Industry, Academia & Rare Diseases
‘Elevator Pitch’ Workshop

8 July 2013

The Wellcome Collection,
183 Euston Road, NW1 2BE
Dear Colleagues,

On behalf of the UCL Rare Diseases Steering Committee and the UCL SLMS Knowledge Transfer and Enterprise Board, it is my pleasure to welcome you to the Industry, Academia and Rare Disease ‘Elevator Pitch’ Workshop.

Rare disease presents many unique challenges for the pharmaceutical industry however the field offers many possibilities for the development of novel diagnostics, drugs and treatments. We hope this workshop will catalyse further industry/academia collaborations to exploit these opportunities.

The EU definition of a rare disease is one that affects fewer than five in 10,000 of the general population. In the UK, it is estimated that 6% (approximately 3.5 million) of the population will be affected by a rare disease at some point in their lives. UCL, The National Institute for Health Research Biomedical Research Centres and our Academic Health Science Centre, UCLPartners (UCLP) are committed to developing our capacity to tackle rare disease. We serve a patient population of over six million across the North Thames region, approximately 0.5 million of whom will have a rare disease. Our research is very diverse with over 100 clinical academic research groups investigating more than 350 rare diseases. Of the 70 NHS nationally commissioned specialist services in England, over 30 are based at UCLP.

Today’s workshop is highly interactive and includes a range of presentations from speakers representing both clinical academia and industry. The aim is to provide the audience with an understanding of the unique challenges faced by industry during the development of orphan products and the key criteria to make a successful partnership. We are also hosting an ‘elevator pitch session’ which will offer a platform for knowledge exchange between our researchers and our industry guests.

We are grateful to UCL Enterprise for their sponsorship of this event and extend special thanks to our speakers and chairs.

We hope you find our workshop interesting and informative. Please take the opportunity to interact with other delegates throughout the day and during the Networking Reception which will be held in the Williams Lounge.

Professor Philip Beales
Chair, UCL Rare Disease Steering Committee

UCL Rare Disease Steering Committee
The UCL Rare Disease Steering Committee comprises key members of the UCL academic and clinical community and aims to develop and implement strategies for rare diseases, linking resources and supporting the further advancement of clinical and academic research.

Professor Phil Beales (Chair)
Dr Ruth Jamieson (Experimental Medicine Domain Co-ordinator)
Cassie Harley (Communication and Events Officer)
Dr Detlef Bockenhauer
Mr Jose Tomas Bras
Dr Gerard Conway
Professor Ingemar Cox
Dr Sarah Creighton
Professor Perry Elliott
Dr Daniel Gale
Dr Julian Gilmore
Dr Paul Gissen
Dr Paola Giunti
Professor Mike Hanna
Professor Simon Heales
Professor Henry Holden
Dr Robin Lachmann
Dr Helen Lachmann
Dr Nick Lench
Dr Juan Pedro Martinez-Barbera
Dr Nadia Miceli
Dr Hannah Mitchison
Dr Sara Mole
Professor Gudrun Moore
Professor Francesco Muntoni
Dr Ros Quinlivan
Dr Gill Rumsby
Dr Rachael Scallion
Professor Sanjay Sisodiya
Dr Andre Strydom
Professor Sarah Tabrizi
Dr Aoife Waters
Professor Lucy Wedderburn
Professor Nick Wood.
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<td>Welcome</td>
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<td>13.40 – 14.05</td>
<td>Cryopyrin Associated Periodic Syndrome – a tale of a rare orphan disease leading to a new NHS treatment service</td>
<td>Professor Philip Hawkins National Amyloidosis Centre, Royal Free Hospital</td>
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<td>GSK and Rare Diseases</td>
<td>Dr Carlo Russo GSK Interim Head, Rare Disease R&amp;D, GSK</td>
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<td>Tracking an innovative discovery from Academia to a startup company and finally to Alexion: A case study</td>
<td>Dr Jeremy Springhorn Alexion Pharmaceuticals Inc.</td>
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<td>Successful industry partnerships: an academic’s perspective (Lysosome Storage Diseases)</td>
<td>Dr Derralyn Hughes Royal Free &amp; University College Medical School</td>
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<td>16.25 – 16.35</td>
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<td>16.35 – 18.30</td>
<td>Elevator Pitches (UCL investigators) 3 min max. per pitch</td>
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UCL Enterprise
Enterprise is important to all universities, but resonates particularly with UCL. From our inception we were created as an enterprising institution, with a bold ambition to create a University dedicated to the greatest good for the greatest number. This principle has underpinned the evolution of modern-day UCL, a confident and enthusiastic community of enterprising researchers, educators, and scholars, working together for the immediate, medium and long-term benefit of society. UCL Enterprise provides UCL’s structures for engaging with business for commercial and societal benefit. It includes three units: UCL Advances, UCL Business and UCL Consultants. Together, they provide access to the capabilities and resources of the UCL community to help businesses start, grow and develop.

Information provided by UCL Enterprise - further information can be found at:

www.ucl.ac.uk/enterprise/files/Enterprise_Review_1.pdf

UCL Business PLC
UCL Business PLC (UCLB) is a leading technology transfer company that supports and commercialises research and innovations arising from UCL, one of the UK’s top research-led universities. UCLB has a successful track record and a strong reputation for identifying and protecting promising new technologies and innovations from UCL academics. UCLB has a strong track record in commercialising medical technologies and provides technology transfer services to UCL’s associated hospitals; University College London Hospitals, Moorfields Eye Hospital, Great Ormond Street Hospital for Children and the Royal Free London Hospital. It invests directly in development projects to maximise the potential of the research and manages the commercialisation process of technologies from laboratory to market.

UCLB supports UCL’s Grand Challenges of increasing UCL’s positive impact on and contribution to Global Health, Sustainable Cities, Intercultural Interaction and Human Wellbeing.

Information provided by UCLB - for more information please visit their website:

www.uclb.com
UCL

UCL was established in 1826 to open up education in England for the first time to students of any race, class or religion. UCL was also the first university to welcome female students on equal terms with men. Academic excellence and conducting research that addresses real-world problems inform our ethos to this day.

UCL is among the world’s top universities, as reflected by performance in a range of international rankings and tables. According to the Thomson Scientific Citation Index, UCL is the second most highly cited European university and the 15th most highly cited in the world.

The University has one of Europe’s largest and most research active groupings of biomedical scientists, which includes four world class postgraduate institutes - the Institute of Ophthalmology, the Institute of Neurology, the Institute of Child Health and the Eastman Dental Institute. UCL, together with the Medical Research Council, Cancer Research UK and the Wellcome Trust, has founded the Francis Crick Institute to bring together scientists from all disciplines.

With dedication and a creative approach, UCL research is helping to address the world’s most urgent problems.

University College London Hospitals (UCLH)

UCLH is committed to delivering top-quality patient care, excellent education and world class research.

Situated in the heart of London, UCLH is one of the largest NHS trusts in the United Kingdom and provides first-class acute and specialist services in seven hospitals.

Together with UCL, Moorfields Eye Hospital, The Royal Free Hampstead, Barts and The London NHS Trust, Queen Mary University of London and Great Ormond Street Hospital they have created Europe’s largest and strongest Academic Health Science Partnership. UCLP pools resources and expertise to produce outstanding research and delivers the benefits more rapidly to patients.

