100,000 Genomes Project Update: The Cancer Programme

Cancer Testing Strategy & NHS Genomic Medicine Service

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Consultant Medical Oncologist at St George’s Hospital
Clinical Lead for Molecular Oncology at Genomics England

17 March 2018
Genomics

• The science of **all** the DNA in the genome

• How it is
  • sequenced
  • analysed
  • interpreted
The first human genome sequence

- Completed in 2000 - cost $3 billion and took more than 10 years
How many genes would they find in the human genome?

• 50,000?

• 100,000?

• 150,000
We have about 20,000 genes.....

The same number as a starfish
Human genome
~3 billion nucleotides

For comparison, there are around 5,500,000 (5.5 million) letters in the whole of the Harry Potter book series.
Cost of sequencing

Today it takes 24 hours costs less than $1000
The 100,000 Genomes Project

Announced by the Prime Minister in December 2012

An Olympic Legacy

Genomics England announced by Secretary of State for Health in speech during NHS 65th Anniversary Celebrations, July 2013
The 100,000 Genomes Project

- **100,000** genomes
- **70,000** patients and family members
- **21** Petabytes of data. 1 Petabyte of music would take 2,000 years to play on an MP3 player.
- **13** Genomic Medicine Centres, and **85** NHS Trusts within them are involved in recruiting participants
- **1,500** NHS staff (doctors, nurses, pathologists, laboratory staff, genetic counsellors)
- **2,500** researchers and trainees from around the world

Focus on rare disease and cancer NHS patients
The Cancer Programme was initiated in 2012 to develop infrastructure for routine high throughput whole genome tumour sequencing for NHS cancer patients.

1. To establish a national research platform of molecular data with linkage to longitudinal clinical data.

2. Transform delivery of molecular testing in NHS clinical cancer care.

Prime Minister announces 100,000 Genomes Project

February: 50,000 genomes sequenced

Fast track cancer results returned within 18 working days

First main programme cancer results returned

Genomics England Clinical Interpretation Partnership (GeCIP) established: >2600 researchers registered in 41 domains

First 11 GMCs (Genomic Medicine Centres) announced

Illumina wins competitive tender for sequencing

First collection of samples for cancer programme pilot (FFPE and FF)

Genomics England set up as a Department of Health-owned company

Sequencing moves to new NHS sequencing centre at Hinxton, Cambridgeshire

First recruitment to main cancer programme

10,000 patients recruited to Cancer Main Programme

First research users access data (WGS and clinical) within research environments
How it works

Patient consent

- Samples
- Clinical Data
- Longitudinal Data

Biorepository

Sequencing Centre

Genomics England Informatics Architecture

Clinicians

GeCIP
Scientific and Clinical Users

GENE Consortium
Industry Users
NHS Genomic Medicine Centres

- 13 Genomic Medicine Centres covering England, and Northern Ireland, Scotland and Wales now on board
- Responsible for identifying and recruiting participants and for clinical care following results
Cancer Analysis & Interpretation for Main Programme (>95% FF samples)

GMC self-reported recruitment to 9 Mar 2018

- 14,799 cancer samples (inc. tumour sample and germline)
- 4 weekly average = 360 samples
Registered participants by tumour type – as of 8\textsuperscript{th} March 2018

