Participant Information Sheet

Research Study Title:

How can we make it easier for people living with rare diseases to take part in rare disease research across the Island of Ireland including Northern Ireland? A Mixed Method Study

We are inviting you to take part in a research study. Before you decide you need to understand why the study is being done and what it would involve for you. This study involves a 15 minute survey with a subsequent optional 45-minute interview online via Microsoft Teams or via telephone. Please take time to read the following information carefully and discuss it with others if you wish. You should not take part in this study until you have had all your questions answered satisfactorily. If you require further information or would like this information in another format, please contact us below using the subject line, "Rare Disease Research 2026".

raredisease@qub.ac.uk

a.j.mcknight@qub.ac.uk (Professor Amy Jayne McKnight, Responsible Academic)

We acknowledge that the term 'rare diseases' may be uncomfortable for some as it may have negative misconceptions. However, as the term 'rare diseases' is widely used and referred to by patients, carers, healthcare professionals, industry members and policymakers, we will be using it throughout this document and the survey for consistency.

Purpose of the study

To capture the voice of those affected by a rare disease on how research can be more accessible.

Why should I participate?

You should consider participating if you live and/ or are affiliated with an organisation on the Island of Ireland (Republic of Ireland or Northern Ireland) and find yourself in any of the categories below:

- have been diagnosed with a rare disease
- are a carer and/ or are a family member of a person with a rare disease
- a researcher investigating rare diseases
- a rare disease advocate and/ or a rare disease charity representative.

Research is an important process in helping understand more about how conditions develop, helping people live with rare diseases, and for the development of treatments. We know that it may be difficult for everyone to access research, including clinical trials. Therefore, this research will explore strategies and that could be implemented to overcome those barriers, making research more accessible. We also welcome hearing success stories from people who live on the Island of Ireland (Republic of Ireland or Northern Ireland) and have participated in rare disease research.

Why have I been invited to take part?

You have been invited to part of this research because you live in or are affiliated with an organisation on the Island of Ireland (Republic of Ireland or Northern Ireland) and may find yourself in any of the categories below:

- have been diagnosed with a rare disease.
- are a carer and/or a family member of a person with a rare disease.
- are a researcher investigating rare diseases.
- are a rare disease advocate and/ or a rare disease charity representative.

Do I have to take part?

Participation in this research is completely voluntary, and you have the right to refuse participation and any questions. Please note that once you have completed this survey, you will only be able to withdraw your responses before it is anonymised. Similarly, once the interview has been transcribed, you will be unable to withdraw your responses. Your survey or interview responses may be included in our study anonymously. If you wish to withdraw your survey response before it has been anonymised, or withdraw from participating in the interview before it is conducted, or withdraw from the prize draw, please contact us at raredisease@qub.ac.uk. If you chose to withdraw from this study once the survey has been anonymised or after the interview has been transcribed, your responses may be quoted anonymously in our study.

If you do decide to take part, you will be given this information sheet to keep and you will be asked to provide consent before providing your responses for the survey and a recorded verbal agreement before commencing a telephone or video interview.

What do I have to do?

The survey is expected to take 15 minutes of your time, and the optional interview is expected to take 45 minutes. The survey is available online, or we can arrange for it to be completed over the telephone with a researcher. The survey will be hosted within Qualtrics. Qualtrics will leave a cookie on your browser when the survey is completed to prevent duplicate responses. Please note that this survey is only open for 6 weeks and closes on 17th of November 2026. The survey will no longer accept responses past this date. The interviews will be conducted online via Microsoft Teams or via telephone. All data collected, from survey responses, interview transcripts, will be anonymised and no identifying information will be included in any publications or reports arising from this research. We have developed the survey and interview guide, with patient and public involvement.

Are there possible disadvantages and/or risks in taking part?

There are no expected risks of participating in this study. Some questions may cause psychological distress, anxiety or might be challenging, but this is not expected. However, in the unlikely event, you can choose to not answer those questions, and the researcher will move to the next question. You may also ask the researcher to stop the interview and resume later or not at all.

