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The 100,000 Genomes Project

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Seattle, October 2015

The 100,000 Genomes Project

Project announced by the Prime Minister in December 2012

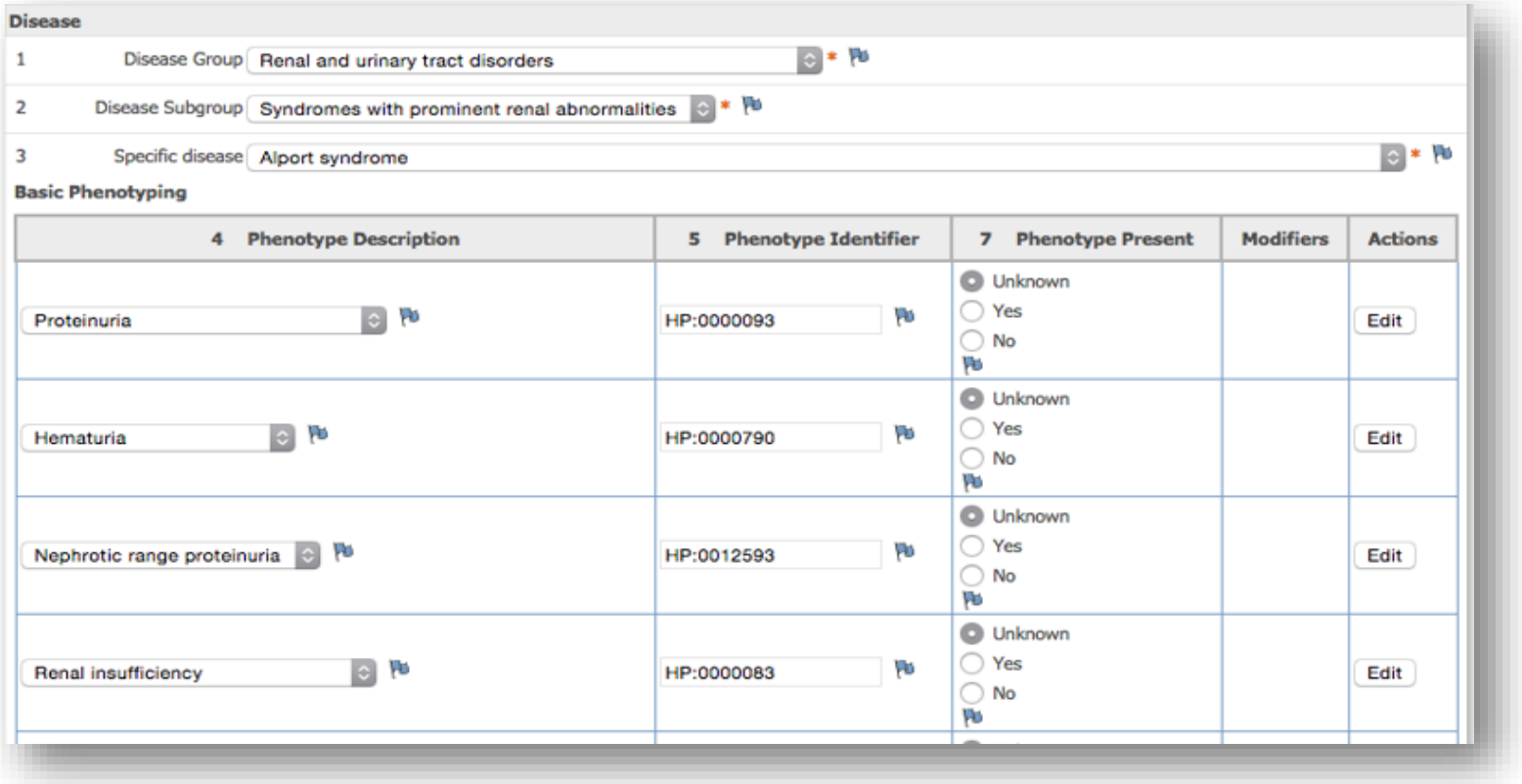
Genomics England announced by Secretary of State for Health in July 2013

