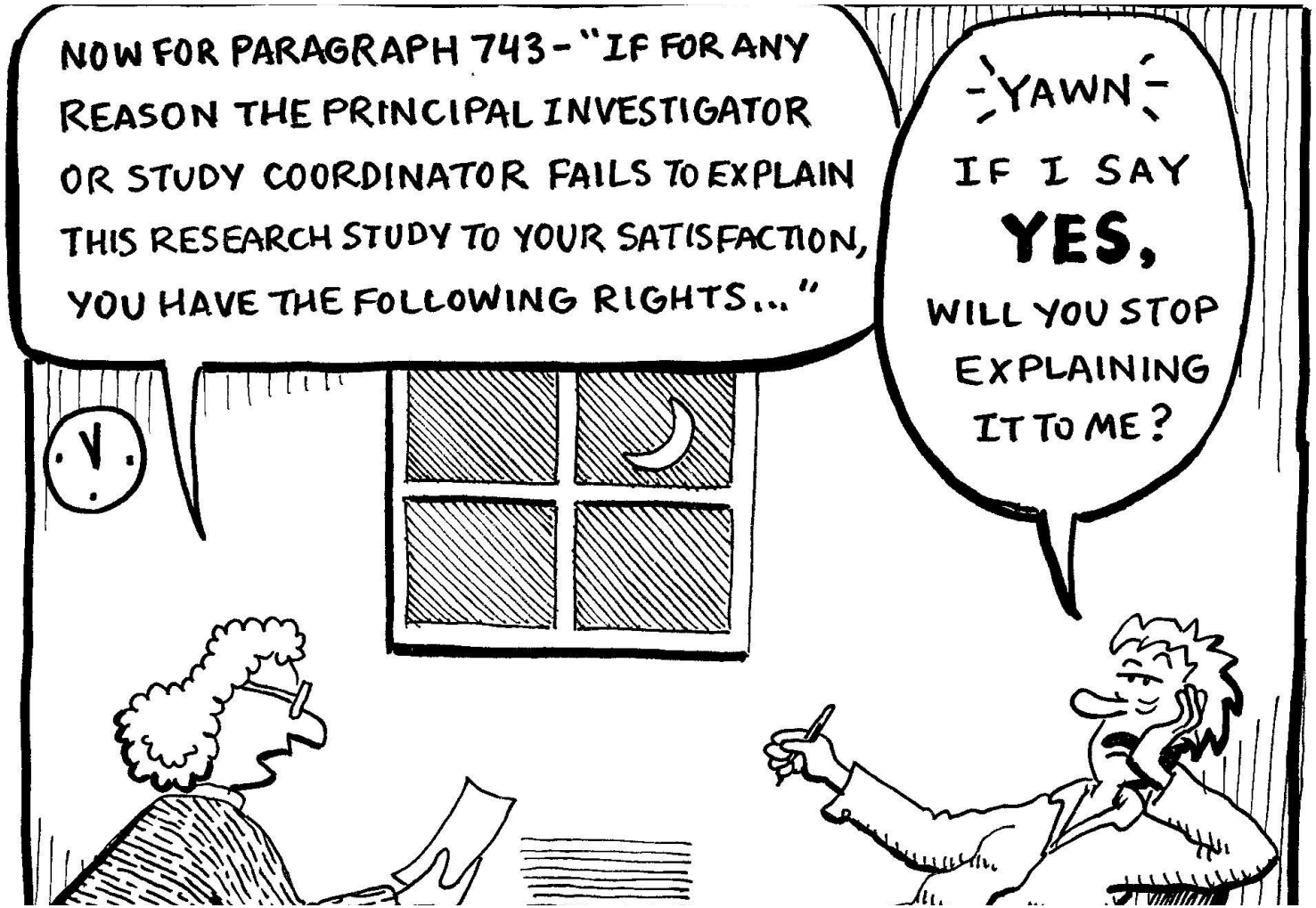
# How do we ‘do’ consent? Clinical & research models in genomic medicine 100,000 Genomes Project and beyond

Christine Patch

Principal Staff Scientist, Society and Ethics Research Group, Connecting Science, Wellcome Genome Campus

Clinical lead for Genetic Counselling, Genomics England, London

Visiting Professor, Faculty of Health and Wellbeing, Sheffield Hallam University


NOW FOR PARAGRAPH 743 - "IF FOR ANY REASON THE PRINCIPAL INVESTIGATOR OR STUDY COORDINATOR FAILS TO EXPLAIN THIS RESEARCH STUDY TO YOUR SATISFACTION, YOU HAVE THE FOLLOWING RIGHTS..."
YAWN
IF I SAY YES, WILL YOU STOP EXPLAINING IT TO ME?

