Genomic Counselling

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wellcome trust sanger institute

EMBL-EBI

BIODATA INNOVATION CENTRE

CONNECTING SCIENCE

Genomics england

CONGENICA
GENOME BASED MEDICINE

specific technologies

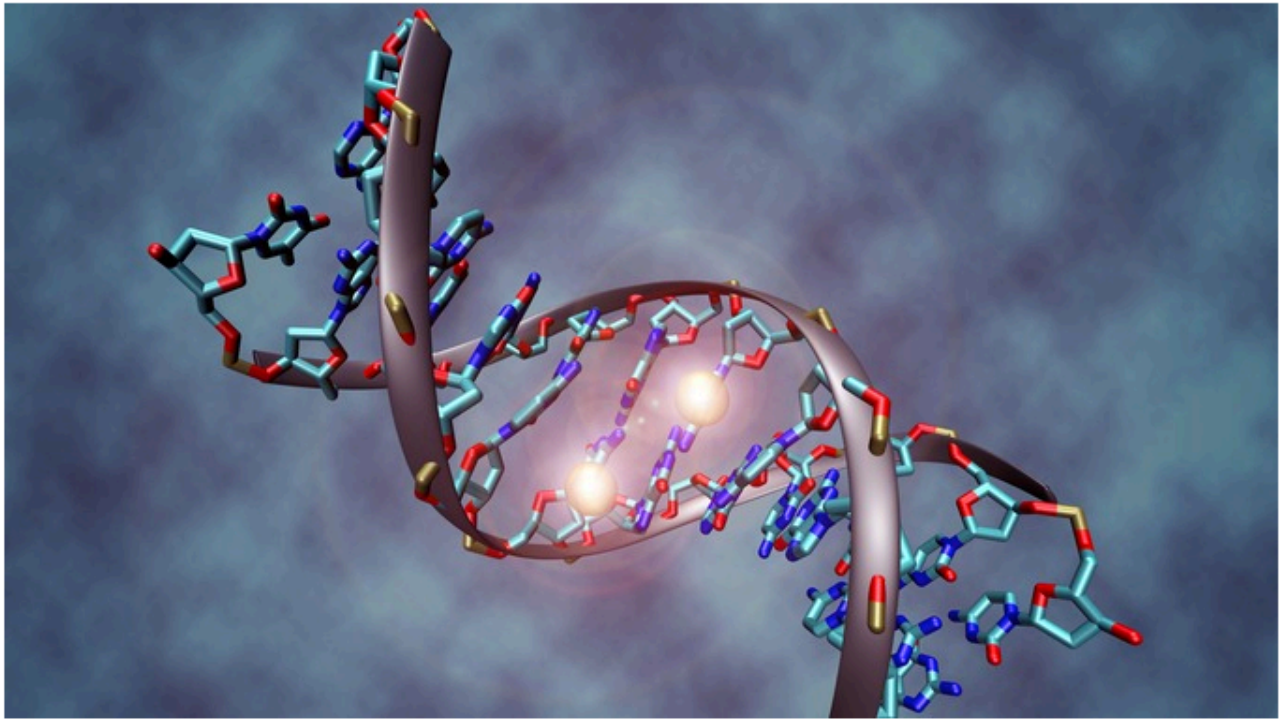
Global Gene Corp

- What is Genomic Counselling?
- New MSc Genomic Counselling
- Training in genomics + bioinformatics for experienced genetic counsellors
- Reality of sequencing in the NHS

22 December 2014 at 1:26am

NHS starts new era of DNA medicine

ALOK JHA SCIENCE CORRESPONDENT



What is Genomic Counselling??

