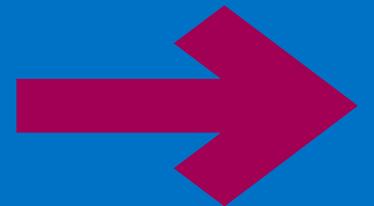


# Integrating genomics into mainstream care: the new NHS Genomic Medicine Service

Prof Sir Malcolm Grant  
Chair, NHS England  
Director, Genomics England Ltd

Jan 2018



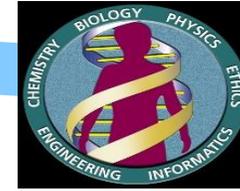
# Why genomic medicine? Why now?



**NHS Genetic Labs**  
working since the 60s



**Long term investment by Gov't in genetic services & workforce**

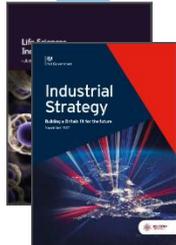


**UK single biggest contributor to Human Genome Project**



**Major parliamentary reports setting out strategic direction**

**Cornerstone of Industrial Strategy to develop UK plc**



**FOR PATIENTS:**



- Enabling a quicker diagnosis & ending the diagnostic odyssey
- Matching people to most effective medications & interventions
- Increasing people surviving cancer through accurate diagnosis & precision therapy



**Building on our Inheritance – HGSG (2012)**

**Commitment to genomics in Mandate and 'Next Steps' plan for NHS**



**>£600 million investment in 100,000 Genomes Project & NHS contribution**



**Generation Genome - CMO (2017)**

# Headlines: 2017

**Feb'17:**  
Completed sequences tops 20,000

Genomes Sequenced

2	0	4	2	9
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**Mar '17:** NHS England Board approves creation of Genomic Medicine Service

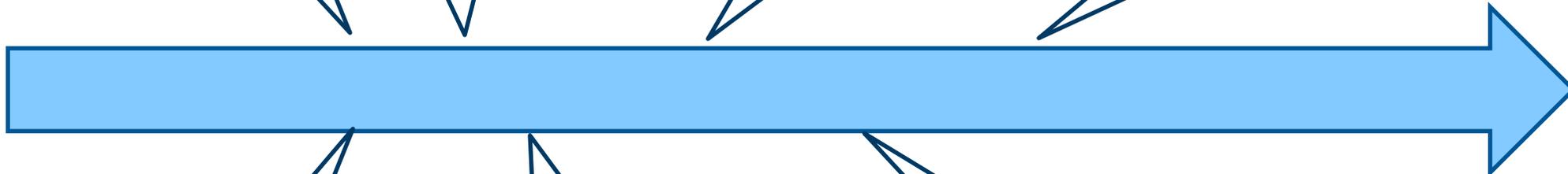


**Jul'17:** Completed sequences top 30,000

Genomes Sequenced

3	1	7	3	0
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**Oct '17:** Cancer programme moves to FF only

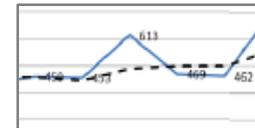


**Feb '17:**  
More than 50 LDPs now part of Project

**Apr'17:**  
Consensus statement on Biopsy developed



**Sep '17:**  
First 800+ samples week



# The 100,000 Genomes Project

## – December 2017

### Samples



**67,446**  
Samples collected from NHS GMCs

Biopsy live and a high calibre cancer genome achieved & enrolment in cancer is growing

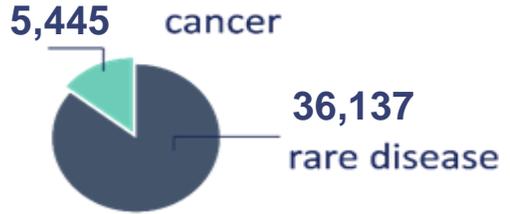
**87** cancer fast track samples collected & turned around in 20 days