Rare Disease

- incidence for each disease < 0.05%
- > 7000 diseases
- cumulative incidence in population ~5%
- cardiovascular, endocrine and metabolism, gastroenterology and hepatology, hearing and sight, immunology and haematology, inherited cancer predisposition, musculoskeletal, neurological, paediatric sepsis, paediatrics, renal, respiratory, skin
- 50,000 participants

Rare Disease

- 30x coverage whole genome sequences
- phenotyping using Human Phenotype Ontology



Disease

1 Disease Group: Renal and urinary tract disorders *

2 Disease Subgroup: Syndromes with prominent renal abnormalities *

3 Specific disease: Alport syndrome *

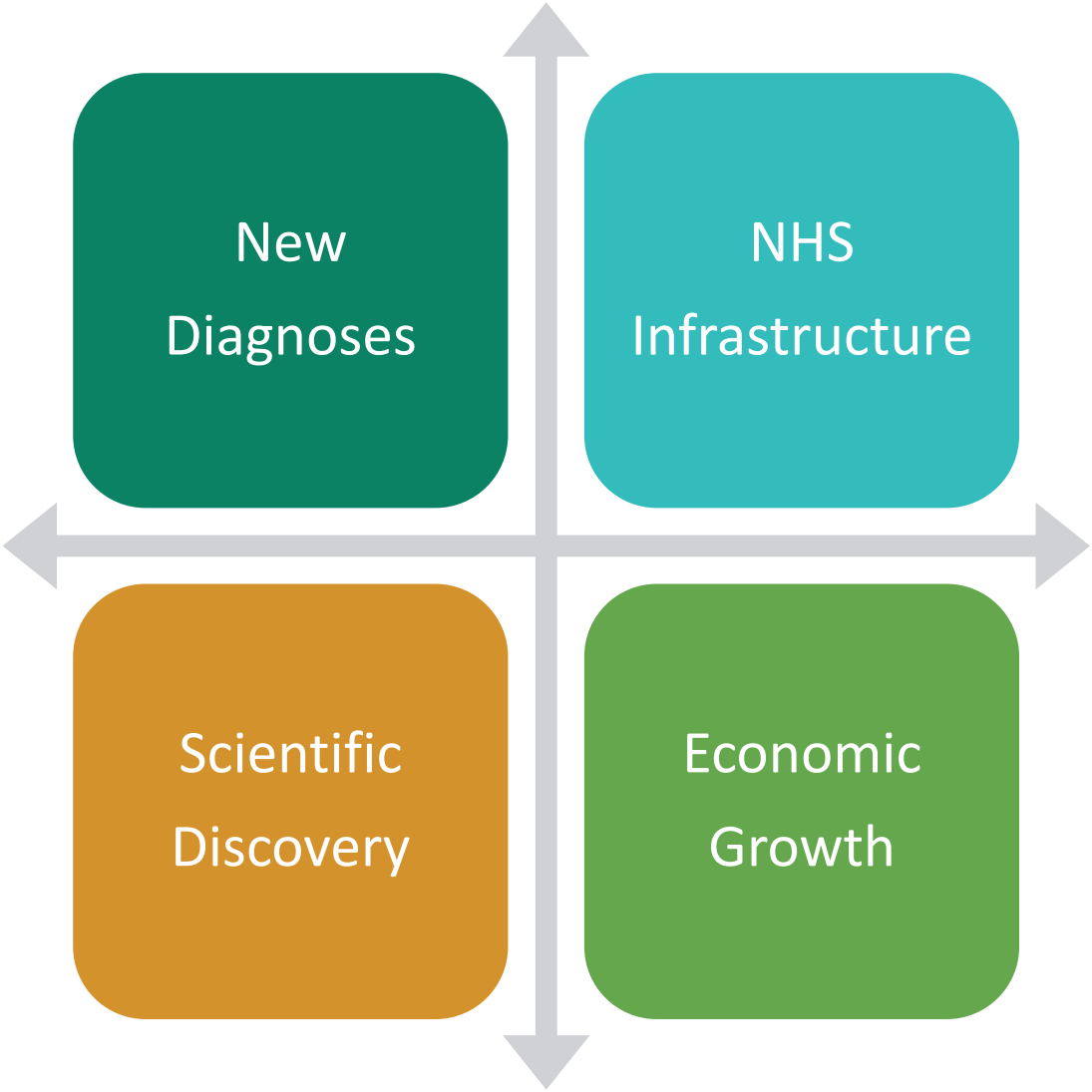
Basic Phenotyping

4 Phenotype Description	5 Phenotype Identifier	7 Phenotype Present	Modifiers	Actions
Proteinuria	HP:0000093	Unknown / Yes / No		Edit
Hematuria	HP:0000790	Unknown / Yes / No		Edit
Nephrotic range proteinuria	HP:0012593	Unknown / Yes / No		Edit
Renal insufficiency	HP:0000083	Unknown / Yes / No		Edit

Cancer

- ovarian, breast, colorectal, prostate, lung, leukaemia, sarcoma, renal, rare and childhood cancers, cancer of unknown primary
- 25,000 participants
- 75x coverage somatic, 30x coverage germline
- dataset reflects requirements for genomic medicine

Expectations



Participants

NHS England

NHS Genomic Medicine Centres

Consented participants, samples, phenotyping, clinical and laboratory data, clinical reports, confirmatory testing, and follow-up

Biorepository

biocentre uk
The centre for biomedical services

DNA & samples for multi-omics

Sequencing

illumina®

Data

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UNIVERSITY OF OXFORD

