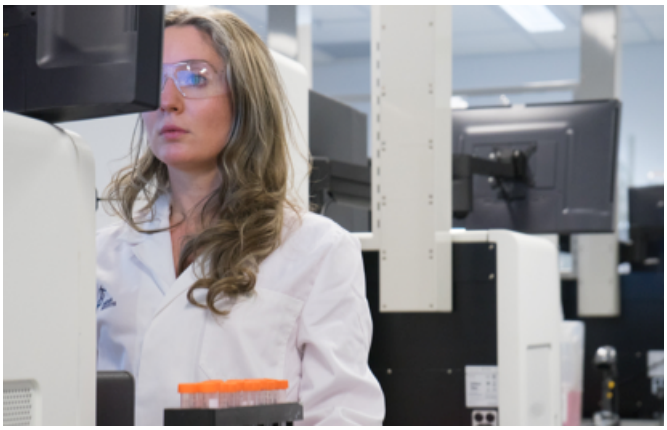
READING GENOMES

In the last two decades, there has been an amazing leap in information and insight into our human genome. This scientific advance has been enabled by new technologies, which can 'read' or 'sequence' the more 6000 million letters in a person's genome – their entire genetic make-up – in days, rather than years.



In 2003, the Human Genome Project was completed and a 'reference' human genome sequence was published: the result of a $3,000,000,000 international effort. This data provided a global infrastructure for biology: researchers around the world could access a common collection of data, and overlay information about DNA variation and function.

Just over 10 years later, a genome sequencing technology company called Illumina, Inc. announced that their new machines could sequence 350 whole human genomes per week, at a base cost of less than 1000 US dollars. The Garvan Institute of Medical Research in Sydney was one of the first institutes in the world to acquire this new technology.

What can you learn from a sequence?

Genomic technologies offer the potential to use personal genomic information to guide medical treatment. Genomic medicine has the potential to transform healthcare because all diseases have a genetic component -- from inherited disorders to complex diseases such as cancer and diabetes.

How could health-related DNA variants be used?

To speed up diagnosis of existing genetic disorders

- To accurately characterise complex disease
- To guide treatment or predict a person's survival
- To better use pharmaceuticals and avoid adverse effects
- To identify new therapeutic targets and design/ develop new therapies
- To better understand biological pathways
- To make reproductive decisions
- To estimate risk of developing disease

What are the issues?

A person's genome contains information that:

- is both unique and shared with family
- can identify a person and their relatives
- may be of interest to others (eg. insurers, employers)
- is mostly stable throughout a person's lifetime.;

Suggested Resources

Genomics information, animations, interactives, and activities for teachers: www.yourgenome.org

Video: 'Whole genome sequencing and you' https://www.youtube.com/watch?v=IXamRS85hXU

'Genome Generation': a card based discussion activity: http://www.yourgenome.org/activities/genome-generation

Science by Doing: Evolution and Heredity https://www.sciencebydoing.edu.au/

Connect with the Garvan Institute's Kinghorn Centre for Clinical Genomics:

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