Project Title: A community approach to developing a rare disease network for patient-centred research

Authors
Dr Rona Smith, University of Cambridge
Mrs Jo Balfour, Cambridge Rare Disease Network
Mrs Laura Cowley, Patient Led Research Hub, University of Cambridge

Plain English Summary

This section of the report has been written with members of the public who have been involved in the research.

There are over 10,000 rare diseases, each affecting fewer than one in 2000 people. Most of these diseases cannot be cured and do not have an effective treatment. More than a third of people wait over 5 years for a diagnosis. For many people, living with a rare disease significantly impacts their quality of life, affects their mental wellbeing, and introduces immense healthcare needs.

Research is vital for many reasons, including:

- improving our understanding of rare diseases,
- achieving faster and more accurate diagnoses,
- developing new treatments and cures to improve physical and mental health,
- understanding patient experiences.

Most research is led by healthcare experts, and often asks questions that patients do not consider as important as researchers do. Many organisations are improving the way that researchers work with patients when designing new studies, but there remain few opportunities for patient communities to develop their own research ideas.

We invited over 20 adults living with rare diseases to create a new online platform where patients, healthcare providers, and researchers can share ideas and work together on new research projects. The platform, called the Rare Disease Research Network (RDRN), launched in November 2024. It provides training and guidance to help patients develop their own research ideas. It also helps to build new research teams by connecting people with similar research interests.

Further work is required to promote the RDRN to healthcare and research organisations. This will make sure that patient research ideas are supported by relevant experts and developed into high-quality studies. In the long-term, RDRN aims to run as a non-profit community-led platform.

Summary of research findings

Aims and Objectives

This project aimed to support the rare disease community (people living with a rare disease, their family members, carers, advocates, patient group representatives) build a network of partnerships and resources to facilitate patient-centred research with individuals affected by rare diseases. Objectives included

1. identifying partners and building a network;
2. conducting preliminary scoping activities to inform future research methodology;
3. developing and growing a sustainable network.

Methods

As an NIHR 'Public Partnership' Programme Development Grant, CamRARE and Patient Led Research Hub (PLRH) co-created an innovative collaborative platform, the Rare Disease Research Network (RDRN), with over 20 adult community members (people living with a rare disease, their family members, carers, and advocates). To recruit community members to the working group, the project was advertised widely via CamRARE, PLRH and partner organisations via social media and routine communications. Key collaborations with UK patient organisations included Beacon for Rare Diseases, Genetic Alliance UK, Rare QoL, Unique, Metabolic Support UK, RareMinds, Breaking Down Barriers, as well as numerous disease-specific patient support groups and internationally-based charities.

Any interested adult affiliated with the UK rare disease community was invited to submit an expression of interest form, sharing basic demographics and research experience. CamRARE and PLRH invited a diverse group (age, gender, experience) of 29 members to join and 24 remained active contributors throughout. This group co-created all aspects of the platform, meeting monthly (virtual or in-person) and working from home. Additional members, including those from outside the UK, were able to contribute via surveys, webinars and virtual meetings. Four rare disease researchers, selected because of their experience supporting patient-centred or patient-led research, also assisted with project development to ensure the platform would be relevant to non-community stakeholders. Activities and community communication were managed by an appointed RDRN PPI Coordinator with oversight from CamRARE and PLRH senior management.

Our Public Partnership 'research' plan was designed to be flexible and iterative, as is essential when using a coproduction approach. As a result, there were minor changes to the original plan to accommodate the community members' feedback and preferences, including:

- The plan for 3 working groups with 10 community members each, working in parallel to develop the platform instead transitioned into one large working group, allowing better continuity and flexible working. The community working group convened monthly and stayed connected between meetings through a WhatsApp group, optional virtual drop-in sessions, newsletters, and email exchanges. This group developed the platform, determining its user-interface, accessibility features, functionality, identifying and creating new resources, and identifying and engaging with the wider community and external stakeholders to develop a multi-stakeholder branding matrix.

- Smaller community task and finish groups were created to allow members with particular interests and skillsets to lead on outputs, including 1) resource and training development, 2) communication and marketing strategies, and 3) sustainable business management.
- The community's preference for informal peer support meant the mentorship program intended to operate during development will instead be developed after the platform is live.

All community members received NIHR-recommended remuneration of £25/hr for attending meetings and completing agreed tasks; meetings were scheduled at a time decided by a majority vote, with in-person, virtual, and independent working from home options available. All resources, including grant documents and finance, were securely shared with the community team to encourage transparency and to support collaborative working. Travel and accommodation were booked ahead of time by staff as requested, to ensure community members were not impacted financially. Remuneration was paid directly via BACS or donated to a charity on behalf of the individual as preferred. Advice for those in receipt of benefits was sensitively provided.

