Great Ormond Street Hospital for Children NHS Foundation Trust

GREAT ORMOND STREET INSTITUTE OF CHILD HEALTH

Version 3, 07 May 2024

ADULT PARTICIPANT INFORMATION SHEET

Understanding the medical features of Norrie disease

We would like to ask your permission to include you in this study.

Before you decide it is important for you to understand why the research is being done and what it will involve. The researcher or a friend or family member may read this information sheet to you if required. Please ask questions if anything is unclear or if you require more information and discuss it with others if you wish. You can contact the research team if you need any more information. Our contact details are at the end of this sheet. Take time to decide whether or not you wish to take part.

Why is the study being done?

The study aims to give people and families with Norrie disease better information about the condition. We want to understand what kind of vision and hearing problems it causes, when these problems begin, their severity, and how commonly it causes any other problems such as seizures, learning difficulties or problems with blood vessels. We also want to understand the variation in people of different ages who have Norrie disease; this is because in the future we will want to be able to tell if new treatments have been successful. For example, if we want to develop a treatment to slow down or prevent hearing loss, how do we tell when it has worked if we don't know at what age hearing loss starts and how quickly it deteriorates.

We want to link information about severity and type of hearing loss, blindness and other problems with the specific gene changes that have caused Norrie disease in different people. We want to know if some genetic changes can cause Norrie disease to be more or less severe, we want to better understand how the blindness and hearing loss develop, and we want to find out whether there is anything that can be done to change this.

In order to do this, we would like to get in touch with as many people with Norrie disease (children and adults) as possible and to ask them for a detailed medical history, examine them and ask their permission to review their medical records and previous test results.

We hope that in the future this will improve information given by doctors about the course of hearing loss, blindness and other problems, to families with Norrie disease. Although this part of our research is not looking for a cure, we hope that this research, together with our laboratory work, can pave the way for other

research in the future.

Why have you been invited to take part?

You have been invited to take part because you have been diagnosed with Norrie disease.

Do you have to take part?

No. This decision is up to you entirely. If you do decide to take part, please keep this information sheet. You will be asked to sign a consent form. If you decide to take part, you are still free to withdraw at any time and without giving any reason. This will not affect the standard of care you receive.

What will happen to you if you take part?

If you want to take part in this study, we will ask you questions about your Norrie disease in the past and now. We will also ask you about any other medical problems you may have experienced. With your permission, we will look through your medical records and results of your clinical tests to understand more about your condition.

If you have not had hearing and eye tests in the last few years, we may suggest that you have these in an NHS clinic and help to arrange it. If you do not want to have these tests repeated, we may ask your permission to write to your GP to ask for details of past tests.

What are the benefits of the study?

We hope that in the future this will improve the information given about the course of hearing loss, blindness, and other problems to families with Norrie disease. Although this research is not looking for a cure, we hope that this research can pave the way for other research in the future.

What happens if I change my mind about taking part?

If you agree to participate and then later decide you do not wish to take part, please contact us by phone, email or letter, and we will destroy any sample you have donated, and write to acknowledge that we have received your notification that you no longer wish to be part of the study.

Will my taking part in this study be kept confidential?

We request your permission for restricted access to your medical records and the information collected about you during the study. All information collected about you will be kept strictly confidential. Any information that is included in a research publication will have all identifying features removed, so that you cannot be recognised from it.

How will my data be stored?

All of the information you give will be anonymised so that those reading reports from the research will not know who has contributed to it. Nobody other than

the researchers will have access to your data. All information collected about you will be saved securely on password-protected NHS computers and stored securely for 15 years in accordance with the Data Protection Act 2018.

Data will also be managed and analysed using the Digital Research Environment (DRE). The DRE provides technical safeguards and processes for strong data governance by design that support compliance with EU General Data Protection regulation (GDPR), UK Data Protection Act (DPA) and data security standards.

Will the use of my data meet GDPR rules?
GDPR stands for the General Data Protection Regulation. In the UK we follow the GDPR rules and have a law called the Data Protection Act. All research using patient data must follow UK laws and rules.

Universities, NHS organisations and companies may use patient data to do research to make health and care better.

When companies do research to develop new treatments, they need to be able to prove that they need to use patient data for the research, and that they need to do the research to develop new treatments. In legal terms this means that they have a 'legitimate interest' in using patient data.

Universities and the NHS are funded from taxes, and they are expected to do research as part of their job. They still need to be able to prove that they need to use patient data for the research. In legal terms this means that they use patient data as part of 'a task in the public interest'.

If they could do the research without using patient data, they would not be allowed to get your data.

Researchers must show that their research takes account of the views of patients and ordinary members of the public. They must also show how they protect the privacy of the people who take part. An NHS research ethics committee checks this before the research starts.

What will happen to the results of the research study?
We plan to publish the results of the research in medical and scientific journals and meetings during or at the end of the study. No participants will be identified.

Who has reviewed the study?
A Multicentre Research Ethics Committee has reviewed this research.

What are the arrangements for compensation?
The project has been approved by an independent Research Ethics Committee who believe that it is of minimal risk to you. However, research can carry unforeseen risks and we want you to be informed of your rights in

the unlikely event that any harm could occur as a result of taking part in this study.

No special compensation arrangements have been made for this project, but you have the right to claim damages in a court of law. This would require you to prove fault on the part of the Hospital and /or any manufacturer involved.

If you are unhappy or wish to make a complaint about the care you receive from the NHS, PALS can help: more details are at http://www.gosh.nhs.uk/parents-and-visitors/clinical-support-services/pals/, or telephone them on 020 7829 7862.

Contact for further information.
Please contact a member of the research team on the email addresses shown below for further information. Thank you for taking part in this study. This is your copy of the information sheet.

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