

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 £14 million platform to advance research into rare diseases, jointly funded by MRC and NIHR. The platform is made of 11 research nodes, led by various universities across the country, involved in both disease-area-specific and cross-cutting research, and a coordinating hub - hosted by Newcastle University, Newcastle upon Tyne Hospitals NHS Foundation Trust and Genetic Alliance UK, to connect the nodes and provide a well signposted route into the platform for external collaborators, including patients, researchers, industry and international partners.

We're thrilled to share the fifth 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.

NEWS

UPNAT Symposium - 2025

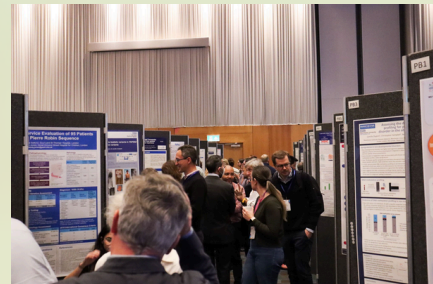


The UPNAT Node recently hosted their 2nd Annual Symposium, with a focus on personalised therapies. Over 150 attendees were welcomed from a wide range of multi-disciplinary background, across academia, charity, industry, NHS, DHSC, regulators, and manufacturing. Invited speakers reflected UPNAT engagement both internationally, with Dr Alban Ziegler (University Hospital of Toulouse) and Dr Marlen Lauffer (Dutch Center for RNA Therapeutics), and on a national level, with Prof Matt Brown (Genomics England), Dr Kath Bainbridge (DHSC), Dr Ella Whittle (UCL) and Dr Amy McTague (GOSH). Across industry and manufacturing, we heard from with Rachel Smith (Parexel), Jeff Milton (La Jolla Labs), Claire MacLeod (Centre for Process Innovation), Dr Andrew Roddam (EveryOne Medicines), and Assoc Prof Peter Oliver (NATA). Claire Andersen provided important PPIE perspective with an update from Unique (Understanding Rare Chromosome and Gene Disorders).

Details about the event, along with full speaker line-up and presentation topics, can be found [here](#). A report of the Symposium is being drafted, so do reach out to upnat@rd-research.org.uk if you were unable to attend or would be interested in hearing more about other UPNAT events.

EpiGenRare Symposium - 2025

The EpiGenRare Symposium was an integral part of the Manchester Dysmorphology and Developmental Disorders Conference (MDC 2025). A multi-day conference held in Manchester between the 16th - 19th November 2025. This high profile specialist conference attracted an international audience of over 300+ delegates congregating together from over 30+ different countries. Chaired by Prof Siddharth Banka (The University of Manchester) and Dr Cristina Dias (King's College London) of the EpiGenRare Node, the EpiGenRare Symposium was an integral part of the conference which featured numerous oral presentations on epigenetic research and began the day's proceedings on Monday 17th November receiving great interest and engagement from the live audience.



Workshop: ELSI Barriers to Mainstreaming

The workshop focused on the 'Barriers to Mainstreaming Genomics' research project which tracks the process of making genomic testing a normal part of mainstream medicine. The workshop presented research data gathered over the past year using a combination of presentations and workshops. The workshop was well attended and gathered feedback on the ELSI Node's initial ideas on research findings and made headway into identifying any gaps or inconsistencies in our analyses to incorporate and take forward into 2026.

NEWS

RDR UK Hub Team at the CamRARE Fest

Our co-lead Vicki Hedley and senior PM Anna Halstead attended RAREsummit 2025, where they had an engaging day filled with inspiring stories. They also caught up with friends from across the rare disease community and connected with others working in the field.



Workshop: UK Regulatory and Access Frameworks in Rare Disease – Current status, opportunities and challenges

The RDR UK Hub ran a regulatory workshop on 19 and 20 November, alongside our CAPTIVATE and UPNAT nodes, the LifeArc Centre for Acceleration of Rare Disease Trials (ARDT), and the Rare Therapies Launch Pad (RTLTP).

The goal of the workshop was

- To bring together a broad range of stakeholders to better understand the goals and plans of key projects and structures working on this topic
- To share and reflect on recent survey data on the status quo of regulatory processes and access in the UK, relevant to rare disease (RD)
- To discuss and build consensus, at a high level, on a range of relevant topics under the broad heading of 'regulatory and access processes and frameworks'
- To develop and clarify current and future collaborative plans for further work or workshops to advance the more specific topics and questions under this broad area (in light of MHRA and other bodies' activities ongoing in this space).



BOOK YOUR CALENDAR

3rd Annual Rare Disease Research UK Conference

16 April 2025

The Birmingham Conference and Events Centre, B5 4E



Take a look at our node showcase sessions, where our nodes shared their progress and updates from the past year.

[Node Showcase Session I](#)

[Node Showcase Session II](#)

Reach out to us at hub@rd-research.org.uk if you are interested in getting involved in our PPIE activities.

If this newsletter has been forwarded to you, and you'd like to register to receive it directly, please subscribe to our mailing list below.

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Rare Disease Research UK.

UK Platform for Nucleic Acid Therapies - UPNAT

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

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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

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Lipidomics and Metabolomics for Rare Disease Diagnosis

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Epigenomics of Rare Disorders - EpiGenRare

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Parkinson Plus Syndromes - ExPRESS

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HistioNode: Histiocytic Neoplasms and HLH

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Changing Clinical Practice Through Innovative Trial Designs - CAPTIVATE

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