UCLH is a world-class leader in clinical research and has one of the largest portfolios of studies. At any one time over 1,000 research projects can be taking place across the hospitals network.

Research at UCLH is delivered through:

- The NIHR UCLH Biomedical Research Centre (BRC) which focuses on experimental medicine
- Our Joint Research Office which provides research management and support with biostatistics, governance and professional development
- The UCLH/UCL NIHR Clinical Research Facility in the Elizabeth Garrett Anderson wing of UCLH which provides a dedicated clinical environment and specialist trained staff for research.
- The UCL Clinical Trials Unit which is a specialist unit with a specific remit to design, conduct, analyse and publish clinical trials and other well-designed studies.
NIHR University College London Hospitals Biomedical Research Centre

The NIHR University College London Hospitals BRC is the result of an outstanding partnership between UCLH NHS Foundation Trust and UCL.

UCLH is one of the largest NHS trusts in the United Kingdom and UCL is one of the leading biomedical research universities. By working together we have become leaders in translating fundamental biomedical research into clinical research that benefits patients.

The NIHR awarded us BRC status in 2007. With BRC status, we have been able to build on our expertise in areas such as cancer, cardiovascular disease and neurosciences.

The BRC supports research by investing in staff posts, equipment, facilities and training.

The BRC takes innovations in basic science and helps to turn them into treatments and therapies which have a direct effect on patients and often saves lives.

In particular the BRC supports experimental medicine research which tends to be ‘first in man’ studies such as research into new therapies and devices or the mechanisms of disease. The BRC will be investing over £100m in experimental medicine over the next five years.

We have organised our research into four programmes:

- Cancer
- Cardiometabolic
- Infection, immunity and inflammation
- Neuroscience

Information provided by the NIHR University College London Hospitals Biomedical Research Centre - for more information please visit their website:

www.uclhospitals.brc.nihr.ac.uk/
NIHR Biomedical Research Centre at UCL Institute of Ophthalmology and Moorfields Eye Hospital
The BRC, established in 2007, has a strategy that is based around six themes defined by major common eye problems or disease processes. We target disease areas that are both clinically significant and identified as important to patients and their relatives. We continue to prioritise areas where there is an outstanding existing track record of translational achievement and expertise at an international level. We have built support and infrastructure around these themes, growing strategic partnerships and collaborations and building capacity to support and consolidate all the links of the translational bridge. As a result, new treatment techniques and practices that improve patient health are brought through as quickly as possible, benefiting patients as well as the NHS, universities and the UK as a whole.

Regenerative medicine and pharmaceutics
Scarring, arising as a natural reaction to surgery performed to improve an underlying ophthalmic problem such as glaucoma, can cause significant impairment of vision. Our site has one of the strongest track records in the world in the fields of preventing ocular scarring, promoting cell regeneration and developing novel medicines. The overarching aim of this research theme is to accelerate the availability and range of innovative cellular and molecular therapies which meet many unmet needs in ophthalmology and in the human body.

Gene therapy
The aim is to develop gene therapy as a treatment for a range of eye diseases, including rare (currently untreatable) forms of inherited diseases as well as common eye disorders (age-related macular degeneration and diabetic retinopathy) that cause haemorrhaging and fluid leakage and irreversible damage to the overlying retina. Gene therapy offers the prospect of local treatment with reduced risk of systemic side effects.

Visual assessment and imaging
This theme provides enabling technologies to all other themes to help with early diagnosis of disease; assessing rate of disease progression; targeting treatments to patients; assessing the effectiveness of treatments and measuring the impact of low vision on everyday tasks.

Genotyping, phenotyping and informatics
This theme aims to further develop a research infrastructure to maximize involvement of our patients in clinical trials of new therapies. The focus will be on genetic variants that predispose people to common eye disorders such as glaucoma and myopia so that we can identify gene targets for new therapies.

New technologies and devices
The aim of this theme is to transform diagnosis and therapy for eye disease by building next-generation devices through the exploitation of recent and ongoing advances in optics, laser and other technologies.

Inflammation and immunotherapy
Inflammation is increasingly understood to underpin tissue damage in the eye. Uveitis, ocular surfaces diseases and corneal graft rejection are obvious areas where inflammation causes problems but it is also central to the progression of age-related macular degeneration and diabetic retinopathy.

Information provided by the NIHR Biomedical Research Centre at UCL Institute of Ophthalmology and Moorfields Eye Hospital - for more information please visit their website:

www.brcophthalmology.org/
Organisational Overview

**NIHR Biomedical Research Centre at Great Ormond Street Hospital for Children Hospital NHS Foundation Trust and UCL**

This BRC is a partnership between Great Ormond Street Hospital NHS Foundation Trust (GOSH) and UCL Institute of Child Health (ICH) and is recognised globally as a leading centre in paediatric experimental medicine research.

The NIHR first awarded us BRC status in 2007 and this award was renewed in 2012, which will allow GOSH UCL BRC to continue to lead on the discovery of treatments for childhood diseases.

GOSH is the largest recipient of nationally commissioned NHS funding in the country, and so our doctors see more children with rare diseases than anywhere else in the world. Our BRC will therefore focus on rare diseases and with our core clinical and research resource, we will deliver patient benefit nationally.

BRC funding will be used to support experimental medicine research and the pull through of basic scientific discoveries into ‘first in man’ studies. The BRC will be investing over £35m into its research programmes over this five year funding period.

We have organised our research into four themes:

**Molecular basis of childhood diseases**

GOSH UCL BRC aims to bring together clinicians and researchers from all disciplines to work toward understanding the molecular causes of childhood diseases, including rapid gene identification in uncharacterised genetic diseases.

**Diagnostics and imaging in childhood diseases**

GOSH UCL BRC will fund research to improve diagnostics for rare diseases and the development of new biomarkers and novel imaging strategies to monitor disease progression and response to experimental therapies.

**Gene, stem and cellular therapies**

We aim to further develop somatic gene and cellular therapies for immunodeficiency disease and a wide range of rare inherited and acquired disorders.

**Novel therapies for childhood diseases**

Through access to some of the largest paediatric cohorts in the UK, we will undertake novel ‘first in man’ studies to develop and deliver novel experimental therapeutic interventions.

Information provided by the NIHR GOSH UCL BRC - for more information please visit their website:

UCLPartners - Delivering partnership solutions to major challenges in health and health care

UCLPartners is one of five accredited academic health science systems in the UK. Our purpose is to translate cutting edge research and innovation into measurable health gain for patients and populations – in London, across the UK, and globally.

To achieve this, we develop integrated, value-for-money, outcome-driven solutions to the most pressing health care challenges. These solutions include faster drug discovery and development; innovative technologies; new approaches to clinician education and professional development; and models of care that drive both quality and value. Our solutions are:

- Patient-led, organising care around patients’ needs and preferences
- Population-focused, taking a system-wide view to drive improved health outcomes at speed and scale
- Cross-boundary, spanning primary, secondary and tertiary care, and connecting different phases of academic research

We take a partnership approach to developing solutions: we work with patient groups, universities, NHS Trusts, community care organisations, commissioners, government and industry. Our role is not to mandate change or to ‘own’ solutions ourselves. Rather, we work with our partners to co-create, test and implement solutions, ultimately embedding these solutions in our partners’ everyday working.

