



# Genomics Information Toolkit for patients

The Genomic Information Toolkit is designed to support individuals and families to engage with and access genetic health services and testing in Queensland.

The Genomics Information Toolkit includes the following fact sheets:

- Introduction to genetics and genomics
- Searching for a diagnosis?
- Clinical Geneticists in Queensland (public and private)
- Your guide to genetic and genomic tests
- Genetics and genomics for GPs

Queensland Genomics would like to thank all our stakeholders who have contributed to the creation of this toolkit, in particular the Queensland Genomics Community Group.

This toolkit is also available to download in the following languages:

- Arabic
- Persian
- Japanese
- Vietnamese
- Simplified Chinese

Visit [queenslandgenomics.org/pct](https://queenslandgenomics.org/pct) for links to download translated versions.

“Genomic testing is helping doctors to pinpoint a diagnosis much sooner, particularly for people with very rare diseases. Like many other people around the world, before genomic testing we had eight years of different tests, and driving from Bundaberg to our specialists in Brisbane close to 200 times for our son Dallas.

When his genetic specialist was able to get whole genome sequencing for Dallas, he was then able to diagnose Dallas – with Van Maldergem Syndrome 2. Dallas is one of only 12 people in the world with this condition.

The Genomics Information Toolkit was an idea I suggested in the Queensland Genomics Community Advisory Group - as a way of giving families accurate information about genetic testing, the different kinds of tests, and where to go for genetic and genomic testing in Queensland.”



**Gary Hondow, Consumer Advocate and Queensland Genomics Community Group member.**

# Introduction to genetics and genomics

Genomics is the study of genomes.

A genome is an organism's complete set of genetic information, also known as DNA. All living organisms have their own genome that is unique to them.

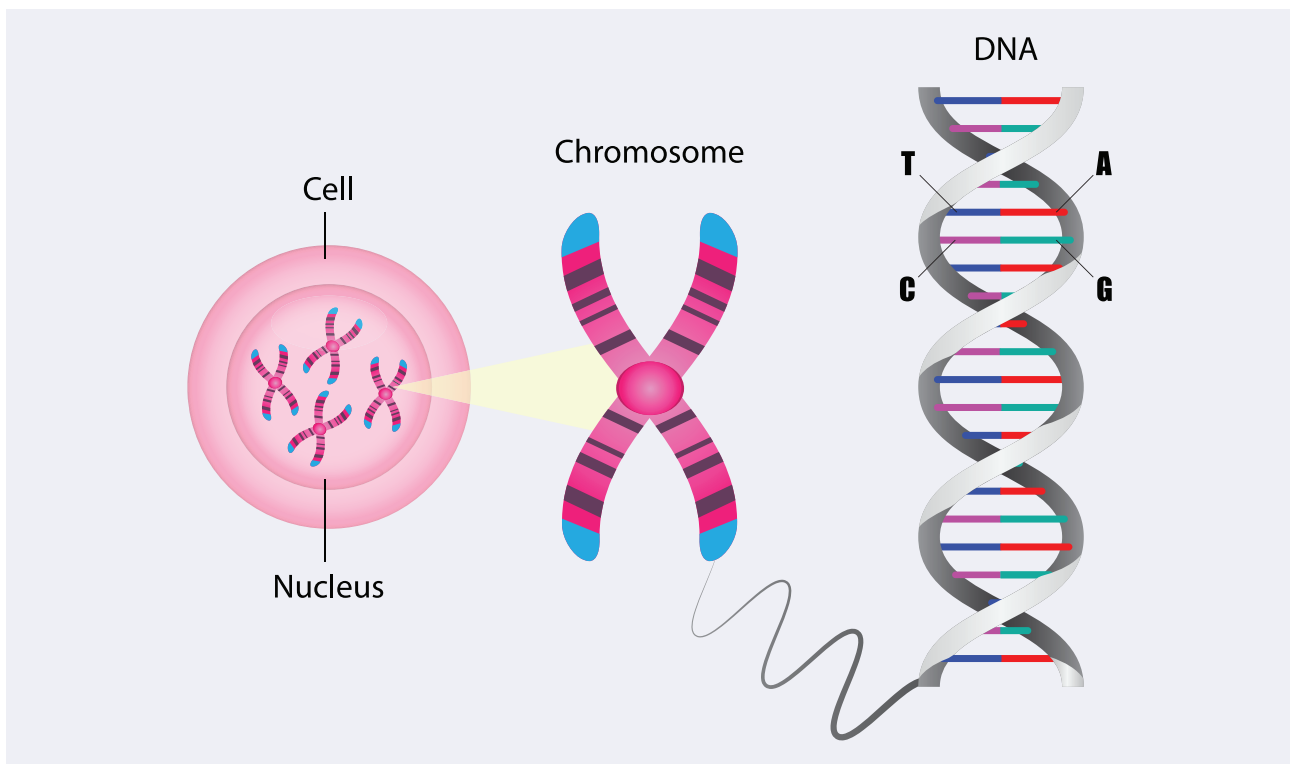
Our bodies contain millions of cells, which are the building blocks that make up all the tissues in our body (for example skin, bones, and muscles).

