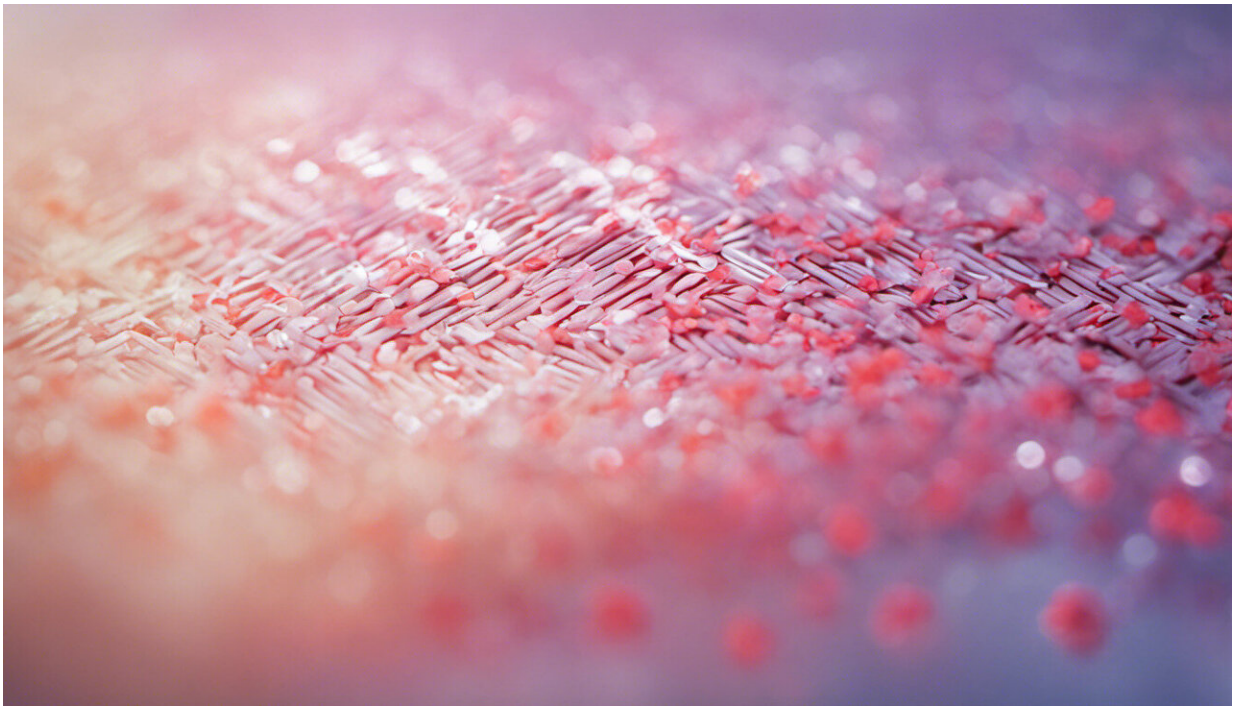
Cheap genome tests to predict future illness? Don't hold your breath

January 20 2014, by Clara Gaff



Credit: AI-generated image (disclaimer)

Sydney's Garvan Institute is this week promoting its acquisition of an Illumina machine which it says can sequence the whole human genome for $1,000. The institute hopes genomic sequencing will become widely available in the near future, so Australians can understand and reduce their personal risk of certain diseases.

Genome sequencing is currently used to diagnose and treat some inherited disorders. But before the technology can be used more widely, we need to ensure its use will bring improvements in health, quality and duration of life.

Genome sequencing

The first human genome was sequenced 15 years ago at a cost of US$3 billion dollars and took over a decade to compete and assemble.

Sequencing an entire genome is a large task: if the human genome were a novel, sequencing it would be the equivalent to reading War and Peace one thousand times. The human genome comprises approximately 25,000 pairs of genes that we each inherit from our parents. These provide the instruction code for all our bodily functions from the moment of conception.

Each of our individual genomes is different. There are about 3 million differences (variants) in the genomes of any two unrelated people. Many of these variants account for physical differences, such as eye colour, between individuals. Some influence the risk of diseases such as diabetes.

Rare variants that disrupt a gene's normal function (called mutations) may cause diseases that may be inherited. Knowing this information may help decision-making about management of disease or even enable individuals to reduce their disease risk by modifying diet and exercise, for example.

