Rare Diseases, UCL

Wednesday 13\textsuperscript{th} February 2013
08.30 – 19.00

The Kennedy Lecture Theatre
Institute of Child Health
30 Guilford Street
London
WC1N 1EH
Symposium Steering Committee:
Professor Phil Beales (Chair)
Dr Ruth Jamieson (Co-ordinator)
Professor Perry Elliott
Dr Paola Giunti
Dr Robin Lachmann
Dr Sara Mole
Professor Gudrun Moore
Dr Andre Strydom

Lectures:
Will take place in the Kennedy Lecture Theatre, ground floor.

Posters:
Will take place in the Winter Gardens.

Lunch and Refreshments:
Will be served in Balcony and Winter Garden areas.

Drinks reception:
Drinks and nibbles will be served from 17.45 in the Balcony and Winter Garden.

UCL Rare Disease Steering Committee
Professor Phil Beales (Chair), Dr Ruth Jamieson (Co-ordinator), Dr Detlef Bockenhauer, Mr Jose Tomas Bras, Dr Gerard Conway, Dr Sarah Creighton, Professor Perry Elliot, Dr Daniel Gale, Dr Julian Gilmore, Dr Paul Gissen, Dr Paola Giunti, Professor Mike Hannah, Professor Henry Holden, Dr Robin Lachmann, Dr Helen Lachmann, Dr Nick Lench, Dr Juan Martinez-Barbera, Dr Nadia Micalli, Dr Hannah Mitchison, Dr Sara Mole, Professor Gudrun Moore, Professor Francesco Muntoni, Dr Rosaline Quinlivan, Dr Gill Rumsby, Dr Rachael Scahill, Professor Sanjay Sisodiya, Dr Andre Strydom, Professor Sarah Tabrizi, Dr Aoife Waters, Professor Lucy Wedderburn.

The UCL Rare Diseases 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.

A first aim of the Committee is to achieve a comprehensive UCL-wide rare disease portfolio that connects databases, registries, biobanks and clinical bioinformatics for rare disease research and management. During this event, we will be collecting data relating to the research and clinical activities being undertaken at UCL.

PLEASE PROVIDE DETAILS OF YOUR RARE DISEASE RESEARCH AT THE REGISTRATION DESK.
Dear Colleagues

On behalf of the UCL Rare Diseases Steering Committee, it is my pleasure to welcome you to the 2013 UCL Rare Disease conference. We have a thrilling agenda that reflects the diverse interests and initiatives in this exciting field.

The EU definition of a rare disease is one that affects fewer than 5 in 10,000 of the general population. In the UK, it is estimated that 7% (4.2 million) of the population will be affected by a rare disease at some point in their lives. Collectively, rare diseases represent a significant burden to the health services and in many cases, the systems and structures within the NHS are not ideally suited to meet the needs for those affected. The need for a coherent strategy to deal with rare diseases is recognised by European governments who will deliver an integrated EU Plan for Rare Diseases in 2013.

UCL and our Academic Health Science Centre, UCL Partners, are committed to developing our capacity to tackle rare diseases, working with our extensive, nationally commissioned services, partner healthcare organisations and patient groups to contribute to the UK’s overall strategy in this area. We serve a patient population of over 6 million, approximately 0.5 million of whom will have a rare disease, across the North Thames region and have an extensive network of world-leading health scientists who can deliver to the national agenda.

We hope you enjoy hearing of the outstanding work taking place across UCL and by our external colleagues. Please take the opportunity to interact with other delegates throughout the day and during the Evening Reception which will be held in the Balcony and Winter Gardens.

We are grateful to our sponsor Genzyme for their support of this event and extend a special thanks to all of our speakers and chairs. We hope you enjoy the day!

Professor Phil Beales, Chair, UCL Rare Diseases Steering Committee
08.30 – 09.00  Registration

Welcome and Introduction

09.00 – 09.10  Professor Sir John Tooke, Vice Provost (Health)

09.10 – 09.15  Professor Philip Beales, Chair - UCL Rare Diseases Steering Committee

SESSION 1: Disease Mechanisms and Pathways
Chair: Professor Philip Beales

09.15 – 09.45  Professor Patrick Chinnery - Newcastle University
Mitochondrial disorders.

09.45 – 10.15  Dr Veronica Kinsler - Institute of Child Health - UCL
The genetics of congenital melanocytic naevus syndrome.