We harness academia (across biomedicine, the humanities and other disciplines) to solve the broader health problems of populations – for example, cultural barriers to cancer screening in certain ethnic communities. We facilitate dialogue between academics, clinicians and populations to ensure that research is focused on impact.

Information provided by UCLPartners - further information can be found at:

Overview

Pamphlet
Professor Philip Beales is Professor of Medical Genetics at UCL and a Wellcome Trust Senior Fellow in Clinical Science. He is also currently Director of the Centre for Translational Genomics (GOSGENE) and has been Head of the Cilia Disorders Laboratory at UCL Institute of Child Health for the last 10 years.

Professor Beales is best known for his clinical and genetic research (17 years) into rare diseases, especially the ciliopathies, leading research culminating in novel gene discoveries for Bardet-Biedl syndrome, Jeune Asphyxiating Thoracic Dystrophy, Craniocutaneous dysplasias, Acrocallosal Syndrome and several other disorders. He was the first to attribute the Bardet-Biedl syndrome phenotype to dysfunctional primary cilia. Professor Beales continues to pursue his interests in early onset obesity, retinal and renal disease and more recently in translational science and therapeutics for ciliopathies. Professor Beales is a Consultant in Clinical Genetics at both GOSH and Guy’s Hospital, London and National Lead for the Department of Health specialist commissioned Bardet-Biedl syndrome clinical and diagnostics service. Professor Beales is currently a member of the Wellcome Trust Rare Disease Strategy Group. He was elected Fellow of the Academy of Medical Sciences in 2011.

Professor Lucy Wedderburn is a Professor and Clinician Scientist in Paediatric Rheumatology based at UCL UK and GOSH. She runs a busy immunology research group focusing on the Immunology of childhood arthritis and myositis, exploring both basic and translational questions. In addition, she is Chief Investigator of the UK Juvenile Dermatomyositis (JDM) Cohort and Biomarker study (JDM is a rare and serious childhood disease affecting approximately 2 million children under 16 per year). She is also Director of the Arthritis Research UK Centre for Adolescent Rheumatology at UCL, ULCH and GOSH.

Professor Perry Elliott is Clinical Director of the Inherited Cardiovascular Disease Unit at UCL. He studied Medicine at St. Thomas’s Hospital Medical School, London. After qualifying in 1987 he trained in General Medicine, gaining membership of the Royal College of Physicians in 1991 and completed his general cardiology training at St. George’s Hospital Medical School, London. He was first appointed as Senior Lecturer at St. George’s Hospital in 1999 and then at UCL in 2003. He was promoted to Reader in Inherited Cardiac Disease in 2005 and became a full Professor in 2012.

Over the past 20 years he has established an international reputation in the field of heart muscle disease, authoring more than 230 peer reviewed papers on the subject. Professor Elliott was elected as a fellow of the European Society of Cardiology (ESC) in 2005, is past Chairman of the ESC Working Group on Myocardial and Pericardial Diseases (2010-12), and chairs the ESC Guideline Task Force on Hypertrophic Cardiomyopathy and the Executive Committee for the European Outcomes Research Programme (EORP) registry on cardiomyopathies. In the UK he is Vice-President of the Cardiomyopathy Association and a member of The National Confidential Enquiry into Patient Outcome and Death (NCEPOD) Surgery in Children study and The Cardiac Devices National Action Group (CDNAG). In 2009, he was appointed as Deputy Editor of The Heart Journal.
**Professor Philip Hawkins** trained in medicine at St George’s Hospital Medical School, London. At Hammersmith Hospital he undertook specialist training and obtained his PhD in studies on amyloidosis in 1990. He was appointed Head of the NHS National Amyloidosis Centre at Royal Free Hospital in 1999 and is Head of the UCL Centre for Amyloidosis and Acute Phase Proteins. His clinical and research interests are focussed on diagnosis, pathogenesis, monitoring and treatment of amyloidosis and inherited autoinflammatory diseases with an emphasis on translational, early phase and novel treatment approaches.

**Dr Carlo Russo** is Senior Vice President, Alternative Development Program, Development Head, Alternative Development and Discovery and Interim Head, Rare Disease Development for GlaxoSmithKline. He is also a founding member of the National Institutes of Health, Geriatrics and Rehabilitative Medicine Study Section.

Dr Russo has a range of academic, scientific, biotech, and pharmaceutical industry experience. He earned his medical degree from the University of Genoa Medical School and is board certified in haematology. After Clinical training in oncology, he pursued research in molecular immunology as a fellow at the Scripps Research Institute in La Jolla (California) and subsequently held a series of academic appointments at the College of Physicians and Surgeons, Columbia University, and at Cornell University Medical and Graduate Schools, where he still serves as an Adjunct Associate Professor. He has published extensively on functional and molecular characterisation of the major histocompatibility complex, cell-mediated immune response, tumour immunology, and the immunobiology of aging.

Prior to joining GSK, Dr Russo was the President and Chief Executive Officer at VaxInnate Corporation, a start-up Biotechnology Company which develops and manufactures vaccines and vaccine technologies. Dr Russo raised $30 million Series B Venture Capital Funds to finance the development of VaxInnate’s first project; an M2e universal influenza vaccine against pandemic flu. In 2011 VaxInnate received $196 million from The Defense Advanced Research Projects Agency (DARPA) to develop the influenza vaccine. The vaccine Phase I studies are ongoing. He previously served as Executive Director and Head in the Department of Global Strategic Regulatory Development, Merck Research Laboratories, where he oversaw and ratified development of global regulatory strategy for all vaccines and biologics. During his time at Merck, Dr Russo was responsible for all regulatory filings in the US and worldwide for new vaccines, including those for HIV, the recently approved human papilloma virus, Gardasil®, and rotavirus vaccine, Rotateq®.

Dr Russo was recruited at GSK to develop an Alternative Development Program focusing on the development of albiglutide, a GLP-1 mimetic drug for Type 2 Diabetes.

In addition, he was a founding member of GSK Biopharm R&D Unit. In that function, Dr Russo was responsible for the development of GSK Biopharm products in all therapeutic areas and for 6 Phase III Biopharmaceutical Programs in therapeutic areas ranging from T1DM to CLL, to MS and RA.

**Dr Jeremy Springhorn** is Vice President of Corporate Development at Alexion and is responsible for Corporate Development, Academic Licensing, Business Development and Competitive Intelligence.

Dr Springhorn was one of the original scientists at Alexion and played an integral role in the engineering and development of Alexion's antibody therapeutics as well as being one of the original inventors of Soliris. Dr Springhorn started his career in Research and transitioned to Business Development leveraging much
of his drug development experience into the review of opportunities for ultra-orphan diseases. Dr Springhorn has led many of the recent and current deals for Alexion and has served in other key positions in the company including Vice President Business Development and Corporate Strategy; Senior Director Protein Therapeutics and Head of Collaborative Research; and Director of Small Molecule Drug Discovery. He has also served as a scientific advisor to Arradial, Inc. and currently serves as a Director of the Greater New Haven Chapter of the Juvenile Diabetes Research Foundation.