### Genomes



**41,582**  
genomes



5,445 cancer  
36,137 rare disease

Scaling up

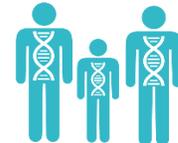


**3,457** genomes since last month

### Analysis and Reports



Reports for **3,337** families sent to GMCs



Equivalent to **6,992** genomes

20-25% actionable findings

**19,865** Genomes and clinical data in the Research Environment



# Building on 100,000 Genomes Project: Establishing the approach for future care

## 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

Project has shown **4-5x** increase in diagnostic yield in Rare Disease and **65%** actionability in Cancer

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

## GENOMIC MEDICINE - CORE PATHWAY



# Moving forward : delivering genomic-based personalisation for patient benefit & service sustainability building upon Project

Technology available, feasible & affordable

Patients/Public ready for change

Clinical Leadership & workforce upskilling

Proof of concept for routine care established

Capturing high quality data & samples for multiple purposes

New service networks & patient pathways being embedded system-wide

Consent & data sharing for multiple purposes demonstrated

System planning & commissioning/economics aligned

## GENOMIC MEDICINE FOR PERSONALISATION



<http://bit.ly/PMvision>

Transformed pathways of care based on careful characterisation of patients facilitating tailored interventions

At home

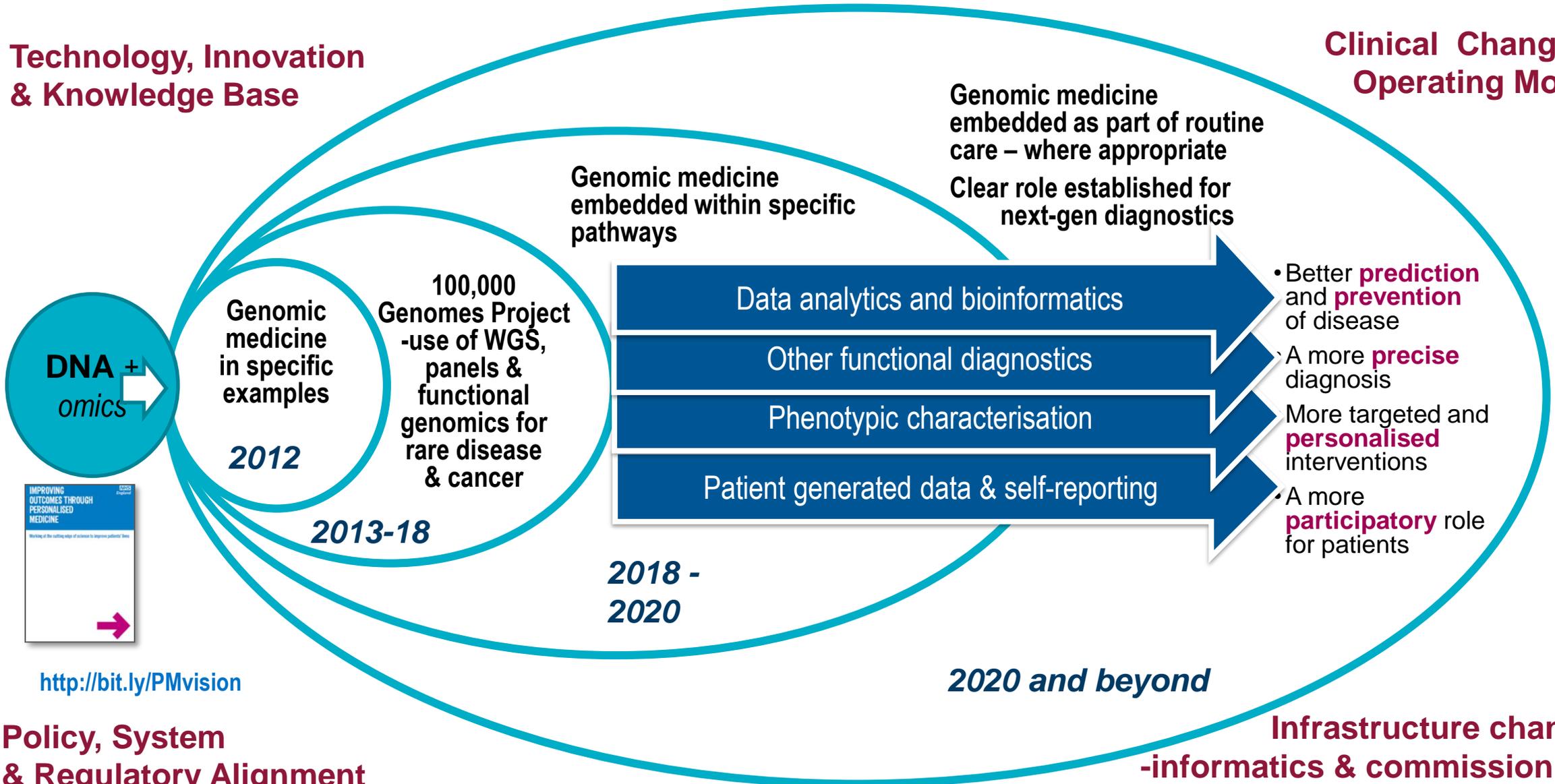
Spectrum of delivery

Specialist Care

# The personalisation journey

Technology, Innovation & Knowledge Base

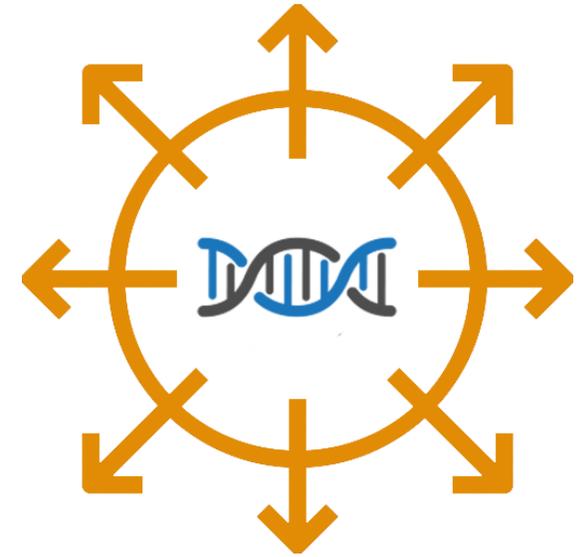
Clinical Change & Operating Model



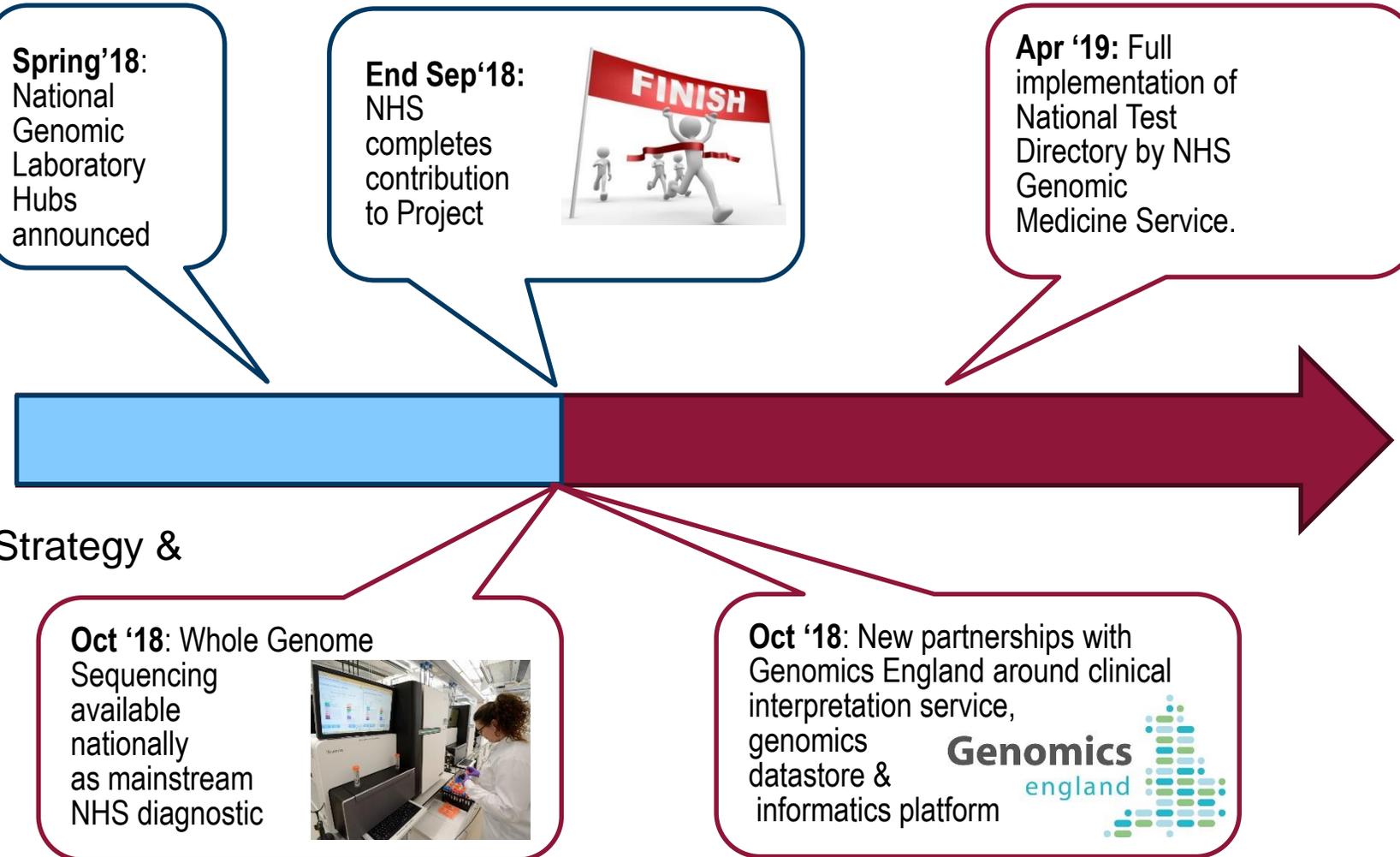
# NHS Genomic Medicine Service: Our aim

The NHS will have:

- A national Genomic Medicine Service providing consistent & equitable care for the country's 55 million population
- Operating to common national standards, specifications & protocols
- Delivering to a single national testing directory – covering use of all technologies from single gene to whole genome sequencing
- Building a national database that will inform academic & industry research & discovery including clinical trials recruitment



# Into the future



Life Sciences Strategy &  
Sector deal



# Assembling & mobilising the building blocks of the service – 2018/19 to 20/21

**Political oversight:**  
Dept of Health & Ministerial Board

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



**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**

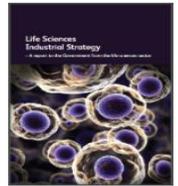
**Whole Genome Sequencing Provision** – NHS requirement combined with others

