Monash IVF

Genetic Carrier Screening
Fact Sheet

So, you're thinking about having carrier screening done. What does that involve?

What does it mean to be a genetic carrier?

Everyone has two copies of each gene. One passed down from their biological mother and one from their biological father. An individual is a genetic carrier when they have inherited one copy of a gene change associated with a specific genetic condition from one of their parents. One altered copy of a gene is usually not sufficient to cause any health concerns in the carrier as they still have the second functional copy. However, if both biological parents are carriers, there is an increased chance that they could have a child who is affected with a genetic condition.

What is genetic carrier screening?

Genetic carrier screening is a test that assesses your chance of having a child with a single gene condition. Having this screening completed will provide you with actionable results, allowing you to be better prepared and explore all your reproductive options with the guidance of our genetic counselling team.

Globally, genetic disease is one of the leading causes of childhood mortality. 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. Some examples of single gene conditions include Cystic fibrosis, Fragile X syndrome, Spinal muscular atrophy, Thalassaemia and Sickle Cell Disease.

What conditions are being screened for?

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 200 couples find out they have a high chance of passing on one of these conditions to their child.

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 30 couples find out they have a high chance of passing on one of the 400+ conditions to their child.

Which test option is right for me?

Monash IVF strongly recommend that patients consider the expanded panel, as it offers significant additional information and insight about serious genetic conditions that may be passed onto children.

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 200	Approx. 1 in 30
At-home saliva or in-clinic blood test options	✓	✓
Support from an in-house, qualified Genetic Counsellor	✓	✓
Support from fertility experts in case of 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		✓

How do I order the test?

Monash IVF now provides at-home genetic carrier screening. The process is simple:

1. Order the screening kit online at **https://www.monashivf.com/services/genetic-testing/carrier-screening/**
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. When the kit arrives, follow the instructions to provide a saliva sample.
5. Mail your sample back using the reply-paid envelope in your kit or drop it off at an eligible clinic. See our list of eligible clinics at www.monashivf.com/services/genetic-testing/gcsk-instructions
6. Receive your results and supporting information via email or phone from our genetic counselling team (usually within 4-6 weeks).

Who should consider carrier screening?

The Royal Australian 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).

Carrier testing only needs to be performed once, rather than every time you have a child. If you change partners, your partner would need to be screened, but you would not need to be retested.

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 your reproductive options and help you decide what may be best for your individual situation. 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 screening.

How much does the screening cost?

The three gene genetic carrier screening test has no out of pocket costs thanks to the Medicare rebate. 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 test, Monash IVF strongly recommend this option as it offers significant additional information and insight about serious genetic conditions that may be passed onto children.

All tests include saliva testing kits, detailed instructions and support from our genetic counselling team.

***Pricing is correct as at 1 November 2023. Please check the Monash IVF website for up-to-date pricing.**

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 30 reproductive couples who have carrier screening will find out they have an increased chance of having a child with a single gene condition.

Why Monash IVF?

Monash IVF have been providing leading reproductive care for 50 years. Our team are experts in their fields, passionate about supporting people throughout their fertility journey. Our patients have access to our experienced genetic counsellors, scientists, obstetricians, gynaecologists, fertility specialists and support teams.

With clinics all over Australia, if you screen positive as a carrier, you're able to receive in-person help and support. If there are any other stumbles along your road to parenthood, we're here to help too. Learn more about us at MonashIVF.com

More Information

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