# The 100,000 Genome Project

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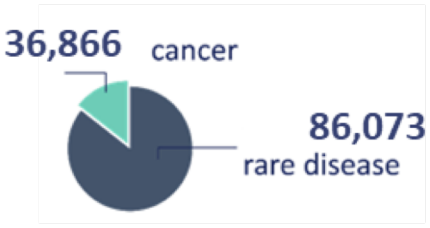
## Samples

**122,939**
Samples collected and received at the UK Biocentre




## Genomes

**119,290**
Genomes sequenced




## Analysis and Results

**105,621**
genomes returned to NHS with summary findings


**Rare disease**
- 20-25% actionable findings

**Cancer**
- 50% cases contain potential for therapy or a trial stratification


**Protocol-based medicine**

**Volume-based healthcare**

**Genomics**

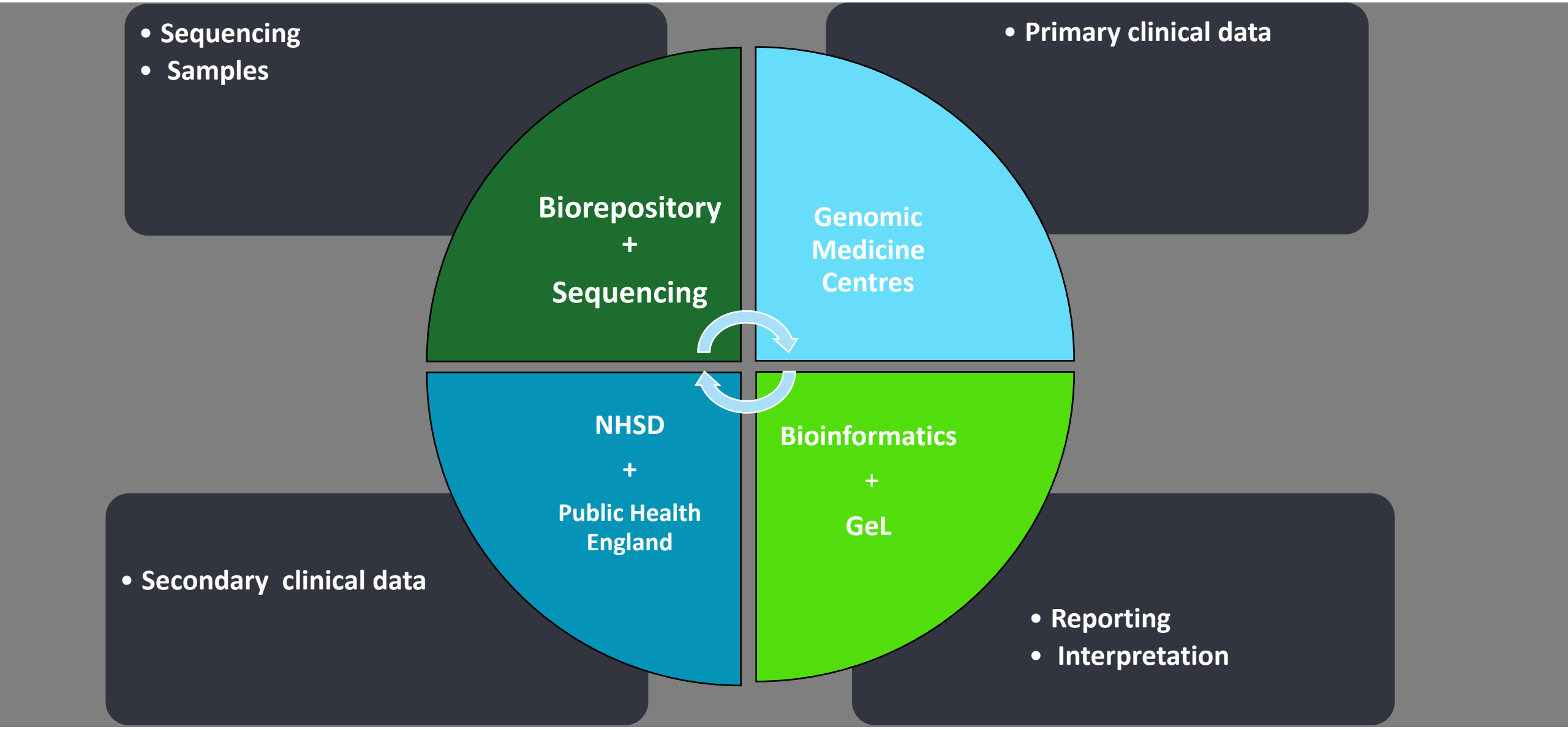
**Data-driven healthcare**

**Value-based healthcare**

# Thank you to everyone who has taken part in the 100,000 Genomes Project

# 100,000 Genomes Project Data Sources and Types





# Consent what does it mean?

Consent to treatment
Common Law
Professional standards and Guidelines


Lawful basis for processing data

- Consent
- Public task
- Legitimate interests
- Contract
- Legal obligation
- Vital interests

Consent to research
Health Research Authority

# Consent in the 100,000 Genomes Project



Participants gave permission for:

Whole Genome Sequencing

- Return of main findings (findings related to reason for recruitment)
- Option of return of limited list of additional findings

Consent materials created and revised (2016)

- Collaboration with participants
- Recruiters
- Independent ethics committee

Materials available https://www.genomicsengland.co.uk/taking-part/patient-information-sheets-and-consent-forms/

