100,000 Genomes Project

Participant Information Sheet

This information sheet describes the Project and key points that you need to consider when deciding whether or not you want to give consent for your child to take part. If you don’t want your child to take part in this Project, they don’t have to. You don’t have to give a reason for your decision and their care won’t be affected if you say no. If you are joining this Project as a participant yourself, information will be provided to you separately about your own participation as a family member. This sheet focuses on your child’s potential participation.

1. INTRODUCTION

About the 100,000 Genomes Project

Genomics England is creating a lasting legacy for patients, the NHS and the UK economy through the sequencing of 100,000 genomes, all with the consent of participants. This is known as the 100,000 Genomes Project. The main goal of the Project is to transform the application of genetics to healthcare in the NHS. The Project is run by Genomics England, a company owned by the Department of Health.

Your child’s genome

The genome is all of the genetic information in the body’s instruction manual. Some of your child’s genome is unique to them, but much of it they share with their relatives. The genome is made of DNA, which is like a series of ‘letters’ that can be looked at using a technique called sequencing. Recent developments in science and technology mean it is now possible to sequence the whole genome.

What can sequencing a whole genome tell clinicians and researchers?

Learning more about genomes can identify the cause of genetic diseases and may help to improve treatments. When the genome sequences of patients with the same condition are compared, it is possible to see patterns. These patterns can be put together with health information about everyone taking part. Once this is done we may be able to link particular patterns with whether people become ill and how severe their illness is likely to be. Sometimes these patterns can also give clues as to the best treatments for a patient, or may reveal information that could be important to other family members.

Why is the 100,000 Genomes Project being done in the NHS?

Fast, low-cost, whole genome sequencing is now affordable for the NHS. Using whole genome sequencing for the benefit of patients is called ‘genomic medicine’. Although genomic medicine is already happening in the NHS on a small scale, we want to develop the evidence that may expand its use across healthcare.

What is the 100,000 Genomes Project planning to do?

This Project will sequence 100,000 whole genomes, half in people with rare genetic diseases and half in patients with cancer. The results will be linked with patients’ medical records and stored securely. By combining this information and allowing authorised researchers to access it, the Project aims to: Provide a diagnosis for some patients with rare diseases.

- Provide a diagnosis for some patients with rare diseases.
- Learn how to adapt cancer treatment by looking at the genome of the tumour.
- Make new discoveries that will help us understand why some people get ill and others don’t.
• Develop a genomic medicine service for the NHS.
• Support clinicians and researchers in hospitals, universities and companies of all sizes to develop new medicines, therapies and diagnostic tests.

2. TAKING PART

Why has my child been invited to join the 100,000 Genomes Project?
Your child has been invited to join the 100,000 Genomes Project because they or a close relative have a rare disease or condition for which a cause or explanation has not yet been found. We may invite some of your child’s blood relatives to take part, as it is often helpful to compare the genomes of the affected person and their relatives.

Key point
Some patients with rare diseases may get a genetic diagnosis for their rare disease for the first time but many will not. It is most likely that, for now, the main benefit will be for patients with similar conditions in the future.

What will my child be invited to do?
After the Project has been explained, and you have had as much time as you need to consider whether you would like your child to take part, you will be asked to:

• Read and fill in a consent form.
• Have your child donate blood samples (up to 3 tablespoons of blood); occasionally other samples such as saliva might also be requested.
• Give your child’s personal details, contact details and health information to the Project. Your family’s contact details are confidential to the clinical team and the 100,000 Genomes Project team and will not be shared with anyone else. More information about what information will be collected and how this will be used is given in section 4.

Your child’s GP and any other doctors involved in their care may be informed that they are taking part. You can ask your child’s clinical team questions about the Project at any stage.

3. SAMPLES

What will happen to my child’s samples after we have donated them to the 100,000 Genomes Project?
This relates to points 3 and 4 on the consent form.

• DNA will be extracted from your child’s sample for whole genome sequencing. This is most likely to take place in England, but it could occasionally be done overseas.
• We will also take blood samples from your child to allow us to do other tests to understand how genes work. In future, we may be able to do new tests on the samples (tests that are yet to be developed).
• Your child’s DNA and other samples will be stored by Genomics England in a secure ‘bank’ called a bio-repository. This is located in the UK.
• Instead of your child’s name being used to label the sample(s), they will be labelled with a unique code number before they are sent to the bio-repository. This means your child cannot be directly identified from the sample(s).
• Your child’s samples could be used up soon and then their clinical team may ask for more, or their samples could remain stored for many years. You don’t have to agree to your child giving more samples if you are asked.
• There may be certain circumstances where your child’s DNA will not be sequenced. If this happens, their clinical team will explain why.
4. ACCESS TO MY CHILD’S DATA AND CONFIDENTIALITY

What information are you asking my child to donate to the Project?

This relates to point 5 on the consent form.

**Key point**
You are agreeing that past medical records, as well as current and future information about your child’s health can be collected by the 100,000 Genomes Project. You are agreeing that this information can be studied indefinitely.

