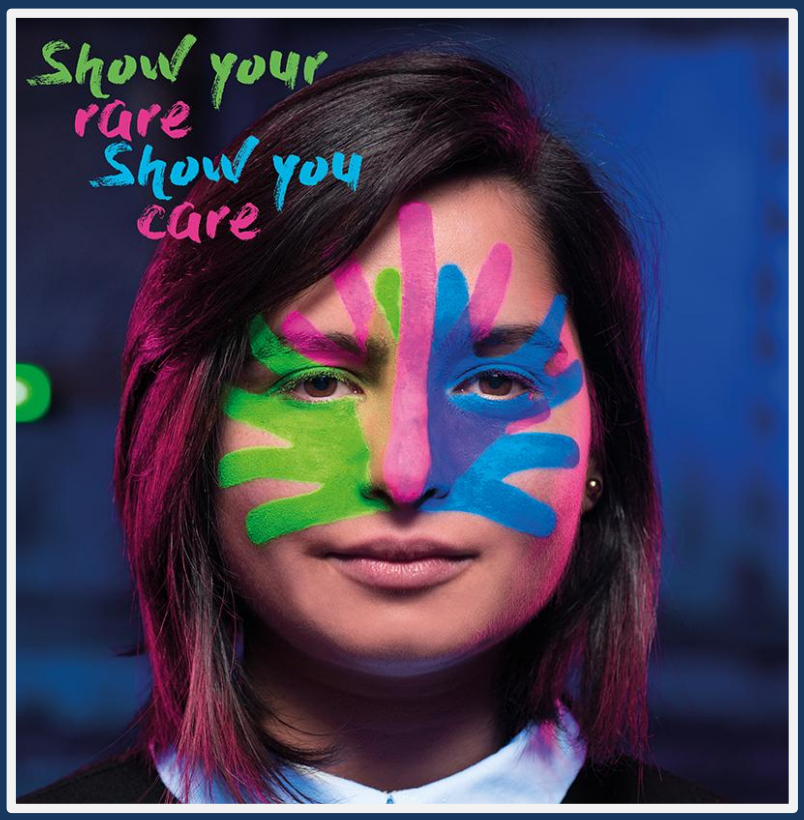
RARE DISEASE SYMPOSIUM



03rd May 2018
8.30 am – 1.00 pm

George Moore Auditorium
UCD O'Brien Centre
for Science

#UCDRDS2018
#ShowYourRare

UCD DUBLIN

medical research charities group
Unity is our Strength

IPPOSI

Rare Diseases Ireland

EURORDIS
Rare Diseases Europe

SCHEDULE

0830-0900	**Registration & Networking | Music by Ms Alison Sweeney**	
	Chair: **Philip Watt**, Chief Executive, Cystic Fibrosis Ireland	
0900-0910	Welcome Address	**Prof Orla Feely**, UCD Vice President for Research Impact and Innovation
0910-0920	Rare Diseases- Progress at National & European Context	**Prof Eileen Tracey**, Clinical Lead, National Clinical Programme for Rare Diseases
0920-0930	UCD Academic Centre on Rare Diseases updates	**Dr Sean Ennis**, Director UCD Academic Centre on Rare Diseases
0930-1010	**Conversation: Innovative Approaches to Rare Diseases** Chair: **Dr Avril Kennan**, CEO, Medical Research Charities Group Participants: • **Prof Thilo Kroll**, Professor of Health Systems Management, HRB PPI Ignite Lead for UCD, UCD School of Nursing, Midwifery and Health Systems ▪ **Dr Atif Awan**, Consultant Pediatric Nephrologist, Temple Street Children's Hospital ▪ **Dr Sally Ann Lynch** , Consultant Clinical Genetics, Our Lady's Children's Hospital Crumlin ▪ **Jean-Louis Roux,** Senior Director Government Affairs EUMEA, BioMarin Europe Ltd.	
1010-1030	**Coffee Break**	
	A Cross-Border Perspective on Rare Diseases	
1030-1050	Social Care in Rare Diseases 'INNOVA Care Project'	**Raquel Castro**, Head of Social Policy EURORDIS
1050-1110	Stronger- Together	**Julie Power**, Vasculitis Ireland