Identified Data

Collected against agreed models, linked to secondary data from other sources.

Other Data

Hospital Episode Statistics, registry data, mortality, *and more*

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Farr

Research Data

De-identified versions of the data made available via a virtual desktop infrastructure

Tools and Services

Quality assurance, clinical interpretation, tools for analysis, reference data

Researchers

Researchers

Genomics England Clinical Interpretation Partnerships, Industry Consortia, and other approved researchers

Infrastructure

- Genomic Medicine Centres (NHS England)
- NIHR national biosample centre
- Hinxton sequencing centre (Wellcome Trust)
- secure data centre (NIHR, MRC)

biosample centre

sequencing centre

data centre

Data Acquisition

- registration, consent, family history, phenotyping via web-based interface
- clinical and laboratory data submitted as XML messages, reports upon clinical events
- secondary data from clinical audits and national reporting datasets
- integrated, longitudinal record via LabKey server for review and quality assurance

- patient-reported data

Research Environment

- separate data centre
- a research ‘embassy’ for each group
- access to shared LabKey server
  - explore data within policy
  - take snapshots for analysis
- private cloud
- ‘airlock’ system

- virtual desktop infrastructure

Progress

- 11 Genomic Medicine Centres
- 5000 whole genome sequences
- 125 rare diseases
- 5 cancers
- 1 pathogen
- 4 tool providers
- 10 pharma companies

- 2500 researchers

Contributors (include)

