

The Botnar Research Centre
The University of Oxford
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Study Telephone number: 07775541615

Participant Information Sheet
Rare and Undiagnosed Diseases Study (RUDY)
Main Study

We would like to invite you to take part in our research study. Before you decide whether to take part, we would like you to understand why the research is being done and what it would involve for you. Talk to others about the study if you wish. Ask us if there is anything that is not clear. **This information sheet describes the main RUDY study, there are other studies within RUDY that you may also be interested in. Please contact a member of our team or visit the RUDY study website 'More Information' section.**

What is the purpose of the study?

We are interested in understanding more about all aspects of rare diseases. Some of this new understanding will be used to develop new tests and treatments for patients with rare diseases. A first step in doing this is to gather information in order to describe in more detail the different types of rare diseases and what it is like to live with them. We intend to do this through questionnaires over a period of five years, and in the future, we hope to introduce some sub-studies such as a physical assessment, blood and urine and a skin biopsy (for certain rare diseases). We are recruiting as many participants to this study as possible because of the rare nature of these diseases. We are also recruiting unaffected relatives - such as partners and blood relatives (parents, siblings) - to try to understand the effect the rare disease can have.

Why and how am I being invited?

You are being invited to take part because you have been diagnosed with a rare disease. You also may have been identified by a hospital doctor or family member or you may have seen a link to Rudy through a charity website or on social media.

Do I have to take part?

No. It is up to you to decide whether or not to join the study. This information sheet will explain the study to you. You are free to withdraw at any time, without

giving a reason. This would not affect the standard of care you receive or any future treatment you may need.

What will happen to me if I take part?

If you agree to participate we will ask you to register online. You have the option to consent online or on paper.

If, however, we are unable to contact you after you have registered your details we will send an email reminder to arrange a consent call 4 and 8 weeks after the date of registration. If there is no response 12 weeks after registration you will be sent an email to withdraw after registration.

Because the study aims to collect information over a five year period, we are aiming to make your involvement as simple and flexible as possible. You can decide what parts of the study you want to be a part of, and how you would like us to contact you: whether by letter, telephone, email or text. You will be free to decide how much you want to take part and your preferences at any time during the study.

Beginning of the Study

Gathering information

We will ask for your permission to access and use your NHS and social care records to get more information about you and the services you have accessed.

We will also ask your permission to contact you if there are new studies for which you may be eligible. Agreeing to be contacted does not oblige you to participate in any studies.

Once you have registered on the RUDY study website and we have received your consent decision we will contact you to confirm that you have been given access to your own secure part of the website to complete:

- A history of any symptoms and treatments you have received. The amount of time this will take to complete will vary by person and you do not have to complete it in one session.
- A variety of questionnaires
We are also testing different questionnaires to see how well they assess you compared to other people. This may help us develop more “personal” approaches to treatment in the future. For this reason some of the questionnaires may ask similar but not the same questions. This may seem repetitive, but it is an important part of the testing of these questionnaires. The questionnaires will take approximately 30 minutes to complete.
- We would like to establish a family tree for each participant, this may include affected or unaffected family members (i.e. have not been diagnosed with a rare disease) who have consented to participate in the study. The reason for this is testing for differences between family members often gives a lot more information than comparing people from different families. If you are happy to

complete it, there is a family history map where you can add the full name and date of birth of any family member and then add if you think they are affected or not. You will also be able to add if you consent for your data to be linked to theirs. We will only link data if you both agree and you will not be able to see each other's data.

- Given how rare these diseases are, it is possible you may already be part of another study. To avoid duplicating information and making the most of all your research data, if you have consented to take part in other research studies, with your permission, we would like to link the data you have provided to those other studies with your RUDY account.

This is only possible if you have given consent to the other research team which you are involved with, to share your data. To enable this link between studies we will use your NHS number, date of birth and full name.

Tissue removed as part of regular care

If you have an operation or other procedure and tissue is removed as part of this treatment, some of the sample is kept as part of your medical record so that it can be examined again as part of your care, now and in the future. If you agree, we would like to have access to this tissue. Enough will always be kept for your medical record: your clinical care comes first.

Subsequent years:

We would like to know how your symptoms and treatments change over time. We will ask you to tell us if, how and when you want to be reminded to update your information. These reminders could include completing questionnaires and providing RUDY with follow up information by letter, telephone, text message, or e-mail.

We hope you will access your part of the website to record any new changes in your symptoms or treatment as that happens, or every 6 months to repeat the questionnaires.

What are the possible disadvantages and risks of taking part?

This study has a low risk of harm. With the exception of sharing your NHS number, date of birth and full name with other authorized research administrators whose research you have consented to, in order to link your data across research studies, the secure and confidential parts of the website will only be accessible to those authorized individuals, yourself and our website administrator.

While we will remove names and addresses at all other times, given the rarity of some of the diseases it may be possible to identify some individuals from their age, height, weight and diagnosis. Where this is likely, we will contact individuals for permission for their information to be used in any publications or reports using your data.

What are the possible benefits of taking part?

We cannot promise the study will help you personally but we will use the information you give us to help develop new tests and treatments for rare diseases.

From the website, you will be able to keep a diary of your symptoms and treatments and if needed print out a list of all the treatments and complications you may have had. This may be useful when you see other health professionals. Participants will also see on the database links to current research for the rare diseases and related disorders.

There seems to be a lot to do if I take part.