# Learning from the 100,000 Genome Project: Review of consent materials

## Wide stakeholder consultation

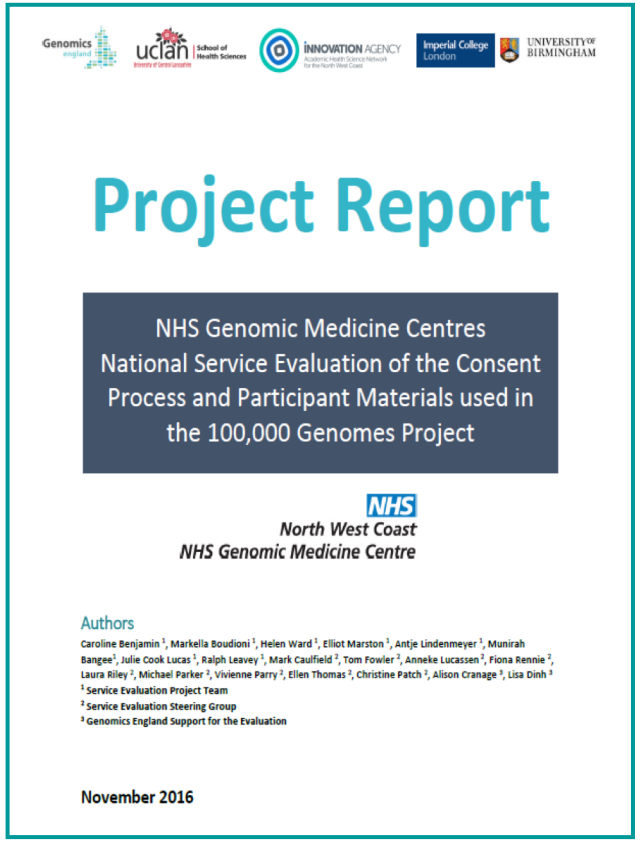
— Including Genomic Medicine Centres, recruiters and participants and PPI representatives

## Main changes

— reduction of approximately 20% in the text overall
— improvements to readability, plain English Crystal Mark
— small reduction in the number of different versions
— shorter consent form, reduction of 30%.

## Accessibility

— Easy read version for people with intellectual disability
— Versions accessible for individuals with visual impairment


Project Report

NHS Genomic Medicine Centres National Service Evaluation of the Consent Process and Participant Materials used in the 100,000 Genomes Project

NHS
North West Coast
NHS Genomic Medicine Centre

Authors

November 2016

https://www.genomicsengland.co.uk/consent-evaluation/

# Examples of revised consent materials



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**C1**

For adults with cancer (or suspected cancer).

[INSERT NHS GMC LOGO]

Crystal Mark 22296
Clarity approved by Plain English Campaign

## 100,000 Genomes Project

### Information sheet

You have been invited to take part in the 100,000 Genomes Project because you have, or potentially have, one of the types of cancer which are being studied by the project. We hope to recruit as many patients as possible with your type of cancer to the project so we can learn more about it.