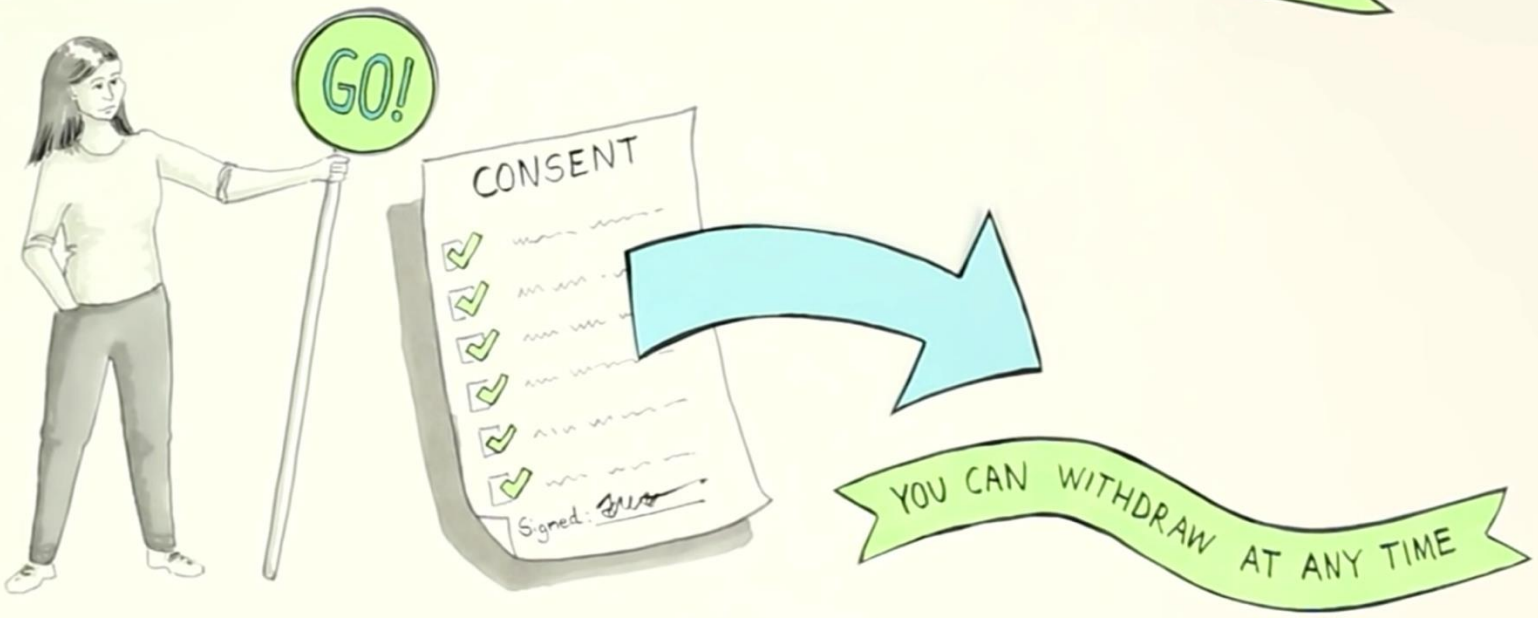
- NIHR Biomedical Research Centres
- NIHR Bioresource for Rare Diseases
- NIHR Health Informatics Collaborative
- NHS England, Public Health England
- Medical Research Council
- Cancer Research UK
- Wellcome Trust
- ...

