Organised by

PCD Research
PCDresearch.org

Sponsored by

ReCode Therapeutics
LifeArc
Weatherden
UKRI MRC National Mouse Genetics Network
Congenital Anomalies Cluster
Pinsent Masons

Rare Diseases Industry Accelerator Day

Getting Cilia Moving

Conference Programme

1 OCT 2024

Francis Crick Institute, London | 9:00-17:30
followed by a drinks reception

Foreword

By Dr Harriet Holme, Founder and Exec Chair of PCD Research

Welcome to the 1st Rare Disease Industry Accelerator Day – Getting Cilia Moving, a pivotal gathering to unite the diverse ecosystem of stakeholders driving transformational change across the rare disease landscape.

It's easy to hear the word "rare" and think it implies a small problem. While each rare disease affects fewer than 100,000 people in Europe, there are over 10,000 rare diseases in total. In the UK alone, 4 million people live with a rare disease, and more Americans live with a rare disease than those who have HIV, heart disease, or stroke combined.

Beyond patients, these life-limiting conditions significantly impact families and caregivers, with an estimated 1 in 7 people, or 9.4 million in the UK, affected. 95% of rare diseases do not have an approved treatment. Worldwide, the number of affected individuals is staggering, highlighting the urgent need for collective action.

Despite their distinct nature, rare diseases share many similarities in terms of their drug discovery and development pathways. These similarities mean infrastructure can be created to serve the community as a whole, fostering economies of scale and driving a new era of investment and therapies.

Here, we showcase primary ciliary dyskinesia (PCD), that affects 1:7,500 births and impairs mucociliary clearance leading to decline in lung function similar to cystic fibrosis (CF). Unlike CF, no approved treatments exist for PCD, despite a similar global market size and significant unmet need.

I see PCD as a test case, with wider applicability across rare disease and personalised medicine showing how we can connect all the dots to market. This event looks to leverage intellectual property from cystic fibrosis into PCD, together with a wider conversation on how we can drive transformational change in rare disease from development through to policy and regulation.

We stand on the brink of a new era where we can truly revolutionise the process for millions of people with rare diseases through molecular characterisation, data integration, advanced genomic technologies, progressive regulation and improved clinical trial design. By aligning the UK landscape to drive commercial value, we can transform the lives of those affected by rare diseases, ensuring this population is not excluded from scientific progress. This represents a tractable, scalable solution, not just for rare diseases, but the future of personalised medicine.

I hope this conference will be more than just a series of presentations and discussions; instead, I see an opportunity to break down silos, share knowledge, and create a cohesive strategy that harnesses the strengths of each sector within our ecosystem and aligns them to a shared goal. By bringing together clinicians, researchers, industry leaders, investors, policymakers and patient advocates, we can forge new pathways for innovation, accelerate the development of life-changing therapies, and ensure that no one is left behind.

Thank you for your vision, your expertise, and your passion. Let this conference be the catalyst for the change we all wish to see, and may our collective efforts lead to a brighter, healthier future for all those living with rare diseases. We are grateful to our generous sponsors and tireless volunteers: LifeArc, the LifeArc Centre for Rare Respiratory Diseases, Recode Therapeutics, Weatherden, MRC National Mouse Genetics Network (NMGN), NMGN Congenital Anomalies Cluster, Pinsent Masons, and Axon-com.com, together with Florence Barkats, Caroline McHugh, Amy Dréan, and AXON.

Dr Harriet Holme

MA MBBS PhD MRCPCH(2009)

Founder and Exec Chair PCD Research

Drug Development Clinician, Weatherden

Parent testimonial

Living with Primary Ciliary Dyskinesia

At the heart of every rare disease journey are personal stories that remind us of the pressing need for disease-modifying therapies. These stories not only illustrate the daily challenges families face but also highlight the urgency for treatments that can provide hope. Michael Dann and Mary Neal are parents whose son is living with primary ciliary dyskinesia (PCD), speak for countless parents who find themselves navigating uncharted territories with little evidence-based guidance or treatment options:

Michael's and Mary's words will resonate with many in the rare disease community, emphasising the emotional, physical, and mental toll rare diseases can take on entire families.

"Living With Primary Ciliary Dyskinesia is to live with great uncertainty. From the moment of our newborn son's breathing crisis through today, we do not know what the future will hold. We spend an hour a day on chest physiotherapy and nebulised saline solution without any knowledge of its effectiveness. We have no way to know the severity of our son's condition from his genotype or from his current symptoms. We see other PCD patients in their 30s on lung transplant lists and feel terrified for his future. Yesterday, as we dropped our child off at preschool for the first time, he loved it, while we could only hear the other children coughing and worried for his safety. Even milestones that should be happy and joyous are clouded by fear and doubt that our actions could be shortening his life. He is a bold, joyous boy, and we would like to keep it that way.

The lack of a treatment or even basic information about PCD is a barrier to living our lives."

