100,000 Genomes Project

Monday 8thth December 2014

Dr Tom Fowler

Director of Public Health

About the 100,000 Genomes Project

- 100,000 whole genome sequences of NHS patients with rare inherited disease, cancers and pathogens

- Project announced by the Prime Minister, David Cameron in December 2012
- An Olympic Legacy

- Genomics England announced by Secretary of State for Health during NHS 65th Anniversary Celebrations - July 2013

Rare Inherited Disease
Cancer
Pathogens

Genomics England Pilots

- Phase 1 - Sequencing and Annotation Competition – finished
- 4 providers 15 samples Testing Sequencing QA and annotation

- Phase 2a - 2000 Rare Disease WGS- 30x depth – over 2014
- Partnering NIHR BioResource and Translational Research Collaborative- 5 centres - 2000+ patients since November 2013.

- Phase 2b - 3000 Cancer Patients (Lung, Breast, Ovary, Prostate, Colon and Leukaemia) over 2014-15
- Somatic (60-80x) and germline (30-40x) – tendering now
- Optimise Molecular Pathology pipeline

- Pathogens will be with Public Health England

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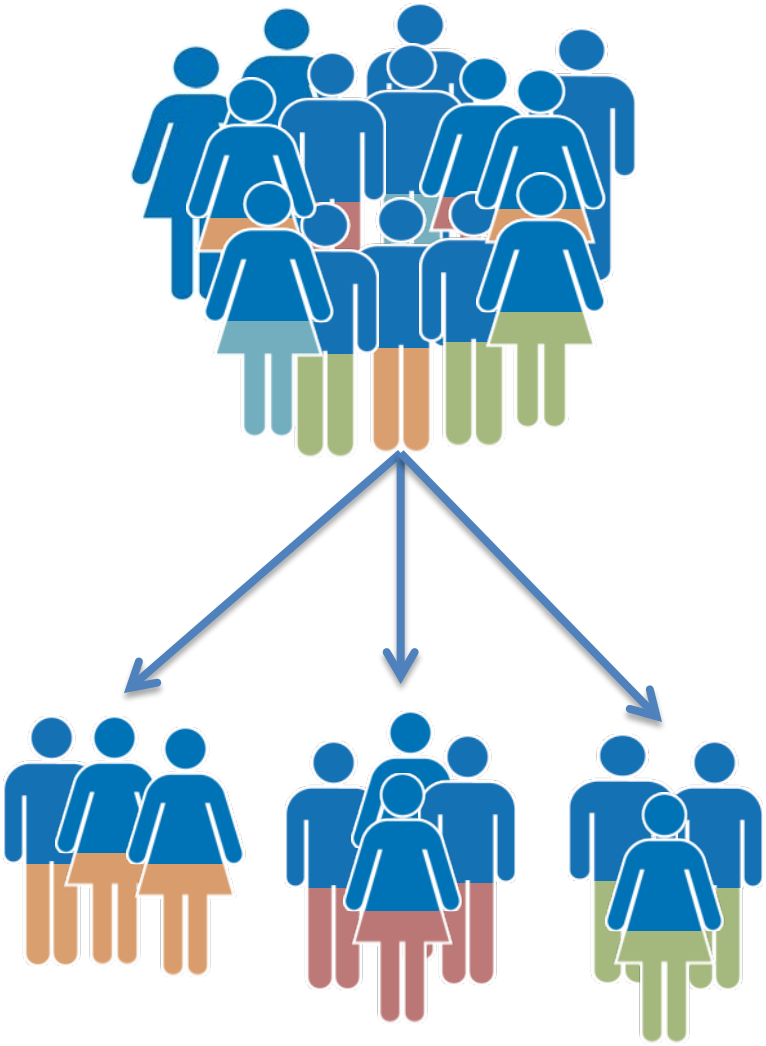
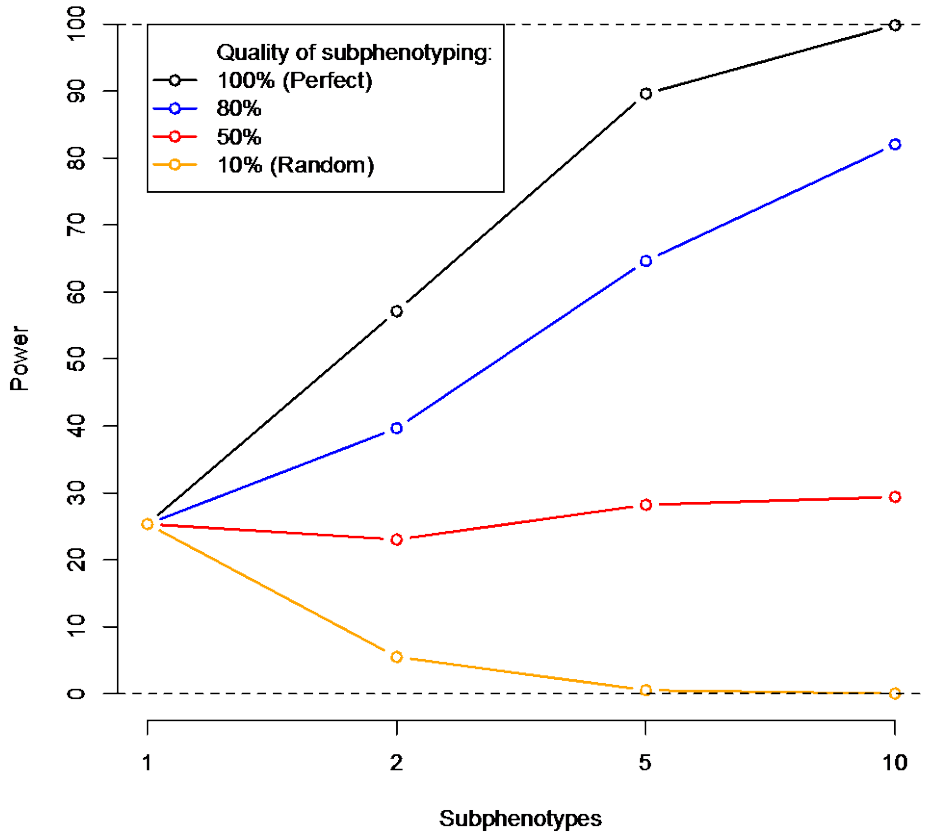
Phase 3 - 15k in 2015, 25k in 2016, 45k in 2017

