



Understanding reproductive carrier screening options

Quick reference guide

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 below.

	Reproductive carrier screen	Beacon expanded reproductive carrier screen
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 severe 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	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	\$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	Up to 2 weeks	Up to 5 weeks
Testing laboratory	Accredited Sonic Pathology Australia laboratory	Accredited US laboratory Fulgent Genetics
Counselling (pre-test)	Available through local healthcare providers or privately	
Counselling (post-test)	Provided free-of-charge* within 3 days of receiving a referral for genetic counselling of an eligible couple; 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 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 August 2020.

[^]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.

*Correct at time of printing