10.15 – 10.45  Dr Anna Gloyn - University of Oxford
PTEN Mutations as a cause of constitutive insulin sensitivity and obesity: Cowden syndrome.

10.45 – 11.15  Professor Sarah Tabrizi - Institute of Neurology - UCL
TRACK-HD and beyond: yielding new insights in Huntington’s Disease.

11.15 – 11.45  Coffee and poster viewing
SESSION 2: Centre for Translational Genomics - GOSgene
Chair: Professor Gudrun Moore

11.45 – 12.05  Dr Chiara Bacchelli - Centre for Translational Genomics - UCL
New genes, old genes: dissecting the genetics of Primary Immunodeficiencies.

12.05 – 12.25  Dr Miriam Schmidts – Institute of Child Health - UCL
Use of whole exome sequencing in diagnostics of ciliopathies.

12.25 – 12.45  Dr Daniel Kelberman – Institute of Child Health - UCL
Identifying novel genes involved in childhood blindness.

12.45 – 13.45  Lunch and poster viewing

SESSION 3: Therapeutic Development
Chair: Professor Bobby Gaspar

13.45 – 14.05  Professor Paul Matthews - Imperial College and GSK
Opportunities for Industry-Academic-NHS collaboration with “Big Pharma”.

14.05 – 14.35  Professor Robin Ali - Institute Ophthalmology, UCL
Gene and cell therapies for retinal diseases.

14.35 – 15.05  Professor Tim Cox - University of Cambridge

15.05 – 15.35  Professor Francesco Muntoni - Dubowitz Neuromuscular Ctr., UCL
Antisense oligonucleotide therapeutic approaches for Duchenne Muscular Dystrophy.
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<td>15.35 – 16.30</td>
<td>Coffee and poster viewing</td>
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<td>Session 4: Policies and Programmes</td>
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<td>Chair: Alastair Kent, Genetic Alliance</td>
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<td>16.30 – 17.00</td>
<td>Dr Ségolène Aymé – ORPHANET – INSERM</td>
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<td>Rare disease research strategies for the EU.</td>
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<td>17.00 – 17.30</td>
<td>Professor Patrick Chinnery - Newcastle University</td>
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<td>NIHR Rare Disease Research and Development.</td>
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<td>17.30 – 17.45</td>
<td>Closing remarks</td>
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<td>Professor Malcolm Grant, UCL Provost</td>
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<td>17.45 – 19.00</td>
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Professor Philip Beales is Professor of Medical Genetics at UCL and a Wellcome Trust Senior Fellow in Clinical Science. He is 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, Cranioectodermal 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 Great Ormond Street Hospital for Children 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.

Gudrun Moore is a UCL Professor of Clinical and Molecular Genetics and the UCL-Institute of Child Health Deputy Director (Research). She has built an international reputation for her work on the genetics of intrauterine growth restriction and common pregnancy complications as well as genomic imprinting in the human. Her research has been generously funded by many charities including most recently WellBeing of Women, Sparks, the Wellcome Trust and the MRC. She has published more than 140 research papers covering both clinical and scientific research. She has close affiliations with clinical academic specialists, including Professor Lesley Regan, Imperial College London with whom she is building an International resource for researchers in common pregnancy failures: the Baby Bio Bank (https://www.ucl.ac.uk/tapb/sample...ucl/biobanks-ucl/baby-biobank). Professor Moore has trained 19 PhD students and many MSc Students. She has lectured extensively in the UK, Europe and the US and has built up numerous national and international collaborations. She has sat on a number of influential committees including five years with the medical advisory board for Sparks. In 2012 she was made an honorary fellow of the Royal College of Paediatrics and Child Health.
Professor Bobby Gaspar is a physician/scientist working in paediatric immunology at the Institute of Child Health (ICH) and Great Ormond Street London. He initially trained in paediatrics and then became interested in primary immunodeficiencies at an early stage in his career and undertook a PhD at the Molecular Immunology Unit at ICH. From there he has continued his academic and clinical career. His interests are in many different aspects of primary immunodeficiency including understanding the molecular and cellular defects and disease pathogenesis, bone marrow transplantation for severe immunodeficiencies and the development of gene therapy for these conditions. Over the last decade, his team have conducted clinical trials that have shown that gene therapy can successfully correct the immune defect in specific immunodeficiency conditions.

Alastair Kent OBE is the Director of Genetic Alliance UK – the national charity of over 150 patient organisations, supporting all those affected by genetic conditions. Genetic Alliance UK’s mission is to promote the development of the scientific understanding of genetics and the part that genetic factors play in health and disease, and to see the speedy transfer of this new knowledge into improved services and support for patients.