The case for deep phenotypic data





Luke Jostins and Gil McVean

Genomic Medicine Centres

NHS ENGLAND

INVITATION TO TENDER

STAGE TWO ITT

NHS GENOMIC MEDICINE CENTRE SELECTION - WAVE 1

- Designated local NHS Lead and extended team
- Capacity and capability networks
- High fidelity phenotypes
- Access to data and samples
- February start date

http://www.england.nhs.uk/wp-content/uploads/2014/10/nhs-gmcs-stg2-itt-fin.pdf

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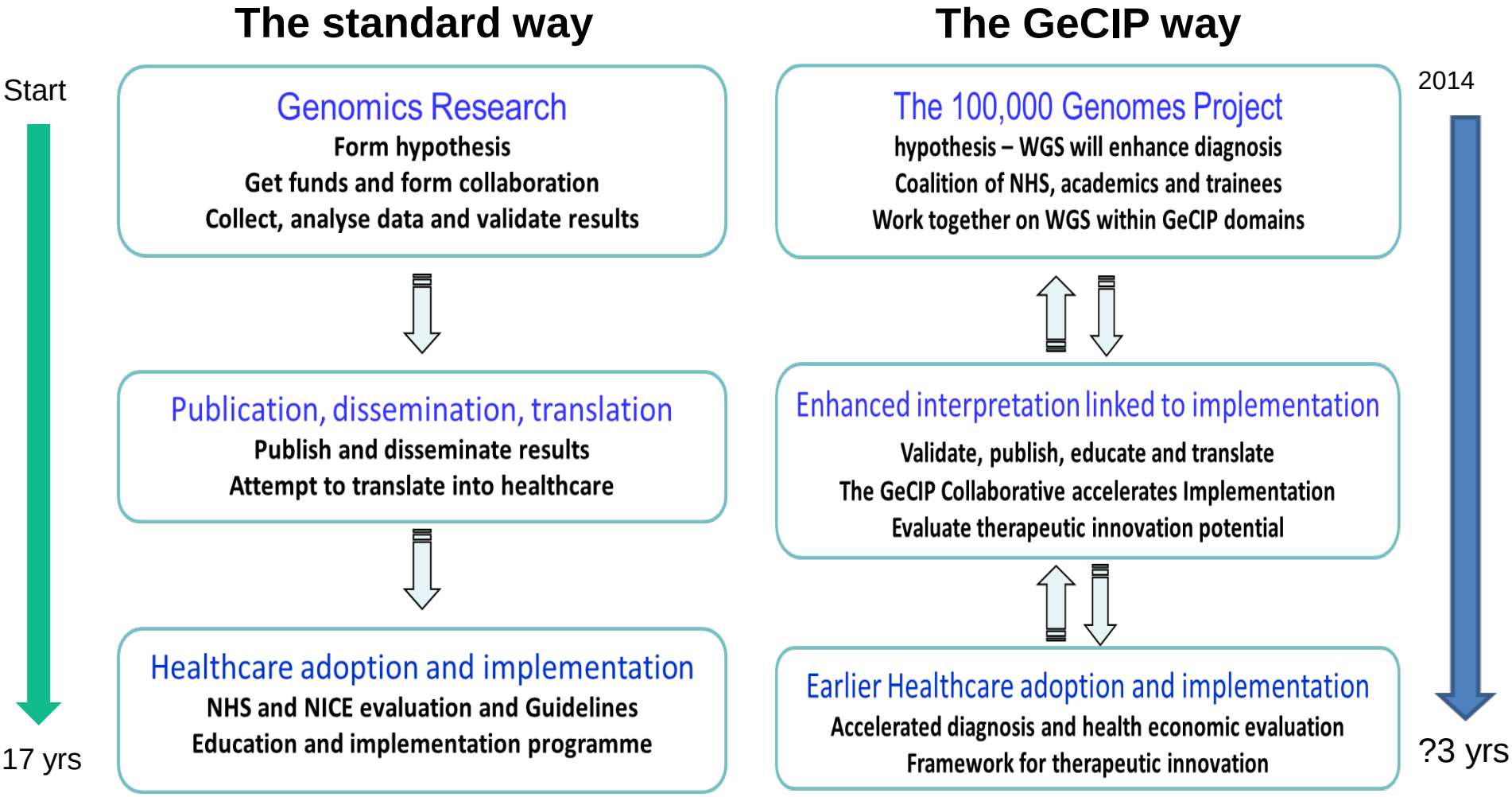
Clinical Interpretation Partnership

Why do we need a Clinical Interpretation Partnership?



Securing Patient Benefit

Genomics England Clinical Interpretation Partnership

- Drive up the fidelity of clinical interpretation of WGS
- UK –led self-organised into domains
- Partnership with researchers, the NHS and Trainees with skills.
- Can bring international collaborators
- Possible formation of a precompetitive consortium of a limited number of industry partners.
- All data generated contributes to the Genomics England Dataset and are available to all.
- Designed to accelerate academic/industry partnership and development of diagnostics and therapies.
- Recognises that to get to a therapy will require significant additional R&D which we aim to stimulate here in the UK.
- IP owned by Genomics England but freely licensed