Prior to 1992, Dr Springhorn was an Instructor of Medicine at Harvard Medical School (1991-1992) and a Postdoctoral Research Fellow in the Cardiovascular Division of the Department of Medicine at the Brigham and Women’s Hospital. He is a member of the Licensing Executives Society and Biotechnology Industry Organization.

Dr Springhorn received a Doctorate degree in Biochemistry and Molecular Biology from the Louisiana State University Medical Center and a Bachelor of Arts degree from Colby College.

Dr Derralyn Hughes is a Senior Lecturer in Haematology at the UCL, and has clinical responsibilities in the area of haematology and lysosomal storage disorders. She directs the research programme in the lysosomal storage disorders unit research laboratory.

Dr Hughes studied medicine at Oxford University and joined the research group in the Sir William Dunn School of Pathology as an MRC training fellow, writing a doctoral thesis in the area of macrophage biology. Her interest in the role of the macrophage in inflammatory and developmental processes has endured and now, as a haematologist, she has focused her laboratory research interests towards the role of inflammatory cell interactions in the pathophysiology of the lysosomal storage disorders Gaucher and Fabry disease. Major laboratory projects are currently aimed at understanding the pathophysiology underlying Gaucher-related bone pathology, increased incidence of malignancy in Gaucher disease and phenotypic variation in Anderson-Fabry disease.

Dr Hughes has a clinical research commitment and is actively involved in a number of trials examining the efficacy of enzyme replacement therapy and other new therapies in the treatment Gaucher, Fabry, Pompe and MPS disorders. A particular interest relates to the clinical and biological effects of Anderson-Fabry disease in women. Recent publications included the effects of enzyme replacement therapy on the cardiomyopathy of Fabry disease and females with Fabry Disease, the development of an age-adjusted and prognostic scoring systems for Anderson Fabry disease, the use of substrate reduction in Gaucher disease, recommendations for the management of haematological complications of Gaucher disease, and the effect of delayed diagnosis in myeloma.

Dr Kevin Lee is Chief Scientific Officer and Head of the Rare Disease Research Unit at Pfizer. Prior to joining Pfizer, Kevin conceptualised and led epigenetics research at GSK and was responsible for the creation of the EpiNova Discovery Performance Unit (DPU) as well as leading the formation of multiple strategic commercial and academic partnerships for the company. Kevin studied Pharmaceutical Sciences at Nottingham University followed by a PhD in Pharmacology at Cambridge. He undertook postdoctoral training as a Wellcome Trust International Prize Fellow before joining the Parke Davis Research Unit in Cambridge.

Prior to joining GSK, Kevin lectured at Warwick Medical School, founded Cambridge Biotechnology (acquired by Biovitrum) and Neurosolutions (now Neurodiscovery - ASX code: NDL). Kevin is an author on over 100 peer reviewed scientific publications, has an MBA from Warwick Business School and has been awarded an honorary Chair in Molecular Pharmacology from the University of Warwick.
**Dr Rob Cooke** is Head of the Biomolecular Structure Department at Heptares Therapeutics, where he is leading research in structural biology and biophysics, computational chemistry and informatics, and protein expression. He is also responsible for the management of alliances with Pharma partners. Prior to this, Rob was at Glaxo, then GlaxoWellcome, then GlaxoSmithKline. Starting as a Structural Biologist, Rob eventually led Departments covering research in several disciplines including structural biology, computational chemistry and analytical sciences. Rob also initiated the proceedings which led to the Structural Genomics Consortium, and has been an advisor to several academic institutes, as well as a member of grant funding committees.

Rob received his BSc and PhD in Inorganic Chemistry from the University of Sydney, and was a post-doctoral researcher in the Department of Biochemistry at the University of Oxford.

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*See page 11 for more information.*

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*See page 10 for more information.*

**Dr Peter Sazani**

Executive Director, Medical Affairs, Sarepta Therapeutics

Sarepta Therapeutics is developing RNA therapeutics for the treatment of Duchenne muscular dystrophy (DMD).
1) Dr Daniel Gale - *A new strategy for complement inhibition in kidney disease*

Complement is a network of proteins in the blood that is crucial to innate defence against infection. Activation of complement in the kidney, usually by antibodies, is central to the pathogenesis of many common kidney diseases, including IgA nephropathy – for which there is no effective treatment and which results in many thousands of people worldwide needing expensive and limited dialysis and transplantation therapies. We recently identified a monogenic disease, CFHR5 nephropathy, caused by mutation of the circulating complement protein CFHR5 leading to activation of complement (independently of antibodies) in the kidney. This results in disease otherwise almost indistinguishable from IgA nephropathy. UCL owns the intellectual property on the use of all or part of this protein to treat kidney disease. CFHR5 dimerizes in the circulation via interaction of the N-terminal 2 domains of the protein. The dimers compete with the circulating complement regulator CFH to deregulate complement, especially at surfaces. The mutation in CFHR5 nephropathy results in duplication of these 2 domains, potentially leading to production of higher order complexes. This pitch is to develop a drug to block this interaction, either a small molecule that binds to the dimerization interface or recombinant protein (currently under production) comprising only the N-terminal 2 domains. This may provide a novel and effective therapeutic strategy to protect the kidney from damage by complement that would be applicable across a wide range of common kidney diseases, including IgA nephropathy, lupus nephritis and kidney transplant rejection, in which complement is activated in the kidney.

2) Dr Sara Mole - *Using yeast to identify new drug targets and therapeutic strategies for disease*

Neurodegeneration is one of the most significant challenges facing healthcare provision and drug development. Very few effective therapies are available for dementias of any form, and cures for neurodegenerative disease are still a distant prospect. Current therapeutic strategies aimed at specific pathological features of individual conditions have not provided the clinical outcomes hoped for. Consequently, the past few years have seen a shift in focus from specific features of individual conditions to features common to neurodegeneration as a whole. Lessons learned from rare diseases can be used to inform therapeutic development in more common conditions that share pathological features. Monogenic inherited neurodegenerative diseases represent ideal candidates for such an approach, as they can be easily modelled in experimental systems. Many of the genes mutated in these diseases are conserved across eukaryotic species. This lends them to investigation in simple model systems such as yeast species which are particularly amenable to image or fitness-based high throughput screens and provide the opportunity for systems level analysis. The yeast model has already proved invaluable in the study of multiple neurodegenerative conditions. The aim of our work is to take full advantage of the strengths of the yeast model system to perform unbiased genetic screens to identify the genetic interactions of inherited neurodegeneration. This will highlight potential therapeutic targets and key neuroprotective factors. To complement this first aim, we also intend to use yeast to identify small molecule therapies for these conditions, in addition to small molecules that influence mutation specific phenotypes in multiple strains.

3) Dr Adam Giangreco - *Small molecule therapy to reverse thymus involution and restore immune function in elderly and immunocompromised patients*

4) Dr Jasper de Boer - *Drug repurposing and leukaemia oncogenes*

We have demonstrated that certain genes have a central role in leukaemia development. Inactivation of these oncogenes led to full remission, even after accumulation of numerous additional genetic changes, indicating that they are the essential drivers of the leukaemia. We are currently running a drug repurposing screening that measures the activity of these oncogenes and have found some early leads that inactivate these genes. We are looking for extra help and funding to scale up these screens and the downstream validation process.