If the results of your tests show that you do not have cancer, we would not include you in the project and would not continue to keep any of your information or samples.

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 about it 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.

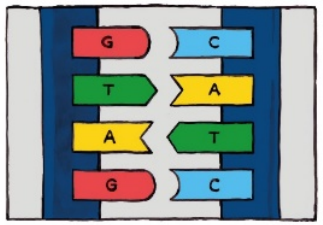
You can take as much time as you need to think about joining the project. Only you can choose if you want to take part. If you don't want to join, you don't have to say why. And, if you don't join, you will still receive the best available care.

### 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.



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**R1**

For adults with a rare genetic condition or an adult family member.

### 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.

# Learning from the 100,000 Genome Project:

Health-care professionals need to be prepared to answer patients' questions about genetics to facilitate genome sequencing consent.

Interviewees' decisions to participate in 100 kG P were based on interpersonal and institutional trust in the NHS, and on an investment in improving care for the future.

Opening the "black box" of informed consent appointments for genome sequencing: a multisite observational study.

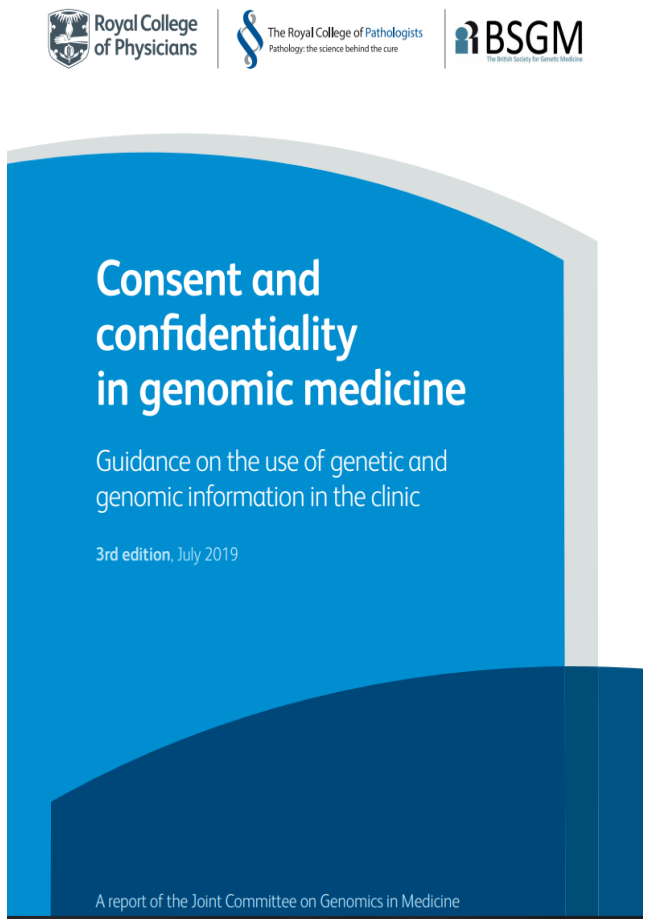
Sanderson SC, et al Genet Med. 2018 Oct 1. doi: 10.1038/s41436-018-0310 [Epub ahead of print]

Fostering trust in healthcare: Participants' experiences, views, and concerns about the 100,000 genomes project.

Dheensa S et al Eur J Med Genet. 2018 Nov 29. pii: S1769-7212(18)30352-5. doi: 10.1016/j.ejmg.2018.11.024. [Epub ahead of print]

# NHS Genomic Medicine Service

- National Service covering all DNA based testing including testing of cancers
- Operating to common national standards specifications and protocols
- 7 Genomic Laboratory Hubs contracted to deliver tests
- Rare and inherited disease testing all commissioned centrally by NHSE
- Aim to build a national genomic knowledge base to inform academic & industry research & discovery including clinical trials and recruitment
- All patients where WGS is the technology used for the test will be offered the opportunity to give permission for the data to be accessed through National Genomic Research Library




# The process of seeking consent ensures that:

- a person understands the nature and purpose of the procedure or intervention thereby asserting a right to self-determination

- may be evidenced by good documentation but a signature on a form will not necessarily indicate what consent has actually been given.

# Patient choice in NHS Genomic Medicine Service

- The patient choice framework – clinical care and research both presented as part of the clinical pathway.

