Genomic Diagnostics

LEADING THE WAY TO IMPROVE HEALTH

Genetic Carrier Screening

Informed Reproductive Decision Making

FOR MEDICAL PROFESSIONALS

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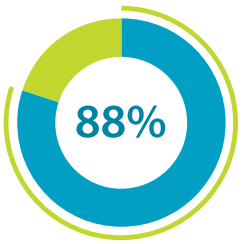
Carrier Screening – Informed Reproductive Decision Making

Genetic carrier screening is DNA-based testing to identify individuals or couples at increased risk of having children with severe inherited genetic disorders, performed in order to inform reproductive decision-making.

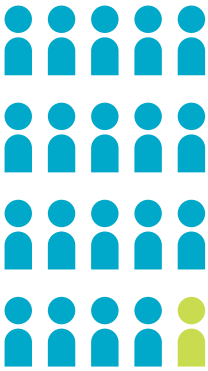
Genomic Diagnostics' Genetic Carrier Screen (GCS) tests for three of the most common inherited genetic conditions: cystic fibrosis (CF), fragile X syndrome (FXS), and spinal muscular atrophy (SMA).

One in twenty Australians will be a carrier for at least one of these conditions, however most will have no family history of disease[1]. This is due to the relatively rare nature of the conditions and their inheritance patterns, either autosomal recessive or X-linked.

Carrier screening for CF, FXS and SMA is now funded by Medicare and should be offered to those considering pregnancy or in their first trimester.



88% of carriers have no family history[1]



1 in 20 is the combined carrier frequency for these three conditions[1]

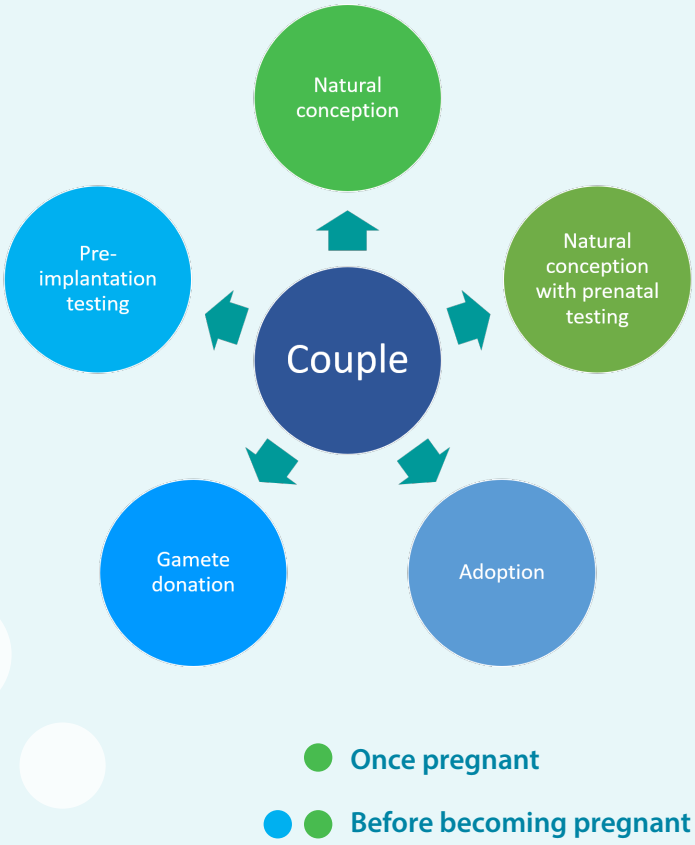
Why and when should patients be screened?

RANZCOG recommends that information on carrier screening be offered to all women planning a pregnancy or in the first trimester of pregnancy[2]. This includes screening for CF, SMA, FXS, along with thalassaemia. There is international consensus that GCS is best performed prior to pregnancy, however, carrier screening may still be offered in early pregnancy.

The purpose of genetic carrier screening is to inform reproductive decision-making.

When a carrier couple is identified (both partners are carriers for CF or SMA, or the female partner is a carrier for FXS), the couple can consider a range of reproductive options; these options become more limited and time-sensitive if carrier testing is left until post-conception.

The recommended screening pathway is to test the female partner first, followed by testing of the male partner if the female is identified as a carrier for CF or SMA.