Parents Mary Neal & Michael Dann

This story is not just one of personal struggle, but of the profound need for transformational change in the rare disease landscape. It serves as a poignant reminder that without significant investment and research into therapies for rare conditions like PCD, countless families are left in uncertainty, forced to cope with more questions than answers.

As we gather to push forward the conversation about rare diseases, Michael's testimony underscores why this work is so vital. Families like his deserve more than hope—they deserve tangible solutions, therapies that can ease the burden, and a future that isn't defined by limitations.

Dann Family 2024

Agenda

1 OCT 2024
Francis Crick Institute
London | 9:00 - 19:00

8.30 **Registration Opens**
Registration and Morning Coffee

Session I: Opportunities in Rare Disease – PCD as an Exemplar For Change

Session Chair:
Prof Pleasantine Mill

9.00 **Welcome and Introduction**

Dr Harriet Holme MA MBBS PhD
Founder and Chair of PCD Research and Drug Development Clinician, Weatherden.

Welcome and introduction to highlight the shared challenges across rare diseases, and the huge opportunities for innovation. While the foundations are in place, a step-change is needed to overcome existing hurdles and drive both investment and therapies.

9:20 **Strategic Horizon Scanning for Rare Diseases in the UK**

Dr Kath Bainbridge PhD
Head of Rare Diseases and Emerging Therapies
Department of Health and Social Care

Exploration of HM's Government's vision for progress in rare diseases and outline of current roadblocks in rare disease. We will outline the efforts to overcome these challenges and set the scene as to where the opportunities for rare disease developments are.

09.40 **What is PCD?**

Prof Carlos Milla
Professor of Pediatrics and of Medicine at Stanford University School of Medicine

PCD is not unique in being a spectrum of clinical phenotypes and severities, which challenge patient diagnosis and stratification for trials. As with most rare diseases, the time to diagnosis remains a huge hurdle and burden to the patient and family. How do we improve this?

10.00 **Navigating Heterogeneity in Rare Disease**

Prof Steven Brody
Professor of Pulmonary Medicine and Physician-Scientist at Washington University in Saint Louis, MO, USA

Merging evidence supports specific genotypes are associated with the most severe disease phenotypes. We will explore the importance of understanding disease mechanisms to reverse clinical phenotypes, and define which patient groups should be targeted first.

10.20 **Questions led by Prof Pleasantine Mill**

Professor Pleasantine Mill
Group Leader, MRC Human Genetics Unit, University of Edinburgh

10.30 **Networking Break**

Session II: Rare Disease in an Age of Precision Medicine – Where Do We Need to Be?

Session Chair:
Dr Sam Barrell, CEO LifeArc

11.00 **Shared Commonalities of Rare Disease and the Value of Large Scale Data**

Prof James Chalmers
Professor James D Chalmers, Asthma and Lung UK Chair of Respiratory Research at the University of Dundee

Many rare diseases share tractable commonalities, either involving the same organ or sharing a mechanism. These domains, or commonalities allow researchers to study multiple diseases within a large database. One such example is the EMBARC database, which is the world's largest bronchiectasis dataset. This exemplifies the importance of data driven decisions in driving improved clinical decisions.

Agenda

11.30 **The Impact of the GECKO Study For Future Clinical Trials**

Prof Amelia Shoemark
Professor of Respiratory Research, University of Dundee

Understanding the landscape of genotype-phenotype correlations is important for progress in rare disease. We will hear about the Gecko study, and advances in characterisation of PCD.

Session III: Lessons Learned From Successes in Rare Disease Translation

Session Chair:
Prof James Chalmers

11.45 **Lessons Learnt From The Success of Cystic Fibrosis**

Prof Jane Davies OBE
Professor of Paediatric Respirology and Experimental Medicine, NHLI Imperial College

CFTR modulator drugs have been a huge clinical success, with hundreds of patients with cystic fibrosis now reaping substantial clinical benefits. We will discuss the approach taken to clinical trial design, progress into younger patients, and lessons learnt along the way.

12.00 **Insights Gained From The First RCTs in PCD**

Prof Thomas Ferkol
Professor of Pediatrics at the University of North Carolina at Chapel Hill

Over the past forty years, advancements in understanding of the genetics and pathophysiology of cystic fibrosis (CF) lung disease have led to new treatments. Highly effective medical therapies have dramatically changed the lives of many affected people, and the success of these treatments in CF shows what could be possible for other inherited, suppurative lung diseases, like PCD.

While CF and PCD have some similarities, there are significant differences between the two diseases. This talk will examine how the CF model could be used to accelerate clinical development for treatment of PCD and describe the obstacles that must be overcome to be successful. Additionally, we will review results of a recent, multicenter, international randomised clinical trial (RCT) specific for PCD, and discuss lessons learned from these efforts.

