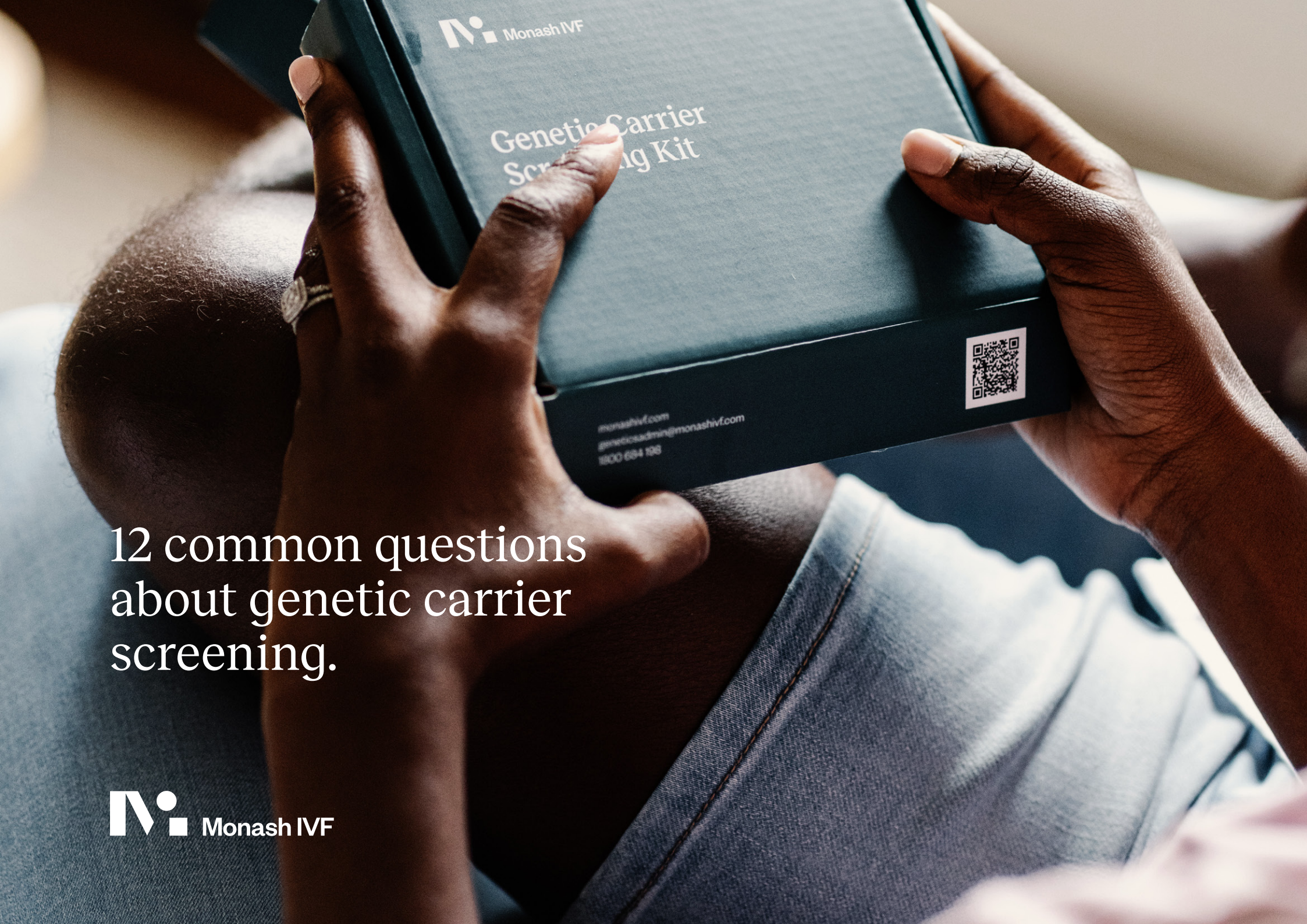


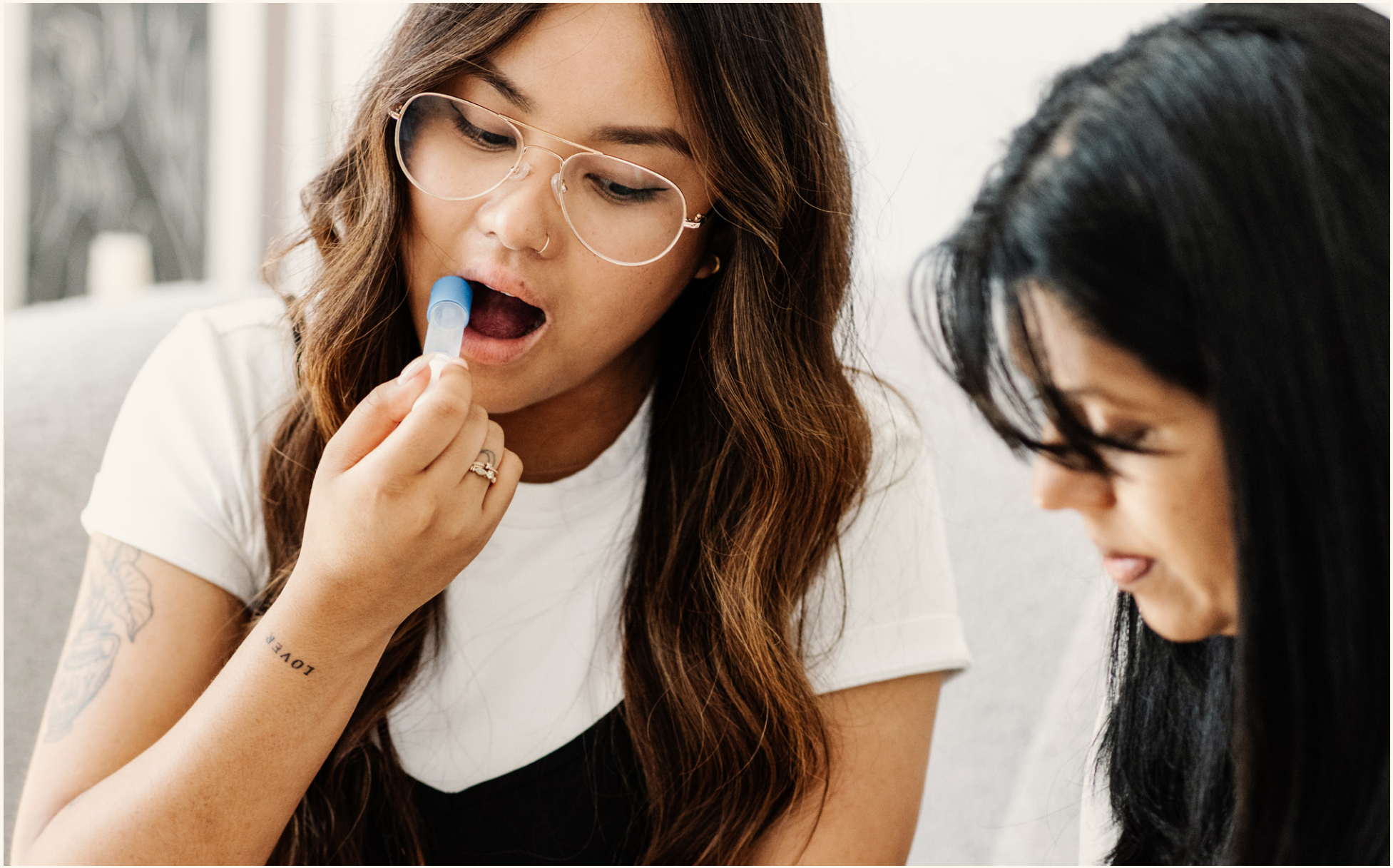
12 common questions about genetic carrier screening.

Monash IVF

Genetic Carrier Screening is a test that assesses your chance of having a child with a single gene condition like Cystic Fibrosis or Fragile X Syndrome.

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

Have questions about it? You're not alone. Here are some of the most common queries.


LOVER

01

When is the best time to have a genetic carrier genetic screening test?

The earlier the testing, the better – and for both parents. Ideally, genetic carrier screening is performed before pregnancy, because the information may guide care during pregnancy or at the time of delivery (where testing can be performed if the child is symptomatic).

Genetic carrier screening can take 4 to 6 weeks to return results. For pregnant patients we routinely recommend using a blood sample instead of a saliva one (so that we don't need to allow time for posting at-home test kits back and forth) and screening both biological parents at the same time.

This way, any follow up testing in pregnancy can be organised as soon as possible. If screening is done around the time of a first dating scan (at 6-9 weeks) then results will be available to allow testing by CVS, and if ordered at the same time as chromosome screening will allow testing by amniocentesis later in pregnancy.

02

Who should have the test?

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.

Importantly, both the egg provider and the sperm provider need to be screened, because the test compares the genetic material of both reproductive parents to determine the chance those two people have of passing on a single gene condition to any child(ren).

If you are planning to have a baby with your partner or planning to use a known donor (someone you know personally), the couple test is recommended. This will help you to understand you and your partner or donor's combined genetic information, so you can make informed decisions about your options for conceiving. If you are planning to use a clinic-recruited donor or an international donor, the single test is recommended. Clinic-recruited or international donors have usually already been screened for single gene conditions.

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.

03

How common is it to have a child with a single gene condition?

The chance of having a child with a single gene condition is very similar to the chance of a having a child with other health conditions we commonly screen for during pregnancy.

Most of the population will have some form of screening for chromosome conditions during pregnancy (whether it's first trimester screening, non-invasive prenatal testing or both) and will have ultrasound screening in pregnancy for anatomical changes and fetal wellbeing.

Although individual single gene conditions are quite rare, collectively they're common and affect as many children or pregnancies as chromosome conditions do.

04

Which genetic conditions should a genetic carrier screening test for?

Genetic carrier screening is often offered via two options: a limited screening panel and an expanded screening panel.