**Clinical Interpretation Pipeline**

with **Genomics England**

**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*

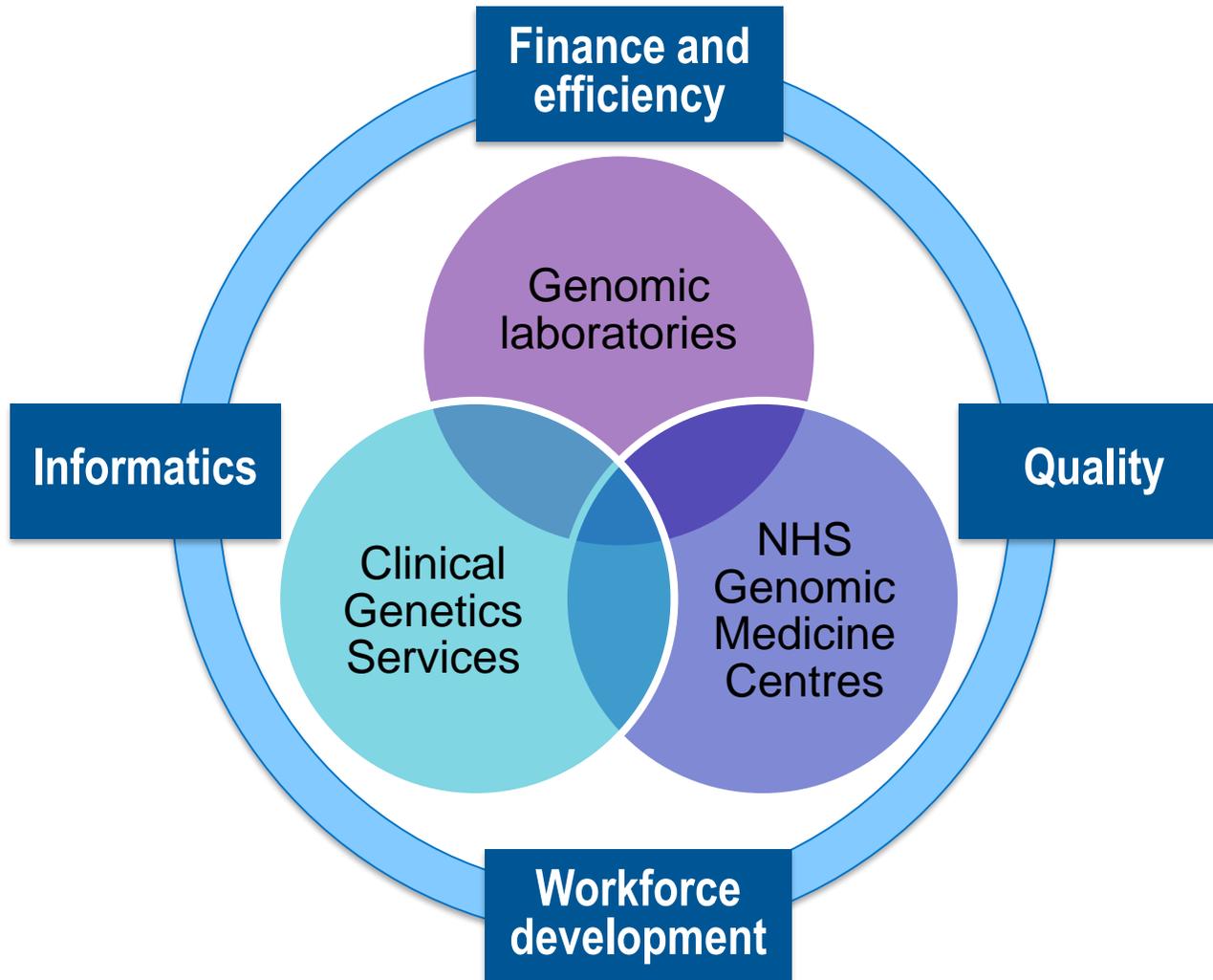


**Overall service timetable:**  
**Procurement:** *Currently live*  
**Mobilisation:** *Q1 18/19 – Q2-18/19*  
**Operational:** *Q3 '18 onwards*

