RARE DISEASE RESEARCH UK

NEWSLETTER

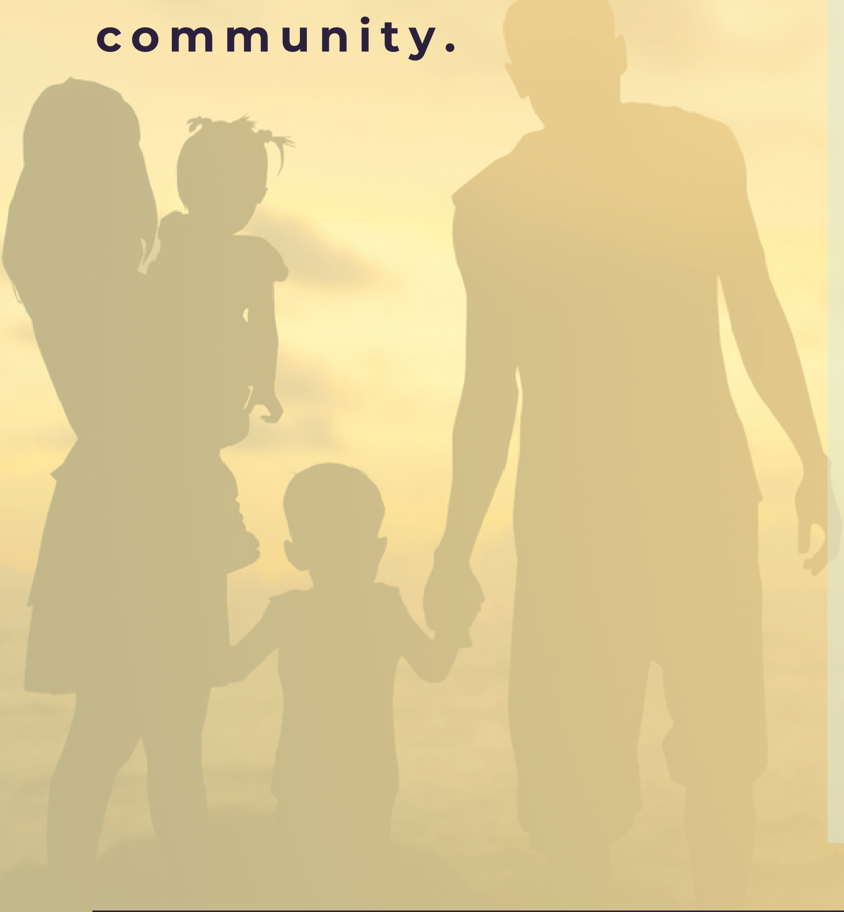
A forum to illuminate the latest advancements within the rare disease research community.

Rare Disease Research UK (RDR UK) is a £14 million platform to advance research into rare diseases. Jointly funded by MRC and NIHR, 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 significantly impact the rare disease research landscape and **improve** the lives of those directly or indirectly affected by rare diseases.

We're thrilled to share the fourth 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.

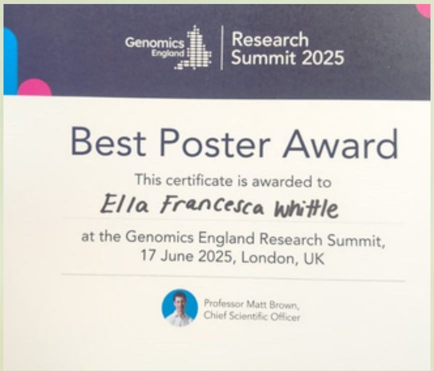


Rare Disease
Research UK.

Issue 4 August 2025

NEWS

UPNAT Wins Best Poster Award at Genomics England Research Summit



Genomics England | Research Summit 2025

Best Poster Award

This certificate is awarded to

Ella Francesca Whittle

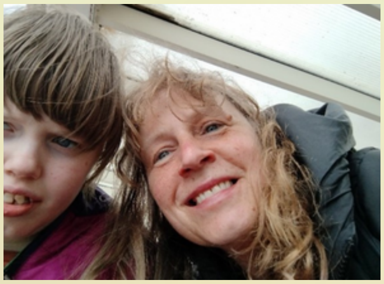
at the Genomics England Research Summit,
17 June 2025, London, UK

Professor Matt Brown,
Chief Scientific Officer

Congratulations to Dr Ella Whittle for receiving the Best Poster Award at the Genomics England Summit 2025. Ella, who is a Research Fellow for the UK Platform for Nucleic Acid Therapies (UPNAT) Node, presented a poster titled “Developing Target Selection Guidelines for Nucleic Acid Therapies". Find out more...