12.15 **Discussion - Experts and Audience**

Led by Prof James Chalmers

12.30 **Lunch**

Session IV: Opportunities for Acceleration within the UK

Session Chair:
Prof Emma Tinsley

13.30 **Translation Across Academia From Biology to Therapeutics**

Prof Pleasantine Mill
Group Leader, MRC Human Genetics Unit, University of Edinburgh

Looking to connect from basic cell biology through to therapeutic development, this talk will share the importance of deep molecular characterisation critical to understanding function, druggable pathways and routes to genetic disease modification.

The LifeArc Centre for Rare Respiratory Diseases will build the foundations needed to make progress in rare respiratory disease. Using PCD as an exemplar, it will look to provide the tools to develop genetic therapies and build partnerships with industry.

13.55 **Importance of Patient Derived Models**

Dr Sara Wells MBE and Prof Pleasantine Mill, Prof Emma Rawlins, Emma Tinsley (moderator)

This panel will explore the importance of patient led models, as well as challenges and strategies with regards to making the right one.

Patient derived models play an important role in improving patient outcomes through personalised medicine and precision approaches. Aligning genetic capabilities with patient derived modelling would aid effective translation.

1 OCT 2024
Francis Crick Institute
London | 9:00 - 19:00

Agenda

14.15 **Accelerating Development in Cell and Gene Therapies**

Matthew Durdy
Chief Executive of the Cell and Gene Therapy Catapult

Using real world case studies this presentation will explore how collaboration has led to acceleration of development in cell and gene therapies.

Genetic therapy technology such as LNP mRNA or ASO platforms have the potential to be powerful tools in the treatment of genetically heterogeneous rare diseases. By aligning progressive regulation with manufacturing the value proposition for these approaches would strengthen, opening up a world of personalised medicine.

14.30 **Joint Talk: Creating A Progressive Regulatory Framework To Support Innovation Across Rare Disease**


Dr Kirsty Wydenbach, Dr Dan O'Connor

Huge commercial investment in areas such as oncology have helped shape regulation and develop regulatory flexibilities. We need similar approaches for rare disease, to drive progress using shared molecular endpoints and for genetic approaches in genetically heterogeneous rare diseases. This would drive the value across the development chain, positively impacting therapeutic development and market and patient access. This talk will include potential novel strategies for clinical trial design and real world data collection, alongside future possibilities for streamlining regulatory approvals and access.

14.50 **Panel: Opportunities for Acceleration and Collaboration Within the UK**




Dr Sara Wells MBE, Dr Peter Oliver, Matthew Durdy, Prof Pleasantine Mill (moderator)

This discussion will focus on how we align patient derived models, with progressive regulation and manufacturing, to build on the genetic and NAT capabilities in the UK to drive advanced therapy development worldwide.

15.10 **Genomics England Generation Study**

Dr David Bick
Principal Clinician for Newborn Genomes Programme, Genomics England

The Generation Study aims to recruit 100k newborn babies and sequence them for over 200 treatable genetic conditions.

Early genetic diagnosis and treatment has the potential to significantly improve the outcome for the children identified. Newborn genomic sequencing can eliminate the diagnostic odyssey for families, reduce the financial burden on DHSC and enabling cohorts for clinical trials.

1 OCT 2024
Francis Crick Institute
London | 9:00 - 19:00

15.25 

Questions: led by Emma Tinsley

15.35 **Networking break**

Session V: Getting New Therapies into Patients

Session Chair:
Dr Matthew Lumley, Medicxi

16.05 **Using Computational Modelling for Next Generation Clinical Trial Endpoints**

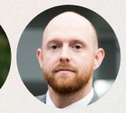
Dr Joe Jacob
Centre for Medical Image Computing, University College London

Biomarkers are critical to effective drug development but pose challenges in rare diseases due to limited data, patient numbers, heterogeneity of the population etc. For respiratory disease FEV1 is a well established endpoint, but is very noisy, and is slow to react to functional change.

Instead, computational models can harness the power of new digital endpoints. This will include success in CF and wider applicability.

16.20 **Panel: How Will We Achieve Trial-ready Cohorts of Patients?**



Dr Rick Thompson, Dr Lucy Allen, Dr John Matthews, Dr Matthew Lumley (moderator)

The panel will discuss how we can achieve pre-recruited cohorts to streamline clinical trials in rare disease. We will discuss ideas to engage and educate a diverse patient community with regards to research, consent and engaging with difficult to reach communities.

Agenda

16.50 **Horizon Scanning For Orphan Assets**

Steve Robery
Strategy Director, Weatherden

The orphan market is worth $150B and set to double in the next 5 years. However, only 5% of rare diseases have an approved therapy. What does the future look like?

17.00 **Attracting Investment for Rare Diseases**

Emma Tinsely
CEO Weatherden

The success of clinical trials depends on more than biology. Clinical trial failure rates remain the single largest economic barrier to the development of many medicines and have remained stubbornly high at 90% over the last 20 years, despite the scientific advances made in that time.