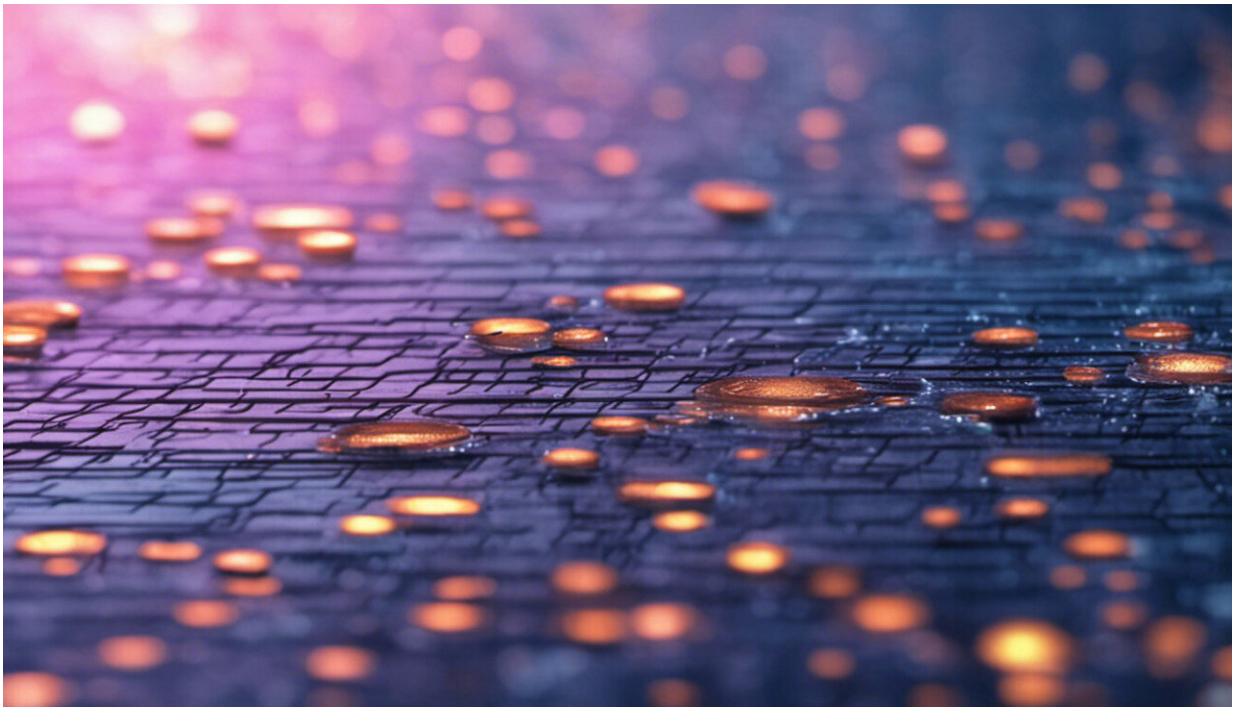
If applied on a wide scale, this technology could help reduce the burden of genetic disease in society.

Current use

In Australia, access to genetic testing has been limited largely to single gene tests for single genetic disorders. Only a handful of these tests are currently paid for through Medicare.

We are starting to see the introduction of gene panels that can screen for dozens or hundreds of high-risk genes in a single test. This is particularly useful for complex gene tests such as those used to identify patients with a high familial risk of developing cancer.

It is also possible to order gene tests from laboratories overseas that sequence the 1% of the genome that codes for the protein–coding genes (so called exome) or which can scan for many thousands of single risk variants. These tests are only available in Australia in a research setting but are likely to become routinely available to high-risk individuals and families in a year or so.



Credit: AI-generated image (disclaimer)

Genome sequencing is currently used to diagnose and treat some inherited disorders. Image from shutterstock.com

Currently, this technology is being used to identify acquired mutations that arise in cancer cells to identify mutations for which there are specific therapies (so called targeted therapies). The aim here is to screen the patient's tumour for particular cancer mutations in order to match the right treatment to the right patient.

This personalised approach to medicine is being quickly embraced by oncologists and their patients.

But the true cost of performing this testing is higher than the base cost of $1,000 and includes the costs of timely sample processing, analysis, complex interpretation and reporting. These labour costs are presently unknown but could be several times the cost of the sequencing.

Death calculator? Not quite

Understandably, there is a lot of community interest in healthy people having their genomes sequenced to predict, and hopefully prevent, disease in the future.

But while we may be able to work out what an individual's genetic sequence is, we don't necessarily know what it means or know how to use the information. We are certainly a long way off being able to predict anyone's quality or duration of life.

One difficulty is that what causes a condition in one person may not affect another. For instance, one researcher points out that he carries a mutation known to cause a severe childhood condition, yet has no signs or symptoms of the condition.

Over time, large studies will build good evidence for the effects of variants associated with common conditions, as well as on the effect of prevention strategies based on this information.

In the meantime, ethical issues are arising over preventive testing. The United States' National Institute of Health recently announced funding to investigate the use of genomic sequencing in screening of newborns.

As well as diagnosing conditions affecting these babies sooner, genomic sequencing could also predict the development of rare inherited adult onset conditions, such as hereditary breast cancer. This is information that some healthy, high-risk adults prefer not to know, while others may want to learn as much as possible.

There is currently no consistent view on how to deal with unexpected findings, which may or may not have health implications some time in the future, particularly when testing would be performed at birth or during pregnancy.

Genetic counselling, however, aims to facilitate decision-making based on personal values and accurate information, as well as promote adjustment after any results are received. This may help people considering testing to fully consider these complexities and their own preferences before undertaking any genetic testing.

Regulation

In Australia, laboratories offering genetic tests for medical use must be

accredited to perform the particular tests. This helps ensure that all tests are both safe and effective and are performed only in an appropriate clinical context.

Government funding for such tests also requires the demonstration that the test and any associated treatments are cost effective.

These regulatory and funding requirements will present challenges for the new genomic technologies that are able to screen for all potential inherited disease risk in a given individual.

Much more research is needed to understand how best to use the vast amounts of information generated by these machines and to identify those individuals who will derive most benefit from or be harmed by this technology.

We also need to determine how to use genomic information cost effectively to improve quality of life and health outcomes.

Source: The Conversation