- Aim to give a **clear, informed and separate choice** about the genetic test (initially only those involving WGS) and participation in research

- All patients where WGS is used as the test technology to be given the **opportunity to participate in research**

  — **Access to de-identified data by researchers-for profit and not for profit**

  — **Recall for research**

  — **Lifelong collection of other routinely collected health data**

  — **Also includes samples**

| First name | NHS number (or postcode if not known) |
|---|---|
| Last name | Date of birth d d m m y y y y |

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# Record of Discussion Regarding Genomic Testing

***This form relates to the person being tested. One form is required for each person.***

*All of the statements below remain relevant even if the test relates to someone other than yourself, for example your child.*

**I have discussed genomic testing with my health professional and understand the following**

### *Family and wider implications*

1. The results of my test may have implications for me and members of my family. I understand that my results may also be used to help the healthcare of members of my family and others nationally and internationally. This could be done in discussion with me or through a process that will not personally identify me.

### *Uncertainty*

2. The results of my test may have findings that are uncertain and not yet fully understood. To decide whether findings are significant for myself or others, my data may be compared to other patients' results across the country and internationally. I understand that this could change what my results mean for me and my treatment over time.

### *Unexpected information*

3. The results of my test may also reveal unexpected results that are not related to why I am having this test. These may be found by chance and I may need further tests or investigations to understand their significance.

### *DNA storage*

4. Normal NHS laboratory practice is to store the DNA extracted from my sample even after my current testing is complete. My DNA might be used for future analysis and/or to ensure that other testing (for example that of family members) is of high quality.

### *Data storage*

5. The data from my genomic test will be securely stored so that it can be looked at again in the future if necessary.

### *Health records*

6. Results from my genomic test will be part of my patient record, a copy of which is held in a national system only available to healthcare professionals.

### *Research*

7. I understand that I have the opportunity to take part in research which may benefit myself or others, now or in the future. An offer to join a national research opportunity is available on the following page.

For any further questions, my healthcare professional can provide information. More information regarding genomic testing and how my data is protected can be found at www.nhs.uk/conditions/genetics

**Please sign on page three to confirm your agreement to the genomic test.**





NHS Genomic Medicine Service. Record of Discussion Form version 4.0.0 (February 2020). To be used for WGS go-live

| First name | NHS number (or postcode if not known) |
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# Participation in The National Genomic Research Library

**The NHS invites you to contribute to the National Genomic Research Library, managed by Genomics England**

Genomics England was set up in 2013 by the Department of Health and Social Care to work with the NHS to build a library of human genomes for researchers to study. Combining data from many different patients helps researchers to better understand disease and spot patterns in the data.

By agreeing to share your data you might get results which could lead to your own diagnosis, a new treatment, or offers to take part in clinical trials. Your taking part could enable diagnoses for people who don't have one.

Please read the following statements. Feel free to ask any questions before making a decision.

**By saying 'yes' to research, I understand the following**

### *Security*

1. Any samples and data stored by Genomics England and the NHS will always be stored securely. Genomics England will take all reasonable steps to ensure that I cannot be personally identified.

### *Re-contact*

2. NHS staff, or Genomics England together with the NHS, can contact me if the data or samples reveals any clinical trials or other research that I might benefit from.
3. If something is relevant to me or my family, there is a process by which this will be shared with my NHS clinical team.

### *Data and sample usage*

4. Researchers may include national or international scientists, healthcare companies and NHS staff. To access the data, these researchers must all be approved by an independent committee of experts, including health professionals, clinical academics and patients. There will be no access to the data by personal insurers and marketing companies.

### *Data storage*

5. Genomics England will collect different aspects of my health data from the NHS and other data from organisations listed at www.genomicsengland.co.uk/understanding-genomics/data. The collection and analysis of my health data for research will continue across my entire lifetime and beyond.

### *Withdrawal*

6. I can change my mind about taking part at any time.

More information regarding research in the National Genomic Research Library can be found at www.genomicsengland.co.uk
For any further questions, my healthcare professional can provide information.

