

New to genetic carrier screening?

You're in the right place to learn why it's so important if you're planning to conceive. The good news is that genetic carrier screening for the three most prevalent conditions, cystic fibrosis (CF), spinal muscular atrophy (SMA), and fragile X syndrome (FXS), is now Bulk-Billed.*

What is genetic carrier screening?

It's a simple blood test that determines your carrier status (risk of having a child with a genetic condition) for the tested conditions.

I have no family history. Do I still need to be tested?

As 90% of carriers for genetic conditions do not have a family history, Australian clinical guidelines recommend offering testing to every woman or couple who are either planning or in the first stage of pregnancy, regardless of their risk factors.

Which genetic conditions are screened for?

Clinical Labs' Gene Access Carrier Screening test screens for the three most common genetic conditions: cystic fibrosis (CF), spinal muscular atrophy (SMA), and fragile X syndrome (FXS). This test is now Bulk-Billed.*



Cystic fibrosis (CF)

A severe genetic condition that causes lung and gastrointestinal problems, affecting about 1 in 2,500 people. Approximately one in 25 people are carriers of CF.

Spinal muscular atrophy (SMA)

An inherited neuromuscular disease historically associated with high morbidity and mortality, affecting about 1 in 6,000 people. Approximately 1 in 35 people are carriers of SMA.

Fragile X syndrome (FXS)

The most common inherited form of intellectual disability, affecting approximately 1 in 3,600 men and 1 in 6,000 women. FXS carrier screening is recommended for females, as it is inherited in a different way to CF and SMA.

Additional screening options:

Clinical Labs also offers Comprehensive Carrier Screening, evaluating an individual's carrier status for over 100 inherited conditions, and Ashkenazi Jewish Carrier Screening for eight genetic conditions most common in individuals of Ashkenazi Jewish ancestry. For more information, visit antenatal.clinicallabs.com.au/patient/carrier-screening/expanded.

Is genetic carrier screening recommended?

Australian clinical guidelines (RANZCOG & RACGP)^{1,2} recommend that every woman or couple who are either planning or in the first stage of pregnancy are offered genetic carrier screening for common genetic conditions, such as cystic fibrosis, spinal muscular atrophy, and fragile X syndrome, regardless of their probability of having these conditions.

When should I be tested?

Ideally, screening is performed before pregnancy to offer greater reproductive choice, such as pre-implantation genetic diagnosis through IVF, using donor eggs (or donor sperm for CF and SMA), donor embryos, or adoption.

What happens if I'm a carrier?

If you are found to be a carrier for CF or SMA, your partner can then be tested for that specific condition to determine your risk as a couple of having a child with CF or SMA. The test for each condition is covered once in an individual's lifetime.

Positive cases are offered one genetic counselling session per couple at no cost, upon your doctor's request.



Are blood tests preferred over saliva testing for genetic carrier screening?

Yes, blood samples are preferred, as blood typically contains more DNA than saliva. DNA extraction results demonstrate that blood samples may yield a greater quantity of higher-quality DNA compared to saliva samples. The amount and quality of extracted DNA will reduce the chances of test failures and the need for sample recollection.

This is particularly important for certain types of genetic testing, such as when testing for genetic mutations associated with reproductive genetic carrier screening.

The three genetic carrier screening tests currently offered by Clinical labs are all blood tests.

I don't have an obstetrician; can I still be tested?

Yes, GPs as well as obstetricians, can provide a referral for genetic carrier screening blood testing.

To find answers to more questions you may have about genetic carrier screening, please visit the Frequently Asked Questions page on our website:

antenatal.clinicallabs.com.au/patient/carrier-screening/faq

1. The Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG) Guidelines.
2. The Royal Australian College of General Practitioners (RACGP) Guidelines.

*Subject to Medicare eligibility criteria.