Genomic Information booklet

Why is health research important?

- Can help answer questions about what makes people healthy and what can make people sick.
- Can help develop medical guidelines on who we should treat and how
- May help people, families and communities to make decisions about their health and live healthier lives – now and in the future.
- May lead to better treatments, better ways to identify health conditions early, or better ways to run health services.

Participation in such research is always voluntary. You can choose to participate or not to participate in health and medical research.

How does taking part in health research benefit me?

For some research studies, there may be a direct benefit to you. Some examples of how taking part in health research that could benefit you:

- A research study may provide you information about your risk of developing a health condition, such as heart disease. This information could help you take the steps, with the help of your GP, to reduce your risk of disease (e.g. through medication or lifestyle changes).
- If you have severe illness but none of the prescribed medications are helping, you may be able to take part in research where you get access to the newest treatments that are being developed.

For other research studies there may not be any direct benefit to you. However, by participating in a research study and providing your data, you will help researchers better understand human health. This work can lead to better treatments and healthcare in the future, not just in Australia but worldwide.

What is genetic/genomic research?

Genomic research is a type of health research that looks at how people's **genetic information** affects health and the way bodies work.

Genetic information contains the instructions that bodies need to develop, grow and function. The instructions are mainly found in sections of DNA called **genes**. DNA can be found in all parts of your body, including blood and saliva.

All of a person's genetic information is called their **genome**.

Changes in genome sequence are called genetic variants. It is normal to have variations in your DNA. It is these variations that makes us all unique. Most of the time, these variants have no effect at all. But sometimes, variants can cause health conditions, or increase the chance that we might develop health conditions in the future. Some variants also affect whether you respond well or badly to certain medications.

For many common diseases, including cancer, heart disease, diabetes and depression, both our genetics and lifestyle play an important role. Though we cannot change our genetics, knowledge of our genetic risk may help us to change our lifestyle to try and reduce disease risk.

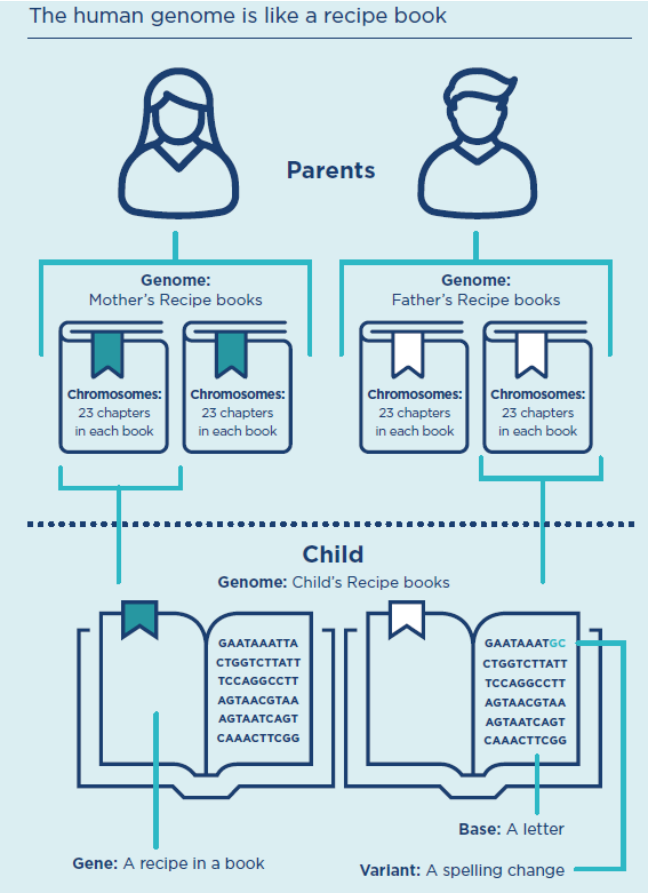


Figure 1. From Centre for Genomics Education: Fact sheet 01

Many genomic research studies aim to use information from peoples' genomes and their health to better understand what variants contribute to disease. This knowledge can help researchers to develop better ways of preventing, diagnosing and treating disease.

Every person's genome is unique (except for identical twins), but you may share some parts of your genome with your blood relatives. This includes parents and grandparents, brothers and sisters, and children. People from the same ancestry also have some similar genetic information.

