Genomics for All:
From 100,000 genomes to a national NHS Genomic Medicine Service

Professor Dame Sue Hill *@CSOsue*

Chief Scientific Officer for England

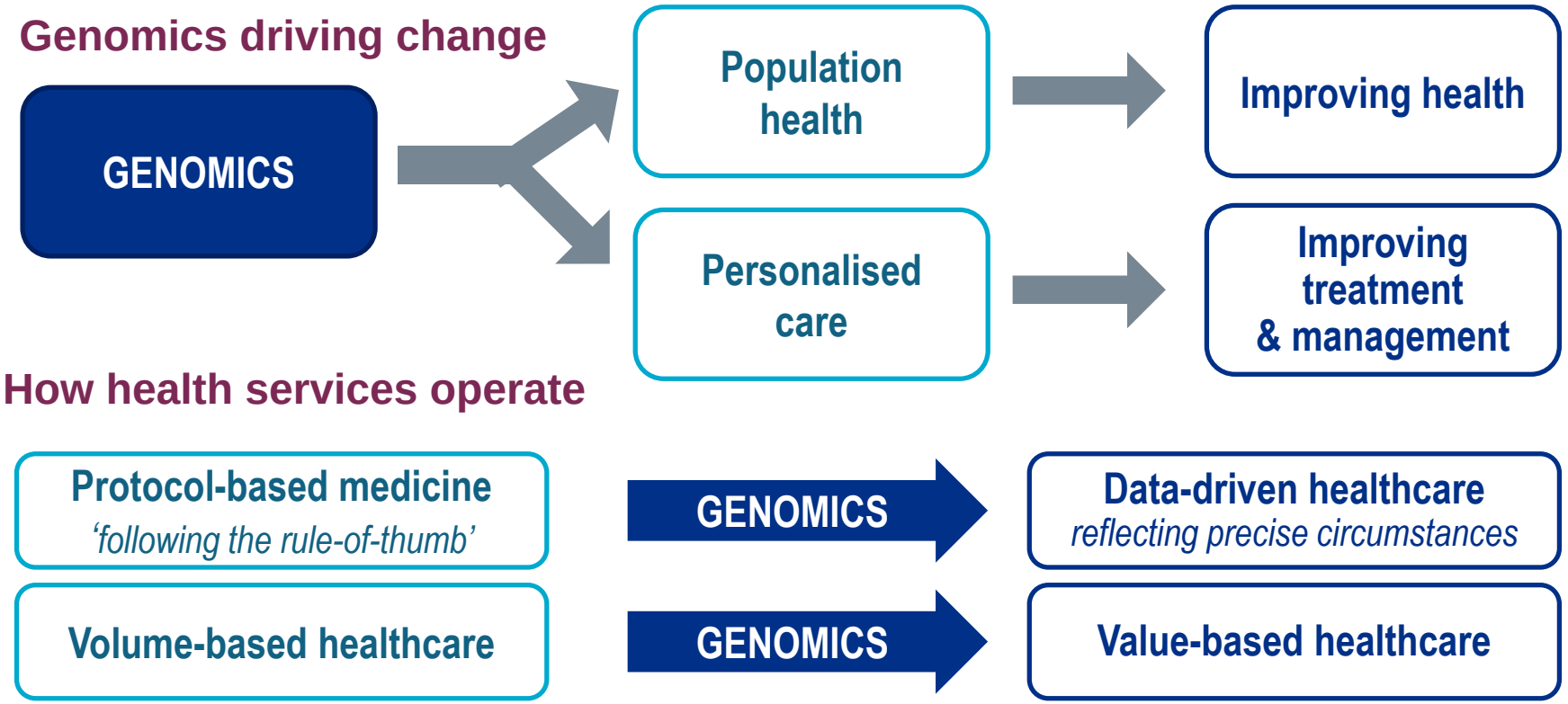
SRO Genomics, NHS England

September 2018

Genomics:
the change that healthcare needs

NHS England

Genomics has the power to drive significant advance in both **what** we do in healthcare and **how** we do it – allowing healthcare systems to tackle the issues of growing demand and maintaining sustainability



Why the genomic revolution now?

