Pre-Conception Carrier Screening

Modified in part from ACOG: Preconception Carrier Screening https://www.acog.org/Patients/FAQs/Preconception-Carrier-Screening

**What is pre-conception carrier screening?**
Pre-conception carrier screening is screening that you can have before becoming pregnant to help predict your chances of having a child with a genetic disorder.

**What is a carrier?**
A carrier is a person who has no symptoms of a disorder but can pass on the gene for that disorder to his or her child.

**How is carrier screening done?**
Carrier screening involves testing a sample or blood or saliva. The sample is sent to a laboratory for testing. Often the partner who is most likely to have a defective gene is tested first. If test results show that the first partner is not a carrier, then no additional testing is needed. If test results show that the first partner is a carrier, the other partner is tested.

**Does pre-conception carrier screening test for all genetic disorders?**
Carrier screening does not detect all genetic disorders. Carrier tests are available for a limited number of diseases, including cystic fibrosis, fragile X syndrome, sickle cell disease, and Tay-Sachs disease. As of 2011, the American College of Obstetricians and Gynecologists (ACOG) recommends carrier screening for Cystic Fibrosis be offered to all women or reproductive age regardless of ethnicity.

**Does a normal test guarantee my child will not have a genetic disorder?**
Screening can reduce, but not eliminate, the chance for some genetic disorder. Because test results can be wrong, it is possible for you to have a child with a genetic disorder even if your and your partner’s test results are negative. A false-positive test results when a person tests positive for being a carrier but does not actually have the gene. A false-negative test result is when a person tests negative for being a carrier but actually does have the gene.

**What can the results of a carrier screening test tell me?**
If both you and your partner are carriers for the same disease, there is a 1 in 4 (25%) chance that the child will get the abnormal gene from each parent and will have the disorder. There is a 50% chance that the child will be a carrier of the disorder, just like the carrier parent. If only one parent is a carrier, there is a 50% chance that the child will be a carrier of the disorder and a 0% chance that the child will have the disorder.

**What decisions do I need to make if I am a carrier?**
If you and your partner are both carriers of a genetic disorder, you have several options. You may choose to proceed with becoming pregnant, with the option of considering prenatal diagnosis. You may choose to use in vitro fertilization to create fertilized eggs in the laboratory, follow by preimplantation genetic diagnosis on each of the embryos for the genetic disorder before transferring the embryo into the uterus to achieve a pregnancy. You may also use donor sperm or donor egg to achieve pregnancy. You may also choose not to become pregnant.

**Who should I speak to if I have more questions about pre-conception carrier screening?**
If you have questions about pre-conception carrier screening or genetic disorders in general, and especially if there is a family history of a genetic disorder, genetic counseling with a board certified geneticist is strongly recommended.

**References**
- Pre-conception Carrier Screening [https://www.acog.org/Patients/FAQs/Preconception-Carrier-Screening](https://www.acog.org/Patients/FAQs/Preconception-Carrier-Screening)
- Genetic Disorders [https://www.acog.org/-/media/Committee-Opinions/Committee-on-Genetics/co486.pdf?dmc=1&ts=20160123T1739300004](https://www.acog.org/-/media/Committee-Opinions/Committee-on-Genetics/co486.pdf?dmc=1&ts=20160123T1739300004)
- Cystic Fibrosis Screening For All [https://www.acog.org/-/media/For-Patients/faq094.pdf?dmc=1&ts=20160123T1735398281](https://www.acog.org/-/media/For-Patients/faq094.pdf?dmc=1&ts=20160123T1735398281)
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Family Fertility Center offers pre-conception carrier screening to all women of reproductive age. Currently we utilize the services of two different genetic testing companies for pre-conception carrier screening:

Counsyl’s Family Prep Screen covers screening for 100+ genetic disorders, including cystic fibrosis, fragile X, and spinal muscular atrophy. Counsyl participates in-network with most common insurance carriers in Pennsylvania, such as Aetna, Capital Blue Cross, Cigna, Coventry, First Health Network, Great-West Healthcare, Highmark, Medicare, Oxford, United Healthcare, and Valley Preferred. Some plans cover the cost of this test 100% so there is no cost to you. Some plans may have an out-of-pocket cost due to deductibles and/or copays ranging from $500 to $900, although the average out-of-pocket cost is $75 to $160. Counsyl would notify you by email or text prior to performing the test to advise of your out-of-pocket cost; and you would have an opportunity to decline the testing or elect to pay for the test out-of-network at a fee of $349. https://www.counsyl.com/services/family-prep-screen/

Progenity’s Preparent Screen offers similar testing but is out-of-network for all insurance carriers. Your insurance company would be billed but, in the event you incur any out-of-pocket expense, you can call Progenity and request your bill be reduced to a self-pay price of $99. https://progenity.com/sites/default/files/resources/Preparent_Global_Panel_Performance_05-20-2015_FINAL.pdf

PLEASE SIGN BELOW TO DESIGNATE WHETHER YOU WISH TO PROCEED WITH PRE-CONCEPTION CARRIER TESTING OR DECLINE TO UNDERGO SUCH SCREENING:

[       ] I wish to DECLINE pre-conception carrier screening despite being advised of its benefits.

[       ] I wish to PROCEED with pre-conception carrier screening by Counsyl*. I understand I will have 48 hours to decline testing after I have been advised of any costs.

[       ] I wish to PROCEED with pre-conception carrier screening by Progenity*. I understand Progenity will bill my insurance company even though it is out of network; and that I can request my cost be reduced to $99 whether or not my insurance company makes payment.

*Family Fertility Center has no financial relationship with and does not receive any kick back from any testing company. You have the right to undergoing your pre-conception carrier screening at any laboratory of your choice. The estimated cost is current as of January of 2016. Family Fertility makes no guarantee the cited cost is up-to-date. It is your responsibility to contact the particular testing company for an exact quote and to find out from your health insurance company your expected out of pocket expense. Family Fertility Center reserves the right to change the testing laboratory without further notice.

Patient Signature: ___________________________ Date:_________________