

Quick reference guide

Understanding reproductive carrier screening options

Sonic Genetics offers two types of reproductive carrier screening. We have summarised the key points for each test below. An online education course is also available for patients. The course intends to supplement pre-test discussions. Complete your registration at patientedu.sonicpathology.com.au

	Reproductive carrier screen (three-gene panel)	Expanded reproductive carrier screen (Beacon)
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, spinal muscular atrophy, fragile X syndrome	>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 44 couples (2%) ¹
Medicare rebate	Yes [#]	No, (\$595* per person)
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	Provided at no additional cost 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.

[#]Medicare rebates apply, subject to criteria being met.

*Please refer to our website for current pricing, sonicgenetics.com.au/rcs/beacon

Reference

- 1 Berbic M, Hesson L, Clarke J, et al. Reproductive carrier screening (RCS) to identify Australian couples at risk of having children with autosomal recessive and X-linked conditions, RANZCOG Annual Scientific Meeting, October 2022.