Genomics strategy and implementation in the NHS in England

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Chief Scientific Officer for England and Senior Responsible Officer for Genomics in the NHS

Genome Canada leaders’ roundtable on the future of genomics in Canada

5th October 2021
Developing the strategy
History of political and strategic buy-in over the last 10 years

Key commitments

1m WGS (Industrial Strategy)
500k WGS in UK Biobank
100K Genomes Project
~1.5 million other genomic tests
500k WGS in NHS
Ongoing NHS GMS investment
5 Million Early Detection Cohort

GENOMICC and COVID research
Genomics England investment – newborns, cancer tech, diversity
Delivery enabled by whole genomics ecosystem

**NHSX**
Support NHS and care organisations to digitise their services and connect systems through technology.

**HRD UK**
Unite the UK’s health and care data to enable discoveries that improve people’s lives.

**NHS Digital**
Supplies data to the health service and provides technological infrastructure.

**Genomics England**
Partners with the NHS to deliver WGS service and provide Trusted Research Environment to enable access to genomic data to drive scientific discovery and accelerate its translation into patient care.

**NIHR**
Delivers world-leading health and social care research. All NHS sites connected through our national network.

**UK Research and Innovation**
Work with partners to shape a dynamic, diverse and inclusive system of research and innovation in the UK.

**Academic Health Science Networks**
Connect NHS, academics, local authorities, third sector and industry to spread innovation at pace and scale.

**Health Education England (HEE)**
Responsible for the development of a multi professional workforce to meet the demands of a rapidly expanding service.

**Government: Department of Health (DHSC) and Office for Life Sciences (OLS)**
Funds provision through the NHS and research in England. Office for Life Sciences (OLS) is part of DHSC and BEIS and has set the 10-year strategy for sector.

**NHS England and NHS Improvement**
Responsible for setting the overall strategy and commissioning services, including genomics.

**Integrated Care Systems and NHS Trusts**
Delivery of NHS services for defined populations.

**NIHR**
Delivers world-leading health and social care research. All NHS sites connected through our national network.

**Data and Digital**

**Strategy and Policy**

**Clinical Delivery**

**Workforce development**

**Research and innovation**

Genomics ecosystem
Drivers for change – the need to address healthcare challenges

The future direction for healthcare systems will require a balanced focus between:

### Population Health
**Focus on improvements to population health**
- A step change in prediction and prevention of disease
- Earlier diagnosis of disease
- Enhanced screening and prediction
- Influencing lifestyle choices

### Personalised Care
**Increasing personalisation and management approaches**
- Tackling the limits of 'one size fits all' medicine & blockbuster drugs
- Medicines optimisation
- Managing adverse drug reactions
- Identification of new targets and treatment approaches
- Improving outcomes

Challenges for healthcare systems:
- Providing equity of access
- Partnering with other sectors to address social determinants of health
- Ageing – increasing numbers of multi-comorbid populations
- Meeting individual’s needs; coordinated, convenient and accessible care
- Delivering high-quality care
- Delivering affordable healthcare – value to payers
- Increasing rates of non-communicable diseases

Meeting individual’s needs; increasing numbers of people living with multiple long-term conditions

Delivering high-quality care

Delivering affordable healthcare – value to payers

Increasing rates of non-communicable diseases

Ageing – increasing numbers of multi-comorbid populations

Meeting individual’s needs; coordinated, convenient and accessible care

Delivering high-quality care

Delivering affordable healthcare – value to payers

Increasing rates of non-communicable diseases
Supports delivery of disease specific strategies:

UK Rare Disease Framework has four key priorities:
• Helping patients get a final diagnosis faster
• Increasing awareness among healthcare professionals
• Better coordination of care
• Improving access to specialist care, treatments and drugs

An action plan will be published to support implementation

NHS LTP ambitions for cancer:
• by 2028, 55,000 more people each year will survive their cancer for five years or more; and
• by 2028, 75% of people with cancer will be diagnosed at an early stage (stage one or two)

DHSC Cardiovascular Outcomes Strategy:
• Focus on prevention through risk minimisation and genomics
• Support early detection
• Reduce premature deaths from cardiovascular disease
• Ensure equity of access to services

Improving Outcomes through Personalised Medicine
10-year framework for delivery of personalised medicine across the NHS, including:
• improved prediction and prevention based on predisposition
• more precise (and prompt) diagnosis based on cause
• targeted interventions through the use of companion diagnostics to personalise effective treatments
Genome UK Strategy

Published in **September 2020**, Genome UK sets out how the genomics community will work together to harness the latest advances in genetic and genomic science, research, and technology for the benefit of patients.

