Insert GMC logo here

Genomics england

Introduction to the 100,000 Genomes Project

This leaflet covers the major aspects of the Project which you need to think about when considering whether to take part or not. It does not cover everything and you should also read the patient information sheet that you will be given. If you have any questions, your clinical team will be able to help you.

The 100,000 Genomes Project

What is the 100,000 Genomes Project?

This Project aims to sequence 100,000 whole genomes from NHS patients (and their relatives) by 2017, to help patients, to put genomic medicine services in place for the NHS, to do research and to develop new treatments. The Project is run by Genomics England, which is a company owned by the Department of Health.

What is a genome?

Your genome or genetic code is your body's instruction manual containing the information needed to make you, run you and repair you. It's made of DNA, which can be read letter by letter using a technique called sequencing.

Looking at the genome of a person affected by a rare disease can help find which DNA changes might be causing the problem. This can be especially helpful when you can also compare that person's genome with the genomes of some of their blood relatives.

For people with cancer, the cancer cells have developed a different genome to the healthy cells. Comparing the two genomes may give clues about possible ways to treat the cancer.

Putting together information about your genome with details of your illness or condition (your health data) and then analysing it, may give

your medical team helpful information about your condition.

Who can take part?

There are several separate parts of the Project, and different people will be able to join these:

1. Those with a rare disease, and often some of their blood relatives
2. Those with certain cancers
3. Those who have had a severe reaction to an infection

You may also be reading this because you are the parent of a child who could join the Project, or if you have been asked to represent a person affected by a rare disease or cancer.

What is involved?

If you consent to take part, you will need to agree to:

- **Give samples of your blood. If you have cancer, you will also need to donate a small piece of your cancer when you have a biopsy or operation.**
- **Allow information about your health, including scans and other test results (your health data) to be sent to Genomics England to help in the analysis of your condition.**
- **Allow your genome sequence data and your health data to be linked to relevant parts of your medical records, including information about your health in the future.**
- **Agree to be contacted in the future by your clinical team and by Genomics England.**

What results could I get?

If you consent to take part, you will need to agree to:

- **Have results about your main condition fed back to you**

This may take about six months, but could take longer. Results might not be back in time to affect any treatment you may need. There is a chance your 'result' might not contain any helpful information. However we will continue to work on your genome, as long as you let us. If we find anything we think is important we will always tell your clinical team.

- **Decide what you feel about Genomics England looking for 'additional' conditions**

Examining your genome can identify some serious but potentially treatable genetic conditions such as rare cancers or heart disease. Most people won't have them, but if you are the sort of person who would like to know if we find something, we can tell you. We're still learning how to use the information in your genome, so what we think is important now may change, and we won't be able to spot everything now. If you are the sort of person who doesn't want to know, Genomics England won't look.

- **If you might have a baby in the future, decide what you feel about 'carrier testing'**

You will be offered a test to look for conditions that don't affect you, but which might be a problem for your baby if you were found to be carrying them in your genome. This is called carrier testing.

Involving your family

If there is a finding which might have implications for other members of your family, the NHS will support you to tell them about it and will offer them any tests, advice or treatment they need.

Use of your data

If you consent to take part, you will need to agree that:

- **Researchers and organisations approved by Genomics**

England can look at your data in a way which protects your identity, including for-profit healthcare companies like those developing medicines or diagnostic tests

Who looks at my data and why?

Your sequence data and your health data are sent to the Genomics England data centre by a secure NHS system. The data centre is within a high security site. The data will be de-identified (that's where everything that might identify you has been removed) and stored in the Genomics England data centre along with data from tens of thousands of other people. Approved researchers can then look at the de-identified data, but they can't take any of it away.

Your data can still be linked back to your name and date of birth by the Genomics England Project team, in order to provide a clinical report which may guide your care. If anything is found which could be useful for your care, even if it years in the future, this information will be passed to your clinical team so they can tell you about it.

Comparing data from many people can give new understanding about the cause of a disease and how to treat it. Drugs have never been developed by the NHS – this is done by for-profit companies. This means that companies that make drugs or diagnostic tests need to use the data, because without this patients won't get the new medicines and treatments they need. Agreements will be put in place to make sure that the NHS receives some benefit from successful discoveries.

What about my privacy?

We make it as hard as possible to identify you from your data, but we can't rule it out. Trying to find out who data belongs to is illegal and could lead to a prison sentence. Genomics England monitors and records everything scientists do with the data, so re-identification is very unlikely.

What about insurance?

You don't have to tell anyone you have taken part in the 100,000 Genomes Project or reveal the results. However, if you have treatment or tests after you get your result, you may be asked for this information if you are taking out an insurance policy, and you will have to declare this in the normal way.

What if I agree and then decide I don't want to be in the project any more?

You can withdraw from the Project at any time. This will not affect your care in any way.

Thank you for considering taking part.

If you think you might be interested in joining the Project, please ask your clinical team to give you the relevant participant information sheet, to give you more specific details about the part of the Project that is relevant for you or your family member.

The 100,000 Genomes Project will create a legacy for generations to come.

INSERT LOCAL CONTACT DETAILS/ LABEL HERE

Genomics England runs the 100,000 Genomes Project. It is a company owned by the Department of Health. For further information please visit www.genomicsengland.co.uk