SCHEDULE

1110-1200	**Conversation: Nothing about us without us - Holistic Care** Chair: **Dr Aoife Brinkley,** Senior Clinical Psychologist, The National Centre for Inherited Metabolic Disorders (NCIMD) Participants: ▪ **Dr Suja Somanadhan**, Assistant Professor of Children's Nursing, UCD School of Nursing Midwifery and Health Systems ▪ **Anne Lawlor**, Parent, Chairperson of the 22q11 Ireland Support Group ▪ **Gary Woods**, My sibling is my hero, Mucopolysaccharidosis III or Sanflippo Syndrome ▪ **Maureen Mason**, Transition Nurse Coordinator, National Rare Diseases Office	
1200-1220	Chair: **Dr Paula Byrne** Associate Professor, UCD School of Medicine Rare Diseases Student's Perspectives ▪ **Kate Bailey,** Medical Student ▪ **John Hutchings,** Nursing Student	
1220-1240	Rare Diseases and Funding Opportunities	**Kay Duggan-Walls,** Health Research Board of Ireland (HRB), EU Programmes Officer & National Contact Point for Health, Horizon2020
1240-1250	Research Support UCD	**Dr Anne-Louise Holloway**, Research Programme Officer, National Programmes, UCD Research & Innovation
1250-1300	Key Learning and Next Steps	**Dr Derick Mitchell**, Chief Executive, IPPOSI
1300-1400	**Refreshments and Networking**	

BIOGRAPHIES

Prof Orla Feely, Vice President for Research, Innovation & Impact, UCD

Orla Feely is Vice President for Research, Innovation and Impact and a Professor of Electronic Engineering at University College Dublin. She holds a BE degree from University College Dublin and MS and PhD degrees from the University of California, Berkeley. Her research interests lie in the area of nonlinear electronic circuits, including MEMS, energy harvesters, power converters and phase-locked loops.

Prof Eileen Treacy, MD, FRCPI, FRCPC, FCCMG (Clinical Genetics) Clinical Lead, National Clinical Care Programme for Rare Diseases

Prof. Treacy trained in Paediatrics and in Clinical and Biochemical Genetics in Canada, France and Australia. She was appointed as Assistant and then Associate Professor in Paediatrics and Human Genetics at McGill University, Montreal, Canada before taking up a consultant appointment as Metabolic Consultant at Children's University and Mater Misericordiae University Hospitals in 2001. Prof. Treacy is a Clinical Professor of Inborn Errors of Metabolism at Trinity College Dublin and is a Full Clinical Professor at University College Dublin. Prof. Treacy has specific research interests in Rare Diseases, Inherited Metabolic Disorders and the treatment of Genetic Diseases and has published extensively in this area. She is an Investigator with the UCD Academic Centre on Rare Diseases, currently Co-Lead of the European Galactosaemia (GalNet) Consortium, Principal Investigator for a HRB-HRA Galactoseamia research grant and national coordinator for the EC 3rd Public Health grant in Rare Diseases (RD-Action 2015-2020).

Dr Sean Ennis, PhD (Genetics) Director of the UCD Academic Centre of Rare Diseases (ACoRD)

Sean has life-long experience in research in medical genetics. He has led and been involved in establishing collaborative approaches to investigating chronic conditions such as Motor Neuron Disease (ALS), Autism Spectrum Disorder and, more recently, Rare Diseases. He has extensive published research studies and is a frequent speaker in the area of genomics. Sean holds a PhD in genetics from University College Dublin. He is a lecturer at the School of Medicine & Medical Sciences at UCD, Investigator with the National Centre for Medical Genetics and National Children's Research Centre. He is director of the UCD Academic Centre of Rare Diseases (ACoRD).