**Vision**
To create the most advanced genomic healthcare ecosystem in the world, where government, the NHS, research and technology communities work together to embed the latest advances in patient care.

**Prioritisation**
Focussed on three key areas and supporting cross cutting themes to drive societal benefit.

**Coordination**
Implementation co-ordination group with representation from 4 countries of UK to support coordinated implementation.
Ambitions included in the strategy

**Diagnosis & personalised medicine**
- Sequence 500,000 genomes and create the most advanced genomic healthcare system in the world
- Establish a proof-of-concept programme, led by Genomics England in partnership with the NHS, to **explore next-generation approach for the diagnosis and treatment of cancer**, integrating multiple data sources
- **Sequence pathogens** quickly and easily using point of care sequencing technology, helping us control outbreaks and fight antimicrobial resistance

**Prevention**
- Continue to develop a **public health and screening system** that uses genomics to intensify screening and interventions in those at high risk
- Formulate a clear evidence-based position use of **polygenic risk scores** at scale in the health service
- Explore how genomic testing can continue to be best used in **reproductive medicine to support parents to make informed choices**

**Research**
- Ensure that clinical genomic testing and genomics research contribute to **powerful national data resources and maximising impact**
- Coordinate the UK’s existing and future genomics ecosystem, enabling **ground breaking-research at scale** for the benefit of patients
- Achieve **greater diversity within our reference genomes**, and future genome-wide association studies (GWAS) will reflect the UK’s diverse populations

**Workforce**
- Ensure that all new graduating doctors, nurses, midwives, pharmacists, allied health professionals, dental and relevant nonclinical staff have a **level of awareness and knowledge of** genomics that is relevant to their role
- Ensure that the healthcare science workforce continues to have **advanced genomic training and education** within their programmes

**Data**
- Establish a **clear set of standards** for genomic and health data
- Develop systems to enable **federated access to data for research** use to enable comparisons across multiple datasets
Strategy to implementation in the NHS
# NHS Long Term Plan implementation

## Ambition for patients

- **Have the best start in life**
- **Delivering world-class care for major health problems**
- **Supporting people to age well**

## Delivery of the ambitions

<table>
<thead>
<tr>
<th>Give people more control over their health and care</th>
<th>Preventing illness and tackling health inequalities</th>
<th>Backing our workforce</th>
<th>Better use of data and digital technology</th>
<th>Value for taxpayers’ money</th>
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## Genomics commitments

- **Targeted investment in innovation**
  - NHS Genomic Medicine Service will sequence 500,000 whole genomes by 2023/24.
  - Improving recruitment to clinical trials and supporting research

### Earlier diagnosis

- **Access to personalised care**
- **Extended access to molecular diagnostics**
- **Genomic testing routinely offered to all people with cancer**
- **Genomic testing to be offered to all children with cancer to be offered WGS**
- **Genomic testing to be offered to seriously-ill children likely to have genetic disorder to be offered WGS**

### Expanding access to genomic testing for FH

### Cardiovascular

- **Early detection and treatment**
  - Rapid identification of high-risk conditions

### Research

- **Linking and correlating genomic, clinical data and patient data**
  - Routes to new treatments, diagnostic patterns and help patients make informed decisions about their care
Strength of the NHS to delivery of genomics strategy

- Can work at a **whole system level**
- **Longitudinal health records**, inclusive of primary and secondary care, can be leveraged to enhance delivery of individual clinical care and improve health outcomes at population level
- Ability to **adopt at scale**, coordinate and consolidate to drive efficiency and value for money