NHS England

A combination of factors in recent years have allowed the potential of genomics for routine care to finally be delivered

Technological advance
eg WGS, wearables

+

Computing power
data capture & analytics

×

Cheaper tech
Rapid falls in cost

Genomics will be a disruptive innovation

BUT we will have to make sure that advances can be adopted and delivered rapidly to deliver patient benefit

NHS

The 100,000 Genomes Project

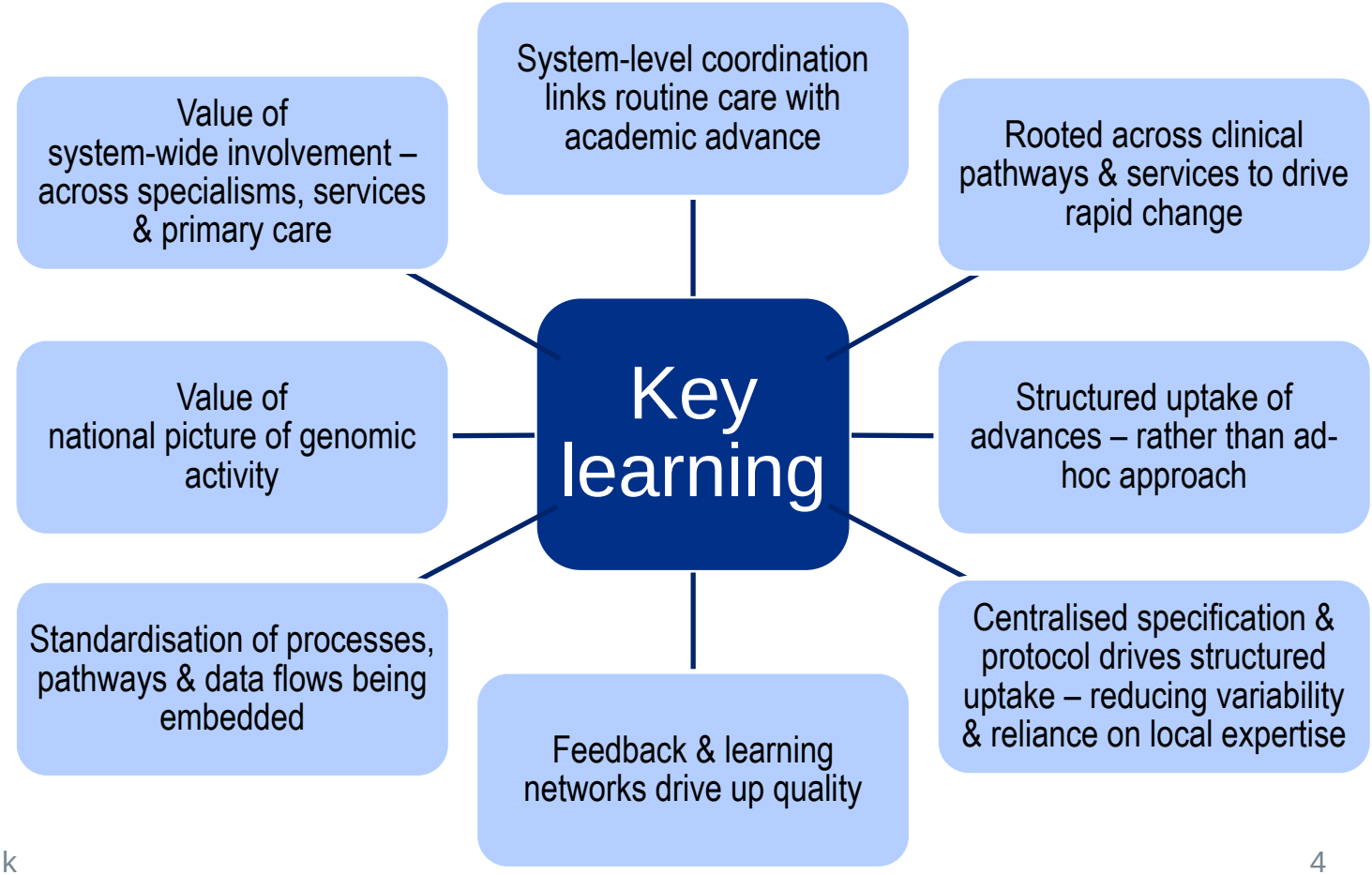
INVENTION → EVALUATION → ADOPTION → DIFFUSION

