Genomics England Experience

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Our history

December 2012
Announced by former Prime Minister David Cameron

July 2013
Genomics England formally launched by then Secretary of State for Health during NHS 65th Anniversary Celebrations

November 2016
Former Prime Minister Theresa May opens a new Sequencing Centre

July 2017
Chief Medical Officer launches Generation Genome and the Life Sciences report

December 2018
Genomics England reaches goal of sequencing 100,000 genomes

January 2019
Long Term Plan “an NHS where access to secure linked clinical, genomic and other data will support new medical breakthroughs and consistent quality of care”
Our vision is a world where everyone benefits from genomic healthcare.

We do so by enabling others to deliver genomic healthcare and accelerate genomic research.
These means we work across the ecosystem:

- **Academia** ~4,000 researchers
- **BioPharma & Start-ups** (over 100 Discovery Forum members, 8/10 Tier 1 Pharma)
- **Tech** (e.g., AWS, Nvidia, Lifebit, Congenica)
- **Sequencing Tech** (e.g., Illumina, Oxford Nanopore)
- **Funders** (e.g., UKRI & MRC & InnovateUK, CRUK, Wellcome, BHF, Lifearc)
GEL Strategy: An Infinity Loop

Evolving genomic healthcare

Accelerating genomic research

Patient

Researcher

Patients
Healthcare teams
Researchers
<table>
<thead>
<tr>
<th>Genomics</th>
<th>Cancer</th>
<th>Rare Disease</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Participants</td>
<td>17K</td>
<td>73K</td>
<td>90K</td>
</tr>
<tr>
<td>Genomes</td>
<td>15K</td>
<td>72K</td>
<td>120K</td>
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- Germline + Tumour
  - 30x
  - 100x

- Germline
  - <20% Singleton

150K 30X genomes equivalents
100,000 Genomes Project Data

Genomics

Clinical Data

- Tumour staging
- Tumour location
- Histological subtype
- Treatment regimen
- Pathology full-text
- Radiology full-text

- Hospital Episode Statistics
- Mental Health Services Data Set

- Mortality data ONS

- COVID-19 status
- Diagnostic outcomes

- Primary Care Data (coming soon)
100,000 Genomes Project Data

Genomics

Clinical Data

Consent

Clinically accredited pipelines for diagnostics

Lifetime follow-up + full retrospective data

Re-engagement
re-phenotyping
re-sampling
re-recruiting
Diagnostic odyssey of children born 2003 onward
Rare Diseases Pilot

68 hospital appointments and 6 years to obtain a diagnosis

18 hospital appointments over 10 years from birth, for unaffected relatives

183,273 episodes of hospital care and cost of £87m, for participants

53,706 episodes of hospital care and cost of £21m, for unaffected participants

25% of cases with diagnosis had actionable findings

Required continued investment in:

- Stronger engineering
- Products and services mindset
- Operational excellence and accreditations
- Implementation in healthcare
NHS Genomic Medicine Service

National Genomic Test Directory

WGS Cancer indications

• Wave 1: Acute Leukemias, Paediatric Tumors, Sarcomas
• Wave 2: Ovarian HGS, Triple Negative Breast, Glioma, Other Heam Onc, Various relapse & refractory

WGS Rare Disease indications

• Wave 1: 20 rare conditions
• Wave 2: +10 rare conditions

>20,000 genomes sequenced in the last 18 months.

How do academics and biotech/pharma work with us?
The National Genomics Research Library

Access review committee

Project proposals

Research Domains

Researcher

Researcher

Researcher

Research projects (Pharma / biotech)

Cloud based Research environment

Data

Data

Data

New science / insights

Publication

Diagnostic discovery

Treatments
Where next?

- Newborns screening
- Increasing data diversity
- Cancer 2.0

**Long-read Sequencing**
To support rapid, precision diagnostics and treatment planning;

**Multi-modal Data**
Enriching our data set for cancer research with the addition of pathology and radiology imaging data alongside whole genome data.
In the long run we aim to enable a lifelong relationship with the genome

- Screening; Rare genetic diseases; NICU PICU
- e.g., Autoimmune disease, Mental health
- e.g., Polygenic risk of common diseases; Pharmacogenomics
- e.g., Cardiovascular; Cancer
- e.g., Neurodegenerative disease