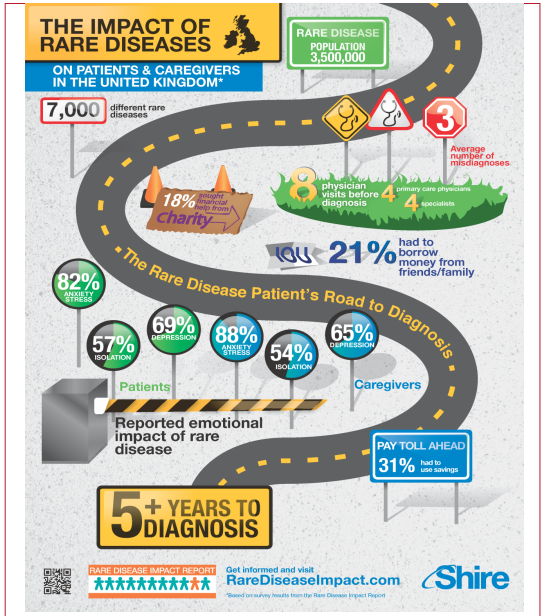
The 100,000 Genomes Project

G R. Wallace

[1]Academic Unit of Ophthalmology and [2]Centre for Endocrine, Diabetes and Metabolism,
Institute of Biomedical Research, College of Medical and Dental Sciences, University of Birmingham, Wolfson Drive, Edgbaston, Birmingham B15 2TT, UK



GMC Patient Identification & Consent Process Map for Rare Disease Patients

Identification
- Patient identified as eligible (via clinic list/disease register)
- CNS or Cons provides 1 page brief as 'takeaway' &/or DVD
- Or send with appointment letter

Face 2 Face recruitment
- Usually at routine OPD visit, could be separate apt at patient request.
- REC approved information sheet (1 of 6 variants)
- +/- relevant REC approved consent
- +/- blood samples
- Register on EST
- Family member consent samples at same time or in separate apt

Withdrawal
- Patient can withdraw at any stage from any further inclusion in the study
- +/- destruction of existing samples/data

Phenotype data
- Entered up to 12 weeks later
- Data set to be confirmed

Possible Findings

Results likely to be AT LEAST 6 months initially (later 2 weeks)
- Related to Clinical Diagnosis (validated in NHS lab)

Cancer:
- Molecular changes in tumour (not present in blood) (somatic changes)

Rare diseases:
- Normal
- Known pathogenic or likely pathogenic (via GeCIPs)

Unrelated to clinical diagnosis

Additional 'looked for' findings
- Affecting individual (cancer predispositions and Familial hypercholesterolemia)
- Carrier status (Recessive and X-linked conditions)

Opt in

Developing list- may include further clinically useful findings as evidence develops

Incidental findings not usually fed back

The 100,000 Genomes Project

The project will sequence 100,000 genomes from around 70,000 people. Participants are NHS patients with a rare disease, plus their families, and patients with cancer.

The aim is to create a new genomic medicine service for the NHS – transforming the way people are cared for. Patients may be offered a diagnosis where there wasn't one before. In time, there is the potential of new and more effective treatments.

Genomics England, a company wholly owned and funded by the Department of Health, was set up to deliver this flagship project which will sequence 100,000 whole genomes from NHS patients by 2017. Its four main aims are; to create an ethical and transparent programme based on consent; to bring benefit to patients and set up a genomic medicine service for the NHS; to enable new scientific discovery and medical insights; and to kick start the development of a UK genomics industry.

What are patients consenting to?

Whole genome sequencing of proband and parents or two relatives.

Storage of samples for other 'omic' analysis

Feedback of pertinent, clinically relevant findings:

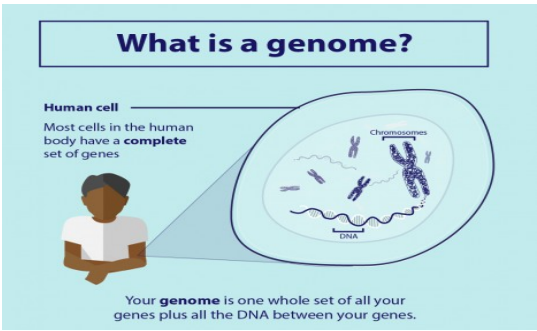
Verified locally
- Known pathogenic, causative results
- Possible causative results (via GeCIPs)
- Looked-for additional clinically important, actionable findings (opt-in)

Linked access to participants health records in perpetuity

Anonymised data and samples shared with approved partners in genomic embassy

Agreement to re-contact to invite to participate in further studies (up to 4 times pa)

Patient DVD



Relevance to Birdshot Uveitis

BU is not a selected disease on the 100,000 GP

We can nominate BU for inclusion

ww.genomicsengland.co.uk