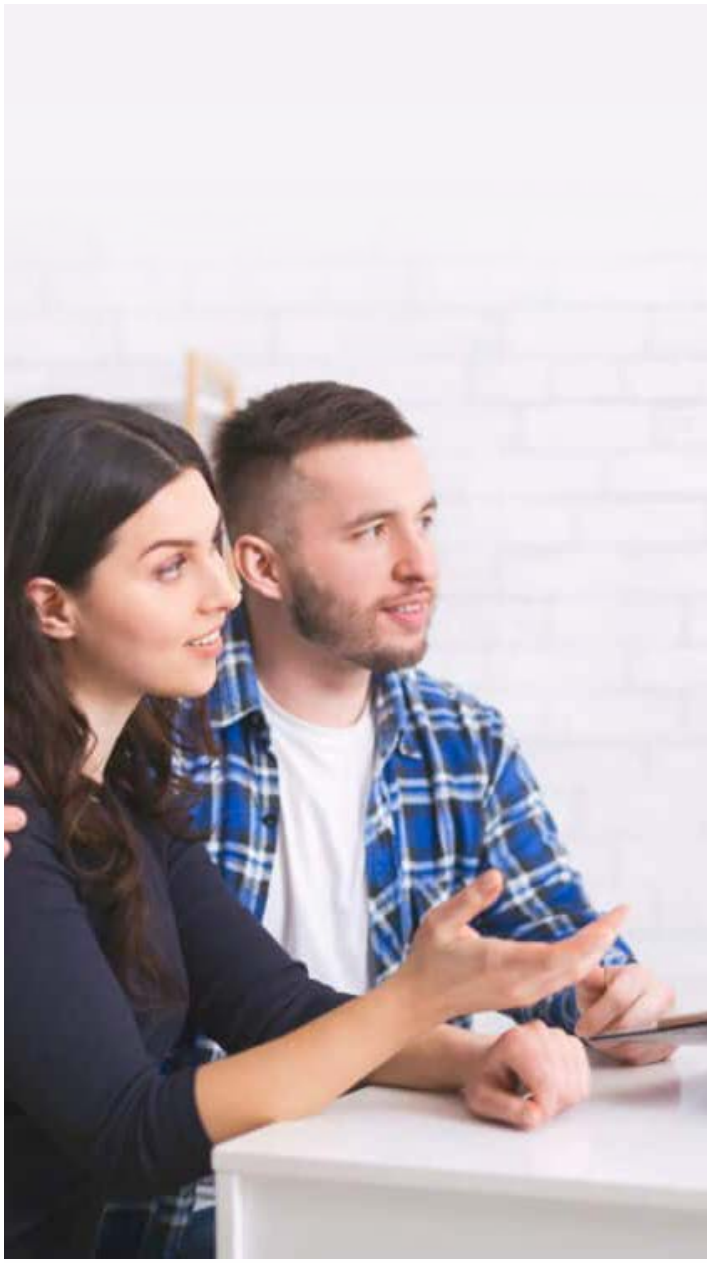


Understanding the results from GCS

GCS tests for the most common genetic changes associated with CF, SMA and FXS. The assay can detect:

- >95% of cystic fibrosis carriers
- >99% of fragile X carriers
- >95% of spinal muscular atrophy carriers

The tests used for GCS are highly accurate diagnostic tests that reliably identify carriers for these conditions. However, GCS cannot identify everyone who is a carrier for these conditions because some very rare genetic variants cannot be detected by the test. This means that even when GCS does not identify individuals or couples as carriers, there is a very small post-test or residual chance that a couple may still have a child affected with CF, SMA or FXS.



Genetic counselling

Carrier couples can be offered genetic counselling to provide more information and enable detailed discussion regarding their options and potential impacts of any decisions.

Genomic Diagnostics offers post-test genetic counselling free of charge to all carrier couples. A referral to a qualified genetic counsellor will be arranged, on request, following the confirmation of a positive carrier couple result.

Conditions Screened

CF, SMA and FXS can have devastating effects on life-expectancy and quality of life. The combined affected pregnancy rate for these disorders is equivalent to the population risk of having a child with Down syndrome.

DISORDER	CARRIER RISK[3]	PEOPLE WITH THE CONDITION[3]	TESTING APPROACH	INHERITANCE
Cystic Fibrosis Most common inherited disorder in Caucasians	1 in 25	1 in 2,500	Testing for the 50 most common CFTR variants that are associated with more than 95% of CF cases	Autosomal recessive; both parents must be carriers to have an affected child
Spinal Muscular Atrophy Most common genetic cause of mortality in children under two	1 in 35	1 in 10,000	Testing for the SMN1 gene deletion that is associated with more than 95% of SMA cases	Autosomal recessive; both parents must be carriers to have an affected child
Fragile X Syndrome Most common form of inherited intellectual disability	1 in 200	1 in 3,600 males 1 in 6,000 females	Testing for triplet repeat expansions in the FMR1 gene that are associated with more than 99% of FXS cases	X-linked; the mother must be a carrier to have an affected child

Medicare Criteria

As of November 1 2023, genetic carrier screening will be listed on the Medicare Benefits Schedule.

Medicare Item 73451 – screening for a female who is pregnant or planning pregnancy

Testing of a patient who is pregnant, or planning pregnancy, to identify carrier status for pathogenic or likely pathogenic variants in the following genes, for the purpose of determining reproductive risk of cystic fibrosis, spinal muscular atrophy or fragile X syndrome:

(a) CFTR
(b) SMN1
(c) FMR1

One test per lifetime.

Medicare Item – 73452 – screening for a reproductive partner of a carrier

Testing of the reproductive partner of a patient who has been found to be a carrier of a pathogenic or likely pathogenic variant in the CFTR or SMN1 gene identified by testing under item 73451, for the purpose of determining the couple's reproductive risk of cystic fibrosis or spinal muscular atrophy.
One test per condition per lifetime.

Explanatory note: The intent of MBS item 73451 is to test an asymptomatic patient of female chromosomal sex who is either planning a pregnancy or is already pregnant. The intent of MBS item 73452 is to test an asymptomatic patient of male chromosomal sex who is the reproductive partner of the female patient tested under item 73451.

Arranging Genetic Carrier Screening

Step 1: Patient consultation

- Discuss carrier screening with your patient as recommended by clinical guidelines
- Order Genetic Carrier Screening on a standard request form, noting any family history or pregnancy , and if the reproductive partner is a known carrier

Step 2: Sample collection

- Patient attends collection centre with their signed request form
- Blood is collected
- Genetic Carrier Screening is performed

Step 3: Result discussion

- Results are delivered to you by your preferred method
- Genetic counselling is provided for couples who are identified as carriers

References:

1. https://www.nature.com/articles/gim2017134
2. https://ranzcog.edu.au/wp-content/uploads/2022/05/Genetic-carrier-screeningC-Obs-63New-March-2019_1.pdf
3. MSAC application 1573 http://www.msac.gov.au/internet/msac/publishing.nsf/Content/1573-public

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