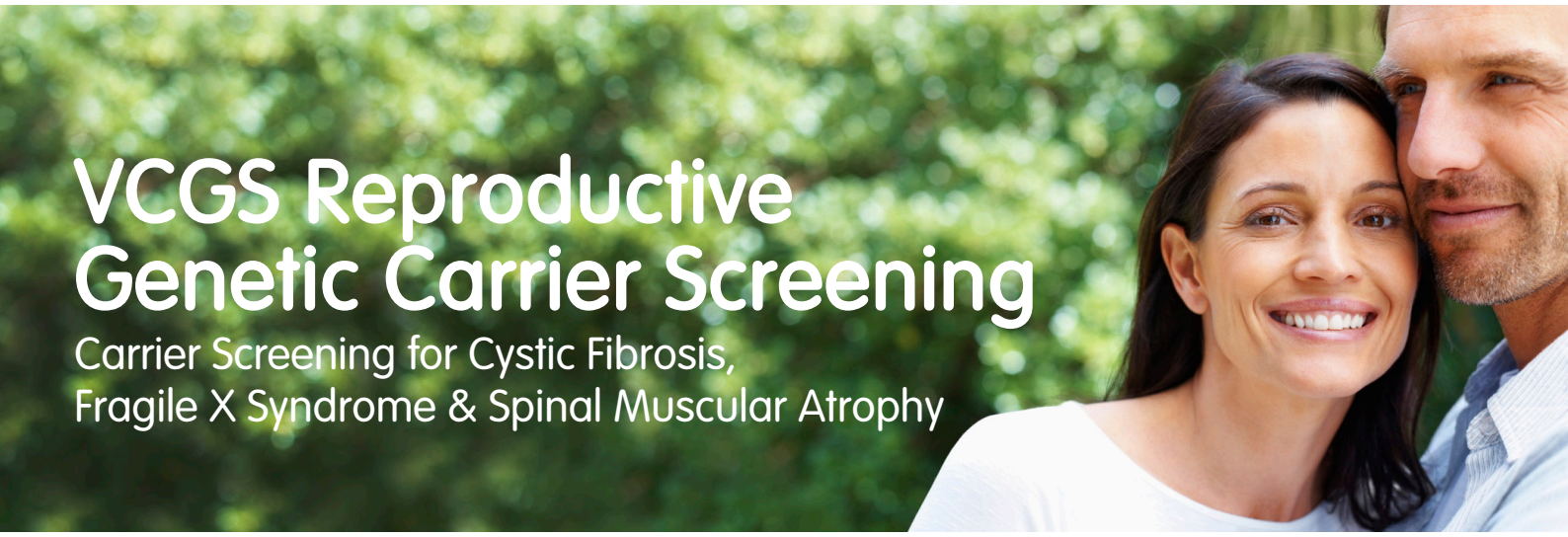
VCGS
Leaders in Genetic Health

FOR FURTHER INFORMATION CONTACT:
Victorian Clinical Genetics Services
Murdoch Childrens Research Institute
Flemington Road, Parkville VIC 3052 Australia
P (03) 8341 6201 W vcgs.org.au



VCGS Reproductive Genetic Carrier Screening

Carrier Screening for Cystic Fibrosis, Fragile X Syndrome & Spinal Muscular Atrophy

Genetic carrier screening

Genetic carrier screening gives individuals and couples information about their risk of having a child with a genetic condition. The VCGS reproductive carrier test is a simple blood test which can be arranged through your doctor. It will tell you if you are a carrier for three common inherited conditions: cystic fibrosis (CF), fragile X syndrome (FXS) and spinal muscular atrophy (SMA).

Many people are carriers of CF, FXS or SMA even though they do not have anybody in their family who has the condition.

The VCGS reproductive carrier screening test looks for the most common gene changes associated with CF, FXS and SMA in Australian children. It will identify about 90% of people who are carriers of CF, 95% of people who are carriers of SMA and over 99% of people who are carriers of FXS. However, the test will not detect every person who is a carrier.

What is the chance that I could be a carrier?

	NUMBER OF PEOPLE WITH THE CONDITION	NUMBER OF PEOPLE WHO ARE CARRIERS OF THE CONDITION
CF	1 in 2500	1 in 25
FXS	1 in 4000	1 in 150
SMA	1 in 6000 – 1 in 10,000	1 in 40

What causes these conditions?

Genetic conditions are caused by changes in genes, which provide the instructions for our bodies. Babies inherit one copy of each gene from each parent. Some people carry changes in a gene which can mean there is a chance that they may have a child with a genetic condition.

- CF and SMA occur when a baby inherits a gene change from both parents.
- FXS is caused by an increase in the length of the *FMR1* gene. The FXS carrier test will tell you whether you carry the low risk gene or the increased risk gene. Women who carry the increased risk gene may have a child with FXS.

