PARENT INFORMATION LEAFLET
NIHR BIORESOURCE - RARE DISEASES

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Many people in the UK are said to be living with a rare disease. Living with a rare disease can have a major impact on a person's quality of life and on their close relatives. There are an estimated 7000 different rare diseases and most of these are inherited. We would like to invite you and your child to join the NIHR BioResource - Rare Diseases (NIHRBR-RD). Please take the time to read the following information carefully and discuss it with others if you wish. Ask us if there is anything that is not clear, or if you and your child would simply like more information.

What is the purpose of the NIHR BioResource - Rare Diseases?
The BioResource has been establishing a panel of thousands of volunteers with and without health problems. It is now being expanded to include rare diseases. Volunteers will be asked to donate a small blood or saliva sample and give consent to be contacted and invited to participate in medical research studies on the basis of data gathered from samples and information they have supplied..

The two immediate aims of the NIHRBR-RD are (1) To develop more affordable DNA-based tests for the diagnosis of rare diseases for which the gene is known and (2) To discover genes causing rare diseases; only half of the genes for rare diseases are currently known. Information and samples from the BioResource will be made available to researchers and doctors working in medical research in both the public and private sector, in the UK and overseas.

Why is this important?
Discovering genes causing rare diseases is the start of a new journey. Accurate tests for rare diseases can be developed to obtain more rapid diagnosis. This is important for selecting the best care and possible treatment, but also to provide accurate information to the wider family about risks to other individuals. Secondly, once the gene causing a rare disease has been identified, the search for better treatments can start. This is not always successful, but for several rare diseases new treatments have already dramatically improved care, giving hope that this will extend to many more in the future.

Why has our family been invited to join the NIHR BioResource - Rare Diseases?
To support research for rare diseases, doctors, nurses and researchers are inviting thousands of people affected by rare diseases across the UK to join as volunteers together with members of their family. Your child's doctor or another member of their clinical care team at their hospital or GP surgery have agreed to join the BioResource initiative on rare diseases.

Do we have to join the NIHR BioResource - Rare Diseases?
It is completely up to you and your child to decide whether either or both of you wish to join. If you, your child or both of you decide not to join, your decision will not affect the healthcare you and your

child receive in any way. You, your child or both of you will be free to withdraw at any time and without having to give a reason.

What will happen if my child and I agree to join the NIHR BioResource - Rare Diseases?
If you, your child or both of you agree to join, we will ask you to sign two consent forms, one for yourself and another for your child. Children between 6 and 15 years of age will be asked to sign an assent form. You and your child would be asked to donate a small blood sample (one to three teaspoons depending on your child's age, 3-4 teaspoons for you) or saliva sample, which would be used for different research tests. You will also be asked to provide contact details for all participants (including email and mobile phone if available) and answer a questionnaire about you and your child's health and lifestyle. With your permission further relevant information about you and your child's health may be retrieved from medical notes and other records held on databases.

What will happen to the samples I give?
We will isolate, analyse and store you and your child's DNA and other components from the donated samples for use in medical research. We may measure a range of chemicals in these samples and may determine the genetic code of you and your child.

Genes are made out of DNA. RNA is the version of the DNA code that the body uses to direct how proteins are made. We may determine the DNA/RNA code of the samples taken. This may include determining the sequence of all or part of the DNA code of you and your child.

What will happen next?
All the information provided by you or your child, or retrieved from you and your child's medical notes or other health records and the results of tests performed with the donated samples will be held on a research database for use in medical research. In the future a number of studies will be carried out and we may contact you, to ask whether you or your child want to take part. An invitation for further medical research studies will be on basis of the data held about you and your child on the research database.

You and your child will be provided with full information regarding each of these studies and will be free to decide whether or not to participate.

How often will my child and I be contacted?
We closely monitor the number of times your child and you are approached and invited to studies. The maximum number of invitations to studies will be 4 each year. We greatly appreciate the effort made by volunteers and are happy to contribute towards travel / parking costs incurred by volunteers participating in studies.

What are the risks and disadvantages of joining the NIHR BioResource - Rare Diseases?
Joining will involve donating a small sample of blood or saliva. Experienced staff will collect the blood. The taking of a blood sample has a small risk of bruising, inflammation or fainting and there may be some discomfort. Saliva samples can be collected by yourselves in your home.

