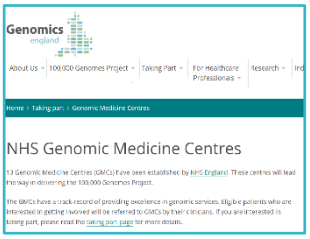
100,000 Genomes Project – consent overview

April 2017

FIRST STEP – LEAD ORGANISATION

Your first point of reference and primary resource for consenting patients is the Lead Organisation at your local Genomic Medicine Centre (GMC). Lead Organisations have a track-record of providing excellence in genomic services. You can find contacts for all Lead Organisations here: https://www.genomicsengland.co.uk/taking-part/genomic-medicine-centres



Lead Organisation contacts

CONSENT DOCUMENTATION

Patient information leaflets and consent forms can be accessed here or by clicking on the graphic below. Introductory leaflets, flyers and videos that can be used to explain the project to potential participants are also available at the same link.

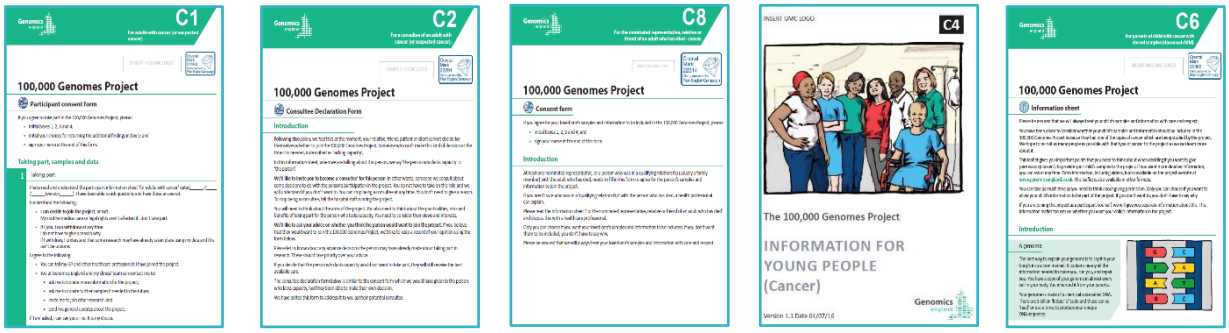


Figure 1: Examples of consent documentation

RETROSPECTIVE CONSENTING

When the 100,000 Genomes Project was initiated, the proposal was taken through full ethics approval at the HRA REC. However, there is both a diagnostic and a research arm to whole genome sequencing. To clarify the distinction between the two a consensus documents was agreed between:

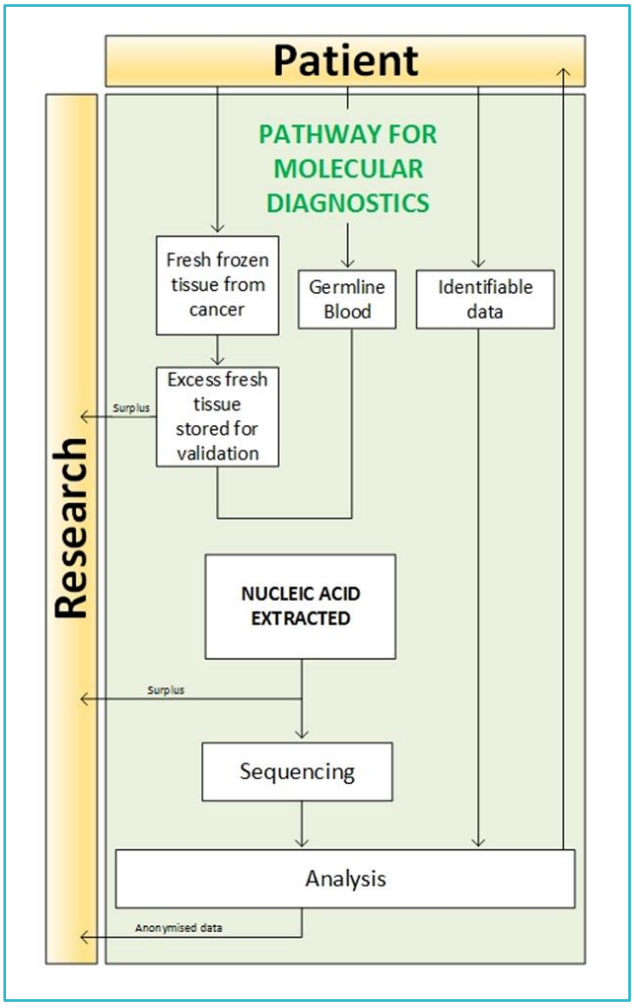
- Royal College of Pathologists
- Human Tissue Authority
- Health Research Authority
- NHS England
- Clinical Directors of the Genetic Medicine Centres
- Genomics England

The consensus recognises that genomic and molecular analysis can be a part of diagnostic care in patients with cancer and that acquiring, handling and local storage of fresh tissue samples can therefore be undertaken as part of a routine diagnostic pathway. Fresh frozen tissue is required to ensure a high quality of genetic analysis which is required for whole genome sequencing and other

large scale molecular genomic analysis. The diagnostic pathway requires multiple patient identifiers and a report is returned ultimately to the patient concerned.

The consensus recognises these samples as being diagnostic based on:

1. Their impact on patient care (including treatment with licensed or unlicensed drugs; entry to clinical trials and disease monitoring)
2. The way clinicians handle the samples and validate the results
3. How patients perceive the testing



The consensus statement responds to concerns expressed by some clinicians about the appropriate HTA licensing requirements needed for them to acquire and handle fresh frozen tissue for the 100,000 Genomes Project, where these samples are considered to be research samples (on the grounds that previously in NHS cancer diagnostics, such a method of storing samples would never, or only very rarely, have a diagnostic application. This consensus makes clear that fresh frozen tissue for this project can be acquired and handled as diagnostic when this is their purpose.

If tissue remains after use in diagnostics and is intended for research purposes then that tissue will fall under the remit of the Human Tissue Act 2004. The separate research and molecular diagnostic pathway are laid out in the attached diagram.

The consensus confirms that patients can now have a sample taken and handled in a genomic friendly manner as part of their diagnostic work up. If the patient is then found to be eligible for the 100,000 Genomes Project, that sample can be stored until the patient is consented and then sent for whole genome sequencing, all without implying any HTA licensing obligations.

Cancer samples for genomic testing are considered diagnostic which means consent can be taken after sampling at a time appropriate for the patient. Doing this means only patients that are eligible and have adequate tissue for DNA testing will be approached.

The full text of the consensus is available as a PDF in the 100,000 Genomes MDT Pack.

100,000 GENOMES PROJECT INFORMATION LINE

Eligible potential participants can be signposted to the information line with any questions they have about taking part in the Project. The information line can answer ad-hoc questions, or people can book an appointment to go through the consent materials in detail. Please note that it can't offer individual or medical advice, or advise on eligibility. Current participants and the public can also call the information line.

0800 389 8221 – open from 9.00am to 5.00pm Monday to Friday.