The aim of RUDY is to improve our understanding of rare diseases. We value any information you can give us no matter how small. We have designed RUDY to be flexible so you are in control of how much you want to contribute to the project, including questionnaires and diaries. You don't have to fill in everything at once, but can choose when you have time to answer questionnaires or fill in the diaries to suit you. How much of your time and effort you give is completely up to you. The more information you give, the better we will be able to answer important questions about rare diseases.

What happens at the end of the five years?

Our intention is to develop this study as a core national project and continue the research. After five years we will review the project and inform you of whether and how we will continue the research, and invite you to continue to take part. In the unlikely event we plan to end the study, we will send you a copy of all the information you have entered for your own record.

What if there is a problem?

Any complaint about the way you have been dealt with during the study or any possible harm you might suffer will be addressed.

The University of Oxford, as Sponsor, has appropriate insurance in place in the unlikely event that you suffer any harm as a direct consequence of your participation in this study.

NHS indemnity operates in respect of the clinical treatment with which you are provided.

In the event that you wish to complain about any aspect of the way in which you have been approached or treated during the course of this study, you should contact Dr. MK Javaid 01865 737831 or kassim.javaid@ndorms.ox.ac.uk or you may contact the University of Oxford Clinical Trials and Research Governance (CTRG) office on 01865 572224 or ctrig@admin.ox.ac.uk.

The Patient Advisory Liaison Service (PALS) is a confidential NHS service that can provide you with support for any complaints or queries you may have regarding the care you receive as an NHS patient. PALS is unable to provide information about this research study.

If you wish to contact the PALS team please refer to the PALS website <http://www.ouh.nhs.uk/patient-guide/pals.aspx>.

Will my taking part in the study be kept confidential?

Yes, we value your information and take security and confidentiality very seriously. The website is password protected, and you will be the only one aside from the authorized research administrator(s) that can access your secure area. Your personal information is encrypted within the University of Oxford and available only to the authorized administrators so they can match your account with your information. If you consent to be approached in the future regarding other ethically approved research your personal details we hold will be kept separate from your anonymised data and will only be accessed by an authorized research administrator. Agreeing to be contacted in the future does not oblige you to take part in any research, it is consent to be approached only.

Any samples or information about you that leaves the hospital/surgery will have your name and address removed so that you cannot be identified. It will be stored in an anonymised form where, again, only the authorized study administrator(s) will be able to link you to the anonymised data. Your anonymised information will be held in a secure database, which is located at the The Botnar Research Centre Oxford NIHR Musculoskeletal Biomedical Research Unit.

Responsible members of the University of Oxford or NHS Trusts may be given access to data for monitoring and/or audit of the study to ensure we are complying with regulations.

What will happen to any samples and data I give?

If you agree, your data will be stored indefinitely in data bases and surplus tissue samples collected during operations you may have as part of standard care will also be stored indefinitely in a tissue bank. Your data will be pseudoanonymised (this is where your data will have any personal identifying information removed and only the admin team will still be able to know your personal details). This material and these data will be used in the future for research that cannot yet be specified and may in the future be deposited in research tissue banks where they can be used by other researchers in the UK and internationally. Pseudoanonymised samples and / or data collected during the course of the study may also be passed on to other organisations which may include commercial organisations. All research will need to be reviewed and approved by an ethics committee and the RUDY network.

Your donated samples may be used in genetic research aimed at understanding the genetic basis for rare diseases. Any results that are clinically important as judged by the Rudy Data Oversight Governance Committee will be sent to the clinical team caring for you.

Pseudoanonymised data collected about you during the study may be looked at by both national and international industry researchers. These researchers will first be approved by the Rudy Data Access Committee to ensure their involvement will help contribute to the aims and objectives of Rudy.

The RUDY network includes doctors and patients. If you would like to volunteer to join our network; who meets via Skype every 2 months to work with the study, please contact Kassim Javaid (rudy@ndorms.ox.ac.uk).

What will happen to the results of the research study?

Results from the study will be presented in professional journals and at conferences. We will send you a newsletter every year to keep you informed of the study's key findings. You will not be able to be identified in the results.

What if relevant new information becomes available?

We expect new information to become available during the time you are taking part in the study. If there is a new test that we wish to use within the study, we will ask the ethics committee for permission to include it in this study and then ask your permission to use it. If a new study starts for which you may be eligible we will contact you to let you know.

What will happen if I don't want to carry on with the study?

You are free to withdraw from all or part of the study at any time. On your page of the website you will be able to indicate which parts of the study you do not want to carry on with, and whether you want to withdraw samples and information that have already been collected, or simply stop any involvement in the future. It will not be possible to remove any information that has been anonymised and already used in research analysis.

If you die while taking part in the study, data and samples you have provided would continue to be available for use in the study, as per the provisions of your consent at the time of death.

Will my General Practitioner/Family doctor (GP) / Clinician be informed about my participation?

We will only contact your GP /clinician if we require a copy of your GP /hospital records now and in the future.

Who is organizing and funding the research?

This study is funded by a research partnership between the National Institute of Health Research (NIHR) Rare Diseases Translational Collaboration, NHS and the University of Oxford at the Musculoskeletal Biomedical Research Unit, Oxford.

Other centres around the country are helping to see patients in their clinical research facilities

Who has reviewed the study?

All research in the NHS is looked at by an independent group of people, called a Research Ethics Committee, to protect participants' interests. This study has been reviewed and given favourable opinion by Berkshire B Research Ethics Committee.

Thank you for reading this information sheet.

For further information about the RUDY study visit www.rudystudy.org or call 07775541615.

General information about participating in research can be found at:

<http://www.invo.org.uk/>

Yours sincerely



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