Australian Government
National Health and Medical Research Council

Sequencing your genome

Rapid advances in DNA sequencing technologies now allow an individual's whole genome to be sequenced. Although this is still relatively expensive, it is likely that in the near future it will become affordable and readily available.[1]

What does sequencing the human genome mean?

Genome sequencing involves working out the order and arrangement of the genetic code or DNA sequence, which is made up of sequences of four building blocks known as nucleotide bases: adenine (A), thymine (T), guanine (G) and cytosine (C). DNA sequencing is technically difficult and expensive. However, being able to read this code and understand how cells translate the information is fundamental to our knowledge of how our genetic make-up influences our health, growth and behaviour.

How much have we learnt about our genes?

Over the past three decades, we have gained a better understanding of the information contained in our DNA and the role of our genes. Understanding all of our genes and how they interact with each other will take considerably more time. We are also a long way from knowing how our genes interact with the environment, which is an important consideration for common health issues such as diabetes, dementia and heart disease.

How much would it cost to sequence my entire genome and can it be done?

In 2005, it cost about US$10 million to sequence a human genome, although the cost was 50 times lower than a decade earlier. In June 2007, Dr James Watson, who discovered the structure of DNA with the late Dr Francis Crick, received his full genome sequence recorded on a computer hard drive for less than US$1 million. By 2012, the price for sequencing a genome had decreased to under US$10,000.

1 In addition to the costs of sequencing, there are costs for analysis and interpretation of the data generated.

Today, the cost of sequencing an exome (the coding regions of your genes) or a genome is in the order of $1,000-2,000, and it is expected that even this cost may fall further in the near future. This cost is for the actual sequencing, and does not include the additional cost of detailed analysis or medical interpretation of this information.

What personal value is there in having your genome sequenced?

This depends on the reason you are undertaking the test. Where a medical practitioner requests a genetic test from an accredited laboratory following careful consideration of your circumstances, this can provide a diagnosis of your clinical condition and may enable you to obtain treatment.

In other cases, people may decide to undertake a test out of curiosity about ancestry or their predisposition to a range of conditions. It is important to understand that if you request whole-genome sequencing in relation to a particular condition, it is likely you will also receive information that you did not request about other genetic susceptibilities. This might raise issues because some of this genetic information could relate to the risk of future onset of other diseases that you might not want to know about and that doctors might not be able to prevent or treat.

If you were to have your genome sequenced today, you would be given a very large amount of information about the billions of bases that make up your DNA sequence. The great majority of this information will have no practical meaning because, at this stage, we do not know enough about the genome and how it works. Even when we know more about the human genome, we will need to understand how genes and the environment can interact before the human genome sequence will make complete sense.

Any genetic information you receive might also have health implications for other family members. Before deciding to have whole-genome sequencing, it is important to consider how the results could affect your family.