**Please use page three to indicate your research choice.**





# Patient Choice and Record of Discussion

## Confirmation of Genomic Test and Research Choices

**I confirm that I have had the opportunity to discuss information about genomic testing, I agree to the genomic test, and my research choice is (circled) below**

**A. I have discussed taking part in the National Genomic Research Library** **YES | NO**

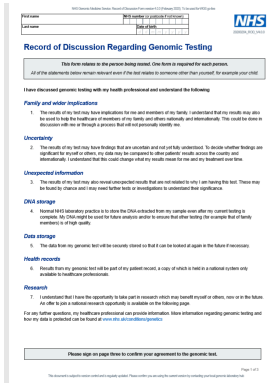
*If your answer to A is **NO** then please ignore B and sign directly below*

**B. I agree that my data and remainder sample may contribute to the National Genomic Research Library** **YES | NO**

| Patient name | Signature | Date |
|---|---|---|
| | | d d / m m / y y y y |

**If you are signing this form on behalf of someone else (children, adults without capacity or deceased patients) then please sign below**

| Parent \| Guardian \| Consultee name* *please amend as appropriate | Signature | Date |
|---|---|---|
| | | d d / m m / y y y y |



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Page 1 of 3

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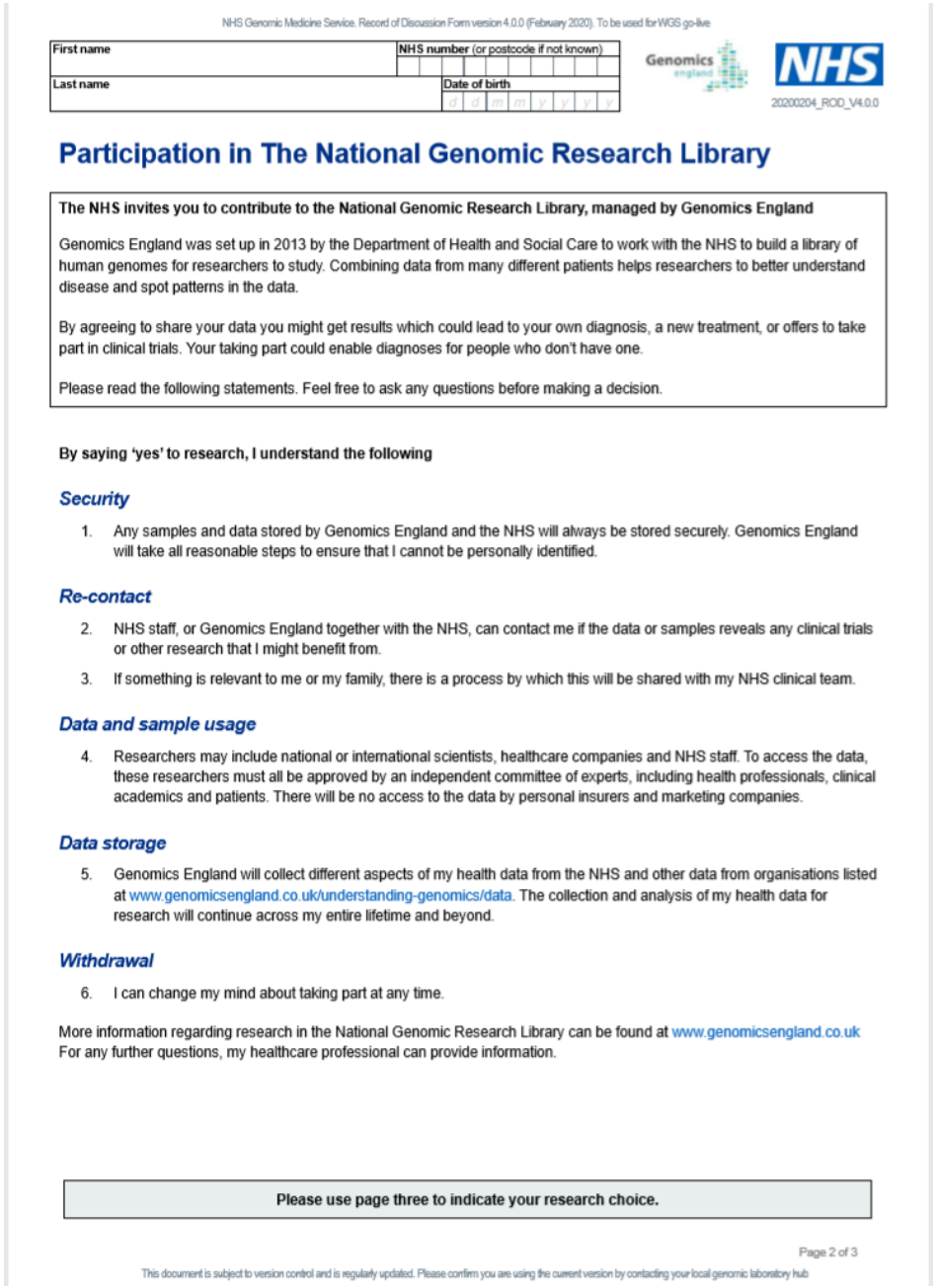

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Page 2 of 3

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- Security
- Re-contact
- Data and sample usage
- Data storage
- Withdrawal

omics/data/

# Data Access and Use

Participant privacy and confidentiality is vital in the 100,000 Genomes Project.