EpiGenRare Node supports Manchester discovery of 2 new genetic disorders that will improve diagnoses for patients with neurodevelopmental conditions

Discussing the next steps for the research, Professor Banka, who is also the Rare Conditions Co-Theme Lead at the NIHR Manchester BRC and Co-Lead for the EpiGenRare Node, said “We now want to understand these conditions better, such as how do their symptoms evolve over time, are there any associated complications and discover potential treatment options. We also need to understand the mechanism of the disease better – how exactly are these genetic mutations causing neurodevelopmental conditions? With this information, we hope to identify treatments and therapies, and make these available for patients.” Find out more...



NEWS

mTOR Node at Tuberous Sclerosis Association (TSA) Big Day event

Leila Ben-Chaabane, the Research Assistant for the mTOR Pathway Diseases Node, was delighted to speak at the recent Tuberous Sclerosis Association (TSA) Big Day, where she shared an update on the world's first mTOR pathway disease registry. The event was a fantastic opportunity to connect with patients, families, clinicians, academics and the wider TS community — full of powerful conversations and shared experiences. We're incredibly grateful to the TSA and all those who welcomed Leila so warmly.

A Day of Discussion: ELSI Conference 2025

What really matters in rare condition clinical trials?
The RDRUK ELSI Node hosted their first conference early this year, bringing together patients, families, researchers, and healthcare professionals in rare disease for a day of important discussion, shared experiences, and honest opinions. Check out their article capturing the voices and priorities shared on the day.

Rare Diseases Trial Collaboration

The LifeArc Centre for Acceleration of Rare Disease Trials (ARDT), is building a UK-wide infrastructure to make delivering rare disease trials faster, more efficient, and more patient-centred. Whether you're an academic researcher, clinician, patient organisation or company, their expert trials team can support you to design, set up, and run meaningful studies that make a difference.

Find out how you can collaborate with them on your next rare disease trial.

EARLY CAREER RESEARCHER AWARD IN PPIE - 2025

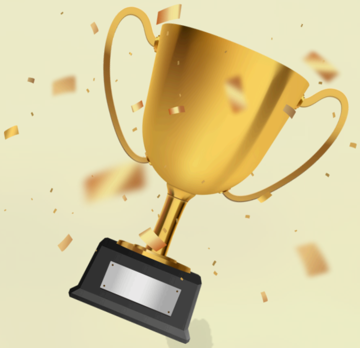
Are you an ECR working with the rare disease community?
Tell us about your PPIE!

This is an exciting opportunity for early career researchers in rare disease and their public partners to celebrate and raise the profile of their PPIE collaboration!

RDR UK in association with Genetic Alliance UK is launching the second round of the annual award for successful PPIE

- **Open to all UK-based ECRs (including clinician scientists)**
- **Must apply with a key PPIE contributor**

PRIZE INCLUDES



- Funding to attend the 2026 Annual RDR UK Conference with a PPIE contributor
- A live award ceremony
- A blog feature on the RDR UK and Genetic Alliance UK websites.

FIND OUT MORE & APPLY NOW!

DEADLINE

16 NOVEMBER 2025

We define early career researchers as post-doctoral researchers, including clinician scientists, who are establishing their career and have not yet applied for grants or fellowships as an independent PI.

GENETIC ALLIANCE UK

BOOK YOUR CALENDAR

Online course: The Power of Patient Advocacy in Genomics

September 2025

📍*Virtual*

This course has been specifically designed to inform and inspire anyone who is interested in learning more about the role that patient advocates can play in improving genomics research or medicine. It also encourages learners to reflect on any advocacy role they may be developing, and includes a peer-reviewed exercise to help them develop a plan to take this further.
Find out more and register.

20th Manchester Dysmorphology Conference

16 - 19 November 2025

📍*The Hilton, Deansgate, Manchester M34LQ*

Immerse yourself in the latest developments in rare disease research, connect with leading experts, and contribute to breakthroughs that shape the future of patient care. Find out more...

UPNAT Symposium

121 November 2025

📍*London*

Join UPNAT node as they host their first symposium.
Speakers:
- Professor Matt Brown (Genomics England, CEO)
- Dr Kath Bainbridge (DHSC)
- Dr Rachel Smith (Paraxel)
- Dr Marlen Lauffer (Dutch Center for RNA Therapeutics)
- Dr Alban Ziegler (University Hospital of Toulouse)

The Centre for Process Innovation (CPI) will introduce their UK-based Good Manufacturing Practice site of RNA Centre of Excellence. More details about the programme will be released closer to the time.

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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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

Rare Disease Research UK.
Rare Early Onset Lower Urinary Tract Disorders - REOLUT

Rare Disease Research UK.
mTOR Pathway Diseases

Rare Disease Research UK.
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

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HistioNode: Histiocytic Neoplasms and HLH

Rare Disease Research UK.
Changing Clinical Practice Through Innovative Trial Designs – CAPTIVATE

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