Website programmer, RedWeb, was appointed in January 2023 to create the online platform envisioned by the community using an iterative approach. Group members designed pages and key functionalities of the website, including: 1) the sitemap, 2) homepage and other public-facing pages, 3) creating a profile and sign-up process, 4) the members area and 'match-making' function, 5) the resources section. For each, the group set the meeting agenda, shared ideas through discussions, designed wireframes, and collected online resources and examples. Using this information, RedWeb built the RDRN platform in sections. Each section was in turn shared with the community for review and user-testing.

Results

This project aimed to support the rare disease community to build a network of partnerships and resources to facilitate patient-centred research with individuals affected by rare diseases. Now launched, RDRN aims to support patient-driven research by:

1. Providing resources and training to support individual patients and support groups conceive and develop their own healthcare research ideas;
2. Showcasing patient-led research ideas and building relevant research teams;
3. Match-making members based on research interests.

Key objectives with the relevant outputs are listed below:

<u>1. Identifying partners and building a network</u>

We began identifying partners and building a network in October 2023 through our recruitment strategy. We shared accessible information materials about RDRN through CamRARE's mailing list and social media channels. This generated significant interest, particularly within the rare disease community, which enabled us to create a community working group of 29 members, 24 of whom remained involved until the end of the project. In an early meeting this group identified our key intended partners for the project, defined as the following stakeholder groups:

1. Rare disease community - individuals: advocates, people living with a rare disease, carers, supporters, parents, families, charities, patient groups, entrepreneurs;
2. Rare disease community - groups: charities, patient groups

3. Researcher - researchers within public and private health organisations, pharma, biotechnology, medical tech, and academia;
4. Healthcare - healthcare organisations, hospital trusts, and healthcare professionals;
5. Industry - representatives within health research, pharma, biotechnology, and medical tech other than researchers and funding;
6. Funders - commercial, public, academic, philanthropic and charitable funding bodies.

RedWeb released the RDRN platform for internal testing in October 2024. Following technical updates, the RDRN was promoted and launched at CamRARE's RAREfest in Cambridge, 22-23 November 2024. RAREfest is a free, public-facing biennial event, providing a unique platform for change through interactive exhibits, talks, film, and art showcasing ground-breaking science, visionary technology, and pioneering organisations, improving lives and bringing hope to those affected by rare conditions. The event unites patients, advocates, and experts to address the challenges faced by people affected by rare diseases, encouraging community collaboration. This year RAREfest attracted over 800 attendees across the weekend.

At the time of writing, only 6 weeks after launch, the platform has 193 members and 9 research questions. Members represent each stakeholder group and we have received widespread support from existing research networks, PPI initiatives, medical charities, and industry partners from across the UK and internationally. We've been invited to present the RDRN and our approach to co-design and co-production at numerous events, including the UK's International Rare Disease Mirror and Action Group. Over 980 people worldwide have subscribed to the RDRN mailing list and we have an involved, active following on LinkedIn with 900 followers. Two community members with relevant experience were appointed as freelance consultants in graphic design and managing RDRN social media campaigns with targeted outreach, engaging non-community stakeholder groups. This aspect of work is ongoing and will be incorporated into the iterative process as we update the platform following feedback gathered from all stakeholders in 2025.

2. Conducting preliminary scoping activities to inform future research methodology

At the start of the project we conducted a mapping and gapping activity and created a collaborative document where any member of our community group could gather and share relevant resources to support patient-led research from the following categories: training, websites and organisations, research resources, PPI resources. Our community group populated this document using their wealth of knowledge within the UK's rare disease landscape. In May 2024, we hosted a community working group meeting which focused on the topic of resources, assessing what is already publicly available and what the team felt was required to facilitate patient-driven research through the RDRN platform. Following the outputs of this meeting, the community group decided that a smaller task and finish group (the resources development team) should be established to create templates, guidance, and video tutorials. This team created the decided-upon resources to be housed under the 'members-only' section of the platform between July and September 2024. Further work and expert input are now required to expand resources and training available within this self-directed RDRN 'patient research pathway'.

