Presenters

Vicky Ford
MP for Chelmsford

Vicky Ford MP is Chair of the All Party Parliamentary Group (APPG) on Rare, Genetic and Undiagnosed Conditions and member of the Science and Technology Select Committee. She was a Member of the European Parliament from 2009 to 2017.

As well as spearheading digital policy and single market reforms to boost trade, she led the negotiations to end mobile roaming charges and changes to European gun laws after the Paris terrorist attacks.

Vicky was ranked in the top 10 most influential MEPs in Europe by Politico magazine. During her time·as an MEP Vicky led negotiations on banking reform in the aftermath of the financial crisis, and helped establish an €80 billion Horizon fund for science and research. This brought over £500 million into the East of England.

Prof Patrick Chinnery

Patrick Chinnery is Professor of Neurology and Head of the Department of Clinical Neurosciences at the University of Cambridge. He is co-chair of the NIHR Translational BioResource for Common and Rare Diseases.

Patrick qualified in medicine in Newcastle in 1992, trained in clinical neurology and developed a research programme studying the genetic basis of rare mitochondrial disorders, leading to new treatment trials for this group of diseases.

A Wellcome Trust Senior Fellow in clinical science, his laboratory is based in the MRC Mitochondrial Biology Unit on the Cambridge Biomedical Campus where he continues to study the inheritence of rare mitochondrial diseases and their treatment.

Prof Allan Colver

Professor Allan Colver is Emeritus Professor of Community Child Health at Newcastle University. He was appointed Consultant Paediatrician with Special Interest in Community Child Health in 1986. From 1997, he occupied research positions at Newcastle University, retiring in November 2017. His earlier research included studies of child accident prevention, health surveillance of preschool children and screening. Professor Colver's research is currently directed to:

- Participation and Quality of Life of children with cerebral palsy (SPARCLE);
- Transition of young people with complex health needs;
- A randomised controlled trial of different medications for managing drooling.

Dr Patrick Yu-Wai-Man

Dr Patrick Yu-Wai-Man is an academic neuro-ophthalmologist with a major research interest in mitochondrial genetics and inherited eye diseases. His research programme is currently focused on dissecting the disease mechanisms leading to progressive retinal ganglion cell loss in mitochondrial optic neuropathies by using diseased patient tissues and animal models, in addition to therapeutic drug screening and clinical trials.

Through his affiliations in Cambridge, Newcastle and London, Dr Yu-Wai-Man has built a national referral network for the investigation and management of patients with mitochondrial eye diseases. He is actively collaborating with an international network of academic and industrial partners in an effort to fast track the development of effective therapies for this group of patients, including novel gene therapy approaches.

Maria Kokocinska

Maria Kokocinska is the Rare Disease and Nephrology Research Coordinator at Birmingham Children's Hospital and works closely with Dr Larissa Kerecuk, Rare Disease Lead. Maria coordinates the 100,000 Genomes Project at the hospital, which is the second highest recruiting hospital for rare diseases and top recruiting hospital for paediatric cancer. She also coordinates research studies for rare renal conditions.

Maria is undertaking an MSc in Genomic Medicine, and her project is looking at the drivers and barriers to genomics testing among families with rare diseases. Maria is also involved with the newly-launched NIHR Children and Young People MedTech Co-operative. This is dedicated to the development of technology in paediatric specialty areas including rare diseases, where disease burden and mortality risk is high.

Presenters

Janet Turberville-Greenley

Janet Turberville-Greenley is Rare Disease Transition Lead Nurse, supported by Roald Dahl Marvellous Children's Charity, at Birmingham Women's and Children's Hospital.

In this role she is involved in leading the way in designing a service that is at the forefront of care for children with rare or undiagnosed conditions.

Janet has been a nurse for 20 years, after graduating from Birmingham's University of Central England. She is experienced in many areas of practice including cardiac services, paediatric intensive care, management of a general surgery ward and renal, where she was lead nurse for renal transition and transplant workup.

Dr David Adlam

Dr Adlam studied at the University of Oxford and is an interventional cardiologist and researcher based in Leicester. His research interests include rare conditions which lead to heart attacks including Spontaneous Coronary Artery Dissection (SCAD) and Coronary Aneurysm Ectasia (CAE).

In 2013, in collaboration with a group of SCAD survivors in UK and Ireland, he launched and became clinical director of the SCAD UK/EU research initiative and the patient website portal hosted by the NIHR Leicester Biomedical Research Centre.

This initiative has been supported by the British Heart Foundation, the British Cardiovascular Society, the British Cardiovascular Intervention Society and Beat SCAD.