You are being asked to:

- Allow information about your child’s health to be sent by their clinical team to the 100,000 Genomes Project.
- Allow the Project to obtain health-related information and past medical records that span your child’s lifetime, including current and past illnesses or stays in hospital. This could include medical records held by the NHS, your child’s GP, and other bodies like local and national disease registries. (Registries are collections of information about patients with a particular condition.)
- Allow the Project to continue to access your child’s health records electronically into the future, so we can update the medical records that we hold on them over the rest of their life. This will include receiving information from their clinical team charting the course of any illnesses they may have. The data we keep for the Project will be in the form of electronic copies and not your child’s original records which will remain in the NHS.
- Allow researchers to view (but not take away) any digital images that are collected from your child’s health records, such as MRI scans and photographs. These images when combined with other data and your child’s genome sequence can help to identify the cause of a rare disease.
- Allow your child’s data to continue to be studied throughout their lifetime as well as after their death, to collect as much useful information as possible, for the benefit of future healthcare and research.

What will happen to my child’s data after we have shared it with the Project?

This relates to points 5 and 6 on the consent form.

**Key point**
The 100,000 Genomes Project links your child’s genome sequence with their medical records and information about their health. Researchers will only have access to this data if they are approved by an Advisory Committee to Genomics England. All the information is held securely, and will only be used for scientific and healthcare purposes.

- You are agreeing that your child’s healthcare information can be linked to his or her sequence data and stored to be compared with many thousands of other sets of information donated by other participants.
- Your child’s data may be used to study many different medical conditions, not just the rare condition that led to your family taking part, or future conditions that your child might go on to develop.

Who will have access to my child’s data?

This relates to point 7 on the consent form.

- To get the most value for healthcare from the information your child has donated, it needs to be accessible worldwide, in a form which protects your child’s identity.
- Genomics England will put the names of all the companies or organisations that have been approved to access data and for what purpose, on the 100,000 Genomes Project website: [www.genomicsengland.co.uk](http://www.genomicsengland.co.uk).
- Access to any of the data held by Genomics England for marketing or insurance purposes will not be allowed.
Access for commercial researchers and organisations

This relates to points 7 and 8 on the consent form.

Key point
You are agreeing that approved researchers from for-profit companies can have access to this data under specific conditions and rules.

- You are agreeing to let not-for-profit organisations access your child’s data, such as research charities, universities or hospitals.
- You are also agreeing to let for-profit (commercial) companies access your child’s data, such as drug companies, or companies making diagnostic tests or developing faster ways to analyse large amounts of data.
- This is to enable new medicines, treatments and diagnostic tests to be developed as quickly as possible.
- Companies will have to pay to access the data we hold, although access may cost them less if they make their results available to all other researchers. If any financial profits are made by Genomics England, these will go back to the NHS.
- Neither you, nor your child, will benefit financially if a product or test is successful because of your child’s participation in the Project.

Can I access my child’s data?

- Genomics England owns the data from this Project. We protect it on your child’s behalf and decide who gets access to it, within the terms of your consent.
- You can ask for a copy of your child’s data, although there will be a charge for providing this.

How will you keep my child’s information secure and confidential?

- Your child’s data is stored in a controlled access database in the UK, and its security level meets national and international data standards.
- Your child’s name and other personal details will be available to your child’s clinical team and the Project team, who need these to analyse your child’s individual results and provide a report to discuss with you. Your child’s name and personal details will not be included in the data which is provided to researchers.
- Researchers looking at your child’s data will be tracked by Genomics England to check that they are following the laws and ethical guidelines that apply to biomedical research.
- Although researchers can look at your child’s data or that of their participating family members and ask questions about it, they can only take away the answers to their questions (their results). They can’t copy or take away any of your child’s individual data.
- Revealing your child’s data on purpose in a way that identifies them would be a legal breach against the permitted use of samples and data from the Project and is against good research practice. Any individual or institution that misused data in this way could face criminal charges or substantial fines, and would be barred from accessing our Project again. Major research funders in the UK have said that they will withdraw funding from any researcher that did this.

Will this affect my insurance or my child’s insurance?

- Any medical treatment you or your child have may need to be disclosed to an insurer that asks about it. However, under an agreement between the Department of Health and the Association of British Insurers, the results of your child’s whole genome sequencing carried out in the 100,000 Genomes Project are not disclosable to insurers.
- You don’t have to tell an insurer that your child is part of the 100,000 Genomes Project, and they won’t have access to the results.
5. FUTURE CONTACT

What might you contact me about in the future?

This relates to point 9 on the consent form.

Key point

You are being asked to agree to be contacted in the future by your clinical team, or by the Genomics England Project team, to ask for additional health information about your child, or to invite you or your child to take part in further research.

- In the future you may be contacted by your child’s clinical team, or directly by Genomics England, for you or your child to be invited to take part in further research. This might include clinical trials of new medicines, or research about your views on aspects of the Project.
- If you don’t want your child to take part in research (or you don’t want to take part yourself) when you are contacted then you and they don’t have to.

6. RESULTS

What information will I receive from whole genome sequencing?

This relates to points 10 and 13 on the consent form.