What we have learned

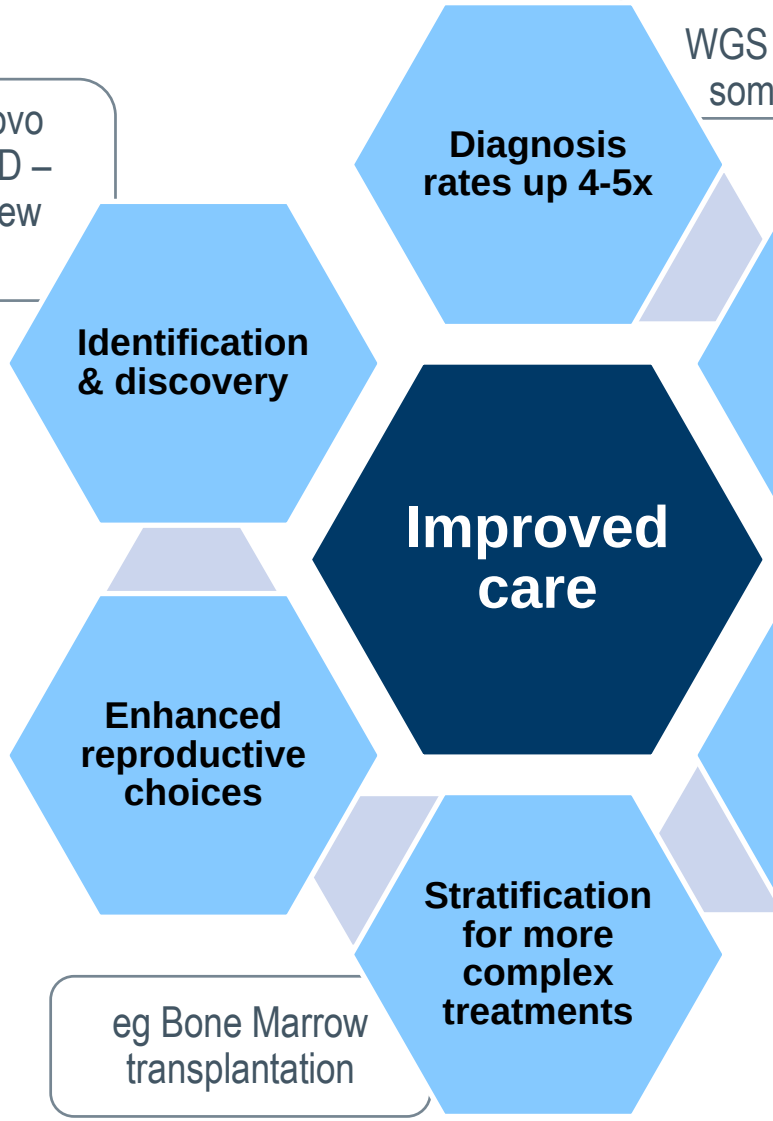
The unique nature of the NHS contribution to the 100,000 Genomes Project has significantly levered learning – firmly linking routine care with academic advance and through taking a system-wide approach *(unlike other genomics initiatives)*