Rebecca Breslin

Rebecca Breslin had a heart attack in 2012 aged just 34, with no known risk factors for heart disease. A rare condition called Spontaneous Coronary Artery Dissection (SCAD) caused her heart attack – but the cause of SCAD is unknown.

Rebecca ('Becks') had many unanswered questions about SCAD and set about finding other SCAD survivors online. After finding 30 survivors in the UK, Becks approached her cardiologist Dr Adlam and together they began a UK SCAD research study in Leicester, which is funded by the NIHR and the British Heart Foundation.

In November 2015, she launched the charity Beat SCAD (www.beatscad.org.uk) with fellow SCAD survivors to raise awareness of SCAD, provide support to people affected by SCAD and raise funds for research.

Prof Paul Gissen

Professor Paul Gissen is Head of the Genetics and Genomic Medicine academic programme at the UCL Great Ormond Street Institute of Child Health (ICH) and Honorary Consultant in Paediatric Metabolic Diseases at Great Ormond Street Hospital.

Prof Gissen studied in Glasgow and completed his Paediatrics training at Manchester, Sheffield and Birmingham Children's hospitals. During his PhD at Birmingham University Prof Gissen studied genetics of rare paediatric liver disorders and became interested in molecular and cellular basis of intracellular trafficking disorders.

Since 2011 his clinical work has been located at the Great Ormond Street Hospital for Children. His clinical research focuses on development of novel therapies for children with inherited metabolic disorders.

Prof John Bradley

Professor John Bradley completed medical training in Nottingham and Cambridge, before undertaking research fellowships at Harvard, Yale and Cambridge Universities.

He is a Consultant Renal Physician at Cambridge University Hospitals, where he is also Director of the NIHR Biomedical Research Centre, Co-Chair for NIHR BioResource for Translational Research and Director of Research for Cambridge University Hospitals.

As an NIHR Senior Investigator, Professor Bradley leads a research programme in vascular biology, with a focus on TNF (tumour-necrosis factors) signaling in the microvasculature.

Professor Bradley was made a Commander of the Order of the British Empire (CBE) in the Queen's birthday honours in 2015 for services to health research.

Consent Panel (Facilitator: Dr Amanda Stranks)

Dr Amanda Stranks

Dr Amanda Stranks is the Patient and Public Involvement/Engagement and Communications Strategy Lead for the NIHR Cambridge Biomedical Research Centre. Prior to this role she held research fellowships at Boston Children's Hospital/ Harvard Medical School and the Jenner Institute at the University of Oxford, and she recently completed an MPhil in Public Health at the University of Cambridge.

During her MPhil studies, Amanda developed an interest in the communication of research and how the public experiences and understands the research undertaken by scientists and clinicians. She hopes to improve the accessibility of the world-class research undertaken on the Cambridge Biomedical Campus and foster connections between its clinicians and scientists an d members of the public who would like to be more involved in research.

Amanda Hunn

Amanda is currently the Joint Head of Policy for the Health Research Authority. She started working in health research in 1989 with Boots Pharmaceuticals as Head of Business Information before moving to the Department of General Practice in 1995 at the University of Sheffield as a Research Fellow.

She later took up the post of Director of Research & Development at the Northern General Hospital in Sheffield. Staying with the NHS, she became the regional manager of the National Research Ethics Service for Yorkshire & the Humber from 2002 to 2006. From 2007 to 2012 she worked as Head of Research and Evaluation for Tribal Health / Capita Health before rejoining the NHS. She also served as a Research Ethics Committee member from 1997 to 2002.

Dr Natasha Kriznik

Dr Natasha Kriznik is a sociologist and qualitative researcher based in the Cambridge Centre for Health Services Research (CCHSR). Prior to joining CCHSR in 2016, Dr Kriznik worked as the Post-Doctoral Research Associate for the St John's College Reading Group on Health Inequalities.

Dr Kriznik has a broad interest in medical sociology, particularly in relation to health inequalities and public health, as well as the processes related to the production of social policy. She is currently working on a project looking to develop the content for a tiered consent process to be used when whole blood donors donate samples for long-term research. This research involves interviewing stakeholders in the research and consent process, and the use of consensus-building techniques to develop and refine the content of the new consent process.

Dr Tom Fowler

Dr Tom Fowler is Genomics England's Director of Public Health and Deputy Chief Scientist, where he also supports the science stream around rare diseases, infectious diseases and cancer. In particular, he has led the rare disease pilot phase of the 100,000 Genomes Project and also coordinates the Project's infectious disease strand.

