Genomic Medicine and the NHS in England

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The NHS – harnessing its unique potential to deliver future care

- Integrated Population health system for 65 million people in UK

In England
- ~250 NHS provider organisations
- ~7,500 general practices
- 1 million patients every 36 hours
- Collected data and longitudinal health records – all have a unique NHS number
- Coordinated commissioning system that influences and shapes provision of care, including new models
- Partnerships at system level and within local health economies
Vision: addressing the challenges in health systems

- Driving down disease burden
- Managing multimorbidity
- Standardisation & industrialisation
- Capturing & harnessing data
- Science & technology driving new care approaches
- Personalisation to address heterogenous disease
- Moving care outside of hospital
- Patients & Society

‘Five Year Forward View’ cross-system strategy identifies three main areas for action:
- Care & Quality gap
- Health & wellbeing gap
- Finance & efficiency gap
Recognising role of science and technology in future NHS sustainability

“The NHS needs to adapt to take advantage of the opportunities that science and technology offer patients, carers and those who serve them.

“...Most countries health care systems have been slow to recognise and capitalise on the opportunities presented by the information revolution. We will exploit the information revolution”
Into the next generation – science moving beyond genomics alone

Profound scientific & technological innovation influencing NHS transformation and effectiveness
Harnessing genomics for a significant advance in diagnostic yield & reach

Building on history and tradition of genetic advance and genetics in NHS

Exponential growth in information – much more that is actionable today and currently available in NHS

Informatics: Combining information to objectively define disease phenotypes

Opportunities to further increase diagnostic yield, coupling with functional genomics

POCT developments providing real-time clinical opportunity

Patient Push | GENOMICS | Science Pull
Genomics securing sustainability & improved outcomes: value-based care

The future direction for healthcare systems is clear – with, at one end, a focus on improvements to population health and, at the other, increasing personalisation of care and management approaches.

• A step change in prevention
• Earlier diagnosis of disease
• Enhanced screening and prediction
• Influencing lifestyle choices

• 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

Delivering improved outcomes and improved system sustainability

VOLUME-BASED HEALTHCARE

VALUE-BASED HEALTHCARE
100,000 Genomes Project: a world-leading model for healthcare transformation

Project announced in 2012 by then PM David Cameron and remains a Prime-Ministerial commitment as a world-leading project

Key principles underpin the Project and the NHS contribution:

- **WGS extends** current NHS funded diagnostic repertoire
- Recruitment of patients with Cancer and Rare Disease from routine care
- **Participants consent** to sharing of de identified data for R&D and for access to longitudinal records
- Moving from proof of concept to implementation in 3 years and aligned to major system priorities *(UK Rare Disease strategy and Cancer Taskforce)*
- A model for transformational change in the NHS as well as delivering science and partnerships with industry

Project ends Dec 2018 – always an expectation the NHS would commission WGS & embed Genomic medicine from 18/19

Integrating advanced genomics into mainstream NHS

100,000 Genomes – the major legacy

Increased discovery of new pathogenic variants

Stimulating and advancing UK life sciences industry

Increasing public understanding & support

New collaborative partnerships

Leading to new diagnoses & treatments, devices & diagnostics

Raising awareness & engagement of use of genomic data

Building in knowledge, pathway & approaches to core care of the future

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100,000 Genomes – the major legacy
NHS Genomic Medicine Centres: delivering the genomic future

• Established to provide NHS contribution to 100,000 Genomes Project - **patients from routine care, treated through routine channels**
• Coordinating care for popn’s of ~3-7 million, responsible for pathways & services across their geography.
• Lead organisation builds networks with other trusts as Local Delivery Partners (>90 Trusts involved at completion of Project)
• Capture improvements in clinical outcomes & drive change in clinical utility
• Networking and sharing to drive transformation
• Underpinned by HEE Genomics Education Programme and investment in genetic scientists and bioinformatics

**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
Sharing and transforming practice: Professional networks

NHS England coordinates a series of network groups bringing together staff from the NHS GMCs in areas of key transformation:

- Clinical Directors
- Informatics
- Programme Leads
- Patient & Public Involvement
- Laboratories
- Rare disease
- Cancer
- Pathology

+Various transition & working groups

These professional networks exist to share experience, ideas and good practice to accelerate the development, adoption and mainstreaming of new genomic approaches.
Enhanced referral & consent processes & new genomic practitioners