Philip Watt, CEO of Cystic Fibrosis Ireland

Philip Watt has been CEO of Cystic Fibrosis Ireland since 2009 and since 2013, he has been Chairperson of the Medical Research Charities Group (MRCG). Philip has worked in both the voluntary and statutory sector in Ireland, including secondments to the Department of Justice and Equality and has written extensively on health, equality and human rights issues

Kay Duggan-Walls, Health Research Board of Ireland (HRB), EU Programmes Officer & National Contact Point for Health, Horizon2020

Kay Duggan-Walls is EU Programmes Officer at the Health Research Board (HRB), the leading agency supporting and funding health research in Ireland. She works as National Contact Point for the EU Horizon 2020 funding programme in the Health, Demographic Change and Well-being challenge as part of the National Support Network. She leads the delivery of support to Irish health researchers providing expert advice and hands-on support to maximise successful involvement in the Horizon 2020 process. She also works with European counterparts in the development of the programme and associated policy initiatives in research and innovation. Kay is also the National Focal Point for the EU Public Health Programme and also represents Ireland on the Management Board of the EU Joint Programming Initiative on Antimicrobial Resistance. Kay has been employed by the HRB since November 2000 in research funding management, managing a variety of award programmes in health research. She holds a BSc in Microbiology from NUI Galway and an MSc from the Open University. Kay was also National Contact Point for FP7 Health, and has supported many successful health researchers in FP7.

Prof Thilo Kroll Professor of Health Systems Management

Thilo Kroll is Professor of Health Systems Management in the UCD School of Nursing, Midwifery and Health Systems and Executive Team Member of the UCD Geary Institute for Public Policy. He has a PhD in Psychology and over 25 years of interdisciplinary research and academic experience. Prior to coming to Ireland he was Professor of Disability and Public Health at University of Dundee and Co-Director of the interdisciplinary Social Dimensions of Health Institute (SDHI) of Universities of Dundee and St Andrews. He has conducted interdisciplinary research with a focus on disability- and health-related topics and inclusive methodologies across the life span in Germany, Norway, England, Scotland, the United States and Ireland. In his youth, he worked as an advocate for young people with arthritis. His PhD focused on social inclusion and school re-entry of children and adolescents with cancer in Germany. In the United States he worked with young people with spinal injuries surviving gun violence. Currently, he supports the implementation of Public and Patient Involvement in health- and social care related work at UCD and beyond as part of the HRB-funded PPI Ignite Initiative.

Dr Sally Ann Lynch, Consultant Clinical Geneticist, Our Lady's Hospital, Crumlin, Children's University Hospital, Temple Street, National Rare Diseases Office, MMUH

My research interests are in rare diseases (RD). Our group have had success in identification of several RD genes and developed simple cost-effective genetic tests, which have been translated into the diagnostic laboratory. Some of these disorders are unique to Ireland. Local research is important as researchers can feedback results quickly which generates interest and new collaborations. I have also an interest in education using new technologies. We have developed a microsite, http://www.ucd.ie/medicine/rarediseases/ which contains information for use by GPs and other health care professionals on common genetic topics. We have developed twenty five educational videos that have been up-loaded to the UCD YouTube channel and have in excess of 370,000 views in more than 200 countries to date. More recently I have catalogued RDs in the Irish Traveller population and together with a specialised group we are at the early stages of developing an online tool to facilitate early diagnosis of RDs found amongst the Irish Traveller population.

Avril Daly, Vice-President of EURORDIS

Avril Daly is the CEO of Retina International the global patient-led umbrella group dedicated to the promotion of research into Rare and Inherited as well as Age-related forms of retinal degenerative conditions. She previously held the position of CEO at Fighting Blindness Ireland for eight years. Avril is the Vice President of EURORDIS (Rare Disease Europe) and chairperson of the Irish National Alliance for Rare Diseases, RDI and was a member of the steering committee working towards the publication of the Irish National Plan for Rare Diseases, (2014) and now represents the patient voice on the working group of the Clinical Programme for Rare Diseases at the Irish Health Service Executive (HSE).Avril was a member of the Medical Research Charities Group (MRCG) and, the Irish Platform for Patients' Organisations, Science and Industry (IPPOSI).She served two terms as Board Member of the European Patients Forum (EPF) for two terms and as Board member of European Platform for Patients Organisations Science and industry (EPPOSI) for two terms.