PREVIOUSLY:

- **Variable access**
- **Variable approach**
- **Variable quality**
- **Data often not linked**
- **DNA quality poor**
- **Testing not integrated**



100,000 Genomes: benefits for patients



Up to 10x in some conditions ~ 50% of testing
WGS speeding & simplifying some complex conditions

eg New genes, de novo changes & mutation ID – research informing new clinical diagnoses

Sometimes after decades of previous testing

As simple as vitamin supplements or new diets

eg Bone Marrow transplantation

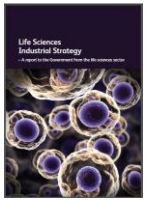
NHS Genomic Medicine Service: vision

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
- All patients to be given **the opportunity to participate in research** *(for individual benefit and to inform future care)*
- Building a **national genomic knowledge base** to provide real world data to inform academic & industry research & discovery *inc. clinical trials recruitment*

The national infrastructure for genomic medicine

NHS England



Political oversight:
DHSC & Ministerial Board

NHS Genomic Medicine Service

NHS England
Strategic Oversight

- Genomic Medicine Centres & Genomic Clinical Services
- National Network of Genomic Laboratories
- National Test Directory

with Genomics England

- Informatics systems & data store
- National Whole Genome Sequencing Provision
- Genomic Data Interpretation

with HEE GEP

- Workforce development
- Industry/ academic/ international partnerships

Built on existing provision – developing capacity to deliver future technologies

+ Strong ethical framework & effective consent + Reflecting diversity of the population

The national genetic testing directory in detail

- The National Test Directory defines all the genetic and genomic tests available through the NHS in England – specifying the appropriate test for each clinical indication. *(~500k tests/yr)*
- Directory has **~300 Rare disease** clinical indications identified across **22 test technologies** with ~75 panels/subpanels and 22 conditions identified for WGS
- Developed through national & international scientific review built upon approach developed by UKGTN
- Annual review process led by expert panel to keep pace with scientific advance

RD Clinical Indication Groups
Cardiology
Deafness
Endocrinology
Eyes
Gastrohepatology
Haematology
Immunology
Inherited cancer
Metabolic
Musculoskeletal
Neurology
Renal
Respiratory
Skin

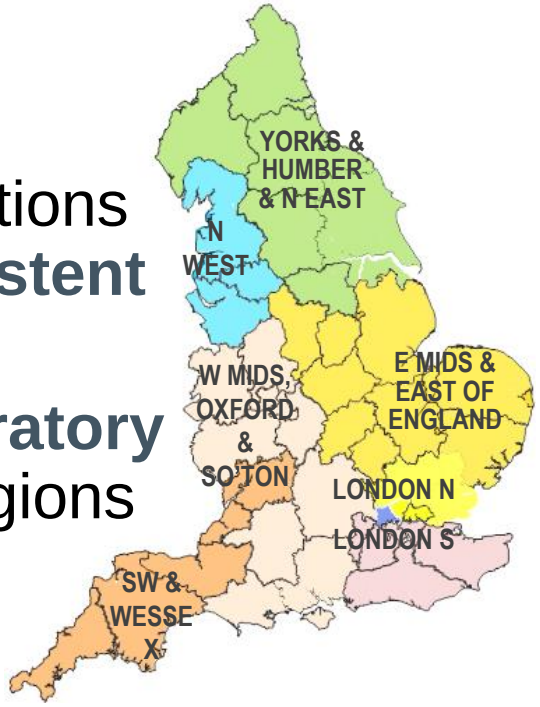
22 clinical indications identified for Whole Genome Sequencing, including Congenital malformation; moderate, severe or profound intellectual disability; neonatal diabetes; cerebral malformation; rare neuromuscular disorders & cystic renal disease

