Sonic Genetics

Quick reference guide

Understanding reproductive carrier screening options

SONIC HEALTHCARE

Sonic Genetics offers two types of reproductive carrier screening

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 below.

	Reproductive carrier screen (three-gene panel)	Beacon reproductive carrier screen (expanded panel)
Specific request form required	No	Yes
Specimen required	Blood sample	Blood sample (or cheek swab, by arrangement)
Number of conditions screened (female)	3 common genetic conditions: Cystic fibrosis, fragile X syndrome, spinal muscular atrophy	>400 serious childhood-onset conditions with limited therapies
Number of conditions screened (male)	2 common genetic conditions: Cystic fibrosis, spinal muscular atrophy^	>350 severe childhood-onset conditions with limited therapies
Chance of a person being shown to be a carrier	1 in 20 individuals (5%)	3 in 4 individuals (75%)
Chance of a couple being shown to be at increased reproductive risk	1 in 240 couples (0.4%)	1 in 20 couples (5%)
Cost	$385* Male partners of carriers for cystic fibrosis or spinal muscular atrophy are tested free-of-charge	$595* per person
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	No	Yes
Result turnaround time for individual test (or couple tested together)	Within 2 weeks	Within 5 weeks
Testing laboratory	Accredited Sonic Healthcare laboratory (Australia)	Accredited Fulgent Genetics laboratory (US)
Counselling (pre-test)	Available through local healthcare providers or privately	
Counselling (post-test)	Provided free-of-charge to couples tested by Sonic Genetics and found to be at high risk of having an affected child (details on how to refer eligible couples will accompany the results).	

^Sonic Genetics does not recommend reproductive carrier testing for fragile X syndrome in unaffected male patients as this is an X-linked disorder. Testing can be arranged if clinically indicated on the basis of family history or clinical features.

#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 GP or specialist may request MBS-rebated carrier testing of a person for cystic fibrosis if a third-degree relative (or closer) is known to be a carrier of (or is affected by) cystic fibrosis and the mutation is documented. A specialist may also request an MBS rebated test if the reproductive partner is either affected or a carrier. Also, if the fetus has an echogenic gut. These conditions are stipulated in the Medicare Benefits Schedule July 2021.

*Correct at time of printing | July 2021

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

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