What are the benefits of joining the NIHR BioResource - Rare Diseases?
There will be no direct benefit to you or your child by joining but you will make a contribution to science and future improvements in NHS care, particularly for patients with rare diseases such as the one in your family.

Will the details about my child and me be kept confidential?
Yes. Best ethical and legal practice will be followed to ensure that all information collected will be handled in confidence. Samples will be labelled with a unique sample study number before being banked and information from genetic and other tests will be linked to this unique number but stored separately from the personal details of your child and you. The database linking unique sample study numbers to personal details will only be accessed by authorised members of the BioResource team who do not have access to the results of the genetic and other tests. Information from these tests will not be used or made available for any purpose other than for research and improvements in health care. Neither you nor your child will be identified personally in any report or publication, including information about BioResource studies which will be released on the Internet.

On occasions we may ask you and your child for separate written consent to contribute a personal story for education purposes. Telling a personal story may be of enormous value to other families affected by rare diseases and to increase awareness in society about rare diseases in general. It is up to you and your child to decide how much, if any, additional information you wish to give.

Can my child and I know the results obtained from the study samples?
It is not planned to routinely feedback the results from genetic or other tests obtained from the donated samples. However, if the research does identify a cause of the rare disease in your family with your permission we would let your doctor and your clinical care team know.

All research results that are identified will need to be confirmed in an accredited diagnostic laboratory before being used in the clinical management of you and your family members. We hope that the NHS will make reasonable efforts to introduce new tests for rare diseases. We will support these efforts where possible by sharing the results of our studies.

Please be aware that the government has extended the genetic test insurance moratorium until 2017. This means there are restrictions which prevent providers from using genetic test results to deny people insurance cover until that set date.

What if my child or I or both of us no longer want to be a member of the NIHR BioResource - Rare Diseases?
Volunteers are free to withdraw from the NIHR BioResource - Rare Diseases at any time without giving a reason. If your child or you choose to withdraw, then you and/or your child will not be contacted again and stocks of the linked samples held at the BioResource will be destroyed. It will not be possible to destroy samples already prepared for testing or to withdraw samples that have been

distributed to other laboratories with whom the BioResource team collaborates. It will not be possible to delete information obtained from your samples or health records from the research databases or laboratory notebooks. However, no new data will be added to the research databases or notebooks from the moment the BioResource team has confirmed your withdrawal. Your personal details will not be held in the research database or laboratory notebooks at any time.

What happens if a discovery is made using the donated sample?
The samples donated to the NIHR BioResource - Rare Diseases are given as an "absolute and non-returnable gift", i.e. without receiving a payment and without conditions. For example if results from the research undertaken with the donated samples are used to develop a new blood test to improve diagnosis or better medicines for treatment, then you or your child will not receive any compensation nor will funds be forthcoming to either of you. The BioResource team will work in partnership with others in the public and the private sector (e.g. pharmaceutical or biotechnology industry, etc.) to successfully develop any discoveries for the benefit of patients.

What will happen to the results of the research study?
To speed up developments of new diagnostic tests and better treatments, results of the studies will be made available to the public through scientific publications, including placing information on the Internet, in press articles and in project leaflets. This information may include part or the entire DNA code of you and/or your child or the results of other tests performed with you or your child's samples and other relevant information from the research database, e.g. age of your child in years, gender, the type of Rare Disease, etc. Under no circumstances would identities be disclosed in any publication, although BioResource will be identified as the source of the material.

Who funds and sponsors the NIHR BioResource - Rare Diseases?
The BioResource is currently funded by the NHS National Institute for Health Research (NIHR) and jointly sponsored by Cambridge University and Cambridge University Hospitals NHS Foundation Trust. This study has been reviewed and approved by Cambridgeshire South Research Ethics Committee.

Further information
If you or your child want more information before deciding, or have any queries about anything concerning the NIHR BioResource - Rare Diseases, please feel free to contact the BioResource team on freephone 0800 0853650 or e-mail us on rarediseases@bioresource.nihr.ac.uk

Thank you to you and your child for considering joining the NIHR BioResource - Rare Diseases.