Inside our cells is our DNA, which tells our cells how to function. DNA can be thought of as the instruction manual that tells your body how to work.

Cells function differently depending on which parts of the DNA are used to get their information.

Your individual genetic information determines many of your traits (for example your eye and hair colour or blood type).

Although not all diseases are genetic, changes (or mutations) in your DNA can sometimes cause health problems.



A genome is an organism's complete set of genetic information, also known as DNA. (iStock)

## Key terms

### Chromosome

The structures in cells where your DNA is packaged.

### DNA

The chemical structure that makes up your genetic material.

### Exome

All the genes in a genome are known as the exome.

### Gene

The part of your genetic material that controls how your traits are developed. Humans have approximately 20,000 genes in their genome.

### Genome

All of your genetic material, including genes and the DNA between genes.

### Mutations

These are the differences in a gene that cause people to have different traits. These can be typical traits like eye colour or detrimental traits that can cause diseases.

## What is the difference between genetics and genomics in health care?

Genomics examines many genes in a diagnostic test. Genetics examines a few genes (1 to 5 genes) in a diagnostic test.

While the technology used in diagnostic tests differs between genomics and genetics, in clinical discussions with patients the words are often used interchangeably without issue.



**Queensland  
Genomics**

Accelerating change in healthcare

Queensland Genomics is working to bring genomics into everyday healthcare in Queensland, to transform the delivery of health services with faster diagnosis, new treatments, and more cost-effective delivery.

This information was produced by the Queensland Genomics Community Group for a Genomics Information Toolkit for patients. This information is correct as at September 2020.

For more information or to view other fact sheets visit [queenslandgenomics.org/pct](https://queenslandgenomics.org/pct).  
For more information on Genetic Health Queensland visit [bit.ly/GHQId](https://bit.ly/GHQId)

# Searching for a diagnosis?

If you have a rare disease or family history you are concerned about a consultation with a genetic health specialist could help you find out if your condition is genetic.

Genetic Health Queensland (GHQ) is Queensland Health's statewide specialist and public healthcare service for people and families with suspected or known genetic health conditions.

## WHAT DOES GHQ DO?

GHQ provides clinical geneticist and genetic counselling services for patients in Queensland's public health system.



A **clinical geneticist** is a doctor with specialist training in diagnosing genetic conditions.



A **genetic counsellor** is an allied health professional with specialised training in genetics.

Both work with individuals, couples and families who are at-risk or diagnosed with an inheritable condition. They also help to interpret results, educate and support patients and their families.

## HOW DO I ACCESS GHQ SERVICES?

Your GP or medical specialist can refer you to a GHQ genetic specialist.

Some medical specialists can also arrange diagnostic genetic tests for conditions within their speciality area.

