5th Annual Dublin Cystinosis Workshop 2019

Speaker Profiles

Dr Olivier Govaere, Institute of Cellular Medicine, Newcastle University, UK is a senior research associate based in the lab of Professor Dr Quentin Anstee and Professor Dr Ann K. Daly.

He is currently working on the EU Horizon 2020-funded EPoS 'Elucidating Pathways of Steatohepatitis' project. This consortium focuses on understanding the pathophysiology of non-alcoholic fatty liver disease and examining the determinants of individual risk for disease progression using a "multi-omics" translational-science approach.

Dr Govaere obtained his PhD in Molecular and Stem Cell Medicine at the KULeuven, Belgium. He did his project in the lab of Professor Dr Tania Roskams at the Department of Imaging & Pathology, focusing on the role of hepatic progenitor cells in liver regeneration and carcinogenesis. He continued his work during a 3-year postdoctoral fellowship investigating phenotype switching in human hepatocellular carcinomas and characterisation of the human hepatic progenitor cell niche in chronic liver disease.

Dr Koenraad Veys is a paediatrician and fellow in paediatric nephrology at the University Hospitals of Leuven in Leuven, Belgium.

He has been working on his PhD thesis since 2015 in the Laboratory of Paediatric Nephrology headed by Professor Dr Elena Levtchenko at KU Leuven, which he will be finalizing this year entitled: "Innovation in therapeutic monitoring and treatment of nephropathic cystinosis".

His research is focused on improving several contemporary aspects of the clinical management of cystinosis patients, including the development of alternative biomarkers for therapeutic monitoring, unravelling the infertility in male cystinosis patients, and developing cell-based gene therapeutic approaches to provide a cure for the kidney disease in cystinosis.

Christian Koeppl works as physiotherapist at the Centre of Social Paediatrics in Traunstein, Kliniken Suedostbayern AG, Germany.

As specialised therapist for children with developmental disorders or handicaps, Christian attended several advanced training courses such as neurodevelopmental treatment (Bobath-Therapy).

In 2011 he completed his bachelor studies leading to a Bachelor of Arts degree in Physiotherapy at THIM University of Applied Sciences in Physiotherapy in Nieuwegein/Netherlands.

Since 2014, Christian Koeppl is member of the interdisciplinary cystinosis consultation in Germany. Together with Dr Nadine Herzig he provides the orthopaedic /physiotherapeutic consultation, during which, amongst other things, he gives individual advices to the patients for activity in everyday life, for sports as well as for training and therapy.

Improving Motor performance and capacity as a long term and sustainable process in patients with cystinosis is an enduring challenge. Therefore, he is currently establishing and leading the Galileo-Study together with Dr Nadine Herzig and other health experts.

Dr Nadine Herzig works as senior consultant at the Specialist Centre for Paediatric and Neuro-Orthopaedics, Schoen Clinic Munich Harlaching in Munich, Germany.

Dr Herzig is member of the interdisciplinary cystinosis consultation in Germany since 2012. As orthopaedic surgeon she provides cystinosis patients with advice for conservative therapies and additionally performs surgery when necessary (e.g. correction of leg axis, foot corrections).

Together with Christian Koeppl she is currently working on a concept to improve motor performance in cystinosis patients and in this context will be co-leading the Galileo-Study.

Ms Christine Knerr is a speech and language therapist, Castillo-Morales®-Therapist and a Bobath instructor.

With over 25 years of working experience in the social paediatric centre in Traunstein, Germany, she has treated children with feeding disorders, dysphagia or other difficulties in speech or oral functions.

Since 2014 she is part of the cystinosis consultation, which is offered three to four times a year in Traunstein and Munich.

Patients are diagnosed and treated by up to 14 health professionals in one day in the sense of a one-stop clinic. Both children and adults are cared for.

Professor Dieter Haffner is Professor of Pediatrics and Head of the Department of Pediatric Kidney, Liver and Metabolic Diseases at Hannover Medical School.

His main interests are the genetic and mechanistic exploration of rare kidney diseases such as hereditary cystic kidney disease, X-linked hypophosphatemic rickets and mineral and bone disorders in children with chronic kidney disease (CKD-MBD).