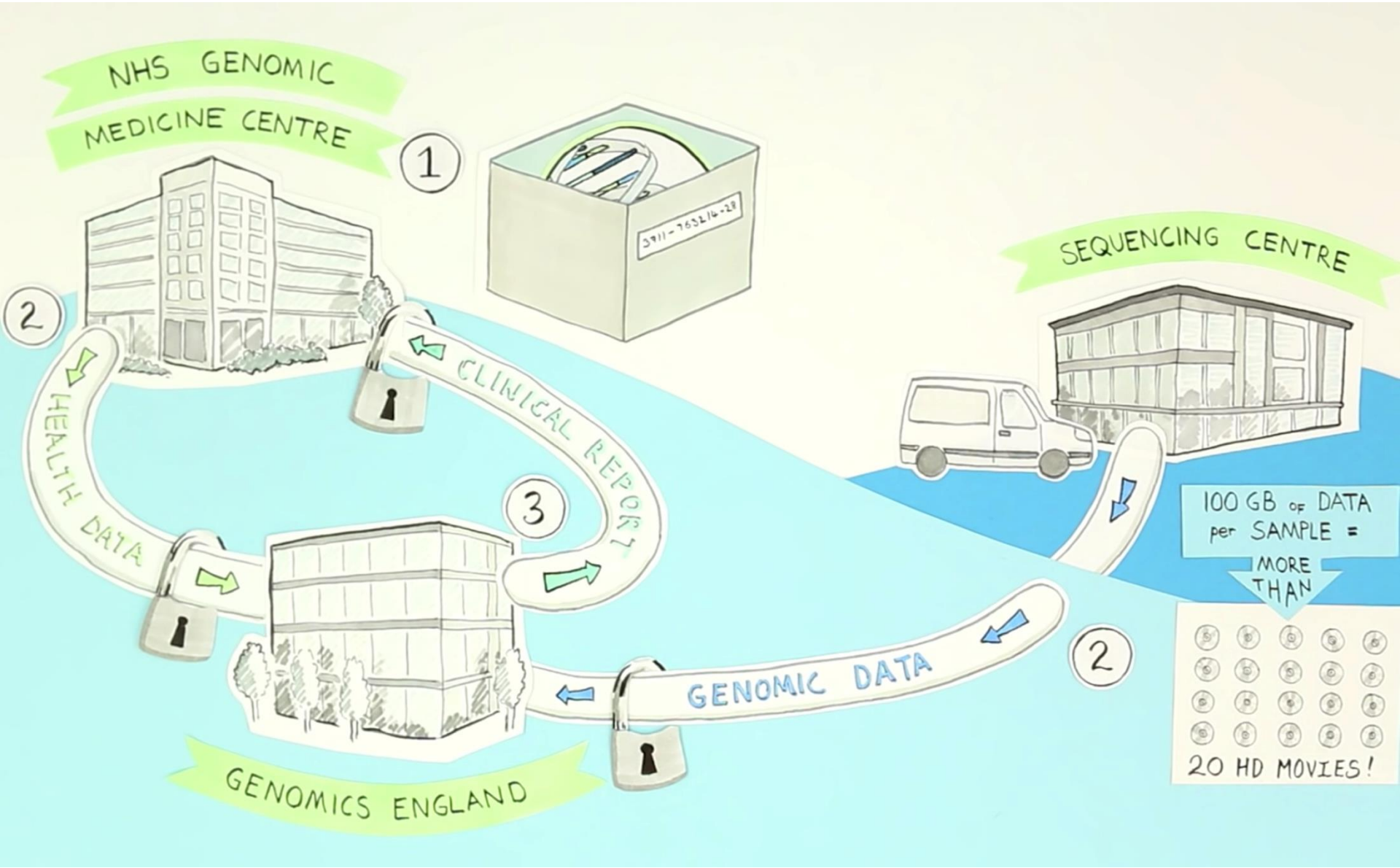
Challenges

- earn the trust of:
  - patients
  - clinicians
  - researchers

- help to transform:
  - clinical informatics
  - research informatics
  - culture

WHAT DOES GENOMICS ENGLAND DO WITH YOUR DATA ?




NHS GENOMIC MEDICINE CENTRE
1
2
HEALTH DATA
CLINICAL REPORT
3
GENOMICS ENGLAND
GENOMIC DATA
2
SEQUENCING CENTRE
100 GB OF DATA per SAMPLE =
MORE THAN
20 HD MOVIES!