Ms Raquel Castro, Social Policy Senior Manager at EURORDIS-Rare Diseases Europe

Raquel Castro is Social Policy Senior Manager at EURORDIS-Rare Diseases Europe, where she is responsible for managing EURORDIS' activities related to the integration of rare diseases into social services and policies. Raquel is currently in charge of INNOVCare project, focused on promoting integrated care for people living with a rare disease. She is also involved in the RD-Action, the Joint Action for rare diseases. Previously, she was assigned to the European Union Committee of Experts on Rare Diseases (EUCERD) Joint Action, conducting the mapping of specialised social services, compiling social care good practices and supporting the development of the Commission Expert Group on Rare Diseases Recommendations to Support the Incorporation of Rare Diseases into Social Services and Policies. Prior to joining EURORDIS, Raquel coordinated the Portuguese Help Line for rare diseases (Linha Rara) and worked with international NGOs, in humanitarian and development projects in Europe and in Asia. Raquel holds a Bachelor of Arts in Communication as well as a post-graduate degree in Project Management at the Lisbon School of Economics and Management.

Dr Suja Somanadhan, Assistant Professor in Children's Nursing

Dr Suja Somanadhan is an Assistant Professor in Children's Nursing at the UCD School of Nursing, Midwifery & Health Systems. She obtained her PhD from the University College Dublin (UCD) in collaboration with National Centre for Inherited Metabolic Diseases (NCIMD) and Temple Street Children's University Hospital. The purpose of pursuing this inquiry was to gain a deeper understanding of what it is like to be a parent of a child, adolescent or young adult with Mucopolysaccharidoses (MPS) a rare genetic disease. Dr Somanadhan's clinical background is children's nursing, and she has over 18 years of clinical experience in paediatric health care settings mainly focused on neonatology, metabolic and endocrine disorders (rare diseases) and general paediatrics. Her research interests are psycho-social aspects of rare genetic disorders and chronic illness in children, adolescents and young adults, and service user engagement in health care to inform policy and practice. She has more specific passion towards Evidence-Based Health Care, Quality & Patient Safety and holds vast experience utilising clinical audit as a tool for Quality Improvement in the clinical settings. Currently, reviewer of the high impact journals such as Orphanet Journal of Rare Diseases and The International Journal of Health Care Quality Assurance (IJHCQA). Also, significant experience in grant writing and obtaining research grant as a principal investigator.

Dr Avril Keenan, CEO, Medical Research Charities Group (MRCG)

Avril has a PhD in genetics and many years subsequent lab experience working on rare human genetic conditions. She moved from the lab in 2007 to become a patient advocate with the rare disease patient organisation DEBRA Ireland. In her role there, as Head of Research and Advocacy, she led a number of international initiatives including the development of evidence-based clinical guidelines and an international patient registry. Avril has sat on many Boards and Committees, including the DEBRA International Executive Committee and the Rare Diseases Ireland Board. In her role as CEO of the MRCG she is passionate about supporting medical research charities and promoting their role in society. She was one of the founders of the Irish Health Research Forum which is currently managed by the MRCG.

Julia Power, Vasculitis Ireland

Prior to diagnosis in 2005 with a rare disease called Granulomatosis with Polyangiitis Vasculitis, Julie worked for 17 years as an Occupational Therapist, specialising in Neurology and Palliative care. Following diagnosis, her life and that of her family's was totally changed. The uncertainty and isolation Julie experienced on her patient journey has fuelled her interest in raising awareness, improving care and research. Julie believes knowledge is power and that patient involvement throughout the entire treatment and research process is mutually beneficial to the Researchers, Clinicians, Service Providers and Patients. In 2010, she founded Vasculitis Ireland awareness, an All-Ireland support group for anyone affected by Vasculitis in Ireland, liaising closely with Vasculitis UK and the Vasculitis Foundation. Julie is a patient representative in the Irish Rare Kidney Disease Registry and Bio bank Steering Committee, the recently formed Vasculitis Ireland Network (VINE) and in planning for a pilot NI Vasculitis service. She graduated as a EUPATI (European Patient Academy in Therapeutic Intervention) fellow in 2016 after completing the 14month intensive research and development course. Julie has been a member of the Northern Ireland Rare Disease Partnership(NIRDP) board of directors since 2015. This year, she was invited onto the Irish Platform for Patient Organisations Science and Industry board (IPPOSI) and is actively involved in improving care for those affected by rare disease both in NI and ROI.