Professor Haffner is coordinating the ESPN working group on CKD-MBD, the IPNA Best Practices and Standards Committee, and the Guidelines and Pathways Task Force of the European Reference Network for Rare Kidney Diseases (ERKNet). He has led, or been heavily involved in, several randomised controlled clinical trials, patient registries, and clinical practice recommendations. He has also published 181 original scientific articles, reviews, editorials and book chapters.

Professor Craig Langman, Isaac A. Abt, MD, Professor of Kidney Diseases, and Professor of Pediatrics at the Feinberg School of Medicine, Northwestern University, Head of Kidney Diseases at the Ann and Robert H Lurie Children's Hospital of Chicago, and director of the two Davita Children's Dialysis Centers in Chicago.

Professor Langman's funded research focuses on the basic and clinical expression of inherited or acquired disorders of calcium, phosphorus vitamin D, and FGF23 metabolism, mechansims of cardiovascular diseases in children with obesity or chronic kidney disease, the proteomics of inherited stone disease, inherited genetic diseases (cystinosis, oxalosis, kidney stones, atypical HUS, hypophosphatasia), and the rehabilitation of patients around the world with chronic kidney disease. Professor Langman has published more than 250 articles, reviews and chapters in his discipline.

Professor Langman has the following major areas of hypothesis-driven research that are studied and supported: urinary proteomics in genetic kidney stone diseases; mechanisms of progression in chronic kidney diseases; rare genetic kidney diseases including atypical HUS, cystinosis, and Alport Syndrome. His clinical focus

is rare genetic kidney diseases; osteoporosis in children Inherited and acquired bone disease in children; disorders of calcification; disorders of phosphorus metabolism; chronic kidney diseases of childhood.

Professor Emeritus Herbie (David Richard) Newell, Emeritus Professor of Cancer Therapeutics at Newcastle University, England, UK.

Professor Herbie Newell is Professor of Drug Development at the University of Sunderland and Emeritus Professor of Cancer Therapeutics at Newcastle University, England, UK. He was Director of Translational Research at Cancer Research UK from 2006 to 2009, and the founding Scientific Director of the Northern Institute for Cancer Research at Newcastle University.

Professor Newell was involved in the development of the registered cytotoxic anticancer agents carboplatin (Paraplatin®) and ralitrexed (Tomudex®), and the first-in-class PARP inhibitor rucaparib (Rubraca®). He is an author of over 230 scientific articles and was Editor-in-Chief of the journal Cancer Chemotherapy and Pharmacology until 2016.

In addition to his academic work, Professor Newell has consulted extensively for both pharmaceutical and biotechnology companies, and until 2016 he chaired the Medical Research Council Development Pathway Funding Scheme. In 2011, Professor Newell was elected to the UK Academy of Medical Sciences and received a C.B.E. in the 2019 New Year Honours list.

Prior to the tragic early death of Professor Roz Anderson, Professor Newell was acting as an advisor on the cystinosis project and agreed with Roz before her death that he would take over the role of Principal Investigator.

Professor Paul Goodyer is an MD, pediatric nephrologist and scientist. He is the Professor of Pediatrics and Human Genetics, McGill University in Canada. He is an

Professor Goodyer heads a research laboratory at the McGill University Health Center Research Institute focused on molecular control of kidney development as it pertains to hereditary renal disease.

With Nicoletta Eliopoulos of the Jewish General Hospital and an international team of collaborators, he is engaged in an effort to develop stem cell therapy for cystinosis, that is particularly prevalent among French Canadians.

Dr Manoe Janssen is a postdoctoral researcher in Experimental Pharmacology at the Utrecht Institute for Pharmaceutical Sciences in Utrecht University in The Netherlands.

Dr Manoe Janssen completed an MSc Molecular Mechanisms of Disease at the Radboud University Nijmegen in 2008 and started her PhD project at the department of Gastroenterology and Hepatology at Radboudumc. In January 2013 she moved to Leuven in Belgium to perform her postdoctoral project at the Stem Cell Institute.

She joined the group of Roos Masereeuw at the department of Pharmacology and Toxicology in Nijmegen in January 2015, and moved with the group to Utrecht in July 2015.