Molecular Genetics & Genomic Medicine *Explore this journal >*

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Invited Commentary

Genetic counselors and Genomic Counseling in the United Kingdom

Anna Middleton, Georgina Hall, Christine Patch

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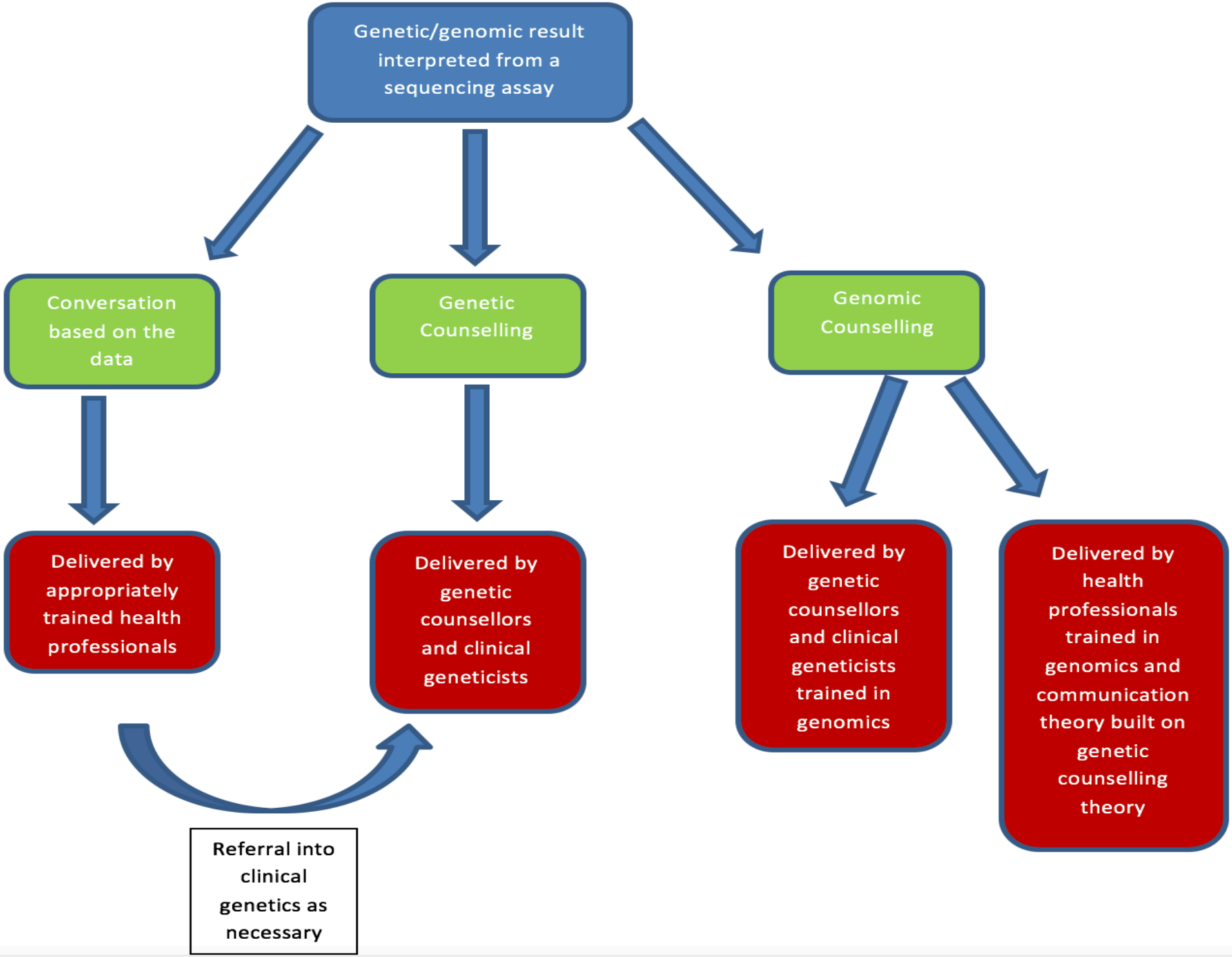
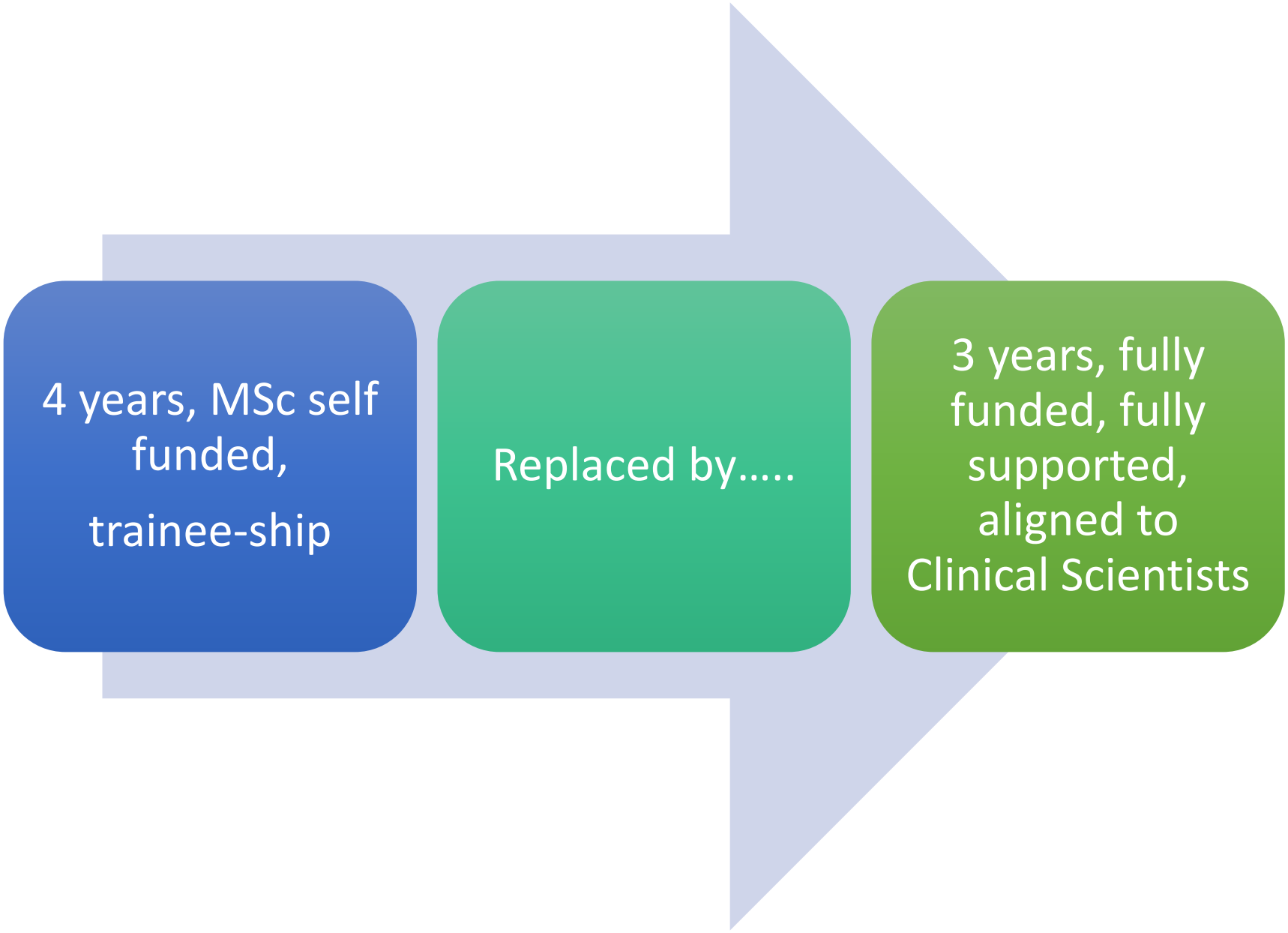

