Gene testing for the public—a way to ward off disease, or a useless worry?

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Credit: AI-generated image (disclaimer)

The launch in Australia of a genomic testing service aimed at healthy people heralds a new era of individual patient care. A scan of your genome, which is the complete set of your genes, to find out if you are at risk of particular diseases, can mean you can then go on to take preventive measures against them.

The CEO of the Garvan Institute's Genome.One lab, which is offering the testing, said it would transform the health system, making it more focused on prevention than treatment of disease.

Genomic testing can have tremendous benefits, as in the case of diagnosing children with rare diseases. When applied to the right patients, genomic testing can provide a diagnosis for more than half of patients with unusual symptoms. And the cost of this to the health system is much lower than for traditional diagnostic tests.

Certainly that all sounds like a good thing, but genomic testing is not yet the precision diagnostic and treatment tool we hope it will one day be. And all genetic knowledge is not necessarily helpful. As with any medical intervention, genomic testing carries risks as well as benefits.

Why genomic testing?

Genomic testing takes advantage of recent advances in our knowledge of genetic causes of disease, as well as technology. It's a test of all 23,000 genes in the body at once.

The success of genomic testing in diagnosing rare disorders has raised the question of whether these tests should be performed in healthy people before they become sick. The potential benefits of testing healthy people are obvious, especially when it comes to conditions that have a proven treatment or prevention.

Cancer is a good example of where genomic testing can save lives. A person found to carry a genetic predisposition to bowel cancer can choose to have regular colonoscopies, which can detect and remove pre-cancerous growths before they cause harm.

And because genetic disorders run in families, potential health benefits

can extend to other family members who may have the same genetic predisposition.

The ultimate goal of genomic testing, as part of personalised medicine, is that it will be available to everyone, allowing each person's health care to be tailored to their individual genetic make-up. In the future, this "lifetime health resource" promises to improve health care from conception to death.

Are we ready for this?

A considerable challenge of genomic testing is the extraordinary complexity of each person's genome. To try to interpret a single human genome is to grapple with literally millions of genetic variants, or points where the person's genetic code differs from the average person's.

Perhaps a handful of these variants will cause disease, but the rest will most likely be harmless. Determining which is which is far from straightforward.

Another problem is that even when specific genetic variants are judged to be harmful, the benefits of knowing this information are not always as clear cut as in the case of bowel cancer. It is an unfortunate reality that most disorders detectable by genomic testing have no proven treatment or means of prevention.

For instance, particular gene variants may put you at risk of developing dementia in your 60s. But if there was nothing you could do to prevent it, would you want to know?

Even when treatments are available, the benefits of knowing you have a certain genetic predisposition may not outweigh the disadvantages.

Consider that genomic testing finds you carry a predisposition to sudden heart death, such as Long QT syndrome. This is an outcome you would certainly wish to avoid. But what if knowing this information caused you to worry more, and the treatment required you to give up sport and take a medication that caused you to feel lethargic every day?

And what if, in the absence of symptoms, your risk of actually dying was only slightly increased compared to the general population? Would you still want to know this information, or perhaps prefer to remain ignorant?

Should we get the test?

The Genome.One clinic at the Garvan Institute in Sydney has addressed some of these concerns by taking a cautious approach.

Genetic counselling is provided before and after testing, and although the whole genome is sequenced, analysis and reporting is limited to just 1% of all genes. Most of these selected genes are associated with heart conditions and cancers, and have been chosen because these diseases are well understood, with treatment strategies available.

Genes that cause untreatable diseases, such as dementia, have deliberately been excluded from analysis. This strategy minimises the risk of harm that may come from the test, but the trade-off is that the likelihood of actually finding something useful is greatly diminished. In fact, Genome.One reportedly estimates only 5-10% of people tested will receive an abnormal result; that is, one that will show them to be at risk of disease.

While it is hard to argue against a test that just might save your life, currently there is insufficient evidence that the benefits of genomic testing outweigh the risks. Even for those who can afford the price tag of

A$6,400, there are probably more effective targets for our health-related spending. Like many years of gym membership, for example.

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