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Project lead

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Realising genomics in clinical practice

Whole genome and whole exome sequencing (WGS/WES) technologies promise to revolutionise health care. However these disruptive technologies are likely to result in significant changes in the process of diagnosis and treatment and in clinician: patient interaction.

In this new project we aim to identify some of the ethical, legal and social issues these technologies present. We will consider the implications for clinicians, the impact upon patients and on health services, policy makers and regulators. We will use our findings to make recommendations for tackling some of the challenges of implementing genomic technologies in clinical care in the NHS.

Our aims and objectives

In our 2011 report, *Next Steps in the Sequence*, we analysed the implications of WGS for the NHS. Some of our recommendations have been implemented such as to minimise the generation of unexpected findings. However, important questions remain, including:

- How should we seek and take consent from patients for genomic testing?
- How should we categorise unexpected findings and when should they be reported to the patient?
- Does the distinction between clinical and research ethics and practice make sense in the genomics era?
- Will genetic testing using WGS require extensive redesign of patient pathways?
- How will clinicians and patients assess what is relevant and manage the volume and complexity of the information generated by these genomic technologies?

National initiatives, such as the UK's 100,000 Genomes Project, raise similar issues and reinforce the need for this work.

To what extent will new genomic technologies revolutionise health care? What will they mean for health professionals, patients and policy makers?

In this new project the PHG Foundation aims to identify some of the novel ethical, legal and social issues these disruptive technologies present.



Alison is a trained lawyer and nurse and has a Masters in medical ethics. She contributes legal and ethical expertise in policy development, particularly the need for adaptation of consent, data protection and feedback procedures in the light of new technologies.

To find out more about the *Realising Genomics* project email Alison at

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In the *Realising Genomics* project we will explore the views and experiences of stakeholders through a series of four workshops; undertake rigorous conceptual and ethico-legal analysis of the issues; and formulate and prioritise policy advice that supports the successful implementation of genomic technologies in health care.

Working together

Our four multidisciplinary invited workshops will focus on the challenging areas identified from the most recent research:

Workshop 1 Empirical research on clinical implementation of whole genome sequencing technologies (July 2013)

Researchers from around the world met to consider the preliminary findings of their research, generating discussion about the need for new approaches to gaining informed consent and guidance for handling unexpected results.

Workshop 2 The research/clinical interface (late 2013)

Research and clinical activities have been viewed as distinct: we will question whether it is possible - and necessary or desirable - for the boundary between research and clinical care to be maintained in its current form once these genomic technologies are implemented.

Workshop 3 The patient pathway (early 2014)

The introduction of these genomic technologies will impact upon existing patient pathways: we will explore the ways in which existing patient pathways may need to be revised, and the ethical, legal, social and practical issues that are likely to be generated.

Workshop 4 Implications for policy (summer 2014)

In our final workshop, we will build upon the findings from previous workshops and, involving a wide range of stakeholders, formulate policy recommendations and guidelines.

We shall produce a range of outputs throughout the project including policy briefings, as well as a definitive final report, which will be published autumn 2014.

Please visit www.phgfoundation.org/realisinggenomics for updates on the project.

The PHG Foundation is a not for profit health policy think tank based in Cambridge, UK. We work to stimulate the fast, efficient and responsible translation of emerging biomedical science into practical applications to improve population health.

www.phgfoundation.org