The critical step remains early clinical development, often termed the “valley of death” for drug development. Shared commonalities provide an avenue to drive development efficiency to improve value.

17.20 **Closing Remarks**

Prof Kev Dhaliwal
Chair of Molecular Imaging & Healthcare Technology / Consultant Physician in Respiratory Medicine

17.30 **Networking Drinks Reception**

Speakers

Dr Harriet Holme MA MBBS PhD

Founder and Chair PCD Research
Drug Development Clinician Weatherden

After reading medicine at the University of Cambridge, Harriet worked in the NHS as an NIHR academic paediatrician in London for nearly a decade, ultimately specialising in paediatric oncology. Harriet completed her PhD on cancer drivers and potential novel therapeutic targets in osteosarcoma at UCL and the Institute of Cancer Research, in Professor Alan Ashworth’s laboratory. This included investigation of synthetic lethality using siRNA screening of the kinome, integration with proteomic and genetic data, together with creation of models of loss of function using gene editing (CRISPR-Cas9).

Harriet currently works as a Drug Development Clinician at Weatherden, assisting biopharm with strategy, commercial positioning, asset and indication selection, and clinical trial design with a special interest in oncology and advanced therapeutics. Harriet is passionate about improving the outcomes for people with rare diseases, in particular PCD, and ensuring access to disease modifying treatments, together with risk stratified, evidence-based care.

Dr Kath Bainbridge PhD

Head of Rare Diseases and Emerging Therapies, Department of Health and Social Care

Kath’s remit includes genomics science policy, sustainable policy on genetics and insurance, advanced therapies and rare diseases policy, including the UK Rare Diseases Framework. All these areas benefit from cross sector working with a wide range of stakeholders across the rare diseases community, delivery partners and research partners including the National Institute for Health Research (NIHR), to ensure an approach to evidence-based policy making in government with a strong focus on the political, ethical, social, legal and technological implications.

Kath has a PhD in genetics from the University of York and worked as a research scientist before joining the civil service.

Professor Carlos E. Milla, MD

Professor of Pediatrics and (by courtesy) of Medicine at Stanford University School of Medicine.

Associate Director for Translational Research at the Center for Excellence in Pulmonary Biology at Stanford.

Director of the Stanford Cystic Fibrosis Center, the Stanford Primary Ciliary Dyskinesia Center, and the Stanford CF Translational Therapeutics Development research program.

Dr Milla has actively participated in multiple clinical research studies and has accumulated substantial experience on the diagnosis and development of novel outcomes for paediatric pulmonary disorders. In addition, Dr Milla has long-standing expertise in drug development through a large number of clinical trials, from early phase to pivotal trials, as well as participating in multiple advisory boards for drug development focused on CF, PCD and other airway disorders. This includes holding several patents and being a corporate board member of startup biotech companies with a focus on pulmonary disorders. Dr Milla has published and lectured extensively on the topics of cystic fibrosis and PCD, and the genetics of rare lung diseases. Current areas of research include early lung disease development and the pathophysiologic mechanisms involved in the defective mucociliary clearance characteristic of CF, PCD and bronchiectasis. Additional research interests include active programs for remote monitoring and biomarker discovery for chronic pulmonary conditions.

Speakers

Professor Steven Brody

Professor of Pulmonary Medicine and physician-scientist at Washington University in Saint Louis.

Steven's laboratory group investigates fundamental aspects of airway epithelial cell biology and the impact of genetic and acquired diseases on the airway. His focus on the mechanisms for control of multiciliated cell differentiation and assembly of motile cilia has uncovered altered pathways in primary ciliary dyskinesia. Steven's laboratory leverages multidisciplinary and collaborative approaches for the establishment of platforms to investigate future therapies for primary ciliary dyskinesia.

Dr Sam Barrell MD

Chief Executive Officer, LifeArc

Dr Sam Barrell CBE is the recently appointed Chief Executive Officer for LifeArc, a self-funded, organisation which bridges the gap between academic research and unmet patient need, the availability of diagnostics, novel solutions and therapies for some of the most under-served conditions: motor neuron disease, chronic respiratory infection, global health, rare disease, and childhood cancer. Sam takes up this position on 1st October.

Between September 2017 and September 2024, Sam was the Deputy Chief Executive Officer of the Francis Crick Institute, one of Europe's largest biomedical research institutes. There she led on strategic and operational management of the institute and deputised for the Director, Sir Paul Nurse.

A GP by background, Sam joined the Crick from a career in the NHS as a notable healthcare leader. She was Chief Executive of the Taunton and Somerset NHS Foundation Trust and, prior to that, Chief Clinical Officer for the South Devon and Torbay Clinical Commissioning Group.

Sam is a member of the British Land Innovation Advisory Council and a Non-Executive Director of Assura plc, a large property company specialising in healthcare premises, and the York Health Economics Consortium (YHEC).