Genetic/genomic result interpreted from a sequencing assay
Conversation based on the data
Genetic Counselling
Genomic Counselling
Delivered by appropriately trained health professionals
Delivered by genetic counsellors and clinical geneticists
Delivered by genetic counsellors and clinical geneticists trained in genomics
Delivered by health professionals trained in genomics and communication theory built on genetic counselling theory
Referral into clinical genetics as necessary

Training new genetic counsellors

MSc Genomic Counselling

Part of the Clinical Scientist pathway


4 years, MSc self funded,
trainee-ship
Replaced by…..
3 years, fully funded, fully supported, aligned to Clinical Scientists

3 year integrated MSc Genomic Counselling

- Integrated 3 year training programme
  - Academic study (Manchester)
  - Work-Based training (regional Clinical Genetics services)
  - Keep working through university holidays
- NHS commissioned (i.e. increase in numbers when workforce demands this)
- Paid positions (starting on £26,300)
- Access to the same genomics and bioinformatics that the Clinical Scientists do

Core Content

- Counselling skills, advanced counselling skills
- Role-playing, video recording, same as current MSc
- Genomics
- Bioinformatics (variant interpretation)
- ElSi issues
- Clinical Genetics

- (no evolutionary genetics, no fruit fly genetics etc)

Training experienced GCs

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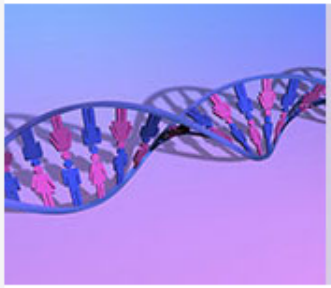
HOME EVENTS ABOUT US SPONSORSHIP CONTACT



Genomic Practice for Genetic Counsellors

3-4 February 2016
Wellcome Genome Campus, Hinxton, Cambridge, UK

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Genomic Practice for Genetic Counsellors
3-4 February 2016
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WELLCOME TRUST ADVANCED COURSES AND SCIENTIFIC CONFERENCES

Genomic Counselling for Genetic Counsellors
2-3 July 2015
Wellcome Trust Genome Campus, Hinxton, UK

Training non-specialist staff


100,000 GENOMES PROJECT
Preparing for the consent conversation

Role playing genetic counselling

Masters in
Genomic Medicine

Jeffrey Arun Rubasingham

Core Medical Trainee, Oxford Deanery

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UNIVERSITY OF CAMBRIDGE
Institute of Continuing Education

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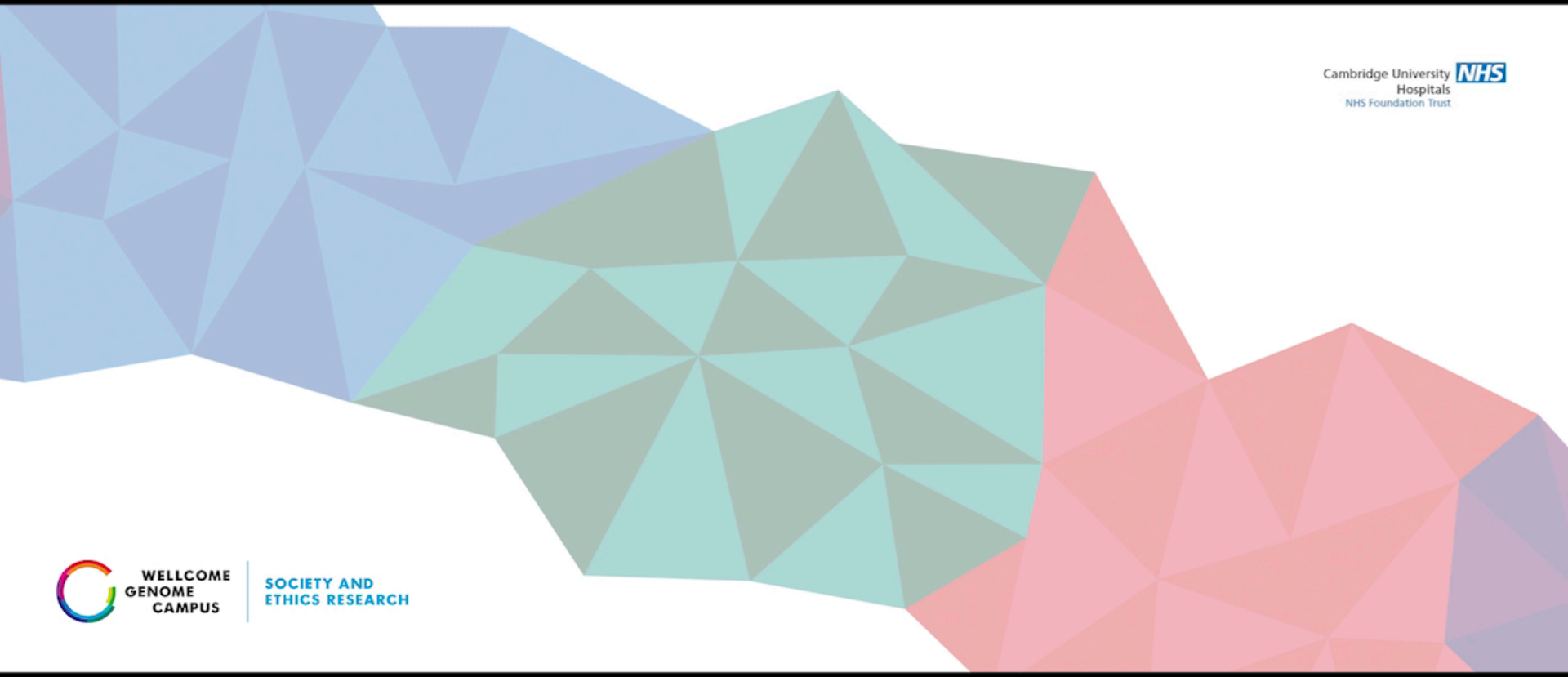
Cambridge University Hospitals
NHS Foundation Trust
WELLCOME GENOME CAMPUS
SOCIETY AND ETHICS RESEARCH