**NHS system delivery**
- Integrated national system of **universal health care coverage**

**Specialised Commissioning**
- National Commissioning
  - NHS E/I Specialised Commissioning nationally commissions services and secures standardisation
- Genomics Unit
  - Commissioning
  - Data led performance management
  - National investment and business planning decisions
  - Policy and strategy

**Commissioning**
- National investment and business planning decisions
- Policy and strategy

**Finance**
- Labs and Scientific
- Transformation

**Integrated Care Partnerships**
- Integrated Care Providers
  - Commissioners
  - Providers
  - Local Authorities
  - NHS Trusts
  - GPs
  - Voluntary Sector
  - Social Enterprise
  - Private

**Provide direction** through mandating approach, messaging and system to accountability
- Link to **broader system priorities** such as cancer, rare diseases, medicines access and personalised medicine
- Use commissioning to enable **adoption of new technologies**, aligning outcomes with improvements and negotiating ahead of outcome of study
- **Collaborative working** has been strengthened by the pandemic experience
Proof of concept
The 100,000 Genomes Project was announced by the Prime Minister, December 2012

An Olympic Legacy

Genomics England was announced by the Secretary of State for Health at NHS 65th Anniversary Celebrations, July 2013

The NHS "urgently" needs to develop the tools and expertise needed to take advantage of a revolution in genetic testing, June 2011
Strategic approach and infrastructure

- Co-ordinating genomic knowledge to make the UK a world leader
- Sequencing 100,000 genomes to advance genomic knowledge
- Turning genomic knowledge into health interventions
- Ensuring the NHS workforce is skilled & able to deliver for patient benefit
- Using genomic knowledge for prevention & health protection

Frontline services involved
13 NHS Genomic Medicine Centres

Samples + clinical data + longitudinal data

Biorepository
Sequencing centre - Illumina

Validation & reporting in NHS labs

Genomics England Informatics Architecture

Discovery Forum
Industry Users

GeCIP
Scientific & Clinical Users

MDTs & return of results to participants

Patient consent

107,513 genomes available in the Research Environment
- 33,333 cancer
- 74,180 rare disease

122,945 Samples collected and received at the UK Biocentre

Creating direction
Leading transformation
Building readiness
Outcomes for participants

Key findings

• Rare disease – **1,200 disorders with unmet need**
• Diagnostic yield **overall 20%** in rare disease including:
  – **55% in Cystic Kidney disease**
  – **51% in Osteogenesis Imperfecta**
• Cancer – **24 cancer types** included and total of 18,500 participants
  – 50% of all cancer participants to have a **known actionable or potentially actionable gene identified** – tumour specific early phase clinical trials

Activities to complete 100,000 Genomes Project

• Ensure all participants receive **additional findings** both secondary looked for findings and pharmacogenomic profiles
• Review the evidence on reanalysis
• Ensure continued feedback of learning and further determine variants of unknown significance
• Ensure participants can continue to be involved in future developments
NHS Genomic Medicine Service
## Whole system transformation in genomics in the NHS

