100,000 Genomes Project

Information sheet

You have been invited to take part in the 100,000 Genomes Project because you have a rare condition or disease for which no clear cause has yet been found. Or, we are asking you to take part because you are a close relative of someone who does have one of these conditions.

This leaflet gives you important information you will need to think about when deciding whether to take part in the project. If you want more detailed information about something, you can ask at any time. You can also find extra information, including videos, on the project website at **www.genomicsengland.co.uk**. This leaflet is also available in other formats.

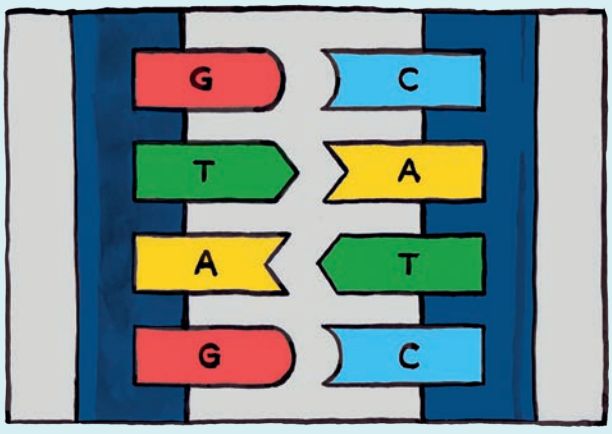
You can take as much time as you need to think about joining the project. Only you can choose if you want to take part. If you don't want to join, you don't have to say why. And, if you don't join, you will still receive the best available care.

Introduction

Your genome

The best way to explain your genome is to say it is your body's 'instruction manual'. It contains nearly all the information needed to make you, run you, and repair you. You have a copy of your genome in almost every cell in your body. You inherited it from your parents.

Your genome is made of a chemical code called DNA. There are 3 billion 'letters' of code and these can be 'read' one at a time, to produce your unique DNA sequence.



You may have already had a genetic test looking for specific changes in genes. Genes are bits of the DNA code that give instructions for specific processes in your body. But genes only make up a tiny part of the DNA in your genome. The rest of your genome's DNA is important too. It may help diagnose or understand a condition. That's why we're offering you **whole** genome sequencing.

About the 100,000 Genomes Project

We are sequencing 100,000 genomes from families affected by rare conditions and from people with cancer. We will link the results to your medical and other records and store them securely.

We aim to:

- provide a diagnosis for some patients with rare diseases;
- learn how to improve cancer treatment;
- make new discoveries that will help us understand why some people get ill and others don't;
- develop future NHS healthcare services; and
- support healthcare professionals and researchers to develop new medicines and tests.

What do you do and what could the project do for me and my family?

We are hoping that some people with a rare genetic condition may receive a diagnosis for the first time. But many will not.

For some conditions, we are more likely to find the cause of a condition if we compare the affected person's genome with their blood relatives' genomes. This is because people in the same family have some parts of their genome in common. So we may ask several people in a family to join the project.

To look for a diagnosis, we will analyse each participant's DNA sequence together with information about their health.

We put all your data together with that of thousands of other people, including others like you. Being able to compare all this data is very useful. Researchers look for patterns that might give new clues about the cause of people's conditions, or patterns that might help them understand a problem better or find a new way to treat it. And they carry on looking for many years to come.

We keep track of what happens to your health for the rest of your life. This means we and those who work with the project can continue to study data even after your death, with the aim of benefiting future healthcare and research.

Knowing what happens to you over the course of your life is critical. For example, some people with the same condition may find it gets worse as they get older, but others may not. If we know how things turn out for people and what they eventually die from, we might be able to link it with particular changes in the genome. In the future, this might help predict or prevent diseases.

Are there any risks?

Some people worry about being identified as someone taking part in the project. The chance of this happening is very small, and we will do everything we can to prevent this from happening.

Taking part

Important points

Some people with a rare condition may get a genetic diagnosis for the first time, but many will not. At the moment, the main benefit will be for people in the future who have similar conditions to you (or your family member).

What will I be invited to do?

If you decide to take part, we will ask you to:

- read and fill in a form giving your permission to take part (consent form);
- give some blood samples (up to three tablespoons), or saliva or other sorts of sample; and
- allow us to collect, store and analyse health and personal information about you.

If you have any questions, you can ask your clinical team.

Your samples

This relates to section 2 on the consent form.

- We will take DNA from your sample. We will then send some of the DNA for whole genome sequencing. This is usually done in England but, rarely, it can be sent overseas.
- We will store the rest of your DNA and any other samples in a secure 'bank' called a bio-repository. This is in the UK.
- We label your samples with a unique barcode instead of your name. We use the code to keep track of your samples.
- Labs we have approved may analyse your samples. They look for clues to help understand more about how genes work. There may be new ways of doing this in the future and the results will go into our data centre.
- Your samples could be stored for many years. If we use them up, we might ask you for more. If you don't want to give more, you don't have to.
- Very rarely, a DNA sample can't be sequenced. If this happens, your clinical team will explain why.

