VCGS
Leaders in Genetic Health

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prepair™ genetic carrier screening

prepair™ by VCGS: reproductive genetic carrier screening

Genetic carrier screening gives individuals and couples information about their chance of having a child with a genetic condition. Specifically, prepair™ identifies carriers of cystic fibrosis (CF), fragile X syndrome (FXS) or spinal muscular atrophy (SMA). These are considered common inherited conditions with significant health impacts.

Why should I offer prepair™?

These conditions are common: 1 in 20 people screened are a carrier for at least one of CF, FXS or SMA.

Often no family history: Most identified carriers have no family history of the condition.

Accurate tests available: Detection rates are high with 90% of CF, 95% of SMA and >99% of FXS carriers detected.

RANZCOG/HGSA & research support screening: The joint RANZCOG/HGSA committee for prenatal screening recommend offering screening* for CF, FXS and SMA to all women, as does published research.

Cystic fibrosis

CF is an inherited condition affecting breathing and digestion. Individuals with CF require daily chest physiotherapy to clear mucus from their lungs, frequent courses of antibiotics, and need to take medicine to aid digestion. There is no cure for CF but better treatments are under research and development. CF is a life shortening condition.

Fragile X syndrome

FXS is the most common cause of inherited intellectual disability. Males and females with FXS can have developmental delay, learning difficulties, anxiety, autism and epilepsy. Some females who are carriers of FXS may have early menopause.

Spinal muscular atrophy

SMA is a condition that affects nerves in the spinal cord and causes muscles to get weaker. SMA type 1 is the most common and most severe. Babies with SMA type 1 usually do not live past two years of age.

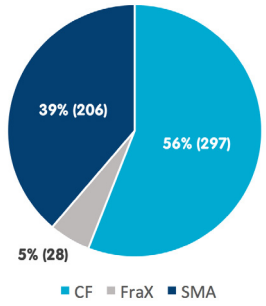
There is no cure for these conditions although treatments and interventions are available to help improve quality of life.

Carrier rates in the population

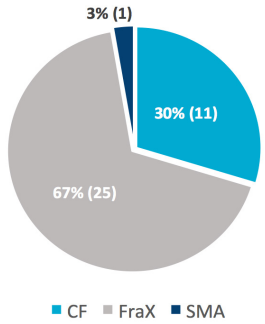
	Number of people who are carriers of the condition	Number of people with the condition
Cystic fibrosis	1 in 25	1 in 2,500
Fragile X syndrome	1 in 150	1 in 4,000
Spinal muscular atrophy	1 in 40	1 in 6,000 - 1 in 10,000

Our experience with carrier screening

10,000 tests	
528	carriers of one or more conditions (9 carriers of 2 conditions)
37	couples at increased risk
25	pregnant at time of testing (CF-8, FXS-16, SMA-1)
22	opted for prenatal diagnosis (CF-8, FXS-13, SMA-1) (6 affected: CF-3, FXS-2, SMA-1)
	couples at increased risk used prenatal diagnosis and/or PGD for subsequent pregnancies



Proportion of carriers by condition (n=528)



Proportion of increased risk couples (n=37)

VCGS is a not-for-profit provider of a comprehensive range of clinical and laboratory genetics services. We provide genetic counselling support before and after testing. This program is offered in partnership with patient support groups.

*HGSA/RANZCOG joint committee on Prenatal Diagnosis and Screening, March 2015 - C-Obs 59.