The reality of sequencing in the UK

- Sequencing is now ‘mainstreamed’

- Re-configuration of diagnostic lab services
  - Currently evolving
  - Different types of panel testing for the same condition
  - Different interpretation of the same variant
  - Plans for genomic data sharing

- 100,000 Genomes Project is changing practice

Broad Consent

Broad Consent

- The consent conversation is more generic
- [Comes from the biobank world]
- Shift in focus from pre-test to post-test, discuss results at the point when you know what is relevant and how
- Sign-posts to more detailed information if needed

What are we expecting patients to consent to?

- Test for the condition of interest
- Donation of their data for research
  - No say on the type of research
  - Non-profit
  - For-profit (commercial companies)
- Large element of 'trust' that the data will be secure, results appropriately handled etc
- Additional Looked For Findings

'Additional Looked For Findings'

[incidental findings, secondary findings]

Additional Looked for Findings

- For 100kGP the list may change and the patient needs to consent to an uncertain list

- Only particular variants looked for in list (i.e. negative result doesn't rule out other variants)

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The Original 100kGP List

Bowel cancer predisposition:

- *MLH1* (adult only), *MSH2* (adult only), *MSH6* (adult only), *APC* (adult and child) , *MUTYH* (adult only)

Breast and ovarian cancer predisposition:

- *BRCA1* (adult only), *BRCA2* (adult only)

Other cancer predisposition:

- *VHL* (adult and child), *MEN1* (adult and child), *RET* (adult and child)

Familial hypercholesterolaemia:

- *LDLR* (adult and child) , *APOB* (adult and child), *PCSK9* (adult and child)

Autosomal recessive carrier status:

- *CFTR* (Cystic fibrosis)

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“Additional Looked for Findings”

“It is not possible, at the point of taking consent, to confirm what conditions might be added to the list. What you can say is that they will be serious, clinically actionable conditions that they may want to be tested for. Participants can only ‘opt in’ to the entire existing list of additional conditions PLUS the future unknown list; they cannot select particular ones” (HEE website)

Pros of returning additional looked for findings

- ‘Potential’ to predict future disease (but patient is unselected for these conditions)
- ‘Possible action’ can be taken to screen for disease

Cons of returning additional looked for findings

- Difficulties in interpretation when patient is unselected for condition
- Long term outcomes unknown (i.e. does the opportunistic screen prevent/reduce mortality from future disease?)
- Potential to cause psychological harm?

Genomics England

"It may help to explain the degree of uncertainty which surrounds the clinical utility of these [*additional looked for findings*] at the current time: the research effort to help us understand and interpret these findings will be ongoing throughout the project, and we will not know for certain what risks patients carry for some time."

The Reality of Broad Consent

- It may feel uncomfortable to health professionals as there is more uncertainty pre-test and less detail
- There is evidence that this is an acceptable approach in the biobank world (Garrison et al 2015)
- Empirical data needed from patient perspective in a clinical setting

Professor Michael Parker
Non-executive Director; Chair of the ethics committee

Broad consent is acceptable and compatible with autonomy

What is Genomic Counselling??

Summary

- Genetic counselling, but working with genomic data
- Shift to broad consent (which changes the dynamic to a post test conversation)
- Counselling to manage uncertainty more?
- Training linked more with clinical scientists
- Do we claim the title?