During her PhD she received the Young Hepatologist Award from the Dutch Society for Hepatology (NVH) and the IGMD Junior Award from the Institute for Genetic and Metabolic Disease (IGMD). In 2016 she received the Kolff Junior Postdoc Grant from the Dutch Kidney Foundation to continue her research into genetic kidney diseases.

As part of the Masereeuw group, Dr Janssen focusses on understanding pathways that can be pharmacologically triggered to enhance the repair and regeneration of the kidney. She focusses on genetic factors and gene mutations that affect proximal tubule function and the development of human in vitro cell models for drug screening. She makes use of the unique, patented, human renal cell lines from the Masereeuw group who have a high predictive value for drug and waste product transport and metabolism. Dr Janssen compares cells from heathy controls and patients to understand which pathways in the cell are affected, to search for druggable targets and evaluate phenotypic improvements. As part of an international consortium on the genetic kidney disease cystinosis she is testing different drug compounds as well as CRISPR/Cas9 mediated gene repair to find a new therapy for this disease.

Dr Laura Rita Rega is a postdoctoral researcher in the Department of Genetic and Rare Diseases at the Bambino Gesù Children's Hospital and Research Institute, Rome.

Dr Rega received her BSc in Biotechnological Sciences in October 2005 and MSc Specialization in Medical Biotechnology in July 2007 at the University of Naples "Federico II". Two months after Specialization, Dr Rega moved to the laboratory of Professor Maria Antonietta De Matteis in the Department of Cell Biology and Oncology at the Consorzio Mario Negri Sud Research Institute (Chieti, Italy). During these years she developed light microscopy methods for the visualization and stabilization of membrane contact sites between the endoplasmic reticulum and the trans-Golgi network. These structures are highly dynamic and only detectable at ultrastructural level. This ambitious project allows her to acquire cell biology and confocal microscopy skills. Dr Rega was awarded her PhD in Life Sciences in November 2012.

In January 2013, Dr Rega moved to the laboratory of Dr Francesco Emma at the Department of Genetic and Rare Diseases at the Bambino Gesù Children's Hospital and Research Institute in Rome. In the last years, she is collaborating on projects aimed at developing better therapies for nephropathic cystinosis. Dr Rega recently showed that stimulation of the activity of the transcription factor EB (TFEB) corrects some cystinotic cellular defects. She is currently confirming these in-vitro studies in animal models of cystinosis.

Dr Suja Somanadhan is Assistant Professor in Children's Nursing at the UCD School of Nursing, Midwifery & Health Systems.

Dr Somanadhan obtained a PhD, MSc (Clinical Practice), Higher Diploma in Children's Nursing and Graduate Certificate in Nurse Education from University College Dublin (UCD). She is a Registered Children's Nurse (RCN), Registered General Nurse (RGN) and Registered Nurse Tutor (RNT).

Dr Somanadhan has 18 years of clinical experience in paediatric health care settings working as a Staff Nurse, Clinical Nurse Manager, and Clinical Nurse Specialist and most recently (February 2013-April 2017) as Clinical Audit and Nursing Research Facilitator at Temple Street Children's University Hospital.

Dr Somanadhan has been involved in many patient quality and safety projects (PEWS, Sepsis, Consent, Clinical Handover etc.) and some International research studies including Evaluating processes of care and outcomes of children in hospital (EPOCH). Her PhD focused on qualitative research inquiry to understand parents experience of living with and caring for children with Mucopolysaccharidosis (MPS), a rare genetic life-limiting conditions.

Dr Somanadhan serves as an academic committee member at the Rare Disease Taskforce in Ireland, leading out on HRB Funded Rare Disease Research Partnership (RAinDRoP). She has extensive working experience and passion towards service user engagement in health care to inform policy and practice, with a focus on Evidence-Based Health Care, Quality & Patient Safety and holds vast expertise utilising clinical audit as a tool for Quality Improvement in the clinical settings.

