100,000 Genomes Project Closure and Future of Genomic Testing

Summary Update for Cancer Clinicians – December 2018

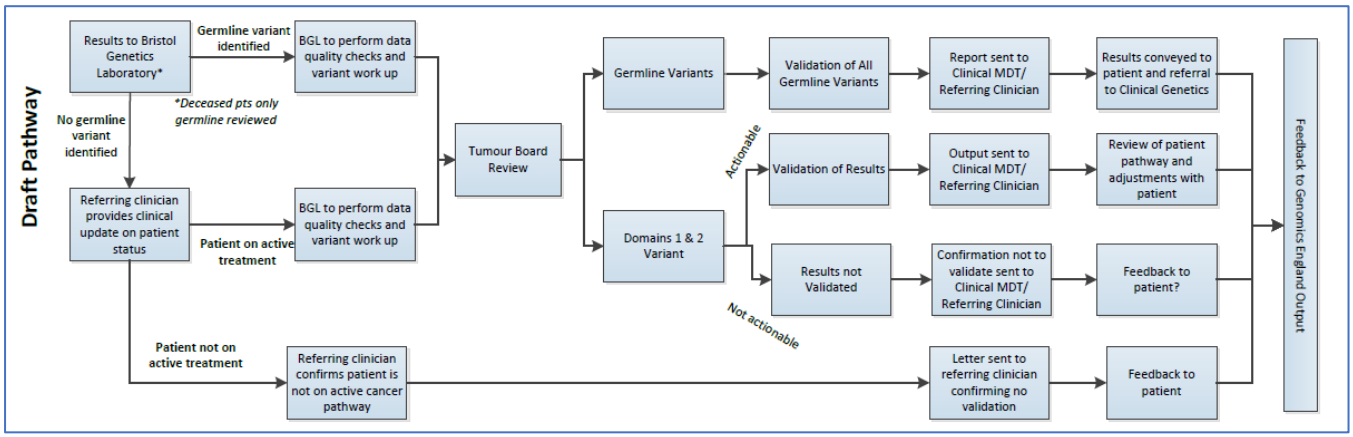
100,000 Genomes Project Closure & delivery:

1. The 100,000 Genomes Project closes to cancer patients at the end of December 2018
2. Nationally, the 100,000 genomes samples completed sequencing on 5th December 2018. This total included 29,000 tumour and blood samples from patients with cancer
3. Across the West of England Genomic Medicine Centre (GMC) region, 635 people with cancer consented to take part in the project and over 1,000 samples will have been submitted.

Proposed Return of Results Process for 100,000 Genomes Project patients:

1. All samples will be sequenced at the national sequencing centre in Cambridge and genome sequence data will be returned to Clinical Scientists at the Bristol Genetics Laboratory (BGL)
2. Genome sequence data will be examined for changes in cancer susceptibility genes (germline findings)
3. Genome sequence data from tumour tissue will also be examined (somatic findings) for patients who are being actively treated for their cancer
4. Germline findings and somatic findings for patients under active management will be evaluated by the Bristol Genetics Laboratory team and discussed at a regional genomics tumour advisory board (GTAB) meeting
5. Any findings that potentially influence patient care are confirmed locally by the Bristol Genetics Laboratory team and then reported back to the clinician responsible for the active cancer care for that patient, and relevant clinical colleagues.
6. The GMC or BGL will not communicate actionable results directly to patients, but expect this to be the responsibility of the clinician responsible for the active cancer care for that patient
7. The GMC will send a standard letter to patients who are not having active cancer treatment and who have no actionable germline findings confirming that there is 'nothing actionable'
8. If the patient's clinical status changes, particularly if there is relapsed disease then we encourage clinical teams to contact the GTAB team to request a re-analysis
   Contact details: nbn-tr.gtab@nhs.net

Diagram 1. Proposed pathway for managing return of results for cancer patients



Future genetic and genomic testing for cancer patients:

1. Germline cancer testing for inherited cancer susceptibility genes is included as part of the national rare disease test directory and is therefore subject to new commissioning arrangements. The provisional date for implementation of this is 1st April 2019
2. The eligibility criteria and process for genetic testing for inherited cancer susceptibility disorders is documented in the NHS genetic test directory for rare diseases: https://www.england.nhs.uk/publication/national-genomic-test-directories/
3. An informatics system called the "National Genomics Informatics Service" (NGIS) is being developed to help clinicians choose the right test for their patients and to give information about how to order tests for inherited cancer susceptibility disorders and how to record the patient's consent for diagnosis and participation in research. The first release of this system will be in 2019
4. The indications and process for genetic testing tumour samples for somatic variants is still under national review. We anticipate somatic testing will be via a provider to provider funding arrangement at least until April 2020
5. Whole genome sequencing will be available from 2019 for adult and children with certain sarcomas, acute leukaemia and for children with other solid tumours. Pathways to enable access to whole genome sequencing for these patients are now being developed with the help of local clinicians.
6. Some other tumour types will be eligible for gene panel testing. Detailed national policy is still in development and will be disseminated in a national cancer genetic test directory in 2019.
7. All cancer NHS genetic testing will be provided by one of seven nationally commissioned Genomic Laboratory Hubs (GLH). The GLH provider for hospitals in the West-of-England hospitals is the South West Genomic Laboratory Hub, based at North Bristol Trust.
8. The transition to the new genetic testing arrangements for cancer has already begun and will continue through a transition period during 2019. The exact timings of transition have not yet been finalised nationally. Transition will be supported by the teams at our local GLHs and GMC.

For queries regarding the 100,000 Genomes Project within the West of England Genomic Medicine Centre please contact: ubh-tr.wegmc@nhs.net

For queries regarding future genetic or genomic testing please contact the Bristol Genetics Laboratory team at: nbn-tr.geneticsenquiries@nhs.net