Arizona Associates for Reproductive Health

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GENETIC CARRIER SCREENING

Genetic carrier screening is done to determine if one or both parents have abnormal genes that increase the chance that their child will have a specific genetic disease. For many genetic diseases, if someone has an abnormal gene, that person is considered a carrier for that genetic disease. If this abnormal gene is passed to the child, the child will usually not be affected with that genetic disease but will be a carrier for that genetic disease. If both parents are carriers of the abnormal gene for the same genetic disease, there is a 25% chance that their child will inherit one abnormal gene from each parent and be affected with that genetic disease. Genetic screening is typically done on one partner first, and if the first partner tests positive, then the other reproductive partner is tested.

The American College of Obstetricians and Gynecologists (' ACOG ') and the American College of Medical Geneticists and Genomics ('ACMG') recommend routine screening for certain genetic diseases and additional screening when indicated due to ethnicity, family history, or other known risk factors. Two of the recommended standard genetic carrier screening tests include Cystic Fibrosis ('CF') and Spinal Muscular Atrophy ('SMA').

During the course of your evaluation and treatment at Arizona Associates for Reproductive Health, your physician may recommend additional genetic carrier screening for specific genetic disease(s) which may be indicated based on your medical history and/or family history to determine whether or not you are a carrier for the specified genetic disease(s). Also, there are certain genetic carrier screening tests that may be ordered by your physician based on your ethnic or racial background.

Although genetic carrier screening tests are a valuable tool there are limitations:

- ***Negative result*** - The genetic testing laboratory usually tests for the most common mutations (change in gene structure) and may not identify the less common mutations. So it is possible to have a negative test result but still have a genetic mutation that was not or could not be identified by the testing laboratory due to limitations of current technology.
- ***Positive result*** - A positive test result indicates that you are a carrier for a genetic mutation that can cause a specific genetic disease or can put you and/or your child at risk for developing a disease. If you are determined to be a carrier, your reproductive partner will then be advised to undergo genetic carrier testing.
- ***Inconclusive result*** - Sometimes it is not possible for the testing laboratory to determine genetic mutations. In this case, the genetic carrier test may need to be performed again at the same or different testing laboratory.

It may take 2 to 3 weeks to receive the genetic carrier screening test results and testing should be performed prior to beginning fertility treatment. Additionally, if you are concerned with passing an inherited (genetic) disorder to your offspring, you have the option of speaking with a genetic counselor, who may suggest additional preconception tests or post-conception (prenatal) procedures such as amniocentesis.

I/We acknowledge:

- The genetic carrier screening tests that have been recommended to me are voluntary and if I choose to have them performed, it will be to determine if I am a genetic carrier (within the limitations of the test) of the conditions being tested.
- Fertility treatment **cannot** proceed until I/we have made a decision regarding genetic carrier screening and, if genetic carrier screening is not declined, the genetic carrier screening results are returned indicating genetic carrier screening for my reproductive partner is not indicated.
- The importance of genetic counseling to further discuss the genetic test(s) and how it may affect me and my family. Information about genetic counseling services has been provided to me.
- The cost of the recommended testing may not be covered by my insurance plan and I will be responsible for the costs associated with the genetic carrier testing.
- It may be possible that even with a negative result I still could be a genetic carrier or predisposed to the genetic condition being tested due to the limitations of the genetic testing.

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Initials

- The genetic test results will be contained in my medical record and the Center will take all appropriate safeguards to protect the confidentiality of my personal health information. However, I hereby consent to the release of these results to my partner, the physicians and nursing staff directly involved in my care for the purpose of my treatment, my current insurance carrier in order to seek coverage or reimbursement, and others specifically authorized by law to gain access to my medical records.
- Current Federal & State laws prohibit discrimination by insurance carriers based on results of genetic carrier screening tests.
- I will release my genetic carrier screening test results to my partner. It has been recommended that I discuss the results of my genetic carrier screening test with family members.
- The recommended testing should be performed and results should be reviewed prior to initiating any fertility treatment. Proceeding with fertility treatment without the benefit of my genetic carrier status could result in an unfavorable outcome including a child with a genetic disorder.

ACKNOWLEDGMENT

I/We have met with my/our physician and healthcare team to discuss genetic carrier screening. I/We have been fully informed of the purpose of the genetic carrier screening, reliability of the screening results, risks and benefits of agreeing to or declining genetic carrier screening and/or genetic counseling and available alternatives to genetic carrier screening. I/We have had the opportunity to ask questions and have all my/our questions answered to my/our satisfaction.

I/We have made the following decision regarding genetic carrier screening, free from coercion:

_____ _____ I/We consent to genetic screening and elect:

_____ _____ Arizona Associates for Reproductive Health selected Counsyl Panel

_____ _____ Counsyl Full Panel

_____ _____ Genetic screening by blood tests

_____ _____ I/We have already had genetic screening prior to coming to Arizona Associates for Reproductive Health

_____ _____ I/We decline genetic screening and understand that this is against the advice of Arizona Associates for Reproductive Health physicians. I/We agree to hold Arizona Associates for Reproductive Health, it's physicians, agents and staff harmless from any and all liability should any consequences occur secondary to my declination of genetic carrier screening.

_____ _____ I/We would like more information about genetic carrier screening before making a decision. I/We understand that fertility treatment **cannot** proceed until I/we have made a decision to consent to or decline genetic carrier screening, have signed and dated the informed consent form, and, if the genetic carrier screening has not been declined, the genetic carrier screening results have been returned indicating that the genetic carrier screening for my reproductive partner is not indicated.

_____
Signature - Female Patient

_____
Signature - Reproductive Partner

_____
Print Name:

_____
Print Name:

_____
Date:

_____
Date:

_____
Initials

Picture Identification

Female Patient:

Type: _____ Exp. Date _____

Reproductive Partner (if applicable):

Type: _____ Exp. Date _____

Picture Identification Confirmed on: _____
Date

_____ _____
Witness - Print Name and Title Signature

_____
Initials