The 100,000 Genomes Project

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Genomics England- mission

• 100,000 whole genome sequences in NHS patients with rare inherited disease, cancers and pathogens from the NHS in England
• Whole Genome Sequencing
• Generate health and wealth
• Legacy of infrastructure, human capacity and capability
• World-leaders in the application of Genomic Medicine for healthcare
Rare Disease- adding future value

- <5% of the population or about 5/10,000 people
- 7000 rare diseases
- Working on 146 disorders
- Detailed and genomically primed phenotyping
- Diagnostics- genomic, imaging, pathology
- Human Phenotype Ontology
- Disease progression
- WGS 30X
- National Rare Disease Registry
Cancer- adding future value

- Disease of disordered genomes
  Drugs target. Tumour heterogeneity, Evolution of cancer, Stratified medicine
- Lung, breast, colon, prostate, ovary and, Leukaemia
- Rare and Childhood Cancers, unknown primary
- Sarcoma, Renal
- Molecular Pathology
- Sequential biopsy of cancer
- WGS at 75x somatic and 30x germline
Infections and Pathogens
Creating the Innovative Platform for Future Health & Wealth

11 Wave 1 NHS Genomic Medicine Centres
Rare diseases, cancers and pathogens
Broad consent, characteristics, molecular pathology and samples

NIHR Biosample Centre
DNA & multi-omics Repository

Sequencing Centre
Wellcome Trust £27m

Refreshable identifiable
Clinical Data
Life-course registry

Linked to anonymised
Whole Genome Sequence

MRC £24m Research Data Infrastructure
Sequential builds of pseudonymised data and WGS
Safe haven - users work within

Primary Care
Hospital episodes
Cancer Registries
Rare Disease Registries
Infectious Disease
Mortality data
Patient entry

Annotation & QC
Scientists & SMEs
Product comparison

Clinicians &
Academics

Training &
Funders

Industry
gene consortium

Firewall
Patient data stays in safe haven

Only processed results pass outside
Establishing infrastructure

11 NHS Genomic Medicine Centre
Awarded 20th December 2014

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Awarded 20th December 2014

Wave 1 GMCs
2nd wave to commence

http://bit.ly/GMCwv1

Greater Manchester
NHS GMC

North East and North
Cumbria NHS GMC

North West Coast
NHS GMC

West Midlands GMC

Oxford GMC

South West GMCs

Wessex GMC

Imperial College Health
Partners NHS GMC

East of England NHS
GMC (inc E Midlands)

University College
London Health
Partners NHS GMC

South London
NHS GMC

NHS GMCs working in partnership with academia, patients and industry through the AHSNs. Regional Genetics Labs and Clinical Genetics Services central to all GMCs

Contracted NHS GMC Lead Organisation working with Local Delivery Partners across the geographical footprint

Biosample centre March 2015

Sequencing centre 31/8/2015

MRC funded Data Centre November 2014
Innovative patient outcomes and data

**Patient Reported Outcomes**

**Multi-omics**

In rare disease and cancer NHS is funding plasma and serum sampling
- RNA transcriptomics, micro RNAs
- Epigenetics, Proteomics and metabolomics

In cancer NHSE is funding
- Cell free circulating DNA (liquid biopsy)
- Sequential biopsies & WGS (trials)

Potential if funds available
- Cancer Cell Lines for drug sensitivity, single cell physiomics or functional genomics (not funded)

**Experimental models**
- Genome editing
- International Mouse Phenotyping Consortium – MRC Harwell
Genomics England Clinical Interpretation Partnership
2500 clinicians and scientists

The standard way

Genomics Research
- Form hypothesis
- Get funds and form collaboration
- Collect, analyse data and validate results

Publication, dissemination, translation
- Publish and disseminate results
- Attempt to translate into healthcare

Healthcare adoption and implementation
- NHS and NICE evaluation and Guidelines
- Education and implementation programme

The GeCIP way

The 100,000 Genomes Project
- hypothesis – WGS will enhance diagnosis
- Coalition of NHS, academics and trainees
- Work together on WGS within GeCIP domains

Enhanced interpretation linked to implementation
- Validate, publish, educate and translate
- The GeCIP Collaborative accelerates Implementation
- Evaluate therapeutic innovation potential

Earlier Healthcare adoption and implementation
- Accelerated diagnosis and health economic evaluation
- Framework for therapeutic innovation

Securing Patient Benefit
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<th>Cancer</th>
<th>Functional</th>
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Building the future of genomic medicine

• 100,000 WGS on NHS patients and pathogens
• Concentrating the UK Genomics Knowledgebase in one location
• The NHS, academics and industry partnerships at the outset to drive Genomic Medicine into the NHS and create wealth
• Building the human capacity and capability
• Key international partnerships to add value
• Leave a legacy of NGS Centres, sample pipeline and biorepository, large-scale data store that makes this usable by the NHS
• New diagnostics and therapies and opportunities for patients

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