<table>
<thead>
<tr>
<th>Tumour Type</th>
<th>n</th>
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</thead>
<tbody>
<tr>
<td>Adult Glioma</td>
<td>399</td>
</tr>
<tr>
<td>Bladder</td>
<td>325</td>
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<tr>
<td>Breast</td>
<td>2207</td>
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<tr>
<td>CUP</td>
<td>116</td>
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<td>Childhood</td>
<td>41</td>
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<tr>
<td>Colorectal</td>
<td>1887</td>
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<tr>
<td>Endometrial Carcinoma</td>
<td>541</td>
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<tr>
<td>Haemonc</td>
<td>198</td>
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<tr>
<td>Hepatopancreatobiliary</td>
<td>294</td>
</tr>
<tr>
<td>Lung</td>
<td>1397</td>
</tr>
<tr>
<td>Malignant Melanoma</td>
<td>219</td>
</tr>
<tr>
<td>Nasopharyngeal</td>
<td>7</td>
</tr>
<tr>
<td>Oral Oropharyngeal</td>
<td>108</td>
</tr>
<tr>
<td>Ovarian</td>
<td>582</td>
</tr>
<tr>
<td>Prostate</td>
<td>722</td>
</tr>
<tr>
<td>Renal</td>
<td>910</td>
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<tr>
<td>Sarcoma</td>
<td>681</td>
</tr>
<tr>
<td>Sinonasal</td>
<td>1</td>
</tr>
<tr>
<td>Testicular Germ Cell Tumours</td>
<td>45</td>
</tr>
<tr>
<td>Upper Gastrointestinal</td>
<td>157</td>
</tr>
</tbody>
</table>
Progress: Cancer Programme
Delivering Clinical WGS

Technical/Logistical Challenges

**LOCAL**
- Pathways for tissue acquisition and processing
  - Tissue preparation (FF/FFPE) [QUALITY]
  - Sample type (biopsies/resections) [QUANTITY]
  - Molecular pathology (Tumour assessment, Digital Pathology) [PURITY]
- Pathways for patients
  - Consent
- Pathways for results and decision-making
  - Tumour sequencing Boards vs tumour-specific MDTs

**CENTRAL**
- High throughput, cost-effective sequencing
  - DNA requirement [QUANTITY]
  - Turnaround time
- Automated analysis, annotation
Progress: Cancer Programme Delivering Clinical WGS

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NHS Molecular pathology transformation

Biopsies into ‘genomics-friendly’ transport media (PBS, alcohol)

Snap-freezing in OPD and pathology: moving away from liquid nitrogen

Digital Pathology assessment driving tissue sampling

Removing formalin from theatres via vacuum-packing

Cellular pathology samples (in vivo/ex vivo)

Lead by Professor Louise Jones & Dr Clare Craig
Sample Handling

AT drop – 4.4
Evenness of coverage – 12.9

AT drop – 15.8
Evenness of coverage – 50.7

Calculated as median for the root mean square deviation (RMSD) of coverage calculated in non-overlapping 100 kb windows
Biopsy overview

- **Total 180** of biopsy samples have been submitted in 2017 and 2018 so far
- 7 GMCs: South London (110), East of England (13), North East and Cumbria (9), North Thames (38) Greater Manchester (6) and Wessex (1), Oxford (3)
- 12 cancer types: Adult Glioma, Breast, Bladder, Carcinoma of unknown primary, Childhood, Colorectal, Haemonc, Lung, Ovarian, Prostate, Sarcoma, Testicular Germ Cell

![Breakdown of biopsies by tumour type](chart.png)
Whole genome sequencing of biopsies

• A total of 180 biopsies have been received in 2017 and 2018 so far
• 2 breast biopsies have failed QC at UKB
• 132 samples have reached Illumina for sequencing so far
• 102 samples have passed QC at Illumina, 11 have failed QC and 19 are awaiting results
• 68 WGS have been delivered back to Genomics England
Progress: Cancer Programme
Delivering Clinical WGS

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‘Samples pertaining directly to the diagnostic function of the 100,000 Genomes Project i.e. tumour samples and germline samples are diagnostic samples. These samples do not fall under the Human Tissue Act 2004 and their storage is not subject to licensing.’

Consensus Statement – Version 1.1 20/03/17

Lead by Dr Clare Craig
Progress: Cancer Programme
Delivering Clinical WGS

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Phase 2
Fast Track summary - weeks 1-25

4 NHS GMCs have been part of the first 25 weeks of Fast Track.

We have recently increased capacity to **40 slots per week**, this is split across the 4 GMCs. It has been agreed that **96 more** samples will be processed in a medium turnaround time (< 20 days) at Illumina from December.

