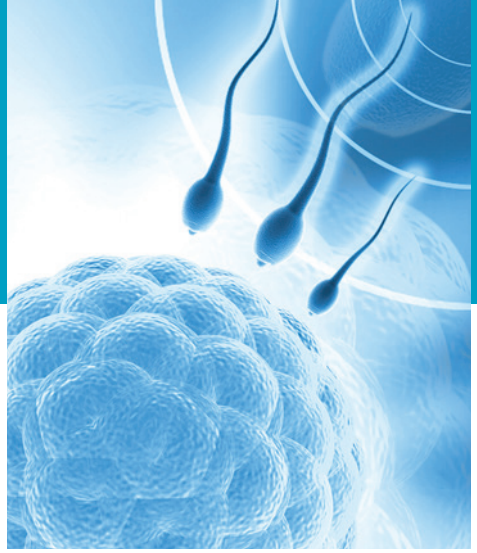
Genetic Carrier Screen

Genetic Counselling Service



Genomic Diagnostics is pleased to announce access to a genetic counselling service in conjunction with *Genetic Counselling Australia* for patients identified as carriers of CF, SMA or Fragile X syndrome using the *Genetic Carrier Screen* test. This is a high quality, free telephone service designed to enable patients to spend time talking to an expert genetic counsellor to answer any questions they may have about the implications of their test results.

1. When should a healthcare practitioner consider referring a patient to the *Genetic Carrier Screen* Genetic Counselling Service?

The *Genetic Carrier Screen* Genetic Counselling Service is designed to enable patients to discuss any questions they may have about Genomic Diagnostics *Genetic Carrier Screen* test and to further understand and explore the issues and implications of their result/s from this test.

The *Genetic Carrier Screen* Genetic Counselling Service is only open to patients who have been identified as a carrier from the *Genetic Carrier Screen* test. The doctor should refer any eligible patient that would benefit from talking to a genetic counsellor.

Please note: Patients can be referred for genetic counselling before and/or after their partner has been tested.

2. How is a patient referred to the *Genetic Carrier Screen* Genetic Counselling Service?

Once the doctor has disclosed the result to the patient/s, they can refer them to the *Genetic Carrier Screen* Genetic Counselling Service using the specific referral form provided by Genomic Diagnostics at the time of result notification.

If the referral form has been misplaced, please call Genomic Diagnostics on 1800 822 999 or email info@genomicdiagnostics.com.au. The referral form must be fully completed and faxed to the *Genetic Carrier Screen* Genetic Counselling Service number found on the referral form.

It is essential that the patient's current telephone contact details are written on the referral form.

3. When will the service be available?

The *Genetic Carrier Screen* Genetic Counselling Service will be available from 10am-4pm, Monday to Friday AEST. A Genetic Counselling Australia genetic counsellor will contact patients directly to arrange an appropriate appointment time.

4. What level of counselling will be provided to the patient?

The *Genetic Carrier Screen* Genetic Counselling Service will make every effort to answer all questions raised by patients regarding their respective test result.

5. Should my patient's partner be tested?

If an individual is identified as a carrier, their partner may need to have carrier testing. **If partner testing is required, it should be organised by the requesting healthcare professional without delay.**

6. My patient does not currently have a partner, can I still refer them to the *Genetic Carrier Screen* Genetic Counselling Service?

Yes, the *Genetic Carrier Screen* Genetic Counselling Service will be able to discuss the implications of the test result for them, their family and discuss the importance of partner testing before they have children.

7. How will the *Genetic Carrier Screen* Genetic Counselling Service be made aware of the patient's results?

Once Genetic Counselling Australia have received the patient referral form, they will contact Genomic Diagnostics customer care team and request a copy of the patient's results. This will be used as a reference during the consult. Doctors can also send a copy of the results with the referral.

8. What are the response times once a referral form has been submitted to the *Genetic Carrier Screen* Genetic Counselling Service?