Alastair is also the Chair of Rare Disease UK (RDUK) the national alliance for people with rare diseases and all who support them. RDUK has over 1,200 members including over 220 patient organisations, health professionals, researchers, the pharmaceutical industry and individual patients and families.

Alastair has worked in the field of genetic and rare disease healthcare for over 15 years. Alastair represents the interests of patients on numerous platforms; he is the president of the European Genetic Alliances Network (EGAN), Director of the European Platform for Patient Organisations, Science and Industry (EPPOSI) and he sits on the Human Genetics Commission and the EU Committee of Experts on Rare Diseases amongst others.
**SPEAKERS’ PROFILES**

**Professor Patrick Chinnery** is a Wellcome Trust Senior Fellow in Clinical Science, Professor of Neurogenetics and Honorary Consultant at the University of Newcastle. His principal interest is in the clinical and molecular basis of neurological disease. He currently runs research projects studying the molecular aetiology of monogenic disorders (including inherited ataxia and spastic paraparesis), and multifactorial disorders (including Parkinson’s disease). Human mitochondrial disorders form a major focus of his work, particularly the interaction between nuclear and mitochondrial genes (focussing on Leber’s hereditary optic neuropathy) and the inheritance of mitochondrial DNA mutations. The research is supported by the Wellcome Trust and Ataxia UK.

**Dr Veronica Kinsler MA MB BChr MRCPCH PhD** Veronica Kinsler studied Medicine at Cambridge University and then went into Paediatrics. She developed an interest in Paediatric Dermatology early on in her career, through working at Great Ormond Street Hospital for Children (GOSH) in London. She now specialises in paediatric pigmentary conditions and genodermatoses there, and sees patients from around the UK.

Veronica is the academic lead clinician for the Dermatology Department at GOSH, and undertakes her research within the Clinical and Molecular Genetics Unit at the neighbouring UCL Institute of Child Health. This research has focused on the clinical and genetic characterisation of Congenital Melanocytic Naevi and their associated complications. She is Director of the International CMN Resource, a worldwide registry for affected individuals. More recently she has established the GOSH rare dermatology diseases resource, a tissue bank for facilitation of research into all rare diseases seen in the GOSH department.
Dr Anna Gloyn is currently a Wellcome Trust Senior Fellow in Basic and Biomedical Science based at the Oxford Centre for Diabetes Endocrinology and Metabolism at the University of Oxford. Anna's research is focused on using naturally occurring mutations in humans as tools to identify critical regulatory pathways and insights into normal physiology. Anna’s early post-doctoral research led to the identification a new genetic aetiology for permanent and transient neonatal diabetes due to \textit{KCNJ11} mutations and resulted in one of the first examples of the determination of the molecular genetic aetiology leading to improved treatment options for patients (Gloyn et al New England Journal of Medicine 2004). Recently the Gloyn group have reported a novel genetic cause of constitutive insulin sensitivity in humans due to mutations in the \textit{PTEN} gene (Pal et al New England Journal of Medicine 2012).

Current research projects are focused on the translation of association signals for type 2 diabetes and glycaemic traits into molecular, cellular and physiological mechanisms and clinically useful tools. Anna is an active member of several international consortia, including DIAGRAM (Diabetes Genetics Replication and Meta-analysis), MAGIC (Meta-analysis of Glucose and Insulin traits Consortium) and the Genetics of Type 2 Diabetes (GoT2D).

Anna’s work has been recognized both nationally and internationally as she is a recipient of a Diabetes UK RD Lawrence research fellowship (2003), a European Association for the Study of Diabetes Rising Star Award (2005), a Medical Research Council New Investigator Award (2007), the RD Lawrence Named Lecturer (Diabetes UK Annual Professional Conference 2009) and most recently a Wellcome Trust Senior Fellowship in Basic Biomedical Research (2011).

