

SightScore™
GLAUCOMA

Understanding your genetic risk of glaucoma.



What is Primary Open Angle Glaucoma?

Primary Open Angle Glaucoma (called 'glaucoma' in the rest of this document) is an eye condition in which loss of vision results from optic nerve damage, most often due to increased pressure within the eye. It mainly occurs when the eye's drainage canals become clogged over time and the correct amount of fluid can't drain out of the eye. Loss of vision in glaucoma is usually gradual and affects peripheral (side) vision first. There can be significant loss of vision before there is an awareness of any problem.

Glaucoma is a leading cause of irreversible vision loss worldwide. In Australia, 300,000 people have glaucoma; about 1 in 200 people have glaucoma by age 40, increasing to 1 in 8 by age 80.

Vision lost to glaucoma is unfortunately irreversible once it has occurred. However, once diagnosed, glaucoma can usually be managed successfully with eye drops, laser therapy, or surgery.

More information about glaucoma and the risk factors for glaucoma can be found at Glaucoma Australia. Visit glaucoma.org.au.



300,000
Australians
have glaucoma



Glaucoma
risk increases
with age



1 in 8
people have
glaucoma by
age **80**¹



1 in 8 glaucoma
suspects develop
glaucoma within
2 years³



Glaucoma has a
strong **genetic**
component



1 in 4 first degree
relatives* of glaucoma
patients will develop
glaucoma in their
lifetime

*parents, siblings and children

1. Mitchell P, Smith W, Attebo K, Healey PR. Prevalence of open-angle glaucoma in Australia. The Blue Mountains Eye Study. Ophthalmology. 1996 Oct;103(10):1661-9.

2. Keel S, Xie J, Foreman J, et al. Prevalence of glaucoma in the Australian National Eye Health Survey. Br J Ophthalmol. 2019; 103(2):191-5

3. Miki A, Medeiros FA, Weinreb RN, et al. Rates of retinal nerve fiber layer thinning in glaucoma suspect eyes. 2014; 121(7):1350-8

What are the main risk factors for glaucoma?

The risk that a person will develop glaucoma in the future, or that already-diagnosed glaucoma will get worse, depends on a wide range of factors. These include:

- Risk factors such as an age over 50, a family history of glaucoma, or being of African descent.
- Clinical features such as high eye pressure, certain types of vision loss, or signs of optic nerve damage.
- Variations in genes that influence the risk of glaucoma.

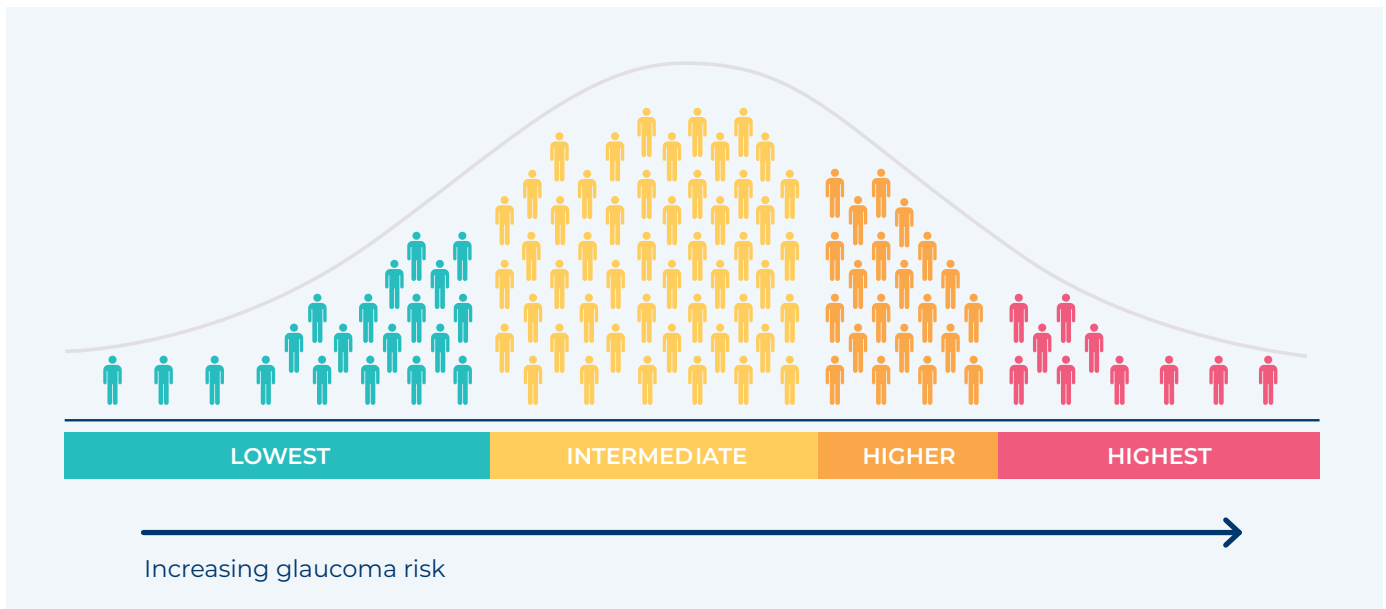
What is a Polygenic Risk Score?

Humans share the same genetic code apart from some differences, called genetic variants, that make each of us unique.

Some of these variants are known to increase the risk of developing certain health conditions, while others reduce the risk.

It is possible to combine the risks from hundreds or thousands of these genetic variants to estimate the overall risk of a person developing a particular health condition, such as glaucoma. This overall risk is known as a polygenic risk score. In some cases, a polygenic risk score may also be linked to the potential severity of the health condition and the way it might develop over time.