Your doctor or medical specialist can find clinical prioritisation criteria for general genetics and cancer genetics online:

- **General Genetics** - [www.metronorth.health.qld.gov.au/specialist\\_service/refer-your-patient/genetic-health](http://www.metronorth.health.qld.gov.au/specialist_service/refer-your-patient/genetic-health)
- **Cancer Genetics** - [www.metronorth.health.qld.gov.au/specialist\\_service/refer-your-patient/cancer-genetics](http://www.metronorth.health.qld.gov.au/specialist_service/refer-your-patient/cancer-genetics)

When your appointments with GHQ are complete, your clinical geneticist will write a report for your referring doctor, and send a report and/or letter to you as the patient. Your clinical care will then be managed by your GP or medical specialist based on the report findings.

## WHERE CAN I ACCESS GHQ SERVICES?

GHQ is a statewide service, based in Brisbane with regional clinics.

Where clinically appropriate, you can also book telehealth appointments, which are available to all regional areas in Queensland.



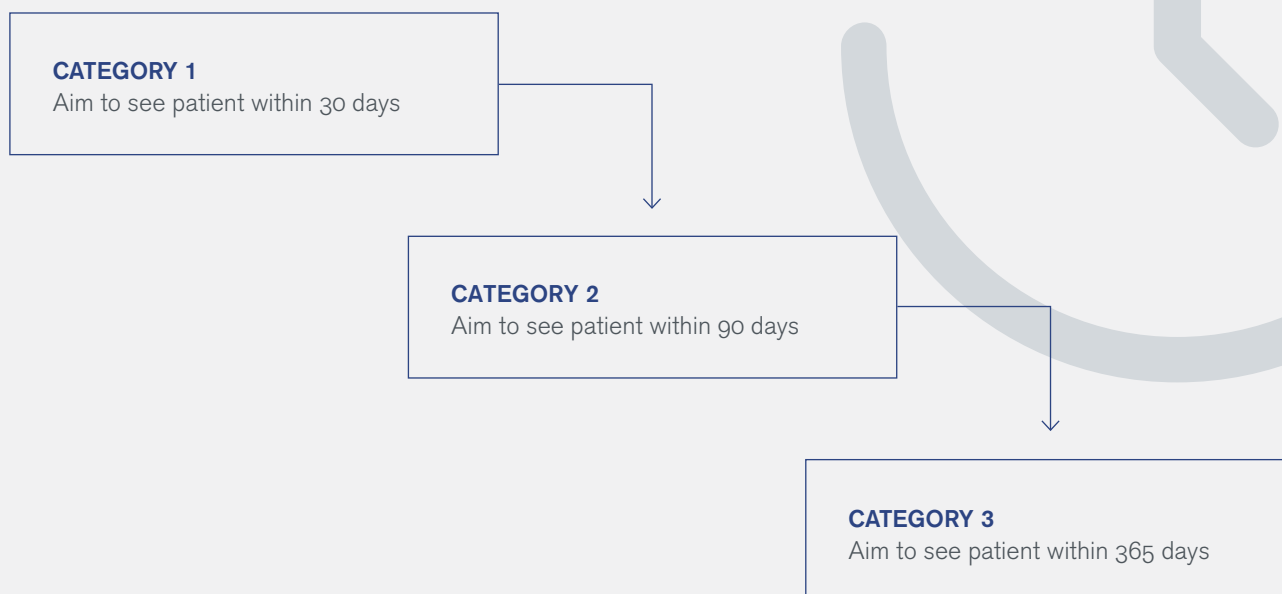
## HOW MUCH DO GHQ SERVICES COST?

GHQ's public health services are provided by Queensland Health. Clinically indicated testing is funded by the hospital or through Medicare. This means you have no out-of-pocket expenses for appointments or clinically indicated testing.

Clinically indicated means testing results may alter the health management or social care of the patient and/or their family.

## HOW LONG ARE WAIT TIMES FOR GHQ SERVICES?

Wait times for GHQ appointments vary depending on urgency - for example, if you are pregnant or have a terminal illness.



## PRIVATE GENETIC SERVICES

### CAN I ACCESS PRIVATE SERVICES?

There are private clinical geneticists working in Queensland. Some offer telehealth services to patients in regional areas.