Initial contact with patients will be made by a Genetic Counselling Australia genetic counsellor within 48 hours of receiving a completed referral form. The genetic counsellor will book an appointment time with the patient or if the timing is suitable, may discuss the situation immediately with the patient.

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9. What happens after a patient receives genetic counselling via the *Genetic Carrier Screen* Genetic Counselling Service?

Once the genetic counselling consultation is complete, the *Genetic Carrier Screen* Genetic Counselling Service will:

- Send the patient back to their referring healthcare practitioner to organise partner testing, for all follow-up, ongoing care and to arrange appropriate management during family planning and pregnancy.
- Fax the referring healthcare practitioner a summary of counselling recommendations. (Fax is being used to ensure efficient speed for communication.)
- Should the couple require genetic counselling following partner testing, the requesting healthcare practitioner is able to refer the patients back to the *Genetic Carrier Screen* Genetic Counselling service for further understanding and information about this result.
- If the couple are pregnant and testing shows that the couple is at risk of having a child with one of these conditions (CF, SMA or Fragile X) we recommend that the requesting healthcare practitioner refers them to a specialist genetic counselling service or prenatal diagnostic service for further management.

Please note: Genetic Counselling Australia does not arrange prenatal diagnostic testing or manage at-risk couples.

10. What happens if the patient's partner is also identified as a carrier of a condition?

If the pregnancy is at risk of Fragile X syndrome or both the patient and their partner are identified as carriers of Cystic Fibrosis (CF) or Spinal Muscular Atrophy (SMA):

- Referral for further genetic counselling should be arranged without delay
- Genetic testing for these conditions in the fetus can be done early in the pregnancy. If the tests diagnose CF, FXS or SMA, a considered choice about whether to continue or terminate the pregnancy is available.
- Family planning options should be explored, including:
  - Establishing a natural pregnancy and prenatal diagnostic testing (CVS or amniocentesis)
  - In-vitro fertilisation (IVF) and pre-implantation genetic diagnosis (PGD)
  - Adoption, or
  - Donor egg/sperm.

11. Can a patient be referred to the *Genetic Carrier Screen* Genetic Counselling Service for genetic counselling regarding other genetic conditions?

The *Genetic Carrier Screen* Genetic Counselling Service is designed to enable patients to discuss any questions and explore any issues and implications related to being identified as a carrier of CF, SMA or Fragile X syndrome from their *Genetic Carrier Screen* test. The service is not designed to offer genetic counselling regarding other genetic conditions. If your patient has a family history of a genetic condition or is concerned about a genetic condition, please consider referring them urgently to a State Based Genetic Counselling Service (www.hgsa.org.au/asgc/find-a-genetic-counsellor).

WHO ARE GENETIC COUNSELLING AUSTRALIA?

Genetic Counselling Australia is an Australian company that specialises in genetics and offer genetic counselling Australia-wide. Led by Associate Professor Leslie Sheffield, an experienced Clinical Geneticist and staffed by passionate and empathetic genetic counsellors, Genetic Counselling Australia aims to inform, educate and empower patients to assist them with understanding and making informed decisions regarding their genetic health.

A/Prof Leslie Sheffield. Clinical Geneticist MBBS, MSc, FRACP, HGSA Cert Clin Genet

Les Sheffield has worked as a clinical geneticist in the field of prenatal diagnosis and counselling since the 1980s. He has developed prenatal diagnostic programs in the area of ultrasound screening for fetal abnormalities, screening for Down syndrome and other chromosomal abnormalities and several Molecular diagnostic tests. He has led a team of clinical geneticists and genetic counsellors at the Royal Women's Hospital, Royal Children's Hospital and Monash Medical Centre, Melbourne. Currently he is the Medical Director of MyDNA Life, a genetic testing company and Genetic Counselling Australia. He is on the Editorial Board of the Pharmacogenomics Journal and is the Genetics Editor of the Internal Medical Journal.

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