A key feature of the Project is that an individual's data will not be released. Instead, de-identified data is analysed within a secure, monitored environment.

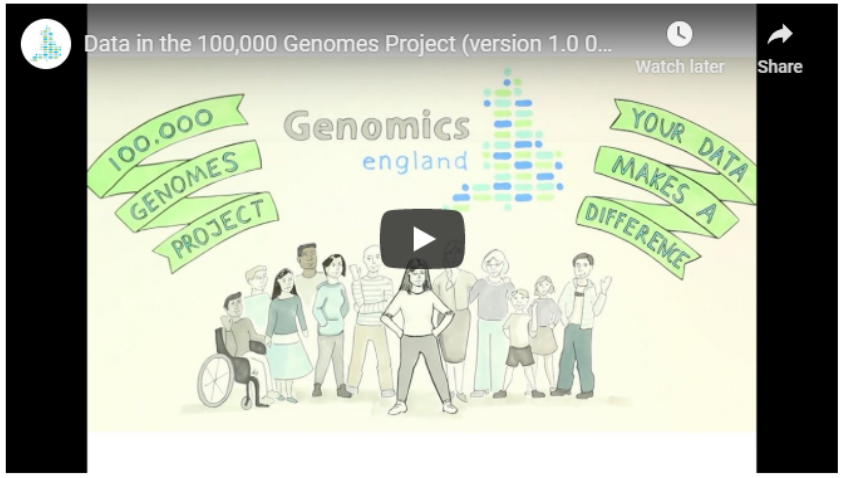
**Who has access?**

Doctors, nurses and other healthcare professionals in NHS Genomic Medicine Centres have access to information about the patients they are caring for. Researchers need to apply to access the de-identified genomic and health data in the data centre. Read more about who has access to the data and current research projects.

Scientists and clinicians will access the data for a variety of research. This could be to:

- Improve diagnosis of rare disease.
- Understand how genomics impacts on health and healthcare.
- Suggest which treatments might work best for an individual.
- Understand the causes of disease better.
- Develop new treatments.