Refer to the *Clinical Geneticists in Queensland (public and private)* fact sheet for more information.

### WHAT ARE THE COSTS OF PRIVATE SERVICES?

Private patients can self-fund their genetic testing, prices range from a few hundred to several thousand dollars.

When specific criteria are met, some genetic tests can be bulk billed. You can talk to your doctor to find out if this applies to you.

When comparing providers, check what services are included in the price. Some laboratory providers don't include data analysis to generate results and there may be an additional cost for this service.

### WHAT DOES A TEST INVOLVE?

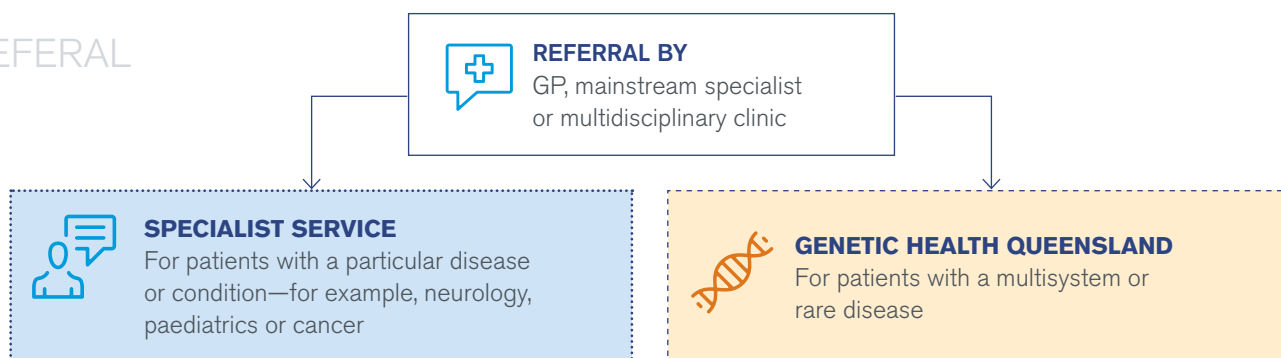
If you choose to go ahead with genetic testing you will need to supply a sample of your DNA - this will be in the form of a saliva or blood sample.

### OTHER SPECIALISTS (PRIVATE AND PUBLIC)

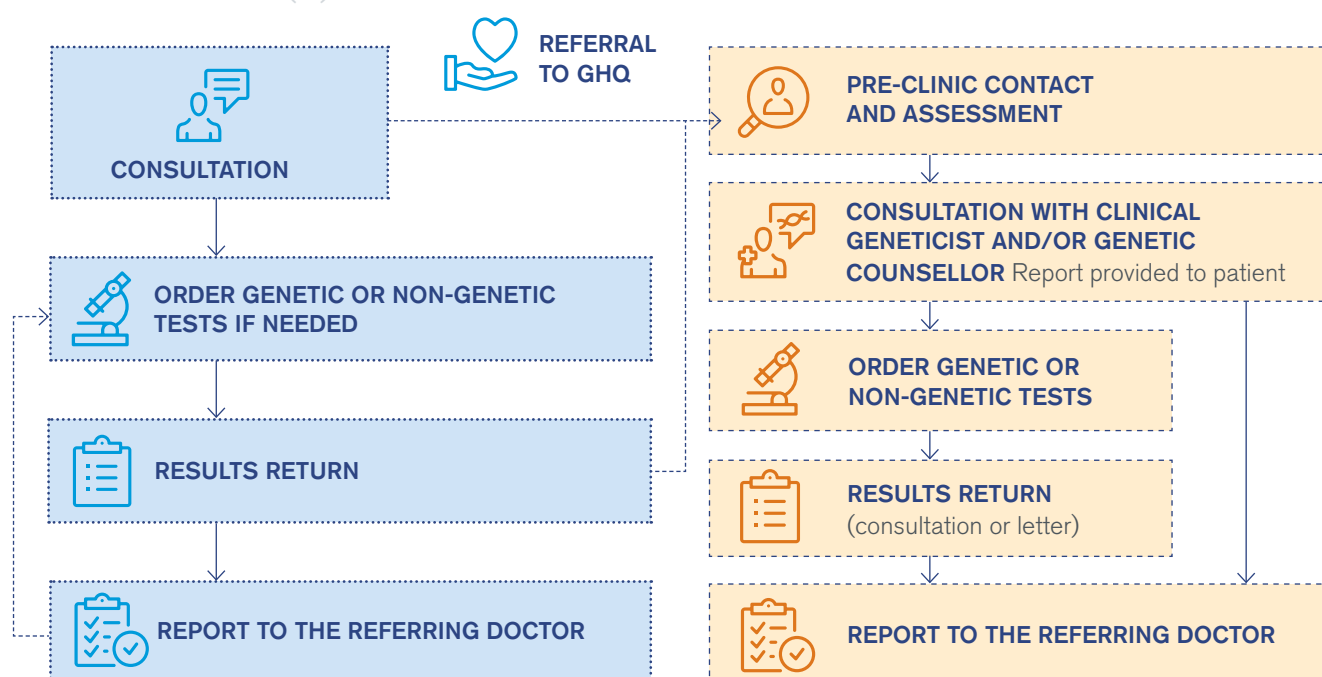
Some specialists with experience in genetic conditions related to their field may order genetic tests or diagnose genetic conditions.