Key point

You are agreeing to be given the results of your child’s whole genome sequencing. The results might not show the cause of the rare condition in your family, and the results might take many months to come back.

The ‘main condition’ is how we describe the rare condition in your family that was the reason your child was invited to join this Project.

- After your child’s genome is sequenced, the results will be sent to your child’s clinical team who will check them and discuss them with you.
- In time, findings will be returned within a couple of weeks as the technology and our understanding of the results develops. At first this will take longer, so some or all of the findings may not be available for many months.
- The results may provide information that helps guide treatment for your child or other members of your family, but alternatively it is also possible that there will be no information to report.
- If something is found which could be important for the health of other members of your family, your clinical team will advise you about what information would be helpful to pass on to them, and will support you in doing this.
- Your child’s genome sequence will be compared with other participants in the Project. This could show that your child is genetically related to someone else who has joined the Project, who you did not know about. This information will be used to help analyse your child’s results, but no identifiable information will be made available to participants who didn’t join the Project as part of your family unit without your consent.
- You will not be told personally about the results of research that has included your child’s data, unless the result is important for your child’s individual diagnosis or treatment. Details of all studies using the Project’s databases will be published and made available on the Genomics England website.

What other results can I ask for from the 100,000 Genomes Project?

This relates to point 10 on the consent form.
Your child’s genome sequence contains information which might be relevant for their health, as well as other types of information such as how they are related to members of their family or their eye colour. It also contains a lot of information we cannot fully understand.

When your child's genome sequence is analysed in this Project, we can look for information which might be relevant for their future health, or the health of other members of the family. We call these ‘additional findings’ (because they are different to the main condition in your family) and you can choose whether we look for these.

- Information from your child’s genome which is relevant to the main condition in your family will routinely be provided to you by the clinical team, as described above.
- Information about health-related additional findings will only be looked for and provided to you if you request this in the consent form.
- Any other information in your child’s genome which is not relevant to your main condition and is not an additional finding will not be looked for or provided to you.

If you ask us to look for health-related additional findings in your child’s genome, all of the diseases we will look for could affect your child before adulthood (below the age of 18), and can either be treated, or screening can be offered aiming to pick them up at an early stage.

Although we know that treatment or screening can be useful in these diseases, there are some people who will receive an additional finding, but who will never go on to develop the disease. As the Project continues, we hope to understand better who will benefit from treatment or screening.

The diseases we will look for are uncommon, and the chance of your child having one of them is not high. If your child is found to have an additional finding, you will be given advice about this, and what treatment or screening they could have.

Even if no additional findings are reported in your child’s genome, there is still a small chance that your child could develop one of the diseases we have looked for. This is because we do not yet know whether genome sequencing will pick up all important 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, the additional findings which we will look for will be regularly updated. If this new information shows that your child has an additional finding in the future, this information will be provided to the clinical team to discuss with you.

Part of the purpose of offering you information about additional findings is to help us decide whether or not we should look for these as part of NHS standard clinical care.

Can I change my mind about additional findings in the future?

- You can change your consent choices about health-related additional findings on behalf of your child at any time by filling in an Opt-in or Opt-out form, which you can get from the clinical team or from the Genomics England website: [www.genomicsengland.co.uk](http://www.genomicsengland.co.uk).

What will happen after the initial reports are sent to my child’s clinical team?

- If you decide to receive your child’s additional findings results these may be returned separately from any main results.
- After you receive any initial reports, all your child’s information will continue to be held on the Project’s...
research databases, and will be added to as new records become available.

- If more information about your child’s genome becomes available after more research has been done, the clinical team will tell you about this. This means you could get further reports in the future, as well as any initial reports on the main results, and your child’s additional findings (if you choose to receive these).

**What are the benefits of joining the 100,000 Genomes Project?**

- Your child might benefit in terms of their healthcare personally but they might not benefit at all.
- Other patients may benefit from the knowledge generated by this Project.

**What are the risks of joining the 100,000 Genomes Project for my child?**

- **Being identified as someone taking part in the Project**: We will always do everything we can to stop this happening. There are penalties in place, but there is still a remote risk that this could happen.
- **Giving blood samples**: There is a small risk of bruising, inflammation or fainting but no more than with any other blood sample. Experienced staff will collect the blood.
- **Future risks**: There might be new ways in the future to link information back to your child directly, but we will keep up with these developments in order to protect your child’s data.

### 7. LEAVING THE 100,000 GENOMES PROJECT

**How can I withdraw my child from the Project?**

This relates to **point 14** on the consent form.

**Key point**

You are free to withdraw your child from the 100,000 Genomes Project at any time in the future. You do not have to give a reason.

If you want your child to leave the Project after you have joined, ask the clinical team for a Withdrawal form, complete the form and return it to them. There are two options for withdrawal, as below. More information about these options is available from the clinical team or in the withdrawal form.

1) **No further contact, but continue to include my child’s samples and information in the Project**

OR

2) **No further contact and no further use of my child’s samples or information.**

Your child’s 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.
For parents of participating children

INSERT LOCAL CONTACT DETAILS/
LABEL HERE