How the test process works

1. Decide whether or not to have the test:

   It is your choice whether or not to have the test. Read through the information provided and discuss the test with your doctor. **It is recommended that the female member of the couple is screened first as FXS testing is not necessary for the male partner.**

   If you would like to talk to a genetic counsellor about the test please call 8341 6201.

2. Consider whether it is the right time to have the test:

   To ensure there is time to make decisions it is best to have this test either before pregnancy or early in pregnancy. If you are pregnant, we recommend having screening before 12 weeks of pregnancy.

3. Request the test:

   If you would like to have the test, your doctor will need to complete the attached test request slip. As yet there is no Medicare rebate for this test and there will be a charge ($385).

4. Have your blood sample taken:

   Take the completed request slip with you to have your blood sample taken. This can be done at any pathology service. Your blood sample will be sent to VCGS where it will be processed.

5. Receive your result:

   It will take approximately 10 working days for your test to be processed and your results to be sent to your doctor. Your doctor will discuss your results with you.

6. If the test shows you are a carrier, talk to a genetic counsellor:

   Your result will tell you if you are a carrier of CF, FXS or SMA. If the test shows you are a carrier, your partner may need to have carrier testing and it is recommended that you speak to a genetic counsellor. Genetic counsellors are trained to talk to people about genetic conditions and genetic testing, to help with making decisions and to provide support. To speak to a genetic counsellor at any point please call: 03 8341 6201.

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UPDATED APRIL 2013

What is cystic fibrosis?

CF is an inherited condition affecting breathing and digestion. CF causes thick mucus which traps bacteria, resulting in recurrent infections that damage the lungs. Thick mucus in the gut also makes digestion of food difficult. Infants, children and adults with CF require daily chest physiotherapy to clear mucus from their lungs, frequent courses of antibiotics, and need to take medicine to aid digestion. Until recently, many children with CF died in early childhood but now many live to be 30, 40 or more. There is no cure for CF but better treatments are under research and development.

What is fragile X syndrome?

FXS is the most common cause of inherited intellectual disability. People with FXS can have developmental delay, learning difficulties, anxiety, autism and epilepsy. The features of FXS vary from mild to severe with males more likely to be severely affected than females. There is no cure for FXS although some educational, behavioural and medical interventions can improve outcomes for people with FXS.
Some females who are carriers of FXS may have early menopause.

What is spinal muscular atrophy?

SMA is a condition that affects nerves in the spinal cord and causes muscles to get weaker. There are four types of SMA. SMA type 1 is the most severe. Babies with SMA type 1 have weak muscles from birth and usually do not live past 2 years of age. SMA types 2 and 3 progress more slowly than type 1. Most children with SMA type 2 or 3 are unable to stand and walk without help. Children with types 2 and 3 SMA can live into early adulthood, depending on the severity of the condition. People with SMA type 4 do not develop symptoms until adulthood. There is no cure for SMA, however there are treatments and interventions available aimed at managing symptoms and improving quality of life.

If the test shows that I am a carrier, what is the chance I will have a child with the condition?

If your test shows that you have one copy of the gene change for CF or SMA, you are a carrier of this condition. However, a couple is only at risk of having a child with CF or SMA if both the father and the mother are carriers of that condition. If you are a carrier of CF or SMA your partner will be offered testing. Two people who are carriers of the same condition have a 1 in 4 (25%) chance of having a child with the condition for each pregnancy.

For FXS, only women who carry the increased risk gene are at risk of having a child with FXS. This means that your partner will not need to be tested for FXS. Female carriers of the increased risk gene have a 1 in 2 (50%) chance of passing that gene onto each child that they have.

If the test shows that I am a carrier, can I develop symptoms?

If you are a carrier of CF or SMA you have a change in one of your two genes associated with these conditions and carriers do not develop symptoms. Some female carriers of FXS may develop fertility problems and go through menopause early (before 40 years of age). Some male and a small number of female carriers of FXS may develop a late-onset neurological condition which causes tremors and balance problems which worsen with age.

If the tests show that we are at risk of having a child with CF, FXS or SMA, what happens next?

If you or your partner are carriers of CF, FXS or SMA you will be offered genetic counselling. The genetic counsellor will talk to you about your results, options for further testing and can help with making decisions and providing support. A number of options are available to women who are carriers of FXS and couples who are both carriers of CF or SMA. Genetic testing for these conditions can be done during pregnancy. If the tests diagnose CF, FXS or SMA, you have a choice about whether to continue or terminate the pregnancy. If you are identified as a carrier prior to pregnancy, there are a number of family planning options available to you including, pre-implantation genetic diagnosis using in vitro fertilisation. If you are a carrier, genetic carrier testing is also available to your blood relatives.

Do I need to have carrier testing every time I have a baby?