Sam was awarded the CBE in 2014 for services to healthcare and was named one of the 50 most inspirational people in healthcare by the Health Service Journal.

Professor James Chalmers MD PhD

Asthma and Lung UK Chair of Respiratory Research at the University of Dundee.

Chair of the European Bronchiectasis Network (EMBARC)

James holds a number of leadership positions with international respiratory societies including chair of the science and research committee of the British Thoracic Society and Chief Editor of the European Respiratory Journal. He runs a large translational research group and has published over 450 peer reviewed papers on respiratory infections, immunology and disease.

Professor Amelia Shoemark

Asthma and Lung UK Chair of Respiratory Research at the University of Dundee.

Chair of the European Respiratory Society Clinical Research collaboration BEAT-PCD Management committee of EMBARC European Bronchiectasis Registry

Amelia's translational research program aims to target ciliary dysfunction as a therapeutic strategy in chronic lung disease. She completed her PhD in respiratory medicine at the National, Heart and Lung Institute, Imperial College London. Her PhD work investigated inflammation in the lungs of patients with the chronic respiratory condition bronchiectasis helping to define causes of the disease (http://www.nhs.uk/conditions/bronchiectasis/).

In her role as a clinical scientist at the Royal Brompton Hospital Amelia conducts diagnostic testing for primary ciliary dyskinesia (PCD) and has been involved in discovery of several PCD genes as leading a program of work to improve diagnostic testing. Amelia has authored 100 scientific articles on PCD and bronchiectasis.

Professor Jane Davies MD OBE

Professor in Paediatric Respirology & Experimental Medicine at the National Heart and Lung Institute.

Honorary Consultant in Paediatric Respiratory Medicine at the Royal Brompton & Harefield NHS Foundation Trust.

Jane has had a long career in cystic fibrosis - as a global lead investigator on CFTR modulator trials, as clinical lead and strategy group member in the UK CF Gene Therapy Consortium, establishing and directing the Lung Clearance Core Facility on behalf of the European CF Society, leading the UK NIHR CF National Research Strategy Group and most recently, as President of the European CF Society

Professor Davies graduated MB ChB from the University of Dundee Medical School in 1987 and undertook clinical training in Paediatrics in London. A post at the Royal Brompton Hospital led to an interest in cystic fibrosis (CF), particularly host-pathogen interactions and her MD, under the supervision of Andy Bush and Eric Alton, focussed on the pathogenesis of Pseudomonas aeruginosa in the lung. Her clinical training was subsequently completed at Great Ormond Street Hospital in the departments of Respiratory Medicine and Infectious Diseases/ Immunology. She returned to the NHLI in 1999, initially as Senior Lecturer, promoted to Reader in 2009 and Professor in 2013. She is based in the Dept of Gene Therapy, led by Professor Eric Alton, with whom she collaborates closely.

Professor Davies remains engaged in translational research in the field of CF. She is the clinical lead and Strategy Group member of the UK CF Gene Therapy Consortium; this Consortium was established in 2002 from 3 groups in the UK with CF gene therapy clinical trials experience (Imperial College/ Royal Brompton Hospital, Oxford University and Edinburgh University). The aim is to develop clinically relevant CFTR gene therapy. On the consortium trials she conducts the bronchoscopic assays and leads on the electrophysiological measurements (potential difference) both in the nose and lower airway.She is also heavily involved in clinical trial design and the validation of outcome measures, leading the Core Lung Clearance Index facility for the European CF Society Clinical Trials Network. She works closely with Prof Diana Bilton, the Adult CF clinical lead, on a number of commercially-sponsored clinical trials in CF, largely involving novel small-molecules directed at CFTR function.

Speakers

Professor Thomas Ferkol MD

Professor of Pediatrics at the University of North Carolina at Chapel Hill. American Lung Association Edward Livingston Trudeau Scholar and recipient of the Primary Ciliary Dyskinesia Foundation Golden Cilium Award.

Prof Ferkol's' research has largely focused on the development of cell and animal models to study inherited, suppurative lung diseases, defining genetic and molecular factors that contribute to chronic airway infection, inflammation, and epithelial injury with almost 30-years of continuous funding from the National Institutes of Health (NIH).

Thomas co-leads the NIH-supported Genetic Disorders of Mucociliary Clearance Consortium, an established, multicenter collaborative that is defining the genetics, pathophysiology, and clinical features of primary ciliary dyskinesia. He has served on numerous international review groups and study sections, and written or co-authored over 200 original articles, scholarly reviews, and book chapters. Prof Ferkoll was previously the President of the American Thoracic Society, only the second paediatrician to serve in this capacity during the 120-year history of the organisation.

