

ADVANCES TOWARDS PERSONALISED MEDICINE: WHAT IS THE COST OF KNOWING YOUR GENOME?

By Kimberley Bryon

Since the completion of the Human Genome Project (HGP) in 2003, scientists have been working towards making whole genome sequencing a useful clinical diagnostic tool. The aim of many of the big biotechnology companies is to make this technique affordable and accessible so that it can be routinely used to diagnose rare genetic disorders and tailor medical treatment to an individual's genetic code – a practice called personalised medicine.

The cost of sequencing the human genome has rapidly fallen from an estimated cost of about \$3billion for the HGP towards a more affordable \$1000. The '500 project' – a collaboration between the University of Oxford and the DNA sequencing company, Illumina, aims to completely sequence 500 genomes from people with a variety of different conditions – from cancer to immunological disorders and rare inherited diseases - to look at how this kind of knowledge may impact diagnosing and treating diseases in the future.

Recently, a 4-year old girl became the first Briton to have their entire genome sequenced as part of the 500 project. The girl was subsequently diagnosed with a rare genetic mutation on the X chromosome leading to craniosynostosis, a condition that hinders expansion of the skull and restricting brain growth. Now that her condition is known, doctors can determine the best possible treatment options.

The concept of personalised medicine isn't new. Herceptin is a drug used to treat breast cancer patients with mutations that are Human Epidermal growth factor Receptor 2 – positive. It has proven to be a more effective treatment than other drugs whereas patients with breast cancer caused by different mutations respond better to alternative treatment options.

The advantages of personalised medicine are clear – fewer costly clinical tests and less time spent taking ineffective treatments. However, could a move to personalised medicine have more sinister repercussions?

In Britain we are fortunate in that we have the NHS. If you have a heart attack, suffer from a stroke or get diagnosed with cancer, unless you opt for private care, this treatment is free. However, some countries such as America depend entirely upon private medical insurance. If whole genome sequencing became a routine medical procedure, it is possible that insurance companies would gain access to the medical information encoded in your genome. Undoubtedly, for many this would push insurance premiums up, even if they never developed the predicted condition.

According to *The Times*, geneticists recently attended a 2-hour seminar organized by David Cameron, to offer advice on how the NHS can make personalised medicine routine. In an interview with *The Times*, Sir John Bell, the UK Government's chief medical advisor said that the NHS is 'unprepared' (M. Henderson, 2011, a) and lacks the infrastructure to store the large amounts of sensitive information generated by personalised medicine safely, leading to concerns of privacy breaches (M. Henderson, 2011, b).

It appears that personalised medicine is fast moving from a futuristic technique towards being a routine clinical procedure. If it does become a commonplace practice, we will need to address a number of ethical questions so that we can appreciate the true cost of knowing your genome.

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Ph.D. MRC LMCB
Faculty of the Life Sciences

References

Henderson, Mark (a), 'All in the Genes' *The Times*, 3 August 2011.

Henderson, Mark (b), 'Revolution in genetics exposes NHS flaws' *The Times*, 3 August 2011.