Your Guide to Genomic Testing

Queensland Genomics

So, what is genomics?

Genomics is the study of a person's complete set of genetic information including all the genes and also other DNA that sits between genes. While genetics looks at only a few genes (1–5), genomics looks at the big picture. Think of your genome like a large instruction manual for what makes you 'you'.

Genomics can be used to:

- Predict disease risk
- Diagnose disease more accurately
- Guide treatment for diseases

Using genomics in healthcare will become more prevalent in the next few years. Genomics is one of the technologies that's enabling precision medicine — where patients are treated based on their unique genome rather than the uniform approach currently used.

Understanding the language of genomics

DNA	A chemical structure that makes up a person's genetic material.
Gene	A unit of DNA that controls one or more traits, e.g. eye colour.
Variant	A modification in a gene that causes a different trait, for example blue or brown eye colour.
Genome	The complete set of a person's DNA, including all of their genes.

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Accelerating change in healthcare

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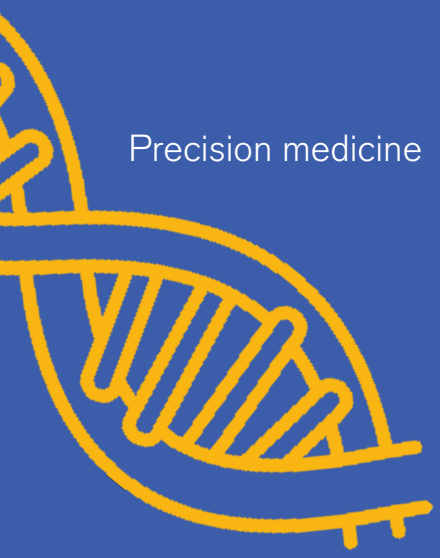
Genome Sequencing	Process that reads a genome to a computer so it can be studied.
Chromosome	In cells, long strands of DNA are tightly coiled and packaged to form structures called chromosomes.
Karyotype	A picture of a person's chromosomes that can be used to diagnose some diseases.
Precision medicine	An approach to patient care that allows doctors to select specific treatments that are most likely to help patients based on an understanding of the genetics of their disease. This is sometimes also referred to as personalised medicine.



Types of Diagnostic Tests

Diagnostic test	Used to determine the cause of a current condition within a patient.
Predictive test	For a person with no signs or symptoms to predict if they are likely to develop the disease in the future. For example, a sibling of a patient with a diagnosis.
Prenatal testing	For an unborn child or the mother of an unborn child to determine if the child has a genetic condition.
PGT	Pre-implantation genetic testing (IVF only) For an embryo prior to implantation to check if the embryo has a genetic variation associated with a genetic condition.
Carrier screening	For couples planning on becoming pregnant to determine their risk of having a child with a known genetic condition.