Technology	Est prop'n of reports
Targeted mutation testing	20-25%
Microarray	10-20%
WGS	10-25%
Small panel	10-15%
STR testing	10-15%
WES or large panel	2-14%
MLPA or equivalent	5-7%
Common aneuploidy testing	5-7%
Karyotype	3-5%
Single gene sequencing	3-5%
FISH; DNA repair defect testing; Methylation testing; UPD testing; X-inactivation; Identity testing; Microsatellite instability; NIPT; NIPD; PGD	each <2%
Other	2-5%

Provided by a new national laboratory system

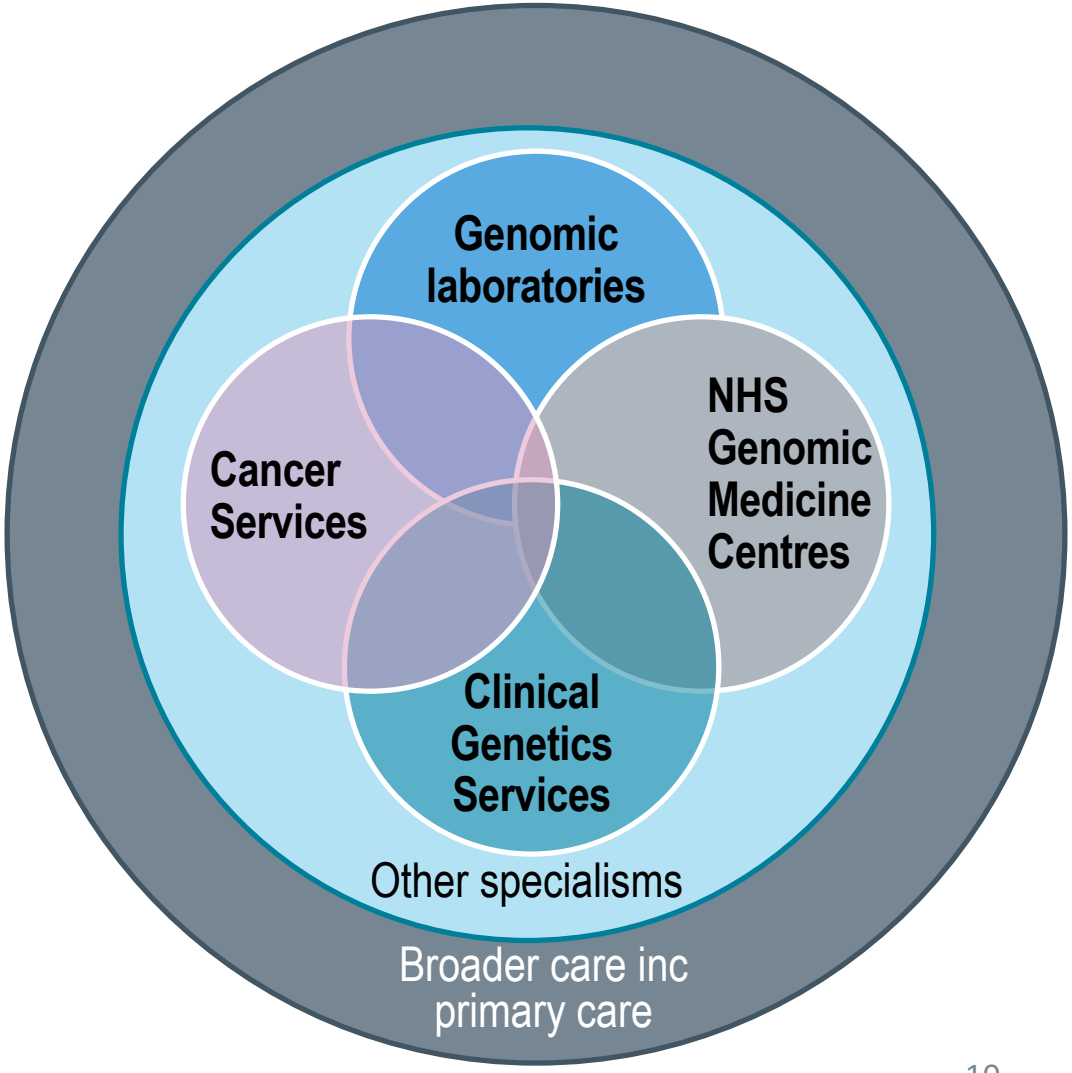
- NHS England have brought together the country's laboratory provision to form a **single national system** providing **equitable & consistent access** to the testing set out in the Test Directory *(initially rare disease & cancer)*
- Standard operating procedures, specifications and common informatics to ensure **consistent high quality** & comparability of results
- Delivered through seven **Genomic Laboratory Hubs** coordinating testing across their regions
- Driving consolidation to ensure **efficiency & quality** – with clear transition plans