Sarah Tabrizi is Professor of Clinical Neurology in the Department of Neurodegenerative Diseases at the UCL Institute of Neurology and Honorary Consultant Neurologist at the National Hospital of Neurology and Neurosurgery. Her research focuses on understanding the basic cellular mechanisms of neurodegeneration, in particular Huntington's disease and prion biology. Sarah leads two major, multidisciplinary research initiatives, TRACK-HD and Track-On HD, aimed at understanding the neurobiology of the neurodegenerative changes in premanifest and early stage HD gene carriers and identifying sensitive measures of disease progression. After studying biochemistry, then medicine from the University of Edinburgh, Sarah moved to UCL for her clinical training and PhD as an MRC Fellow. She obtained a Department of Health National Clinician Scientist Fellowship in 2002 and became a full professor in
2009. Sarah has published over 150 peer-review articles, and her research has been the subject of a review article in New England Journal of Medicine, scientific articles in The Economist, Lancet Neurology, and Scientific American, and a personal profile in The Lancet (2nd June 2012). She serves on several executive and advisory panels including the UK HD association, the European HD Network and NINDS/NIH and co-founded the UK All Party Parliamentary Group for HD in 2010.

Dr Chiara Bacchelli is the Manager of the Centre for Translational Genomics-GOSgene at the UCL Institute of Child Health in London, led by Professor Phil Beales. Funded by the Great Ormond Street Hospital Biomedical Research Centre of the NIHR, GOSgene opened in February 2010 to facilitate rapid gene identification in uncharacterised genetic diseases. Her work and interest in genetics began during her PhD working on rare congenital disorders with Professor Peter Scambler (Molecular Medicine Unit, ICH). Dr Bacchelli then undertook a position in the Molecular Immunology Unit (ICH) working with Professors Bobby Gaspar and Adrian Thrasher on the genetic characterization of primary immunodeficiencies. After a period at the Institute of Cancer Research working in the Section of Cancer Genetics lead by Professor Nazneen Rahman, she took up the position of Manager of GOSgene. During the course of her career, Dr Bacchelli acquired a vast repertoire of molecular genetic techniques with focus on disease gene identification through linkage analysis, homozygosity mapping and nowadays next generation sequencing. Her current work in GOSgene focuses on the use of exome sequencing and advanced data analysis as tools for gene identification in affected individuals to help improve diagnostic testing, genetic counselling, family planning options and prenatal service development.

Dr Miriam Schmidts studied medicine at the University of Freiburg in Germany and undertook paediatrics specialist training at the Centre for Pediatrics and Adolescent Medicine in Freiburg from 2005 to 2009. In 2009, Dr Schmidts joined Professor Philip Beales and Dr Hannah Mitchison’s research group at the Institute of Child Health, University College London as a Clinical Research Fellow funded by the German Research Foundation (DFG) and is supported by an Action Medical research Clinical Training Fellowship since 2011. Dr Schmidts has a longstanding interest in both motile and non-motile hereditary ciliary diseases with special emphasis on disorders with renal involvement.
**Dr Daniel Kelberman’s** research interests involve all aspects of human genetic disease from mutation discovery, the molecular consequences of pathogenic variation on the underlying biology and natural history of disease, together with the role of molecular diagnosis in clinical management. He undertook his PhD studies at the UCL Institute of Child Health investigating the molecular genetics of craniofacial birth defects. He subsequently focussed his research on genetic disorders of the pituitary, forebrain and eye in the Clinical and Molecular Genetics Unit at ICH identifying and characterising mutations in patients with varying pituitary disorders and associated syndromic phenotypes, investigating the developmental role and expression of the genes involved.

More recently, he joined Professor Jane Sowden in the Ulverscroft Vision Research Group, a newly formed cross-disciplinary initiative with the remit to strengthen genetic research within the Birth Defects Research Centre at ICH and improve clinical care and services to patients with genetic ophthalmic disorders at Great Ormond Street Hospital. This role specifically involves the development of new translational research projects utilising modern molecular genetic techniques, as well as collaboration between a number of basic scientists and clinicians in a variety of specialties aiming to improve knowledge of the causes, diagnosis and management of children with inherited eye disorders.

**Professor Paul Matthews**, OBE, MD, DPhil, FRCP is Head of the new Division of Brain Sciences in the Department of Medicine of Imperial College, London. His research is noted for innovative translational applications of clinical imaging for the neurosciences. This has developed with exploitation of the powerful synergies between the physical and quantitative sciences and medicine. He was the founding Director of two internationally leading research imaging centres, the University of Oxford Centre for Functional Magnetic Resonance Imaging of the Brain (FMRIB) and, later, of GlaxoSmithKline’s Clinical Imaging Centre (now a public “spin out” as Imanova, Ltd). He continues to hold a role in GlaxoSmithKline (GSK) as a Vice President in Medicines Discovery and Development and, therefore, is one of the small number of senior clinical academics in the UK with industry experience.
Robin Ali is Professor of Human Molecular Genetics at UCL Institute of Ophthalmology, London where he is also Head of the Department of Genetics. He also holds a faculty position at UCL Institute of Child Health and is the Theme Leader for Gene Therapy at NIHR Biomedical Research Centre for Ophthalmology, Moorfields Eye Hospital.