5) Dr Laurent Bozec - *A UK-wide academia, industry and patient network and Biobank for rare dental anomalies*

Tooth development involves a series of complex, cellular and molecular interactions between the oral epithelium and underlying connective tissue. A dental anomaly occurs when there is a disruption in these processes, and can be localised to one tooth or generalised to affect all teeth. These anomalies can represent trigger signs for undiagnosed underlying conditions such as rare diseases. These include inherited diseases of the enamel (Amelogenesis Imperfecta) and dentine (Dentinogenesis Imperfecta).
Elevator Pitch Abstracts

Imperfecta). Our vision is to create a UK-wide network comprising clinicians, patients groups and researchers (both academic and industrial) to share knowledge, expertise, resources in order to further the research of dental anomalies for patients' benefits. This network would aim to facilitate referrals from dental practitioners to chartered experts as well as providing a common platform for scientists to further the research into these anomalies. This network will have four mains goals:

- Raise awareness of the conditions to the general public and educate dentist practitioners about the conditions
- Provide excellence in both fundamental and translational research for patients' benefits
- Improve the care of the patients by facilitating referral
- Improve the access to study samples for scientists across the UK in order to further the knowledge and understanding of these anomalies

UCL Eastman Dental Institute would act as a centre of excellence, using our pre-existing local expertise (both clinical and research)

- UCL EDI would administer a web-based support network for both patients and practitioners and would also act as a repository of samples (Biobank) accessible to our partners.

6) Dr Virginia Arechavala-Gomeza - Streamlining drug candidate selection for Duchenne Muscular dystrophy
For the last 8 years I have been part of an effort to develop new drugs for Duchenne muscular dystrophy, a fatal devastating disorder affecting boys. We are trying to restore a missing protein, but we lack the methods to measure this particular protein accurately when selecting drug candidates for future trials, so we most commonly use some surrogates of their effect. Despite all this, we have been quite successful and some of our candidates are at late stage clinical trials. The development of these new drugs has gained such momentum that we cannot afford to use the traditional methods for selection anymore and this is why I aim to develop a new method that can accurately measure this protein in a more streamlined method.

7) Dr Petra Disterer - APO-SKIP, a splice-switching oligonucleotide therapeutic for familial hypercholesterolemia
We are an expert team all with a background in oligonucleotide therapeutics. The team consists of Dr. Bernard Khoo, a clinical endocrinologist, Prof. James S. Owen, a Lipidologist, Dr. Paul Simons, a mouse model specialist and I, Dr. Petra Disterer, the technical specialist. Our product is a splice-switching oligonucleotide therapeutic for familial hypercholesterolemia, a genetic disease which is characterized by extremely high levels of plasma LDL and premature cardiovascular disease. Current drugs to reduce LDL, such as statins, are inadequate and recently approved treatments Mipomersen and Lomitapide suffer from severe side effects and are available under a Risk Evaluation and Mitigation Strategy only. We expect our Apo-Skip treatment to avoid these side effects with at least comparable efficiency due to the mechanism of action. We have demonstrated a 50 % reduction in LDL levels in a mouse model and the patent is submitted. At this point in time we are looking to progress into the pre-clinical stage in preparation for a first-in-man study and would welcome company expertise and input. We can offer expertise, well-established assays and disease models.

8) Professor Edward Tuddenham - A gene therapeutic cure for severe factor VII deficiency
The product is a gene therapeutic cure for severe factor VII deficiency. This often fatal recessive type of haemophilia affects about one per million of the population and is caused by mutations in the factor VII gene. Currently the only therapy is with intravenous infusion of recombinant factor VIIa given at least daily due to very short half life in the circulation. We have previously demonstrated successful conversion of severe factor IX deficiency (a less rare type of haemophilia) to a mild form by gene therapy. Our preclinical work shows that a vector based on the successful factor IX treatment but containing factor VII is highly effective in mice with severe factor VII deficiency protecting them from early death and giving a normal response to injury. Our plan is to take this new vector through the necessary stages for clinical translation with the aim to start clinical trials in 2014/15. Our extensive experience with gene therapy for the commoner related conditions of haemophilia B and A will ensure a smooth transition. We have intellectual property to protect our Factor VII vector and will apply for orphan drug status. We estimate that there are up to a hundred suitable cases for treatment in Europe whose current management costs up to £1,000,000 annually due to the extremely high price of
Elevator Pitch Abstracts

recombinant factor VIIa and necessity for frequent infusion. A single dose treatment will both prevent all bleeding and is likely to be effective for many years even lifelong.

9) Dr Paul Winyard - A new treatment to reduce cyst progression in Polycystic Kidney Disease
PKD is an inherited condition which causes kidney failure, which can only be managed by dialysis and transplantation. We have a new treatment for the blood vessels that significantly reduces cyst progression.

10) Dr Shabbir Moochhala - What type of kidney stones do I have? Fast and accurate diagnosis in the clinic
Kidney stone disease is becoming more common worldwide. It causes pain, often recurs and can cause kidney failure. We have a new way of diagnosing the exact type of kidney stone in a clinic setting, allowing rapid and directed treatment/advice.

11) Professor Roger Gunn - Reducing Risk in Translation: Imanova - driving innovation in imaging sciences
The acceleration of drug development through experimental medicine and ‘fast-to-fail’ are important concepts, especially in rare diseases. The use of molecular and multi-modal imaging can provide early information in-man on disease processes, drug-target engagement and dosing, which can help to reduce risks in decision making in small data rich studies. Imanova, a novel JV company between the MRC, UCL, ICL and Kings, provides a full range of imaging services to support early-stage drug development. With a pharmaceutical company pedigree and a world-class science base, we understand the drug development process and the importance of collaboration, high quality and working to deadlines.

12) Dr Khalid Hussain - A new medical therapy for congenital hyperinsulinism title tbc
Congenital hyperinsulinism (CHI) causes severe hypoglycaemia in the neonatal and infancy periods. The hypoglycaemia causes brain damage leading to mental retardation, cerebral palsy and epilepsy. Typically patients with CHI do not respond to medical therapy and require a near total pancreatectomy resulting in life long diabetes mellitus. We have pilot data to show that these patients might be managed with a new form of medical therapy thus avoiding a major pancreatectomy. We wish to take this project further.

13) Professor Simon Heales - Diagnosis, target identification and biomarker discovery for rare diseases
Pathology at Great Ormond Street Hospital and the Neurometabolic laboratory at the National Hospital, Queen Square. These laboratories have an international reputation for the diagnosis and monitoring of rare inherited disorders. The take home message of my pitch is that our NHS/university labs are open for business and that we can and do work successfully with industry in mutually beneficial ways. We have a wide range of expertise, methods and equipment. Furthermore, our laboratories are fully accredited which means that we work and adhere to very high quality standards. The collaborations that we have and would like to develop further are in the areas of diagnostics, elucidation of disease mechanisms for the identification of potential therapeutic targets and biomarker discovery with an emphasis on reporting treatment efficacy.