Professor Pleasantine Mill PhD

MRC Investigator at the MRC Human Genetics Unit at the University of Edinburgh
Co-Lead LifeArc Centre for Rare Respiratory Diseases
Co-Lead MRC NMGN Congenital Anomalies Cluster
Co-lead NIHR/MRC Rare Disease Research Node CILIAREN

Pleasantine leads a programme to understand genetic disease and disease mechanisms arising from dysfunction of mammalian cilia, called the ciliopathies. With 20 years of expertise in developmental genetics and cell biology, her work spans from forward genetics screens through to candidate discovery in human disease genetics. Her lab focuses on phenotype-driven projects that disrupt cilia structure and/or function to uncover underlying genetic changes, understand disease mechanisms and move towards much needed therapeutics for rare disease.

Pleasantine's work is funded by the UK Medical Research Council, NIHR, LifeArc and the European Research Council. She sits on the Leadership Team for the UK Cilia Network, and is co-lead for the MRC/NIHR Rare Disease Platform CILIAREN, the MRC NMGN Congenital Anomalies Cluster and the LifeArc Translational Centre for Rare Respiratory Diseases.

Professor Emma Rawlins PhD

Professor of Developmental Physiology in a joint appointment with the Department of Physiology and the Department of Development and Neuroscience, University of Cambridge, UK.

Emma Rawlins obtained her PhD in developmental biology from the University of Edinburgh in 2002. For her PhD she worked with Dr Andrew Jarman on cell fate specification in the developing Drosophila PNS. She performed postdoctoral work with Prof Brigid Hogan at Duke University from 2004 – 2009 where she identified stem cell populations in the developing, homeostatic and repairing mouse lungs. In 2009 she started her lab at the Gurdon Institute, University of Cambridge. In 2024 she was promoted to Professor and her lab works on lung stem and progenitor cell biology, combining innovative human organoid models with mouse genetics.

Dr Sara Wells PhD MBE

Director of the Mary Lyon Centre, MRC Harwell
Chief Biological Research Facility (BRF) Officer at the Crick

Sara has an academic background in genetics and since joining MRC Harwell in 2002, she has worked across biomedical fields, advancing best practices in animal research and enabling great science through the provision of the highest standards of services for researchers.

Sara's outstanding track record in supporting and delivering complex research projects involving in vivo studies, led to her being appointed as the Crick's Chief BRF Officer in early 2024. In this role, Sara provides valuable support to the Crick's leadership team on strategic and practical decisions regarding the BRF and animal research.

Matthew Durdy

Chief Executive Officer, Cell and Gene Therapy Catapult (CGTC)

Matthew was one of the founding team in 2012 of the CGTC. Prior to 2020, he was Chief Business Officer and was responsible for strategy, communications, marketing, and business development. He is credited with leading the design and implementation of the commercial model for the highly successful CGTC manufacturing centre and being a global champion for the early integration of healthcare economics and reimbursement expertise into decision-making and clinical product design. He began his career in international investment banking and venture capital, and has successfully invested in and managed a number of biotechnology SMEs and regional operations of multinational organisations. He has an MA from the University of Oxford in Pure and Applied Biology, an MBA from the University of Chicago, and is a Fellow of the Chartered Institute for Securities and Investment. He is also Chair of Parkinsons Research Ventures Ltd and Vice Chair of the Board of Trustees of the charity Parkinson's UK.

Dr Joseph Jacob MD

Centre for Medical Image Computing, University College London, London, UK.

Joseph is a recipient of successive Wellcome Trust Research Fellowships, with the most recent Fellowship awarded in October 2023. He leads the Satsuma Lab at the Centre for Medical Image Computing, UCL which performs computer-analysis of clinical CT imaging in chronic lung diseases and lung cancer. The Satsuma lab aims to develop imaging biomarkers of globally important lung diseases to accelerate their early prognostication and in turn, allow an increased focus on managing early-stage modifiable respiratory disease.

Dr Kirsty Wydenbach

Head of Regulatory Strategy, Weatherden

Dr Kirsty Wydenbach is a renowned regulatory expert and pharmaceutical physician who leads the strategy division at Weatherden, a global integrated clinical consultancy. Kirsty joined Weatherden from the MHRA where she was an expert medical assessor in the Clinical Trials Unit for more than a decade, with a particular interest in advanced therapies, first-in-human trials and novel trial designs. She currently chairs the Clinical Trial Science and Regulations Expert Group of the Faculty of Pharmaceutical Medicine, is an expert on the Scientific Advisory Board of CRO Simbec-Orion, a committee member of the Association for Human Pharmacology in the Pharmaceutical Industry (AHPPI) and a visiting lecturer for King's College London.

Speakers

Dr Dan O'Connor MD PhD

Director of Regulatory and Early Access Policy at The Association of the British Pharmaceutical Industry (ABPI).

Editor-Author of the Oxford Specialist Handbook in Pharmaceutical Medicine.

Dan joined the ABPI from the Medicines and Healthcare products Regulatory Agency in 2023. At the MHRA he was Deputy Director of the Innovation Accelerator and Regulatory Science. Dan has special interests in drug development, rare diseases, regulatory science, health innovation, patient engagement and drug repurposing. He completed higher medical training in Pharmaceutical Medicine.

