RARE DISEASE RESEARCH UK

NEWSLETTER

A forum to illuminate the latest advancements within the rare disease research community.

Rare Disease Research UK (RDR UK) is a new £14 milliion platform to advance research into rare diseases. Jointly funded by MRC and NIHR, RDR UK is made of 11 research nodes - involved in both disease-area-specific and cross-cutting research, engaging leading universities across the country - and a coordinating hub, hosted by Newcastle University, Newcastle upon Tyne Hospitals NHS Foundation Trust and Genetic Alliance UK.

As a UK-wide platform, we hope to be able to significanty impact the rare disease research landscape and imporve the lives of those directly or indirectly affected by rare diseases.

We're thrilled to announce our very first edition of our quarterly newsletters, packed with insights, activities and updates from across the RDR UK Platform that we're eager to share with you.

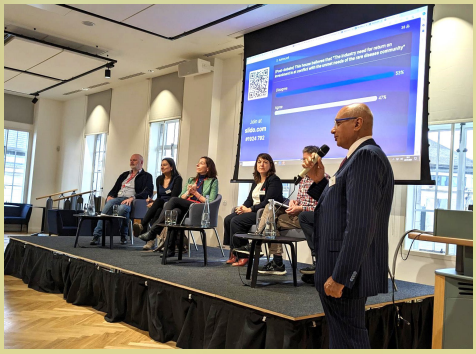
With each edition, we aim to illuminate the latest advancements within the rare disease community, share compelling stories, and foster collaboration among researchers, industries, and patients.

HIGHLIGHTS

CILIAREN Node Workshop
09 Feb 2024

Rare Disease Day 2024
29 Feb 2024

RDR UK 1st Annual Conference
26 March 2024



mTOR Pathway Diseases Node 2024 Symposium
24 April 2024

NEWS FROM OUR NODES

Check out our website to find out the latest happenings within our nodes!

PATIENT VOICE

The Platform, in partnership with Genetic Alliance UK is delivering a program of work to support good PPIE in rare disease research.

Hear from Lynn Laidlaw, Patient Contributor and Researcher

'A four-year diagnostic odyssey before being diagnosed with a rare auto immune disease prompted my interest in Patient and Public Involvement (PPI) in research, so I was chuffed to be invited to become a panel member for a session discussing PPI at the recent Rare Disease Research UK Conference in Newcastle.

I have had the privilege of working with multiple academics, institutions and organisations, as well as co-producing research and working as a patient/peer researcher. In the session I highlighted that, for me, PPI is built on developing trusting relationships, having honest conversations and meaningful collaboration. It's as much as state of "being", working in a values and principles led way, as "doing"- the methods used to involve people. There is emotional labour involved for both researchers and public contributors and it can impact on people's identity. We become involved because of our lived experience; our aim is to help the research process to ensure research meets people's needs. If the people involved in research didn't feel that their unique experiences and expertise could help researchers, they wouldn't waste their time as research is too important. PPI can't thrive without reflexivity, that is understanding how our life experiences, assumptions, biases and values can impact on the way we work together. It requires us to embrace "productive tensions" and have an understanding of how academic culture and power imbalances can impact on the way we work together in partnership.

I have embraced the joys and challenges of PPI because fundamentally research is hope to people like me who live with rare, poorly understood and treated diseases.'

Reach out to hub@rd-research.org.uk if you are interested to know more about our PPIE activities.

EVENTS

Resources and Funding Opportunities
06 June 2024, 9:30 - 18:00
Advance Training Centre, MRC Harwell

This event aims to provide valuable insights into resources available in the UK and funding opportunities for rare disease research in the UK and Europe. We want to bring various stakeholders from academia and industry together and discuss how to overcome the challenges in public-private sector collaborations in rare disease research.

The UK Platform of Nucleic Acid Therapy for Rare Diseases (UPNAT) Node inaugural symposium
27 June 2024, 10:0 - 17:00
UCL Institute of Child Health, London

The symposium will explore
1. Target selection and pre-clinical development,
2. Tolerability, toxicity and scale-up synthesis of NATs, and
3. Clinical trials and regulatory approval process.

The agenda will include, among others, presentations by:
- Piotr Kosla, Biogen (also parent of a patient, and Simons Searchlight Community Advisory Committee member)
- Sara Tabrizi, UCL
- Ana Lisa Tavares, Genomics England
- Willeke van Roon-Mom, Leiden University
- Daniel O'Connor, the Association of the British Pharmaceutical Industry

We hope to see many of you there. Register now!
For queries, contact UPNAT@ucl.ac.uk.

1st Scientific Meeting of the MRC Rare Early Onset Lower Urinary Tract Disorder (REOLUT) Node

27 June 2024, 14:00 - 16:45

UCL Great Ormond Street Institute of Child Health, London

This half-day meeting will comprise: an overview of the REOLUT Node, short presentations of ongoing and planned genomic, biology and novel therapy research, and a PPIE session. There will also be a keynote lecture presented by Dr Alina Hilger from the Research Centre on Rare Kidney Diseases, University Hospital, Erlangen, Germany on “Genetics of lower urinary tract disorders”. Register now!

24th European Nephrogenesis Workshop

28 June 2024

UCL Institute of Child Health, London

The Workshop hosted by REOLUT Node Co-I, David Long' provides an excellent forum for young investigators (PhD students and post-docs) to present their on-going work in a friendly environment. Primarily focused on kidney development, the workshop has expanded in recent years to include researchers working in the stem cell field, vascular biology, models of renal disease and novel therapeutic interventions. Register now!

ExPRESS Pilot Study Meeting - London 2024

July

UCL Queen Square Institute of Neurology, London

The meet will bring all the UK Node PIs and study team members together, to review and discuss the pilot phase of the ExPRESS study. Stay tuned to our webiste for more details.

Reach out to hub@rd-research.org.uk for queries.

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SUBSCRIBE

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UK Platform for Nucleic Acid Therapies - UPNAT

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Ethical Legal and Social Issues in Rare Conditions Research and Clinical Practice - ELSI

Rare Disease Research UK.

Renal Ciliopathies National Network - CILIAREN

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Rare Early Onset Lower Urinary Tract Disorders - REOLUT

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mTOR Pathway Diseases

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Cardiovascular Initiative

Rare Disease Research UK.

Lipidomics and Metabolomics for Rare Disease Diagnosis

Rare Disease Research UK.

Epigenomics of Rare Disorders - EpiGenRare

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Parkinson Plus Syndromes - ExPRESS

Rare Disease Research UK.

HistioNode: Histiocytic Neoplasms and HLH

Rare Disease Research UK.

Changing Clinical Practice Through Innovative Trial Designs – CAPTIVATE

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