<table>
<thead>
<tr>
<th>Issue</th>
<th>Action</th>
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<tbody>
<tr>
<td>Variation in access</td>
<td>National commissioning and finance mechanisms to ensure standardisation and service stability:</td>
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<tr>
<td></td>
<td>• Secured funding for infrastructure</td>
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<tr>
<td></td>
<td>• National standards and specifications for delivery of services</td>
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<td></td>
<td>• Regular assurance monitoring</td>
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<tr>
<td>Dispersed infrastructure</td>
<td>Comprehensive clinical and organisational infrastructure to deliver consistent, high quality genomic medicine. Key components of infrastructure include:</td>
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<tr>
<td></td>
<td>• Consolidated laboratory infrastructure</td>
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<td>• Standardised clinical pathways</td>
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<td>• Embedded workforce, education and training</td>
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<tr>
<td>Lack of performance data</td>
<td>Performance monitoring to ensure service is being delivered in line with expected standards</td>
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<tr>
<td></td>
<td>• Benchmarking to understand trends between GLHs</td>
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<td></td>
<td>• Monitor access to testing and improve equitability</td>
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<td></td>
<td>• Inform future commissioning arrangements and understand where additional actions or support may be required</td>
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<td>Clinical leadership concentrated in clinical genetics</td>
<td>A dedicated multidisciplinary clinical leadership across the system</td>
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<td></td>
<td>• Drives and delivers leadership and partnership at all levels including a bottom-up approach to learning and embedding</td>
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<td>• Collaborative governance, system leadership and strategic partnership</td>
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<tr>
<td>Embedding of innovation and new technologies</td>
<td>• Significant NHS investment in 100,000 Genomes Project as proof of concept for whole genome sequencing as part of routine clinical service – set up of 13 NHS Genomic Medicine Centres, extensive clinical leadership and pathway transformation</td>
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<td></td>
<td>• Infrastructure to support rapid adoption of new technologies for example long read sequencing technology and liquid biopsy</td>
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<tr>
<td>Lack of IT connectivity</td>
<td>Developed understanding of fundamental need for data sharing – while ensuring confidence in confidentiality and data security</td>
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<tr>
<td></td>
<td>• Created NHS informatics and data infrastructure for genomics</td>
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<td>• Brought multiple systems together to collate and curate clinical data for an individual</td>
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<td>• Developed a rich clinical dataset with standard nomenclature and data models (HPO, SNOMED CT)</td>
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<td>• Established national database of consented and deidentified genomic (WGS) and clinical data</td>
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Building the infrastructure: NHS Genomics Medicine Service

Overarching aim:
To enable the NHS to harness the power of genomic technology and scientific advances to improve population health and patient outcomes

Key principles:
- Be clinically and scientifically led
- Have patients and public involved at all levels
- Ensure equity of access for all patients
- Have a standardised model of delivery and commissioning across the country
- Be responsive to innovation and new technologies
- Inform and drive change using data led insights

The NHS Genomics Medicine Service Pathway

1. Patient is referred for genomic testing by ‘mainstream’ clinicians or clinical genetics service
2. 7 NHS Genomic Laboratory hubs perform genomic testing, analysis and interpretation and feed back results to clinicians

NHS England and NHS Improvement: Commissions and funds services and is responsible for national oversight, assurance and direction

7 NHS Genomic Medicine Service Alliances: Responsible for multidisciplinary clinical leadership to embed genomic medicine across end-to-end pathways

Collaboration with Health Education England to support workforce development, training and education

Clinical Genomics Services support complex care and other clinical teams. They provide input into genomic MDTS

*Currenting updating the clinical genomics service specification
Delivering the full breadth of genomic testing with horizon scanning

NHS GMS and National Genomic Test Directory has a deliberate focus on the **whole continuum of genomic testing**. To keep pace with **scientific and technical developments** the Test directory is updated annually through the Test Evaluation Working Groups and review of evidence submitted through consultation.

**The testing strategy**

**Continuum of Genomic Testing**

- **Disease focussed**
- **Informing treatment decisions**
- **Population based**
- **Predictive**
- **Horizon scanning**

**Targeted Testing**

- For known causes of genetic disease, e.g., Huntington's Disease

**Panels (10s-100s of genes)**

- Small and Extensive Next Generation Sequencing

**Exome Sequencing**

- Standardising rapid NICU/PICU testing
  - Fetal Exome testing
  - Interim testing while WGS established

**Whole Genome Sequencing**

- Looking across the genome for diagnostic, therapeutic and predictive information

**Examples in Cancer**

- Aim to test 30,000 patients/year: Biomarker testing including for BRCA and Lynch Syndrome
- Standardising cancer panels for pan-solid tumours – high throughput cutting edge technology
- Referrals from all providers
- Establish transport pathways

- Phase 1: 3 cancers, including all paediatric
- Phase 2: pilot triple negative breast and high grade serous ovarian cancer

**Examples in Rare Disease**

- Cascade testing for Familial Hypercholesterolaemia.
- NGS gene panel standardised across England for 323 panel tests

- Phase 1 – 21 rare disease conditions
- Phase 2 – additional 25 rare disease conditions

**Core testing** – delivered by all 7 GLHs

**Specialist testing** – delivered by between 2-5 GLHs with specialist knowledge for interpretation

**National provision** – one provider for the whole country
Developing the testing strategy

National Genomic Test Directory outlines the testing strategy for the NHS and has a deliberate focus on the **whole continuum of genomic testing**.