Dr David Bick MD PhD

Principal Clinician for the Newborn Genomes Programme at Genomics England

Prior to his work in England, Dr Bick was the Chief Medical Officer and a faculty investigator at the HudsonAlpha Institute for Biotechnology. Dr Bick also served as the Medical Director of the Smith Family Clinic for Genomic Medicine at HudsonAlpha and the Laboratory Director of the HudsonAlpha Clinical Services Laboratory. He came to HudsonAlpha from the Medical College of Wisconsin where he was Professor in the Department of Pediatrics and the Department of Obstetrics & Gynecology and Director of the Clinical Sequencing Laboratory, Director of the Advanced Genomics Laboratory, Medical Director of the Genetics Clinic at Children's Hospital of Wisconsin, and Chief of the Division of Genetics in the Department of Pediatrics at Medical College of Wisconsin. Dr Bick is board certified in Pediatrics, Clinical Genetics, and Clinical Molecular Genetics. Dr Bick has published numerous peer-reviewed articles, chapters, and reviews. His laboratories at the Medical College of Wisconsin and Children's Hospital of Wisconsin were the first in the world to offer whole genome sequencing as a clinical test. He also developed the first Genomic Medicine Clinic in the United States.

Dr Peter Oliver PhD

Head of Biology, Nucleic Acid Therapeutic Accelerator (NATA)

Pete has over 25 years' experience in cell and molecular biology. After a degree in Biochemistry and a PhD in Genetics, he led projects studying new models of neurological disorders at the University of Oxford, before becoming a group leader at the Department of Physiology, Anatomy and Genetics in 2012 funded by an ERC Consolidator Award. His group's work in rare disease modelling using the mouse resulted in a new position as an MRC Programme Leader at the MRC Mammalian Genetics Unit in 2017. Pete joined NATA in February 2022, bringing an understanding of multi-disciplinary approaches to translational science; from biochemistry and cell biology to the application of preclinical genetic tools. Pete remains a visiting academic member of the University of Oxford and is an active co-investigator in the MRC National Mouse Genetics Network.

Dr Lucy Allen PhD

Director of Research and Healthcare Data

Lucy Allen joined the Cystic Fibrosis Trust in late 2019. Lucy's previous experience includes building and leading research collaborations with industry, other research charities and clinical academics across a wide range of disease areas such as respiratory, mental health, and cancer and nutrition for the NIHR. She has also led R&D projects for GE Healthcare developing radiopharmaceuticals for cancer and Alzheimer's disease. Lucy also has a PhD in lung inflammation and infection.

Dr Matthew Lumley MD PhD

CEO Myosana Therapeutics

Venture Partner at Medicxi

Matthew is a clinician scientist with 10 years of experience in academic medicine and 8 years in drug development. He was previously global clinical lead for Moderna's first mRNA therapeutics in Rare Disease and has developed expertise in translational medicine and early clinical development in complex paediatric diseases. Previously he has held roles in medical affairs and market access roles at Pfizer.w

Dr John G. Matthews MBBS MRCP PhD

Chief Medical Officer, Recode Therapeutics

Dr. Matthews brings over twenty years of proven leadership in clinical trial design and drug development, and extensive patient care experience. He joined ReCode from 23andMe, where he was the senior clinical development lead for rare disease therapeutic strategy. While there, he provided leadership to drug development programs and phenotype working groups aimed at discovering novel genetic targets.

Prior to 23andMe, Dr. Matthews served as vice president of clinical development at Aimmune Therapeutics supporting late and early-stage programs. He spent the previous ten years at Genentech, a subsidiary of Roche, in leadership roles including senior group medical director. Before Genentech, Dr. Matthews held positions of increasing responsibility at Novartis Pharmaceuticals and was a clinical scientist in the department of early development for respiratory and inflammation products at GlaxoSmithKline, where he worked on a novel inhaled steroid among other molecules.

Dr. Matthews trained in pulmonary medicine at the Royal Brompton Hospital and the Lane Fox Respiratory Unit, St Thomas' Hospital, London. He earned his Ph.D. in respiratory clinical pharmacology from Imperial College London. He is a member of the Unbiased BIOmarkers for the Prediction of Respiratory Disease Outcomes (U-BIOPRED) program and serves as executive director of RASP-UK, a large-scale research project that aims to target treatments effectively in patients with severe asthma.

Dr Rick Thompson PhD

CEO, Beacon

Rick joined Beacon (previously known as Findacure) as the charity's third member of staff and first-ever Scientific Officer. His aim was to drive forward the charity's work in drug repurposing.

Rick became CEO in 2017, and has since been involved in almost all of Beacon's projects, driving the organisation's growth and strategy

Before Beacon, Rick secured a PhD in Evolutionary biology and studied moles in Cambridge's Museum of Zoology. He has written articles, given talks and provided training across the European rare disease community. He is continually amazed by the work, knowledge and commitment of our patient group leaders.

