"What if we could tick that box on our investigation request screen?"

Personal genotyping and general practice

The future is here. *Right now*, it's happening in front of us! 11 years ago, finishing my first degree in pharmacology, the sequencing of the human genome was the talk of the scientific world. The consequences of unravelling the genetic material associated with not only who you are but also what you will become spanned across all scientific communities. My interest at this time was pharmacogenomics — the idea that medicines will be tailored to your individual genetic profile. People were sceptical and dismissed the idea as fanciful, futuristic and likely to be crushingly expensive.

Three weeks ago, I paid a sum less than that required to attend an outpatient appointment to have my DNA commercially analysed for 1 million common genetic variations. The results were quick, comprehensive and provided information I thought a distant dream. Prior to the testing I sought to inform myself of the risks to my mental health, my fiscal health and indeed my future health by undertaking the test. Many people reacted negatively to the thought of undergoing personal genotyping but my feeling was that learning more about myself could only help me plan for my future.

The results were astounding and remarkably accurate. They depicted me as of Northern European ancestry, having brown straight hair, blue eyes, and identified hundreds of third-to-fifth cousins the world over. It identified me as a bitter-tasting, lactose-tolerant, malaria-sensitive adult who was more likely to be a sprinter rather than a marathon runner. Importantly, however, it also highlighted me as extremely unlikely to develop type 2 diabetes mellitus but it demonstrated my higher than average risk of restless legs syndrome and of gallstones. The results reflected my increased likelihood of responding to typical antidepressants and of developing atrial fibrillation; subsequently it delineated my increased sensitivity to warfarin. It revealed that I have a normal sulphonylurea clearance and am low-risk for fluorouracil toxicity; all-important when dealing with pharmacokinetics and drug toxicity.

After several pages of debriefing one can choose to review their risk of

Alzheimer's disease in the future. The well-studied ApoE4 gene and its allelic variations can provide information into a future preponderance to developing the disease. Similarly for females, the BRCA gene and its mutations can provide valuable information. These test results can allow those, who feel specifically able to deal with this information to plan for their future health. I am someone who likes to plan, but I understand that others may prefer not to know.

After reviewing my HIV-resistance, smoking behaviour, response to hepatitis C treatment, and adiponectin levels, I began to think about my results in the context of general practice. What if we could tick that box on our investigation request screen? How soon until we start requesting single-nucleotide polymorphism tests before initiating antidepressants or clopidogrel? If genetic profiling became available the whole care paradigm as it stands would change. We could direct interventions to those at higher risk of developing conditions before it happened. We could tailor medication to those who are sensitive. We could write an advanced decision should our risk of Alzheimer's disease be higher than average. General practice would tilt even further towards proactive risk reduction, less about reactive management of unsuspected illness.

Whether the doctors of today are prepared for an onslaught of genetic information is unclear and the results clearly need to be contextualised. Many diseases are polygenic and consideration must be given to a variety of epigenetic and non-genetic factors. But to provide the saliva sample and receive a taste of who I am, what I am, and what I will potentially become, was something I would never take back.

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