**Strategy**

Covers **over 3,000 rare disease and majority of solid and haem-onc cancers**

Strategy to move away from single gene testing to **more comprehensive DNA and RNA panel testing** through to whole exome and whole genome sequencing to drive efficiency and productivity.

Developed through review of scientific and clinical evidence

Supported by **clinical leadership and patient and public involvement**

**Evidence**

Evidence from the 100,000 Genomes Project and other national and international studies supported the implementation of WGS in routine clinical care.

WGS clinical indications chosen based on:

- **clinical assessment** – based on current and emerging evidence relating to analytic and clinical validity, and optimal testing technology
- **operational assessment** – technical requirements, laboratory infrastructure, equity of access, pathway implications, cost and affordability elements and commissioning requirements

**Investment and oversight**

**Commissioned and funded nationally** to provide clarity and ensure equity of access – required investment decision

**Mandated for use** in all NHS GLHs to ensure standardisation

Collect data to enable benchmarking between GLHs

Test Directory is updated annually to keep pace with **scientific and technical developments** from evidence submitted by NHS, academia, industry, patient groups

**POLYCYSTIC KIDNEY DISEASE**

**Problem**

- ‘1 in 500 people affected with PKD – autosomal dominant
- Renal failure tends to occur in middle age
- Tolvaptan can slow progression of renal failure if you detect people early enough

**Solution:**

- PKD1 and PKD2 are the main genes
- PKD2 tends to be milder
- PKD1 is technically really tricky to sequence so most families don’t have a genetic diagnosis
- Lots of PKD diagnoses were made through 100,000 Genomes Project

**Impact:**

Having a genetic diagnosis helps with:

- Predicting progression and medicine
- Working out who in next generation needs monitoring (without the genetics can’t be sure until 30 that someone isn’t affected)
- Reproductive options such as PGD

WGS testing now increasingly being arranged by renal physicians with support from clinical genetics
Governance, leadership and partnership – a multi-layered approach

**NHS E/I Genomics Unit** provide national direction:
- Service specifications
- Protocols Quality monitoring

**Shared governance and risk**
- Dives and delivers leadership and partnerships **at all levels**
- Works **across boundaries** – national, local, clinical, organisational, participants and local innovation bodies.

**NHS Genomic Laboratory Hubs and NHS GMS Alliances**
- Provide **population-based care** across organisational boundaries
- Drive local service and professional networks, each with **Medical, Scientific, Operational and Clinical leadership**

**Formal partnerships between NHS England and Genomics England**

- **New partnerships**
- **Data and reporting**
- **Population based care**
- **A coordinated system**
- **National Direction**

**NHS GMS Alliances** working across the geography to bring together **Alliance Networks** engaging all Integrated Care Systems, Trusts, Cancer Alliances, AHSNs Primary Care Networks, pathology networks etc.

Data collection and performance reporting to **drive challenge and change**
NHS System Level

Working with all partners across a geography; includes the ICS, Cancer alliances, regional NHSE/I teams and local clinical leadership and partnership with key partner boards. Governance links with the Genomic Laboratory Hubs.

Clinical Leadership

Genomic medicine focused ‘Supra regional’ clinical directorates
- Engage with clinical leaders, advocates, and champions across a geography
- Creation of clinical genomic senates and other advisory structures

Transformation

Service models and projects to drive embedding of genomics:
- Local and National approach
- 7 key national transformation projects and multiple projects running in the regions

Workforce Development

Working with Health Education England at national and regional levels for appointment of education and training leads by linking needs with priorities.

7 NHS Genomic Medicine Service Alliances across the country, aligned to the geographies of the NHS GLHs, providing:

Cohesive approach

Forward looking

Right people, right place
Example: North Thames NHS GMS Alliance structure

North Thames population approx. 5 million
Lead national transformation project on pathology and Lynch Syndrome

Partnership with patients and the public will ensure scrutiny of our approach and acceptability of solutions. With a paid independent chair, the model is built on a successful model of partnership and participation.