Following a PhD in Behavioural Genetics in 2003, his career began as a Specialist Registrar in Public Health. He has worked in both commissioning and specialised commissioning as well as roles such as regional epidemiologist and health protection consultant. As a locum Public Health consultant at the Chief Medical Officer's Private Office in 2011-2013, Tom was editor-in-chief of the 'Annual State of the Public's Health' (Vol. I), a comprehensive review of health data for England, and worked on thought leadership around public health, including wellbeing and antimicrobial resistance.

Dr Mary Kasanicki

Dr Mary Kasanicki obtained a degree and PhD in biochemistry before qualifying as a solicitor, specialising in intellectual property law and litigation at a city law firm.

She is currently head of the legal team at the Biomedical Research Centre (BRC) at Cambridge University Hospitals. Her responsibilities include the legal aspects of large projects such as the 100,000 Genomes Project, oncology research collaborations and oversight of the management of BRC intellectual property rights.

Mary advises on consent, data protection law and the use of genomic and other data which has been collected in large research projects.

Consent Panel

Prof Bobbie Farsides

Professor Bobbie Farsides is a bioethicist who studied for her undergraduate and postgraduate degrees at the London School of Economics. She is currently Professor of Clinical and Biomedical Ethics at Brighton and Sussex Medical School. Bobbie is a member of the British Medical Association (BMA) Medical Ethics Committee and the Genomics England Ethics Advisory Committee. She is also a board member of the Human Fertilisation and Embryology Authority (HFEA).

With Anneke Lucassen from Southampton University, Bobbie is about to commence a study funded by the Wellcome Trust looking at ethical preparedness and genomic medicine. Bobbie is interested in ethical issues relating to children and young people, and has chaired the Nuffield Council Working party on Children and Clinical Research and been a member of the NICE End of Life Care for Children guidance writing group.

Ping Coutts

Ping Coutts is currently on secondment as Renal Nurse specialist - where she is involved in renal research, including in rare kidney conditions - at the Queen Elizabeth Hospital, King's Lynn.

Ping, who is from China, has worked in the UK since 2004 in renal medicine, mainly in haemodialysis care. In 2014 she started working in renal research, covering renal anaemia and bone disorders, renal transplant, vasculitis, myeloma and lupus studies. In 2016 she relocated to North Cambridgeshire and started working in cancer trials; her permanent post at the Queen Elizabeth is as a specialist cancer research nurse.

Her research duties include all stages of the clinical trial process, including registration, trial set-up, randomisation, frontline patient recruitment, and follow-up clinics.

Dr Lydia Drumright

Dr Lydia Drumright is the University Lecturer in Clinical Informatics in the Department of Medicine at the University of Cambridge, where she directs the Cambridge Clinical Informatics Centre. She is also the Chair of the Central Cambridge Research Ethics Committee and Co-Chair of the Cambridge Infectious Diseases Interdisciplinary Research Centre.

Dr Drumright's work focuses on observational and experimental health research in humans to better understand health risks and how to prevent these. With a very translational focus, she has conducted research in a multitude of settings, across many different countries, on a variety of health problems.

Dr Drumright has a particularly strong interest in human diversity, health equity, public and patient engagement and the ethics of science, medicine and the involvement of human participants in research.

Trainer: Data and Research

Dr Neil Walker

Dr Neil Walker is Clinical Bioinformatics and Statistical Genomics Manager at the NIHR National BioResource for Translational Research – which must be a contender for longest job title ever invented to describe the IT lead on a big research project! He is also IT lead for the East of England Genomic Medicines Centre, which is rolling out the 100,000 Genomes Project for both rare diseases and cancer in East Anglia.

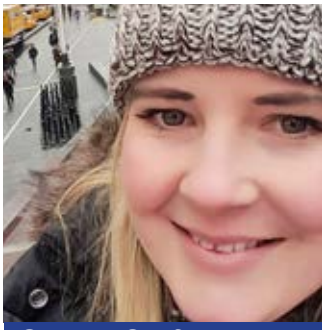
Neil studied social science at Cambridge, and has spent almost 30 years organising data for medical research. Projects have ranged from the small-but-startlingly-complex – a 40 person clinical trial; to the large-and-not-that-simple – checking facts and figures for the national breast screening program. He has a deep interest in making data available for re-use – to the right researchers, with the right questions. He is Deputy Chair of the METADAC – a data access committee that reviews applications to use data from various big long-running studies - and was a signatory of the "Toronto Accord".

Trainers: Social Media

Emma Damian-Grint

Emma Damian-Grint spent 5 years working for the rare disease charity the Tuberous Sclerosis Association. Responsible for growing their fundraising and audience, she successfully launched Facebook, Twitter and YouTube for the charity. Focusing on bringing new ways of interacting with the patient community, clinicians and researchers on a small budget, she was able to grow the charity's reach and voluntary income.

