

100,000 Genomes Project

Participant consent form

If you agree to take part in the 100,000 Genomes Project, please *initial* each box and sign this form.

1 I understand that my participation in the 100,000 Genomes Project is voluntary and that if I refuse, I don't need to give any reason and that my present or future medical care or legal rights will not be affected. I confirm that I have read and understood the information dated ___ / ___ / ___ (version_) for the 100,000 Genomes Project. I have had the opportunity to ask questions and have had these answered satisfactorily.

Initial here to indicate your agreement

2 The 100,000 Genomes Project allows medical researchers, healthcare teams and commercial organisations to access your samples and health and related information we collect. I agree to join this Project and specifically understand the following:

Initial here to indicate your agreement

3 I agree to give a sample of blood and/or for samples already collected as part of my medical care to be used, and if essential, saliva to be collected, and for details about me and any samples I provide to be stored securely by Genomics England.

Initial here to indicate your agreement

4 I agree that my donated sample(s) can be used to collect DNA for whole genome sequencing, or for studies looking at proteins or other components of my cells. I understand that my samples may be processed in other ways that have approval from the 100,000 Genomes Project. I understand that my samples or DNA could be sent for secure processing or analysis outside of the UK by approved organisations. I understand that future research on my samples may use new tests or techniques that are not yet known.

Initial here to indicate your agreement

5 I agree to give access to my health records and personal information to be used alongside my samples for scientific or medical purposes and research relating to medical condition(s) affecting me, or other people. I understand that this can be at any point during my life and will continue after my death.

Initial here to indicate your agreement

6 I agree that my samples, my DNA sequence, and information from my health records and any other information I give to the Project can be collected and stored securely by the Project as a resource for use by approved researchers from around the world, for future scientific or medical purposes during my life and after my death. I understand that they won't be allowed to copy or remove any of my information.

Initial here to indicate your agreement

7 I agree that the 100,000 Genomes Project can collect, store and analyse information from my medical notes and health records (in general practice (from my GP) or hospital or social care records, or other sources such as local or national disease registries). I consent to this access now and in the future (including after my death). I agree that these notes and records or the samples I give may be looked at by approved individuals from Genomics England or from the NHS Trust and other study monitors at any time.

Initial here to indicate your agreement

8 I understand that all information about me held by the Project will be treated as confidential. I understand that information from my samples, records or other information I give to the Project will only be accessible in a form which protects my identity.

Initial here to indicate your agreement

9 I understand that these research organisations could include commercial (for-profit) companies.

Initial here to indicate your agreement

10 I understand that I will not benefit financially if research undertaken through the 100,000 Genomes Project leads to new treatments or medical tests.

Initial here to indicate your agreement

11 If, based on results obtained from my samples and/or information about me, I might be eligible to participate in future research studies, I agree to be contacted by my clinical team in order to invite me to participate. This may be about this Project or other ethically approved research studies, including clinical trials or research about ethics. I will be provided with full information about these studies, when and if I am contacted. I understand that agreeing to be contacted does not oblige me to participate.

Initial here to indicate your agreement

12 I agree to being asked to provide further samples or health information by my clinical care team for the purposes of the Project. I understand that I am not obliged to give this permission. I also agree to be contacted directly up to four times in any year by the Project (not via my doctor), for additional information about my health or lifestyle. I understand that this does not oblige me to participate.

Initial here to indicate your agreement

Return of results

13 I understand that it is not possible to guarantee that we will find anything of significance now or in the future.

Initial here to indicate your agreement

14 Main genetic findings (*non-optional*): I give consent for the Project to run tests on my samples and health information relating to the cause or management of the main condition that led me (or my relative) to join the Project (sometimes called the 'primary' or 'pertinent' findings). I agree that these results can be reported to my extended clinical care team. I agree that my clinical care team can discuss these results with me. I also understand that the results may **not** be able to provide a diagnosis or to provide information to influence my clinical care (or the care of my relative), assuming that these results can be returned in a relevant time frame, which may not be possible.