Additional scrutiny and collaboration opportunities from UCLP hosting arrangements and ICHP support

The senate provides a critical opportunity to involve the full health delivery network alongside the genomics partners. ICSs and primary care are particularly important for transformation ambitions

The GMSA provides an opportunity to widen the reach of existing enabling infrastructure (e.g., Education and Training) and consolidate across the partnership geography

Established genomics provision provides the foundations of the Alliance. Over time this will be further harmonised across North Thames and West London
Builds on infrastructure from NHS and 100,000 Genomes Project

- A regional Clinical and Scientific Directorate will enable clinical and scientific partners to work with the executive team to ensure delivery of the business plan (including transformation plans).
- Membership of the Directorate will be adjusted as appropriate once the business plans and transformation projects have been agreed. For example, there will be additional non-recurrent resources that will be put in place for delivery of the transformation plans that may be asked to join the directorate.
- In addition, given the number of specialty leads, attendance may be requested of specific individuals rather than the full cohort. Given they all feed into the Clinical Director, there will be access to clinical leadership for all specialties.

The speciality leads for Genomics Testing ensure clinical leadership is wide reaching. In partnership with the executive team, including Clinical Director, Chief Nurse, Chief Midwife, and Chief Pharmacist, this provides further opportunities to bring together the existing genomics work with the ambitions of the GMSA.
Research driving clinical improvements
Research driving improvements in clinical care

The single biggest driver of genomic medicine success is the ability to build and create partnerships – within and across organisations and across the globe.

Discovery
- Discovery of new drugs and genomic variants
- Exchanging information, sharing resources and best practice

Translation
- Establishing evidence synthesis against agreed policy domains
- Proof of concept studies
- Real world evidence studies
- Example liquid biopsy / long read sequencing

Adoption
- Reviewing the evidence for commissioning and contracting
- Trailblazers / early adopters
- Agreeing common outcome metrics incorporating the design of policy levers

Spread
- Commissioning and finance alignment
- Standardisation of services
- Equity of access

Evolving genomic healthcare:
- Collaborating with researchers to test and confirm variants
- 141 diagnoses returned to NHS laboratories since January 2020
- Further 135 variants undergoing triage to establish suitability for return to the NHS
- Proband diagnosed with osteogenesis imperfecta (OI)
- Researchers identified compound heterozygous variants in a known OI gene
- One variant was a 1.7Mb deletion, and the other was an intronic variant predicted to affect splicing
- Result had immediate relevance for the next generation in the family

Accelerating genomic research:
- 95,600 participants recruited as part of 100,000 Genomes Project
- 117,000 genomics in reading library
- 53 petabytes of genomic and medical data
- 32 bioinformatic pipeline workflows
- 21,000 cancer genomic signatures
- 84 academic institutes
- 3,680 registered academic researchers
- 8/10 top pharmaceutical companies
- 160+ publications using GEL assets
- 550 registered GeCIP projects

Over 94% of NHS GMS WGS patients consented to data being used for research
Better coordination and understanding of genomic research to ensure developments fed into the NHS and patients benefit through dialogues on consent, diagnosis, prognosis and treatment.

Increasing volume of high-quality genomic research in the NHS with reliable genomic insights that are easy to request and interpret.

Researchers through data, infrastructure, insights and environment to collaborate and accelerate fundamental and translational research.

Benefits

Goals

Review proposals and collaborative bids from: Commercial/industry, NHS, academic, voluntary sector/charities etc

Facilitated access to new genomic samples from the NHS GMS

Identify need and opportunities to support further development of genomic medicine

Commissioning through evaluation projects

Respond to emerging technologies

Identification of unmet need

Capabilities and capability monitoring

- Understanding genomic research across England to support more strategic approach to use of resources
- Understand capacity of NHS GMS infrastructure to support research projects

Report submitted from each region detailing ongoing research

Monitoring via NIHR CRN portfolio and databases

Research ongoing via Genomics England Research Environment

323 genomics studies on NIHR CRN portfolio
Engaged in 34 NIHR Programmes
664 NIHR Central Commissioning Facility studies involving genomics
Including clinical trials, genotype/phenotype studies, pharmacogenomics and COVID studies
Using real world evidence to support product introduction