Now working for Genetic Alliance UK, she is responsible for 3 individual brands with different audiences across a range of online platforms, and growing the national awareness campaign Rare Disease Day. She has a passion for harnessing the power of online tools and social media for high impact and low cost to benefit patients, and their charities, through campaigning, fundraising and to raise awareness of their work and the condition they support.



Gemma Seyfang

Gemma Seyfang is a young mum of two boys who runs the family business and also has Pompe disease, a rare condition that affects the body's muscles, with patients experiencing progressive muscle weakness, including the muscles that control breathing.

After suffering symptoms throughout her 20s Gemma was diagnosed in 2016 and began a double-blinded clinical trial last year; there is currently only one drug available in the world to treat Pompe disease.

Gemma loves using social media to raise awareness of this disease and says: "The people and groups I've met through social media have become like a family to me." Gemma looks forward to sharing her stories and ideas in the social media workshop.

Trainers: Genomics

Gemma Chandratillake

Gemma Chandratillake is trained as a molecular geneticist and genetic counsellor. She has a long-standing interest in rare genetic diseases, having used fruit flies and worms to model alpha-1-antitrypsin deficiency and Niemann-Pick type C disease in the lab.

Gemma previously worked at the Stanford University spin-out company Personalis to develop a clinical genomic sequencing test for the diagnosis of individuals with rare disease. Since moving back to the UK, Gemma has focused on healthcare professional education in genetics. She is the Education & Training Lead for the East of England Genomic Medicine Centre (NHS) and coordinates the Masters course in Genomic Medicine at the University of Cambridge. Gemma is a Trustee of the Cambridge Rare Disease Network and works with stakeholders to find creative ways to mainstream genomics within the NHS for the benefit of patients with rare disease.

Dr Karola Rehnström

Dr Karola Rehnström has worked on several large-scale genomics projects, including the 100,000 Genomes Project and the UK10K. She is currently the Programme Lead for the East of England Genomic Medicine Centre, delivering the 100,000 Genomes Project.

The 100,000 Genomes Project aims to deliver a better genomics service for NHS patients and the East of England Genomics Medicine Centre is delivering this service across its partner hospitals in Cambridge, Derby, Leicester, Norwich, Nottingham and Papworth for patients with rare diseases and cancer in the East of England.

Previously, Karola has worked with identifying genetic risk factors in families with autism spectrum disorders and learning disabilities.

Trainers: Working with Industry

Katie Kliss

Katie Cliss is Fundraising Officer at Findacure, the UK charity that builds the rare diseases community to drive research and develop treatments.

Katie is responsible for maximising Findacure's revenue through a range of fundraising opportunities, including charitable trusts, corporate grants and sponsorship, and fundraising events.

She works on developing Findacure's community fundraising efforts as well as building strong relationships with the charity's supporters and varied rare disease stakeholders.

Libbie Read

Libbie Read is the Projects and Communications Officer at Findacure, a UK-based rare diseases charity. Libbie started at Findacure in 2015 as the Fundraising and Communications Officer, where she was responsible for maximising the charity's revenue from a range of sources and their outgoing communications.

In September 2017, she moved to the Projects and Communications Officer role, in which her responsibilities are to manage the charity's peer mentoring programme, training webinars, online resources portal and outgoing communications, including their website, social media, monthly newsletter and press releases. Libbie is passionate about raising awareness of rare diseases and helping patient groups to solve their most pressing challenges. She has previously mentored patient groups as part of Findacure's peer mentoring programme.

Tanya Collin-Histed

Tanya Collin-Histed became involved in the Gaucher world in 1996 when her daughter Maddie was diagnosed with Type 3 Gaucher disease. A year later she became a volunteer for the Gauchers Association and started to support patients and their families with Type II and III Gaucher Disease through family conferences, information booklets and providing friendship and emotional support.

In 2001 Tanya received the Alan Gordon Memorial Award in recognition for her work with Neuronopathic Gaucher disease and her support to families. She is Chief Executive of the Gaucher Association UK and Chief Executive Officer of the European Gaucher Alliance (EGA), where she supports patients globally to access treatment through compassionate programmes in partnership with pharmaceutical companies and supporting evolving patients' groups in countries across the globe.

Information for Consent Panel Session

THE CONSENT PANEL (ON STAGE)

Dr Mary Kasanicki

Amanda Hunn

Ping Coutts

Dr Lydia Drumright

Dr Natasha Kriznik

Dr Tom Fowler

Prof Bobbie Farsides

Dr Amanda Stranks
(FACILITATOR)