Genomic Laboratory Hubs mapped to NHS STPs
(Sustainability & Transformation Partnerships)



Mainstreamed across clinical pathways

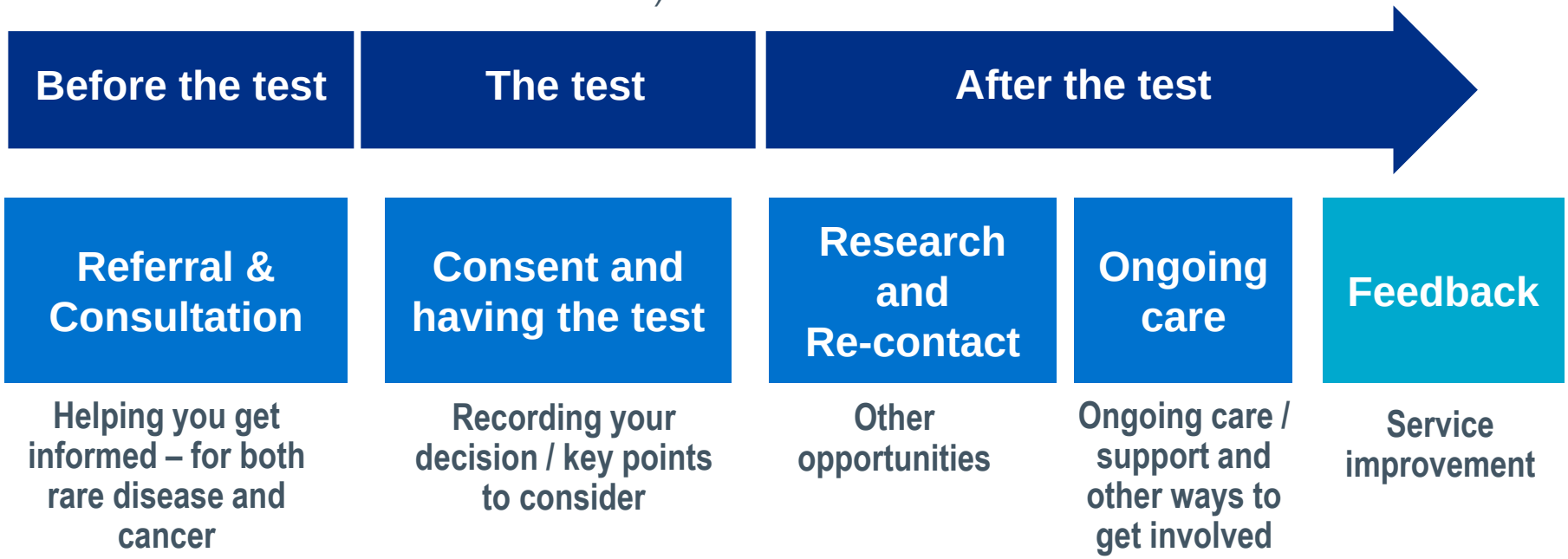
- Core genomic services form a nucleus to drive mainstreaming & access to genomic medicine
- Role of NHS GMCs and Clinical Genetics will evolve to support a focus on end-to-end pathways for patients
- Requires workforce development and upskilling



