

# the partumpost

## Carrier screening for CF, FXS & SMA

Volume 1 Issue 5 November 2017

### Why is carrier screening important?

Genetic carrier screening gives individuals and couples information about their chance of having a child with an inherited genetic condition. Carrier screening for autosomal recessive and X-linked conditions is available to those planning a pregnancy or in the early stages of pregnancy. Collectively, these genetic conditions affect approximately 1 in 700-1000 pregnancies (1,2).

Recognising the importance of screening for these conditions, RANZCOG and HGSA have issued a consensus-based recommendation that carrier screening for CF (cystic fibrosis), FXS (fragile X syndrome) and SMA (spinal muscular atrophy) may be offered to women at low risk for these conditions (i.e. those without a family history) (3, recommendation #6).

This position has been strengthened by a recent statement from the American College of Obstetricians and Gynecologists that recommends 'information about carrier screening should be provided to every pregnant woman' (4).

### What conditions should be screened?

Technological advances are enabling the development of panels that provide carrier screening for a larger number of conditions.

Criteria exist for evaluating which conditions warrant inclusion in these panels and indicate that the conditions should be (i) relatively common, (ii) have a serious impact on the affected individual and their family, (iii) have a highly sensitive test available, and (iv) can be diagnosed prenatally (5). CF, FXS & SMA are 3 conditions that clearly satisfy these criteria.

### Why should carrier screening for CF, FXS & SMA be offered?

We have recently published a review of outcomes of 12,000 *prepair* genetic carrier screening tests (Fig 1) for CF, SMA and FXS which highlights the importance of offering carrier screening for these conditions (1).

*Anybody can be a carrier.*

1 in 20 people were identified as a carrier of one or more of these conditions. 88% had no known family history of the condition prior to testing which is consistent with the published literature (6).

*Collectively, these conditions are as common as Down syndrome.*

Of those couples pregnant at the time of testing, 1 in 240 were found to be at high risk of having a child with one of these conditions.

1 in 1006 women pregnant at the time of testing had a pregnancy affected by one of these conditions. This is equivalent to the chance of a 29 year old woman having a baby with Down syndrome (7).

While screening for chromosome conditions like Down syndrome is available to all pregnant women, research suggests that most healthcare providers are not routinely offering carrier screening to their patients pre-conception or in early pregnancy (8).

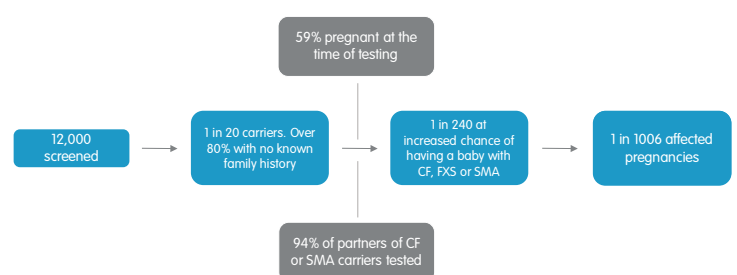


Figure 1. Outcomes from 12,000 *prepair* carrier screening tests.

*Clinical support is key to offering carrier screening.*

RANZCOG and HGSA recommend that people considering carrier screening should be appropriately informed of the benefits and limitations and any associated costs (3), (recommendation #6).

Local research has shown that women can be offered testing and make informed decisions about carrier screening without adverse psychosocial consequences; the researchers highlighted the importance of testing being offered with available genetic counselling (9).

Our research indicates that carriers identified through *prepair* testing valued access to accurate information and support from genetics experts (10). It is important to note that identifying a carrier has implications for the broader family.

Our genetic counsellors are available to provide expert information about tests and results and can help co-ordinate further testing including carrier testing of other family members. Appointments with a genetic counsellor and clinical geneticist are available if required.

## What do I need to know when offering *prepair*<sup>TM</sup> carrier screening?

**We recommend testing the female partner first:** It is important to screen the female partner first because carrier testing for FXS is not relevant for males.

Although males can be carriers of FXS, they are not at risk of having a child with FXS because the repeated sequence does not expand to cause FXS when passed from father to daughter. Screening the woman first also minimises the cost for couples; if she is not a carrier of any of these conditions, their risk of having an affected child is low and no further testing is required.

***prepair*<sup>TM</sup> is a comprehensive carrier screening service which includes genetic counselling for anyone considering or undergoing screening.**

**Testing can be performed using blood or saliva:** simply complete the *prepair* test request form or contact the *prepair* team for saliva kits. Testing with saliva is accurate and often more convenient for patients.

## References

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6. McClaren, B.J., et al., A case for cystic fibrosis carrier testing in the general population. *Med J Aust*, 2011. 194(4): p. 208-9.
7. Hook, E.B., et al., Chromosomal abnormality rates at amniocentesis and in live-born infants. *JAMA*. 1983. 249(15):2034.
8. Valante, G., Population cystic fibrosis carrier screening in Victoria: Health professionals' attitudes, knowledge and practice patterns. Master of Genetic Counselling, Department of Paediatrics, University of Melbourne, 2015.7.
9. Metcalfe, S.A., et al., Informed decision making and psychosocial outcomes in pregnant and nonpregnant women offered population fragile X carrier screening. *Genet Med*, 2017.
10. Beard, C.A., et al., "I'm Healthy, It's Not Going To Be Me": Exploring experiences of carriers identified through a population reproductive genetic carrier screening panel in Australia. *Am J Med Genet A*, 2016. 170(8): p. 2052-9.

**Results are available within 10 working days:** all carrier results are phoned through to the referring doctor. Genetic counselling is available to all individuals and extended family members at no further cost.

## What about other carrier screening options - is more better?

Expanded carrier screening panels that test for well over 100 genetic conditions are available. Understanding the conditions included in these panels is important as not all meet recommended inclusion criteria.

**Expanded panels can be problematic if conditions are included that are not relevant to pregnancy planning or when results reported have uncertain outcomes, resulting in unnecessary anxiety for couples.**

Careful consideration for expanded panels is required regarding the conditions to be included and the clinical utility of the information screening may provide.

Our multidisciplinary team of medical geneticists, genetic counsellors, specialist physicians and laboratory scientists is currently developing our own unique expanded carrier screening panel.

This panel will include only those genetic conditions that meet the recommended criteria listed above. A review of expanded carrier screening panels will follow in a future edition of the *partumpost*.

*For more information about *prepair*<sup>TM</sup> carrier screening please contact the screening team on +61 3 9936 6402.*

**Please email [vcgs@vcgs.org.au](mailto:vcgs@vcgs.org.au) for any questions, topic suggestions or to subscribe to the *partumpost*.**