14) Professor Francesco Muntoni - Antisense oligonucleotide therapies for neuromuscular disorders
Following the successful development of morpholino antisense oligonucleotides to induce exon skipping and restoration of the reading frame in Duchenne muscular dystrophy, we are currently working at 2 different areas in which antisense oligonucleotides could be used: exon retention (relevant for spinal muscular atrophy) and allele specific exon skipping to induce out-of-frame deletions in mutant alleles in autosomal dominant neuromuscular conditions. We are currently targeting collagen VI related muscular dystrophies and RYR1 related congenital myopathies, for which we have good model systems.
### Attendees

<table>
<thead>
<tr>
<th>Name</th>
<th>Institution/Company</th>
<th>Position/Role</th>
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</thead>
<tbody>
<tr>
<td>Dr Gioia Altobelli</td>
<td>UCL Division of Medicine</td>
<td>Design/analysis of transcriptomics in clinical trials for early treatment of rheumatoid arthritis (RA)</td>
</tr>
<tr>
<td>Neta Amior</td>
<td>UCL Institute of Neurology</td>
<td>Periodic Paralysis and intracellular signaling pathways</td>
</tr>
<tr>
<td>Dr Virginia Arechavala-Gomeza</td>
<td>UCL BioCruces Health Research Institute</td>
<td>Dystrophin quantification in Duchenne muscular Dystrophy</td>
</tr>
<tr>
<td>Dr Chiara Bacchelli</td>
<td>UCL Centre for Translational Genomics – GOsgene</td>
<td>Manager of the Centre for Translational Genomics-GOsgene at the UCL Institute of Child Health</td>
</tr>
<tr>
<td>Professor Philip Beales</td>
<td>UCL Institute of Child Health</td>
<td>Clinical and genetic research in ciliopathies, including Bardet-Biedl syndrome, Jeune Asphyxiating Thoracic Dystrophy</td>
</tr>
<tr>
<td>Dr Lesley Bergmeier</td>
<td>Queen Mary University of London</td>
<td>Behcet's disease and innate immunity at the oral mucosal barrier</td>
</tr>
<tr>
<td>Dr Mike Bond</td>
<td>UCL Laboratory for Molecular Cell Biology</td>
<td>Neurodegenerative disease</td>
</tr>
<tr>
<td>Dr Elliot Bland</td>
<td>Queen Mary Innovation</td>
<td>Business Development &amp; Licensing (Tech Transfer)</td>
</tr>
<tr>
<td>Dr Laurent Bozec</td>
<td>UCL Eastman Dental Institute</td>
<td>Establishment of UK Research Centre on Dental Anomalies</td>
</tr>
<tr>
<td>Dr Paul Brogan</td>
<td>UCL Institute of Child Health</td>
<td>Pathogenesis, including genetic contribution, diagnosis, treatment, and prognosis of rare autoimmune and autoinflammatory diseases of the young. Rare disease clinical trials</td>
</tr>
<tr>
<td>Dr Thomas Brown</td>
<td>TPP Global Development Ltd.</td>
<td>A UK-based, biotech company focused on developing preclinical drug development programmes to a commercialization point at which they can be licensed to large pharmaceutical/biotech companies, or spun out into separate standalone operating companies</td>
</tr>
<tr>
<td>Chris Chaney</td>
<td>Great Ormond Street Hospital</td>
<td>Establishing a Centre for Children's Rare Disease Research in partnership with Great Ormond Street Hospital and the UCL Institute of Child Health</td>
</tr>
<tr>
<td>Dr David Chau</td>
<td>UCL Institute of Neurology</td>
<td>Parkinson's disease</td>
</tr>
<tr>
<td>Dr Gioia Cherubini</td>
<td>Queen Mary University of London</td>
<td>Business Development Manager</td>
</tr>
<tr>
<td>Dr Rob Cooke</td>
<td>Heptares Therapeutics</td>
<td>Structure-based drug discovery for GPCRs</td>
</tr>
<tr>
<td>Dr Kevin Cox</td>
<td>Imanova Limited Imaging</td>
<td></td>
</tr>
<tr>
<td>Dr Nara Daubeney</td>
<td>NHS</td>
<td>Diabetes</td>
</tr>
<tr>
<td>Dr Piers Daubeney</td>
<td>NHS</td>
<td>Diabetes</td>
</tr>
<tr>
<td>Dr Jasper de Boer</td>
<td>UCL Institute of Child Health</td>
<td>Drug discovery for childhood leukaemia</td>
</tr>
<tr>
<td>Dr Audrey Duncanson</td>
<td>The Wellcome Trust</td>
<td>Senior Portfolio Developer</td>
</tr>
<tr>
<td>Professor Perry Elliott</td>
<td>The Heart Hospital, UCL</td>
<td>Inherited cardiovascular diseases</td>
</tr>
<tr>
<td>Elisa Fassone</td>
<td>UCL Institute of Child Health</td>
<td>Mitochondrial disease, inborn errors of metabolism</td>
</tr>
<tr>
<td>Andrea Ferreira</td>
<td>UCL Institute of Neurology</td>
<td>Neurosciences, Cancer Research and Genetics</td>
</tr>
<tr>
<td>Dr Daniel Gale</td>
<td>UCL Centre for Nephrology / Royal Free Hospital</td>
<td>Genetic kidney disease. Current work focusses on complement regulation and metabolism</td>
</tr>
<tr>
<td>Dr Amir Gander</td>
<td>UCL Department of General Surgery</td>
<td>Lead for Tissue Access for Patient Benefit (TAPb)</td>
</tr>
</tbody>
</table>
Attendees

Dr Adam Giangreco  
UCL Internal Medicine  
Epithelial cell signalling in tissue homeostasis, repair and disease

Dr Francesca Gliubich  
Queen Mary University of London  
Focus on support external partnership with industry and academic centres of excellence and the not for profit sector

Professor David Galton  
Barts Medical School  
Molecular genetics of common disease

E. Glynn  
Great Ormond Street Hospital Children’s Charity

Professor David Goldblatt  
Great Ormond Street Hospital / UCL Institute of Child Health  
Director, NIHR BRC  
Immune response to vaccines and infectious diseases in childhood

Dr Kirstin Goldring  
UCL School of Life and Medical Sciences  
Use of human samples, panels and cohorts for research purposes

Professor Roger Gunn  
Imanova Limited  
Imaging

Dr James Hagan  
Global Medical Excellence Cluster

Dr Sarah Hardy  
The Wellcome Trust Technology Transfer Division: Representing the Wellcome Trust’s Pathfinder scheme which aims to catalyse partnerships between industry and academia in orphan and neglected diseases

Cassie Harley  
UCL School of Life and Medical Sciences  
Communications and Events Officer

Professor Stephen Hart  
UCL Molecular Immunology Unit  
LipTide Nanoparticles for Genetic Therapies of Cystic Fibrosis

Professor Philip Hawkins  
UCL Institute of Child Health  
Pathogenesis, diagnosis, clinical evaluation and treatment of amyloidosis and inherited periodic fever syndromes

Professor Simon Heales  
UCL Professor of Clinical Chemistry / Head of Chemical Pathology, Great Ormond Street Hospital / Head of Neurometabolic Unit, National Hospital

Rachel Heatley  
UCL School of Life and Medical Sciences  
Communications and Events Officer

Dr Steven Heggie  
UCLH Biomedical Research Centre  
Cancer Programme Operations Manager

Dr Rachel Hemsley  
UCL Business PLC  
Ophthalmology

Dr Daniel Herron  
UCLH Biomedical Research Centre  
Neurosciences Programme Operations Manager

Dr Wendy Heywood  
UCL Institute of Child Health  
Translational biomarker research

Dr Dayle Hogg  
Imperial Innovations Ltd.

