex spinal surgeon with a special interest in paediatric spinal surgery. He works at Salford Royal Foundation Trust in Manchester which is the largest spine surgery centre in Europe. He also practices at the Royal Manchester Children's Hospital. A significant part of his practice includes correction of complex spinal deformities of the cervico-thoracic and thoracolumbar spine, and he regularly deals with neuromuscular scoliosis, metabolic bone diseases and neurofibromatosis.

SPEAKER

Dr Sue Huson
Manchester, United Kingdom

Dr Huson trained in medicine in Edinburgh and after general training in adult medicine and neurology, moved to a research post Professors Peter Harper and Alastair Compston in Cardiff. She started the post as a would-be neurologist and switched to a clinical genetics career mid-way. Her Welsh population based study of neurofibromatosis type one (NF1) and gene mapping studies led to the award of MD with gold medal and distinction. She then trained in clinical genetics at the Kennedy Galton Centre, London. As a consultant, first in Oxford and then in Manchester, she developed her interest and expertise in the diagnosis and management of all forms of neurofibromatosis. From April 2009 – April 2017 she led the nationally commissioned service for complex NF1 in the north of England. Sue has co-edited two books, numerous book chapters and research papers on neurofibromatosis.

Dr Jaishri Blakeley
Maryland, USA

Dr. Jaishri Blakeley is Associate Professor of Neurology, Oncology and Neurosurgery, director of the Johns Hopkins Comprehensive Neurofibromatosis Center (JHCNC) and director of the Neurofibromatosis Therapeutic Acceleration Program (NTAP). NTAP was founded in 2012 to harness intellectual power, essential resources and the power of collaboration to develop effective therapies for the nerve sheath tumors afflicting patients with neurofibromatosis type 1 (NF1). Dr. Blakeley is also an active member of the Department of Defense Neurofibromatosis Clinical Trials Consortium where she serves as the Johns Hopkins site principle investigator and committee chair for Schwannomatosis, a member of the executive committee for the Response Endpoints in Neurofibromatosis and Schwannomatosis International Consortia where she helps develop and validate endpoints for clinical trials addressing NF1, NF2 and Schwannomatosis and a co-investigator on the first dedicated Specialized Programs of Research Excellence award focused on NF1. She was the co-chair of the 2015 CTF International Neurofibromatosis Conference and the 2016 Society for Neuro-Oncology Education day and has leadership roles at the American Society for Clinical Oncology, the Society for Neuro-Oncology and the American Academy of Neurology (currently chair of the Neuro-Oncology section). She is also invested in mentoring the next generation of physician scientists through the Francis Collins Scholars Program (through NTAP) and as the director of the UCNS Neuro-Oncology Fellowship at Johns Hopkins. Dr. Blakeley's research expertise is in the development of clinical trials for nervous system tumors and specifically, early clinical-translational studies including tumor pharmacokinetic and pharmacodynamic investigations, imaging biomarkers for rare tumors and incorporation of patient focused, functional endpoints into efficacy studies. These efforts are all in the service of improving the outcomes for the patients for whom Dr. Blakeley provides care as an active clinician in neuro-oncology with a focus on neurofibromatosis. The quality of her patient care and her contributions have been recognized with several awards including the Thomas Preziosi Award for Clinical Excellence in Neurology at Johns Hopkins, the Frank Ford Memorial Neurology Teaching Award at Johns Hopkins, the Neurofibromatosis Mid-Atlantic "Make a difference award" and the Children's Tumor Foundation Humanitarian Award.

Professor Pierre Wolkenstein
Paris, France

Dr Pierre Wolkenstein is MD, PhD, dermato-oncologist at Henri-Mondor Hospital (Assistance Publique-Hôpitaux de Paris). He is Professor at University Paris Est Creteil, Vice-Dean for Academic Affairs and Head of the Department Cancers Immunity Transplantation Infections. He is also the coordinator of the National Referral Centre for Neurofibromatoses in France.

Pierre Wolkenstein focused his interest in care, basic, translational, and clinical research in the field of neurofibromatoses, and mainly on neurofibromatosis 1 (NF1). The French Network managed more than 3000 patients with NF1. The papers of Pierre Wolkenstein are centered on severe phenotypes, genotypes-phenotypes correlations, identification of modifying genes and animal models. His index H is 41. From 2-6 November 2018, the French team will organize the first Global Joint Neurofibromatoses Conference.

Professor Scott Plotkin
Boston, USA

Professor Scott Plotkin, MD, PhD, is a Professor of Neurology at Harvard Medical School and is Director of Cancer Neurology at Massachusetts General Hospital. He graduated magna cum laude from Harvard University and received an MD and PhD in neuroscience from Tulane University School of Medicine. He completed his internship in internal medicine at Tulane and then neurology residency at Massachusetts General Hospital and Brigham & Women's Hospital in Boston, MA. Upon completion of his residency, Dr. Plotkin completed a clinical fellowship in neuro-oncology at Massachusetts General Hospital and was a post-doctoral fellow at the Schepens Eye Research Institute of Harvard Medical School.

Dr. Plotkin joined the faculty of Harvard Medical School in 2003 in the Department of Neurology. He is currently Director for the Family Center for Neurofibromatosis as well as director of the MGH/DFCI/BWH Neuro-Oncology Fellowship program. Dr. Plotkin's research focuses on developing clinical trials for patients with NF1, NF2, and schwannomatosis. He has served as principle investigator on single and multi-center clinical trials and is an active member of several national consortia. In 2011, he co-founded the Response Evaluation in Neurofibromatosis and Schwannomatosis (REiNS) International Collaboration. This collaboration has published consensus recommendations for clinical trial endpoints in trials for NF patients and continues to work closely with the FDA, Cancer Therapy Evaluation Program, and other agencies.

SPEAKERS

Professor Rajiv Sarin

Mumbai, India

Professor T Rajkumar

Chennai, India

Professor Meena Bhatt

Banglore, India

Professor Vic Riccardi

California, USA

Dr Anat Stammer

INTERNATIONAL SYMPOSIUM ON NEW DEVELOPMENTS IN NEUROFIBROMATOSES AND RASOPATHIES

Their Management, Diagnosis, Current and Future Therapeutic Targets

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