

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

Participant Information Sheet for parents/legal guardians

Rare and Undiagnosed Diseases Study (RUDY)

Main Study

Your child is being invited to take part in our research study. Before you decide whether you would like your child to take part, we'd like you to understand why the research is being done and what it would involve for you and your child. 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 and your child may also be interested in. Please visit the RUDY study website 'More Information' section.**

What is the purpose of the study?

We are interested in understanding more about and developing 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. We are recruiting as many participants to this study as possible because of the rare nature of these diseases.

Why and how is my child being invited?

Your child is being invited to take part because he/she has been diagnosed with a rare disease or is a blood relative of someone that has. He/she 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.

Does my child have to take part?

No. It is up to you and your child to decide whether or not to join the study. This information sheet will explain the study to you and we will answer any questions you may have. You are both free to withdraw at any time, without giving a reason. This would not affect the standard of care your child receives or any future treatment your child may need.

What will happen to my child if he/she takes part, and what will I have to do if my child does take part?

If you agree for your child to participate we will ask you to register on behalf of your child online. You have the option to consent online or on paper.

If, however, we are unable to contact you after you have registered your child's 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.

As 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 your child to be a part of; and you will be able to consent for the parts of the research that you are happy for your child to participate in. You will also be able to tell us how you would like us to contact you: whether by letter, telephone, email or text. You will be free to change your mind about your child's participation and yours or your child's preferences at any time during the study.

Beginning of the Study

Gathering information

We will ask for your permission to access and use your child's NHS and social care records to get more information about him/her and the services he/she has accessed. With regards to your child who is not affected by the rare disease, we would only ask for your permission to access any records if we felt it was relevant to the study.

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

Once you have registered on the RUDY study website 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 he/she has 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. This may not be relevant if you are consenting on behalf of a sibling of a child with a rare disease, but we are aware some siblings may have other medical issues not related to the rare disease and so it is possible for you to tell us anything you would like to tell us about your child's medical condition.
- A variety of questionnaires
We are also testing different questionnaires to see how well they assess him/her 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 family tree 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 on behalf of your child, 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 by the rare disease or not. You will also be able to add if you consent for your child's 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 your child may already be part of another study. To avoid duplicating information and making the most of your child's research data, if you have consented on their behalf to take part in other research studies, with your permission, we would like to link the data you and your child have provided to those other studies with their RUDY account. This is only possible if you and your child have given consent to the other research team which your child is involved with, to share their data. To enable this link between studies we will use your child's NHS number, date of birth and full name.

Tissue removed as part of regular care

If your child with the rare disease has an operation or other procedure and tissue is removed as part of this treatment, some of the sample is kept as part of your child's medical record so that it can be examined again as part of his/her 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 child's medical record: your child's clinical care comes first.

Subsequent years:

We would like to know how your child's symptoms and treatments change over time. We will ask you to tell us if, how and when you and your child want to be reminded to update your child's information.

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

What are the possible disadvantages and risk of taking part?

This study has a low risk of harm. With the exception of sharing your child's NHS number, date of birth and full name with other authorized research administrators whose research you have consented to, in order to link your child's data across research studies, the secure and confidential parts of the

website will only be accessible to those authorized individuals, yourself, your child 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 the report.

What are the possible benefits of taking part?

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

From the website, you will be able to keep (if it is relevant) a diary of your child's symptoms and treatments and if needed print out a list of all the treatments and complications they may have had. This may be useful when your child sees 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 my child takes part.

The aim of RUDY is to improve our understanding of rare diseases. We value any information you and your child can give us no matter how small. We have designed RUDY to be flexible so you and your child are in control of how much you want to contribute to the project, including questionnaires, diaries and potentially other RUDY sub-studies, such as blood and urine tests. 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 yours and your child's 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 and your child 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 child's own records.

What if there is a problem?

Any complaint about the way you or your child have been dealt with during the study or any possible harm you or your child 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 trial.

NHS indemnity operates in respect of the clinical treatment with which your child is provided.

In the event that you wish to complain about any aspect of the way in which you or your child has been approached or treated during the course of this study, you should contact Dr. MK Javaid 01865 737831 or rudy@ndorms.ox.ac.uk or you may contact the University of Oxford Clinical Trials and Research Governance (CTRG) office on 01865 572224 or ctr@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 child’s taking part and my contribution to my child’s taking part in the study be kept confidential?

Yes, we value your child’s information and take security and confidentiality very seriously. The website is password protected, and you and your child will be the only ones aside from the authorized research administrator(s) that can access your secure area. Your child’s personal information is encrypted within the University of Oxford and available only to the authorized administrators so they can match your child’s account with their information.

If you consent to be approached in the future regarding other research your personal details we hold will be kept separate from your child’s anonymised data and will only be accessed by an authorized research administrator.

Agreeing to be contacted in other ethical research in the future does not oblige your child to take part in any research, it is consent to be approached only.

Any samples or information about he/she that leaves the hospital/surgery will have his/her name and address removed so that he/she cannot be identified. It will be stored in an anonymised form where, again, only the authorized study administrator(s) will be able to link your child to the anonymised data. Only researchers and regulatory authorities will have access to your child’s anonymised information. Your child’s 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 the 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 my child gives?

If you agree, your child's data will be stored indefinitely in data bases and any remaining tissue samples collected during this study will also be stored indefinitely in a tissue bank. Your child's data/ tissue sample will be pseudo-anonymised (this is where your child's data will have any personal identifying information removed and only the admin team will still be able to know your child's personal details). This material and collected 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 that may include commercial organisations. All research will need to be reviewed and approved by an ethics committee and the RUDY network.

The RUDY network includes doctors and patients. If you, as a parent of a participant, would like to volunteer to join the group, who meet via Skype every 2 months to work with the study, please contact Kassim Javaid (rudu@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 and your child informed of the study's key findings. Your child 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 your child is 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 your child may be eligible, we will contact you to let you know.

What will happen if we 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 child's 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.

In the event of the death of a participant, data and samples provided by the donor would continue to be available for use in the study in perpetuity, as per the provisions of consent at the time of donor 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 participants 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.

Further information and contact details

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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