Dr Steven Howe  
UCL Institute of Child Health  
Gene therapy for metabolic diseases using retroviral vectors and investigating blood stem cells and their application in treating inherited disease

Dr Derralynn Hughes  
Royal Free Hospital / UCL  
Lysosomal Storage Disorders

Dr Bethan Hughes  
The Wellcome Trust Technology Transfer Division: Representing the Wellcome Trust’s Pathfinder scheme which aims to catalyse partnerships between industry and academia in orphan and neglected diseases

Rachel Hughes  
Great Ormond Street Hospital  
Raising funds for rare disease research taking place within the ICH

Dr Julian Hughes  
Great Ormond Street NIHR Biomedical Research Centre  
Facility Manager

Dr Khalid Hussain  
UCL Institute of Child Health / Great Ormond Street Hospital  
Paediatric Endocrinology

John Irwin  
ViroPharma Europe  
ViroPharma's development and commercialization work is centered on rare diseases. ViroPharma is actively engaged in the identification and acquisition/licensing of post-POC clinical opportunities
# Attendees

<table>
<thead>
<tr>
<th>Name</th>
<th>Institution/Position</th>
</tr>
</thead>
</table>
| **Professor Ann Jacklin**    | Imperial College London
Patient safety research, Previous role as THE pharmacist on the Advisory Group for National Specialised Commissioning |
| **Dr Dan Jagger**            | UCL Ear Institute
Ciliopathies, genetic syndromic/non-syndromic deafness                                |
| **Dr Ruth Jamieson**         | UCL School of Life and Medical Sciences
Research Strategy Coordinator for School of Life and Medical Sciences' Experimental Medicine Domain |
| **Professor Parmjit Jat**    | UCL Institute of Neurology
Cellular senescence, ageing, cancer and neurodegeneration                             |
| **Dr Louise Johnston**       | Newcastle University
Neuromuscular field                                                                   |
| **Bernard Jolles**           | D-Gen Limited
Development and commercial exploitation of diagnostic tests and therapeutics for prion and other neurodegenerative diseases including Alzheimer's Disease |
| **Annika Jones**             | Great Ormond Street Hospital
Most of the children treated at Great Ormond Street Hospital are affected by a rare disease, which is why it is the ideal location for the new Centre for Children's Rare Disease Research, opening in 2017 |
| **Dr Xiayi Ke**              | UCL Institute of Child Health
Genetic epidemiology, epigenetics, bioinformatics                                        |
| **Assel Kashkenbayeva**      | UCL Neuroscience                                                                    |
| **Bridget Lacey**            | ViroPharma Europe                                                                  |
| **Catherine Lavery**         | Queen Mary University of London
Rare diseases                                                                           |
| **Dr Kevin Lee**             | Pfizer
Guest presenter                                                                       |
| **Jonathan Lees**           | UCL Division of Biosciences
Use network biology and high throughput datasets for target prioritisation of diseases |
| **Dr David Long**            | UCL Institute of Child Health
Renal Development and Disease, Vascular Biology                                        |
| **Dr Luis Lopes**            | The Heart Hospital, UCL
New genetic determinants of the phenotype in hypertrophic cardiomyopathy               |
| **Richard Lynn**             | British Paediatric Surveillance Unit
Scientific Coordinator                                                                 |
| **Professor Raymond MacAllister** | UCL Division of Medicine
Two main areas; organ protection during ischaemia and approaches to understanding and treating pulmonary hypertension |
| **Professor Tom MacDonald**  | Blizard Institute, Queen Mary University of London                                  |
| **Natasha Makengo**          | UCL Institute of Child Health / Great Ormond Street Hospital
Childhood Arthritis
Prospective Study coordinator                                                        |
| **Dr Thomas McCorvie**       | Structural Genomics Consortium, Oxford University
Determining the structural basis of missense mutations associated with disease and protein misfolding, specifically on cystathionine beta-synthase deficiency |
| **Dr Barry McGuinness**      | A UK-based, biotech company focused on developing preclinical drug development programmes to a commercialization point at which they can be licensed to large pharmaceutical/biotech companies, or spun out into separate standalone operating companies |
| **Dr Lou Metherell**         | William Harvey Research Institute, Queen Mary University of London
Genetics of endocrine disease                                                          |
| **Dr Tim Meyer**             | UCL Cancer Institute
Gastrointestinal cancer; Clinical trials; Antibody targeted therapy                  |
| **Dr Andrew Millar**         | Genzyme Therapeutics
Rare Diseases                                                                           |
| **Dr David Miller**          | UCL Translational Research and Industrial Partnerships
Translational research at UCL                                                           |
Dr Hannah Mitchison
UCL Institute of Child Health
Molecular genetics of childhood rare disease including ciliopathies (primary ciliary dyskinesia, short-rib polydactyllys) and neurodegeneration (Batten disease)

Dr Sara Mole
UCL Division of Biosciences
Using yeast to identify new drugs and drug targets

Dr Shabbir Moochhala
Royal Free Hospital / UCL
Accurate diagnosis of kidney stone disease

Professor Gudrun Moore
UCL Institute of Child Health
Clinical Genetics

Professor Francesco Muntoni
UCL Institute of Child Health
Antisense oligonucleotides to induce splice modification of genes involved in neuromuscular disease

Dr Tamara Nicolson
Trinzyme UK

Dr Libby Oakden
UCL Business Plc.
Medical and biomedical innovation

Professor Chris O'Callaghan
UCL Institute of Child Health / Great Ormond Street Hospital
Primary ciliary dyskinesia and respiratory problems affecting cila. Developing novel therapies in these areas

Dr Reeba Oliver
Royal Free Hospital / UCL
My research area and interest lies in Immunology and dendritic cell immunomodulation

Dr Maria Martha Papachatzaki
QMUL & Barts Health NHS Trust
Cortical neurodegeneration and demyelination in MS

Dr Dipali Patel
Structural Genomics Consortium, Oxford University
Misfolding diseases e.g. Amyotrophic lateral sclerosis/Huntington disease – understanding the structural and biological properties of the proteins

Dr Beth Payne
UCL Cancer Institute
Developing new models of hematopoietic diseases that will facilitate our understanding of their genetic basis and development of novel treatments using high throughput screening of whole organisms

Professor Sir Mark Pepys
UCL Wolfson Drug Discovery Unit
Amyloidosis and acute phase proteins; drug discovery and development

Dr Clarissa Pilkington
UCL Institute of Child Health / Great Ormond Street Hospital
Clinical research into rare diseases in paediatric rheumatology including juvenile dermatomyositis and SLE

Dr Jamie Plumer
Imperial Innovations
Early stage venture capital investment in exciting rare disease projects

Dr Shamima Rahman
UCL Institute of Child Health
Development of biomarkers for the diagnosis of mitochondrial disease

Pavithra Rallapalli
UCL Division of Biosciences
Analysis of mutations in the Factor 8 and 9 genes and studying their impact on structure and function. Development of tools for effective management of mutational data; structural analysis

Sara Rees
Great Ormond Street Hospital
Centre for Children's Rare Disease Research at GOSH with UCL’s ICH

Dr Malcolm Roberts
Eisai Ltd.

