phg foundation
making science work for health

Whole genome sequencing

The impact of new DNA sequencing technologies for health

In the light of rapid developments in DNA sequencing technologies, we find ourselves on the brink of a new era of genomic medicine in which whole genome sequencing (WGS) becomes an affordable clinical reality. There is no longer any question that within the next few years it will be possible to sequence a full human genome at a speed and cost that will open up unprecedented new opportunities for health care. Pressure is now mounting to ensure that this potential is promptly, effectively and responsibly harnessed for a real impact on health. To meet this challenge, important questions must be considered:

- **What is the role of WGS technologies in medicine and public health?**
- **How will they affect routine clinical practice and the wider public, and what ethical and legal issues do they raise?**
- **What operational barriers exist to adoption within health services, and how should they be tackled?**

The PHG Foundation has launched a major new programme to evaluate and address the implications of WGS for health and society. As an independent genetics think-tank and acknowledged founder of the field of public health genomics, we have been at the forefront of efforts to catalyse the timely and responsible application of useful biomedical innovation into health practice for over a decade.

Our aim is to identify the best health applications of WGS technologies and recommend strategies that will be pivotal in directing their adoption into health services to improve patient care. We believe that the best approach is to work together with forward-thinking sector leaders to make this happen, and are seeking partners and supporters for this programme.

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Genomics

Our understanding of the genetic basis of disease has increased dramatically over the past decade. There are now thousands of genetic tests for single gene disorders, allowing diagnostic and carrier testing for individuals and families affected by known forms of inherited disease. Genomic analysis is also starting to be used to uncover previously unidentified causes of inherited disease, as well as to guide and monitor treatment strategies through pharmacogenetic testing and tumour profiling.

Sequencing technologies

At the same time, the exponential development of new high-throughput and massively parallel DNA sequencing technologies has radically reduced both the cost and the time required to sequence an entire human genome. The first human genome took around five years to sequence and cost several billion dollars; today, the same process can be completed within weeks for tens of thousands of dollars using next generation sequencing technologies. Just a few years from now, whole genome sequencing (WGS) will be possible in a matter of days, for less than the current cost of sequencing a single gene. In addition, using the same technologies, gene expression profiling and epigenetic analyses are becoming simpler and cheaper.

The future

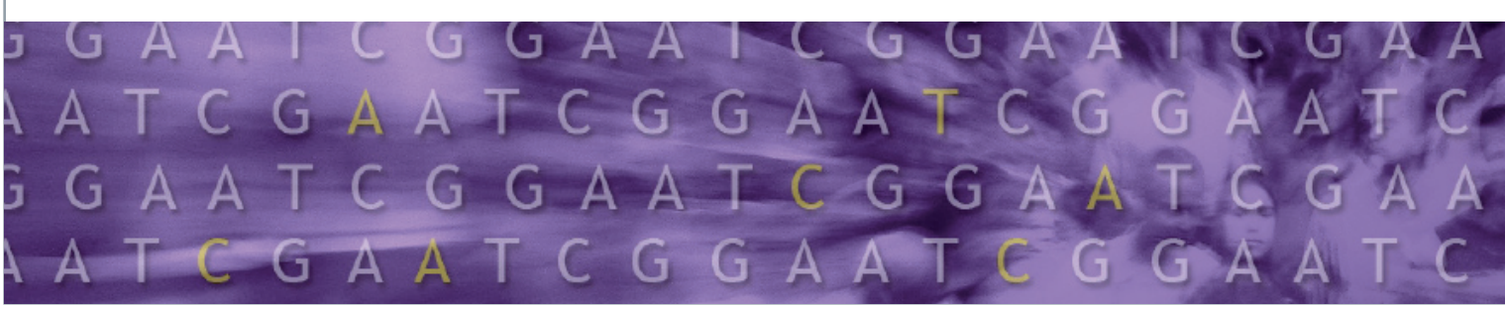
There is no doubt that WGS will become technically and economically feasible in the near future, and will have a significant impact on the practice of medicine and public health. However, efforts to consider how best to harness these scientific advances to improve the health of individuals and populations are lagging behind. Prompt development of a strategic vision focused on clinically actionable health applications of WGS will maximise the health benefits and efficiency savings for the future.

