Implementing a hybrid clinical/research model in genomic medicine: post 100,000 Genomes Project

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NHS
Health Education England

Consent in the 100,000 Genomes Project

- Participants consented to
  - 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
  - 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

Health-care professionals need to be prepared to answer patients' questions about genetics to facilitate genome sequencing consent. Health-care professionals' education also needs to address how to effectively listen and elicit each patient's questions and views, and how to discuss uncertainty around the disease risks associated with secondary findings.

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]

We found that 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. Interviewees relied upon receiving good ongoing NHS care for managing their own or their child's rare disease, but they worried about what their relationships with NHS healthcare professionals would be like in future. …….. To honour and foster participants' trust - which may easily be lost - and their clinical labour, we therefore recommend ongoing public engagement and consultation about how genomics is being integrated more widely across specialties (especially given current funding and staffing constraints in the NHS) within the newly formed NHS Genomic Medicine Service.

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]

Learning from the 100,000 Genome Project

- Complexity of recording consent status at scale
- Complexity of sustaining consent choices over time
  - Withdrawal
    - Partial
    - Full
  - Additional findings
    - yes to no
    - no to yes
  - Changes in capacity
    - Gaining
    - Losing

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**

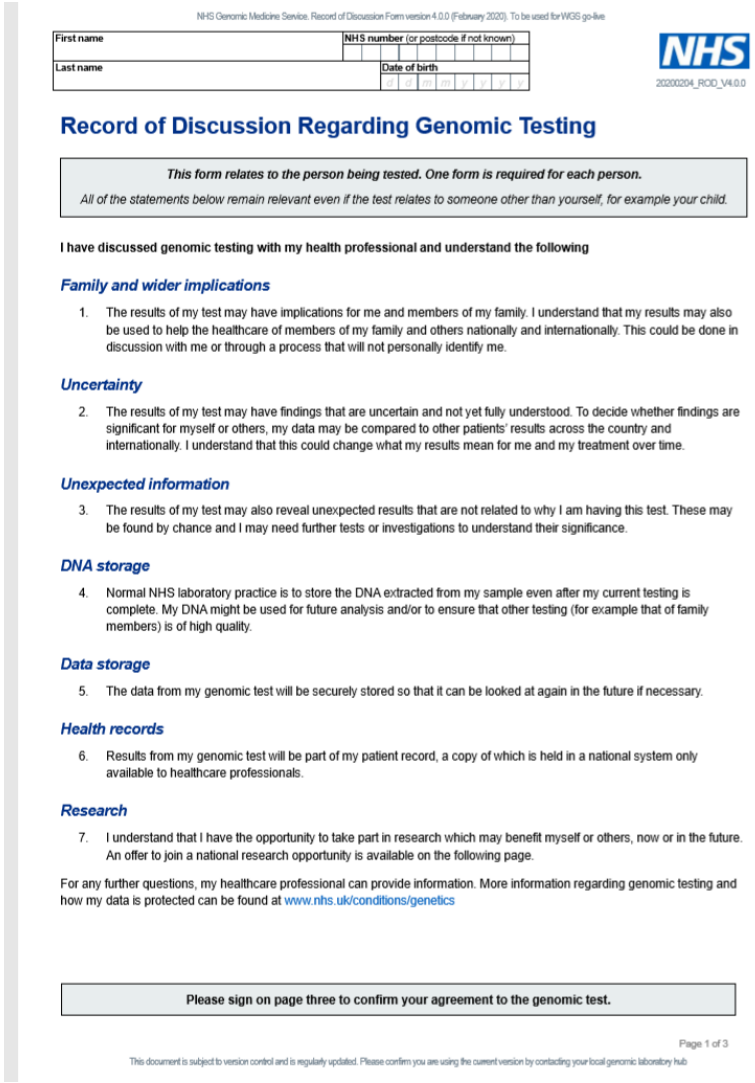
Clinical Choice

All tests on offer as part of GMS are NHS-commissioned diagnostic tests for patients presenting with unexplained symptoms

The consent conversation with these patients is unchanged from current consent standards

With the exception of:

1. National storage of data
2. Discussions of pertinent germline results (for some cancers)



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)
Last name	Date of birth

20200204_ROD_V4.0.0

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.

Page 1 of 3

This document is subject to version control and is regularly updated. Please confirm you are using the current version by contacting your local genomic laboratory hub

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.

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.

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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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NHS

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.

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

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

Considerations for implementation

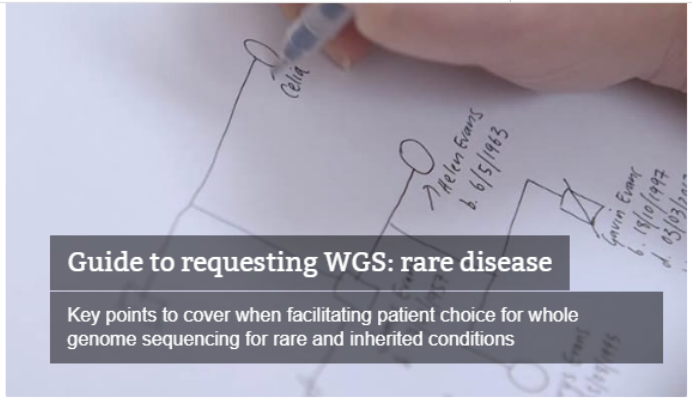
100,000 genome project

- Complex project within the health care system
  - Regulated under Health Research Authority as a Tissue Bank

- Creation of NHSE Genomic Medicine Service
  - Clinical service and Research combined
  - System transformation –
    - Including development of patient choice module within test ordering system

- Work force transformation
  - Health Education England Genomics Education Programme

  - https://www.genomicseducation.hee.nhs.uk/



Training resources

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

New partnership to sequence human genomes in the fight against coronavirus

Posted on May 13, 2020 at 12:01 am

- Genetic susceptibility to coronavirus to be tested in ground-breaking nationwide study
- Genomes of thousands of patients with coronavirus will be sequenced to understand how a person's genetic makeup could influence how they react to the virus
- Genomics England partners with University of Edinburgh to lead research drive to support the search for new treatments

GenOMICC

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Find out how to register interest to take part in the GenOMICC study.

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

NHS

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