The main focus of his research is the development of gene and cell therapy for the treatment of retinal disorders. He established the world’s first clinical trial of gene therapy for retinopathy. The results from this trial reporting an improvement in vision (New England J Med, 2008), along with results from two other trials, established proof-of-principle of gene therapy for inherited retinal degeneration and are widely regarded as a landmark for the whole gene therapy field. His group has also provided the first proof-of-concept for effective transplantation of photoreceptors (Nature 2006; Nature, 2012) which has provide the basis for ES cell-derived photoreceptor transplantation, now a major programme in his laboratory. Robin Ali and his team have received numerous prizes and awards for their work on developing new treatments for retinal degeneration including the Pfizer/ARVO Karl Camras Translational Award in 2010 and Alcon Research Institute Award in 2009. In 2007 he was elected to the UK Academy of Medical Sciences and in 2009 appointed Senior Investigator of The UK National Institute of Health Research. He serves on the advisory boards of a number of funding bodies including the UK MRC as well as the advisory boards of pharmaceutical and biotech companies, including Alcon/Novartis and ReGenX.

Tim M Cox is Professor of Medicine at the University of Cambridge and Honorary Consultant Physician at Addenbrooke’s NHS Foundation Trust Hospitals. A trained Internist, his particular interest is in the pathogenesis and treatment of inborn errors of metabolism. He is an Editor of the Oxford Textbook of Medicine.
Professor Francesco Muntoni is a clinical paediatric neurologist with an interest in clinical, pathological and molecular aspects of neuromuscular disorders. The Dubowitz Neuromuscular Centre where he works at UCL Institute of Child Health and Great Ormond Street Hospital is the largest paediatric neuromuscular centre in UK; in 2001 it was designated by the Department of Health as the National reference centre for clinical, pathological and genetic aspects related to the rare conditions congenital muscular dystrophies and congenital myopathies. Professor Muntoni’s research activities are focused on the genetic diversity of neuromuscular disorders, with the identification of 26 neuromuscular disease genes in the last 13 years; on understanding pathophysiology of mutant neuromuscular genes; and on translational research focused on Duchenne muscular dystrophy. In this area he has previously obtained funding and completed investigator led studies focused on novel genetic therapy- antisense oligonucleotides for skipping exon 51 of the dystrophin gene. In November 2012 he obtained funding from the European Commission to coordinate a multicentric study using a new antisense oligonucleotide targeting a different exon of the dystrophin gene.

Ségolène Aymé is a medical geneticist and Emeritus Director of Research at the French Institute of Health and Medical Research (INSERM). She was the founder of Orphanet in 1997 (www.orpha.net) and its Executive Manager from 1997 up to 2011. Orphanet is the webportal dedicated to rare diseases and orphan drugs which is currently funded by the French Ministry of Health, the INSERM and the European Commission as a Joint Action (DG Public Health). She chairs the European Union Committee of Experts on Rare Diseases (www.eucerd.eu) and the WHO Topic Advisory Group for Rare Diseases. She serves as Editor-in-Chief of the Orphanet Journal of Rare Diseases (www.ojrd.com). She is the project leader of “Support IRDiRC”, which provides the services of a scientific secretariat to (www.irdirc.org).
1. Rosella Abeti, UCL  
Addressing Mitochondrial function in a mouse model of Friedreich’s Ataxia (FRDA).

2. Katherine Aitchison, UCL  
Developing lentiviral vectors for gene therapy of type I Gaucher disease.

3. Michael Bond, UCL  
Modelling inherited neurodegeneration in fission yeast.

4. Stuart Brown, UCL  
Linear morphea fibroblasts possess distinct TGFβ responsiveness

5. Rachel Brown, UCL  
A transposon-based mutagenesis system in the Sz.pombe model of juvenile CLN3 disease.

6. Valentina S Caputo, Imperial College  
Polycomb-mediated, bivalent chromatin domain-associated transcriptional repression as a novel pathogenetic mechanism in inherited disease.

7. Anna Cariboni, UCL  
Semaphorin signalling in the control of GnRH neuron development and Kallmann Syndrome.

8. Caroline Coats, UCL  
Sub-Clinical Left Ventricular Dysfunction in Anderson Fabry Disease Assessed By Two-Dimensional Speckle Tracking Echocardiography.