3

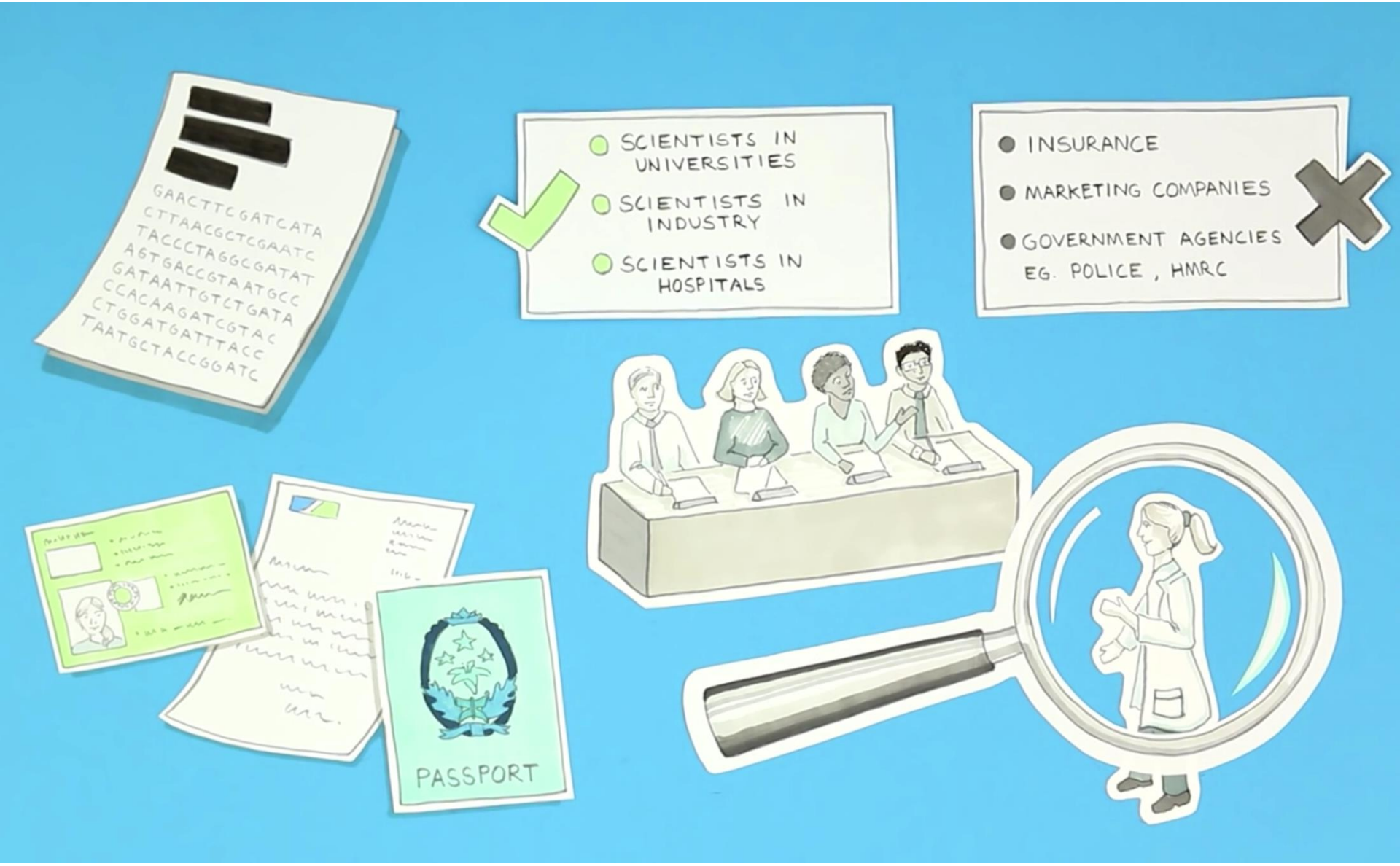
- YOUR HEALTH DATA
- YOUR MEDICAL RECORD DATA
- YOUR GENOME DATA

WHAT WILL SCIENTISTS DO WITH THE DATA ?

UNDERSTAND WHICH DIFFERENCES ARE IMPORTANT

BEST TREATMENT

- USE CURRENT MEDICINES BETTER
- NEW DRUGS
- NEW DIAGNOSTIC TESTS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SCIENTISTS IN UNIVERSITIES
SCIENTISTS IN INDUSTRY
SCIENTISTS IN HOSPITALS
INSURANCE
MARKETING COMPANIES
GOVERNMENT AGENCIES EG. POLICE , HMRC
PASSPORT

100,000 GENOMES PROJECT

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YOUR DATA MAKES A DIFFERENCE

Thanks

- The Genomics England team: in particular,
  - Sir John Chisholm (Executive Chair)
  - Professor Mark Caulfield (Chief Scientist)
  - Vivienne Parry (Communications and Outreach)
  - Ed Stafford (Director of Informatics)

- LabKey
  - enjoy the rest of the conference!