Sonic Genetics
Quality is in our DNA

Understanding reproductive carrier screening options

Quick reference guide

The great majority of children are born healthy without their parents taking any particular precautions or tests. However, approximately one in 25 children (4%) is born with some permanent condition which developed during prenatal growth. There are different approaches that parents can take in response to this possibility. Various tests are available before and during pregnancy that can provide information for parents and their doctors.

A couple can choose whether they want to learn about or have these tests. There is no obligation.

Sonic Genetics offers two types of 'reproductive carrier screening'

1) A three-gene panel (Reproductive Carrier Screen)
2) A comprehensive panel testing for >400 genes (Beacon Expanded Reproductive Carrier Screen)

To assist patients and their doctors to make an informed decision on this type of testing, we have summarised the key points for each test overleaf.

An important part of pre-pregnancy planning

Most children with a familial genetic condition do not have a family history of that condition. A family history of a genetic condition may be an important clue that there is an increased chance of a condition, but most affected children do not have such a family history.

A reproductive carrier screen is a test of the parents to look for genetic errors (mutations) that are unlikely to cause a problem in the parent, but which could cause a serious genetic condition in a child.

Timing of carrier screening

The ideal time for carrier screening is before pregnancy. This gives a couple time to consider the issues that may arise from the test result and make informed choices about their reproductive plans.

Reproductive carrier screening can also be done in early pregnancy. This is less suitable, as there is less time for issues to be considered and decisions made, and there are fewer options available to the couple.

Accessing the tests

As with all pregnancy-associated tests, reproductive carrier screening can raise questions of ethics and choice that may require time to consider. In the first instance, a couple should discuss any questions or concerns with their doctor.

Choosing a carrier screen to inform a couple's reproductive choices

	Reproductive Carrier Screen	Beacon Expanded Reproductive Carrier Screen
Specimen required	Blood sample (or cheek swab, by arrangement)	Blood sample
Number of conditions screened (female)	3 common conditions in Australia: Cystic fibrosis, Fragile X syndrome, spinal muscular atrophy	>400 severe childhood-onset conditions with limited therapies
Number of conditions screened (male)	2 common conditions in Australia: Cystic fibrosis, spinal muscular atrophy	>350 severe childhood-onset conditions with limited therapies
Chance of a person being shown to be a carrier	6 in 100 individuals (6%)	75 in 100 individuals (75%)
Chance of a couple being shown to be at increased reproductive risk	1 in 160 couples (0.6%)	1 in 20 couples (5%)
Cost (per person)	$385*	$595*
Medicare rebate	May be available if a relative or partner is known to be a carrier#	No rebate available
Combined report for a couple available	Available in 2020*	Yes
Result turnaround time	Up to 2 weeks	Up to 5 weeks
Testing laboratory	Accredited Australian laboratory in the Sonic Healthcare network	Accredited US laboratory by Fulgent Genetics
Prenatal testing available	Yes	Yes
Can the test be performed in pregnancy?	Yes	Yes
Is privacy of data assured?	Yes	Yes
Counselling (pre-test)	Available through local healthcare providers or privately	
Counselling (post-test)	Provided free-of-charge* within 3 days of receiving a result indicating a high chance of having a child with a condition; please refer to www.sonicgenetics.com.au/rcs/gc for eligibility criteria	

A GP or specialist may request MBS-rebated carrier testing of a woman for Fragile X syndrome if she has a relative who is known to be a carrier of (or is affected by) Fragile X syndrome OR a specialist (but not a GP) may request MBS-rebated carrier testing of a person for cystic fibrosis if the reproductive partner or a third-degree relative (or closer) is known to be a carrier of (or is affected by) cystic fibrosis.
These conditions are stipulated in the Medicare Benefits Schedule August 2019

*Correct at time of printing

For further information, please refer to our website, www.sonicgenetics.com.au or call us on 1800 010 447

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