Genomics in the NHS

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100,000 Genomes Project: overview

**PRINCIPLES**

- **100,000** genomes from Rare Disease (families) & Cancer (people & tumours)
- **4** key principles:
  - WGS extends current diagnostic scope
  - Recruitment from routine care, treated through routine channels
  - Participants consent to sharing of de-identified data for R&D & industry use & for longitudinal access
  - Establishes model for transformational change

**INFRASTRUCTURE**

- **13** NHS Genomic Medicine Centres covering populations of **3-7** million
- Networked with **90** NHS hospital organisations (of circa 200) to ensure access – outreach clinics into other NHS orgs & link to other UK countries
- Contractual requirements include common protocols, data sharing, collation & submission against agreed data standards & sets
- National networking, groups & events to drive standardisation, sharing of best practice & to drive improvements

**LEGACY**

- **4** key legacies:
  - Increased discovery of new pathogenic variants
  - Integrating advanced genomics into mainstream NHS
  - Increasing public understanding & support
  - Stimulating and advancing UK life sciences industry

- HEE Genomics Education Programme enhances system capacity across the NHS’s **1.3** million staff

**GENOMIC MEDICINE - CORE PATHWAY**

- Identification of suitable patients from routine care
- Involvement of patients in ethics, data & consent issues
- Supply of high-quality processed samples
- Collection of linked phenotypic and clinical data
- Validation of WGS findings and feedback to patients
100,000 Genomes Project Progress

**Samples**
- **62,003** Samples collected from NHS GMCs

**Genomes**
- **35,918** whole genome sequences
- **4,768** cancer genomes
- **23,780** rare disease genomes

**Analysis and Reports**
- **2,931 families**
  - Reports sent to GMCs 20-25% potential action
  - Equivalent to **5,999** genomes

**Scaling up**
- **4,980** genomes in last 1/4

**Additional**
- **60** cancer fast track samples collected turned around in 20 days
- **23,780** genomes scheduled to move into the Research Embassy in October 2017
- +2500 families reports c5000 Genomes in November
Genomics changing lives

**CHILD D**
- 10 year old girl admitted with life-threatening chicken pox
- Previous unusual infections
- Detailed immune testing not found cause
- Mutations found in CTSP1 gene – not familiar to immunologists
- Curative bone marrow transplant
- No risk to siblings
- New testing planned to identify others with condition

**PATIENT J**
- 24-year-old with intellectual disability & visual problems
- Undiagnosed for 20 years
- Defect identified in SRD5A3 gene
- End of ‘diagnostic oddessy’
- Follow-up modified to reflect risk of coagulopathy
- Will help diagnose other families

**INFANT P**
- ‘Failure to thrive’
- Unclassified immune deficiency
- Recruited with consanguinous parents
- Died age 5 months
- Mother pregnant
- Sibling also affected – condition can be treated with Vit B12
- Sibling responding well
- Defect identified in TCN2 gene – transcobalamin deficiency
Building an NHS Genomic Medicine Service from the 100,000 Genomes Project

Building from proven areas of clinical utility (rare disease & cancer)

Key principles

- To ensure comprehensive and equitable access for the entire population (55 mil)
- To improve the quality, value and sustainability of care by providing prompt diagnosis and personalised care
- To support learning, research & development through new collaborative partnerships between the NHS and with academia and UK life science sector and international collaborators/initiatives.
- To retain and build the political, ethical and moral trust of the UK in genomic medicine
The NHS Genomic Medicine Service: Assembling building blocks 18/19 to 20/21

Policy & strategy, coordinating, contracting & evaluation function with a programme of care & fixed budget

Political oversight:
Dept of Health & Ministerial Board

NHS Genomic Medicine Service underpinning Personalised treatments and interventions

Genomic Medicine Centres & Genomic Clinical Services
Integrated clinical genetics & providing population-based care

National Lab Network
7 'new' Genomic Lab Hubs inclusive of cancer genomics

National Testing Strategy
Inc annually refreshed testing directory (single gene – WGS)

Informatics architecture & data store

Whole Genome Sequencing Provision – NHS requirement combined with others

Clinical Interpretation Pipeline

Workforce development
Upskilling of existing staff & ongoing professional engagement in conjunction with Health Education England

Industry/ academic/ international partnerships
Supporting ongoing research & development through clinical care

Advances in genomic & informatics technologies & other next generation diagnostics informing policy, strategy & regulation

Overall service timetable
Procured: Q3/4 '17/18
Operational: Q2 '18 on
Mainstreamed: Q2 '19 onwards
Success requires effective engagement across the health system & society

CLEAR VISION AND OBJECTIVES

Strategic Engagement
- NHS Boards & CEOs
- Academic & industry Partners (AHSN) & LSS
- National clinical programmes eg Cancer, MH, Diabetes, CVD
- Other national initiatives eg Pathology, STPs/ACOs

Clinical Engagement
- Specialist medical (MRCs)
- Multiprofessional
- Broader secondary care
- Primary care

Patient & Public Engagement
- Patient and condition groups
- Local engagement driven by individual NHS GMCs
- Structured ‘public conversation’
- Targeted engagement of groups of public eg school students
Recognising the need for a cross-sector approach to develop healthcare for all

The 100,000 Genomes Project and NHS future plans recognise the importance of supporting parallel activity in academia, research and industry to maximise the development and potential of genomic medicine. It has been structured to actively engage & support research endeavours by clinicians, academia & the life science industry for patient benefit.

This includes:

- Supporting trial design & patient selection
- Repurposing of existing drugs & treatment
- Providing an evidence base to inform identification of new treatment targets & care approaches
- International collaborations to establish standards & share in mainstreaming genomics into practice