Dr Hannah Blakey, Renal Research Registrar at the Queen Elizabeth Hospital Birmingham

Dr Hannah Blakey is a Specialist Registrar in Renal Medicine based in Birmingham, UK. She is currently undertaking a 'Renal Disease in Pregnancy' research fellowship and is registered at the University of Birmingham in an MD programme.

Her research is focused on exploring the role of complement in the development of pre-eclampsia, both in healthy pregnant women and those with Chronic Kidney Disease.

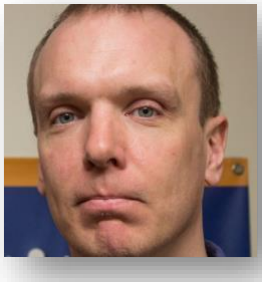
She is supervised by Dr Graham Lipkin, who runs the Cystinosis clinic service at the Queen Elizabeth Hospital Birmingham and was previously the National Lead Clinician for the Cystinosis Rare Disease Group.

They also run a monthly joint renal-obstetric antenatal clinic service for pregnant women with Chronic Kidney Disease, and have recent experience of successfully managing women with cystinosis through pregnancy.

Ms Anne Marie O'Dowd is a founding Director of Cystinosis Ireland and Chair of the Cystinosis Ireland Research Group.

Her day job in the field of adult antenatal education where she is an antenatal teacher and tutor.

She is the mother of Luke aged 18 with cystinosis who is her inspiration for all of this work.



Dr Brendan Keating, Assistant Professor Pediatrics and Surgery (Transplantation)

Brendan Keating, PhD, received his DPhil in molecular genetics from the Department of Clinical Medicine at University of Oxford, UK. He completed a post-doctoral fellowship at the Institute of Translational Medicine and Therapeutics (ITMAT) at UPenn, and was a visiting Scientist at the Wellcome Trust Sanger Institute, Cambridge, UK. Dr. Keating has led or co-authored over 135 genomic publications and has given over 100 presentations in 25 countries.

Dr Keating has designed and developed genome-wide and -omic tools with for cardiovascular and transplant related studies, in > 250,000 DNAs/samples resulting in over a hundred genetic discoveries. His current major research interests focus on the analyses of polymorphisms of transplant donor and recipient's genomes, with the aim of predicting genomic signals that may underpin graft rejection and complications of rejection.

Dr Keating is Principal Investigator of genome-wide association studies (GWAS) for solid-organ transplant cohorts within the Children's Hospital of Philadelphia, and of a number of kidney transplant -omic studies. He instigated the formation of an international genomics consortium (iGeneTRAIN) for large-scale genomic studies using > 56,000 patient samples from a number of international transplant studies (www.igenetrain.org). Dr Keating is also a member of the Pharmacogenomics and Return of Results working groups of the NIH-NHGRI electronic medical record and genomics (eMERGE) network which is integrating clinical genetic data into patient's electronic medical records for clinical decision support for individualize dosing of patients. Dr Keating is also PI of a prospective NIH funded study looking at post-transplant outcomes in 12 North American pediatric renal transplant centers, and 6 international sites.

Dr Ahmed Reda, Postdoctoral Researcher at KU Leuven and Vrije Universiteit Brussels, Belgium

Ahmed Reda, PhD, is currently a postdoc at KU Leuven and Vrije Universiteit Brussel, both in Belgium since 2017.

His main scientific interest is male fertility preservation. He works now on unravelling the mechanism and possible fertility preservation of male cystinosis patients. He was awarded his PhD from Karolinska Institutet, Sweden, in 2016, with the topic "Artificial testis to study early gonadal development and male germ cell differentiation".

He worked also as a researcher in Karolinska Institutet since 2010. Dr Reda also has an MSc in molecular biology and BSc in pharmaceutical sciences.

Mr Mick Swift is Deputy Chief Executive Officer and Research Director at Abbey Capital and Chairman of Cystinosis Ireland.

As Abbey's Research Director, and member of their investment committee, Mr Swift manages and oversees their research and risk management processes as well as portfolio construction.

Mr Swift joined Abbey Capital in May 2002 after fifteen years as a trader and manager of trading teams.

Mr Swift is a long-time supporter of Cystinosis Ireland, where he brings his industry professionalism, knowledge and expertise to the executive committee and the role of Chairman.