Patient choice: informing the journey

To support the implementation of the Genomic Medicine Service, a new patient choice (consent) model is being developed. Key aspects are:

- Aim to set clear and informed choice about use of NHS GMS to patients
- Choice supported by plenty of information to help patients understand choices & implications
- Clear and distinct choice to be part of research programme *(without choice impacting on standard of clinical care of individual)*

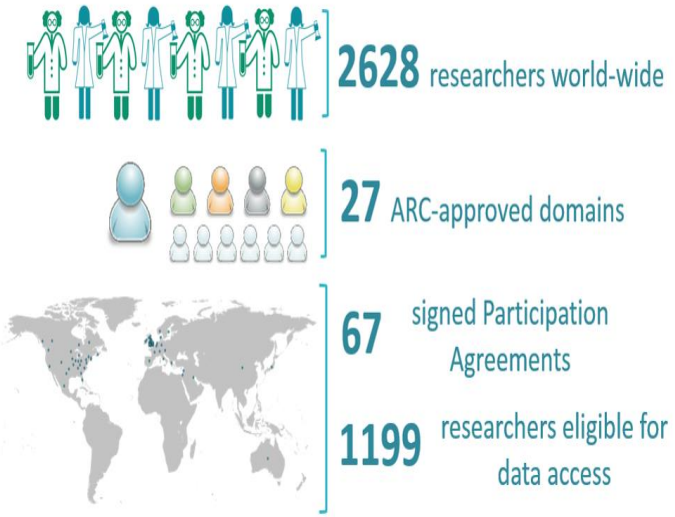


Driving research & discovery through real-world data

NHS England

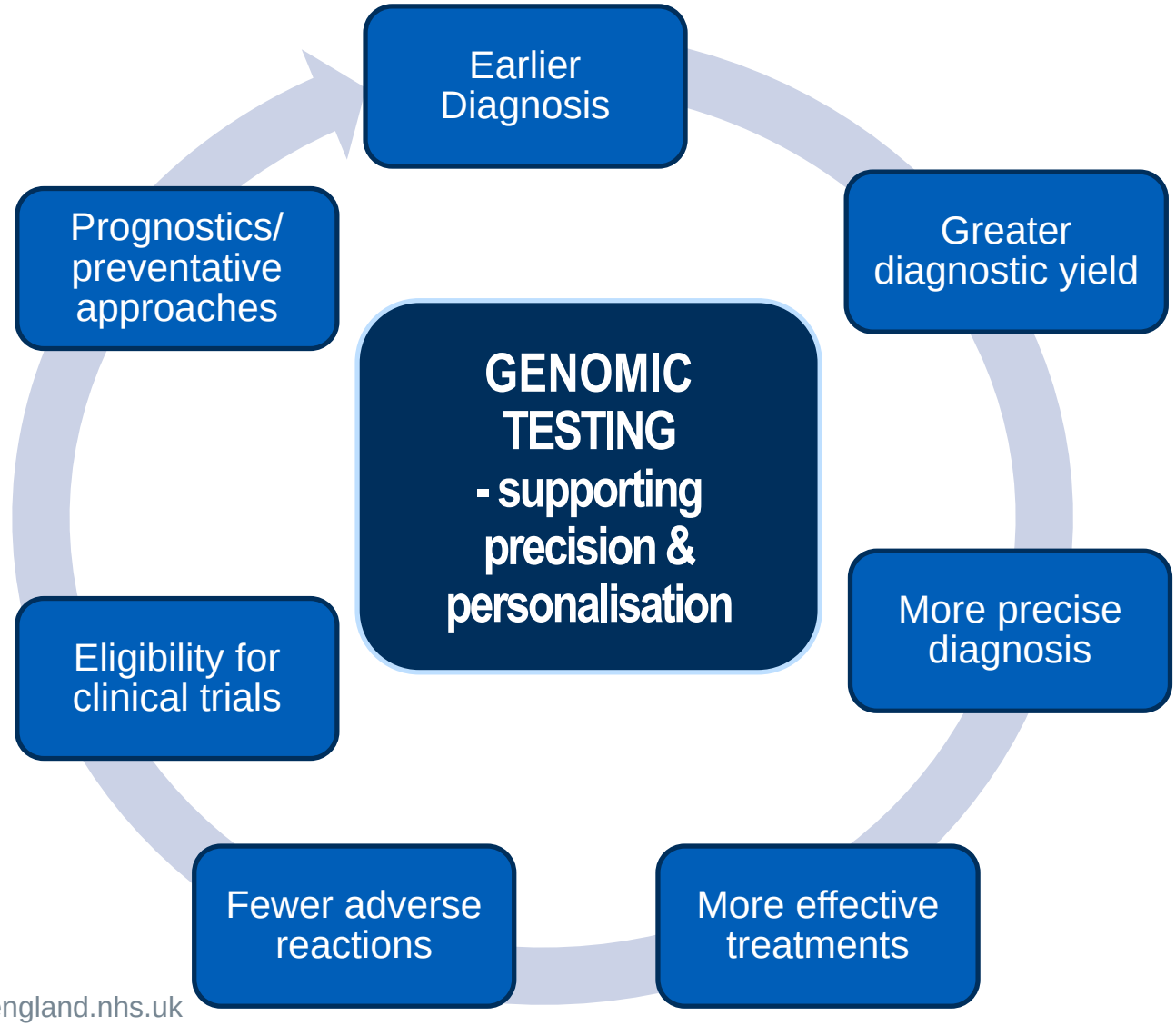
- The NHS Genomic Medicine Service will significantly speed research and discovery due to the significant volume of **real-world data** generated
- This provides **much richer information** to inform the development of new treatment approaches
- The power of this data is being harnessed through new **partnerships with industry** and **supporting academics** through the work of the Genomics England Clinical Interpretation Partnership *(GeCIP)*, driving discovery across 40 research areas (domains) *inc 14 domains covering Rare Disease*