Our response

Our new programme investigates how WGS technologies can be directly used to improve health in the near future, focusing primarily on the UK NHS, with additional consideration of international implications for health and healthcare systems. Our multi-disciplinary knowledge and expertise makes us uniquely well-placed to lead this crucial work.

Method and outputs

This programme will be a key focus for the PHG Foundation over the next year. Guided by expert advisors, it will include major workshops for stakeholders and and policy makers to examine the implications of WGS for health. Our findings will be published in a series of peer-reviewed articles and a final public report.



Specific objectives

1. To review existing and emerging 2nd and 3rd generation sequencing technologies, including different methods of targeted and bioinformatics analysis, and their clinical applications to date
2. To analyse issues relating to the development, maintenance and validation of appropriate informatics platforms for storage, access, annotation and clinical interpretation of genomic data
3. To evaluate the utility and impact of WGS technologies for the prediction, diagnosis and management of diseases with a strong heritable component, and the implications for pathology services, clinical genetics and other medical specialties
4. To evaluate utility and impact of WGS technologies for the diagnosis, monitoring and treatment of cancer, including the potential for personalised and stratified medicine, and the implications for pathology services, oncology and other medical specialties
5. To outline service models and workflow pathways describing how WGS technologies might be used in clinical practice, and their economic implications within the UK NHS
6. To explore the economic, ethical, legal and social implications of implementing widespread WGS for specific medical applications
7. To investigate the possible impact of private companies offering WGS and analysis services to healthcare providers and on a direct-to-consumer basis

Expanding the scope

Other major implications of WGS technologies not currently within the scope of the project include: pathogen sequencing and monitoring infectious diseases; targeting population screening programmes based on genetic risk prediction; forensic and identity testing; and streamlining drug discovery and pharmaceutical development.

If there is interest in broadening the programme to include additional elements, and appropriate funding can be secured, we would be happy to discuss this option with potential partners.

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Partners

We have already secured several major partners for this work, including the National Genetics Reference Laboratories and the University of Cambridge Centre for Science and Policy. We also have strong links with national bodies including the UK Genetic Testing Network, the Human Genetics Commission, the newly established Human Genomics Strategy Group, the Joint Committee on Medical Genetics, the British Society for Human Genetics and the Royal College of Pathologists, as well as local centres of excellence including the University of Cambridge School of Clinical Medicine, the Eastern Sequence and Informatics Hub, and the Wellcome Trust Sanger Institute.

About us

The PHG Foundation is an independent genetics think-tank based in Cambridge, UK - home to one of the world's most successful bioscience clusters and the internationally renowned University of Cambridge. We work closely with academia, the healthcare and corporate sectors.

As a charitable foundation, we use our multidisciplinary expertise to identify and evaluate biomedical innovations with the potential to improve health globally, and to encourage their adoption by health systems. For example, our influential response to the House of Lords Science and Technology Committee Report on Genomic Medicine set out a series of recommendations for the strategic development and implementation of genomic medicine in the UK.

Our strategic aims are to:

- **Identify and promote the most beneficial opportunities for biomedical innovation to improve health and tackle disease.**
- **Develop and promote models and capacities for the effective mainstreaming of biomedical innovation into improved clinical and public health policies and practices.**

How you can help

With our considerable track record and influence in this area, we expect this project to boost awareness of the potential of WGS technologies for health among different audiences, growing the field and generating a resource that will be immensely valuable for market analysis and healthcare planning.

To maximise the impact of the project we wish to combine our expertise with that from other sectors, and are actively seeking partners willing to provide financial support, and to share their knowledge, skills and networks.

To find out more

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