The limited screening panel screens for three common genetic conditions in the Australian population: cystic fibrosis, Fragile X and spinal muscular atrophy. However, testing for three genes is just scratching the surface when it comes to carrier screening. Using the same technology, you can screen for hundreds of conditions, providing greater valuable information about your reproductive health.

Of couples who have an increased chance of having a child with a single gene condition, only a maximum of 20% would be identified using the three-gene test, leaving 80% with false reassurance. For this reason, Monash IVF offer an expanded panel to ensure that the carrier screening is as comprehensive as possible and test for conditions which are known to be common in different populations.

You can see the full list of genetic conditions the expanded panel tests for here.

05

Will the results of the test tell me anything about my own health, or the risk of me developing any conditions in the future?

The chance of finding out something about your own health through genetic carrier screening is very low.

Some people worry that by undergoing genetic carrier screening they might find out something that has implications for their own health, but this is extremely unlikely. Most of the genes tested only have an effect if both copies of the genetic information have an error (autosomal recessive conditions) or the error is in the genetic information on the X chromosome in a male child (X linked recessive conditions). As such, most of the genes tested have no health implications for the parent.

There is a small subset of genes that can have health implications for parents, but these are very rare. An example of one such condition is Duchenne muscular dystrophy (DMD). Female carriers of DMD can have a cardiomyopathy in addition to a chance of having a male child with Duchenne muscular dystrophy. Any such findings are always explained by our genetic counselling team and recommendations for additional healthcare are given by our medical team.

06

Can I avoid finding anything out about my own health by choosing a limited screening panel?

No. The chance of finding out something about your own health isn't avoided by choosing a limited screening panel. The three genes in the limited screening panel (cystic fibrosis, Fragile X and spinal muscular atrophy), if found, can also have future health implications.

For instance, being a carrier of fragile X can lead to the onset of menopause before the age of 40 (premature ovarian insufficiency) and an increased chance of having a tremor later in life.

Cystic fibrosis carriers are usually well but some variants are associated with an increased risk of pancreatitis, and male carriers of two variants in the CFTR gene may have azoospermia (no sperm in the ejaculate).

We recommend speaking to our genetic counselling team if you're worried about discovering things out about your own health.

07

Can’t I just rely on family history?

No. Roughly 80% of children with a single gene condition are born into a family where there is no family history of the condition. If we screened using family history alone, we would not identify these families until after they have had a pregnancy or child affected by a single gene condition. That’s why it’s important to screen during pregnancy or (preferably) before.

Family history is still an important part of a preconception health check, and we encourage you to speak to your doctor about any significant family history of a genetic condition so that individual advice can be provided.

08

I already know a genetic condition is present. Do I still need to proceed with genetic carrier screening?

Yes. The screening is still recommended even when a known genetic condition is present. It's recommended that everyone have genetic carrier screening done regardless of existing conditions, because you, your partner or your donor may carry other conditions as well. Most of the time, it is possible for us to design a test to screen for specific known conditions in the other reproductive partner.

09

Can I have the screening done during pregnancy alongside other tests such as NIPT?

Sometimes the two screening tests get mentioned together, but they are designed to look for very different conditions. Non-invasive prenatal testing (NIPT) and other chromosome screening tests will look for extra or missing copies or parts of chromosomes, which generally occur in the formation of an egg or sperm (and therefore can happen in each pregnancy).

Genetic carrier screening looks for small spelling errors in genetic information, which are inherited from each biological parent. Unlike NIPT, genetic carrier screening does not need to be repeated for each new pregnancy.

10

How can I organise the test? Do I need to see my doctor?

Genetic carrier screening can be ordered by anyone, preferably prior to, or in early pregnancy. With Monash IVF, you can even complete the test in your own home after ordering it on our website.

Our team of healthcare professionals is led by Dr. Tristan Hardy and Dr. Melody Menezes, and they will guide you through the process and any follow up care. If you would like your family doctor or a specialist to be involved, we can include them in the process.

11

Is the cost of the test covered by Medicare or by private health insurance?

Currently, there is no Medicare rebate or private health insurance support for carrier screening, meaning that the cost of this process is all out of pocket.

However, 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.

The Morrison Government has promised to cover the cost of carrier screening for the three most common inherited conditions in Australia – cystic fibrosis, spinal muscular atrophy and fragile X syndrome – if it is re-elected.

We'll know more soon, but even if the promise is fulfilled, the Medicare subsidy won't come into effect until 2023.

12

What happens if the test reveals a high chance of having a child with a genetic condition?

If you are one of the 1 in 20 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, having IVF and testing any embryos produced before having them implanted, or considering IVF with a sperm, egg or embryo donor. The Monash IVF team are here to support you, whichever option you choose.

With you every step of the way.

Remember, we are here to help. Monash IVF have been providing leading reproductive care for over 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. We are passionate about providing the best care in reproductive genetics and happy to help with any questions: just reach out and ask us.

Phone: 1800 674 792 (free call)
Email: enquiries@monashivf.com


Monash IVF genetic carrier screening tests can be ordered on the website: https://www.monashivf.com/services/genetic-testing/carrier-screening/