What is SightScore?

SightScore is a saliva test that looks at thousands of genetic variants in your genome to create a personalised polygenic risk score for glaucoma. It estimates your genetic risk of developing glaucoma in the future or, if you already have glaucoma, the risk your glaucoma might get worse, relative to others in the community with similar ancestry.

The spread of possible SightScore results is shown as a 'bell curve', with most people near the middle, and fewer people at the upper and lower risk ends.

Some people's SightScore will fall into the lowest risk group (■ green in the picture) meaning they have a lower risk than most others in the community. Others will fall into the intermediate risk group (■ yellow) meaning they have a fairly normal risk, while others will fall into the higher (■ orange) or highest (■ red) risk groups, meaning they have a higher risk than most others.

Who is SightScore for?

- ✓ Adults who do not have evidence of glaucoma currently.
- ✓ Blood relatives of people with glaucoma, who do not have evidence of glaucoma currently.
- ✓ People who have suspicious, but not definitive, features of glaucoma (so-called "glaucoma suspects") and those with high eye pressure.
- ✓ People diagnosed with glaucoma.



How might my healthcare practitioner use SightScore?

Depending on your medical history and eye examination, your healthcare practitioner may use SightScore to consider:

- When you should first be checked for glaucoma.
- How often you should be checked for glaucoma.
- Whether to refer you to a more specialised healthcare practitioner for monitoring.
- How to manage your care if you have glaucoma, including some treatment decisions.
- Whether your close blood relatives (e.g. parents, siblings, adult children) should be checked for glaucoma.

SightScore may also be used to help you understand your personal genetic risk so you can be proactive in your healthcare in regard to any recommendations about follow-up appointments and lifestyle changes that might be worthwhile.

Can SightScore guarantee I will or will not develop glaucoma, or have my glaucoma get worse?

SightScore tells you about your future risk. It is not your destiny.

A high SightScore does not mean you will definitely develop glaucoma in future or, if you already have glaucoma, that it will definitely get worse. Equally, a low risk score does not rule out the possibility of developing glaucoma or that existing glaucoma will not get worse in the future.

How does SightScore relate to my family history?

Blood relatives (e.g. parents, brothers/sisters, adult children) share parts of their genetic code and will have some of the same genetic variants. Thus, for conditions with a strong genetic contribution, such as glaucoma, a healthcare practitioner will take account of a person's family history when assessing their risk.

However, a family history is not the same as a person's individual genetic risk.

It is possible to have a high genetic risk of glaucoma without a family history of the condition. It is also possible to have a family history of glaucoma and not have a high individual genetic risk. This is because a family history does not mean a person has personally inherited the genetic variants that increase the risk of glaucoma.

SightScore is a personalised genetic test which uses your own DNA to assess your individual risk of developing glaucoma.

Does SightScore test every aspect of genetic glaucoma risk?

Rarely, a person will have a single variant in a key glaucoma gene (e.g. Myocilin) that has a large impact on glaucoma risk. SightScore may identify some of these rare variants. A healthcare practitioner may order separate testing of these variants in the rare cases it is warranted based on family history and other clinical considerations.

What is the SightScore process?

STEP 1



Talk to your healthcare practitioner about SightScore. If appropriate, they will complete a test referral form.

STEP 2



Order and pay for SightScore test. You can do this on our website or in some cases, your healthcare practitioner may be able to provide a service that includes the test.

STEP 3



Collect your saliva sample and return to us.

STEP 4



Your DNA will be tested and a SightScore generated.

STEP 5



Most results will be available within 2-3 weeks from the time the lab receives your sample. If you were referred by a healthcare practitioner, your results will be returned to them. If you referred through our family service, your report will be returned to you.

Key things to know

- SightScore is a genetic risk test. It does not diagnose glaucoma.
- SightScore only estimates your risk of developing primary open angle glaucoma and not other forms of glaucoma or other conditions.
- A high risk result does not mean that you will definitely develop glaucoma in the future or, if you already have glaucoma, that it will definitely worsen. Equally, a low risk score does rule out ever developing glaucoma or glaucoma worsening in the future.
- SightScore tests for polygenic risk. SightScore may also test for rare single gene variants. It may not take into account clinical features and risk factors like eye pressure, age, or environment.
- Although SightScore has been validated in a range of ancestries, it has been most extensively validated in populations with European ancestry (like most current polygenic risk scores). Results may be less accurate for people with non-European ancestry.
- In a small number of cases, we will be unable to provide you a SightScore, even after you have provided another sample(s) for retesting. You will be refunded if this happens.
- SightScore is not covered by Medicare or private health insurance. Information about the cost of SightScore is available from seonixbio.com.
- Your DNA does not change, so your SightScore will be relevant for your whole life. However, the meaning of your SightScore may change over time depending on your glaucoma status and other clinical features. Improved versions of SightScore may also become available in future as researchers learn more about glaucoma.

Additional resources:

Glaucoma Australia:

Glaucoma Australia provides sight saving education, support and advocacy to people with glaucoma and their families.

Visit glaucoma.org.au
or call 1800 500 880.

Learn more
about the
SightScore test:



seonixbio.com

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This pamphlet is intended to provide you with general information. It is not a substitute for advice from your healthcare practitioner. If you decide to take the SightScore test, your healthcare practitioner will ask you to sign a consent form. Read it carefully and if you have any questions about the test or consent form, ask your healthcare practitioner.