9. Joana Costa, Imperial College  
Cell- and tissue-specific transcriptional regulation of PIGM by the generic transcription factor Sp1 explains the divergent cell and clinical phenotypes in inherited glycosylphosphatidylinositol deficiency.

10. Seema Dhanjal, UCL  
Availability of Preimplantation Genetic Diagnosis for rare genetic disorders at the UCL Centre for PGD.

11. Suzanne Drury, UCL  
Shortening the diagnostic odyssey for primary immune deficiencies – novel molecular findings using next generation sequencing gene panels.

12. Suzanne Drury, UCL  
New tools for diagnosing rare diseases – Next generation sequencing in the diagnostic lab.

13. Elisa Fassone, UCL  
Whole Exome Sequencing In Complex I Deficient Leigh Syndrome, A Rare Mitochondrial Disease.
14. Daniel Gale, UCL
   Whole exome sequencing in familial kidney disease.

15. Alice Gardiner, UCL
   Next Generation Sequencing of Ion Channels in Neurological Disorders.

16. Dianne Gerrelli, UCL
   The Human Developmental Biology Resource.

17. Glenn Irvine, Advocacy for Neuroacanthocytosis Patients
   Differential Diagnosis of Chorea.

18. Ay Lin Kho, King’s College London
   Recessive mutations in EPG5 cause Vici syndrome, a multisystem disorder with defective autophagy.

   Exon Skipping of Hepatic APOB pre-mRNA with Splice-switching Oligonucleotides Reduces LDL Cholesterol In Vivo.

20. Sophia Kleine Holthaus, UCL
   Gene therapy to improve vision in Neuronal Ceroid Lipofuscinosis.

21. Razan Jawdat, UCL
   Investigating the Role of Mitochondria in the Preimplantation Embryo.

22. Richard Lynn, UCL
   Surveillance of rare paediatric disease.

23. Davide Marotta, UCL
   Loss or overexpression of CLN3/btn1 affects the Golgi complex in juvenile neuronal ceroid lipofuscinosis.

24. Suran Nethisinghe, UCL
   The role of interruptions in polyglutamine in the pathology of SCA1.

25. Atul Mehta, UCL
   Lysosomal Storage Disorders - The Royal Free Unit.

26. Ryan O'Shaughnessy, UCL
   Identification of a hyperkeratosis “signature” in lamellar ichthyosis: Mdm2 inhibition prevents hyperkeratosis in lamellar ichthyosis models.

27. Michael Parkinson, UCL
   Optical Coherence Tomography Studies of Retinal Changes in Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS) and Other Genetic Ataxias.

28. James Polke, UCL
   Transcript Analysis of a Variably-Penetrant GTP Cyclohydrolase Intronic Splicing Mutation.

29. Panicos Shangaris, UCL
   Haematopoietic stem cells derived from sheep amniotic fluid engraft after transplantation, a potential source for an in utero autologous cell therapy.
30. **Mariana Vieira, UCL**  
*Strategies for identification of new therapeutic targets for juvenile neuronal ceroid lipofuscinosis. using fission yeast.*

31. **Cathy Woodward, UCL**  
*Next Generation Sequencing as a potential diagnostic tool for mitochondrial DNA diseases.*

32. **Anselm Zdebik, UCL**  
*Mutations in TRPV4 cause Charcot-Marie-Tooth type 2C.*
The **Experimental Medicine Domain** is one of nine domains in the School of Life and Medical Sciences at UCL.

Our aim is to foster innovation in medical diagnosis and to develop new therapeutic approaches. We will stimulate bidirectional interchange between clinical medicine and science in order to open up new avenues of research, and to apply scientific discoveries to improve medical care. A particular focus is proof-of-concept experiments in humans. As part of the domain activities, we are delighted to facilitate the organisation of the UCL Rare Diseases conference and hope you find the event both interesting and stimulating.

**Professor William Rosenberg, Chair, UCL Experimental Medicine Domain**  
**Dr Ruth Jamieson, Strategic Co-ordinator Experimental Medicine**

[http://www.ucl.ac.uk/slms/domains/experimental-medicine](http://www.ucl.ac.uk/slms/domains/experimental-medicine)  
[**r.jamieson@ucl.ac.uk**](mailto:r.jamieson@ucl.ac.uk)

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**ICH Events**  
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