Dr Carlo Russo
GlaxoSmithKline
Guest speaker

Dr Francesco Saverio Tedesco
UCL Department of Cell and Developmental Biology
My research is focused on the study of muscle regeneration, using murine models of muscular dystrophy and stem cell transplantation as a model system. I am also developing novel gene and cell therapy strategies for Duchenne muscular dystrophy

Dr Peter Sazani
Sarepta Therapeutics
RNA therapeutics for the treatment of Duchenne Muscular Dystrophy

Professor Elizabeth Shephard
UCL Division of Biosciences
The inherited disorder trimethylaminuria

Dr Claire Smith
UCL Institute of Child Health
Ciliopathies and respiratory infections
## Attendees

<table>
<thead>
<tr>
<th>Name</th>
<th>Institution/Company</th>
<th>Position/Role</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dr Jon Telfer</td>
<td>UCL Translational Research Office</td>
<td>Translational Project Manager</td>
</tr>
<tr>
<td>Dr Ian Macintyre Thomas</td>
<td>Soft Option</td>
<td>IT statistical analysis</td>
</tr>
<tr>
<td>Dr Natasha Tian</td>
<td>Queen Mary University of London</td>
<td>Business Development Manager</td>
</tr>
<tr>
<td>Dr Jonathan Tobin</td>
<td>Imperial Innovations</td>
<td>Investment into early stage science-based opportunities</td>
</tr>
<tr>
<td>Professor Richard Trembath</td>
<td>Queen Mary University of London</td>
<td>Identify genes and delineate the molecular pathways underlying a range of human genetic disorders using both established and emerging technologies</td>
</tr>
<tr>
<td>Peter Trill</td>
<td>TPP Global Development Ltd</td>
<td>A UK-based biotech company focused on developing preclinical drug development programmes to a commercialisation point at which they can be licensed to large pharmaceutical/biotech companies, or spun out into separate standalone operating companies</td>
</tr>
<tr>
<td>Professor Edward Tuddenham</td>
<td>UCL Cancer Institute</td>
<td>Gene therapy for severe factor VII deficiency based on a vector that is highly effective in preclinical trials</td>
</tr>
<tr>
<td>Dr Pedro Vieira</td>
<td>Baxter</td>
<td>Immune deficiencies, Haemophilia</td>
</tr>
<tr>
<td>Dr Stephen Walsh</td>
<td>UCL Division of Medicine</td>
<td>Renal tubular physiology, specifically ion transport in the distal tubule. I work with patients with defects in renal salt transport, such as Gitelman and Bartter syndromes as well as those with acid base transport problems such as renal tubular acidosis</td>
</tr>
<tr>
<td>Dr George Wang</td>
<td>UCL School of Pharmacy</td>
<td>Pharmaceutics</td>
</tr>
<tr>
<td>Dr Aoife Waters</td>
<td>UCL Institute of Child Health</td>
<td>Haemolytic Uraemic Syndrome</td>
</tr>
<tr>
<td>Dr Tom Weaver</td>
<td>Congenica Ltd.</td>
<td>Congenica Ltd. is a newly formed company that provide genome based medicine with a focus on rare disease, pre-natal diagnostic testing and medical discovery</td>
</tr>
<tr>
<td>Professor Lucy Wedderburn</td>
<td>UCL Institute of Child Health</td>
<td>Paediatric rheumatology</td>
</tr>
<tr>
<td>Laura White</td>
<td>Great Ormond Street Hospital</td>
<td>Work on the Centre for Children’s Rare Diseases at GOSHCC</td>
</tr>
<tr>
<td>Emma White</td>
<td>ViroPharma</td>
<td></td>
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<tr>
<td>Helen Wise</td>
<td>UCL Enterprise</td>
<td></td>
</tr>
<tr>
<td>Dr Hywel Williams</td>
<td>UCL Centre for Translational Genomics - GOSgene</td>
<td>The identification of genetic mutations causing congenital diseases with novel phenotypes</td>
</tr>
<tr>
<td>Dr Chris Williams</td>
<td>UCL Business PLC</td>
<td>Senior Business Manager</td>
</tr>
<tr>
<td>Dr Paul Winyard</td>
<td>UCL Institute of Child Health</td>
<td>PKD is an inherited condition which causes kidney failure, which can only be managed by dialysis and transplantation. We have a new treatment for the blood vessels that significantly reduces cyst progression</td>
</tr>
<tr>
<td>Dr Zhi Yao</td>
<td>UCL Division of Biosciences</td>
<td>My main research focus is on Parkinson’s disease and how cell metabolism and mitochondria function can affect the disease development.</td>
</tr>
<tr>
<td>Dr Anselm Zdebik</td>
<td>UCL Division of Biosciences</td>
<td>Working on Epilepsy, Ataxia, Sensorineural deafness and Tubulopathy (EAST) syndrome due to mutations in KCNJ10. A zebrafish model shows epilepsy and can be “rescued” with human WT but not mutant cRNA, thus allowing targeted drug screening</td>
</tr>
</tbody>
</table>

For further information please contact
Dr Ruth Jamieson
(Experimental Medicine Domain Co-ordinator):

r.jamieson@ucl.ac.uk
+44 (0)20 7679 6165
(Ext: 46165)
Housekeeping Information

Cloakroom
All rooms have coat rails provided, please ensure you keep valuables with you at all times as neither The Wellcome Trust nor the conference organiser has responsibility for any items missing. There is a cloakroom manned by Security on the ground floor if you wish to use this instead.

Use of mobile phones and pagers
Please make sure that you turn off all mobile phones and pagers, particularly if using microphones in the rooms. There is only limited reception in the Auditorium.

WiFi
Whilst in the Conference Centre guests have complimentary wireless access to the internet, please use the username and password below (case sensitive)

Username: Conference
Password: spring13

Toilets
Toilets are situated off the lift lobby on lower ground 2 and between the Franks and Dale rooms on lower ground 1.

Smoking
Smoking is not permitted in any area of the building, guests are asked to move away from the front doors to smoke on Euston Road.

Fire Procedure
There are no fire drills planned.
If the alarm is activated you must leave the building by the nearest exit and make your way to the assembly point, which is:

UCL MAIN QUADRANGLE, ALONG GOWER STREET
If you discover a fire, activate the alarm by breaking the nearest ‘break glass’ point found at each exit and various places around the building and follow the evacuation route:

- Do not use lifts.
- Do not stop to collect personal belongings.
- Do not re-enter the building until you have been told it is safe to do so

Occupants with a disability can make themselves known on arrival to discuss any assistance we may be able to provide including evacuation chairs or escorts required, and should follow the evacuation procedure along with other guests or wait at a refuge area for assistance.

Accident Procedure
Should you or any member of your party require first aid treatment or an ambulance, please call 2200/2222 or security on 8532 directly. This will ensure the correct assistance is provided.
Guests

- GlaxoSmithKline
- Pfizer
- Alexion
- Sarepta Therapeutics
- Eisai
- ProSensa
- Genzyme
- HEPTARES therapeutics
- TPP Global Development
- D-Gen
- Baxter