Your data

This relates to section 3 on the consent form.

Important points

We will collect electronic copies of all your past and future health records for the project. Your clinical team will send information about your rare genetic condition (if you have one) to the project.

- Because any of your data could benefit approved research or future healthcare, we are asking to access electronic copies of all of your records from the NHS, your GP and other organisations. This includes information about any illnesses or stays in hospital – even ones that you may not think are related to the rare condition in your family.
- The data are from different sets of records, including hospital or clinic records, medical notes, social care and local or national disease registries. The data includes images from your NHS records, such as MRI scans, X-rays or photographs.
- We don't remove any originals from your records. These will stay in the NHS.
- We will continue to access all your updated records throughout your lifetime, for as long as you are in the project. This includes information added after your death.
- If you want to know more about what we access, visit our website www.genomicsengland.co.uk/the-100000-genomes-project/data/.
- We may use your data to study many different medical conditions, not just ones that affect you.
- Only your clinical team and people who are involved in the project have your name and other personal details. We need these so we can return your results to you. We may let your GP, or other medical staff who look after you, know you are taking part.
- We at Genomics England protect your data and control who has access to it.
- We will own all the data from this project.

Access for researchers and organisations – including commercial companies

This relates to section 3 on the consent form.

Important points

We can only look at your data for approved scientific and healthcare purposes. Before researchers see data, we 'de-identify' it. This means we take out all names, dates of birth, NHS numbers and other personal details. The researchers may be from not-for-profit organisations, as well as commercial organisations (those whose aim is to make a profit).

An important part of the project is making sure that discoveries get turned into treatments and tests for people with rare conditions as quickly as possible. New drugs and diagnostic tests are developed by companies worldwide, not by the NHS. Almost all of these are commercial companies. Other companies need to use the data to try and find new, faster ways to analyse large amounts of data.

- Approved researchers, including from companies worldwide, can access your de-identified data.
- Approved researchers from charities, universities or hospitals can also access your de-identified data.
- We will not allow access to any data for marketing or insurance purposes.
- Researchers may publish the results of their research in medical journals. They may also present their results at scientific meetings. It is important for scientists and doctors to share results to help research advance as quickly as possible. You will not be identified in any of this.
- Companies are charged to work with us at Genomics England. These charges contribute to the cost of the project. If we make any profit from this, it will go back to the NHS.
- You will not benefit financially if a product or test is successful because you have been involved in the project.

How will you keep my information secure and confidential?

- Your data is stored in a secure data centre in the UK. Its security level meets national data standards. Researchers can only look at de-identified data inside this monitored environment.
- Researchers can only take away the answers to their questions (their results). They can't copy or take away any individual data or images.
- Nobody can access the data without asking us first and we will carry out checks on them. They also have to say exactly what they would like to do.
- We have an advisory group who will assess each request. The group includes patients and participants in the project. We then decide who gets access to the data.
- Our website lists the companies or organisations approved to access data, and for what purpose **www.genomicsengland.co.uk/the-100000-genomes-project/data/**.
- We will monitor researchers looking at your de-identified data to check that they're doing what they asked to do and no more.
- We will put the results of research on our website to show how the project data is being used.
- When researchers publish their research, this is still de-identified. But if you have an extremely rare condition affecting just a few people, someone may be able to work out who you are.
- If anyone reveals your data on purpose in a way that identifies you, it is a legal breach (in other words, they have broken their contract with us or they have broken the law). Any person, institution or company that does this could face criminal charges or substantial fines. They could have their research funding stopped. We would also ban them from accessing the project again.

Can I get hold of my data?

Yes. You can ask for a copy of your data. There may be a charge for this. For more information, speak to your healthcare professional or see our website at **www.genomicsengland.co.uk**.

Will taking part affect my insurance?

You don't have to tell an insurer that you are part of the project, or about your results. They won't have access to your results. If you have any medical treatment or a diagnosis, you should tell your insurer about these if they ask.

Getting your results

This relates to section 4 on the consent form.

Important points

Your genome will be investigated for possible causes of the rare genetic condition in your family. You will get these results, even if nothing is found. Your clinical team will give you the results, as they would for other test results. The results might take many months to come back.

Even if no cause is found at first, we will keep looking. We will continue to study your genome, so results may come back some time in the future. Your clinical team will tell you about any results from further research. This means you might get initial results and then some more later. Or, you may get no results at first, but something later on. We cannot tell you how long this might take.

- After your genome is sequenced, the results are sent to your clinical team for checking.
- You may be called for an appointment to discuss the results, or you may receive a letter.
- If we find something which could be important for the health of your family, your clinical team will let you know and provide support if you want, so you can tell your family members what they need to know.

- We won't tell you personally every time your data is studied, but your clinical team will tell you if something is found which might be important for your health.
- Sometimes genome tests reveal that people may not be related by blood as they expected. Your clinical team won't tell you this, unless it has serious implications for someone's health.

What other results can I ask for?

Additional Findings

This relates to section 5 on the consent form.