Assoc Professor Paula Byrne BA PhD

Paula Byrne is an Associate Professor in the School of Medicine, UCD who has a special interest in Rare Genetic Disorders. She has worked on molecular pathology of neurodegenerative hereditary spastic paraplegias for a number of years in collaboration with Prof Michael Hutchinson. She also has a keen interest in genetics in medical education and has developed innovative methods to equip medical students with the necessary skills and knowledge to become competent and caring healthcare professionals who are life-long learners. In response to evolving advances in biomedical science and technology and to the needs of society she developed an innovative elective module on *Rare Genetic Disorders and the Medical Healthcare Professional* which aims to increase awareness among future medical healthcare professionals.

Marueen Mason, Transition Nurse Coordinator, Nation Rare Diseases Office

Maureen is presently working as the National Rare Disease Transition Coordinator based in the National Rare Diseases Office, Mater Misercordiae Hospital..
She is a qualified RGN (SVUH) and RM (The Coombe Hospital) with nursing experience in Ireland, UK, U.A.E and Australia mainly in midwifery.
She also has diploma's in SHWW (UCD) and FLMt (NCI). Maureen has over 20 years' experience in the Pharma Industry with cross functional roles in Sales, Marketing, Market Access, Advocacy and Strategic Business Development. Her experience is across many therapy areas, particularly Respiratory, Cardiology and Neurology. Maureen is also employed as a Marketing & Medical Information Manager with an SME in radiopharmaceuticals. She is currently on the Advocacy sub-committee in Fighting Blindness with previous experience on projects with Retina International.

Jean-Louis Roux, Senior Director Government Affairs EUMEA, BioMarin Europe Ltd

Jean-Louis is currently serving as Senior Director at BioMarin EUMEA, a California-based biotech pharmaceutical company active in the field of orphan medicines for the treatment of rare to ultra-rare diseases, and currently at the forefront in the research and development of breakthrough cell and gene therapies. At BioMarin since 2017, he is in charge of developing the company's regional Government Affairs department. His professional experience over the past 13 years has been predominantly in public health and rare diseases, with various successive roles in corporate, NGO and consultancy environments, which have seen him develop and run a wide range of successful government affairs and patient advocacy campaigns both at the EU level in Brussels and locally in EU Member States. Prior to BioMarin, Jean-Louis worked as Director at government affairs consultancy Incisive Health International in London and, from 2015 to 2017, as Public Affairs Director at EURORDIS-Rare Diseases Europe, the leading patient advocacy organisation representing people living with a rare disease across the continent, where he played an instrumental role in European and national debates on access to orphan medicines, whilst also establishing more firmly the voice of people living with rare diseases at the United Nations with the inception and launch of the NGO Committee for Rare Diseases in New York on 11 November 2016. A French national, Jean-Louis is a father of two and shares a personal family connection to the cause of rare diseases.

Gary Woods, My sibling is my hero

Gary Woods is a sibling to Rebecca Woods, who has Mucopolysaccharidosis III or Sanflippo Syndrome. Gary plays an active role in caring for his sister and has also previously volunteered for charities related to the condition. Gary works in politics and has a particular interest in the not for profit sector focusing on the wellbeing and support of siblings of those with special needs.

Anne Lawlor, Parent, chairperson of the 22q11 Ireland Support Group

Anne is a founding member and chairperson of the 22q11 Ireland Support Group. The organisation, set up in 2007 supports families affected by 22q deletion syndrome and strives for the improvement of services for those affected by this and other rare conditions. In her leadership position Anne is a driver of real change. She networks extensively with local, national and international researchers, clinicians and mental healthcare professionals. Anne is recognised for her ability to draw people together and is well known both in Ireland and abroad as a true rare disease advocate. A recipient of a 2017 Global Genes Rare Champion of Hope Award Anne also accepted two Charity Impact Awards on behalf of 22q11 Ireland last year. Dedicated to raising awareness of 22qDS as a poorly understood and under-recognised condition Anne completed a Masters in Management of Community and Voluntary Groups and works in a voluntary capacity with 22q 11 Ireland. She lives in Dublin with her daughter Áine aged 34 who was diagnosed with 22qDS aged 15.

Dr Anne-Louise Holloway

Dr Anne-Louise Holloway currently holds a dual role in UCD Research as National Research Programme Officer and Proposal Support team lead. The proposal support team is the first point of contact for UCD researchers applying for research funding. In her capacity as National Research Programme Officer designs pre-call supports for the national research funding schemes. Anne-Louise has first-hand academic research experience gained in both Ireland and the UK and has considerable research administration experience, gained firstly working for a funding agency (EPSRC) in the UK and more recently as a university research officer within the Irish Higher Education sector. Anne-Louise is a UCD graduate with a BSc and PhD in Chemistry

Dr. Derick Mitchell, Chief Executive of IPPOSI

Derick has over ten years experience of working in patient involvement, multi-stakeholder management, scientific communications and advocacy at both the national and European level, and has a strong interest in the area of patient and public involvement in research. Derick leads IPPOSI's participation in the European Patients Academy (EUPATI), an initiative which is training patients to become involved in the medicines R&D process, and has spread the IPPOSI public-private partnership model to over 20 countries. Derick is a member of a number of national and international boards including the EHealth Ireland committee; the oversight committee for the National Rare Disease Plan; the Medical and Life Sciences Committee of the Royal Irish Academy, the International Advisory Board of the HRB-Trials Methodology Research Network; the HIQA Research Ethics & HTA Advisory Boards, among others. Derick graduated with a BSc. (Hons) in Biotechnology from NUI Galway (2000) followed by a PhD in Molecular Medicine from University College Dublin (2004).

Dr Aoife Brinkley, Senior Clinical Psychologist, The National Centre for Inherited Metabolic Disorders (NCIMD)

Brinkley is a Senior Clinical Psychologist who has worked in the area of paediatrics for fifteen years, having completed her doctoral training in University College Dublin. For the last ten years she has worked in Temple St. Children's University Hospital within the speciality of Inherited Metabolic Disorders. During this time she has had the privilege of working with many children and adults with rare diseases and their families, offering psychological support in relation to the varied challenges associated with chronic health conditions. Dr. Brinkley has also conducted research in a range of areas relating to paediatric psychology, including a number of studies investigating neuropsychological sequelae of inherited metabolic disorders.

Alison Sweeney, Music Therapist

Alison Sweeney qualified with a Masters in Music Therapy from the University of Limerick where she studied under Prof. Jane Edwards and Dr. Triona McCaffrey. Alison comes from a very musical background having first obtained a Bachelor of Music degree from University College, Cork. She specialises in paediatric hospital care and has broad experience working with hospitalised children in the areas of neurosurgery, neurology, life limiting conditions and paediatric palliative care. Alison set up and runs the music therapy service in Temple Street Children's University Hospital as well as the National Children's Hospital, Tallaght. Alison's research interests lie in the areas of stress and coping, interdisciplinary team work and augmentative and alternative communication systems. She regularly works jointly with speech and language therapists, occupational therapists, physiotherapists and psychologists and is currently involved in a piece of research examining the use of upper limb stretching songs in the treatment of children with upper limb weakness in the acute neurosciences.

John Hutchings, Children's & General Nursing Student, UCD

John is currently pursuing a degree in Children's and General Nursing in UCD (3rd year). Previously John completed a BSc in Genetics in UCC. Johns strives to incorporate his current knowledge of genetics into his future nursing career.

Kate Bailey, Medical Student, UCD

Kate Bailey is a Stage 4 medical student in University College Dublin. From Ramelton in Donegal, she has a keen in interest in travel and the outdoors.

Justin Dawson
Justin is currently employed as an Audio Visual Technician at the UCD School of Nursing, Midwifery & Health Systems. AVIXA CTS ® holder. 2018 AV Technology Europe Finalist. 2017 AV Awards Finalist. Passion for media, tech and Scouting!

Aisling Jackman
Aisling is currently employed as a Research Administrator at the UCD School of Nursing, Midwifery and Health Systems, and one of the main organiser for the Rare Disease Symposium in UCD.

A special thanks to the Rare Disease Symposium volunteers:

- Dr Claire Magner
- Rachel Howe
- Cian Milofsky
- Emma Dorris
- Alison Sweeney
- Niamh Morgan
- John Hutchings
- Kunal Ntin Patil
- Marie O'Flanagan
- Frances Howlin

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