Genetic Carrier Screening

Helping your patients to make informed reproductive choices

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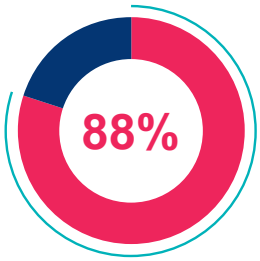
Carrier screening for informed decisions

Genetic carrier screening is DNA-based testing to identify individuals or couples at increased risk of having children with severe inherited genetic disorders, so they can make informed reproductive decisions.

Our genetic carrier screening tests for three of the most common severe genetic conditions: cystic fibrosis (CF), fragile X syndrome (FXS) and spinal muscular atrophy (SMA).

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]

Why screening is important

Genetic carrier screening helps to inform reproductive decision-making.

It gives carrier couples (where both partners are carriers for CF or SMA or the female partner is a carrier for FXS) the opportunity to consider a range of reproductive options.

One in 20 Australians will be a carrier for at least one of these conditions.[1] Most will not have a family history of the disease.[1]

1 in 20 chance of being a carrier for one of these conditions[1]

When to offer screening

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, FXS and SMA.

There is global consensus that genetic carrier screening is best performed before pregnancy. This gives your patients the widest range of reproductive choices and more time to make important decisions.

Carrier screening can still be offered in early pregnancy, although options will be more limited and time-sensitive.

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

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Genetic conditions screened

CF, FXS and SMA are three of the most common severe genetic disorders and can have devastating effects on life-expectancy and quality of life.

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
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
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

Accuracy of screening results

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

>95% of CF carriers

>99% of FXS carriers

>95% of SMA carriers

Genetic carrier screening tests are highly accurate and reliable. However, genetic carrier screening cannot identify a small percentage of carriers (1-5%) because some very rare genetic variants cannot be detected by the test.

Genetic counselling

We offer genetic counselling free of charge for all carrier couples. This professional service provides information about the condition and enables in-depth discussions about a range of options.

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Medicare criteria

Genetic carrier screening for CF, FXS and SMA is now listed on the Medicare Benefits Schedule.

Medicare Item 73451
Patient who is pregnant or planning pregnancy

One test per lifetime for 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 CF, SMA or FXS: (a) CFTR (b) SMN1 (c) FMR1

Medicare Item – 73452
Reproductive partner of a carrier

One test per condition per lifetime for 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 CF or SMA.

Arranging genetic carrier screening for your patients

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.

2

Sample collection

Your patient has a blood sample collected at their local TML Pathology collection centre.

3

Genetic carrier screening

Our expert Genomic Diagnostics team uses the DNA from the blood sample to test for the most common genetic changes that cause CF, FXS and SMA.

4

Results

We deliver the results to you within 10 working days and offer genetic counselling for couples who are identified as carriers.

Learn more:

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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