Your genetic data may reveal undeclared biological relationships but these will never be revealed unless required by the law.

How is genomic data used in Australian healthcare?

In Australia, genomic information is already being used in healthcare. For example,

	**Newborn screening:** Every newborn in Australia is offered free screening for rare, genetic conditions through a simple bloodspot test. This mean that babies with rare conditions can receive early care and support, leading to better health outcomes for the baby and their family.
	**Cancer treatment:** Doctors can prescribe the most suitable treatments to cancer patients, if they know about specific genomic variants that are present in those cancer cells.
	**Disease diagnosis:** Knowing whether someone's condition is caused by a genetic variant can help doctors and patients make decisions about medications, surgery and family planning.

How is genomic data generated?

DNA can be extracted from different types of biological samples, such as blood and saliva. Blood is the best source of DNA. The DNA is then analysed using specialised technology and machines, which produces large amounts of genetic data. We use this genetic data to look at differences and similarities between people's DNA, and understand more about genetic risk of disease.

Why is diversity of participants important in genomic studies?

Some variants that may be important for our health are only present in people from a particular ancestry, making it very important to have people from diverse backgrounds in genomic studies. For some diseases there can be differences in risk, age at which disease starts and which treatments work best, depending on ancestry. We will only know about these differences if people from different ancestries participate in research and we have genomic and health data on these communities. Knowledge of any differences could help improve healthcare for different communities.

What kind of results can I get from a genomic research study?

Information that is found and offered to people usually tell them about genetic variants that could affect their health in some way. However, any results from genetic research have to be confirmed by your doctor through the healthcare system before any medical support is provided to you.

Genetic risk factors for heart disease

Genetic variants that increase a person's risk of getting disease can be:

Monogenic:

Mono = one

Genic = gene

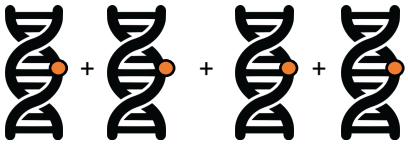
Single variant – big impact

Sometimes a single variant in a single gene is enough to cause disease or cause someone to have very high risk of disease. Such variants are usually rare. Diseases caused by monogenic variants are often called familial (inherited) because these variants can be passed on from parent to child. Genetic testing for monogenic variants is currently offered through the Australian healthcare system for some diseases.

Polygenic:

Poly = many

Genic = genes



Many risk variants, each with small impact

There are many common variants in different genes that on their own do not cause problems. But, they can increase risk of disease by a small amount. We all carry different risk variants for different diseases, and if a person has a large number of these risk variants, their chances of getting disease can be high. We can add up how many risk variants for a particular disease a person carries, and this is called a polygenic risk score. Knowledge of these genetic variants for different diseases and what they mean is still quite new, which is why they are not used in current medical guidelines. In addition, most of the knowledge of these variants is based on data from mostly European ancestry individuals (for example, around 86% of genetic data comes from participants of European ancestry, less than 2% from South Asian individuals, and less than 2% from African ancestry individuals)

What genetic/genomic services are available through the healthcare system?

Genetic Health Queensland (GHQ) is a state-wide service that provides clinical genetics services across Queensland by a team of specialist clinical geneticists and genetic counsellors. GHQ supports individuals and families with a known or suspected genetic condition, offering both face to face and telehealth (telephone or video) appointments for children and adults across the state.
You can find out more at https://metronorth.health.qld.gov.au/rbwh/genetic-health-queensland.

How are my data, privacy and confidentiality protected?

All research in Australia involving humans is reviewed by an independent group of people called a Human Research Ethics Committee (HREC). Approval of a study by a HREC ensures that the research is done in a way that protects the dignity, rights, and welfare of study participants, as outlined in the Australian Privacy Principles (2014), and the National Health and Medical Research Council (NHMRC) Guidelines for Ethical Conduct of Human Research (2023).

As part of this approval, researchers have to take steps to make sure that participant data is stored securely. Personal information (such as your name, date of birth and address) is kept separate from health and genetic data to maintain your confidentiality and privacy. This means that your data is "de-identified". All data is kept on secure, password-protected servers and only researchers who have obtained relevant clearance from a Human Research Ethics Committee and with approval from principal investigators are given access.