The current average TAT from sample submission to sequence return is 12.9 days.

**759** / **847** Samples sent by NHS GMCs were sequenced (**89.6%**)

- Any samples not meeting the QC parameters at UKB have been transitioned to the normal turnaround time pipeline.
- Discussions with further GMCs are underway for a further stage of the rollout.

<table>
<thead>
<tr>
<th>NHS GMC</th>
<th>Samples sent</th>
<th>Samples passed QC at UKB</th>
<th>Samples sequenced</th>
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</thead>
<tbody>
<tr>
<td>East of England</td>
<td>412</td>
<td>402</td>
<td>396</td>
</tr>
<tr>
<td>North Thames</td>
<td>171</td>
<td>149</td>
<td>131</td>
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<tr>
<td>Oxford</td>
<td>100</td>
<td>93</td>
<td>92</td>
</tr>
<tr>
<td>South London</td>
<td>164</td>
<td>148</td>
<td>140</td>
</tr>
</tbody>
</table>
Variant-level and gene-level actionability in Domain 1

- Variants in a virtual panel of 132 potentially actionable genes
- Actionable genes are defined as genes in which small variants (SNVs and indels <50bp) have reported therapeutic, prognostic or clinical trial association, as defined by the GenomOncology Knowledge Management System
## Domain 1 somatic variants

<table>
<thead>
<tr>
<th>Gene</th>
<th>GRCh38 coordinates ref/alt allele</th>
<th>Transcript</th>
<th>cDNA and protein change</th>
<th>Predicted consequences</th>
<th>Population germline allele frequency (1KG)</th>
<th>VAF</th>
<th>Alt allele/total read depth</th>
<th>COSMIC ID</th>
<th>Gene-level actionability</th>
<th>Variant-level actionability</th>
<th>Gene mode of action</th>
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</thead>
<tbody>
<tr>
<td>BRAF</td>
<td>7:140924594 G&gt;A</td>
<td>ENST00000288602</td>
<td>c.110C&gt;T p. (Ser37Leu)</td>
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<td>N/A</td>
<td>0.08</td>
<td>6/74</td>
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<td>oncogene</td>
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<tr>
<td>BRCA1</td>
<td>17:43094768 C&gt;T</td>
<td>ENST00000057654</td>
<td>c.763G&gt;A p. (Glu255Lys)</td>
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<td>16/116</td>
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<td>tumour suppressor</td>
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<tr>
<td>BRCA1</td>
<td>17:43094624 C&gt;T</td>
<td>ENST00000057654</td>
<td>c.907G&gt;A p. (Glu303Lys)</td>
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<td>7/127</td>
<td>N/A</td>
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<td>tumour suppressor</td>
<td></td>
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<th>Gene-level actionability</th>
<th>Variant-level actionability</th>
<th>Gene mode of action</th>
</tr>
</thead>
<tbody>
<tr>
<td>BRCA2</td>
<td>13:32338167 C&gt;A</td>
<td>ENST00000544455</td>
<td>c.3812C&gt;A p. (Ser1271*)</td>
<td>stop_gained</td>
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<td>7/123</td>
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<td>Therapeutic (ovarian ca); Trial (fallopian tube ca); Trial (ovarian ca); Trial (prim peritoneal ca)</td>
<td>Tumor suppressor</td>
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<tr>
<td>EGFR</td>
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<td>c.2573T&gt;G p. (Leu858Arg)</td>
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<td>N/A</td>
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<tr>
<td>FGF3</td>
<td>11:69816341 C&gt;G</td>
<td>ENST00000334134</td>
<td>c.303G&gt;C p. (Lys101Asn)</td>
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<td>N/A</td>
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<td>5/89</td>
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<td>Trial (solici neoplasm)</td>
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<td>N/A</td>
</tr>
<tr>
<td>FGFR4</td>
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<td>ENST00000292408</td>
<td>c.246G&gt;C p. (Trp82Cys)</td>
<td>missense_variant</td>
<td>N/A</td>
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<td>5/101</td>
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<tr>
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<td>N/A</td>
<td>N/A</td>
</tr>
</tbody>
</table>
Potential clinical trials

National Lung Matrix Trial: Multi-drug Phase II Trial in Non-Small Cell Lung Cancer

This study is currently recruiting participants.