Involvement of broad range of clinical specialities & teams

Improved phenotypic characterisation of patients & collation of data from multiple EPR & other systems

Establishment of Rare disease & Cancer Genomic MDTs & laboratory validation of WGS variants

Distributed clinical leadership & new approaches to clinical & professional engagement

New ways of working with patients & patient groups & engaging with BME groups

Clinical & Service Transformation
Project has created a World-Leading Infrastructure for genomic medicine

National elements

- National Biorepository
- Whole Genome Sequencing Provider
- National Genomic integrated Database
- National Genomic Interpretation Pipeline
- National Knowledge Base

Pharma, Biotech and Diagnostic Companies investing in the UK

2500 Researchers providing Insights and Discoveries

Network of 13 NHS Genomic Medicine Centres serving ~3-5m population, inc data hubs

13 Lead organisations & 75 local NHS trust delivery partners

Multiple hospital sites and outreach locations

NHS molecular diagnostic and genomic laboratories

Access through a range of specialties & services
How the elements of the Project fit together
Rare disease: cross-specialism reach to improve diagnosis

• 207 rare and inherited diseases currently in scope with more under consideration with a clear process for assessing new conditions.

• Aims of RD Programme include:
  • To add value with additional biological insights from WGS
  • To develop a programme of functional multiomics pathways & biomarkers
  • To create a unique dataset that may enable therapeutic innovation

• Meets unmet need: ~30% diagnostic yield (cases previously –ve in standard tests)

• Recruitment outside of clinical genetics - average 50%

• Family structures good

• Project shown how senior clinicians across the range of conditions are recognising and engaging with the growing role for genomics

- Neurology
- Endocrinology
- Cardiovascular
- Renal Medicine
- Haemotology & Immunology
- Rheumatology
- Respiratory
- Dermatology
- Oncology
- Infectious Disease
- Surgery
- Intellectual disability & Paeds
- Psychiatry
- Emergency Medicine

Cardiovascular
- Familial Hypercholesterolaemia
- Familial cerebral small vessel disease
- Severe hypertriglyceridaemia
- Familial Thoracic Aortic Aneurysm Disease
- Brugada syndrome
- Long QT syndrome
- Catecholaminergic Polymorphic VT
- Unexplained sudden death in the young
- Short QT syndrome
- Idiopathic ventricular fibrillation
- Arrhythmogenic R Ventricular Cardiomyopathy
- L Ventricular Noncompaction Cardiomyopathy
- Dilated Cardiomyopathy
- Dilated Cardiomyopathy and conduction defects
- Hypertrophic Cardiomyopathy
- Melge disease
- Miliroy disease
- Lymphoedema dietrichsais
- Lipoedema disease
- Primary lymphoedema
- Pulmonary arterial hypertension

0-10 families
11-100 families
101-500 families
>500 families
New disorder
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

Defect identified in TCN2 gene – transcobalamin deficiency
Sibling also affected – condition can be treated with Vit B12
Sibling responding well
Lifting diagnostic & consultation pressure on NHS services

- An analysis of HES data on Project participants showed the extent of NHS contact they currently have
- 2 million Episodes on 31,781 participants matched 98.2% of participants – **average 63 episodes/person**

**Georgia’s story**
Now age 7. Developmental delay. Multiple medical problems including unusual eye and kidney problems
No cause known despite extensive testing. Negative exome result in ophthalmology research study
**HES:** 151 outpatient attendances across 10 specialties
10 day case admissions, 2 inpatient admissions including 12 days for cochlear implant
**WGS:** mutation in *KDM5B* found; not present in either parent (‘*de novo*’)
**Benefits of diagnosis:** Ends 4 year diagnostic odyssey & repeated NHS visits. Informs parents on risk of recurrence in another child (very low)
Recruitment in cancer

- 13 common cancers included in Project:
  - Breast, Ovarian, Prostate, Lung, Colorectal, Sarcoma, Renal, Adult Brain Tumour, Bladder, Endometrial, Melanoma, Upper GI Tumours, Testicular, Haematological
  - Aim of programme to understand tumour evolution and drive drug discovery and clinical trials

- Multiomic samples taken to assess potential technologies eg cfDNA

- Series of initiatives to facilitate and speed recruitment in cancer:
  - Exemplar model of genomic analysis in cancer care developed
  - New pathways established for Fresh Frozen tissue
  - 3 Accelerator sites to build clinical pathway
  - HTA approved consensus statement opening up new biopsy pathway – more engagement across clinical teams and greater role for endoscopic procedures and interventional radiology
  - Fast track pathway developed (currently from submission to sequence to return of results in 17 working days)
Cancer: providing clinical reports based on actionability

Early analysis found 65% of Project cancer cases have variants in actionable genes

- WGS provides extra diagnostic reach (structural variants, copy no. variants etc)
- Global patterns of mutation (‘signatures’) proving informative
- Defining causal drivers from benign ‘passengers’ is critical
Academic partnerships at the forefront of discovery & innovation

Aim is to accelerate the adoption of new approaches from ~17 years to <5 years

- 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
Engagement with the global academic research community

Genomics England Clinical Interpretation Partnership (GeCIP)

- Partnership between over 2,000 researchers across the globe inc NHS staff, trainees, plus international collaborators.
- Designed to accelerate academic/industry partnership and development of diagnostics and therapies
- 40 topics (domains) of research.
- Most cover a single disease or group of diseases within rare disease or cancer.
- In addition several cross-cutting domains (eg epigenomics, health economics and technology.
- Carefully controlled access to pseudonomised data from the National Genomics Dataset
- In turn, All data generated contributes to the Genomics England Dataset
A structured approach to genomic upskilling & education

• Health Education England Genomic Education Programme established alongside 100,000 Genomes Project with the overarching aim of ensuring the workforce of today and tomorrow has the right numbers, skills, values and behaviours, at the right time and in the right place.

• 3 initial objectives:
  - Support 100,000 Genomes Project
  - Increase capacity and capability
  - Embed genomic medicine into mainstream practice

• Structured to deliver
  - Comprehensive reach across 1.3M NHS staff
  - Impact across whole education continuum for prospective and current workforce, across all professions

• 2020 Goal: Embed genomics education at all levels of the current and future workforce.
Continuum of genomic medicine education – applied across the professions

The HEE Genomics Education Programme has developed a range of tailored resources and is incorporating genomic learning into basic undergraduate training across the clinical professions and also postgraduate professional development.

Genomics knowledge into action

*Pre-registration
Core genomics concepts
clinical examples & relevance to patient care

Post-registration
Contextualising the genomics concepts
tailored for specialty/role

Workplace
Genomics in clinical practice
Competences for role undertaken;

The GEP has also developed new professional roles & training

- New professional roles specified (eg Bioinformatics, Genomic Counsellors) with new funded training courses, and routes to formal recognition through statutory registration
- Development & funded growth of existing roles through funded places (eg Molecular Pathology)

Research capacity is being built through multi-level fellowship programme
The new multiprofessional Masters in Genomic Medicine

- A central part of the HEE Genomic Education approach has been the development of a multiprofessional Masters in Genomic Medicine with a curriculum specifically designed to support modern genomic medicine.
- The course has a modular structure with core modules (e.g., Genomics of common & rare inherited disease) and optional modules from Advanced Bioinformatics to those covering ethical, social and economic issues.
- Individual modules can be taken for CPPD. A research project founded on access to the National Genomic Database is a core part of the course.
- The course is delivered through a national network of 10 leading universities, providing breadth of geographical access with individual centres delivering modules building on their existing specialist areas.
- Over 600 fully-funded places offered to NHS staff to ensure broad upskilling – but courses open to students beyond the UK.
Resources

► **WGS MOOC**
  - Online ‘social learning’ style course on FutureLearn
  - Developed for health professionals with limited understanding of the sequencing process
  - 13,500+ registered learners since Sep 16

► **Genomics 101 series**
  - Series of 8 short online modules aimed at health professionals with limited or no genomics knowledge.
  - Designed to take the learner from genes and proteins, to genomics in clinical practice.
  - Currently in development, launching late 2017

► **Genomics Game**
  - Board game for face-to-face interactive group learning activity
  - Developed primarily for nursing workforce
  - Aligned to current curricula
  - Currently in testing phase, and due for release in August 2017

► **Genomics film series**
  - Filmed interviews to raise awareness of genomics in key areas:
    - Cardiology, Pathology, Cardiology, Ophthalmology, Respiratory, Primary Care
    - Nursing, Midwifery, Bioinformatics
    - First films to launch by autumn 2017

Also… introductory online courses, factsheets, articles, videos, animations and infographics

@Genomicsedu  #genomes100k  www.genomicseducation.hee.nhs.uk
Networked approach to Workforce transformation

- Funded 13 NHS GMC training and education leads
- Engaged across full range of professional groups (eg nursing and midwifery, pharmacists, primary care)
- Launched NHS Faculty of Genomic Medicine: alumni and experts to act as genomic champions within wider workforce, with oversight from advisory group informing GEP strategy and policy
- Exploring patient pathways to identify NHS touchpoints (eg familial hypercholesterolaemia)
- Forming the HEE Training and Education Genomics England Clinical Interpretation Partnership to facilitate rapid knowledge transfer from 100,000 Genomes Project into training and education
Delivering genomic-based personalisation for patient benefit & service sustainability

Technology available and affordable

Patients/Public ready for change

Clinical Leadership & workforce upskilling

Proof of concept for routine care established

New service networks and patient pathways set up

Health economics established

System planning & commissioning aligned

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


GENOMIC MEDICINE FOR PERSONALISATION

Spectrum of delivery

At home

Specialist Care
Birth of the NHS Genomic Medicine Service

Mar 2017: NHS England Board agrees strategic approach for building a genomic medicine service from 2018/19, delivering personalisation of treatment and intervention building on the legacy of the 100,000 Genomes Project.

The key principles for the NHS genomic medicine service are:

- To ensure **comprehensive and equitable access** for the entire population
- 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.
- To **retain and build the political, ethical and moral trust** of the UK in genomic medicine
The NHS Genomic Medicine Service: Assembling building blocks & budget

NHS Genomic Medicine Service underpinning Personalised treatments and interventions

- Genomic Medicine Clinical Services: integrated clinical genetics & providing population-based care
- National Lab Network: 7 ‘new’ Genomic Lab Hubs
- National Testing Strategy: inc testing directory (single gene – WGS)
- Informatics architecture & data store
- Whole Genome Sequencing Provision
- Clinical Interpretation Pipeline
- Workforce development: upskilling of existing staff & ongoing professional engagement
- Industry/academic/international partnerships and projects: supporting ongoing research & development through clinical care

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

Procured: Q3/4 ’17/18
Mobilised: Q2 ’18 on
Mainstream arrangements: Q2 ‘19 on
Underpinned by comprehensive genomic testing strategy

A 360° view of current genetic testing

Definitive test list for rare disease & cancer

Evidence based framework for annual directory review

Baseline commissioning & money flows & funding & economic model

WGS & functional tests & replacement of existing tests

Governance & monitoring arrangements

Service specification & informatics & data sharing model

Developing genetic testing strategy

• Methodology for test selection, evaluation & decommissioning of tests
• Expert groups to assess and determine clinical utility across care spectrum
• Authoritative directory of genetic tests linked to clinical conditions – reviewed annually & to include NIPT
• Determine levels of testing – tackling unmet need & variation
• Standardising approach & use of high throughput technology
• Aligned with funding flows & reimbursement model

WGS + informatics genes/ panels/ exomes + biomarkers

Wet genes/ arrays/ panels/ exomes

TRANSFORMATION
Early candidates for WGS testing
Rare disease – first 28 conditions

Intellectual disability / mental health
- Intellectual disability incident cases

Cardiovascular
- Aortopathy
- Paediatric cardiomyopathy
- Primary Lymphoedema

Endocrinology
- Disorders of sex development
- Growth failure in early childhood

Haematology
- Rare anaemias and bone marrow failure disorders
- Bleeding and platelet disorders

Hearing
- Monogenic deafness

Immunology
- Childhood immunodeficiency
- Maternity and child health
- Acutely unwell infants with likely monogenic disease
- Monogenic fetal disorders with multiple possible genetic causes
- Congenital malformation and dysmorphic syndromes with multiple possible genetic causes

Metabolic
- Unexplained metabolic disease
- Neonatal diabetes

Neurology
- Hereditary ataxias, choreas and other movement disorders
- Young onset and familial neurodegenerative disorders
- Epileptic encephalopathy and syndromic epilepsy
- Paroxysmal disorders, pain disorders and sleep disorders
- Mitochondrial disorders
- Neuromuscular (including peripheral neuropathies)
- HSP, leukodystrophies and rare neurological disorders

Ophthalmology
- Inherited retinal disorders
- Bilateral congenital and childhood cataracts
- Other genetically heterogeneous rare ophthalmic disorders

Renal
- Steroid resistant nephrotic syndrome
- Atypical cystic kidney disease

Other
- Ultra-rare and atypical monogenic disorders

Cancer – planned in phases
- Sarcomas
- Paediatric tumours
- Haematological malignancies
Evolution of NHS Genomic Medicine Centres & clinical genetic services

NHS genomic medicine service will have a key role in driving change across the NHS:

- Ensuring **comprehensive coverage** and access across their geography, including all Hospitals, Specialist Providers and Primary Care
- Enabling **access to an approved genomic test directory** up to level of WGS and consolidate learning from 100,000 Genomes Project
- Delivering national **genomic medicine service consent** model
- **Integrating clinical genetics service** to provide specialist advice and expertise including in genomic counselling and **extending genomic MDTs**
- **Further mainstreaming** Genomics outside of clinical genetics and embedding within other clinical specialities to introduce genomic testing within a broader range of conditions and pathways
- Further **informatics and data developments** to underpin delivery and alignment with national/international R and D initiatives
- **Driving medicines optimisation**/ appropriate prescribing and personalisation of interventions
- Providing **evidence for health economic and value assessments**
The NHS framework for personalisation

**DNA + omics**

- Genomic medicine in specific examples
  - 2012
  - 2013-18

- 100,000 Genomes Project
  - use of WGS, panels & functional genomics for rare disease & cancer

**Technology, Innovation & Knowledge Base**

- Genomic medicine embedded within specific pathways
- Genomic medicine embedded as part of routine care – where appropriate

**Clinical Change & Operating Model**

- Clear role established for next-gen diagnostics

- Better prediction and prevention of disease
- A more precise diagnosis
- More targeted and personalised interventions
- A more participatory role for patients

**Policy, System & Regulatory Alignment**

- Data analytics and bioinformatics
- Other functional diagnostics
- Phenotypic characterisation
- Patient generated data & self-reporting

**Infrastructure change**

- Informatics & commissioning


**2018 - 2020**

**2020 and beyond**
A whole pathway approach to developing personalisation across care continuum

Evaluation of candidate conditions assesses:
- Potential for **high impact** of next-generation diagnostics including functional genomics and for **medicines optimisation**
- Evidence base for **utility/cost effectiveness** and health economics
- **Responsiveness & turnaround time** for results
- Evolution of **Informatics and analytical platforms** including decision support
- Whole pathway approach
- Adoption of new technologies

**NHS Clinical priorities**
- Diabetes
- Mental health

**Opportunities for population health approaches**
- Familial Hypercholesterlemia

**Medicines optimisation & reduction of ADR**
- Warfarin
- Respiratory

**Where significant utility from next-gen diagnostic approaches**
- Rare Diseases, Cancer

**Where significant variation in access**
- Renal
- Cardiovascular

Driving new pathways & services now: WM Familial Hypercholesterolemia Service

- Previous service lacks co-ordination;
- Care fragmented, no agreed pathway, standards or protocols or systematic cascade testing
- 22 West Midlands CCGs have committed to commissioning the regional service including funding of nursing posts and genetic testing
- Embedded in WM GMC

• If mutation –ve treat in accordance with NICE guidelines (CVD Risk Assessment and Reduction)
• MECC, lifestyle advice and refer to relevant lifestyle services
• Offer referral to 100,000 genome project
Stratifying conditions: identification and management of genetic diabetes

Maturity Onset Diabetes of the Young (MODY) is a specialised condition that is often misdiagnosed as Type 1, but responds badly to conventional treatment. A combination of phenotypical characterisation (including established diagnostics) with genomic testing identifies precise diagnosis and appropriate treatment options.

DIAGNOSIS

- EIF2AK3 p.E371*: Wolcott Rallison Syndrome
- FOXP3 c.227delT: IPEX syndrome
- GATA6 c.1448-1455del: Syndromic pancreatic agenesis
- STAT3 p.T716M: Multi-organ autoimmune disease

TREATMENT

- Sulphonylurea therapy
- Liver Transplant
- Bone Marrow Transplant
- Insulin and exocrine supplements
- ? STAT3 inhibitor

Basic details (eg BMI) & simple pathology (eg HbA1c)

Probability calculation

Urinary C-peptide creatinine ratio testing/ Antibody testing for pancreatic autoimmunity *(rules out Type 1)*

Genetic testing
Transforming surgical decision making: colorectal cancer

Genomic medicine will revolutionise diagnosis & improve surgical decision making

- WGS can identify likely patient response - informing decisions on surgical approach
- A mutation profile indicating patient likely to have many micro metastases then they could have targeted neoadjuvant chemotherapy before surgery.
- Developing new drugs against metastases might make more primary tumours operable.