GeCIP by numbers



INVENTION → EVALUATION → ADOPTION → DIFFUSION

Genomics: higher quality care for all



Given the potential of genomics to transform care, it is likely to feature strongly in the 10-year Long Term Plan currently being developed for the NHS

Current and future steps to deliver genomics for all

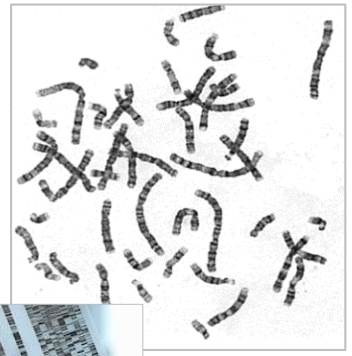
Current focus of NHS activity: consolidating key services & infrastructure – for **standardisation & efficiency**

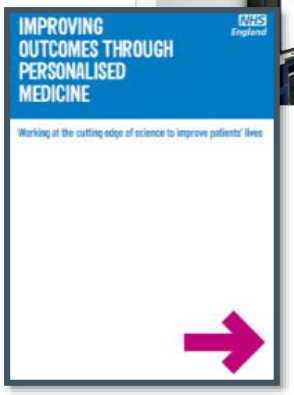
Carefully building a robust & sustainable service – to provide the **platform for future** advances

Crucial to have **public dialogue** on the importance of genomics to build familiarity & comfort with these technologies

Establishing & embedding pathways to drive genomic **mainstreaming across all clinical specialties** *(inc primary care)*

Need ***change across the whole workforce*** – ongoing over next 10-20 years - to deliver the full potential of the technology





http://bit.ly/PMvision