See ▶ Contacts and Locations

Verified October 2016 by University of Birmingham

Sponsor:
University of Birmingham

ClinicalTrials.gov Identifier:
NCT02664935

First Posted: January 27, 2016
Last Update Posted: October 26, 2016

⚠️ The safety and scientific validity of this study is the responsibility of the study sponsor and investigators. Listing a study does not mean it has been evaluated by the U.S. Federal Government. Know the risks and potential benefits of clinical studies and talk to your health care provider before participating. Read our disclaimer for details.

Collaborators:
Cancer Research UK
AstraZeneca
Pfizer
Experimental Cancer Medicine Centre Network

Information provided by (Responsible Party):
University of Birmingham
Structural variants
Mutational density
Coverage and copy number

Mutational signatures

Supplementary Content

Mutation context

Hypermutation rain plots

Cancer types: Signature 1 has been found in all cancer types and in most cancer samples.

Proposed etiology: Signature 1 is the result of an endogenous mutational process initiated by spontaneous deamination of 5-methylcytosine.

Additional mutational features: Signature 1 is associated with small numbers of small insertions and deletions in most tissue types.
1260 cancer whole genomes analysed – Jan 2018
Colorectal genomes (potential actionability)

- Signatures 6 + 15 - defective DNA mismatch repair/MSI tumours
- Signature 10 - recurrent POLE somatic mutations/ultra-hypermutator tumours
- Signatures correlate with high mutation burden. Population MSI deficient and proficient very well distinguished

Trials/therapies
- Green: trial actionability
- Orange: therapeutic actionability

signature signal
- 80
- 60
- 40
- 20
- 0

Cancer signature

Coding SNVs per Mb (log10)

Trials and therapies available for CRC at the gene level

COSMIC
Colorectal genomes (potential actionability)

- This is a subset of variant-level associations with therapies and trials
- Only therapies are associated with specific variants
- KRAS/NRAS variants are mutually exclusive with BRAF
Lung cancer genomes

- Signature 4 - tobacco carcinogens
- Signatures correlate with high mutation burden. Mutation burden distribution is continuous
- We are planning to implement clonal neoantigen analysis into our pipeline
Strategy for utilising WGA

Current:

1. **Enhance recruitment to existing clinical trials**
   - Better curation of existing national stratified oncology trials: FOCUS-4, MATRIX
   - Enhance recruitment to Phase 2 targeted therapy trials

Next steps:

2. **Pan genomic markers**
   - Utilise WGA for stratification as a companion diagnostic (dependent on biopsy pathways, TAT)
   - Better molecular characterisation of tumours/patients

3. **Designing clinical trials**
   - Utilise WGA for stratification (dependent on biopsy pathways, TAT)
   - To better identify potential biomarkers for stratification
   - Better molecular characterisation of tumours/patients
Facilitating recruitment of patients to potential clinical trials

PIK3CA in lung vs colorectal, breast trials

• PIK3CA trials in lung are curated with specific variants, e.g. Q546E, D549N, E542K, H1047L, etc.