NO, you only need to have this test once in your lifetime. If you are not a carrier, you remain at very low risk of having a child with the condition. If you are a carrier and you have a new partner, your new partner may need to be tested.

If no gene change is found, could I still be a carrier?

YES. The current carrier test detects the majority of carriers but it cannot detect every gene change that causes CF, FXS and SMA. If no gene change is found, you are not a carrier of the most common gene changes, but there is still a small risk for each condition that you are a carrier of rarer gene changes.

What if I have a relative who is a carrier or has CF, FXS or SMA?

If you have a family history of one or more of these conditions, YOUR CHANCE of being a carrier is GREATER than most people and you and your partner should consider testing. If you or your partner has a family history of any of these conditions contact VCGS to discuss testing.

Contact Details:

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Flemington Road, Parkville VIC 3052
P (03) 8341 6201 W vcgs.org.au
E carrierscreening@vcgs.org.au

CYSTIC FIBROSIS Victoria

Cystic Fibrosis Victoria
Ph: (03) 9686 1811
www.cysticfibrosis.org.au/vic

The Fragile X Association of Australia

Fragile X Association of Australia Inc
Ph: 1300 394 636
www.fragileX.org.au

Spinal Muscular Atrophy Association of Australia Inc.
Ph: (03) 9545 3633
www.smaaustralia.org.au

To order more information brochures and test request slips call: (03) 8341 6201

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PATHOLOGY REQUEST FORM VCGS REPRODUCTIVE GENETIC CARRIER SCREENING

Victorian Clinical Genetics Services, Murdoch Childrens Research Institute
Flemington Road, Parkville VIC 3052 Australia
P +61 3 8341 6201 F +61 3 8341 6212 W vcgs.org.au

UPDATED APRIL 2013

PATIENT DETAILS

patient LAST NAME | GIVEN NAMES | SEX | DATE OF BIRTH | LABORATORY REF:

patient ADDRESS | POSTCODE | PHONE (home) | MOBILE

TESTS REQUESTED

VCGS REPRODUCTIVE GENETIC CARRIER SCREENING:

4ml EDTA BLOOD

NON-PREGNANT TESTING ☐ PREGNANT TESTING ☐

GESTATION (weeks): _____

EDD: _____

LABORATORY COPY

DOES THE PATIENT OR THEIR PARTNER HAVE A FAMILY HISTORY OF CF, SMA or FXS?

PATIENT ☐ NO ☐ YES

PARTNER ☐ NO ☐ YES

IF YES, PLEASE SPECIFY: _____

NOTE: If the individual or partner has a family history of CF, SMA or FXS contact the screening program genetic counsellor on: (03) 8341 6201

IT IS RECOMMENDED THAT SAMPLES BE TAKEN BEFORE 12 COMPLETED WEEKS GESTATION
PLEASE COMPLETE THE DETAILS OF BOTH PARENTS SO THAT THE RESULTS CAN BE LINKED IF NECESSARY
BLOOD SAMPLES FOR THIS TEST CAN BE TAKEN AT ANY PATHOLOGY SERVICE
Send sample to: Victorian Clinical Genetics Services

SIGNATURE: _____ PERSON COLLECTING SPECIMEN

TIME OF COLLECTION: _____

DATE OF COLLECTION: ____/____/_____

DOCTOR'S SIGNATURE AND REQUEST DATE

X_____ **DATE:** ____/____/____

COPY REPORTS TO

REQUESTING DOCTOR (Provider Number, Initials and Address)

PARTNER DETAILS

partner's LAST NAME | GIVEN NAMES | DATE OF BIRTH

SEND SAMPLES TO:

MOLECULAR GENETICS LABORATORY
VICTORIAN CLINICAL GENETICS SERVICES
MURDOCH CHILDRENS RESEARCH INSTITUTE
4TH FLOOR, EAST BUILDING
ROYAL CHILDRENS HOSPITAL, FLEMINGTON RD,
PARKVILLE VIC 3052

The report will be sent to the requesting practitioner

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patient LAST NAME | GIVEN NAMES | SEX | DATE OF BIRTH | LABORATORY REF:

patient ADDRESS | POSTCODE | PHONE (home) | MOBILE

TESTS REQUESTED

VCGS REPRODUCTIVE GENETIC CARRIER SCREENING.

YOUR SAMPLE HAS BEEN SENT TO:

MOLECULAR GENETICS LABORATORY
VICTORIAN CLINICAL GENETICS SERVICES
MURDOCH CHILDRENS RESEARCH INSTITUTE
4TH FLOOR, EAST BUILDING
ROYAL CHILDRENS HOSPITAL, FLEMINGTON RD,
PARKVILLE VIC 3052

PATIENT COPY

Your result will be sent to the doctor who ordered your test

REQUESTING DOCTOR (Provider No, Surname, Initials, Address)