NHS

NHS
National Institute for
Health Research

BioResource for Rare Diseases

(part of the NIHR BioResource)

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Patient Information Leaflet

27/04/2012 Version 1

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 to join the BioResource for Rare Diseases. Please take the time to read the following information carefully and feel free to discuss it with your family or close friends and ask us if there is anything that is not clear, or if you would simply like more information.

Further information

If you want more information before deciding, or have any queries about anything concerning the BioResource for Rare Diseases, please feel free to contact the BioResource team on freephone **0800 0853650** or e-mail us on **rarediseases@nihrbioresource.org.uk**.

What is the purpose of the BioResource for 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. These 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. Information and samples from this resource may also be made available to other scientists working in biomedical and healthcare research.

> **The two immediate aims of the BioResource for Rare Diseases are (1) To develop more affordable DNA-based tests for the diagnosis of rare diseases for which the gene is known (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 have I been invited to join the BioResource for 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. Your doctor or another member of the clinical care team at your hospital or GP surgery have agreed to join the BioResource initiative on rare diseases.

Do I have to join the BioResource for Rare Diseases?

It is completely up to you to decide whether you wish to join. If you decide not to join, your decision will not affect the healthcare you receive in any way.

You will be free to withdraw at any time and without having to give a reason.

What will happen if I agree to join the BioResource for Rare Diseases?

If you agree to join, we will ask you to sign a consent form. You would be asked to donate a small sample of blood (3-4 teaspoons) or saliva, which would be used for different research tests. You will also be asked to provide your contact details (including email and mobile phone if available) and answer a questionnaire about your health and lifestyle. With your permission further relevant information about your health may be retrieved from medical notes and other records held on databases.

What will happen to the samples I give?

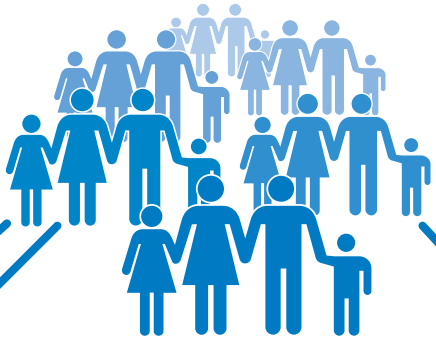
We will isolate, analyse and store your DNA and other components from the donated sample for use in medical research. We may measure a range of chemicals in these samples.

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 your DNA code.

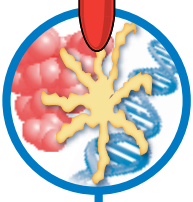
What will happen next?

All the information provided by you, or retrieved from your medical notes or other health records and the results of tests performed with your 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 want to take part. An invitation for further medical research studies will be on the basis of the data held on the research database. You will be provided with full information regarding each of these studies and will be free to decide whether or not to participate.

Study Design



Take blood sample



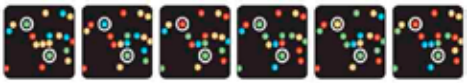
Isolate cells, protein and DNA

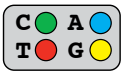
MEDICAL NOTES

Sample donation and data assesment

Samples in the BioResource for Rare Diseases and data in the research database will have a unique study identifier, and will not be labelled with your name or contact details.

Compare genetic alphabet



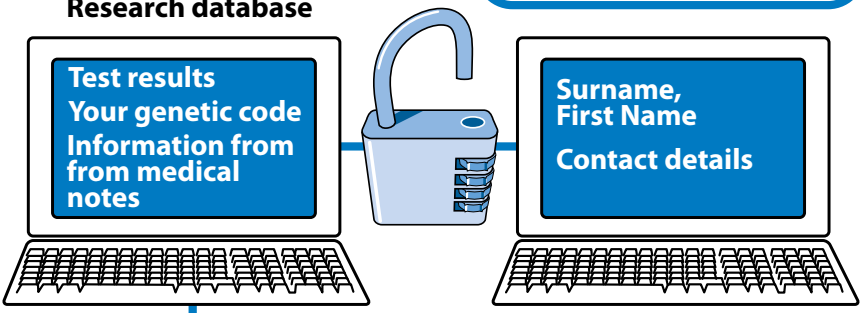


Top: CATCGT
Bottom: CCCCCC

Analysis and storage of results

Research data will be securely stored in a database. Personal data such as your name and contact details will be kept strictly confidential and only authorised BioResouce people will have access.

Research database



Published data released on e.g. the Internet

How often will I be contacted?

We keep a close eye on the number of times 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 BioResource for 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 you in your home.

What are the benefits of joining the BioResource for Rare Diseases?

There will be no direct benefit to you by joining but you will make a contribution to science and for future generations.

Will the details about 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 your personal details (surname, first name, contact details). 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. You will not be identified personally in any report or publication, including information about BioResource studies which will be released on the Internet, newspapers and other media.

On occasions we may ask you 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 to decide how much, if any, additional information you wish to give.

Can 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 the BioResource studies.

Please be aware that the government has extended the genetic test insurance moratorium until 2014. This means there are restrictions which prevent providers from using genetic test results to deny people insurance cover until that set date.

What happens if a discovery is made using the donated sample?

The samples donated to the BioResource for 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 will not receive any compensation nor will funds be forthcoming to 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 project leaflets. This information may include part or your entire DNA code or the results of other tests performed with your samples and other relevant information from the research database, e.g. your age in years, your gender, the type of rare disease, etc. Under no circumstances would identities be disclosed in any publication, although the BioResource will be identified as the source of the material.

What if I no longer want to be a member of the BioResource for Rare Diseases?

Volunteers are free to withdraw from the BioResource at any time without giving a reason. If you choose to withdraw, then you 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 medical 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 (surname, first name, contact details) will not be held in the research database or laboratory notebooks at any time.

Who funds and sponsors the BioResource for 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 2 Research Ethics Committee.

Thank you for considering joining the BioResource for Rare Diseases.