What are the possible benefits of taking part?

There is no intended direct benefit to participants from taking part in this study. The information you provide in this study may help to improve the future of accessibility to rare disease research, including clinical trials. This could lead to increased participation in research, which may lead to greater research into treatments available for those living with a rare disease.

How will information you provide be recorded and stored?

Only named research team members will have access to the data from this study (Priscilla Jeyaraj, Dr. Blánaid Hicks, Professor Amy Jayne McKnight, and Dr. Noleen McCorry). Our research collaborators from outside of Queen's University Belfast (Dr. Ameeta Retzer (University of Birmingham) and Vicki Hedley (Newcastle University)) will only have access to anonymised data and information. Responses from this study will be kept anonymous and no identifiable information will be included in the analysis of this study. Your responses may be quoted anonymously as part of a report and manuscript for publication. All data generated from this study will be stored on a Queen's University Belfast password-locked encrypted computer.

Information that is discussed during the interviews is confidential with the exception that if you disclose information that indicates that you are at risk of harming yourself or others, or in danger of being harmed by someone else, the researcher is legally obliged to pass on this information to the responsible academic, Professor Amy Jayne McKnight and members of the

research team to discuss if further action is required which may include informing emergency medical services (General Medicine, Nursing Council) or the police.

What will happen to the results of the study?

You can provide your contact details at the end of the survey and interview to consent to be contacted with the results of this study. Your contact details will not be linked to your responses. The results of the study will also be sent to our charity and voluntary sector partners. Additionally, the results and will be presented in conferences, seminars, and during research meetings.

This research will be submitted as part of Priscilla Jeyaraj's PhD thesis. The research will be made available in an electronic or digital thesis that is open access. This will be publicly available to all on the Queen's Research Portal. The research will also be made available in forthcoming publications. These will likely be made open access, which means they will be publicly accessible via public platforms (for example, Queen's Research Portal).

What if something goes wrong?

We do not anticipate anything going wrong that would harm participants, however we do appreciate that incidents may occur at any stage. If this study has harmed you in any way, or if you wish to make a complaint about the conduct of the study, you can contact the lead academic at Queen's University Belfast using the details below for further advice and information:

Professor Amy Jayne McKnight (a.j.mcknight@qub.ac.uk)

Should you remain unhappy and wish to make a formal complaint, you can contact the Research Governance Team at Queen's University Belfast (Tel: 028 9097 2529, Email: researchgovernance@qub.ac.uk)

Who is organising and funding the study?

This study is part of Priscilla Jeyaraj's PhD, that is funded by the Department for the Economy (DfE) within the LifeArc Centre for the Acceleration of Rare Disease Trials. This project is supported by LifeArc (ARDT, 10747), Health and Social Care Research and Development Office (COM/5802/24), and the Northern Ireland Kidney Research Fund (NIKRF_Ecosystem).

Ethical review of the study

This study has been reviewed by two expert peer reviewers and the Faculty of Medicine, Health & Life Sciences (MHLS) Research Ethics Committee (490380).

Who should you contact for further information?

Please contact raredisease@qub.ac.uk with the subject title 'Rare Disease Research 2026' if you wish to access these, or if you have any questions.

Professor Amy Jayne McKnight, Responsible Academic: a.j.mcknight@qub.ac.uk

This research will be conducted in compliance with data protection legislation. For more information about how we look after your information, how to access your rights and who to contact if you have any queries or concerns about data protection please visit the Queen's University Belfast website;

https://www.qub.ac.uk/privacynotice/Research/ListofResearchPrivacyNotices/PrivacyNoticeforResearchParticipants.html

The privacy notice for the survey platform (Qualtrics) is available at https://www.qualtrics.com/privacy-statement/.

Thank you for your interest in this study and for taking the time to read through this information sheet.