Monash IVF
Leading the future of reproductive care

Genetic Carrier Screening Test

Genetic carrier screening is available to everyone.
You do not need to be an IVF patient to use Monash IVF's genetic carrier screening test.

What is genetic carrier screening?

Genetic carrier screening is a test that assesses your chance of having a child with a recessive or X-linked genetic condition.

Recessive genetic conditions occur when each reproductive parent is a carrier of a genetic change which is passed on to their child, or a woman carries a genetic change on the X chromosome which is passed on to a male child (XY). Some examples of single gene conditions include Cystic fibrosis, Fragile X syndrome, Spinal muscular atrophy, Thalassaemia and Sickle Cell Disease.

Having this screening completed will provide you with actionable results and peace of mind, allowing you to be better prepared and explore all your reproductive options with the guidance of your doctor.

Who should consider genetic carrier screening?

The Royal Australian and New Zealand College of Obstetricians and Gynaecologists recommends that all individuals planning a pregnancy or in early pregnancy consider genetic carrier screening.

Although genetic carrier screening can be done at any time, it is recommended that you have this test before you are pregnant. If you are already pregnant, you can still have this testing in addition to other screening tests during pregnancy, such as non-invasive prenatal testing (NIPT).

Your carrier status doesn't change over time, so the screening only needs to be performed once per person. If you change partners or donors, your new partner or donor would need to be screened, but you would not usually need to be retested.

What are the options?

There are two options for genetic carrier screening: a three gene panel screening and an expanded panel screening.

The three gene panel tests for 3 conditions: Cystic fibrosis, Fragile X syndrome and Spinal muscular atrophy. About 1 in 240[1] couples find out they have a high chance of having a child with one of these three conditions.

The expanded panel tests for over 400 conditions, including the 3 conditions listed above as well as hundreds of other conditions that are equally serious, such as Thalassaemia, Duchenne muscular dystrophy (DMD), Haemophilia and Sickle Cell Disease. About 1 in 50[2] couples find out they have a high chance of having a child with one of the 400+ genetic conditions tested.

Monash IVF strongly recommend patients consider the expanded panel, as it offers significant additional information and insight into genetic conditions that are collectively more common than the three conditions screened in the three gene panel.

1. Archibald, A. D., Smith, M. J., Burgess, T., Scarff, K. L., Elliott, J., Hunt, C. E., ... Amor, D. J. (2018). Reproductive genetic carrier screening for cystic fibrosis, fragile X syndrome, and spinal muscular atrophy in Australia: Outcomes of 12,000 tests. Genetics in Medicine, 20(5), 513-523. doi:10.1038/gim.2017.134.
2. Mackenzie's Mission (2022), Outcomes, 2 July 2024, www.mackenziesmission.org.au

Genetic Carrier Screening Options	Three Gene	Expanded
Conditions included in screen	3	400+
Patient OOP (for Medicare eligible individuals)	N/A (bulk billed)	$695/individual $990/couple
Number of couples who screen positive	Approx. 1 in 240[1]	Approx. 1 in 50[2]
At-home saliva or in-clinic blood test options	✓	✓
Support from an in-house, qualified Genetic Counsellor	✓	✓
Support from reproductive experts in the event of a high-risk result	✓	✓
Tests for top 3 conditions among Caucasian Australians	✓	✓
Tests for top 10 conditions among Caucasian Australians		✓
Tests for top 3 conditions among other ethnic groups		✓
Customised panels possible where required (e.g., for donor matching)		✓

How to access Genetic Carrier Screening

Monash IVF provides genetic carrier screening for everyone; not just IVF patients.

There are two ways of accessing this test:

Option 1

At-home saliva kit

1. Order the screening kit online at monashivf.com/services/genetic-testing/genetic-carrier-screening-kit/

2. Receive an email from the Monash IVF genetic counselling team with the details of your test and a consent form.

3. If you are pregnant, you will receive a phone call advising you that a blood test is preferred, and we will email you a pathology form. **Otherwise**, look out for the at-home genetic carrier screening test in the mail.

4. Provide a saliva sample by following the instructions in the kit.

5. Mail your sample back using the reply-paid envelope in your kit or drop it off at an eligible clinic.

6. Receive your results and supporting information via email or phone from our genetic counselling team (usually within 4-6 weeks).

Option 2

Monash IVF clinic for a blood test

1. Contact your local clinic for an appt.
For a list of clinic locations visit: monashivf.com/services/genetic-testing/genetic-carrier-screening-kit/

2. Attend your clinic at the pre-organised appointment time.

3. Complete paperwork. You will be required to complete relevant paperwork and pay (if needed) for your blood test when you arrive at the clinic.

4. Blood test. A nurse will meet you for your blood test.

5. Receive your results and supporting information via email or phone from our genetic counselling team (usually within 4-6 weeks).

I don't have a family history of genetic conditions. Do I still need the screening?

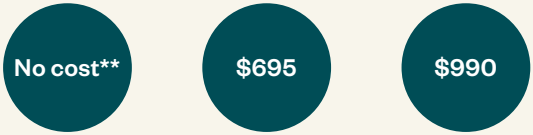
The majority of children with single gene conditions are born into families with no other affected family members, and about 1 in 50[2] reproductive couples who have genetic carrier screening will find out they have an increased chance of having a child with a single gene condition.

How much does the screening cost?

The three gene genetic carrier screening test has no out of pocket costs for individuals eligible for Medicare rebates. To access this rebate you will need a referral from your doctor.

The expanded genetic carrier screening test costs $695 for an individual or $990 for a couple.* While there is no Medicare rebate available for this expanded test, Monash IVF strongly recommend this option as it offers significant additional information and insight into genetic conditions that are collectively more common than the three conditions screened in the three gene panel.

Both tests are available via a saliva kit with detailed instructions or via a blood test taken in the clinic. All of our tests include support from the genetic counselling team.



Three gene panel

Expanded panel - individual

Expanded panel - couple

***Pricing is correct as at 1 July 2024. Please check the Monash IVF website for up-to-date pricing.**

**** for Medicare eligible individuals**

What if the results say we are at-risk?

If you are one of the reproductive couples with an increased chance of having a child with a single gene condition, our experienced genetic counselling team will talk to you about any genetic implications and if needed, refer you to a specialist who can help you to understand your reproductive options. These options may include testing in pregnancy, testing after the birth of a child, considering IVF with a gamete donor or considering IVF with preimplantation genetic testing of embryos. The Monash IVF team are here to support you, whichever option you choose.

If genetic carrier screening does identify you as having an increased chance of having a child with a single gene condition, you may be eligible for Medicare funding for IVF treatment and embryo testing.

Why Monash IVF?

Monash IVF have been providing leading reproductive care for 50 years. Our team is comprised of experienced fertility specialists, clinical geneticists, counsellors, scientists, obstetricians, gynaecologists, ultrasound technicians, donor and surrogacy experts and nursing teams, all of whom are passionate about supporting people throughout their entire fertility journey.

Importantly, this means that our expertise doesn't end with genetics. Rather, we can offer holistic, integrated care from genetic testing to counselling, all the way through to fertility treatment options and even ultrasound services. So we're with you every step of the way. Learn more about us and the services we provide at **monashivf.com.**

More information

For more information or to order a genetic carrier screening kit, scan the QR code below or visit **monashivf.com/services/genetic-testing/genetic-carrier-screening-kit/**

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