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

# NHS Genomic Medicine Service

## – integrating delivery across the system

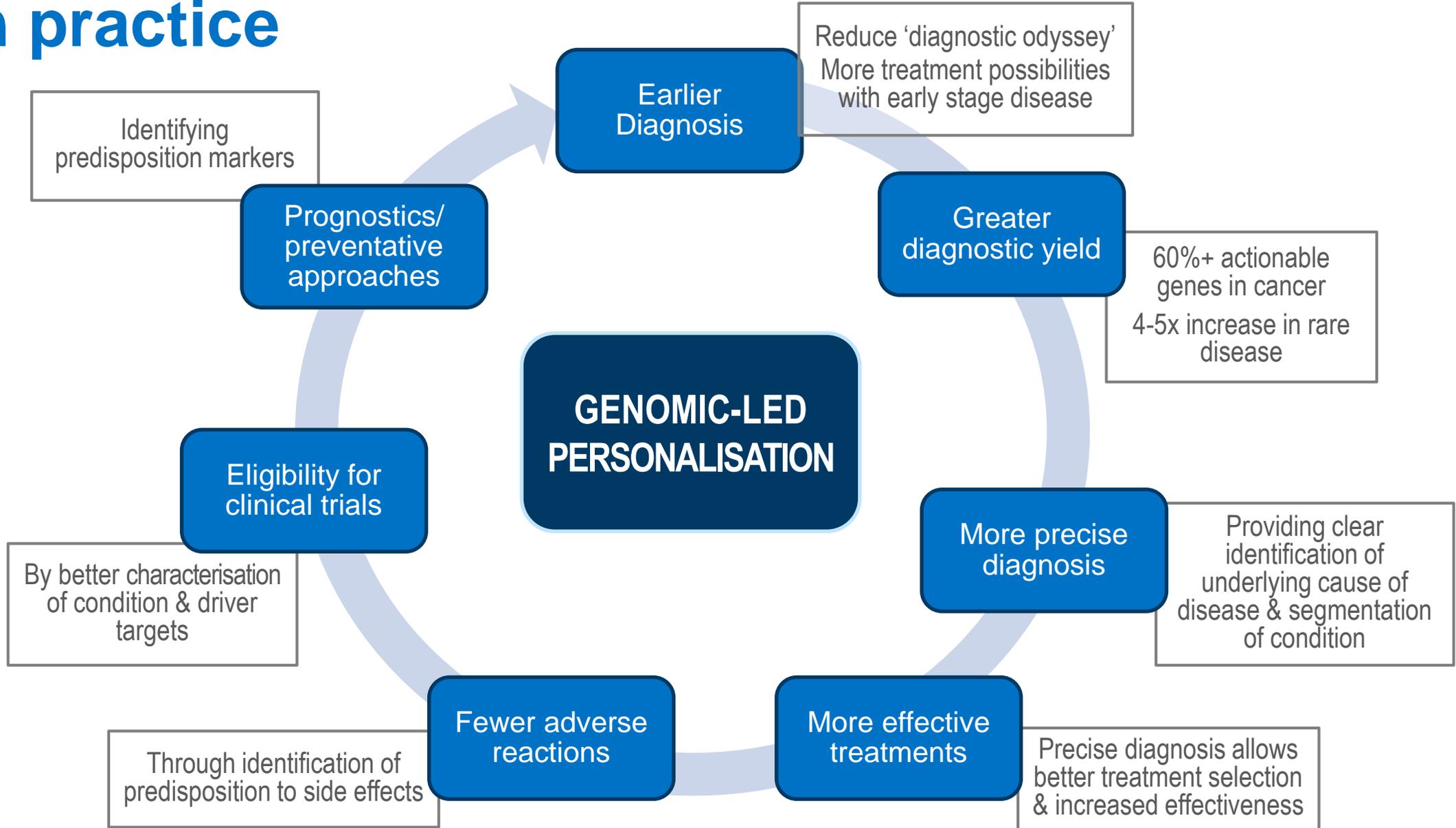


- The Genomic Medicine Service will bring together existing clinical genetics services with NHS GMCs and the new Genomic Laboratory Hubs to provide seamless delivery of service

### Key aspects of future service:

- Ensuring **comprehensive coverage** and access across their geography, including all Hospitals, Specialist Providers and Primary Care
- Enabling access to an **approved genomic test directory**
- **Integrating clinical genetics service** to provide specialist advice and **extending genomic MDTs**
- **Further mainstreaming** & embedding genomics within other clinical specialities
- **Driving medicines optimisation/** appropriate prescribing and **personalisation of interventions**

# Personalisation providing significant advance in practice



# 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 (eg Medical Royal Colleges)

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

# The genomic medicine journey to 2025

## **Today:**

- Variable patient access to cutting-edge genetic technologies
- Proof of concept project demonstrating benefits
- 'One size fits all' treatment based on symptoms
- Limited use of genomic markers
- Diagnostic & clinical data not linked

## **By 2020:**

- National Genomic Medicine Service driving personalised treatments and interventions with consistent & equitable access across the country – underpinned by a National Genomic Test Directory
- Improved diagnosis of rare conditions and better understanding of cancer
- Integrated informatics platform to support comprehensive linking of genomic and clinical data to give a full picture to patients
- Routine care and treatment closely linked through to clinical research, academia and industry with many more patients eligible for clinical trials

## **By 2025:**

- New taxonomy of medicine based on underlying case & personal response
- Integrated clinical services taking a 'whole pathway' approach
- Routine use of Whole Genome Sequencing and newer genomic technologies embedded across multiple clinical pathways
- Genomics included as a fundamental part of clinical training across all professions and levels
- Tailored, optimised & more effective therapies for better outcomes

# Integrating genomics into mainstream care: the new NHS Genomic Medicine Service

Thanks to:

Prof Sue Hill, Chief Scientist, NHS England

Ellen Graham, Deputy Director Genomics, NHSE

Sir John Chisholm, Chair, GEL

Prof Mark Caulfield, Chief Scientist, GEL