## WHAT IS THE PATH TO RECEIVING CARE?

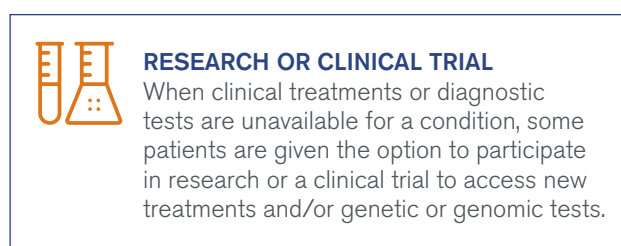
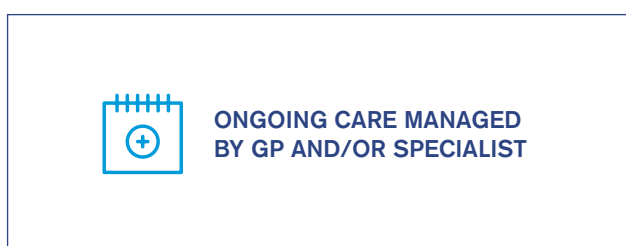
### REFERRAL



### CONSULTATION(S)



### ONGOING CARE



Genetic or genomic testing may or may not result in a patient's diagnosis. As our understanding of genetic disease improves, some test results can be reanalysed or reassessed. Reanalysis can result in a change to the patient's diagnosis. Your genetic specialist, mainstream specialist or GHQ may recommend you recontact them over time to discuss whether retesting or reanalysis is appropriate for you.



# Your guide to genetic and genomic tests

Genetics and genomics are rapidly advancing fields with new terms and different tests emerging regularly.

This fact sheet is your guide to understanding these terms and what some of the many different genetic and genomic tests do.

If you are seeking diagnosis and treatment, your clinical geneticist or relevant medical specialist will review the available information and recommend the best test for you.

## Key terms

### **Chromosome**

The structures in cells where your DNA is packaged.

### **DNA**

The chemical structure that makes up your genetic material.

### **Exome**

All the genes in a genome are known as the exome.

### **Gene**

The part of your genetic material that controls how your traits are developed. Humans have approximately 20,000 genes in their genome.

### **Genome**

All of your genetic material, including genes and the DNA between genes.

### **Mutations**

These are the differences in a gene that cause people to have different traits. These can be typical traits like eye colour or detrimental traits that can cause diseases.

## What are some of the test options?

### **Chromosomal tests**

*Karyotype or chromosomal microarray analysis*

Analysis of the chromosomes: These tests check the number of chromosomes or if any chromosome has extra or missing pieces. They may also identify large structural rearrangements.

### **Condition specific**

*Single gene test or specific multi-gene test*

These tests check one or more genes for mutations that cause specific diseases like cystic fibrosis.