Drivers for Change
- Burden of disease (economic & humanistic)
- Patient journey and current treatment algorithms
- Patient subgroups and competition
- Unmet needs

Research
- Define the need
- Define the desired outcomes
  - Patient outcomes
  - Treatment costs & resource use
  - Adherence
  - Effectiveness/comparative effectiveness
  - Safety/comparative safety

Cost/benefit
- Value Proposition

Introduction
- Product Introduction

Responsive and adaptive pathway
NHS GMS delivery of the genomics strategy

7 NHS Genomic Laboratory Hubs all with high throughput cutting edge technology

203 cancer clinical indications covering majority of solid and haem-onc cancers

357 rare disease clinical indications covering over 3,000 rare diseases

500 gene cancer next generation sequencing panel

Over 323 specialist rare disease panels

600,000 genomic tests performed in England every year

Whole genome sequencing service for 21 rare disease clinical indications and 3 cancers with an average diagnostic yield of 33% - up to 50% in some conditions

250 referrals to rapid Fetal Exome Sequencing Service with diagnosis identified in around 40% of cases

1,200 referrals to date for national rapid whole exome sequencing for NICU / PICU with diagnostic yield of around 40%

3 NHS GLHs are delivering a Non-Invasive Pre-Natal Testing (NIPT) service as part of an evaluative roll out

Over 2,000 NHS staff dedicated to the delivery of the NHS GMS

7 NHS Genomic Medicine Service Alliances led by Clinical Directors

7 national genomic transformation projects

26 local genomic transformation projects

Over 200 funded posts in GMS Alliances to support multiprofessional clinical leadership

Over 920 genomic research projects being supported across the NHS

Over 920 genomic research projects being supported across the NHS

17 clinical genetics services
Genomics is a global initiative with opportunities for international collaborations

A policy-framing and technical standards-setting organisation, seeking to enable responsible genomic data sharing within a human rights framework.

An independent not-for-profit charitable organization identifying opportunities to foster global collaboration to demonstrate value and the effective use of genomics in medicine.

Ongoing challenges to overcome

**Changing end-to-end pathways (engaging clinicians across care spectrum)**
- Expanding existing roles & responsibilities & addressing capacity gaps
- Resources to upskill clinicians & develop genomic literacy
- Formal education & training programmes
- Evolving role of clinical genetics
- Involving all NHS providers
- Embedding genomics and driving change across end-to-end pathways

**Working within an ethical framework**
- Working at speed of public acceptance
- Choice & consent
- Central role for public
- Ethics of diagnostics can be dynamic – cf HIV testing
- Big questions about when appropriate to carry out WGS within the life course

**Retaining & Building public trust**
- Creating a societal “contract” for people to share and contribute their genomic data based on reciprocity, altruism & solidarity
- Ensure appropriate governance frameworks
Themes for success for genomics

**Government and senior healthcare leader interest**
Demonstrate benefit of genomics to population and also to broader agendas such as life sciences or the economy.

**Proof of concept studies**
Alignment with strategy and interface between clinical and research. Support end to end innovation including plans for adoption and scale up. Building the evidence base for ongoing transformation.

**Investment in infrastructure**
Co-design with leadership to enable delivery of a high quality, equitable services that offers value for money, efficiency and productivity. Infrastructure supported by national oversight.

**Receiver pull**
Build the evidence base to demonstrate benefit of genomics to the system and society. Engagement with system leaders and alignment with professional networks.

**User pull**
Co-creation with users through public dialogue. Demonstrate benefit to the users and opportunities for research and discovery.
Lee Morris underwent Ocular Gene Therapy at Manchester University NHS Foundation Trust in one of the first gene therapy treatments undertaken by the NHS.

Lee had vision problems from birth and at 8 years old was diagnosed with RPE65 retinal dystrophy, a rare inherited retinal condition caused by defects in one of a number of different genes.

Other patients have been given the treatment since at other NHS hospitals and the patients are reporting that their vision is improving well.
Thank you for listening!

Keep in touch:

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