## Data in the 100,000 Genomes Project




Useful links

**Insurance**
Find out how taking part in the Project may affect insurance.


**About genomics**
Infographics and short films introducing genomics and genome sequencing.


**Taking part**
Information about taking part in the Project

Genomics england

https://www.genomicsengland.co.uk/understanding-genomics/data/

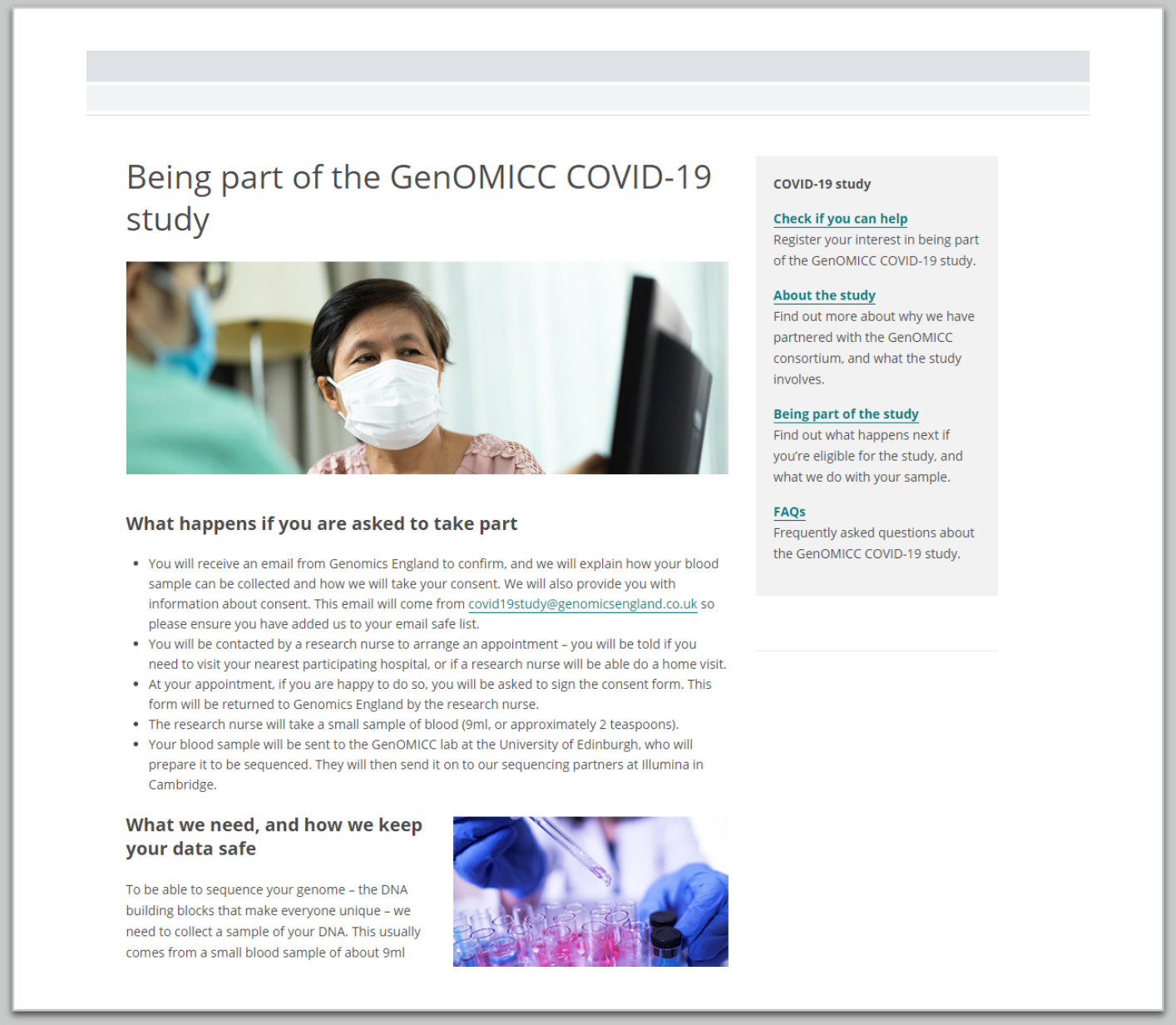
# COVID-19

Delayed the start of Genomic Medicine Service

Approach to simplified consent has been essential to collaborations in relation to COVID 19 research

Facilitating partnership with GenOMICC study

Facilitating co recruitment with other studies



## Being part of the GenOMICC COVID-19 study



### What happens if you are asked to take part

- You will receive an email from Genomics England to confirm, and we will explain how your blood sample can be collected and how we will take your consent. We will also provide you with information about consent. This email will come from covid19study@genomicsengland.co.uk so please ensure you have added us to your email safe list.
- You will be contacted by a research nurse to arrange an appointment – you will be told if you need to visit your nearest participating hospital, or if a research nurse will be able do a home visit.
- At your appointment, if you are happy to do so, you will be asked to sign the consent form. This form will be returned to Genomics England by the research nurse.
- The research nurse will take a small sample of blood (9ml, or approximately 2 teaspoons).
- Your blood sample will be sent to the GenOMICC lab at the University of Edinburgh, who will prepare it to be sequenced. They will then send it on to our sequencing partners at Illumina in Cambridge.

### What we need, and how we keep your data safe

To be able to sequence your genome – the DNA building blocks that make everyone unique – we need to collect a sample of your DNA. This usually comes from a small blood sample of about 9ml



**COVID-19 study**

**Check if you can help**
Register your interest in being part of the GenOMICC COVID-19 study.

**About the study**
Find out more about why we have partnered with the GenOMICC consortium, and what the study involves.

**Being part of the study**
Find out what happens next if you're eligible for the study, and what we do with your sample.

**FAQs**
Frequently asked questions about the GenOMICC COVID-19 study.

# Patient/Participant choices

Ensure patient/participants' choice is free and informed

Enough information available for them to access to make the choice

Choice must be recorded and complied with

— More than a signature on a form

# Questions??????


Genomics england
Thank you to everyone who has taken part in the 100,000 Genomes Project
NHS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