Steven Robery PhD

Strategy Director, Weatherden

Steven is a strategy and business development executive, who has helped to support decision making at both an R&D and Corporate level across both Pharma and Biotech. Steven joined Weatherden after spending 10 years working in roles in biotech, GSK and life science consulting, where he has had various roles across business development as well as R&D and commercial strategy. Steven also holds a PhD in Molecular Biology from Royal Holloway University of London.

1 OCT 2024

Francis Crick Institute

London | 9:00 - 19:00

Speakers

Emma Tinsley

CEO Weatherden

Emma is a leading biotech executive and the CEO of Weatherden, a global integrated clinical consultancy and expert in scientific strategy and clinical development. Emma has deep experience of company formation, clinical development strategy, operating model transformation and investment management and was previously at SV Health Investors in the biotech company creation team creating immunology and oncology companies; and prior to this, in the Life Sciences team at KPMG.

Professor Kev Dhaliwal

Kev Dhaliwal is Professor of Molecular Imaging and Healthcare Technology at the University of Edinburgh and a Consultant Physician in Respiratory Medicine.

Interim Director for Edinburgh Futures Institut.

Lead LifeArc Centre for Rare Respiratory Diseases

Kev's research and innovation activities focus on developing new technologies spanning the 4Ds in health research, data, diagnostics, devices and drugs. His clinical practice is in the New Royal Infirmary of Edinburgh, and the newly established Healthcare Technology Accelerator Facility whose aim is to expedite the clinical and commercial translation of healthcare technologies spanning all specialities. He leads the Translational Healthcare Technologies Group at the University of Edinburgh which brings together basic and translational teams to drive new futuristic approaches for human disease.

Notes

1 OCT 2024

Francis Crick Institute
London | 9:00 - 19:00

Sponsors

We extend our deepest gratitude to our major sponsors, whose generous contributions have made this event possible.

LifeArc Centre for Rare Respiratory Diseases

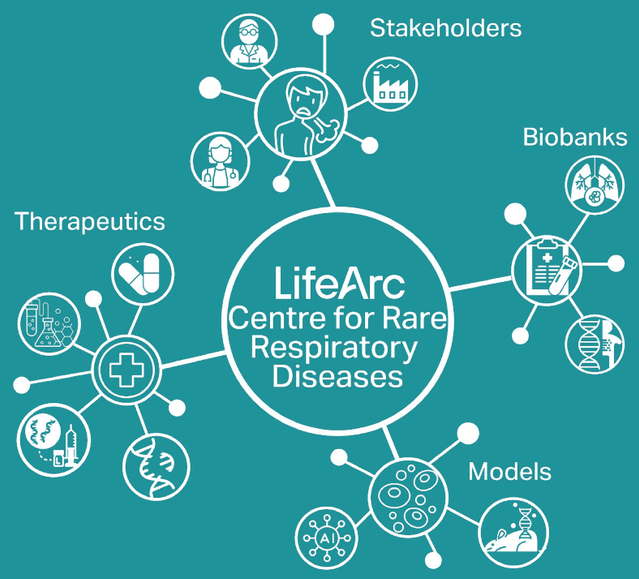
Respiratory health research in general is under-resourced within public funding priorities receiving less than 2% of public funding for health research. Similarly, within the rare disease space, childhood and adult rare respiratory diseases (RRDs) are overlooked, most are poorly characterised and there are few approved therapies, however the burden for patients is significant with high levels of mortality and morbidity. Only 4% (11/277) of European Medicines Agency orphan disease marketing authorisations are for RRD products, with none in children, despite childhood rare diseases representing three quarters of all approved indications.

Launched in September 2024, the LifeArc Centre for Rare Respiratory Diseases marks ~ £15M investment by LifeArc and its University partners to address the huge unmet need, to improve lives of RRD patients. It looks to connect work in the RRD space that has remained fragmented and accelerate funding for disease-modifying research.

The Centre assembles expertise to develop new therapies for RRDs, providing a sustainable and inclusive "go-to" entry point, with 11 regional clinical sites, to connect patients and carers with clinical experts, researchers, investors and industry across the UK. Our Centre will create a UK-wide infrastructure for patient registration and biobanking, discover and advance pioneering therapies, and engage with industry, investors and regulatory partners to 'fast-track' derisked clinical trials in the RRD space. Together, we will work to remove roadblocks to translation to the clinic for RRDs.

We look to boost the capacity and reputation of UK translational medicine, public awareness of the realities of living with RRDs, and patient awareness of resources that can improve their quality of life.

https://www.lifearc.org/project/lifearc-translational-centres-for-rare-diseases/



We would also like to express our heartfelt thanks to our supporting sponsors. Your contributions are invaluable in helping us achieve our goals.