During our monthly community working group meetings, we discussed research design and how to increase opportunities for involvement in research, particularly for those living with a

rare disease. For example, clustering based on phenotype or how to address the barriers of running an international clinical trial. This objective has also been addressed in more detail through a Research Fellowship awarded to grant co-applicant Laura Cowley via NIHR East of England Applied Research Collaborative (EoE ARC). This Fellowship included further outreach, an online survey, and one-to-one semi-structured interviews with people living with a rare disease, their families and carers to understand their research priorities and views on appropriate methodologies. Key findings from this work illustrated the need for inclusive research design which welcomes patients living with multiple co-morbidities and/or ultra rare conditions. The rare disease community was found to be research aware and motivated to be involved in setting the research agenda, extending from disease-specific trials to disease-agnostic adaptive studies which consider overarching themes, as well as mixed-method approaches to capture mental health and family quality of life.

With future development, the RDRN aims to support and promote inclusive methodologies, including drug repurposing and data emulation using routinely collected sources. While we appreciate each new research proposal must be developed in a manner best suited to its aims, objectives and outcomes, RDRN resources and training will introduce design features and disease agnostic outcome measures which could add value to the wider rare disease community.

3. Developing and growing a sustainable network

We have always envisaged that the RDRN would have a legacy beyond the duration of this funding. Developing a sustainable network and exploring longer term sustainability options have been part of the work we have conducted over the last 15 months. Following the initial recruitment and outreach stage of the project which ran between October 2023 and December 2023, we built a core team of members from a range of non-community stakeholder groups (academics, clinicians, funder and industry representatives). These individuals have been integral in building the platform with a wealth of knowledge and lived experience in the rare disease space. The community group also decided that a smaller task and finish group (the community management team) with relevant professional experience should investigate ways to ensure a future sustainable business model and discuss RDRN's transition from grant-funding to independent social venture. The team met 6 times between October to December 2024 to discuss the potential market and create value proposition models.

We also undertook significant outreach by presenting at events and liaising with umbrella charities and other key groups to spread the word about RDRN and grow our network. The voice of our rare disease community has been centric, and we have understood their research priorities and how they would like to lead their own research through co-production methods during our community working group meetings. To understand our non-community stakeholder groups, we undertook separate consultation work with each group, including a collaborative project with MediData. MediData identified key motivators for industry and researchers to engage with the RDRN platform. They found that by aligning with the MHRA's Patient Involvement Strategy 2021-2025, the RDRN's focus on patient involvement has the potential to enhance drug development efficiency and better regulatory outcomes. Industry stakeholders were seen to value its potential for broad collaboration across rare diseases and integrated evidence planning. Academic researchers saw it as a way to enhance publication relevance, enhance grant competitiveness, and connect with patient-led groups

to ensure meaningful engagement. Government institutions recognised the RDRN's role in improving care outcomes and fostering inclusive trials, while non-profit organisations appreciated its ability to align with patient priorities, build partnerships, and increase visibility, enhance capacity and funding. Collectively, these findings underscored the platform's value as a comprehensive resource and network for advancing patient-driven rare disease research.

However, further work is required to understand motivating factors and perceived barriers for non-community organisations interested in supporting RDRN activities.

Because of our success in growing a network, this work has sparked conversations about how we transition RDRN into an independent entity like a charity or social enterprise and create a sustainable future funding model. Our community management team are continuing to consult on this topic, and support-in-kind has been provided by Cambridge Enterprise. This work package will be a core aim of the project moving forward.

Discussion

Further work is now required to understand the challenges associated with patient-driven and co-created research as perceived by investigators, industry partners and funders. Additional funding is necessary to explore these factors, appropriately expand the platform, and establish proof-of-concept by supporting the development of patient-driven disease-specific and disease-agnostic research proposals. Community members have contributed to new grant applications as collaborators and named co-applicants, attempting to secure follow-on funding for further RDRN development.

In the long-term, RDRN aims to 1) develop an adaptable framework de-risking co-design in healthcare research, and 2) offer a brokering service for stakeholders keen to involve patient communities in research design upstream of funding. Opportunities for corporate sponsorship, support-in-kind, and mentorship will also enable low-risk engagement for stakeholders to support patient communities develop their own research projects.

Conclusion

Outreach conducted by CamRARE, PLRH and this project illustrates that the rare disease community is research aware, motivated to help set the UK research agenda, and keen to contribute to patient-driven opportunities. The newly established Rare Disease Research Network is widely perceived as providing a meaningful way for the community to develop and showcase their own research ideas, engage with healthcare and research professionals, increase awareness about rare diseases, advocate for change, and be involved in developing new research. Community feedback gathered reflects the unmet needs outlined in the UK Rare Disease Framework: faster diagnosis, increased awareness among healthcare professionals, better coordination of care, and improved access to specialist care, treatments and drugs. A primary overarching theme is for new research to be inclusive and capture disease-agnostic features through adaptive and mixed-method designs.

