eugene

Our carrier screening options

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Test	 Core	 Comprehensive	 Couples (Expanded)
Genes included	**3 genes** (CFTR, SMN and FMR1)	**787 genes** (View full gene list)	**620+ genes** (View full gene list)
Who	Biological females screened first, male reproductive partner only screened if female is a carrier of CFTR on SMN	For individuals, reproductive couples and those using donor gametes	Reproductive couples (male–female only)
Turn around time	**Up to 4 weeks** Sequential testing of male partner adds 2 weeks to TAT	**Up to 4 weeks**	**6–8 weeks** If either partners' sample fails + 6–8 weeks for recollection. No individual reporting available unless testing is completed
Cost	**$0** Medicare eligibility required*	**$949** Individual **$1499** Couple	**$949** $949 is the out of pocket cost Medicare eligibility required*
When	RANZCOG & RACGP guidelines recommend that all couples planning pregnancy or in 1st trimester should be offered carrier screening		
Reporting	• Individual carrier status • Limitation: only common gene variants are included in this assessment.	• Individual carrier status for all conditions • Secondary finding with potential personal implications. • Reproductive risk (low, increased risk)	• Reproductive risk (low or increased risk) • NB. Individual carrier results for CF, SMA and FXS for female only
Conditions	• Cystic fibrosis (CF), spinal muscular atrophy (SMA) and fragile X syndrome (FXS)	• Screens for 100s of genetic conditions relevant to more ethnicities. • Includes conditions that may have severe presentations (cognitive or physical impairments), early childhood onset, or for which early intervention may be beneficial • Full sequencing of CF, SMA and FXS	• Screens for 100s of genetic conditions relevant to more ethnicities • Focuses on severe childhood onset conditions • Screening for common causes of CF, SMA and FXS
Pick up rates of at-risk couples	1 in 240 couples will be at increased risk (0.42%)	1 in 50 couples will be at increased risk when screened for over 750 conditions (~2%)	1 in 40 couples will be identified at increased risk (2–3%)

*The medicare rebate can only be claimed once per lifetime if the test includes CF, SMA and FXS