### **Symptom-specific test**

*Panel test or broad multi-gene test*

These tests look for mutations in many genes which are associated with similar symptoms or syndromes—for example, a hereditary cancer panel, hereditary cardiac panel or infant epilepsy panel.

### **Whole exome sequencing (WES) and whole genome sequencing (WGS)**

Both tests collect data from nearly all genes. Analysis often focuses on a panel of genes that is associated with the condition of interest.

WES is more cost-efficient and focuses on the most informative parts of the genome.

WGS looks at all parts of the genome. With WGS, it's possible to find out if a genome has extra or missing pieces.

## Who needs to provide a sample?

If you choose to go ahead with genetic testing you will need to supply a sample of your DNA - this will be in the form of a saliva or blood sample.

A **singleton test** only requires a sample from the patient.

A **trio test** requires samples from the patient and their biological parents.

A **couple test** requires samples from the two people who are planning to have a child.

A **paired test** is for cancer patients only and requires a sample of a patient's cancer and normal tissue.

## Test purposes

A **diagnostic test** is used to determine the cause of a patient's condition.

A **predictive test** is for a person with no signs or symptoms of a genetic condition. The test predicts whether this person is likely to develop a disease in the future. Generally, this is only possible when a gene change that caused a condition has already been identified in another family member. For ethical reasons, predictive testing for adult-onset conditions is only offered to adult family members.

**Prenatal testing** is for an unborn child or the mother of an unborn child and is used to determine if the child has a genetic condition.

**Pre-implantation genetic testing** is for an embryo prior to implantation. The test is used to check if the embryo has a genetic variation association with a genetic condition.

**Carrier screening** is for couples who are planning on becoming pregnant. The screening is used to determine the couple's risk of having a child with a known genetic condition.

### Key takeaways



**Chromosomal tests** are used to diagnose a condition and generally only require samples from the patient. In some cases testing of parents is also required.



**Condition specific or symptom specific tests** are used to diagnose a patient or an unborn child, predict a patient's risk of developing a disease, or predict the risk of having a child with disease. These tests require samples from the patient, and in some cases their parents.



**WES or WGS are tests** which can look at many genes at once. These tests require samples from the patient, and in some cases their parents.

# Clinical Geneticists in Queensland (public and private)



Clinical geneticists are doctors who specialise in diagnosing and providing advice to patients with inherited, or genetic health conditions. There are a number of clinical geneticists working in Queensland through the public and the private health systems.

NAME	ORGANISATION	PROVIDER	CONTACT DETAILS	SPECIALISATION	SERVICE AREA	REFERRAL REQUIRED FROM GP
Various Clinical Geneticists	Genetic Health Queensland	Public	<ul style="list-style-type: none"> <li>📞 (07) 3646 1686</li> <li>✉️ GHQ@health.qld.gov.au</li> <li>📍 health.qld.gov.au/ghq</li> </ul>	<ul style="list-style-type: none"> <li>▪ Paediatric &amp; Adult clinical genetics</li> <li>▪ Cancer</li> <li>▪ Prenatal</li> </ul>	Clinics in Brisbane and major regional centres. Telehealth appointments available.	Yes
Professor David Coman		Private	<ul style="list-style-type: none"> <li>📞 (07) 3832 9876</li> <li>✉️ reception@drdavidcoman.com.au</li> <li>📍 drdavidcoman.com.au</li> </ul>	<ul style="list-style-type: none"> <li>▪ Paediatric &amp; Adult clinical genetics</li> <li>▪ Metabolic</li> <li>▪ Prenatal</li> <li>▪ Cancer</li> </ul>	Brisbane. Telehealth appointments available for patients outside Brisbane.	Yes
Dr Di Milnes	Genes Australia	Private	<ul style="list-style-type: none"> <li>📞 (07) 3088 6983</li> <li>✉️ admin@genesaustralia.com.au</li> <li>📍 genesaustralia.com.au</li> </ul>	<ul style="list-style-type: none"> <li>▪ Paediatric &amp; Adult clinical genetics</li> <li>▪ Cancer</li> <li>▪ Prenatal</li> </ul>	Brisbane Telehealth appointments available for patients outside Brisbane.	Varies
Dr Michael Gattas	Brisbane Genetics	Private	<ul style="list-style-type: none"> <li>📞 (07) 3217 8244</li> <li>✉️ email@brisbanegenetics.com.au</li> <li>📍 brisbanegenetics.com.au</li> </ul>	<ul style="list-style-type: none"> <li>▪ Paediatric &amp; Adult clinical genetics</li> <li>▪ Cancer</li> </ul>	Brisbane Telehealth appointments available for patients outside Brisbane.	Yes
	Wellspring Genetics		<ul style="list-style-type: none"> <li>📞 (07) 3556 3896</li> <li>✉️ email@wellspringgenetics.com.au</li> <li>📍 wellspringgenetics.com.au</li> </ul>	<ul style="list-style-type: none"> <li>▪ Preconception</li> <li>▪ Cancer</li> </ul>		No
A/Prof Michael Gabbett	The Mendel Institute	Private	<ul style="list-style-type: none"> <li>📞 (07) 3193 7780</li> <li>✉️ admin@mendelinstitute.com.au</li> <li>📍 mendelinstitute.com.au</li> </ul>	<ul style="list-style-type: none"> <li>▪ Preconception</li> <li>▪ Paediatric &amp; Adult clinical genetics</li> <li>▪ Cancer</li> </ul>	Brisbane Telehealth appointments available for patients outside Brisbane.	Yes

