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## Expert action plan for bringing genomic medicine to NHS patients

**New recommendations from the PHG Foundation address the challenges facing the NHS as it begins the task of delivering the benefits of genomic sequencing to patients.**

Recent developments in genomic sequencing have the potential to revolutionise the diagnosis and treatment of many diseases, particularly inherited diseases and cancers. Rapid technological innovation and falling costs mean the NHS is on the cusp of introducing new whole genome sequencing tests into clinical practice.

***Realising Genomics in Clinical Practice***, a new report from the PHG Foundation (a non-profit organisation focused on the application of genomic technologies to improve healthcare) guides optimal NHS implementation of genome sequencing technologies by identifying the broad range of ethical, legal, social issues (ELSI) and practical concerns that will arise. It addresses these challenges by proposing a comprehensive set of recommendations for using these technologies in ways that improve healthcare while minimising potential harms.

*Realising Genomics* is the culmination of two years of work, informed by five stakeholder workshops attended by UK clinicians, clinical scientists, patient representatives, policy makers and academic experts. Key recommendations from the report include:

- **Genome sequencing and diagnosis should generally begin with deliberately targeted analysis and interpretation of a set of disease-associated genes consistent with the patient's presenting phenotype** (observable features and symptoms). Using this approach as a first-line test, before analysing and interpreting the whole exome or genome, will help avoid generating large volumes of data which will have adverse or unpredictable impact.
- **Patients' autonomous choices must be recognised by enhancing current processes for seeking consent: consent procedures should include a thorough disclosure of the impact, benefits and uncertainties that may arise.** The nature of the test; the generation, interpretation and disclosure of incidental findings (genetic variants not related to the condition under investigation) and variants of unknown significance, the sharing of data, and the potential for later reanalysis and recontact of patients should be explicitly addressed.
- **Mandatory data deposition into a secure, comprehensive and accessible NHS genomic database should be established.** This is to build the evidence to be able to interpret genetic variants more effectively and ultimately to improve patient care.

Alison Hall, Programme Lead (Humanities) at the PHG Foundation commented:

*"These recommendations are a plan of action for implementing genomic sequencing technologies in an ethical and responsible way in the NHS. By involving clinicians, lab scientists and patients throughout the whole process, we hope that this framework will be robust and effective in driving policy forward."*

*“Unlike the 100,000 Genomes Project, which is concerned with research, the Realising Genomics recommendations focus on clinical care, though there are significant synergies between the two initiatives. Whilst it is possible that some of our recommendations might be addressed by the Genomic Medicine Centres and through research that will be undertaken by the Genomics England Clinical Interpretation Partnerships, independent initiatives are also likely to be necessary.”*

Of note, Alison Hall also observed:

*“Our recommendations do not support actively looking for pathogenic variants in individuals without symptoms or a family history (opportunistic screening), since without sufficient evidence for clinical utility, the potential harms are likely to outweigh the benefits.”*

***Realising Genomics in Clinical Practice*** (full report)

***Realising Genomics in Clinical Practice: Executive Summary***

**-ENDS-**

For more information or to request interviews with spokespeople, please contact Dr Philippa Brice at the PHG Foundation press office **+44 (0)7505092081** or **[philippa.brice@phgfoundation.org](mailto:philippa.brice@phgfoundation.org)**

## **Notes to editors**

- The PHG Foundation is an independent, non-profit health policy think-tank based in Cambridge, UK.
- We focus on how genomics and other emerging health technologies can provide more effective, personalised healthcare and deliver improvements in population health
- Our mission is making science work for health – identifying the best opportunities for 21st century biomedical science and technology to improve health, and offering solutions to the operational and societal challenges that inhibit their effective adoption into medical and public health policy and practice
- We generate knowledge, evidence and ideas to inform, educate, and stimulate debate, and promote new strategies for health service organisation and policy
- We also provide expert research, analysis, health services planning and consultancy services for governments, health systems, and other non-profit organisations
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