The RDRN is unique within the UK, offering an alternative way for patient groups and people affected by rare disease to be involved in research. The platform will help address the unmet need for research equity across the rare disease community and complement existing rare disease research networks and patient and public involvement initiatives. The approach to

co-production benefits both patient-driven and traditional investigator-led paradigms, empowering all rare disease stakeholders to collaborate and shape future research. However, further work is required to involve people affected by a rare disease who are not supported by a patient group or charity, and to understand motivating factors and perceived barriers for industry, healthcare and research organisations. To succeed as a long-term platform, the RDRN must create a sustainable business model and transition away from a grant-funded project.

Patient and Public Involvement

This section of the report has been written with members of the public who have been involved in the research.

As an NIHR 'Public Partnership' Programme Development Grant, this project has been entirely co-produced by the rare disease community (people living with a rare disease, family, advocates, patient support group representatives). Over 20 individuals from the community were involved in developing the platform, determining its user-interface, accessibility features, functionality, identifying and creating new resources, and identifying and engaging with the wider community and external stakeholders. Outreach with the wider community, largely via online newsletters, surveys, virtual meetings, in-person events and networking has helped direct working group activities.

One challenge has been involving people affected by a rare disease who are not members of a patient group or charity. We believe our continued work with embedded, trusted, community groups and healthcare providers will spread awareness and encourage these individuals to become research active. Key collaborations with UK patient organisations include Beacon for Rare Diseases, Genetic Alliance UK, Rare QoL, Unique, Metabolic Support UK, RareMinds, Breaking Down Barriers, as well as numerous disease-specific patient support groups and internationally-based charities.

Alongside the main working group, three task and finish groups, each composed of up to 10 members of the rare disease community, were formed to focus on 1) guideline and training development, 2) communication and marketing, and 3) sustainable business management. Two community members with relevant experience were appointed as freelance consultants in graphic design and managing RDRN social media campaigns with targeted outreach, engaging non-community stakeholder groups. In addition, community members contributed to new grant applications as collaborators and named co-applicants, attempting to secure follow-on funding for further RDRN development.

Prior to launch in November, the main working group tested a beta version of the platform, collating technical errors to share with the website programmer. The RDRN launched at CamRARE's RAREfest, a free two-day public-facing event in Cambridge. Launch activities and promotional material were designed and created by the communication and marketing task and finish group. Multiple members attended the in-person event to engage attendees and promote the RDRN; one member joined RDRN staff to present the platform at a VIP launch, and two others joined RDRN staff to present the platform as keynote speakers. The

launch was well attended and RDRN was positively received, entirely because of the community's efforts.

As the platform is now live, the community working group have transitioned into active members, showcasing their research ideas and promoting the RDRN throughout their networks, at rare disease events and conferences.

All community members received NIHR-recommended remuneration of £25/hr for attending meetings and completing agreed tasks; meetings were scheduled at a time decided by a majority vote, with in-person, virtual, and independent working from home options available. All resources, including grant documents and finance, were securely shared with the community team to encourage transparency and to support collaborative working. Travel and accommodation were booked ahead of time by staff as requested, to ensure community members were not impacted financially. Remuneration was paid directly via BACS or donated to a charity on behalf of the individual as preferred. Advice for those in receipt of benefits was sensitively provided.

All community members involved in the project have expressed a desire to remain involved and further develop the platform. Testimonials and survey feedback gathered from community members indicates that RDRN staff were consistently welcoming and inclusive.

Data sharing statement - See link

[https://www.nihr.ac.uk/documents/nihr-position-on-the-sharing-of-research-data/12253] for the NIHR position on the sharing of research data. The NIHR strongly supports the sharing of data in the most appropriate way, to help deliver research that maximises benefits to patients and the wider public, the health and care system and which contributes to economic growth in the UK. All requests for data should be directed to the award holder and managed by the award holder.

Disclaimer

This project is funded by the National Institute for Health and Care Research (NIHR) under its Programme Development Grants Programme (Grant Reference Number NIHR205155). The views expressed are those of the author(s) and not necessarily those of the NIHR or the Department of Health and Social Care.

This project was carried out between July 2023 and December 2024. This final report has not been peer-reviewed. The report was examined by the Programme at the time of submission to assess completeness against the stated aims. These reports do not undergo a peer review process.