NOTE: These clinicians are FRACP specialists in Clinical Genetics. Other specialists may have recognised sub-specialties in genetics but have not been included on this list. Check with your specialist to find out if genetic consultation and testing is a service they are able to provide.



To see a clinical geneticist you will usually need a referral from your GP. Please see our related fact sheet Searching for a diagnosis?

This information was produced by the Queensland Genomics Community Group for a Genomics Information Toolkit for patients. This information is correct as at May 2021. For more information or to view other fact sheets visit [queenslandgenomics.org/pct](https://queenslandgenomics.org/pct). For more information on Genetic Health Queensland visit [bit.ly/GHQld](https://bit.ly/GHQld)

# Genetics and genomics for GPs

If your patient has a rare disease or a family history they are concerned about a consultation with a genetic health specialist could help them to find out if their condition is genetic.

Genetic Health Queensland (GHQ) is Queensland Health's statewide specialist and public healthcare service for people and families with suspected or known genetic health conditions.

## More information

You can find clinical prioritisation criteria for **general genetics** and **cancer genetics** online.

Clinically indicated testing and consultation with GHQ has no cost to the patient. If you are a health professional and need more information about Queensland Health's genetic health services and patient referral process visit [www.health.qld.gov.au/ghq/professionals/referral](http://www.health.qld.gov.au/ghq/professionals/referral) or get in touch.

**(07) 3646 1686**

[GHQ@health.qld.gov.au](mailto:GHQ@health.qld.gov.au)

## Links

**General Genetics** - [www.metronorth.health.qld.gov.au/specialist\\_service/refer-your-patient/genetic-health](http://www.metronorth.health.qld.gov.au/specialist_service/refer-your-patient/genetic-health)

**Cancer Genetics** - [www.metronorth.health.qld.gov.au/specialist\\_service/refer-your-patient/cancer-genetics](http://www.metronorth.health.qld.gov.au/specialist_service/refer-your-patient/cancer-genetics)



## Located in a regional area?

GHQ is a statewide service based in Brisbane with regional clinics. Where clinically appropriate, GHQ offers appointments via telehealth.

### Interested in upskilling in genetics and genomics?

Free educational resources available on the RACP website. Visit [elearning.racp.edu.au](http://elearning.racp.edu.au) and search 'Clinical Genomics for Physicians'.

Queensland Genomics and Check Up partnered to deliver GP education in genetics and genomics. Visit [queenslandgenomics/gp-resources](http://queenslandgenomics/gp-resources) for more information