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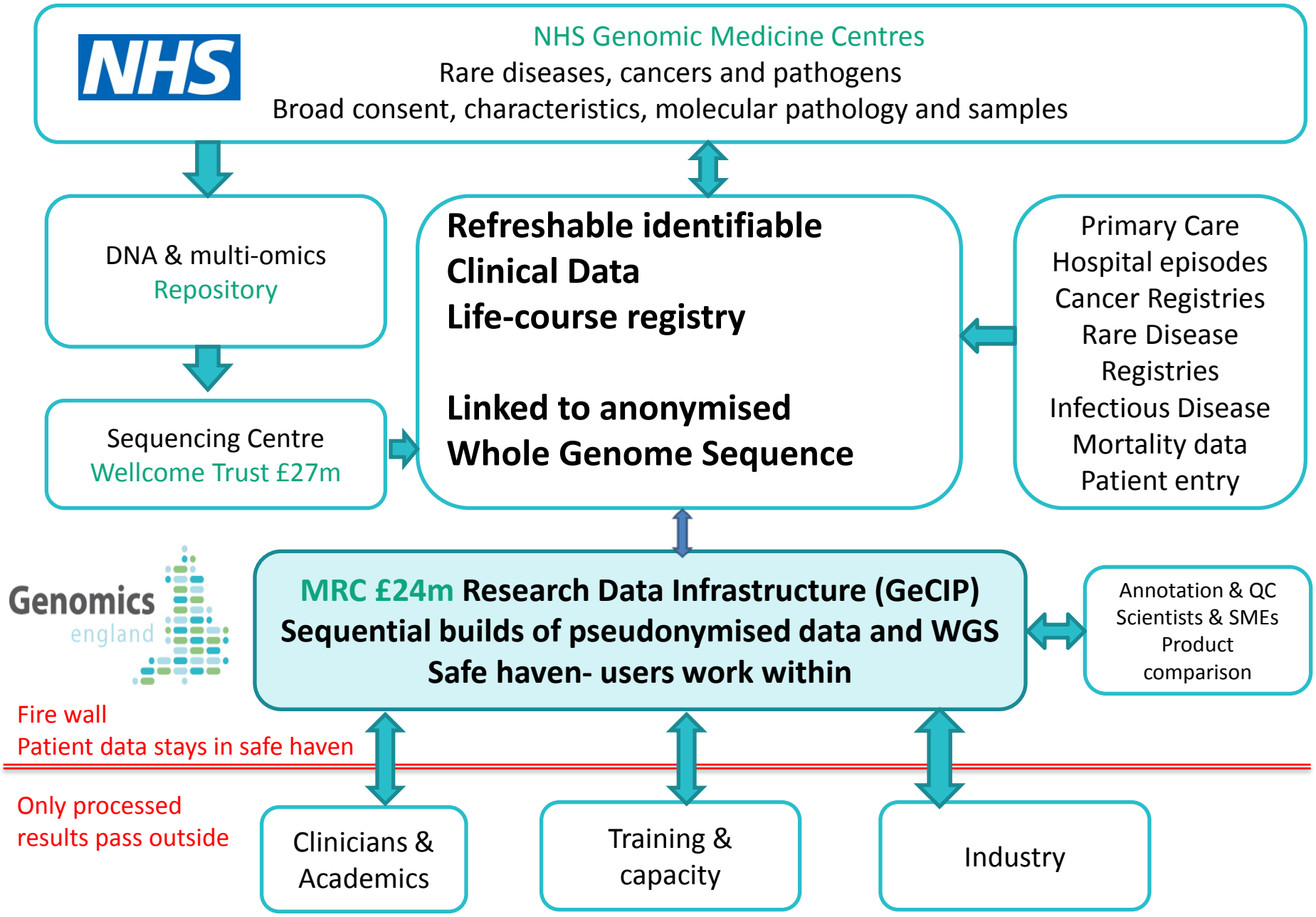
Genomics England Clinical Interpretation Partnership Rare Disease, Cancer and Infection Domains

Organised in Disease Domains	Key functions and outputs
UK led - steering group	Appoint domain leader & sub-groups
Genomics England Chief Scientist's Team	Oversight, informatics and logistics for the programme
Multiple phenotypic sub-groups	Deeper phenotyping & extend programme
Functional characterisation Multi-omics	Single cell or model functional studies RNA, epigenetics, proteomics
Analysts and Bio-informaticians	Novel analytic approaches
Interpretation - NHS and PHE teams and Researchers	Highest fidelity dynamic reporting system Integrated Multi-Disciplinary Team
Training - HEE/GECIP trainees	Genomic Medicine Academy
Precompetitive industry partners	Academic/Industry Collaboration

Genomics England – The main programme



Establishment Phase

- Illumina Partnership
- NHS Genome Sequencing Centre – £27m from Wellcome

- MRC Award of £24m to establish the UK Data Infrastructure for Genomic Medicine

Partnership of QMUL, UCL, The Farr, Oxford Big Data, UK Biobank, Sanger, EBI, Cambridge, Kings

PARTNERSHIP & INNOVATION WITHOUT REINVENTION

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Genomic Medicine

NHS
Health Education England

- Specialist genetics, pathology and specialist clinical workforce in Genomic Medicine
- Increase in specialised scientific training fellows funded over 3/5 years:
  - Molecular Pathology including Infections and Pathogens
  - Genetics / genomics
  - Bioinformatics
- Commission bioinformatics workshops in conjunction with partners
- Develop specialist on line on-line learning and an MSc in Genomic Medicine
- Develop an MSc in Genomic Medicine
- CPD access to MSc modules for specialist practitioners

Genomics England

- 100,000 WGS of NHS patients
- Working with NHS, academics and industry to drive Genomic Medicine into the NHS
- Support that with education
- Leave a legacy of NGS Centres, sample pipeline and biorepository, large-scale data store that makes this usable by the NHS
- New diagnostics and therapies and opportunities for patients
- By end of 2017

NHS England | Public Health England | NHS National Institute for Health Research | NHS Health Research Authority | Department of Health