Important points

You can choose if you want us to look in your genome for genetic changes that could be signs of a small number of other conditions. These conditions could be serious but the NHS can already treat them or use screening to pick them up at the earliest stage possible. We call these 'additional findings'. We will report on these results separately to your clinical team, later than the findings about your rare genetic condition. We may change which conditions we look for.

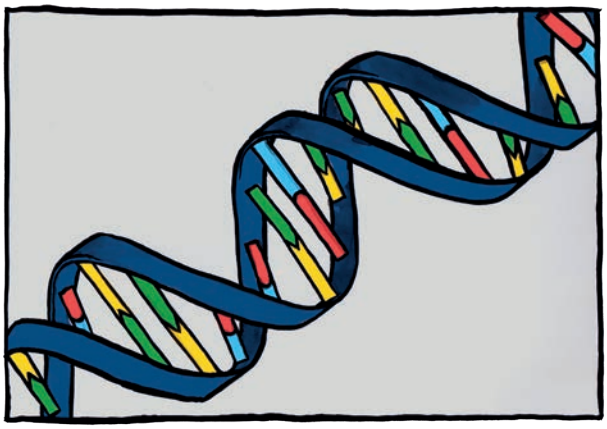
- The diseases we look for are uncommon, and the chance of you having one of them is low.
- As the project continues, we hope to understand better who will benefit from treatment or screening.
- If we find a result pointing to one of these health conditions, your clinical team will give you advice and support. You may not need treatment for the condition until you are older. You may never go on to develop it at all.
- However, you could still develop one of these conditions even if we find no relevant results.
- This is because we do not yet know whether genome sequencing will pick up all relevant genetic changes. We should know more about this by the end of the project.
- As we learn more about how to use genome sequence information over the life of the project, we will regularly update the specific conditions which we will look for. So you could get further reports about different conditions in the future. More details are available on our website.
- If you don't want any additional findings, we won't look for them in your genome.

Carrier testing

If you are not planning to have children in future, this section is unlikely to be relevant to you. You can move to the next section.

Healthy people can 'carry' genome changes that could affect the health of future children. These genome changes increase the risk that children or grandchildren may be born with a serious medical condition. You can tell us if you want us to look for these. This is called carrier testing. We may add to or change which conditions we look for. Carrier testing results are unlikely to be relevant to your own health.

- The chance that you are a carrier for one of these conditions is low.
- If you are found to be a carrier, we will give you more information about the condition and how this might affect your future children or grandchildren.
- Some of the things we may look for only increase the risk of a baby having the condition if your partner is also a carrier. We will only look for these conditions if your partner is also taking part in the project and you both decide you want to find out about these.
- If you have children in the future with a new partner, these results are not relevant as your new partner will not have been tested.
- For other conditions that you may carry in your genome, we can look at your sequence alone, without your partners.



- We will regularly update the specific conditions which we will look for, so you could get further reports about different conditions in the future. More details are on the website.
- If you don't want any carrier testing results, we won't look for them in your genome.

Can I change my mind about having additional findings or carrier testing?

Yes. You can change your choices at any time by filling in an 'opt-in or opt-out form'. These are available from your clinical team or from our website at **www.genomicsengland.co.uk/taking-part/patient-information-sheets-and-consent-forms/**.

Apart from these additional findings and carrier testing no other information will be looked for or reported.

Future contact

In the future, your clinical team or the Genomics England project team may contact you. This could be to ask you for more information. Or to invite you to take part in future research, including clinical trials of new medicines. Or to ask you for your views on the project. It is up to you whether you agree to take part in these studies. We may also send you information about the progress of the project.

Leaving the 100,000 Genomes Project

This relates to section 1 on the consent form.

To leave the project at any time, ask your clinical team for a 'withdrawal form'. Fill this in and return it to them. There are two options for withdrawal, as shown below. You can get more information about these from your clinical team or in the withdrawal form.

1. No further contact, but continue to include my existing samples and information in the project.
2. No further contact and no further use of my samples or information.

What happens if I am no longer able to give permission to take part in the project?

In the future it is possible that you may become unable to make decisions for yourself. This might be if you become very unwell.

If you live in Northern Ireland:
The law says that the permission (consent) that you gave originally will always apply.

If you live in England:
The law says that the permission (consent) that you give to join the project is no longer valid.

If your clinical team believe that you can't make decisions about this project, at the time these are needed, they can look for someone to advise on your behalf. This could be a friend or family member. If the person believes:

- that you would still want to take part in the project, we will arrange this; or
- that you would **not** want to take part, you will stop being a participant. We will not ask for any more samples or information from you or your records. You would also stop being a participant if the clinical team can't find someone to advise for you. If you stop, information that has already been collected about you up until that point will still be available for researchers in the future. This is in line with your original consent.

Your clinical team will be happy to answer any questions you have.

Please share this leaflet with your family and friends if you want to.

Thank you for considering taking part in the 100,000 Genomes Project.

Insert local GMC contact details here
Phone number:
Email address:
Hospital address: