Sharing Resources:
Panel discussion on moving from research into clinical care

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The world is changing

Percentage of whole genomes and exomes that are funded by healthcare systems

- 2012: ~1%
- 2018: ~20%
- 2022: >80%

Areas of clinical uptake: infectious disease, cancer, rare disease, common/chronic
MRI – ($400-$4,000) 100 million scans - pa

PET/SPECT ($1000-$6,000) 50 million scans - pa
If we can enable secondary use of clinical genomic data for research we will have a >60 million virtual cohort by 2025.

Global genomic data sharing can lead to...

- Demonstrated patterns in health and disease
- Increased statistical significance of analyses
- Matching other / similar patients, leading to increased diagnoses
- ‘Stronger’ variant interpretations
- More informed clinical decisions
150+ Genomic Data Initiatives Globally

CLINICAL/GENOMIC MEDICINE

RESEARCH

NATIONAL

COHORTS

40 initiatives
70 initiatives
21 from 15 countries
64 globally
HTA – Government decision making

- Geisinger/Regeneron – fund exomes and evaluate in practice
- Kaiser Permanente – very diligent HTA pre introduction
- Canadian clinical genomes for RD diagnosis or large panels for cancer
  - Several provinces - re-patriate “out-of-country tests” optics vs budget control; secondary use for research versus rather than pdf report; build the competency and capacity in hospital labs versus cost, speed, clinical certification; over capacity in research sequencing but lab not certified for clinical use
  - Other provinces no experience with clinical NGS- only research experience; ministries of health need clear, well articulated case with HTA that they will regard as valid and the competency/capacity – Pop scale 8.5 m vs ~1m or <
  - Work force, wait list, diagnostic odyssey, early identification, role of AI
Five times the diagnosis, one quarter the cost

Prospective comparison of diagnosis
- 80 children <2 years of age
- Features of known mendelian conditions
- 58% diagnosis vs 11% in standard care

Cost effectiveness study of first 40 children
- Cost per diagnosis $5047 vs $27050 std care
- Incremental saving of $2,181 per additional dx
- Bootstrapped & sensitivity analysis

Follow up cost utility study
- Cost saving of AU$1,578 per QALY gained
- No increase in hospital service use.

Evaluation for effective education

Genetic specialists: Current practice and workforce

Other medical specialists: needs, practice, preference

Tools to assist genomic education design and evaluation

Research Topic
Educating Health Professionals in Genomic Medicine: Evidence-Based Strategies and Approaches
Program logic + evaluation framework for genomic education

Goals ≈ Long-term outcomes

Stakeholder engagement

Planning

**Situation Analysis**
Consider:
- Needs assessment
- Project parameters
- mandates/priorities
Define genomic workforce
Define desired ‘level’ of genomic literacy, e.g., competencies

**Opportunity Analysis**
Existing/repurpose resources
Possible partners

**Evaluation plan**

**Checkpoints**
- Approvals in place
- Resources in place

**Deliverable:**
Defined Education Intervention
- Goals/aims
- Target groups
- Learning objectives

**Development**

Consider:
- Theoretical framework
- Curriculum and learning design
- Assessment
- Project management
- Piloting or β-testing
- Promotion or dissemination (marketing)

**Checkpoints**
- Expert review
- Approvals in place
- Resources in place

**Deliverables**
- Educational intervention/s
- Assessment/s

**Delivery**

**Outcomes**

- Short
- Med.
- Long-term

Evaluate / document
Evaluate / document
Evaluate / document
Evaluate
100,000 Genomes Project: structured to build the approach for future care

Proof of concept through the 100,000 Genomes Project

4 principles
1. WGS extends current diagnostic scope
2. Recruitment from routine care, treated through routine channels
3. Participants consent to sharing of de-identified data for R&D & industry use & longitudinal access
4. Establishes model for transformational change

4 key legacies
1. Increased discovery of new pathogenic variants
2. Integrating advanced genomics into mainstream NHS
3. Increasing public understanding & support
4. Stimulating and advancing UK life sciences industry

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Principles for transformation:

- Building readiness
  - Building the evidence & learning for future approach & embedding
- ‘Whole NHS’ approach to delivery
- Participants recruited through routine care
- Integral alignment between clinical care & research
- Creating new pathways & protocols
- Clinical Leadership & workforce upskilling alongside service development
- Working across organisational boundaries & building new partnerships

Creating direction

Aligned with NHS transformational model:

Leading Transformation
## Making the case for genomics

### COST/BENEFIT
- Understanding cost of multiple sequential testing/ unwarranted variation & establishing activity
- Replacement of outmoded technologies – enhanced diagnostic yield above SoC
- Made clinical & economic case for both non-WGS & 500K WGS in mainstream care & mechanism for annual review & prioritisation
- Gained tripling of investment in genomics over next 5 years & centralisation of budgets

### QUALITY & OUTCOMES
- Improving care in key national clinical priority areas (Cancer/ Rare Disease/CVD/Acute Care)
- Supporting and linking with personalisation/ medicines optimisation & ADR reduction *(NHS drugs budget £17bn pa)*
- Established principle & buy in for single national approach, protocols, standards, datasets, data sharing, IG, metrics & scrutiny (quality dashboard) *inc National Genomics Testing Service & National Genomics Test Directory*

### DELIVERY & SERVICE MODEL
- Established WGS deliverability and requirements for whole infrastructure (inc non-WGS) with NHS informatics and data developments
- Service & human cost of diagnostic odyssey
- Reducing inequity and unmet clinical need
- Demonstrated value of new models of care and how existing services could be consolidated & networked
The new national genomic infrastructure

- Coordination, engagement & networks through contracted NHS Genomic Med Centres til 2023/4

- Engaged & informed patients
  - New patient choice & consent model – including sharing data for clinical care & research involvement

- Clinical genetics & other key services
  - Ordering & Genomic MDT clinicians

- National Testing Service for all genomics (single gene – WGS) delivered through 7 Genomic Lab Hubs

- National Genomic Test Directory (Cancer & RD) – specifies tests & approach with annual review

- Clinical Interpretation & decision support
- National Genomic Informatics System (NGIS)
- National WGS provision

- Clinical Data Store (WGS+)
- Research Environment
- Controlled access to inform research, discovery & ongoing NHS engagement

Underpinning ongoing NHS/NIHR clinical research initiatives with contractual requirements to deposit data

- Informed and shaped by contractual requirements for patient participation & societal engagement

- Supported integrated & coordinated workforce development – linked to HEE Genomics Education Programme
- Funded distributed clinical Leadership + AoMRC Clinical Leads

Elements delivered in partnership with Genomics England
Developing the health system workforce alongside service transformation

The Genomics Education Programme was established in 2014 to run alongside the developments in genomic medicine services to driving training and upskilling for the entire 1.3 million NHS workforce.

The GEP provides a wide range of free-to-access resources to provide an ‘anytime, anyplace, anywhere’ approach to education – tailored to suit the range of professional requirements, the extent and time available for learning & the immediate need for education.

Central to the programme is the multiprofessional Masters in Genomic Medicine (& associated CPD modules) and specialist commissioned training places (eg Bioinformatics & genomic counselling) + undergraduate & postgraduate training curricula.

Resources show huge reach
- 50,000+ total staff reached
- 1400+ Masters framework places
- 1.5million+ web page views
- 460,000+ resource views
- 34,440 course registrations
- 19,600+ MOOC registrations

Resources and material at genomicseducation.hee.nhs.uk and www.futurelearn.com
Public Engagement

Public engagement is **not** education

It is:
Making a connection, Building a bridge
No scientific jargon, but culturally acceptable metaphors

Different engagement for publics and patients

We already have 50+ GA4GH films on genomic data sharing, translated into 14 languages, plus 20 other films in English (incidental findings, genetic counselling terms etc)
Light touch, google search as a metaphor for sequencing

(Click on image to open video in YouTube)
Deeper connection, dominant inheritance, used within clinic

Prof Anna Middleton
Wellcome Genome Campus, Cambridge, UK
www.wgc.org.uk/ethics

(Click on image to open video in YouTube)