Initial here to indicate your agreement

15 Additional genetic findings (*optional*): I understand that I can choose whether or not I want the Project to use my samples and health information to **look for additional genetic results, beyond my (or my relative's) main findings** (sometimes called 'secondary' findings). These additional results would relate to my risk of developing certain serious or possibly life-threatening and rare medical conditions which can be cured, made less severe, or prevented via standard NHS treatment. The Project has agreed with the NHS, a limited list of the medical conditions to be looked for under these criteria. This list will be updated and may change over time according to new medical evidence. The person taking my consent has a list available of which genetic conditions will currently be looked for under these criteria. These conditions will be fully explained to me, along with the implications of a positive test, and the support that will be available to me if I then choose to have these tests. If I allow the Project to look for these additional findings, a health professional will discuss these results with me after they have been confirmed by the NHS.

Tick your choice (ü)

Yes, I want this information to be looked for and fed back to my clinical team

OR

No, I do not want this information to be looked for and fed back to my clinical team

Initial here to confirm your choice

16 Pre-symptomatic Carrier Testing for **couples** participating in this Project (*optional*): (*only offered if you and your opposite-sex partner/spouse have both joined this Project and if you tell us that you are thinking about having children together, now or in the future.*)

I understand that my partner/spouse and I have the choice of whether or not to be tested to see if, when looked at together, we both 'carry' a risk of passing on serious or possibly life-threatening and rare genetic medical conditions to our future children. These conditions may (or may not) be able to be cured, made less severe, or prevented via standard NHS treatment.

The Project has agreed with the NHS to look for carrier status for a limited list of medical conditions that fit these criteria. The list of conditions will be updated over time according to new medical evidence.

The person taking my consent has a list available of the genetic conditions carrier status will currently be looked for. I can choose whether or not to discuss this list with them.

I understand that being confirmed to be a carrier of any of these listed conditions is not likely to affect my or my partner/spouse's health. These results will not usually determine whether we ourselves need to have follow up or treatment for a condition on the list.

I understand that these carrier status test results are offered – and will be returned to us – together as a couple, only if each of us give consent to this. If we **don't** each consent together, this information will **not** be looked for and will not be reported to us.

I understand that medical conditions **that can only be passed on to children by one member of the participating couple** are included on the list of tests. Only in this case, results will be given individually to that member of the couple by a health professional. Both partners/spouses do not need to consent together to have this testing, just the participant concerned.

I understand that where relevant regarding testing my partner/spouse and I would be offered appropriate support such as genetic counselling.

Tick your choice (ü)

Yes, I want this information to be looked for and fed back to my clinical team

OR

No, I do not want this information to be looked for and fed back to my clinical team

Initial here to confirm your choice

17 Incidental findings: I understand and agree that any other results of genetic or other analysis of samples, separate to my main findings (if I am the patient), or additional or carrier status findings (where I have consented to receive these), will not be fed back to me.

Initial here to indicate your agreement

18 I understand that information generated by this Project may be of benefit to my family members now or in future. I understand that normal clinical practice will be applied in the use and sharing of this information with other members of my family and their medical teams, based on my relative/s' potential benefit from receiving this information.

Initial here to indicate your agreement

19 I understand that I am free to withdraw my permission for my samples and information to be used at any time in the future. I don't need to give a reason and my medical care will not be affected. I understand if I join and then withdraw from this Project that it will not be possible to remove my data from research that may already have taken place.

Initial here to indicate your agreement

Name of patient (BLOCK CAPITALS)		
<input type="text"/>		
Date of birth	Signature	Date
DD / MM / YY	<input type="text"/>	DD / MM / YY

Name of researcher (BLOCK CAPITALS)	
<input type="text"/>	
Signature	Date
<input type="text"/>	DD / MM / YY

When completed:

- 1 (original) to be kept in the adult participant's 100,000 Genomes Project records.
- 1 copy for participant.
- 1 copy to be sent to Genomics England.

Insert local contact details here	
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Hospital address:	Cambridge University Hospitals NHS Foundation Trust Hills Road Cambridge CB2 0QQ