Integrated analysis of gene expression profiles suggests 4 consensus molecular subtypes in Colorectal Cancer

Careful stratification could justify an organ preserving local approach with much less morbidity & comparable oncological outcomes

Dienstmann R et al. ASCO Meeting Abstracts. 2014
Multilevel approach to personalisation in respiratory conditions

- The multi-faceted nature of respiratory conditions are increasingly make them ideal candidates for a personalised approach at many levels,
- Rather than assign broad diagnostic labels (‘asthma’, ‘COPD’) across the continuum of patient presentation, precise diagnosis will allow the identification of specific ‘treatable traits’ (*pulmonary, extrapulmonary & behaviour/lifestyle*) allowing more personalised care & better outcomes.

Network model of condition drivers

Personalised approach to airway disease

- Adult with symptoms, signs or events suggestive of airway disease
- History, clinical examination and risk factors
- Spriometry/Fdx/blood eosinophils

Strong history and/or risk factors present and/or abnormal test results
- High probability of airway disease
- Assess and manage treatable traits (tables 1-3)

Atypical history, no risk factors and normal test results
- Low probability of airway disease
- Assess and manage nonairway treatable traits (tables 2 and 3), consider alternative diagnosis and follow-up
Genomics and Medicines optimisation

Precise diagnosis leads to precise treatment selection and fewer non-responders

Patient response to a particular pharmaceutical can be predicted by genomic & phenotypic characterisation

£16 bn pa spend on drugs and rising

NHS England working on plans for mainstreaming genomic testing & pharmaco genomic profiling linked to sharing of patient record

Genomic screening can identify potential adverse drug reactions
Understanding health economics to deliver value

As the diagnostic potential of genomics becomes clearer and personalised treatment options a reality, fresh consideration needs to be made of the health economics:

- New elements of value come into play
- A more dynamic approach to value needs to be made – with shares for diagnostics/drugs/payers/patients
- Standards of evidence must change – broader picture of clinical utility

Adapted from: Can and Should Value-based Pricing be applied to Molecular Diagnostics, Office of Health Economics Apr 2012
Opportunities to support future care & innovation through personalisation

- Characterisation & segmentation of disease – targets discovery efforts, builds in disease markers
- More translational research & more candidate products (though for smaller patient groups)
- Mining data for findings that boost understanding of real world response
- Better tests for clinicians to check patients for suitability. Patients will have more confidence in fit of medicine. Patient benefits for adherence, support & monitoring
- Better selection of clinical trial subjects – quicker & more compelling results for safety & VfM

PERSONALISATION
Supporting the Life Sciences Industrial Strategy

- Government has launched the Life Sciences Industrial Strategy, recognising the huge contribution of the sector to UK plc.
- Genomics makes a significant contribution to the Strategy, particularly through its role within the new Health Advanced Research Programme (HARP) which will include:
  - Increasing use of genomic medicine in NHS and aligning this with industry collaboration
  - Increasing genomic testing and screening
  - Developing diagnostics for early asymptomatic disease
- The strategy also sets out the need for a step change in number of clinical trials, recruitment levels and novel methodology.
Effective engagement across the health system & its population

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
The genomic medicine journey over the next decade

**Today:**
- ‘One-size-fits-all’ treatment based on symptoms
- Services and professions organised according to organ/speciality
- Limited use of genomic and molecular markers
- Diagnostic and clinical data not linked

**By 2020:**
- Whole genome sequencing for specific conditions
- Improved diagnosis of rare conditions and better understanding of cancer
- Comprehensive, linked diagnostic data coupled with effective informatics analysis to give a full picture of patients

**By 2025:**
- New taxonomy of medicine based on underlying cause and personal response
- Integrated clinical services taking a ‘whole body’ approach
- Tailored, optimised and more effective therapies for better outcomes
- New NHS relationships with academia, industry, patients & patient groups