• In colorectal, breast cancer are trials associated with ‘PIK3CA Mutation’ will all appear in the ‘gene-level actionability category’

Need for better curation of UK clinical trials

• At the moment we relay on curation of clinicaltrials.gov provided by GenomOncology

• Trial status on clinicaltrials.gov is poorly updated

• Most trials in clinicaltrials.gov are annotated at gene level
Facilitating recruitment of potential patients

National Lung MATRIX Trial SMP2

n=122 patients

- Matrix trial centre
- Metastatic
- Primary
- Unknown if Primary or Metastatic
Facilitating recruitment of potential patients

FOCUS-4 Trial

n=229 patients

2+ Genes (KRAS/NRAS/PIK3CA/BRAF) 37%
KRAS/NRAS 40%
PIK3CA 10%
BRAF 13%

Focus4 Eligible Genes

Metastatic
Primary

Greater Manchester
West Midlands
North Thames
East of England
South London
Wessex
Oxford
Mutation burden across different tumour types

POLE mutation potential MSI
Facilitating recruitment of potential patients

Potential Immunotherapy clinical trials using MSI as a biomarker
Pan genomic markers

Mainstream TMB testing through Genomics England and existing infrastructure

Press Release

Pivotal Phase 3 CheckMate -227 Study Demonstrates Superior Progression-Free Survival (PFS) with the Opdivo Plus Yervoy Combination Versus Chemotherapy in First-Line Non-Small Cell Lung Cancer (NSCLC) Patients with High Tumor Mutation Burden (TMB)

First and only Phase 3 trial to evaluate and show a highly statistically significant PFS benefit with an I-O/I-O combination in first-line NSCLC patients with high TMB, regardless of PD-L1 expression

Results will be shared with regulatory authorities and presented at a future congress

Trial will continue as planned to assess the Opdivo plus Yervoy combination for the other co-primary endpoint of overall survival in patients whose tumors express PD-L1

Monday, February 5, 2018 6:59 AM EST

Obtain supporting evidence: Fast Track all lung biopsies (focus on Fast Track sites)
Clinical review and feedback

- Molecular Tumour Boards established at three laboratory sites for rapid lab validation and clinical interpretation of results in line with local pathways and agreed GMC standards
- All results sent from tumour boards to tumour specific MDTs and leads
- GMC wide Molecular Tumour Board in place to ensure standardisation, consistency and sharing of good practice
- Development of standardised pathways, integrated molecular/pathology reports and generic patient letters
- Networks being developed across GMC region to support LDPs
Moving to a NHS genomics service

Molecular testing: how, when, why?

**HOW?**
- Single gene/Standalone test
- Small panel (eg Amplicon hot spot)
- Large panel (eg Hybridisation-capture)
- Exome
- Genome

**WHEN?**
- LOCAL/REGIONAL DISEASE
  - neoadjuvant chemo-radioRx
  - surgery

- METASTATIC DISEASE
  - Biopsy (diagnostic/recurrence)
  - chemo-Rx targeted drug

- METASTATIC DISEASE
  - +/-biopsy

- Phase II/III Clinical trial

**WHY?**
- Standard of Care ‘actionability’
- Clinical Trials
- Research

- Diagnostic
- Prognostic
- Predictive
- Targeted Drugs

- Single new agent vs SOC
- Multi-arm umbrella/basket
- Experimental n of 1 genomics-drug matching

- Discovery
- Longitudinal patient studies: stratifiers of response
Implementation of a Cancer Testing Strategy

Wave 1
- **Stream Z**
  - Test: Current Methodologies
  - Patients: All eligible patients
  - Targets: Full set of standard of care markers

Wave 2
- **Stream A**
  - Test: NGS panel (up to 50 genes / targets)
  - Targets: Community determined Essential & (Extended) targets
  - Patients: Potential for results to immediately inform management

Wave 3
- **Stream B**
  - Test: WGS
  - Targets: Many : Exemplar cancers
  - Patients: Potential for results to immediately inform management

Movement between streams possible after expert review

Assignment to a specific wave determined after expert review
Acknowledgements

• Genomics England
  • Cancer Team
  • Bioinformatics Team
  • Informatics Team
  • Clinical Data Team

• GeCIPs

• NHS England

• NHS Genomic Medicine Centres

• Health Education England

• Cancer Working Group

• Validation & Feedback Working Group

• Illumina/R&D Team
Thank you

Any questions?
Following the Project

@genomicsengland  #genomes100k

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info@genomicsengland.co.uk

www.genomicsengland.co.uk