Vitamins Health:
Genetic testing – panacea or dystopia?

Genetic testing is moving from science-fiction to mainstream – what are the implications for life expectancy?

Genes are short segments of DNA which tell your cells how to function and encode what traits to express. Each of us has inherited approximately 20,000 genes from our parents, with slight variations in these genes leading to the differences that we observe within and across different populations. Mutations can cause health problems if the mutated gene stops carrying out the instructions needed for the body to function properly. Genetic tests aim to find mutations in particular genes or identify the presence of specific genetic variants that associate with certain medical conditions. This can help diagnose a disease as well as identify the best choice of treatment for that individual.

Since the millennium, great progress has been made in understanding the human genome. In 2003 the first full human genome was sequenced at an estimated cost of $3 billion. It is expected that the cost will soon fall below $100.

There are great hopes for how genetic testing can help us live healthier lives. It is hoped that collecting and analysing this wealth of data will bring in a new era of what is often called 'Personalised Medicine'. This means using our genetic data (along with other diagnostic tools) to determine the most effective interventions to improve our health and fight disease, whether through medicine, lifestyle choices, or even simple changes in diet.

Current uses of genetic testing

Genetic testing is already available for over 2,000 conditions, both rare and common. These tests are used by healthcare professionals for a number of different purposes:

- **Diagnostic testing** – diagnosing an existing genetic condition
- **Predictive testing** – identifying an increased risk of developing a specific disease in the future
- **Carrier testing** – testing for recessive genes that may be passed on to your children
- **Prenatal and pre-implantation testing** – identifying genetic diseases in foetuses and embryos
- **New born screening** – testing new born babies for certain conditions
- **Pharmacogenetic testing** – helping choose the most effective medicine based on your genes

These tests tend to focus on specific genes and diseases. Genetic testing is also carried out for more general research purposes to help us understand how our genome as a whole impacts upon health and disease. As the volume of genetic data increases, so does the potential for further breakthroughs.

The 'Direct to Consumer' genetic testing market has developed in parallel to these healthcare applications, with companies such as 23andMe fast becoming households names. It is worth noting that companies like 23andMe do not perform whole genome testing, but rather rely on spot checking an individual's DNA at specific preselected sites based on known mutations.

These companies have not been without criticism. 23andMe currently test for genetic variants which are risk factors for nine health conditions, including Alzheimer's disease, Parkinson's disease and breast cancer. Concerns have been addressed about the psychological impact of receiving a positive test for one of these conditions, about the accuracy of these tests and about the level of understanding of the results among users.

For example, 23andMe test for three variants in the BRCA1 and BRCA2 genes which are associated with an increased risk of breast cancer. In total, more than 1,000 variants in these genes are known to increase cancer risk, and the particular variants tested for are only common in people of Ashkenazi Jewish descent. Receiving a negative result to such a test, particularly for those with a family history of breast cancer, may lead to an unjustified sense of complacency.

As well as health conditions, 23andMe also looks at things like genetic traits as well as ancestry. Therefore, such tests may currently be best viewed as a bit of fun rather than a useful healthcare tool, albeit one which could come with an unpleasant surprise.

The future of genetic testing

As datasets grow, so does the opportunity for insights. One interesting concept is the 'Polygenic Risk Score', developed using a data-driven approach known as a 'Genome Wide Association Study'. Such studies use statistical methods to identify a whole host of genetic variants which have a positive or negative impact on the risk of developing a particular disease or trait. For example, a recent study calculated polygenic risk scores for heart disease, diabetes, atrial fibrillation, inflammatory bowel disease and breast cancer using genetic data from around 400,000 people in the UK Biobank. Around 20% of participants were identified as having a threefold risk for at least one of these diseases.

By identifying individuals with an elevated risk for a particular disease early in life or even at birth, interventions can be put in place. For example, in the case of heart disease, drugs like aspirin and statins could be prescribed in a more targeted fashion, reducing unnecessary side effects. And individuals could be provided targeted guidance as to how to reduce their risk through healthy eating, exercise and other lifestyle changes.

Genetic testing also has implications for the treatment of existing cancers. Cancer is caused by mutations to the DNA within cells, and cancer genomics is the study of the genetics of tumours. By understanding the specific mutations that led to the growth of a tumour, custom-designed treatments can better target cancerous cells without harming healthy ones.

We have also recently seen some news on the darker side of genetics and genetic testing. Late last year, a Chinese scientist claimed to have created the world's first genetically edited babies using a gene editing tool. We can only hope that legislation and ethics keep up with the technology to ensure that our ever-growing understanding of the human genome is used for the common good rather than individual gain.

Implications for future longevity improvements

It feels inevitable that healthcare will increasingly adopt the use of genetic testing as datasets grow, insights are harvested and costs reduce. This is definitely a good news story, and Personalised Medicine is likely to be one of the key drivers in improvement of life expectancy in the coming decades.

For us to benefit fully from genetic testing, it is likely that individuals will need to modify their behaviour to best play the particular hand they are dealt through their genes. Sadly, the evidence to date is that presenting highly personalised risk information (including genetic tests) does not produce sustained changes to peoples' lifestyles. (In contrast, the evidence shows that individuals do tend to increase their level of insurance on being provided with an adverse genetic test – causing a potential headache for life and health insurers.)

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